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‘WE ARE PRETTY SURE WE CAN NEVER SAY NEVER’: EXPLORING HOW RISK AND UNCERTAINTY COMMUNICATION IMPACTS CARE IN PUBLIC GENETIC COUNSELLING CONSULTATIONS IN SOUTH AFRICA

MEGAN SCOTT

A THESIS SUBMITTED TO THE FACULTY OF HUMANITIES, UNIVERSITY OF THE WITWATERSRAND, JOHANNESBURG, IN FULFILMENT OF THE REQUIREMENTS OF THE DEGREE OF DOCTOR OF PHILOSOPHY

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ABSTRACT

Introduction
Risk and uncertainty are inherently part of genetic counselling (GC) due to the complex nature of the information conveyed. Despite an emphasis on communication within the GC profession little is understood about the process of GC communication in everyday practice, particularly within socio-cultural diverse settings. The study aims to explore how risk is communicated and how uncertainty is managed during public-based South African GC consultations and how these processes impact patient care.

Methods
This qualitative interactional study includes 18 video-recorded patient-genetic specialist consultations, 12 genetic specialist interviews, and researcher field notes collected from genetics clinics at 3 tertiary public hospitals in Cape Town, South Africa. The study utilises a blended interactional analysis approach that draws on principles of conversation analysis and theme-orientated discourse analysis. Thematic analysis and content analysis are utilised for talk-extrinsic data. All data has been triangulated.

Results
Several interactional features are noted to create tension in the communication of risk and management of uncertainty in a local context. The volume and complexity of risk information and formats, together with universal and personal uncertainty are difficult to address in GC consultations. Contextual factors such as limited patient engagement, challenges in gathering patient information, literacy barriers and language mismatches, as well as differing risk perceptions create further interactional uncertainties that must be managed. Findings suggest that engaging with risk and uncertainty topics in GC whilst attempting to achieve GC's goals of establishing and prioritising patient agendas, tailoring information according to a patient's needs, ensuring complete disclosure of risk information and maintaining a neutral stance create complex interactional spaces. Findings in this study suggest that the activities and communication approaches currently used within these complex interactional spaces may be hindering, rather than promoting patient care.

Conclusion
GC risk communication is more complex in reality than what is suggested in theory. The study highlights that some of the goals of GC are not being met in a South African context and existing guidelines create additional interactional uncertainties and tensions. The findings of this study call for the reconsideration of current GC risk communication practice in South Africa. Current practice suggests a heavy teaching focused GC model. More attention is needed on the counselling model of GC which focuses on a patient's concerns and needs. Greater symmetry with the counselling GC model will assist in developing a care relationship within local GC practice that pays attention to tailoring risk information and managing uncertainty in GC consultations. Collectively, this approach may promote contextually attuned South-African based GC guidelines that may assist in achieving positive patient outcomes, and assist local genetic specialists navigate the complexity of practice in a socio-cultural diverse setting. The study adds to the growing literature demonstrating the value of interactional qualitative research within healthcare. The findings are important for South African and international GC training and practice, as well as other healthcare areas where risk is conveyed,

Key Words: Risk, Uncertainty, Care, Genetic Counselling, Communication, Interactional Research, Socio-cultural diversity, South Africa

DECLARATION

I declare that this thesis is my own, unaided work. It is submitted for the degree of Doctor of Philosophy at the University of the Witwatersrand, Johannesburg. It has not been submitted before for any other degree or for examination at any other university.

Signed: 

Megan Scott

19 May 2021

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"Remember to look up at the stars, and not down at your feet. Try to make sense of what you see and wonder about what makes the universe exist. Be curious."

- Stephen Hawking

PEER-REVIEWED PUBLICATIONS & CONFERENCE OUTPUTS BASED ON THIS RESEARCH

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Scott, M., Watermeyer, J., & Wessels, T-M. (2020). Video-recording complex health interactions in a diverse setting: Ethical dilemmas, reflections and recommendations. *Developing World Bioethics, 20(1), 16-26.* DOI: 10.1111/dewb.12226 [ISI accredited].

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ACRONYMS USED IN THIS THESIS

AMA: Advanced maternal age

AUM: Anxiety/Uncertainty Management

TB: Tuberculosis

CA: Conversation analysis

CD: Communicable disease(s)

EBMG: European Board of Medical Genetics

ESHG: European Society for Human Genetics

GC: Genetic counselling

GCOS: Genetic Counseling Outcome Scale

GCSA: Genetic Counsellors South Africa

HAART: Highly active antiretroviral therapy

HBOC: Hereditary breast and ovarian cancer syndrome

HCRU: Health Communication Research Unit

HIV: Human immunodeficiency virus

HPCSA: Health Professions Council of South Africa

HREC: Human Research Ethics Committee

OI: Osteogenesis imperfecta

NCD: Non-communicable disease(s)

NSGC: National Society of Genetic Counselors

PCC: Patient-centred care

REM: Reciprocal Engagement Model

RIAS: Roter Interaction Analysis System

TCS: Treacher Collins syndrome

TODA: Theme orientated discourse analysis

UCT: University of Cape Town

UK: United Kingdom

UMT: Uncertainty Management Theory

USA: United States of America

Wits: University of the Witwatersrand

LIST OF TERMS & CONDITIONS MENTIONED IN THIS THESIS

Advanced maternal age: Women considered to be at an older age for reproduction, which carries a risk for having a baby with a birth defect. Women over the age of 35 years are considered to be of advanced maternal age.

Amniocentesis: An invasive medical procedure where amniotic fluid is removed from around an unborn baby. Amniotic fluid contains fetal cells and the procedure is predominantly used for diagnostic testing for genetic conditions, fetal infections and sex determination.

Chromosomes: Large thread-like structures found in the nucleus of every cell. Chromosomes contain DNA material.

Chromosome translocation: A chromosomal abnormality in which the structure of certain chromosomes in an individual has been altered.

Congenital: Present at birth.

Congenital rubella: An illness caused in a baby due to their mother being infected with rubella during pregnancy. Fetal rubella infections during the early stages of pregnancy can lead to miscarriages, and congenital problems such as deafness, cataracts, and heart defects.

***De Novo*:** A genetic mutation occurring for the first time in the affected individual, rather than it being inherited from a parent.

Down syndrome: A genetic condition caused by a chromosome aberration, namely trisomy 21. Individuals with Down syndrome have physical delays, intellectual disability, and distinct facial features.

Fetal medicine specialist: Also known as a perinatologist. A sub-specialist in the field of obstetrics who specialises in maternal and fetal care, especially pregnancies considered to be high risk or who have complications. Fetal medicine specialists can provide specialised screening and invasive diagnostic procedures.

Gardner's syndrome: A rare genetic condition and sub-type of familial adenomatous polyposis (FAP). It causes a predisposition to develop colorectal cancer at a young age as well as benign and malignant tumours on various parts of the body. It is characterised by the presence of multiple colorectal polyps, osteomas, fibromas, and sebaceous cysts. Individuals are also at risk for developing cancer of the small intestine, stomach, liver, bile duct, thyroid, central nervous system and adrenal glands.

Gene: The basic unit of hereditary. A gene is made up of DNA and codes for a protein that performs particular functions in the body.

Genetic counselling: The process where knowledge about the genetic aspects of illness is shared with individuals who are at risk for developing an inherited condition, or who may pass it on to their offspring.

Genetic counsellor: A trained healthcare practitioner who provides genetic counselling. Genetic counsellors typically have a MSc (Med) degree in genetic counselling.

Genetic mutation/fault: A permanent alteration in the DNA sequence within a gene. The alteration prevents the gene from functioning correctly which has implications on the health of an individual.

Genetic specialist: The term 'genetic specialist' has been used in this study to ensure anonymity in a small, specialised field. It includes genetic counsellors and medical geneticists, as well as individuals who are in their final stages of formal training, namely senior genetic counselling students, genetic counselling interns, and medical genetic registrars.

Hereditary breast & ovarian cancer syndrome (HBOC): An adult-onset cancer predisposition syndrome caused by an inherited genetic fault. It is typically characterized by an increased risk of breast and ovarian cancer, as well as other cancers such as male breast cancer, prostate cancer, pancreatic cancer, and melanoma.

Hypophosphatasia: An inherited disorder that affects the development of bones and teeth due to disruptions in bone mineralisation.

Interpreter: A person who interprets and provides a translation of a language or words.

Lifeworld: The social and environmental aspects and influences of an individual.

Medical geneticist: A sub-speciality within medicine that involves the diagnosis and management of inherited disorders.

Osteogenesis imperfecta (OI): An inherited disorder that affects the development of bones due to interruptions in collagen formation. Individuals with OI are prone to breakages without apparent trauma.

Patient: The word 'patient' in this study includes individuals affected by a genetic condition, or who are at risk for a genetic condition, and/or caregivers (e.g. parents) of an affected or at-risk individual.

Prader Willi syndrome: A genetic condition caused by a deletion in Chromosome 15. Affected individuals have behavioural problems, intellectual disability, developmental delay, short stature and hormone interruptions.

Prenatal: Also known as antenatal. Prenatal care is typically provided during a woman's pregnancy to ensure the wellbeing of the mother and the unborn baby.

Public & private healthcare: South Africa's healthcare system is divided into two parallel sectors, namely private and public healthcare. Individuals who are able to afford private healthcare insurance use private healthcare. Individuals who do not have private insurance, and who cannot afford private healthcare make use of public, or state-funded, healthcare facilities. Most South African individuals make use of public healthcare services.

Registrar: A medical practitioner who is training within a sub-speciality of medicine.

Soft marker(s): A small anatomical variation that is detected during fetal ultrasounds. Soft markers are used as an indicator for risk in an unborn baby, particularly for chromosome abnormalities e.g., Down syndrome. A soft marker does not necessarily mean that an abnormality is present, but that there may be an increased risk. A soft marker is thus a screening tool and further diagnostic genetic testing is required to confirm whether an abnormality is present or not.

Teratogen: An agent or environmental factor that can cause developmental problems in an unborn baby.

Treacher-Collins syndrome (TCS): A genetic condition usually evident from birth, that is characterised by malformations in the bones of the face, giving a distinct appearance. It can range from mild to severe, and although affected individuals are considered intellectually normal, they may have respiratory and visual complications, a cleft palate, as well as hearing loss which may affect learning**.** There are several genetic mutations that can cause TCS, and depending on which gene is involved, can be inherited in an autosomal recessive or a dominant manner. Many cases (up to 60%) involving the autosomal dominant associated gene, are *de novo,* thus if the parents of an affected child are considered normal, then the recurrence risk in another pregnancy is low.

Ultrasound: A screening device that uses sound waves to produce images of an unborn baby in the uterus. It is used as a tool by healthcare practitioners to monitor a pregnancy, particularly the growth and development of an unborn baby.

CHAPTER 1: INTRODUCTION

1.1. Thesis overview

This is a study about risk and uncertainty communication in genetic counselling (GC) consultations and how they impact care within a particular context. Risk and uncertainty are two separate constructs, yet they are closely linked. When an individual considers a risk, they are often faced with uncertainty. Uncertainty is thus something that is communicated when discussing risk, but it is also something that must be managed interactionally. Managing uncertainty facilitates risk-related decision-making and aids in making risk information relevant and useful. Managing uncertainty can be seen as assisting individuals to cope and make sense of risk.

In GC, risk and uncertainty accompany discussions around genetic inheritance, clinical diagnosis and testing. Uncertainties in GC may also stem from communication issues during a GC consultation due to contextual influences such as socio-cultural differences. In turn, interactional uncertainty may lead to additional risks such as misunderstanding between individuals. This study is thus not only about the type of risk information conveyed during GC, but also the communication *process* of GC and how interactional and contextual features may create additional uncertainties.

GC is a hybrid profession that entails risk assessment, conveying risk-related information and assisting in clinical management. Yet GC also emphasises the psychological and emotional impact of genetic conditions on individuals and families, focusing on coping, psychosocial support, and the provision of counselling. The notion of care is thus a suitable lens to study how risk and uncertainty communication process during GC consultations impacts patients since to care, is to maintain and ensure the wellbeing of individuals within different environments.

Part of ensuring care in GC consultations is to understand the process of communication within different healthcare settings. Understanding what contextual factors facilitate or hinder communication between healthcare practitioners and patients is imperative in ensuring that patients benefit from the information that they receive during GC, and way in which they receive this information. Many of these communication processes and contextual influences can only be explored and examined during real-time consultations and cannot be taught or fully

appreciated in classroom teachings or role-play exercises. These may include aspects such as trust, listening for subtle patient cues, the impact of the 'atmosphere' of an interactional space, learning to be flexible and to tailor information to meet different patients' needs, and developing the skills to manage uncertainty and complex psychosocial features that impact communication and decision-making. These authentic moments and lessons from actual healthcare service delivery have been termed the 'hidden curriculum' (Browning et al., 2007; Inui, 2003; Watermeyer et al., 2020a).

By focusing on the communication process and highlighting some of these contextual features that impact everyday GC interactions, practitioners involved in GC services delivery and training may have opportunities to become attuned to the needs of patients attending GC services within a particular setting. By extension, by paying attention to risk and uncertainty communication, the study also places a looking glass on the field of GC, since GC at its fundamental level is about communicating risk and managing uncertainty. The study creates an opportunity to highlight how GC services occur within particular settings, how services impact patients and genetic specialists, and how GC curricula and theoretical guidelines align or create tensions with actual practice.

Being a genetic counsellor myself, I understand that communication in GC is challenging, especially in the South African context. This study is not about describing the 'perfect' GC consultation, as I believe that it does not exist. GC consultations are messy and complex with a mix of communicative moments done well, and other moments where communication could have been done better. The study is thus about deconstructing moments of talk within GC consultations and understanding what influenced these moments and why they occurred in the way that they did. This approach strives to enhance patient care through refining practitioner's communication skills and ensuring that the approaches used to communicate with patients are contextually attuned. Here, research methods such as interactional language-based approaches and video-recordings play a crucial role in providing an opportunity to study the finer details of what happens within real-time GC consultations. Furthermore, although much attention has been paid to how patients experience GC, less is known regarding how practitioners such as genetic counsellors and medical geneticists experience the delivery of risk information and the management of uncertainty. The study provides valuable insight from South African genetic counsellors' and medical geneticists' perspectives about their professional experiences in a public healthcare setting. These insights, together with the video-recorded GC consultation

data, provide a rich understanding of the complexities of communicating risk and managing uncertainty in South African GC consultations.

The study makes use of several existing theories to build a theoretical framework. Theories borrowed from include patient centred care, the voice of medicine and the lifeworld, uncertainty management theory, and the ethic(s) of care. From a methodological point-of-view, the study is qualitative in nature. Beeson (1997) emphasises that GC is a powerful area for qualitative research. Genetic counsellors continuously oscillate between the rapidly advancing and technical world of molecular genetics, whilst equally facing the highly emotional and complicated social worlds of patients and families dealing with the reality, or prospect, of a debilitating and permanent condition. Qualitative research provides opportunities to elucidate what happens when the worlds of medicine and science collide with the psychological and social worlds of people (the lifeworld).

Having an interest in both what happens during consultations at an interactional micro-level and wanting to explore broader macro-level genetic specialists' experiences and contextual influences made choosing an analytical lens particularly challenging. It took several months of deliberation and discussion with my supervisors, other researchers, and experts in the field to finally decide to borrow from different qualitative methods and use a blended socio-linguistic approach. The study's methods thus do not claim to be purist in any way. I believe this decision has helped address the many layers of complexity encountered throughout this research journey.

As beneficial as the decision to use a blended approach has been, it has also come with methodological and epistemological tensions. These tensions have had to be carefully managed and navigated throughout the research process. Some of the tensions comprise the use of multiple sources including video-recorded interactional data from GC consultations, genetic specialist interviews and observational field notes. Using different analytical lenses such as principles of conversation analysis (CA), theme-orientated discourse analysis (TODA), thematic analysis (TA) and principles of content analysis to analyse the different sources, and then attempting to triangulate the findings has been challenging, especially given the volume of data produced. In addition, the project has produced ethical and logistical challenges using video recordings in a clinical setting, as well as studying my own colleagues and professional

field. Discussions and reflections on these experiences are provided in the thesis as they have been important personal and professional lessons.

1.2 Positioning the researcher: Wearing three hats

1.2.1 Hat one: The medical scientist & genetic counsellor

I have always been fascinated by medicine and science. It seemed like a natural choice coming from a family of scientists and medical professionals. I remember spending time as a child and teenager in various government hospital pharmacies in KwaZulu-Natal and the Western Cape, where my mother worked, being captivated by the noise, the people, the conversations and the different diagnoses. Job shadowing in a clinical trial centre and a hospital in the Eastern Cape further cemented my wish to pursue a career in this field. I have also always loved interacting with people. However, I was never quite sure how to blend these two interests. Being a doctor was never my first choice. However, I was unsure what other health career paths to choose. Enrolling in a Bachelor of Science degree at Stellenbosch University in the Western Cape, focusing on a wide range of human health-related topics, seemed like the right choice. Here my love for genetics grew and I soon realised that this was the field I wanted to contribute to.

My postgraduate journey in human genetics began as a quantitative laboratory-based scientist in Cape Town. Adhering to strict protocols, using glass beakers, chemicals, pipettes and statistics to generate large sums of numerical data was an everyday norm in the laboratory. My only experience of 'patients' were blood and amniotic fluid samples, later processed into small, plastic tubes containing extracted, clear-coloured DNA labelled with anonymised reference numbers. Although appreciating the technicality and skills required to undertake this work, I sensed something missing within this controlled environment. I wanted to know more about and connect with the individuals and families behind the samples and to hear their stories. This put me on the path of becoming a genetic counsellor by enrolling in a Master's degree in Johannesburg at the University of the Witwatersrand (Wits) in 2010.

Shifting my career path to becoming a genetic counsellor allowed me to be directly part of the clinical setting and to work with the patients and families. Here I have found purpose and value. During my time as a GC student at Wits and as an intern at various public hospitals and genetic services in Johannesburg, I became increasingly aware of the social, contextual, and psychological factors that impact clinical services. I started to sense that these factors cannot

be separated but are rather an unavoidable part of healthcare provision and that they often create messy and challenging consultations. Although I was taught the principles of 'good' patient-genetic counsellor communication and what to talk about, how to do this in practice was not straightforward. How did I truly know whether I had assisted a patient and provided a helpful service? And if so/not so, where during the interaction and communication process did it go right/wrong? Finding ways to convey complex scientific information in a useful way to another person, whose social context and reality was so different from mine, was not only difficult and frustrating at times, but also humbling. These clinical experiences highlighted for me not only that patient feedback is essential, but that there is a need to better understand how communication and context impact the clinical and scientific processes. The experiences of genetic specialists in the field also seemed imperative to understand what type of support they needed to undertake this complex task.

1.2.2 Hat two: The researcher

The complexities encountered within the local GC clinical setting sparked an interest to look at the process of communication and how it occurs, especially since there is a paucity of such knowledge. My Master's research project explored patients' experiences of South African GC services and found that although GC consultations were generally viewed as a positive experience, the information recall of patients was poor, and many indicated that they had found the information conveyed to them challenging to comprehend and at times overwhelming. Furthermore, contextual factors such as language barriers, poor literacy, and the unfamiliarity with genetic conditions and GC services further impacted their experiences. These findings have led to further interest and exploration of communication practices in South African GC consultations for my PhD.

These GC communication interests have forced me to step out of my comfort zone and delve into the unchartered world of qualitative research. The non-linear, varied approach that qualitative research uses, together with the encouragement to embrace, rather than eliminate the many interweaved layers of health practice and social life that qualitative research highlights have been a challenging, yet enlightening experience for me. Surrounding myself with other qualitative researchers and mentors has also allowed me to appreciate the enormous value that qualitative research brings to healthcare.

1.2.3 Hat three: The patient & mother

The third hat I write about is deeply personal, and it has taken me some time to decide whether to include it in my thesis or not. However, I have decided to include it, as I believe that this hat has most strongly shaped this body of research and how I view genetic risk and uncertainty as both a genetic counsellor and a researcher.

During my PhD project, my husband and I found out that we were expecting a baby. During my routine 20-week prenatal fetal ultrasound scan I was told that soft markers, structural changes that confer congenital risk, (in this case for Down syndrome), had been identified during the scan. Suddenly, going from what I had assumed and what had been reported as, a healthy and happy pregnancy, my husband and I were faced with several genetic risk considerations, including the offer of invasive testing (an amniocentesis). We both had to make a decision under uncertainty in which two different risks had to be considered: 1) accepting the presence of the soft markers and waiting until our baby was born to confirm or eliminate the presence of a congenital disorder, or 2) deciding to have an amniocentesis and a diagnostic genetic test to confirm or eliminate the presence of Down syndrome, but then be willing to accept the small, but significant risk of having a miscarriage from the amniocentesis procedure.

In the end, my husband and I decided to undergo an amniocentesis. We did not want the uncertainty of waiting for the remainder of the pregnancy to know the outcome. The results of the genetic test came back negative and provided clarity that our child did not have Down syndrome.

Reflecting on what happened, never did I consider myself to be at risk for a genetic or congenital abnormality before this experience. Thus, I did not feel worried or concerned when going for my routine 20-week prenatal ultrasound scan. Hearing the fetal medicine's specialist say to me: "I am so sorry to tell you this Megan, but can you see those echogenic foci in baby's heart? They are soft markers for Down syndrome" was the last thing I expected. The joy of having a baby quickly turned into shock and fear. One could have heard a pin drop in the consultation room as the remainder of the consultation hung in vulnerable silence whilst the specialist carefully examined our baby for other clues, trying to decipher what these soft markers could mean.

During the consultation, the specialist discussed risk predictions and outcomes, and reminded me, what I already knew as a genetic counsellor, that soft markers are screening indicators for risk, they are not diagnostic. Further objective risk calculations, such as there being no other soft markers or features noted during the remainder of the scan (or in any of the previous ultrasound scans), my younger age, insignificant family history and that I had already undergone non-invasive prenatal testing (NIPT) which was reported as low risk, should have been motivators for me to view my risk as low. However, in those decision-making moments, my 'hat' as an emotional being and as an expectant mother, took over from my 'hat' of a professional genetic counsellor.

My reaction to being the patient and having my *own* body and baby at risk was completely different to what I expected, from the point of view of being a genetic counsellor. Rather than weighing up the risks in an objective manner, my decisions and thoughts were driven by my emotions. The uncertainty of the situation also made me feel uncomfortable, despite my training to counsel other pregnant women and couples in similar uncertain circumstances.

This personal experience and being placed in the 'patient's' shoes have given me empathy to the incredibly difficult decisions patients have to make during GC consultations, and the implications attached to the offer of invasive genetic testing. When the risks and uncertainties discussed during a consultation have direct consequences for you and your family's wellbeing, making objective, unemotive decisions seems impossible. Also, experiencing the healthcare practitioner, a colleague whom I have worked with for several years, attempt to facilitate decision-making with unclear information for my husband and I, has given me further appreciation for the complexity of risk communication and decision-making under uncertainty.

1.3 Introduction to the structure of the thesis

This chapter, *Chapter One*, provides an introduction and overview of the study, the theoretical and methodological framework and my position.

Chapters Two, Three, Four and Five provide an investigation of the background literature in which the study is embedded. I have used a broad range of literature sources including international and local academic papers, conference proceedings as well as academic presentations attended. The HCRU and Wits' library resources were utilised, as well as work

suggested by academic and GC colleagues. Sources include hard-copy publications as well as electronic publications from platforms such as the Wits e-library, PubMed and Google Scholar. I have tried to ensure that sources are published within accredited journals.

Chapter Two introduces the constructs of risk and uncertainty, how they are different, yet related. The role and impact of risk and uncertainty within healthcare, particularly within communication is further extrapolated. *Chapter Three* describes the field of genetic counselling, including its history, the models, frameworks, and theoretical outcomes, and how risk and uncertainty play a role in GC. The chapter also looks at the impact of risk and uncertainty communication on patient care within GC. *Chapter Four* discusses the impact of context on GC service provision and provides an overview of the South African setting, including local factors thought to impact risk communication and the management of uncertainty in GC consultations. *Chapter Five* provides an overview of interactional research.

Chapter Six describes the theoretical framework of the study, namely a framework of care. The chapter provides details on what the term 'care' means in this study, the existing theories that the framework borrows from, and how they collectively contribute to a framework for GC consultations.

Chapter Seven establishes the niche focus of the study. Existing literature is used to argue the need for evidence-based GC practices using interactional methods. I further argue how this study provides a unique opportunity to address the paucity of knowledge on the process of risk and uncertainty communication within South African GC consultations.

Chapter Eight details the research question, the aim and objectives. It further describes the methods used, including the research design, the research setting, participant details, inclusion and exclusion criteria, data sources, the analytical approach, data saturation, trustworthiness and ethical considerations.

My results and discussion are divided into three chapters, namely *Chapters Nine, Ten* and *Eleven. Chapter Nine* serves as the introduction to the results and provides an overview of the sources of risk, the formats used to convey risk information. *Chapter Ten* presents evidence for different layers of uncertainty identified in local GC consultations. In this chapter, reported and interactional evidence of universal and personal uncertainties are presented. The chapter

also demonstrates how contextual factors and patient characteristics perpetuate the presence of interactional uncertainty during risk discussions. The final results chapter, *Chapter Eleven*, demonstrates misalignment in the theoretical ideals of GC versus the realities of practice, and how particular GC guidelines may hinder patient care in a South African context. The results are presented together with discussions and supporting literature.

Chapter Twelve discusses the study from a general point-of-view, what the findings mean and the implications on South African GC practice and training. The study's findings are also considered within a framework of care and how they align with local patient outcomes. The chapter further deliberates on the overall approach used in the study, particularly the ethical tensions encountered due to the use of video-recordings within different public-based genetic clinics during routine practice.

The final chapter, *Chapter Thirteen*, provides details on the implications and recommendations for South African GC training and practice based on the study's findings, and how the study contributes to both national and international literature on GC communication and interactional health research. Arguments on the strengths of the study and considerations on the study's limitations are also addressed in this chapter, together with concluding reflections.

Regarding the writing style of the thesis, due to the small South African GC field and professionals offering local GC services, I have had to be cautious of maintaining participant confidentiality. Even if unintended, the write-up of my results in this thesis had the potential for practitioner participants and research sites to be identified. I have thus chosen to remove all identifying features such as names of research sites, as well as ages, gender, ethnicity, qualifications and years of professional experience from the study write-up. Choosing rather to use a broad label of 'genetic specialist' to describe genetic counsellor and medical geneticist participants. I also use the term genetic specialist in the literature review chapters of this thesis for literature that have described genetic counsellors, medical geneticists, genetic nurses, or healthcare practitioners who practice GC. I am aware that the nature of qualitative research is to provide detailed descriptions of the participants and settings, however, I am sensitive to the participants' rights to remain anonymous. I believe that I have achieved a balance in this thesis write-up that respects participant confidentiality without losing the richness of the data presented.

1.4. Conclusion

This chapter provides an overview of what the study is about, background detail for the motivation for the topic, and describes my position as a researcher, a genetic counsellor and a patient/mother. The chapter further introduces the theoretical and methodological frameworks and the individual chapters. In this thesis, I will attempt to address the paucity of knowledge on risk and uncertainty communication within South African GC consultations using a blended qualitative approach. I further reflect on the notion of care in local GC consultations and how communication may affect this. In addition, I address how theoretical ideals and guidelines for GC do not necessarily align with actual service provision, and the implications for patient outcomes, as well as training and practice for local genetic specialists.

CHAPTER 2: RISK & UNCERTAINTY

2.1 Introduction

This chapter serves as an introduction to risk and uncertainty. The chapter provides literature on the concepts of risk and uncertainty, how they manifest in everyday situations, and how they are assessed and managed. The literature on risk communication and uncertainty in healthcare is also discussed.

2.2 The concept of risk

Risk has been described in several ways, with a common description being a situation which involves uncertainty and the possibility of a dangerous or negative outcome (Kaplan & Garrick, 1981; Oxford Dictionaries, 2015; Vlek, 1987). Risk is thus a phenomenon in which security can be compromised due to a potentially threatening action or situation, there are two or more possible outcomes to a risky situation, however if, when, or what outcome will occur is unknown (Beck, 2000; Covello & Merkhofer, 1993).

Described as "ubiquitous, pervasive, diverse and global" (Kemshall, 2003, p.3), risk is a popular topic and has been studied within many disciplines each with their own theories and analytical perspectives. Disciplines include, but are not limited to, economics, business management, safety, insurance, decision-making, politics, environmental studies, social science, psychology, ethics, and health (Brown, 2014; Dickert et al., 2015; Luhmann, 1993). Not only is risk diverse in discipline, but there are also many types of risks. There are those risks we are alerted to and can mitigate (e.g., poor lifestyle habits), and those that we have no awareness of (e.g., an asteroid hitting earth). There are those risks we can independently control (e.g., the choice to go skydiving), and those that we cannot (e.g., economic down-grades). Some risks happen instantaneously (e.g., a car accident), some over-time (e.g., global warming), some risks have a low possibility/high impact (e.g., an earthquake), and some risks have a high possibility/low impact (e.g., the chance of losing when gambling with small change). Collectively, all risks involve the potential for loss or a negative outcome, with no, or limited information of the timing or extent of the outcome (Kahneman, 2011).

2.2.1 Assessing & managing risk

Due to the awareness of risks and their associated harms, efforts are made to obtain greater clarity on the type, magnitude, and outcomes of the risk, to avoid unwanted consequences (Bradbury, 1989; Covello & Merkhofer, 1993). *Risk assessments* are a way to provide supportive evidence for decision-making under uncertainty, as well as a precursor strategy to control risk, also known as *risk management*.

Risk assessments involve the practice of recognising, evaluating, and reviewing risks related to dangerous substances, actions, or events to quantify when something is likely to occur (Gigerenzer, 2003; Kaplan & Garrick, 1981; Tannert et al., 2007). In this way, an educated guess can be made with 'imperfect information'. Risk assessments are often complex since different interpretations of probability must be taken into consideration, including subjective probability (gutfeel or personal perception), propensity probability (how certain actions or scenarios present the same outcomes) and frequency probability (the number of times the risk occurs within a particular context) (Gigerenzer, 2003; Hacking, 1975; Sarangi et al., 2003). This level of complexity can make risk assessments extremely challenging, as no singular preferred method, process or outcome exists.

Risk management, in turn, involves knowing that forthcoming events may lead to undesirable outcomes and subsequently entails identifying, choosing and employing suitable actions to try to control the established risk (Covello & Merkhofer, 1993; McNeil et al., 2015). Through risk management efforts are made to not only minimise the possibility of an ill-fated event from happening, but also to be prepared should something negative occur (Hubbard, 2009), the latter considered a "resilience to future events" (McNeil et al., 2015, p. 9). Although risk assessments and management remain an integral part of various fields, the ability to make confident decisions that guarantee certain outcomes remains elusive (Burton-Jeangros et al., 2013; Gross, 2007; Tannert et al., 2007) and may always be.

2.2.2 The perception of risk

Risk can be understood from either an objective or subjective lens. Objectively, risk is something that can be measured and calculated to predict the probability of an event happening (Anscombe & Aumann, 1963; Holton, 2004). Empirical evidence is thus used to identify, predict and control certain outcomes (Otway & Thomas, 1982). Alternatively, a subjective view of risk is a value-driven notion based on an individual's understanding and experience on

whether something is likely to occur or not (Bradbury, 1989; Holton, 2004). Risk may thus be emotionally and motivationally constructed (Arrow, 1951; Tversky & Kahneman, 1974).

Separating subjective and objectives views of risk during decision-making is difficult, with research suggesting that individuals respond to risky situations based on both scientific evidence and personal discernment (Arrow, 1951; Bradbury, 1989; Lippman-Hand & Fraser, 1979; Lloyd et al., 2001; Paling, 2003; Ropeik & Gray, 2002; Vlek, 1987). A study on patients' informed decision-making for surgery found that although patients are provided with evidence-based risk information, patient's personal psychological perspectives may differ, and they may over-estimate or under-estimate the same given risk (Lloyd et al., 2001). Some individuals also view risk differently depending on if the risk is personally relevant or not. A study on young adults' attitudes and lifestyle behaviours shows that young adults display optimistic biases against their personal risk of developing cancer and cardiovascular disease when comparing the same risks for other individuals in the general population (Masiero et al., 2018). Research also suggests that individuals' risk perceptions can change over time or be altered depending on how information is presented to them (Lloyd et al., 2001; Paling, 2003; Smerecnik et al., 2009). Risk is thus not something "'out there' independent of our minds and culture" (Kahneman, 2011 p. 141). A balance is needed between subjective emotions and objective reasoning when making risk-related decisions (Kemshall, 2003; Kahneman, 2011).

2.3 The concept of uncertainty

Uncertainty is a closely linked concept to risk. Uncertainty stems from information being lacking, ambiguous, or overly complex (Brashers, 2001; Gollust et al., 2012). Although risk outcomes are typically negative, some uncertain outcomes are neutral or linked to reward or hope (Brashers et al., 2000; Cairns, 1999). Uncertainty has also been described as a natural occurrence of life and complex systems (Merry, 1995; Mishel, 1990). Despite the acquisition of knowledge, uncertainty may never be eliminated from some situations as they are layered with multiple uncertainties that are difficult to address. Uncertainty in these instances is thus tolerated, rather than eliminated, as an attempt to cope with unknown knowledge (Brashers & Hogan, 2013).

There are two types of uncertainties: 1) personal inadequacies of knowledge i.e., one's own ignorance, and 2) limitations in current scientific knowledge i.e., information that is universally

not yet known or understood. Early philosophers such as Socrates and Plato have alluded to the challenge of these two personal and universal uncertainties and our inability to know all (Fox, 1957). Despite our advancements in technology and research many years later, we are still unable to make confident decisions because of existing daily uncertainties and limited information (Burton-Jeangros et al., 2013; Gigerenzer, 2003). It appears that the more we learn, the more uncertain we become about what we know (Tannert et al., 2007).

2.3.1 Uncertainty & anxiety

As individuals and as a society, it is our natural tendency to seek structure, as it creates harmony, stability, and the ability to anticipate and control processes and events (Kahnemann, 2011; Kurth, 2016). When faced with uncertainty, not only does disorder and randomness occur, but it also brings about several emotional states such as anxiety and fear. Anxiety is particularly common in uncertain situations and is an important determinant of how individuals may respond to re-establish control or stability (Kurth, 2016). Although anxiety often goes together with fear, it is important to distinguish between the two terms. Fear is a response to a clear and apparent risk, while anxiety is a response to a perceived risk where the impact and extent are unpredictable or uncertain (Cheney & Sayfarth, 2007; Kurth, 2016). Anxiety may cause individuals to have a heightened sense of pessimistic risk appraisals, thus they may over-estimate risks within particular contexts, and practice risk-avoiding behaviour. Anxiety is thus a large emotive driver behind how individuals make risk-related decisions during uncertain situations (Maner & Schmidt, 2006).

2.4 Risk communication & uncertainty in healthcare

2.4.1 Risk communication in healthcare

Risk is a fundamental topic in healthcare decision-making. Healthcare risk not only involves the identification, treatment, and management of disease by patients and doctors; but also plays a role in service functioning, staff safety, and monetary decision-making for policymakers, insurance companies, as well as medical organisations (Bodemer & Gaissmaier, 2012; Parker, 2009). Risk from an individual health perspective can be characterised as a disease precursor (including demographic factors, individual behaviour, physiological changes, as well as family and/or environmental history) that is linked to increased morbidity or mortality (Mosby's Medical Dictionary, 2009a). Risk has been studied in multiple health-related fields including cancer screening, clinical trials, vaccinations, therapeutic drugs, chronic disease management,

community health messaging, as well as medical procedures to name a few (Chapman & Coups, 2006; De Wit et al., 2008; Peters et al., 2007; Schapira et al., 2001). Prenatal healthcare has also become increasingly medicalised with pregnant women facing numerous health risk messages about screening and testing options to ensure a healthy baby. Reasons for the increase in risk information in obstetrics and gynaecology are linked to defensive medicine practices which aim to discuss all risks and testing options with a patient due to an increasingly litigious society (Hashiloni-Dolev et al., 2019).

Healthcare practitioners may analytically consider risk, whilst patients may calculate risk based on personal experience and associated meanings (Hunt et al., 2006). Literature suggests that divergences in preference and thinking can create difficulties when making decisions about treatment and management of disease (Ahmed et al., 2012). Furthermore, one of the most vital components of risk is not only how it is identified and managed, but how it is communicated. Risk communication is a two-way interaction amongst individuals, groups, or institutions in which an event or procedure is discussed to 1) exchange information about the associated harms and benefits; 2) to improve comprehension of the risk; and 3) to facilitate informed decision-making on how to manage it (Ahl et al., 1993; Ahmed et al., 2012; Cho, et al., 2015).

Risk is also important in health communication, which involves conveying usable and understandable information concerning health (Calderón & Beltrán, 2004). According to Cho et al. (2015) risk communication may contain several features including images, numbers, stories, formal agreement documentation and rhetoric, all used to influence perception, highlight information and bring about decision-making and action against a given risk. These points stress the importance of proficient communication since this process can influence not only the way individuals view risk, but subsequently how they act upon it during risk-related decision-making. The risk 'communicator' thus plays a critical role in which much power is given over influencing the perception of the message 'receiver,' and a careful balance of information and how it is portrayed needs to be taken into consideration.

Due to the difficulty of explaining risk, much research attention has been paid to this activity (Bogardus et al., 1999; Smets et al., 2007). There are many ways in which a risk can be expressed during communication. Formats include numerical formats; relative, comparative and absolute risks; personalised and generalised risk; as well as narrative explanations (Ahmed et al., 2012; Dowse & Ehlers, 2001; Edwards et al., 2001; Fagerlin et al., 2011; Gaissmaier et

al., 2012). In addition, framing, priming, and anchoring are also used (Edwards et al., 2001; Tversky & Kahneman, 1974). Each format is described in further detail below.

2.4.1.1 Numerical Data

Numerical evidence is made up of abstract, factual figures and numbers, which are based on frequencies and probabilities (De Wit et al., 2008; Schapira et al., 2001). A *frequency* can be presented as a ratio of cases of which an occurrence has happened, over the number of cases at risk for such an occurrence (e.g., the incidence of breast cancer in African American women is 126 of 100,000) (National Cancer Institute, 2011; Schapira et al., 2001). *Probability* is indicated as a percentage (Schapira et al., 2001). Percentages represent a fraction of 100 (e.g., 126 of 100,000 individuals can be shown as 0.126%), (National Cancer Institute, 2011). Fagerlin et al. (2011) has suggested that to comprehend this format of risk presentation, greater cognitive demands are necessary, thus numeracy and analytical skills are vital. Although difficult to understand, research has indicated that patients find frequency representations of risk easier to interpret than probabilities. Furthermore, it is thought that the addition of graphical displays to explain these numerical figures increases understanding, especially amongst lower educated groups or in individuals who struggle to grasp quantitative risk (Ahmed et al., 2012; Schapira et al., 2001).

Although frequencies and probabilities are the most common format to present numerical risk information, calls have been made to use statistical methods in combination with other formats that add context such as narrative explanations, metaphors, analogies, as well as visual and graphical displays. In this way, statistical information can become more tangible, without losing the supposed accuracy that numerical evidence provides (O'Doherty & Suthers, 2007; Smerecnik et al., 2009; Trevena et al., 2013).

2.4.1.2 Personalised versus generalised risk

Generalised and personalised risk is another common topic in healthcare settings. The general population risk is a group of people who are vulnerable to a specific event because they all share a particular trait, whereas personalised risk is calculated more specifically (Mosby's Medical Dictionary, 2009b). For example, in invasive breast cancer risk estimation, a woman's personalised risk considers specific information such as her age, ethnicity, as well as her family and medical history (Ahmed et al., 2012). It is thought that personalised risk estimations are more beneficial to patients since they are more accurate and improve understanding and

decision-making, particularly when patients are faced with different health screening tests (Ahmed et al., 2012; Edwards et al., 2013; Hunt et al., 2006).

2.4.1.3 Narrative explanations

Narrative communication of risk is thought to be one of the easier formats to comprehend, particularly for lay individuals, as narrative explanations can be used to replace complex numerical figures (O'Doherty & Suthers, 2007; Weinstein, 1999). According to Winterbottom et al. (2008) narrative communication involves a story (fictional or real) which represents an example of an event which includes characters, details and an outcome. This way of presenting risk often produces emotionally driven, tangible evidence (De Wit et al., 2008). In medical cases narrative may involve a story about patients, possible treatment and management options, and the subsequent consequences of each choice (De Wit et al., 2008; Winterbottom et al., 2008). Research has shown that narrative explanation together with non-quantitative phrases such as "low", "medium", "high", "more often" or "sometimes" are more commonly used in everyday language, rather than quantitative numerical figures (O'Doherty & Suthers, 2007). Caution is needed when applying this method however, due to bias and potential misrepresentation of the true statistical values of a given risk (Trevena et al., 2013; Weinstein, 1999; Winterbottom et al., 2008).

2.4.1.4 Metaphors

Figures of speech such as metaphors and similes are used to assist in explaining concepts, particularly ones that are difficult to imagine or understand. A quote from Kendall-Taylor et al. (2013) describes metaphor as "a device for thinking and talking with, and a good metaphor leads people to think and talk productively about something that they were not previously proficient in thinking or talking about" (p. 415). Although similes are differentiated from metaphor by the inclusion of the word 'like' (Skelton et al., 2002), for the purposes of this study, the term 'metaphor' includes both similes and metaphors.

Although helpful in some instances, using metaphors can also create communicative trouble. Studies have shown how the metaphor of 'body soldiers' to describe CD4 counts in HIV counselling can result in patient confusion (Penn & Watermeyer; 2018) and that the 'war' metaphor in cancer can make patients believe that they have failed in their commitment to eliminating the disease (Reisfield & Wilson, 2004). Certain metaphors describing the body as a 'broken machine' may also make healthcare practitioners forget that there are emotive and

real individuals behind illnesses (Loftus, 2011). Caution against the assumption that all metaphors are useful is advised. It is also important to note that metaphors are culturally governed and that when used out of context, or used across languages, they may be misunderstood, or unhelpful (Heredia & Cieślicka, 2015; Kimmel, 2004; Maalej, 2004; Sontag, 1978).

2.4.1.5 Visual aids

Visuals aids and pictograms have been used in several areas of healthcare including pharmacy discussions (Dowse & Ehlers, 2001; Watermeyer, 2008); cancer consultations (Brown et al., 2017; Zikmund-Fisher et al., 2008), paediatric care, internal medicine, cardiovascular consultations, ophthalmology and gastroenterology (Dowse, 2020). Visual aids are thought to improve comprehension when discussing numerical information, increase recall and re-enforce verbal messages (Dowse, 2020; Garcia-Retamero & Hoffrage, 2013; Houts et al., 2006; Tait et al., 2010).

In contexts of linguistic and literacy barriers, diagrams and props may facilitate understanding of information (Dowse & Ehlers, 2001; Watermeyer, 2008; Watermeyer & Penn, 2009a). However, caution is also necessary when using visual aids since many are culturally and contextually mediated (Dowse & Ehlers, 2001). The potential for diagrams to cause negative reactions, or patient confusion has been demonstrated in South African GC (Wessels, 2014), pharmacy (Dowse & Ehlers, 2001), and audiology (Penn & Watermeyer, 2018) research.

2.4.1.6 Framing, priming & anchoring

Risk information can also be manipulated in certain ways using framing, priming and anchoring. Framing can be defined as describing logically equal information in different ways, such as positive and negative framing, as well as gain and loss framing (also known as goal framing) (Ahmed et al., 2012; Edwards et al., 2001). Ahmed et al. (2012) have reported that positive and negative framing involves describing a single feature of an item or state, in either a positive or negative manner. For example, a patient can be told that they either have an 82% five-year survival rate after a breast cancer diagnosis (positive framing); or that they have an 18% chance of dying within five years after a breast cancer diagnosis (negative framing). Both approaches describe the same information, but each approach has a different effect on the person receiving the information. Similarly, Edwards et al. (2001) has highlighted that gain and loss framing involves describing the consequence of a particular action. For example, the

benefit of undertaking breast cancer screening (gain framing), or the detriment of not undergoing breast cancer screening (loss framing). Studies have indicated that individuals are more likely to choose a risk-avoiding option when the information is negatively presented to them, whilst riskier behaviour is adopted when positive outcomes are emphasised (Tversky & Kahneman, 1974; Edwards et al., 2001).

Priming is another method that can be utilised in risk presentation. Although it is assumed that people can cognitively process information in a balanced manner, researchers have argued that risk decisions are often affected by subconscious influences, and that decision-making is not necessarily rational (Erb et al., 2002; Loewenstein et al., 2001). Priming, to do with human memory, can be seen as prior exposure to an object, word, concept, or task which subsequently influences unconscious information processing when responding to a seemingly unrelated action or task thereafter (Kahneman, 2011). To elaborate further, a theoretical framework called the stereotype priming model (Pechmann, 2001) has been used in health and risk communication to influence patient behaviour. This approach emphasises either the positive or negative health outcomes of a particular behaviour based on stimulating peoples' memories of stereotypic assumptions of this type of behaviour. For example, Pechmann (2001) infers that if an individual is unconsciously reminded of stereotypical images of reckless, irrational people, they are more likely to respond to health messages that negate alcohol consumption and driving. In this way healthcare practitioners can manipulate how people perceive the outcomes of risky behaviour, thus influencing how they respond to it.

Anchoring is often used in decision-making in which unknown information is 'anchored' or associated with previously known information, to make the unknown more perceptible (Michie & Marteau, 1996). This description supports Kahneman's (2011) explanation in that anchoring occurs when individuals "consider a particular value for an unknown quantity before estimating that quantity" (p. 119). Anchoring is common in risk and health communication. It can occur during explanations of risk when patients 'anchor' themselves to a subjective risk estimate when being presented with objective risk-estimates by a healthcare practitioner. The final perceived estimate is subsequently often under- or over-estimated as the patient never quite grasps the objective risk, because of their previous subjective risk perception (Senay & Kaphingst, 2009). Anchoring is also used when healthcare practitioners, based on their prior medical knowledge, are unable to adjust their level of communication with patients since they assume the patient shares the same amount of knowledge as them, thus over-estimating the

patient's level of understanding (Chapple et al., 1997; Senay & Kaphingst, 2009; Wasserman et al., 2010).

2.4.2 The impact of uncertainty on health-related risk communication

Modern healthcare services are increasingly becoming more uncertain due to the rapid introduction of new knowledge and technologies into healthcare practice. Although these advancements have assisted in learning, they have also demonstrated how little we know, or may ever know about the intricacies of science, medicine and the human body (Getz & Kirkengen, 2003; Hashiloni-Dolev et al., 2019; Lou et al., 2020a; Thomassen Hammerstad et al., 2020).

Uncertainty is experienced by both patients and health practitioners. Patients and their families are thought to experience layers of uncertainty when learning about a new diagnosis or health risk. They may have personal ideas on disease causation, who is at fault, as well as preferred treatment and testing options. They may also wonder about the practitioner and medical institution's competency, and whether they will be able to personally cope psychologically, financially and physically (Brashers, 2001; Stivers & Timmermans, 2016). Peters et al. (2007) explain that patients may feel uncertain how to weigh up the benefit or risk of a procedure; the legitimacy of the evidence presented in an interaction; or the severity/greatness of the risk/benefit involved. Health practitioners in turn may struggle to communicate risk within medical fields where there is limited knowledge.

Uncertainty is particularly present in prenatal care and service provision, especially regarding communication and decisions related to testing and screening for abnormalities in an unborn baby. The offer of tests that can detect fetal abnormalities has the potential to turn a woman or couple's pregnancy into an anxiety-inducing experience, rather than an enjoyable one (Heyman et al., 2006). Some of these tests require an invasive procedure, e.g., an amniocentesis, which has the potential to cause a miscarriage due to the procedure. Deciding to pursue an invasive test may create procedural concerns, whereas alternatively deciding not to pursue a confirmatory test when a screening test has indicated that an unborn baby *may* have a problem, may create further uncertainty whether a baby has a problem or not. Some advanced genetic tests also have the potential to create uncertainty in prenatal settings, especially those that are implemented into medical practice in which the interpretation of results is still unclear (Getz & Kirkengen, 2003).

The potential for introducing and exacerbating uncertainty for pregnant women has created an ethical dilemma and how risks should be identified, communicated, and acted upon. Social researchers such as Katz Rothman (1988; 1993) and Rapp (2000) have written about the expectations placed on pregnant women by medical professionals to consider the tests offered to them, and the dilemma of pregnant women having to make decisions. If choosing not to undergo screening and testing of their unborn baby, a pregnant woman may be perceived to be a 'bad mother' by healthcare practitioners or broader society, especially if their child is born with a permanent disability that could have been detected before being born. However, if a woman chooses to undergo screening and testing, they may be placed into further decision-making dilemmas, such as considerations on termination of pregnancy or being at risk for a procedural-related miscarriage when undergoing invasive diagnostic testing. Getz and Kirgengen (2003) further discuss reporting and disclosure of soft markers during routine ultrasound screening. Soft markers are minor anatomical variations that may indicate an increased risk of chromosome abnormalities in a baby, especially Down syndrome. Soft markers are considered screening features, rather than diagnostic, since soft marker findings may be transient, and their risk needs to be confirmed through further invasive diagnostic testing. Getz and Kirkengen (2003) argue that ultrasound examiners need to consider a pregnant patient or couple's context when deciding to disclose all soft marker findings since reporting may lead to over-estimations of risk, exacerbation of uncertainty and may have several negative emotional consequences for a pregnant patient or couple. However, knowing where to draw the line regarding non-disclosure and full disclosure remains a contentious ethical topic.

Previous research on mothers and parents who underwent termination for a fetal abnormality has indicated that mothers and parents may struggle to adjust to their risk-related decisions due to conflicting moral beliefs, perceived judgement from others, and feelings of guilt, shame, and grief (France et al., 2013; Korenromp et al., 2007). Uncertainties for pregnant women facing prenatal testing decisions include diagnostic uncertainty (Richardson & Ormond, 2018), moral uncertainty (not knowing whether a decision is morally acceptable) (Anderson, 2007), and uncertainty on whether they have made the right decision due to a risk being involved (LeBlanc et al., 2009) e.g., choosing to undergo an amniocentesis where there is a risk for procedural-related miscarriage or choosing not to undergo amniocentesis and then not knowing if their baby will be born with a disability or not. A South African study by Scott (2018) suggests that

decision-making for pregnant women faced with a genetic risk is complex, with several medical and social considerations needed.

2.4.3 Managing uncertainty in health communication

Early theorists on communication describe the goal of risk communication to be the reduction of uncertainty (Shannon, 1948), or to influence the perception and balance between certainty and uncertainty (Blumer, 1969). However, modern research suggests that uncertainty cannot be eliminated. Thus, rather than reducing uncertainty, research has focused on better understanding its acceptance and presence in risk communication, and its impact on health service delivery (Pilnick & Zayts, 2014; Sarangi & Clarke, 2002a; Stivers & Timmermans, 2016; Thomassen Hammerstad et al., 2020).

As uncertainty cannot be eliminated, managing uncertainty in interactions has become of interest within the fields of communication (Hogan & Brashers, 2015; Hong, 2020; Knobloch & Solomon, 2005; Kuang & Wilson, 2017; Pilnick & Zayts, 2014; Stivers & Timmermans, 2016; Watermeyer et al., 2020a). Individuals participating in health interactions may often experience uncertainty due to each other's communication skills and emotional states (Berger, 1995; Brashers, 2001). Misalignments in personal values and goals may create further uncertainty for individuals (Brashers et al., 2003). A practitioner's inability to foresee a sure outcome, or provide a diagnosis, places tension not only on decision-making within a consultation but on patient satisfaction since patients want to be given certain information (Ahmed et al., 2012; Arribas-Ayllon & Sarangi, 2014; Politi et al., 2011; Stivers & Timmermans, 2016). An Israeli study on patients' experiences with doctor encounters found that part of patients' motivation for attending medical consultations is to receive assurance and support for their medical concern. Thus, they may feel disappointed when their medical concern is not adequately addressed by a health practitioner (Arieli & Tamir, 2018). Uncertainty is therefore not only topical, but also a *relational* concern (Han et al., 2011; Hogan & Brashers, 2015; Pilnick & Zayts, 2014; Stivers & Timmermans, 2016).

2.5 Conclusion

In conclusion, risk and uncertainty are two closely linked constructs that impact nearly all areas of life. Together they create dilemmas regarding the potential for harm and how that harm may be mitigated and controlled. In moments where risk outcomes cannot be predicted, uncertainty can arise on a personal level, a relational level and a decision-making level. Risk and the

presence of uncertainty also creates several challenges in the practice of medicine and health communication. Focusing on the interactional process between healthcare practitioners and patients is important to understand how risk and uncertainty shape the provision of healthcare, especially in modern healthcare services where risk and uncertainty are pervasive and foregrounded.

CHAPTER 3: GENETIC COUNSELLING

3.1 Introduction

The chapter discusses a specialised area within healthcare, namely GC, and how risk and uncertainty plays a prominent role within this field. The chapter provides a description of GC, the field's history, the models, frameworks, and goals that inform GC practice, as well as the factors which influence the communication of risk and uncertainty in GC consultations.

3.2 Genetic counselling: A definition

Genetic counselling (GC) is a type of medical service that is aimed at individuals and families. The most popular definition currently used within the GC community as proposed by the National Society of Genetic Counselors (NSGC) is: "genetic counselling is the process of helping people understand and adapt to the medical, psychological, and familial implications of the genetic contributions to disease" (Resta et al., 2006, p. 79).

GC is uniquely different from other medical fields in that it does not offer treatment or advice, but rather information and support (Clarke et al., 1996; Skirton et al., 1998). GC forms part of medical genetic services and is typically offered by genetic counsellors but can also be offered by medical geneticists, genetic nurses or other healthcare practitioners (Abacan et al., 2019; Greenberg et al., 2012; Ormond et al., 2018).

Referrals to medical genetic services are usually made by other practitioners for three reasons. These include: 1) the provision of a diagnostic label so that further prognostic information can be provided for an individual and their family, 2) to obtain information on the likelihood of a condition developing in an unaffected, but at-risk individual and 3) the wish for further information regarding pregnancy-planning and whether a future child may be affected with a congenital condition (Sarangi & Clarke, 2002a).

GC consultations involve complex conversations about disorders that are, or may be, genetic in origin and aim to assist and inform individuals and families about their own and reproductive health. During a typical genetic counselling (GC) consultation, a genetic counsellor will: 1) assess and interpret an individual's personal and family medical history to establish the risk of disease occurrence or recurrence; 2) provide education about genetic inheritance, testing,

management and preventative options; 3) convey information about current resources and research; 4) counsel the individual to promote informed decision-making; and 5) provide emotional and psychosocial support to aid in the individual's adaptation to their risk or condition (Resta et al., 2006). Although present in nearly all healthcare areas, GC is most frequently aligned with preconception and prenatal care, oncology, cardiology, paediatrics and neurology. Apart from clinical services, genetic counsellors also hold research, laboratory, policy, education, and advocacy-based positions (Abacan et al., 2019; Resta, 2019).

3.3 Genetic counselling history

GC services are available in several countries; it is, however, considered a relatively young field when compared to other areas of healthcare. The introduction of genetic clinics and risk assessments commenced in the United States of America (USA) in the 1940s, with Sheldon Reed first mentioning the idea of genetic counselling (Rantanen et al., 2008; Resta, 2019). Medical genetic practices were mainly conducted by medical geneticists and university staff, with the predominant focus being reproductive outcomes for couples and families and reducing disability. Formal GC training commenced in 1969 at Sarah Lawrence College in New York, with the profession expanding globally in the late 1980s and early 1990s with the establishment of training programmes in other parts of the USA, Canada, Australia, United Kingdom (UK) and South Africa (Kromberg et al., 2013a; Leeming, 2013; Ormond et al., 2018; Skirton et al., 1998). During this time, the profession also developed a stronger psychosocial and person-centred focus, largely influenced by works produced by health behavioural psychologists and ethics researchers (Resta, 2019). The expansion of services and training also led to the introduction of GC into other medical areas such as oncology, cardiology, and neurology (Abacan et al., 2019; Resta, 2019). Independence as a profession also occurred in the 1990s and 2000s with the establishment of professional genetic counsellor boards, an academic journal (namely the Journal of Genetic Counseling) and refinement of the definition of GC (Resta, 2019). Currently, it is estimated that there are 7,000 genetic counsellors in approximately 28 countries, with the USA, Canada, the UK, Australia and New Zealand having the most well-established GC services (Abacan et al., 2019; Ormond et al., 2018).

3.4 Genetic counselling models & frameworks

Several models and frameworks have been described in GC academic literature, textbooks and professional guidelines for practice and training purposes, most of which are based within medical, educational, psychotherapeutic, and ethical paradigms (Biesecker et al., 2019; Veach et al., 2003; Walker, 2009). Many of the models, frameworks and guidelines described overlap in focus, goals and approach with no singular approach prioritised. I expand on some of the main models and frameworks that shape GC practice below.

3.4.1 Roger's Person-Centred Theory

Although modern GC has evolved, much of its founding principles are based on Carl Roger's Person-Centred Theory (Biesecker et al., 2019). This psychological theory was not developed specifically for GC, but the profession draws largely from its beliefs and ideologies (Resta, 1997). The theory believes that every person has the inner ability to develop self-awareness and achieve both self-actualization and self-directedness (Rogers, 1992; Veach et al., 2003). Individuals should be viewed in their entirety, rather than only focusing on particular aspects of their being, that individuals strive for continuous personal growth and that individuals behave in rational ways when they can understand themselves (Hjelle & Ziegler, 1992). From a GC perspective, Roger's person-centred theory places emphasis on the counsellor's attitude and the counsellor-patient relationship. Counsellors are expected to be genuine, empathetic, and have unconditional positive regard towards the patient. This, together with respect for patient autonomy, is the foundation for non-directiveness in GC (Fine, 1993; Veach et al., 2003).

Non-directiveness, or neutral communication, is a central tenet of GC. Non-directiveness is informing an individual without persuading them, hence it aims to provide information and facilitate patients to reach a decision that is best for them and to enhance patient autonomy (Benkendorf et al., 2001; Kessler, 1997a). Alternatively, direct communication, or paternalism, can be defined as advice giving, or where the healthcare practitioner makes decisions on behalf of the patient based on what they assume is in the best interest of the patient (Murgic et al., 2015; Sandman & Munthe, 2010). Directive communication is thus influential and may be considered an attempt to undermine a patient's autonomy as it does not prioritise independent decision-making (Kessler, 1997a; Sandman & Munthe, 2010). To remain non-direct in GC practice, three key communicative features to avoid have been described: providing advice,

withholding information and making recommendations (Benkendorf et al., 2001; Brunger & Lippman, 1995; Michie et al., 1997).

A non-directive approach was first introduced into GC practice by Sheldon Reed in 1949, as the field of medical genetics attempted to move away from historic eugenic attitudes to a more ethical method of counselling that aligned itself with patient-centredness (Weil, 2003). Neutral approaches have also assisted genetic counsellors and genetic specialists (which includes medical geneticists and genetic nurses) to navigate the ethical debates in GC which focus on genetic disease and disability. Attempts at reducing 'unfavourable' physical and human features may become a contentious area amongst pro-life and human rights advocacy groups, the medical fraternity, and affected individuals and families (Rantanen et al., 2008; Walker, 2009).

Although a cornerstone of GC practice, there has been debate in the GC literature as to whether GC truly is, or can be, a neutral service (Arribas-Ayllon & Sarangi, 2014; Biesecker et al., 2019; Clarke, 1997, 2017; Pilnick, 2008; Pilnick & Zayts, 2012; Sarangi, 2010; Weil et al., 2006; Wessels et al., 2015; Wessels & Koole, 2019; Wyatt, 2001). Kessler (2001) also points out that although non-directiveness is often referred to in GC, it is difficult to define. Arguments have been made that complete neutrality in GC is unhelpful for patients and that active engagement from genetic counsellors is necessary to assist patients in meaningful ways (Weil et al., 2006). Non-directiveness is thus a fluid process (Bernhardt,1997; Michie et al., 1997) or one that occurs along a continuum (Benkendorf et al., 2001) rather than being completely absent from GC consultations.

An American study by Benkendorf et al. (2001) using interactional methods to study 43 prenatal GC consultations found that genetic counsellors oscillated between direct and non-direct forms of communication during a consultation through their word choice and sentence construction. The topic of conversation also impacted a genetic counsellor's communication style. For example, when providing medical or genetic information the style was typically directive, but when discussing a patient's decision-making, counsellors would revert to non-direct and neutral positions. Although attempting to adhere to GC guidelines, Benkendorf et al. (2001) reported that the genetic counsellor's continuous oscillation between direct and non-direct communication and overt use of indirect speech during particular moments in a

consultation created moments of patient confusion and prevented genetic counsellors from fully exploring patients' individual needs.

A recent study by Clarke (2017) has also spoken about the role of non-directiveness in current GC practice and how some genetic counsellors and practitioners in fields such as oncology and cardiology feel that non-directiveness no longer has a place in these specific GC areas. Oncology and cardiology-related GC often deals with pre-symptomatic individuals, thus promotes the use of genetic testing, as well as the prevention, surveillance, and treatment of disease. This approach differs from prenatal or paediatric GC where a condition may already be present and much of the discussion is on reproductive choice, which may bring about complex moral and ethical discussions. The advancement of GC into different health areas, contexts and the provision of complex tests thus create greater scope for debate on non-direct and direct GC communication.

3.4.2 Genetic counselling teaching & counselling models

With GC's two aims of education and counselling, Kessler (1979) proposed, and later refined (1997, 1999) two GC models that are still used in practice today - namely the teaching and counselling models of GC.

3.4.2.1 The Teaching Model of GC

This model emphasises that patients attend GC to seek information and that the genetic counsellors' purpose is to ensure that patients are educated so that they can make informed decisions (Kessler & Antley, 1981). Genetic counsellors are considered experts and are in authoritative positions. Their goal is not to provide psychosocial counselling, but rather to educate a patient by providing accurate, all-inclusive medical and scientific information in a neutral manner that is non-coercive and impartial (Kessler, 1997a; Kessler & Antley, 1981; Veach et al., 2003; Smets et al., 2007). Even though a patient's psychosocial issues may be attended to, this is not the primary focus of a consultation (Kessler et al., 1982).

3.4.2.2 The Counselling Model of GC

This model promotes an egalitarian stance and proposes that patients attend GC for reasons that extend beyond information seeking and that GC plays an important psychotherapeutic function for patients (Biesecker et al., 2017, 2019; Kessler & Antley, 1981; Redlinger-Grosse et al., 2017; Smets et al., 2007). In the counselling model, GC aims to understand the patient's

background, values and emotions, it aims to alleviate the patient's distress, helps the patient make meaning of the information provided, facilitates decision-making, and allows for a patient to gain a sense of control over their circumstances. Much of the counselling model is built of good communication skills and trust, with genetic counsellors encouraged to be flexible in their counselling approach to best meet the needs of the individual patients. Patients are thus permitted equal control over the agenda-setting of the consultation in this model, and whilst education is considered one of the goals of GC, the model does not perceive it to be the only goal of GC (Kessler, 1997b; Lewis, 2002; Smets et al., 2007; Veach et al., 2003).

3.4.2.3 Which model is favoured in practice?

Although existing separately in theory in early descriptions of GC, much of modern practice is a blend of the teaching and counselling models into what is described as a psychoeducational (Biesecker & Peters, 2001; Kessler, 1997b; Resta et al., 2006; Smets et al., 2007; Weil, 2003) approach which considers both the cognitive and affective impact of genetic disease. However, although both models are acknowledged, there is little understanding of the use and implementation of the two models in actual practice in different settings (Meiser et al., 2008).

Historically, GC practice was about reproductive choice and prevention of disease (Resta, 2019), thus much of GC practice was about information giving and education (Biesecker & Peters, 2001; Meiser et al., 2008; Resta, 2019). An American study by Roter et al. (2006) video-recorded 152 cancer and prenatal GC consultations with simulated patients and found that the teaching model was favoured and that genetic counsellors dominated talk with minimal attention to psychosocial concerns. Post genetic counsellor questionnaires suggested that reasons for an education-focused approach included the enormity of the teaching task, time constraints to build a meaningful relationship and a lack of confidence due to limited teaching and training opportunities for counselling techniques.

More recent literature suggests that the teaching model is still used today particularly with the expansion of genetic research, technology, and testing capabilities in the genomic era. GC is often described in modern media and by other health entities as a profession that provides information, genetic risk assessments for individuals and families, and assists in choosing appropriate genetic tests and interpreting the results (Biesecker et al., 2019). Skirton (2018) further describes how GC practices in the UK and the Netherlands are becoming increasingly education-focused, and that some Master's GC training programmes across Europe have been

noted to favouring biomedical approaches to GC training and practice, with less emphasis on counselling skills.

A greater emphasis on the counselling model and the psychosocial needs of a patient occurred particularly from the mid-late 1990s onwards when GC services moved into areas such as oncology, cardiology and other non-reproductive areas, and the increased interest of behavioural psychology within GC (Resta, 2019). Although research has shown the preferred use of the teaching model in practice (Ellington et al., 2006; Meiser et al., 2008; Roter et al., 2006; Skirton, 2018; Smets et al., 2007), researchers and genetic professionals have called for greater use of the counselling model (Henneman et al., 2008; Redlinger-Grosse et al., 2017; Skirton, 2018) or what Biesecker et al. (2017, 2019) has described as the 'therapeutic model' of GC. The counselling model has been deemed critical in the era of genomic medicine where decision-making under uncertainty and interpretation of ambiguous test results have become increasingly common (Austin et al., 2014; Benjamin et al., 2020; Biesecker et al., 2019), which may exacerbate feelings of anxiety and cause patient confusion.

Due to the hybrid nature of GC, genetic counsellors take on several roles including biomedical expert, educator, negotiator, service provider, gatekeeper, and psychosocial counsellor (Sarangi, 2000; Thomassen Hammerstad et al., 2020). Previous literature suggests that these roles are complex, which may be why genetic counsellors struggle to balance or achieve teaching and counselling expectations. To date, there is still no agreement on which of Kessler's GC models are preferred, nor how to blend these two models and their goals effectively in practice.

3.4.3 The Reciprocal Engagement Model (REM)

One of the first global GC models to be suggested is the Reciprocal Engagement Model (REM). This model was proposed in 2007 after calls were made for an empirically based model to guide GC practice rather than only the theoretical outcomes suggested by Roger's person-centred theory and Kessler's teaching and counselling models (Veach et al., 2007; Weil et al., 2006).

The REM was developed through collaboration amongst 23 programme directors from 20 GC graduate programmes in North America and suggested the following foundational principles: 1) genetic information and education are essential, 2) the patient-counsellor relationship is

integral to the service, 3) patient autonomy must be respected, 4) patients are resilient and 5) patients' emotions impact the GC process (Redlinger-Grosse et al., 2017; Veach, et al., 2007). More specifically, the REM emphasises 17 main goals which broadly include understanding the patient's background and adapting the counselling approach, accordingly, establishing a mutually agreed working relationship with the patient, awareness of the genetic counsellor's influence on the communication process and promoting patient empowerment and control over their situation (Redlinger-Grosse et al., 2016).

It can be noted that although the REM aims to integrate the 'best' aspects from previous GC models, empirical evidence on the REM too remains scarce (Redlinger-Grosse et al., 2016). More evidence-based research is thus necessary to understand the value of this GC model in actual practice.

3.4.4 Suggested components of a genetic counselling consultation

Different sources provide guidelines on how GC should be practiced, what the expected competencies of a genetic counsellor should be and what content Master's Genetic Counselling programmes should entail. The American-based NSGC (National Society for Genetic Counselors [NSGC], 2020) and the European Board of Medical Genetics (EBMG) (European Board of Medical Genetics [EBMG], 2020) both provide publicly accessible documents and guidelines for training and practice for genetic counsellors, genetic nurses, and medical geneticists on their official websites. Further suggestions for practice are provided in GC academic textbooks.

A typical GC consultation, as described by two American-based GC practice manual textbooks (Veach et al., 2003; Walker, 2009) is structured into the following suggested phases:

Introduction and Agenda Setting: In this section of the consultation the genetic counsellor is expected to provide details on GC, establish the patient's reason for referral and enquire and outline patient and counsellor goals and expectations for the consultation.

Gathering Information: In this phase, obtaining personal patient clinical information including chronic conditions, medical diagnosis or complications, as well as information on previous and current pregnancies. Gathering information on family members is also important here and

includes types of relations, relevant clinical information related to the patient's medical concern, consanguinity, infertility history, presence of birth defects and ethnicity.

Risk Assessment: Establishing or verifying a clinical diagnosis is an important part GC and may require input from a clinical geneticist or another medical specialist, before or during the GC process. Genetic testing can be ordered by a GC to confirm a suspected risk or clinical diagnosis too. Risk assessments usually involve a discussion about an individual's personal risk due to a confirmed genetic condition, or who may be at risk for a genetic condition based on their relation to other affected or at-risk family members. Risk assessments also take into consideration the reproductive risk for future children. Risk assessments in GC are usually based on an individual's family pedigree, personal clinical considerations such as phenotypic features, age, gender, medication use, reproductive history and lifestyle factors in relation to previous risk literature and mathematical calculations.

Providing Information: In this section of a consultation explanations of the risk assessments and relevant information are provided, together with the implications for the individual patient and other family members. Descriptions of various testing and management options are provided as well as information on useful sources and further support.

Facilitating Decision-Making: Exploring patients' views, discussing the pros and cons of the various test and management options and their outcomes is important in this phase.

Psychosocial Support: Information on a diagnosis or personal and family risk can bring about strong emotions for patients that need to be acknowledged and addressed. Genetic counsellors need to assist patients to adapt and cope with the reality of a condition and the information received. Genetic counsellors also need to be able to recognise psychological concerns or distress that are beyond their professional scope and to provide appropriate referrals for patients. Acknowledging and exploring a patient's cultural beliefs, value system, experiential, social and educational background is also important. Walker (2009) highlights that these issues need to be attended to throughout the GC consultation and not only within a particular phase.

Ending the consultation: Summarising information from the consultation, arranging next management steps and referrals, providing counsellor contact information, and arranging

follow-up consultations or referrals, if appropriate, is needed in the closing section of a consultation.

3.5 Genetic counselling goals & outcomes

The outcomes of GC have shifted in time with the advancement of science, the various global settings where GC is offered, and the clinical sub-specialities within which GC practices (Resta, 2019; Biesecker et al., 2019). The GC literature also uses the words 'goals' (Biesecker et al., 2019), 'ideals' (Rantanen et al., 2008) and 'outcomes' (Redlinger-Grosse et al., 2016), when describing GC practice. Goals and ideals can be defined as theoretical endpoints or achievements which efforts are directed towards (Dictionary.Com, 2020), whilst outcomes can be described as actual patient consequences or end-results of their counselling interactions (Biesecker & Peters, 2001).

There are several goals and outcomes for GC consultations described in the GC literature. Defined GC goals include reducing and managing risk (Resta et al., 2006), assisting individuals and families in health-related and reproductive decision-making (Bernhardt et al., 2000), education (Biesecker & Peters, 2001; Resta et al., 2006), emotional adaptation to genetic disease (Biesecker et al., 2019), as well as increased personal control and patient empowerment (McAllister & Dearing, 2015). Further goals or 'ideals' include the provision of relevant and unbiased information to patients, checking patient understanding, providing psychosocial support, informed consent, protecting patient privacy, consideration of familial risks, management of appropriate testing and ensuring autonomous patient decision-making. These were described in a literature review by Rantanen et al. (2008) focusing on 56 publications from different global professional, patient, political and ethical organisations.

Various quantitative and qualitative studies have attempted to measure GC outcomes which have included patient knowledge and education (Baldwin et al., 2012); changes in risk perception and patient preferences in risk communication (Braithwaite et al., 2004; Lobb et al., 2003); patient satisfaction (Davey et al., 2005; DeMarco et al., 2004; Guimarães et al., 2013); psychosocial adaptation to genetic information such as distress (Codori et al., 2005); anxiety (Pieterse et al., 2006) and personal control (McAllister & Dearing, 2015); alterations in family communication (Gaff et al., 2005; Mendes et al., 2018); group versus individual counselling

(Hynes et al., 2020) and in-person GC versus online tele-counselling (Buchanan et al., 2015; Otten et al., 2016) to name but a few.

One of the biggest challenges in measuring the outcomes of GC is that there is no agreement within the GC profession, nor in the broader medical field whether GC's primary goal is education or counselling, and how, or which patient outcomes should be measured (Benjamin et al., 2020; Biesecker et al., 2019; MacLeod et al., 2002; Skirton, 2018; Veach, et al., 2003; 2007; Weil et al., 2006). Below are some descriptions of attempts by researchers to understand and consolidate GC outcomes.

3.5.1 The Genetic Counselling Outcome Scale

McAllister et al. (2011) have attempted to provide a comprehensive patient-reported GC outcome framework to try to address challenges in knowing what GC outcomes should be as reported by patients. In their UK-based study, McAllister et al. (2011) conducted a phased approach. In the first phase, 527 participants were recruited from various patient support groups (all of whom represented a wide range of genetic conditions or concerns). Participants were asked to complete an 84-point psychology-adapted questionnaire. Based on the responses, a second questionnaire was developed called the Genetic Counselling Outcome Scale (GCOS-24) which included 24 questions. Two hundred and forty-one patients were then invited to participate in a second phase of the study where they were sent a questionnaire pack which included the GCOS-24 and several other psychology-based assessment scales/questionnaires, much of which was focused on measuring perceived personal control, anxiety, depression, and satisfaction. The participants were then asked to complete the same questionnaire pack several weeks after their GC appointment to assess variations in their answers. Emerging results showed that patient empowerment was considered a particularly important outcome of GC. Further assessment of the GCOS-24 in other research settings has re-emphasised the value of patient empowerment as a GC outcome, and that information provision alone does not make a meaningful difference to the lives of patients (Thomas & McAllister, 2019).

The use and benefit of the GCOS-24 scale, and its adapted version, the Genomics Counselling Scale (Grant et al., 2019) has since been referred to in preceding GC literature (Biesecker et al., 2019; Resta, 2019; Thomas & McAllister, 2019) and has been used in other UK-based (Costal Tirado et al., 2017), Canadian (Inglis et al., 2015) and Danish (Diness et al., 2017) GC settings. The GCOS-24, however, remains problematic in its adaption for all contexts and

individuals, since the foundations of the outcome scale are based on reports from predominantly Caucasian patients (McAllister et al., 2011). GC outcomes may vary across global settings and different ethnic or cultural groups. GC outcomes may also vary amongst sub-groups of GC patients e.g., those referred for cancer versus those referred for prenatal GC (Thomas & McAllister, 2019). Patient needs and GC outcomes in other global settings therefore need further exploration.

3.5.2 Genetic counselling outcomes based on the Reciprocal Engagement Model (REM)

A study by Redlinger-Grosse et al. (2016) highlights the lack of a comprehensive genetic counsellor-reported GC outcome framework and that the necessity for guidelines or a framework has become a priority for the NSGC. The study conducted several focus groups with 27 USA based GC programme directors, senior practising genetic counsellors and experts in GC outcome research to elucidate the outcomes of GC based on the 17 REM goals. Results from the study revealed 197 described GC outcomes. Key outcomes included attention to a patient's social, family and cultural background; focusing on a patient's locus of control, self-efficacy and self-esteem; observing changes in the patient's quality of life, and medical management; a patient receiving relevant genetic and risk information that they can comprehend, the ability for a patient to make informed decisions, and that a patient's emotional needs are acknowledged and met.

Despite the many outcomes described participants in the Redlinger-Grosse et al. (2016) study raised concerns about the ability to measure or achieve all GC outcomes, and how time and context may change GC outcomes. A call for evidence-based outcomes and finding methods that can elucidate the true nature of GC practice and its outcomes thus remains a priority for the GC field.

3.5.3 The FOCUS-GC framework

The Framework for Outcomes of Clinical Communication Services (FOCUS)-GC (Cragun & Zierhut, 2018) is one of the latest frameworks suggested in GC literature and attempts to consolidate and identify many of the existing models to highlight the main outcomes and goals. There are several working groups involved with the FOCUS framework, mainly from the USA and Europe, with much of the framework still under review. Current focuses of the framework include communication process measures, patient care experiences, the impact of GC on families and the impact of GC on patient health. Attention to context is also mentioned in the

framework and how individual, family, social, cultural and institutional factors impact GC services and relationships between patients and providers. Evidence-based research in various healthcare settings is encouraged to understand different contextual variables (Cragun & Zierhut, 2018).

The lack of agreement on what GC outcomes should be and the paucity of the latest evidence-based knowledge is a further motivation for the development of the framework. Apart from understanding how GC improves patient care, many of the latest questions have arisen due to pressures from other healthcare fields for genetic counsellors to provide evidence of their added value and quality of patient care (Cragun & Zierhut, 2018; Hooker et al., 2017; McAllister et al., 2016; Resta, 2019; Zierhut et al., 2016). New understandings of the value of GC are critical since much of GC practice, goals and genetic technologies have advanced and expanded beyond some of the earlier models, definitions and outcomes of GC (Biesecker et al., 2019; Cragun & Zierhut, 2018; Resta, 2019).

3.6 Risk studies in genetic counselling

Although sharing many of the difficulties of typical medical consultations, GC offers a unique field in which to study health risk communication. The information conveyed includes complex explanations of risk, inheritance patterns, as well as scientific and molecular terminology that may be difficult to understand, even for well-educated individuals (Penn & Watermeyer, 2012a).

Although several risk communication strategies have been made in theory, one of the biggest challenges in GC is the application of risk communication knowledge in practice (O'Doherty & Suthers, 2007). Although GC attempts to educate patients on their risk, GC studies have suggested that despite efforts by genetic counsellors, a patient's risk perception does not necessarily alter after a GC consultation (Austin, 2010; Fransen et al., 2006; O' Doherty & Suthers, 2007; Wessels & Koole, 2019). These studies support an earlier finding by Lippman-Hand and Fraser (1979) investigating patients' experiences of being at risk for a genetic condition that patients view risks in a binary manner (it will or will not happen), and that they may not be able to assimilate or adjust to new risk information provided to them.

Much of GC risk communication research has stemmed from cancer-related GC practices where several types of personal, familial and population-based cancer-risk information are typically conveyed (Cicero et al., 2017; Davis et al., 2020; Dorval et al., 2013; Hallowell et al., 2007; Henneman et al., 2020). However GC risk communication has also been addressed in paediatric and adolescent settings (Lipkus, 2010; Metcalfe et al., 2011; Wu et al., 2017); prenatal GC (Georgsson et al., 2017; Hodgson et al., 2010; Pighin et al., 2013); psychiatric GC (Jenkins & Arribas-Ayllon, 2016); GC for multifactorial conditions (Waxler et al., 2012); and GC for chromosome rearrangements (Walser et al., 2016). Studies have also focused on patient experiences and understanding of risk information based on different GC approaches such as face-to-face consultations and technology-based platforms such as video and telephonic consultations (Gordon et al., 2018; Jacobs et al., 2016).

Methodologies to study risk communication have included: quantitative-based patient- and general population- questionnaires and online surveys (Chapman & Coups, 2006; Eichmeyer et al., 2005; Henneman et al., 2020); quantitative coding methods such as the Roter Interaction Analysis System (RIAS) to assess dialogue content (Ellington et al., 2005; Michie et al., 2005); qualitative patient focus groups and interviews (Dorval et al., 2013; Gordon et al., 2012; Kinney et al., 2010; Schapira et al., 2001); as well as genetic counsellor interviews (Arribas-Ayllon & Sarangi, 2014; Henneman et al., 2008).

Understanding the formats used to convey risk information, and patient preferences in this regard, have also been studied. Researchers have claimed that knowing what format a patient prefers when receiving genetic risk information not only improves patient-centred care (Henneman et al., 2020) but that patients will become more engaged in the communication process since the conveyed risk information will be useable and meaningful (Fortin et al., 2001). Several researchers have focused on the various formats to convey genetic risk information in GC including statistical representations, narratives (or verbal descriptions), visual and graphical displays, as well as metaphors and analogies (Dorval et al., 2013; Henneman et al., 2008, 2020; Lobb et al., 2003; Michie et al., 2005). Many studies looking at risk formats in GC conclude that tailoring information according to patients needs and background were important and that a ‘one size fits all’ approach is not recommended. Although numerical information remains a popular method of presenting genetic risk information, studies have highlighted that these formats are best used in combination with verbal or graphical explanations (Dorval et al., 2013; Henneman, 2020; Michie et al., 2005; O’

Doherty & Suthers, 2007) and that numerical risk information is not necessarily helpful to individuals with lower literacy or numeracy abilities (Joseph et al., 2017; Portnoy et al., 2010). Little is known about the use of different risk formats in GC consultations in socio-cultural diverse settings, however.

In attempts to consolidate risk communication knowledge, researchers have created a GC risk communication checklist (Fransen et al., 2006); guidelines (Etchegary & Perrier, 2007; O'Doherty & Suthers, 2007) and a GC risk communication aid (Lobb et al., 2006). The NSGC also has several condition-specific practice guidelines (strict adherence is advised) and practice resources (flexible adherence is recommended that is dependent on context) available on their official website and they are published in their home academic journal, namely the Journal of Genetic Counselling. The European Society for Human Genetics (ESHG) (European Society for Human Genetics [ESHG], 2020) and the EBMG (EBMG, 2020) also provide guidelines for different GC-related areas and recommended practice on their official websites.

Using existing literature and piloting it in Dutch GC consultations, Fransen et al. (2006) developed a checklist for genetic counsellors focusing on three features of risk communication: How risk is presented, how decision-making under uncertainty is facilitated, and to whom the risk information is being conveyed (i.e., the context of the individual patient). The checklist had a 'yes/no' checklist which genetic counsellors referred to when conveying information. The checklist included considerations of the following: The format used to convey risk information (e.g., percentages, proportions, verbal explanations), paying attention to the use of scientific jargon, non-verbal cues and patient emotions, as well as how to start and end a GC consultation. The checklist also included the use of a decision-making tool which was divided into four steps to support a patient when making a decision under uncertainty: 1) helping a patient to identify and define what decision challenge is, 2) creating a decision-making tree for a patient by discussing all possible options and consequences of their different decisions; 3) conveying probability of risk in each option and outcome, and 4) facilitating a preferred choice of action for the patient based on the patient's values and the likelihood of each outcome occurring.

GC risk communication guidelines and cautions for 'pitfalls' in risk communication have also been described by O'Doherty and Suthers (2007). For example, they caution against conveying potentially confusing population risk information; encouragement to use both negative and

positive framing rather than only one type of frame due to the persuasive nature of framing; provision of absolute rather than relative risk information and acceptance that subjective risk perceptions may influence logical decision-making. Providing numerical or statistical risk information together with verbal explanations rather than in isolation is further encouraged, as well as contextualising risk information so that a patient can make meaning of the risk on a personal level.

Etchegary and Perrier (2007) provide further risk communication guidelines by suggesting that genetic counsellors conceptualise risk within an information processing theoretical framework. This framework attempts to elucidate how patients may seek, avoid and respond to risk information based on different theoretical models and how these models may apply in GC risk communication settings.

Lobb et al. (2006) have developed a GC risk communication aid to be used between medical geneticists and high-risk breast cancer families in Australia. The aid was developed in collaboration with medical geneticists, genetic counsellors, psychologists, an epidemiologist, an oncologist, a linguistic, and a patient, and consisted of 13 colour pages that include information on personal and population breast cancer risk, information on BRCA1 and BRCA2 mutations, family risk information, screening and management options. In addition to written words, graphical formats such as pie charts, bar graphs and numbers were also included. Piloting the study with 38 unaffected patients found that patients' knowledge improved, and their risk perceptions were more aligned to the objective risk information.

3.7 Uncertainty studies in genetic counselling

Uncertainty is a natural presence within GC due to the nature of complex risk information and making decisions with limited knowledge. Sarangi and Clarke (2002a) explain that the genetic counsellor/practitioner experiences uncertainty relating to the inability to convey accurate knowledge, and the patient experiences uncertainty with having to make a medical decision with information that is unclear. Not only are the health and social outcomes of genetic conditions difficult to predict, especially those with clinical and age-of-onset variability, but the effectiveness of treatment; preventative options and test interpretations differ between individuals (Bernhardt et al., 2014; Bylund et al., 2012; Clarke & Wallgren-Pettersson, 2019; Hashiloni-Dolev et al., 2019; Henneman et al., 2008; Lou et al., 2020a; Pilnick & Zayts, 2014;

Steel et al., 2014). Uncertainty in GC can also extend beyond the self and can relate to concerns for other family members, as shown in a study by Bylund et al. (2012) which focused on mothers who were at risk for *a BRCA1/2* mutation and their concerns relating to their daughters.

Uncertainty is described in prenatal (Hammond et al., 2020; Kalynchuk et al., 2015, Lou et al., 2020a; 2020b); paediatric GC literature (Lipinski et al., 2006; Steel et al., 2014); cancer GC (Ardern-Jones et al., 2010; Bylund et al., 2012; Rauscher, 2017); psychiatry GC (Hippman et al., 2013); GC for adult-onset conditions (Chiò et al., 2014); GC for mitochondrial disorders (Vento & Pappa, 2013) as well as the delivery of complex genomic test results (Thomassen Hammerstad et al., 2020).

In prenatal GC, uncertainty typically stems from difficulties in deciding to undergo invasive testing e.g., an amniocentesis (which carries a small, but significant procedural-related miscarriage risk) when a genetic risk has been identified (Salomon et al., 2019), or deciding whether to undergo a termination or not when a genetic condition/significant risk has been confirmed (Scott, 2018). Uncertainty in prenatal GC also stems from the inability to interpret ultrasound findings, or diagnostic test results, especially those generated by advanced genetic tests such chromosome microarrays (CMA) and whole-genome and exon sequencing in modern prenatal practice that exacerbate, rather than alleviate uncertainty in some instances (Lou et al., 2020a; Richardson & Ormond, 2018). The uncertainty from these test results is due to the vast amount of test information that is produced, with many results not yet understood from a clinical or risk implication perspective.

Lippman-Hand and Fraser (1979) found that pregnant patients experience three categories of uncertainty during prenatal decision-making in GC: 1) the unknown impact of having an affected child, 2) not knowing how to decide and fears about how other individuals would perceive their decision choice, and 3) not knowing if they would be able to parent a child with a disability. The limitations to gain certain information on the health of their pregnancy and unborn baby causes great anxiety for parents and places genetic specialists in difficult counselling positions.

A study by Hammond et al. (2020), interviewing 16 parents from the UK and the Netherlands who had experienced uncertainty due to the detection of a structural fetal anomaly, found that sources of uncertainty for parents included the diagnosis, prognosis and cause of a fetal

anomaly. Further sources of uncertainty included the lack of clarity from medical professionals on what the anomaly may mean (the outcome may be significant, or the outcome may be insignificant), with one participant describing that 'clinical suspicion' (Hammond et al., 2020, p.5) was not enough for them to make confident decisions, and the short timeframe in which to make decisions. Parents mentioned personal uncertainty management strategies which included doing internet-based searching, seeking support from family and friends, connecting with other parents who had gone through similar experiences, and remaining hopeful.

Research has shown that in the era of modern medicine, pregnant patients aim to seek as much information as possible about their unborn baby (Kalynchuk et al., 2015; van der Steen et al., 2015), however, may regret receiving uncertain information as shown in a Danish study by Lou et al. (2020a) due to the creation of anxiety and fear of having a baby with a severe disability. When realising that the outcome is less severe than what was anticipated, parents may experience further regret having sought advanced prenatal genetic testing.

Uncertainty is also felt by genetic counsellors and medical geneticists about genetic risk information. A study which provided questionnaires to 193 prenatal genetic counsellors in the USA asking about their experience and comfort with providing pre-and post- GC for CMA testing found that only 59% of the counsellors felt comfortable with providing CMA counselling, whilst only 43% felt comfortable discussing the termination of pregnancy based on CMA results (Bernhard et al., 2014). A study interviewing 17 Israeli pregnant, or recently pregnant genetic counsellors further found that being an expert in the field did not make participants feel more comfortable with the uncertainty felt during prenatal screening, and decision-making on genetic testing (Shkedi-Rafid & Hashiloni-Dolev, 2018).

Furthermore, uncertainty within GC consultations, as described in other health literature is also a relational concern (Han et al., 2011; Hogan & Brashers, 2015) that impacts patient-genetic counsellor/practitioner communication (Arribas-Ayllon & Sarangi, 2014; Hammond et al., 2020; Hippman et al., 2013; Lou et al., 2020b; Pilnick, 2002; Pilnick & Zayts, 2014; Rauscher, 2017; Stivers & Timmermans, 2016).

Findings from Hammond et al. (2020) also demonstrate how communication in GC can assist in managing uncertainty. Parents in the study whose unborn babies were found to have a structural abnormality mentioned that the way the healthcare practitioner spoke to them (using

a slow and calm pace), allowing time for questions, and displaying an empathetic manner, were particularly helpful. Addressing patients' emotional responses to uncertainty also assists how patients respond to information as shown in a cancer GC study (Rauscher, 2017).

A study by Hippman et al. (2013) described the difficulty of providing risk information to patients and families affected by bipolar disorder and schizophrenia due to the complexity of psychiatric genetic and molecular information. The study indicated that some genetic counsellors admitted that counselling may create further uncertainties for these patients rather than alleviate concerns. Conversely, however, affected patients and families indicated high satisfaction levels with GC and that their reasons for attending GC extended beyond receiving certain information. Rather, patients felt that GC provided an opportunity for meaning making, improved personal control and hope for the future when faced with an uncertain situation. Using uncertainty as a tool to provide hope and adaptation to a situation is further described in a study focused on 546 caregivers of individuals with Down syndrome in the USA (Truitt et al., 2012).

The above-mentioned studies suggest that the relationship and the psychosocial aspect of GC are highly valued by patients and that patient-centred communication has the potential to aid patients to cope with feelings of uncertainty.

3.8 Risk & uncertainty communication in genetic counselling & their relation to care

Risk and uncertainty communication in GC can be closely linked to the notion of care. GC is often seen as a 'caring' service. This is because GC is not just an opportunity for providing information but is considered a therapeutic process in which a patient and their family receive psychosocial support and facilitation during autonomous decision-making (Austin et al., 2014; Benjamin et al., 2020). The profession thus bridges medicine and psychological counselling where attempts towards meaning-making are explored together with the presentation of risk figures (Lobb & Gaff, 2010).

According to the Oxford English Dictionary, the term 'care' can be defined as ensuring the necessities that maintain the health, wellbeing, and protection of something or someone (Lexico, 2020). The act of care may extend to self, to others and to our environment so as to

ensure that life and how we live in the world may continue in the best way possible (Fisher & Tronto, 1990; Tronto, 1993).

GC guidelines emphasise that genetic specialists should assist patients to adapt physically, mentally, and emotionally to genetic risk and support them through life-changing decisions. Apart from being expected to convey complex information in an understandable manner, genetic counsellors are encouraged to be patient-centred, compassionate, empathetic, trusting, and to have the necessary skills to address psychosocial topics and interpersonal tensions (Balkite & Smith, 2006; Kessler, 1999).

Biesecker (2016) highlights how GC encompasses characteristics of 'care':

> It [GC] engages us in experiences of human suffering and resilience that we would otherwise not have the privilege of participating in. It is intimate work born of compassion and engagement with others at a meaningful level. Strangers admit us into their lives by sharing their narratives, including their thoughts and feelings about their greatest fears and hopes. (p. 621)

In GC consultations, for informed choices to occur, patients need to have a clear understanding of the risk information given to them. The genetic counsellor or practitioner thus carries a responsibility to present risk information in an understandable, sensitive manner using appropriate formats (Henneman et al., 2008). People also have varying beliefs on what causes genetic conditions which may differ to typical scientific explanations such as family transgressions, spiritual causes, and environmental factors such as eating certain foods whilst pregnant (Barlow-Stewart et al., 2006; Morris, 2013; Penn et al., 2010), all of which may impact on how patients respond to genetic risk information and uncertainty.

Although a patient's family members and their risks are discussed during a patient's consultation, contacting and informing family members about their risks is often left to the consulted patient, rather than the genetic counsellor, for patient confidentiality and privacy reasons (Hodgson et al., 2016). While a patient may be encouraged by a genetic counsellor or practitioner to discuss information with their family, conveying complex genetic risk information to their family may be challenging for the patient. At-risk family members may thus remain uninformed or may receive either the incorrect or only partial risk information due

to a patient's misunderstanding or lack of confidence to convey the information received (Hodgson et al., 2016; Mendes et al., 2018). Geelen et al. (2011) further highlights that genetic risk information may bring about social disruption within a family unit and cause tensions.

Collectively, GC consultations may thus take on a 'heaviness' in terms of a patient's and their family's informational and psychological needs that have to be addressed. Several emotive and uncertain conversations may occur both during a GC consultation and the consequences may extend to the broader family. Effective risk communication, providing support, checking of understanding, and assisting patients to become confident and comfortable with genetic risk information and uncertainty is thus a vital task for healthcare practitioners providing genetic counselling (Hodgson et al., 2016; Mendes et al., 2018). Furthermore, enacting and ensuring care is a critical, yet complex, part of GC when discussing risk and dealing with uncertain decision-making. What is considered 'good' patient care may also differ within varying contexts.

3.9 Conclusion

In conclusion, this chapter defines genetic counselling, provides a brief history of the profession, as well as describes the models, frameworks, goals and outcomes of GC. The chapter also discusses the importance of risk communication, and how risk is presented in GC. Lastly, the chapter discusses GC literature regarding uncertainty, and how uncertainty occurs due to a lack of knowledge, and within patient-genetic counsellor/practitioner relations due to communication approaches. Managing uncertainty as part of patient care is also described.

Most of the studies and literature presented in this chapter on previous risk and uncertainty studies in GC are based on quantitative and qualitative surveys, interviews and questionnaires. Further interactional and process-related studies focusing on risk and uncertainty are expanded upon in *Chapter Five.*

CHAPTER 4: GENETIC COUNSELLING IN SOUTH AFRICA

4.1. Introduction

This chapter serves as a background to the genetic counselling profession in South Africa. It provides an overview of the profession's history and status in South Africa, and details local counselling and training facilities and approaches. The chapter also provides literature on genetic disease prevalence, available genetic services and how these services are embedded into the country's broader healthcare system. Details on how South Africa's socio-cultural context and individual patient characteristics may influence local GC practice and risk communication are also provided.

4.2 Genetic counselling history in South Africa

In South Africa, limited informal medical genetics services have been offered since the 1950s by interested cytogeneticists and physicians, however, human genetics was only included into local medical schools' curriculums after the 1970s, after strong advocacy for medical genetic training was emphasised, particularly by Professor Phillip Tobias from Wits University in Johannesburg. Clinical genetics was also initially recognised as a sub-speciality practised by doctors trained in paediatrics, pathology and internal medicine, with medical genetics only officially recognised as a primary speciality in medicine in South Africa in 2007 (Jenkins, 1990; Kromberg et al., 2013b). The first Chairs and Departments of Human Genetics were established at UCT and Wits in the 1970s, with the University of Stellenbosch and the University of Pretoria following soon thereafter. There are currently four universities with medical genetics departments that offer genetic services and formal training, and another three universities offering limited medical genetic services (Beighton et al., 2012; Jenkins, 1990).

South African GC services are offered by genetic counsellors, medical geneticists and genetic nurses (Greenberg et al., 2012; Kromberg et al., 2013a; Ormond et al., 2018). Local GC services provide consultations and care for individuals who may be at risk for a birth defect, or an inherited monogenic or chromosomal disorder for which prenatal genetic diagnosis, genetic screening, as well as diagnostic, predictive, and carrier testing may be available (Kromberg & Krause, 2013; Kromberg et al., 2013b). Medical areas focused on in local GC practice include paediatric, preconception and prenatal care, oncology, ophthalmology and neurology related areas.

4.3 South African genetic counselling training

Formal two-year Master's-level genetic counsellor training was first offered in South Africa in 1988 by Wits University (based in Johannesburg), and later by UCT (based in Cape Town) in 2004 (Kromberg et al., 2013a). In addition to the two-year Master's programme, UCT also recently started offering six-month medical genetics short-course to interested healthcare practitioners with applicants needing a background in medicine, biological sciences, nursing, psychology or social work (University of Cape Town [UCT], 2021a). Whilst locally adapted to some extent, local GC training is largely informed by USA-based GC training models and psycho-educational counselling approaches that blend Kessler's teaching and counselling models (Biesecker & Peters, 2001; Kromberg et al., 2013a; Ormond et al., 2018). To date, UCT and Wits are the only two centres offering formal genetic counselling training in the country.

In addition to genetic counsellors offering GC services, medical geneticists and genetic nurses also provide local GC services. The provision of GC services by other healthcare practitioners is not uncommon where there is a shortage or absence of genetic counsellors and has been described in South America, other African countries, Asia and some European countries (Abacan et al., 2019; Ormond et al., 2018). Various contextual settings affect the content of GC training and services, who offers GC, and how it is offered (Beighton et al., 2012; Bucio et al., 2019; Cordier et al., 2012; Kromberg et al., 2013a; Ormond et al., 2018; Pilnick & Zayts, 2019; Rantanen et al., 2008; Sahhar et al., 2005; Wessels & Koole, 2019, Zayts-Spence et al., 2021). South African training, patient interactions and consultation approaches for medical geneticists and nurse are regulated and are typically aligned with genetic counsellor practice and philosophy (Greenberg et al., 2012; Kromberg et al., 2013a, Ormond et al., 2018).

All qualified and practising genetic counsellors (including interns) are registered with the Health Professionals Council of South Africa (HPCSA) (Kromberg et al., 2013a) which is a large regulatory body that oversees training, education and registration of almost all local healthcare practitioners.

According to the HPCSA, the following outcomes are recommended as part of the national GC training curriculum: 1) understanding and having up-to-date genetic, scientific, medical and counselling information; 2) effective professional and patient communication skills (including setting an agenda, conveying information clearly, exploring psychosocial issues, and

facilitating informed decision-making), and an ability to communicate with all individuals regardless of their background, values or beliefs; 3) having critical thinking skills to assess, apply, and adapt genetic counselling knowledge and skills to various clinical consultations; 4) involvement in education, research and upholding inter-professional relationships; and 5) upholding ethical standards in accordance to HPCSA and GC professional guidelines (Health Professionals Council of South Africa [HPCSA], 2019). Genetic counsellors (and other genetic specialists) are also expected to be able to assess a patient's level of understanding and to modify their counselling approach accordingly. Attention to the psychosocial needs of patients is also highlighted, as well as regular debriefing and reflexivity to improve practice (HPCSA, 2019). These national guidelines have been approved by the Genetic Counsellors South Africa (GCSA) society and by the two local GC training institutions (namely the Divisions of Human Genetics at UCT and Wits University). After completion of training and a two-year internship at an HPCSA-registered institution, local genetic counsellors in training submit an assessment portfolio to the HPCSA for examination, and if successful, are granted registration to provide independent practice (HPCSA, 2019). Regular assessment of compliance and continuing professional development of registered genetic counsellors are carried out by the HPCSA on an annual basis.

For local practicing GCs, the HPCSA's Standards of Practice for Genetic Counsellors document exists to guide practice (HPCSA, 2013). Much of this document overlaps with the philosophies and goals highlighted in the HPCSA (2019) GC training guidelines. emphasises non-directive counselling where individuals have the right to "be informed of their genetic risks and reproductive options" (HPCSA, 2013, p. 4). Furthermore, genetic counsellors are expected to provide accurate risk assessments and to educate individuals about their "medical, genetic and scientific aspects of the condition/s and the associated risks" (HPCSA, 2013, p. 4). The HPCSA (2013) document however highlights the necessity for patient-centered counselling which considers a patient's "educational, socio-economic, ethnocultural, religious and historical backgrounds' as well as their 'traditions, health beliefs, lifestyles and values" (HPCSA, 2013, p. 5).

The most recently published literature in this regard indicates that there are approximately 25 qualified genetic counsellors and 12 medical geneticists in South Africa, with most based in the Western Cape and Gauteng provinces where they predominantly work within genetic service-based healthcare teams (Malherbe et al., 2016; Ormond et al., 2018). However, not all

qualified genetic counsellors are in clinical GC posts, with some working in research or academic posts, or other professional areas (Genetic Counsellors South Africa [GCSA] committee, personal communication, 2021). GC clinical opportunities and posts remain scarce due to the prioritisation of other healthcare areas and conditions by the South African Government (Malherbe et al., 2016).

4.4 Genetic counselling practice within a local context

Genetics transcends national borders, politics, social classes, age, gender, language and ethnicity. How individuals respond to genetic conditions, however, differs between individuals and across contexts (Alliman et al., 2009; Beighton et al., 2012; Cordier et al., 2012; Edwards, 2010; Kromberg et al., 2013a; Pilnick & Zayts, 2012; Rantanen et al., 2008; Sahhar et al., 2005, Zayts-Spence et al., 2021). The South African setting is introduced in this section concerning GC practice.

4.4.1 The prevalence of genetic disease in South Africa & the use of genetic services

Genetic and congenital disorders are a common occurrence, with global estimations of 1 in 15 live births being affected by a congenital disorder. Of these 1 in 15 affected births, 80.5% are thought to be genetic in nature, and the remainder caused by teratogen exposure (Christianson et al., 2006). Despite these statistics, genetic and congenital disorders often receive little attention in developing countries such as those within Africa due to the focus on infectious diseases such as HIV/TB and malaria, prioritisation of maternal and child mortality, as well as poverty (Christianson & Modell, 2004). Collectively, these contextual factors hamper the provision of genetic services, testing and support, with little or no clinical, diagnostic or research capacity or infrastructure despite the role of genetics in monogenic and multifactorial conditions in many African countries (Beighton et al., 2012; Christianson et al., 2006).

South African research has shown that there is a lack of awareness of GC services amongst patients and healthcare practitioners alike (Malherbe et al., 2016; Morris, 2013; van Wyk et al., 2016) and the local GC field remains relatively small (Kromberg et al., 2013a). A study by Malherbe et al. (2016) showed that of 83,118 predicted annual births in South Africa having a congenital disorder in 2012, only 2,714 of these births were recorded by the National Department of Health in that year. These figures indicate under-reporting by nearly 97%. This level of under-reporting suggests that affected individuals and families are not receiving

adequate support by appropriate medical genetic services. Unlike in some developed countries, there are also no sub-speciality GC services in South Africa. Rather, local genetic counsellors tend to be generalists who consult on all GC-related referrals. Reasons for this include challenges in geographic access and the small number of local GCs (Greenberg, et al., 2012; Ormond et al., 2018).

In South Africa, all medical genetic services are offered through a referral system, with 80% of patients seen in public healthcare services (Greenberg et al., 2012). Research by Kromberg et al. (2013a) indicated that for the 2007-2008 period, 3365 national cases were seen - 61% of which were counselled by medical geneticists, whilst 39% were seen by genetic counsellors. There were 57 different types of diagnoses made, with the most common being advanced maternal age (AMA), fetal abnormalities, abnormal ultra-sounds during pregnancy, Down syndrome, Trisomy 18, Huntington Disease, cystic fibrosis, sickle cell anaemia, retinal disorders, haemophilia, muscular dystrophy, developmental delay, inherited breast and ovarian cancer syndrome (HBOC), hereditary colon cancer, neurofibromatosis, multiple miscarriages and metabolic disorders. Albinism, particularly amongst the Setswana population, as well as consanguinity, is also common.

South Africa also has a unique genetic disease profile, much of which still needs to be understood. The recent establishment and prioritisation of the African Genome Variation Project not only hopes to better understand African populations from a genetic and molecular perspective but aims to provide important support for African-based medical genetic research and services (Gurdasani et al., 2015; Kromberg et al., 2013b; Mulder et al., 2018). Particular genetic variants are also unique to some South African groups such as the Afrikaner and Ashkenazi Jewish populations due to founder effects (Schoeman et al., 2013; Seymour et al., 2016).

4.4.2 The healthcare system & social circumstances

The overarching healthcare system, as well as the communities in which these systems exist, are significant influences of health service provision. Even though a health practitioner and a patient may have a positive relationship, if this interaction occurs within a poor operating environment, the desired health outcome may not be achievable (Bensing et al., 2003). Healthcare communication in South Africa is highly influenced by its social, historical, political and health context, which all form part of the broader operating environment.

Although governmental efforts have been made towards improving the quality of basic living standards and service delivery, socio-economic problems such as poverty, unemployment, as well as inadequate access to basic housing and healthcare remain high (Coovadia et al., 2009; Mayosi et al., 2012; Statistics South Africa, 2019). The country's healthcare system is also divided between private and public facilities. Although easily accessible, and used by 83.8% of the population, the public healthcare system is under-resourced, over-stretched, and is unable to provide certain medication, tests and procedures (Maphumulo & Bhengu, 2019; South African Department of Health, 2011). Almost all public healthcare patients access services through a referral system, and many are unfamiliar with specialised services.

South Africa is made up of nine provinces. Each province has its own unique socio-demographic and economic make-up, all of which influence what, and how, services are provided. The Western Cape, where this study took place, is the fourth largest province in the country, compromising of 7 million of South Africa's 59 million people and is considered to have better public access to infrastructure, social and education services, and healthcare than other provinces and cities in South Africa (Statistics South Africa, 2019; 2020). Much of the Western Cape population is made up of Mixed Ancestry, Black African, White, and Indian/Asian individuals, as well as a large migrant population. Although multiple languages are spoken, Afrikaans, isiXhosa and English are the predominantly spoken languages in the Western Cape (Statistics South Africa, 2014; Western Cape Census, 2011).

Despite the relative successes of the Western Cape, 35,7% of individuals in Cape Town (where this study's research sites were based) live below the poverty line (< R3,500 per month); 232,027 households are registered as indigent, and 50.2% of individuals aged ≥ 20 years do not have a matric (Grade 12 schooling) qualification. Furthermore, the Western Cape population is burdened by healthcare problems similar to those across South Africa, such as CDs (TB/HIV); NCDs (diabetes, hypertension and cancer); maternal and child health concerns; as well as interpersonal violence and road traffic accidents. Furthermore, alcohol abuse, fetal alcohol syndrome (FAS), and fetal alcohol spectrum disorder (FASD) has become synonymous with the Western Cape, where some of the highest global cases of alcohol use, FAS and FASD have been reported, especially within impoverished communities (Olivier et al., 2016).

4.4.3 Patient participation in healthcare interactions

Research has indicated that most local patients are used to more direct forms of health communication, rather than autonomous non-directive approaches due to the county's historical paternalistic health approach (Day et al., 2003; Wessels et al., 2015). A South African study by Patel & Dowse (2015), looking at information-seeking behaviour in South African isiXhosa patients, found patients to be largely passive in healthcare interactions. Patients do not ask questions, they are not provided with the opportunity to ask questions, they often have poor health literacy and do not know where to seek relevant information. A study by Keikelame and Swartz (2013), looking at patient interactions in a chronic illness clinic in Cape Town, found similar findings. Patients seldomly made eye contact with the healthcare practitioners, did not ask questions, and provided minimal responses. Similar patient behaviour has been described in other South African healthcare research undertaken in other provinces (Penn & Watermeyer, 2018; Watermeyer & Penn, 2009a; Watermeyer et al., 2020a; Wessels, 2014).

Previous international research has also described similar patient-healthcare practitioner interactions. A Dutch GC study found that patients do not ask questions or indicate when they felt left behind in the conversation (Otten et al., 2015). Other Dutch studies (Harmsen, 2003; Meeuwesen et al., 2007) further show that ethnic minority patients tend to give less verbal and non-verbal 'clues' to doctors that misunderstandings have occurred in an interaction, and an American study focusing on patient-clinician interactions for hand surgery found that patients tended to accept the healthcare practitioner as an expert (Menendez et al., 2017).

Based on these literature findings, passive patient behaviour in healthcare interactions has the potential for communication breakdown and poor patient outcomes.

4.4.4 Health literacy

Health literacy can greatly affect risk communication and can be defined as an individual's cognitive and functional ability to acquire, process, and navigate health information and services, to make suitable healthcare decisions (Institute of Medicine, 2004; Paasche-Orlow et al., 2006). Health literacy is an important factor to a person's health, with elderly individuals, poor, minority populations, those with limited English proficiency, and lower schooling educational levels being most at risk of having inadequate health literacy skills (Joseph et al., 2017; Paasche-Orlow et al., 2006). Individuals with limited health literacy not only struggle with making medical decisions but also become easily overwhelmed by health management

and the greater hospital system. Limited health literacy, together with language limitations, becomes a minefield of communication barriers in healthcare (Joseph et al., 2017; Martin & Parker, 2011).

Poor health literacy is not only confined to developing, or poor countries. Two studies conducted in the USA show that 36% of American citizens have basic (can only follow or understand basic everyday literacy tasks), or below basic literacy levels (Kutner et al., 2006; Nielsen-Bohlman et al., 2004). Health literacy usually requires more than everyday literacy skills, especially with regards to complex topics such as risk and uncertainty. Numeracy proficiency, the ability to understand numbers and statistical information, is also often taken for granted by health practitioners. Inadequate numeracy skills often lead to a lack of understanding of medical information, poor recall of information, lower participation and active engagement in health decision-making (Julian-Reynier et al., 2003; Kaphingst et al., 2016; Lillie et al., 2007; Peters et al., 2007). Collectively, these have the potential for poorer patient outcomes.

In South Africa, interactions in the public healthcare system often involve patients who have poor literacy, numeracy, and analytical skills, which together with language barriers, make mutual understanding difficult (Coovadia et al., 2009; Schlemmer & Mash, 2006; Wasserman, et al., 2010). Health practitioners may struggle in adapting and formulating appropriate explanations and language when communicating with patients (Koch-Weser et al., 2010). Patients having poor literacy skills have previously been described in South African GC literature (Morris, 2013; Shingwenyana, 2020; Solomon et al., 2012).

4.4.5 Cultural & linguistic differences

Cultural and linguistic diversity is challenging in healthcare and medical genetic services as it can affect how individuals communicate, and how they understand health (Levin, 2006; Saleh, et al., 2009; Vontress, 2003). Multicultural GC approaches aim to provide information and support in a manner that is sensitive to cultural norms and values, with genetic counsellors considering the influence of both their own and the patient's cultural and social backgrounds (Barlow-Stewart et al., 2006; Kinney et al., 2010). Although well described, Barlow-Stewart et al. (2006) points out that multicultural GC is difficult to achieve in practice, and that differing beliefs have the potential to create uncertainty and tension within interactions. Research has

also indicated that risk and disease causation amongst ethnic groups are also not understood in the same way.

A review by Machirori et al. (2018), focusing on knowledge construction of prostate cancer and genetics amongst Black men from 25 studies based in various developed and developing countries, found that participants' perceptions and knowledge of cancer is shaped by personal, societal and structural factors. The review indicates that it may be taboo to talk openly about cancer in Black male culture and having an illness or worrying about one's health and taking part in screening is considered feminine behaviour which threatens a man's masculinity and power (Akpuaka et al., 2013; Moyo, 2017; Mulugeta, 2014; Sanchez et al., 2007). Medical misunderstandings such as prostate cancer being a sexually transmitted disease and the stigma of digital rectal examinations during prostate screening being associated with homosexual practice, may further dissuade Black men from participating in cancer-related health practices, together with historical and racial mistrust of the health system (Friedman et al., 2009; Mulugeta et al., 2004; Sanchez et al., 2007).

Another study amongst Chinese Australians by Barlow-Stewart et al. (2006) found that the concept of 'inheritance' is understood differently from western culture. Instead of both parents passing on genetic characteristics, some members of the Chinese population believe that a paternal lineage passes on positive characteristics, whilst a maternal lineage is weaker and has the potential to pass on 'negative' characteristics, for example disability or disease. These different understanding can influence how genetic risk and inheritance are spoken about amongst families and in genetic specialist consultations. A study by Abad et al. (2014) amongst a Filipino population further showed that genetic birth defects are not only thought to be caused by genetic inheritance, but also by maternal behaviour and spiritual influences. Reproductive risk perception amongst communities that practice consanguinity (marriages to blood relatives) has also been examined demonstrating how genetic specialists need to be sensitive to individuals who have different practices to conventional western practices (Teeuw et al., 2014).

In South Africa, language and culture can greatly influence genetic specialist risk communication. The country has eleven official languages, different practising religions, as well as several ethnic groups and sub-groups. Immigrants from other countries also bring their own unique language and cultural identities (Crush & Williams, 2010). Cultural beliefs and practices amongst local communities also differ. Western cultures typically view illness as an

individual concern that has a bio-physical and bio-mechanism cause (known as the biomedical model), whilst African cultures view illness as a collective concern which is caused by divine interference (known as the magico-religious model) (Oosterwal, 2009).

Some cultural beliefs and practices amongst South African communities include fatalism, traditional medicine, faith healing, as well as community and family decision-making (Edwards, 2011; Kromberg & Jenkins, 1997; Morris, 2013; Solomon et al., 2012; Penn & Watermeyer, 2012a; Penn et al., 2010). Local beliefs on the causation of genetic conditions include not following correct ancestral rituals (e.g., albinism and haemophilia); intercultural relations (e.g., Down syndrome); social influences (e.g., eating particular foods or having a traumatic event during pregnancy) and that a maternal lineage can be responsible for causing disability (Morris, 2013; Penn et al., 2010; Solomon et al., 2012).

Language difficulties remain prevalent in local healthcare interactions. Previous South African research in family medicine (Schlemmer & Mash, 2006), mental health services (Kilian et al., 2015; 2020; Swartz & Kilian, 2014) and health communication (Penn, 2007; Penn & Watermeyer, 2018) have indicated that language barriers are problematic in local healthcare practice, having the potential for miscommunication, adverse health outcomes, ethical dilemmas, patient misunderstanding, healthcare practitioners concerns on whether the information is being understood, as well as a lack of rapport building.

Although Black African individuals make up the majority of the South African population, the predominant languages in healthcare are English and Afrikaans, rather than Black indigenous languages (Kilian et al., 2020). Few first language English-speakers in South Africa can converse in an African language, with research suggesting that receiving health information in English may be challenging for some patients (Levin, 2006; Penn, 2007; Wasserman et al., 2010). Furthermore, local medical genetic research has shown that patients in the public healthcare system struggle to understand genetic concepts, modes of inheritance, risk, and English scientific terms such as DNA, genes, chromosomes and cells, since no similar words exist in indigenous African languages (Christianson & Kromberg, 1996; Gillham et al., 2015; Morris, 2013; Penn et al., 2010; Shingwenyana, 2020; Solomon et al., 2012; Wessels et al., 2015).

Despite known language mismatches in local healthcare services, there remains a paucity of professionally trained healthcare interpreters, with the majority of cross-lingual conversations facilitated by family members, nurses, cleaners, hospital administration staff, security personnel and even other patients on an informal basis (Beighton et al., 2012; Kilian et al., 2020; Penn & Watermeyer, 2018; Schlemmer & Mash, 2006; Swartz & Kilian, 2014; Watermeyer, 2020). Hospital staff who can converse in a patient's first language may feel obliged to interpret for doctors and healthcare practitioners in more senior positions. Feelings of coercion may lead to resentment and poor professional relations (Penn & Watermeyer, 2018).

Poor interpretation practices bring about additional risks such as breaches in patient confidentiality, bias in information reporting, medical errors and poor patient outcomes due to misunderstanding and misinterpretation of crucial medical information. Being able to converse in a language does not imply that an individual has the necessary skills to interpret proficiently (Juckett, & Unger, 2014; Krystallidou et al., 2017). Video-recordings of South African psychiatric patient consultations involving ad-hoc interpreters further shows that despite attempting to have the patient's best interests in mind, interpreters and health professionals dominate the 'space' of the consultation through their turn-taking, questioning, gatekeeping and advocacy, leaving little room for the patient's voice (Kilian et al., 2020). Ultimately this impacts patient care.

Language barriers and the challenge of working without professional interpreters in South African GC consultations and research has also been previously highlighted (Beighton et al., 2012; Gillham et al., 2015; Shingwenyana, 2020) with suggestions made for language training and the development of South African specific GC language aids (e.g., a dictionary or language booklet) that can be used in local GC services (Shingwenyana, 2020).

4.5 Conclusion

Context and individual characteristics create several challenges in healthcare, especially during communication between healthcare practitioners and patients. This section discusses GC and context, particularly the South African context. South African GC services follow practice guidelines and training aligned with the global north; however, local contextual factors bring about unique influencers to GC training and practice. The section highlights the importance of

understanding individuals' social circumstances, backgrounds, cultures, literacy levels, and language preferences as they collectively influence how individuals understand and respond to health information and services.

CHAPTER 5: INTERACTIONAL RESEARCH

5.1 Introduction

This chapter introduces observational research and the study of naturally occurring interactions using language-in-interaction focused-methods, particularly via Conversation Analysis (CA) and Theme-Orientated Discourse Analysis (TODA). The chapter provides literature on the use of this method in understanding communication, particularly within healthcare. The chapter further provides literature on interactional research focused on risk and uncertainty in healthcare and genetic counselling.

5.2 Defining interactional research

According to Flick (2014) observational research tries to understand how activities and events occur within particular contexts, not only from an outsider's perspective but also from the participant's perspective. This method has become one of the most reliable ways of gathering evidence during interactions between individuals, as it captures data in a naturalistic way, with minimal researcher interference. The method sheds light on micro-interactional phenomena that include words, as well as phenomena that occur beyond words such as non-verbal gestures, multi-modal aspects such as the use of props and tools, and what occurs during moments of silence (Heath et al., 2010; Heritage & Maynard, 2006; Parry, et al., 2016; ten Have, 2007; Toerien, 2014) .

Language is a cornerstone of human society. Language-in-interaction, or process-focused research, sometimes described as discourse analysis, sociolinguistics, applied linguistics, linguistic anthropology and ethnomethodology is interested in observation, particularly how social activities occur and why, and how and why they matter to individuals. This type of research pays particular attention to the social activity of language, namely how it is produced, responded to and used to achieve certain goals (Heller et al., 2017). Liddicoat (2011) describes how the activity of language in interaction serves a greater purpose than just neutral conveying of information. Interactions between individuals are thus a series of performances bound by the rules and assumptions of conversational language that maintain social order, politeness and governs how individuals speak to one another. Performances in interactions can include several activities such as upholding self-image, achieving particular agendas, attaining mutual understanding as well as negotiating meaning (Chiu & Qiu, 2014; Lambert et al., 1997).

Language in interaction is also situated within context, thus social norms that govern language use may differ between various formal and informal settings as well as cultures (Burke, 2002). Observing and paying attention to the activities of naturally occurring talk provide rich opportunities to understand how language informs practices and relationships between individuals.

5.2.1 Conversation analysis

CA is a type of method that centres on the interactional activities that occur during a conversation (Sacks, 1984; ten Have, 2007; Wooffitt, 2005). Created by Harvey Sacks in the 1970s, and further developed by his colleagues, Emmanuel Schegloff and Gail Jefferson, CA follows the premise that natural interactions do not occur in a random fashion, but rather happen in a co-constructed manner in which particular social orders are followed (ten Have, 2007; Wooffitt, 2005). This may include turn-taking (who speaks); sequence organisation (how things lead to one another); turn-design (how each speaker structures their turn) and repair (how misunderstandings or mishearing are dealt with) (Schegloff, 2007; ten Have, 2007). Interactional displays and utterances such as pauses, breaths, coughs, laughter, and sounds such as 'umm', 'mmm' and 'uh,' are also considered significant (Antaki, 2011; Ellis, 2013; Goffman, 1983). Collectively, these displays serve functions such as: maintaining social order, accomplishing participant goals; demonstrating trouble in an interaction, and achieving (or attempting to achieve) intersubjectivity (mutual understanding of a shared idea) (Antaki, 2011; Ellis, 2013; Schegloff, 1992; Seale & Silverman, 1997). Focusing on these details of talk allows for a deeper understanding of how ordinary conversational activities such as greeting, questioning, advice-giving, option-listing, requesting, misunderstandings, and complaining occur (Heritage & Maynard, 2006; Toerien, 2014).

CA data is typically collected via audio and/or video-recordings and transcribed in fine detail into text (ten Have, 2007). In its purest form, CA takes on a more positive, rather than interpretive stance (Wooffitt, 2005). Founding researchers such as Schegloff (1997) argue that researchers run the risk of imposing particular assumptions onto data if analysing it with a theoretical framework in mind. For these reasons, CA rejects social identities or assumptions, unless demonstrated relevant within an interaction through participants' orientation towards it, or clear evidence that it impacts the way an interaction unfolds (Schegloff, 1997; ten Have, 2007). Thus, if not directly indicated in the interaction, broader contextual factors are

traditionally not seen to be relevant in CA (Waring et al., 2012). Rather, meaning and context are created and shaped directly by the actual sequences of talk (Heritage, 2005).

Although acknowledging that analysis of discourse can at times lead to unwarranted theoretical claims with little proof in the data, some researchers have argued that CA's traditional idea of 'context' may be too narrow (Waring et al., 2012; Wetherell, 1998). Broader social discourses and factors within a particular setting are important influences on how people orientate and display themselves within talk: e.g., who talks first, the title of address, and who has more authority (Cicourel, 1981; Wetherell, 1998). This may include areas such as doctor-patient consultations, emergency services, education, courtrooms, and police interviews where talk and activities are sensitive to the formalities of the setting and are also influenced by broader institutionalised factors (Antaki et al., 2003). Street (2003) highlights that institutionally bound interactions, such as medical consultations are embedded into broader contexts whose features influence individuals' cognitive, emotional, and behavioural responses and how they produce and respond to moments of talk. The broader context is thus a further important consideration when researching communicative processes in these types of settings. For studies using principles of CA in these areas, acknowledgement of these factors may be important (Penn & Watermeyer, 2018; T. Koole, personal communication, February 2019).

5.2.2 Theme-orientated discourse analysis

Theme orientated discourse analysis (TODA) is another type of interactional-focused analysis approach developed by Roberts and Sarangi (2005). It draws from sociological and linguistic frameworks and is considered a type of "free-range discourse analysis, drawing inspiration from many approaches" (Roberts & Sarangi, 2005, p. 639). It pays particular attention to language use within professional practice, most notably in medical settings.

TODA focuses on 1) focal themes and 2) analytical themes. Focal themes relate to broader focal topics such as decision-making, trust, neutrality, and patient-centredness. Analytical themes focus on micro-level features within moments of talk, e.g., how information is framed, and how footing (the way individuals align or negotiate their identities and positions during interaction) or face-work (politeness strategies, claims of knowledge and managing fragile moments) activities occur. Analytical themes also pay attention to rhetoric (persuasive talk) as well as context and social identity (how particular utterances have a meaning which is dependent on the surrounding detail and context of talk) (Brown & Levinson, 1987; Goffman,

1983; Gumperz, 1992; Roberts & Sarangi, 2005; Sarangi, 2010; Thomassen & Sarangi, 2012). Linguistic repertoires e.g., metaphors or figures of speech may also become important in discourse-based methods (Wooffitt, 2005) such as TODA. Through the use of analytic themes, TODA can demonstrate how interactions are organised moment-by-moment, and how sense-making, negotiation and goal setting occur between individuals via implicit or explicit cues.

Although mainly focused on talk-in-interaction, TODA also makes use of additional "ethnographic knowledge" (Roberts & Sarangi, 2005, p. 632) collected in the form of interviews, informal conversations, and observations of the research setting. These are used to understand broader contextual and organisational structures in which the interactional data is embedded.

5.3 Studying patient-healthcare practitioner consultations using interactional methods

Patient-healthcare practitioner interactions lie at the heart of medical care. How communication occurs between these two groups of individuals is thought to impact patients' adherence to medicine, assimilation of knowledge, and satisfaction with service delivery. Furthermore, communication assists healthcare practitioners to accurately diagnose a patient's concern and facilitate in knowing what a patient's needs are, both physically and emotionally. Despite the recognised importance of patient-healthcare practitioner consultations, studying and measuring the effectiveness of communication in these encounters is difficult (Drew et al., 2001).

Observation of everyday patient-healthcare practitioner consultations using interactional methods has become one of the most reliable ways of gathering empirical evidence in medical interactions. This approach has been used both in local (Penn & Watermeyer, 2018; Watermeyer, 2008; 2020; Wessels, 2014) and international (Drew et al., 2001; Heritage & Maynard, 2006; Mahmoodi, 2015; Pilnick & Zayts, 2014; Sarangi & Clarke, 2002a; Sarangi & Clarke, 2002b; Thomassen & Sarangi, 2012; Toerien et al., 2013, Zayts-Spence et al., 2021) research to understand how communication occurs within a variety of healthcare fields.

Language-in-interaction methods such as CA and TODA can elucidate social, moral and technical dilemmas that healthcare practitioners and patients face during moments of talk in healthcare consultations. Furthermore, these methods can focus on the micro aspects of talk

such as power dynamics, mutual understanding, conflict, misalignment in agenda, mitigation and neutrality are enacted and achieved during a conversation (Sarangi & Clarke, 2002a; Heritage & Maynard, 2006). Interactional methods are useful in understanding activities during practitioner-patient activities such as breaking bad news (Maynard & Frankel, 2006), negotiating treatment decisions (Stivers, 2006; Toerien & Jackson, 2019), establishing patient agendas (Wessels, 2014), closing conversations with patients (West, 2006) and checking patient understanding (Watermeyer & Penn, 2009a; 2009b). Furthermore, contextual aspects and non-verbal displays can also be studied using interactional approaches to understand how they impact practitioner-patient relationships (Heath, 2006; Roberts & Sarangi, 2005). Toerien and Jackson (2019) demonstrate how interactional methods can highlight, both implicitly and explicitly, the complexities and tensions of patients' and practitioners' psychosocial and biomedical perspectives during medical negotiations and decision-making.

Drew et al. (2001) and Pilnick et al. (2018) further emphasise that interactional methods provide real-time evidence that has practical value for healthcare practitioner training. Not only does studying patterns of interaction provide reflexive opportunities on communicative behaviour and understanding the outcomes of different communication styles, but it also allows for the identification of interactional features and activities that either promote or undermine, patient-centred care.

CA and TODA have been applied across a range of medical fields and interactions. CA has revealed how trouble in the initial greeting and opening stage of a call to an emergency centre can impact the time taken for an ambulance to be dispatched to the person in need (Penn et al., 2007). Other CA studies conducted in the UK on neurology consultations (Toerien et al., 2013), breast cancer (Mahmoodi, 2015), and neonatal critical care (Shaw et al., 2016) have demonstrated how word choices and ways in which information is conveyed by doctors can impact a patient's or family's decision-making regarding uptake of treatment. TODA has been used to study how genetic counsellors use a non-directive stance to perform certain activities during patient decision-making (Arribas-Ayllon & Sarangi, 2014); and how shared understanding in medical interactions is navigated (Roberts & Sarangi, 2005). Furthermore, research making use of principles of CA and TODA has elucidated health communication practices in patient-pharmacy interactions (Watermeyer, 2008) and the interactional dynamics of genetic counselling consultations (Wessels, 2014). Collectively these studies show the benefit and practical potential of CA and TODA methods.

There is also a growing body of literature emphasising the value of linguistic-orientated research methodologies in medical genetic and GC interactions (Lehtinen, 2013; Pilnick, 2002; Pilnick & Zayts, 2014; Sarangi & Clarke, 2002a; Thomassen Hammerstad et al., 2020; Wessels et al., 2015; Yelei et al., 2013, Zayts-Spence et al., 2021). This type of research may provide opportunities for training and practice modifications, as it yields data that can showcase communication styles, formats and approaches in a real-time sense, allowing for intimate reflection on the process of communication and counselling (Biesecker, 2018; McAllister et al., 2016; Pilnick et al., 2018; Wessels et al., 2015; Yelei et al., 2013, Zayts-Spence et al., 2021).

5.4 Interactional methods to study risk & uncertainty in genetic counselling

Risk communication and uncertainty have been previously studied in medical genetics and GC as described in Chapter Three. Although not as prominent as other research methods, interactional research to understand risk and uncertainty in GC consultations, also exists. This type of research can be seen as process-focused, rather than in-put or out-put focused GC research as described by Paul et al (2015). Process-focused GC research pays attention to the content of GC consultations, how professionals are meeting client needs, what communication techniques are used, and what the challenges and tensions in professional issues are. Process-focused research is also able to highlight the influence of different context and socio-cultural backgrounds of patients and practitioners on GC theoretical ideas and the subsequent outcomes (Paul et al., 2015).

GC risk communication has been investigated in several interactional based studies, many of which have overlapped with focuses on uncertainty and how it impacts GC risk communication (Pilnick & Zayts, 2014; Sarangi & Clarke, 2002a; Thomassen & Sarangi, 2012). Uncertainty has been investigated in prenatal GC risk communication (Pilnick & Zayts, 2012; 2014), delivery of uncertain genomic test results to families (Stivers & Timmermans, 2016; (Thomassen Hammerstad et al., 2020), cancer risk discussion (Thomassen & Sarangi, 2012), discussions on a lack of diagnosis in paediatric and adult GC consultations (Brookes-Howell, 2006), and GC paediatric discussions on disability (Steel et al., 2014).

Sarangi (2000), using a discourse-based analytic approach, demonstrated that GC is an information-rich activity that involves genetic counsellors using several different formats to convey information in a structured and deliberate manner. The study concludes that GC uses hyper-explanations of risk information and that these actions may be attempts by genetic counsellors to ensure that patients are informed, and that the genetic counsellor remains within the neutral boundaries of their profession when facilitating patient decision-making. Pilnick (2002) studied audio-recorded UK-based GC consultations using CA and noted that several interactional challenges occurred between patients and genetic counsellors. Particularly difficult parts of the interaction were knowing what information was relevant for a patient and managing varying risk perceptions of different family members who accompany patients to GC consultations.

Topics on risk are easier to identify, however, identifying topics, or moments of uncertainty in interaction is more challenging. Previous researchers using interactional methods to study uncertainty have however made helpful suggestions on how topics or moments of uncertainty can be recognised. Sarangi and Clarke (2002a) suggest focusing on discourse strategies and disclaimers that individuals use during an interaction, either based on what has been said, or what is about to be said. Discourse strategies and disclaimers include hedging (I may be wrong, I think, I guess etc.), using auxiliaries (could, might, maybe etc.), adverbs (the risk is approximately/about, 50/50 etc.), as well using verbal markers to identify professional boundaries and expertise (I am not a neurosurgeon). Pilnick and Zayts (2014), in their study on uncertainty in prenatal GC consultations for pregnant women identified as high-risk based on their screening results, further indicate that uncertainty is also displayed and introduced in GC risk discussions by a speaker when they produce hitches, word formulation disruptions or restarts during their sentence construction. Similar dysfluency markers have also been indicated to represent uncertainty in a paediatric GC study by Steel et al. (2014). These paralinguistic features, together with others, such as pauses, fillers such as ‘um’ and ‘uh’ as well asymmetry in prosodic contour (e.g., speech rhythm, rate, pitch as well as rising and falling intonation), are further indicators for displays of uncertainty in interaction (Barr, 2001).

How uncertainty is managed interactionally has also been studied in different GC settings. Stivers and Timmermans (2016) used CA-inspired methods to analyse 44 video-recorded GC consultations from the USA where medical geneticists delivered genomic test results to families. Their results showed that in instances where ambiguous test results were conveyed,

medical geneticists went beyond what was indicated on the test report, and reinterpreted the information, typically to reduce the level of uncertainty. The study concludes that by managing the boundaries of uncertainty for families, medical geneticists re-enact a care relationship (further described in *Chapter Six*). A recent paper using TODA to examine Norwegian interactional GC data described how genomic knowledge for colorectal cancer creates several uncertainties during GC consultations, especially when interpreting complex genomic results. Decisions on what information to share, and how to share the information has the potential for professional and family ethical tensions. These tensions need to be managed and negotiated during GC (Thomassen Hammerstad et al., 2020). A discourse-based GC study by Brookes-Howell (2006) in the UK shows how uncertainty is used by parents and genetic counsellors to increase or decrease the likelihood of finding a diagnosis. Unlike other health professions, uncertainty in GC also allows parents to provide diagnostic insight and predictions, thus conversations about uncertainty and diagnosis become a negotiated activity between parents and genetic counsellors when there are ambiguous results or diagnoses. Another discourse-based study by Steel et al. (2014) in Australia shows how genetic specialists show displays of communicative uncertainty when discussing dysmorphic children's features with parents and how this may impact rapport during the GC relationship.

Context has also been considered in GC interactional research. A study by Pilnick and Zayts (2019), investigating the impact of the presence or absence of a shared first language in prenatal clinics in Hong Kong, found that when healthcare practitioners and patients were mismatched regarding language, patients struggled to engage in the decision-making phase of the consult. An earlier study by Pilnick and Zayts' (2012) further suggests that a patient's socio-economic circumstances and health practitioners' perceptions of the patient's ability to look after a child with a disability, influences a health practitioner's direct and non-direct communication with the patient when offering them invasive prenatal testing.

In South Africa, little interactional research exists for GC. A local Master's level study by Perrott (2010) focusing on risk communication in advanced maternal age (AMA) GC consultations in Cape Town made use of a mixed-method approach. Methods included genetic counsellor interviews (n = 3), audio-recorded genetic counselling consultations (n =13), and post-consultation patient interviews (n=12). Analysis of the audio-recorded consultation transcripts did not involve a true interactional analysis approach, but rather a layered approach using thematic analysis (to identify what was communicated and how), and content analysis

(which involved using a quantitative grading scale to count the number of times particular descriptions of risk occurred across the recorded consultations) was utilised. Themes from the consultation data were used to compare against the themes identified in the genetic counsellor and patient interview data. Findings from the study revealed that although patients indicated satisfaction with the service, there were several interpretations between genetic specialists and patients on what 'risk' means with specialists viewing risk analytically, whilst patients viewed risk experientially. Formats used to convey risk included numerical, verbal and graphical and patients struggled to make sense of their risk as they had little expectation or understanding as to why they have been referred to GC or why they were considered to be at risk. Contextual challenges noted to impact GC risk communication included poor communication from referring healthcare practitioners with patients, language barriers and the importance of collective family decision-making on a patient's risk, rather than independent decision-making that was expected to occur in a short period of time during a GC consultation.

Wessels' (2014) PhD study also focused on AMA GC services, but in Johannesburg in this instance. The study focused on the process of communication across whole GC consultations. Thus, although aspects of risk and uncertainty were described, the actual focus was not on these topics specifically. The study approach included 17 video-recorded prenatal GC consultations, researcher observations of the research setting, 12 patient post-consultation interviews, and 14 post-consultation GC interviews. Analysis of the interactional data included principles of CA and TODA, together with considerations from intercultural studies. The study found that South African GC consultations in Johannesburg are largely divided into six phases: opening, information giving, information gathering, decision-making, counselling, and closing. Findings further indicated tensions in the genetic counsellors' expectations to both educate and counsel patients, and that genetic counsellors must integrate complex risk information with a patient's socio-economic disparities, literacy levels, personal needs, and prior perceptions. Genetic counsellors dominated the amount of talk and led much of the consultations through their own, rather than the patients' agendas. Patients were largely passive, and many did not actively engage in the decision-making as suggested by western GC models. Recommendations included the development of a contextual model for GC which considers the social and political context of particular settings since they impact upon how patients engage with genetic counsellors during GC consultations, particularly in the decision-making phases of the service.

Wessels has since expanded on the interpretation of these findings in recent papers. Wessels et al. (2015) describe how decision-making in GC is complex and that patients' responses and orientations towards risk information and the offer of invasive prenatal testing are influenced by historical, political and socio-economic factors. These factors may impact a patient's ability to perceive themselves as autonomous decision-makers in healthcare, rather viewing the genetic counsellor as having decision-making authority. Thus, even if unintended, a genetic counsellor's offer of invasive testing may be seen as implicitly directive communication that invasive testing should be accepted. Wessels and Koole (2019) have further suggested that *how* risk information is presented is less important than the *meaning* of the risk information for a patient. The paper suggests that further interactional exploration is needed in GC to understand whether counselling or education is prioritised by patients and how genetic counsellors choose to pursue either counselling or education as suggested by Kessler and Antley (1981) and Kessler et al. (1982).

5.5 Conclusion

In conclusion, the chapter has described interactional research methods, and their value in health communication research, particularly for risk and uncertainty research in GC. Collectively, risk and uncertainty play important roles in GC communication. Not only does communication affect how risk is spoken about, but it also affects the focus and experience of uncertainty (Stivers & Timmermans, 2016). Further research on risk communication and the management and negotiation of uncertainty in GC in socio-cultural diverse settings, such as South Africa is needed.

CHAPTER 6: THEORETICAL FRAMEWORK

6.1 Introduction

As GC borrows from many different models, frameworks and areas, I have chosen not to focus on a singular theory to support the arguments made in this thesis, but rather to draw from different existing theories to understand the notion of risk, uncertainty and care within a South African GC context. The theories predominantly borrowed from include: Patient-Centred Care, the voice of medicine and voice of the lifeworld (Mishler, 1984), Uncertainty Management Theory (Brashers, 2001; 2007), Anxiety/Uncertainty Management (AUM) Theory (Gudykunst, 1995) and Ethic(s) of Care (Tronto, 1993; 2010).

6.2 Patient-centred care

Modern healthcare practice has become inherently focused on patient-centred care (PCC). The terms person-, client and patient-centred care as well as patient-centredness are used across health literature and although no single definition exists, the concept is generally understood as a healthcare approach that pays attention to the individual needs, background and preferences of a patient, as well as how psychosocial factors impact on healthcare delivery (Epstein et al., 2005; Lambert et al., 1997). PCC pays attention to elements of humanity, empathy, and ensuring shared power and responsibility in decision-making (Hong & Oh, 2020; Ogden et al., 2017), it focuses on the relationship between patient and healthcare practitioners, it encourages patient participation in healthcare decision-making, and it is aware of context (Kitson et al., 2013). PCC is thus not a "technology-centred, doctor-centred and hospital-centred" approach (Stewart, 2001, p. 444).

Although the term PCC has been used theoretically since the 1950s, and health practitioners, policymakers, health educators have been aware of factors such as literacy, language, ethnicity, age, social status and gender impacting health outcomes; how they do so in actual clinical practice is poorly understood (Epstein et al., 2005; Ishikawa et al., 2013; Ogden et al., 2017). Part of the challenge of defining what PCC is, is that the essence of such care is difficult to capture in practice. Thus, much of the idea of patient-centred care exists purely in classroom teaching (Ogden et al., 2017; Stewart, 2001), with "many evangelists but few practitioners" (Hawkes, 2015, p.757).

Although PCC is broadly defined, it's interpretation may vary amongst individuals or settings. A health practitioner may foreground particular information, procedures and/or activities during a healthcare consultation, deeming them essential or beneficial, however a patient may not share the same sentiments. Delivering PCC in practice may also be hampered by entrenched institutional and historical medical practices which make it difficult for individual health practitioners or patients to alter or shape the health services that they are involved in or utilise (Richards et al. 2015a; Stewart, 2001). PCC may also look different in different contexts, and elements of what makes up PCC are not always tangible, easily measurable or concretely observable (Langewitz, 2007; Ogden et al., 2017). There is thus not only a need to gain better insight to what patients' value in healthcare consultations, but greater collaboration between healthcare practitioners and patients is necessary in health policy making to elucidate what PCC means (Stewart, 2001; Richards et al., 2015a). Empirical-based research across different healthcare settings is also necessary to understand the meaning of PCC in different contexts (Ogden et al., 2017).

The foundations of GC lie within patient-centred care, borrowing from Roger's Person-Centred Theory (Biesecker et al., 2019; Veach et al., 2003). Although a person-centred approach is mentioned as an 'ideal' GC consultation where a patient's values and needs are prioritised (Ormond, 2013; Redlinger-Grosse et al., 2020), Biesecker et al. (2019) argues that despite GC guidelines having teaching, counselling and advocacy as central tenets to GC practice, much of these guidelines have been developed without context, or without evidence-based knowledge. Understanding what patients' needs are and how they respond to actual GC practice is not well understood in different settings, thus there is a paucity of knowledge regarding what PCC means within GC practice (Biesecker et al., 2019). Focusing on the communication between patients and healthcare practitioners may be one way to gain insight into how patient care is enacted and how the process is influenced by surrounding contextual details (Lambert et al., 1997; Street, 2003).

6.2.1 Patient-centred care & trust

A significant element in PCC is trust. Trust in the patient-healthcare practitioner relationship, as well as trust in the broader healthcare system, is thought to produce higher levels of patient satisfaction, it improves continuity of care, it promotes a patient's ability to adjust and cope with an illness diagnosis and it helps to alleviate negative patient emotions such as anxiety and fear (Armstrong, 2018; Street et al., 2009). The importance of developing a trusting

relationship in GC interactions is described in GC guidelines and literature (Smets et al., 2007; Thain et al., 2019; Veach et al., 2007; Weil, 2001).

The communication process between a patient and a healthcare practitioner is particularly important in establishing relational trust, however, can be difficult due to patient-healthcare practitioner asymmetries in knowledge. A patient typically does not have the same degree of medical or scientific knowledge as a healthcare practitioner, whilst alternatively, a healthcare practitioner does not have the same insight as the patient regarding their personal symptoms, and family and social backgrounds (Fritz & Holton, 2019). Due to these asymmetries in knowledge between a healthcare practitioner and a patient, a great deal of the success of a health consultation is placed on the ability to trust one another. During the communication process, how a healthcare practitioner conveys information, whether they are able to explain complex knowledge in an understandable manner, whether the information imparted is relevant to the patient's concerns, and whether the patient has felt heard and acknowledged in the consultation contributes to the establishment of trust (Hong & Oh, 2020; Street et al., 2009). The healthcare practitioner also has to trust the accuracy of information provided by the patient and has to trust that the patient is able to make informed health-related decisions (Arribas-Ayllon & Sarangi, 2014; Fritz & Holton, 2019; Veach et al., 2007).

Trust is also an important factor when it comes to uncertainty. In medical consultations that are particularly uncertain (for example in prenatal GC consultations where serious decisions have to be made with unclear information) trust can be particularly beneficial in helping patients and healthcare practitioners cope and adjust to the situation (Fritz & Holton, 2019). Uncertainty has the potential to create anxiety and discomfort in patients. Despite a healthcare practitioner's hesitance to admit uncertainty to a patient, talking about the uncertainty and acknowledging its presence has been argued to enhance trust and rapport, rather than create a breakdown in the relationship (Armstrong, 2018). These discussions are thought important for enhancing patient care.

Despite the claim of trust being important in the patient-healthcare practitioner relationship, trust has received little attention in health research and practice (Chang et al., 2013; Hong & Oh, 2020; Pearson & Raeke, 2000; Smets et al., 2007). Greater insight is needed into what factors impact trust in GC consultations, particularly uncertainty, and how this may impact care.

6.3 The voice of medicine & the voice of the lifeworld

When considering how health practitioners and patients approach healthcare interactions, Mishler (1984) describes it as a clash between the voice of the lifeworld and the voice of medicine. Borrowing from the Theory of Communicative Action (Habermas, 1984), the voice of the lifeworld can be explained as a patient's everyday life experiences that shape how they view the world, whilst the voice of medicine can be described as a healthcare practitioner's focus on biomedical, scientific and technical aspects that are devoid of particular personal or social perspectives (Barry et al., 2001; Mishler, 1984).

Much of academic health literature and 'biomedical' information is focused on western medicine practices and philosophies. However, in varying global contexts, such as South Africa and the Middle East, alternative and traditional healing practices may also be utilised in pluralistic manners (Germond & Cochrane, 2010; Kale, 1995). Many of these alternative health practices are thought more sensitive to lifeworld and contextual concepts than western medicine practices (Barry et al., 2001; Busby, 1999). In western medicine practices, power relations exist where conversations are typically controlled by the healthcare practitioner who initiates questions, develops the topic of the talk, and chooses whether to address psychosocial concerns or not (Barry et al., 2001; Germond & Cochrane, 2010; Mishler, 1984). In this hierarchical, medically focused manner, there is potential for the patient's concerns to not be fully explored and the patient may not have the opportunity to make sense of their situation and what the information may mean for them personally (Barry et al., 2001).

The idea of health and healing differs according to different people and contexts, which in turn influences individuals' health-seeking behaviour and actions. Furthermore, understandings of health are also not static, but rather evolve and are shaped by place, time, society and circumstance (Germond & Cochrane, 2010). How a patient responds to healthcare services and the information received is dependent on the patient's particular context at that particular point in their lives (Barry et al., 2001).

Germond and Cochrane (2010) further describe how individuals have 'health worlds' that exist within their lifeworld. Health worlds and lifeworlds are thought to be characterised by two important notions: Both are *communicative*, in that communication between individuals plays an important role in how the two worlds are shaped, and they are both *relational* in that health

impacts not only the self but also others (Germond & Cochrane, 2010; Germond & Malapo, 2006). Understanding a person's lifeworld and constituent health worlds requires an understanding of a person as a whole. Germond and Cochrane (2010) describe understanding an individual's body as not only constituting the physical aspect that needs to be assessed and medically managed but how the body is also a representation of an individual's emotions, spirit, religion, culture, community, family, children, childhood, environment, and space.

Although it is acknowledged that a healthcare practitioner may never be able to appreciate the fullness of a patient's lifeworld (Germond & Cochrane, 2010), keeping in mind the different lifeworld perspectives of patients and healthcare practitioners can assist in understanding the dynamics of healthcare communication (Barry et al., 2001; Toerien & Jackson, 2019). Furthermore, whilst tensions can exist in health practitioner-patient interactions, they can also be important opportunities for mutual learning. For the patient it may be gaining scientific or health knowledge, and for the healthcare practitioner it may be better understanding of a patient and seeing them as a full and "embodied person" (Germond & Cochrane, 2010, p. 315). Collectively, this mutually sensitive, and active attempt of understanding between a healthcare practitioner and patient can lead to better health outcomes and ultimately improved patient care.

6.4 Uncertainty Management Theory & Anxiety/Uncertainty Management Theory

Uncertainty Management Theory (UMT) (Brashers, 2001; 2007) may have relevance within GC consultations since this theory is interested in how uncertainty impacts communication between individuals, especially during health interactions and decision-making (Brashers, 2007; Redmond, 2015). Genetic information often includes great amounts of uncertainty due to the nature of the conditions, unknown risks, limitations in testing and challenges in interpreting genetic test results. Managing uncertainty during interactions may thus be an important part of ensuring positive patient outcomes (Epstein & Street, 2007; Bylund et al., 2012; Dean, 2016; Hammond et al., 2020; Rauscher, 2017) and by extension enhancing patient care.

UMT is concerned with the relational management of uncertainty during communication and how uncertainty is either maintained to provide hope and optimism (Brashers, 2000; Dean, 2016; Khan et al., 2016; Stivers & Timmermans, 2016; Truitt et al., 2012) or reduced to enable

confident decision-making, to plan, or to predict the actions of others (Brashers, 2003; Hong, 2020). Brashers and Hogan (2013) describe how uncertainty is not simply about information retrieval, but that it is also a complex phenomenon that affects how information is conveyed and pursued by individuals. Individuals may display emotions such as anxiety and distress in response to uncertain scenarios and may behave in different ways, such as actively seeking out or choosing not to seek out informational resources when faced with uncertainty (Brashers, 2007; Brashers & Hogan, 2013; Hong et al., 2020). Hogan and Brashers (2015) further describe individual behaviours around uncertainty including information acquisition, information handling and information use. During uncertain scenarios, individuals either actively (intentionally) or passively (unintentionally) gain knowledge that alters the extent of the uncertainty. Individuals also differ in how that information is retained and organised, and how they act on that information, both emotively, and in their activities thereafter. If, when, how and why they respond to uncertainty and the related information they receive is contextually situated for different individuals. Uncertain situations thus have the potential to impact the rapport and relationship between practitioners and patients, and ultimately care outcomes.

Gudykunst's (1995) Anxiety/Uncertainty Management Theory (AUM) Theory is another useful theoretical framework for managing uncertainty in communication. AUM Theory aims to enhance meaning-making during the exchange of information between individuals by encouraging individuals to be mindful of their own, and the other participating individual's behaviours and ways of conveying information. Thus, attention to the process of information exchange is imperative in reducing uncertainty and anxiety (Gudykunst, 1995; Yoshitake, 2002). Furthermore, researchers have suggested that part of paying attention to the process of managing uncertainty, is identifying the sources of uncertainty that impact communication behaviours (Babrow & Kline, 2000; Brashers, 2007; Mishel, 1990). If sources are identified, individuals may better adapt to the presence of uncertainty and may make greater concerted efforts to establish common ground and connection with one another due to their awareness of the potential for miscommunication and relational breakdowns (Gudykunst & Nishida, 2001; Daniels & Swartz, 2007).

In diverse contexts, it may be particularly important to extend the identification of uncertainty sources not only to the information within the consultation but also to consider sources that stem from the surrounding context that may create additional interactional uncertainties during communication between individuals. Contextual sources of uncertainty may include patient

behaviours, as well as language diversity, literacy barriers and cultural differences that may impact how patients receive and respond to health information as shown in previous GC studies (Barlow-Stewart et al., 2006; Cheng et al., 2018). Paying attention to and identifying uncertainties stemming from complex genetic risk information as well as surrounding contextual features may create challenges. Research focusing on uncertainty in health interactions is thus imperative to better understand how uncertainty may impact patient care.

6.5 Ethic(s) of care

The ethic, or ethics, of care is thought to be based on the notions 1) that individuals have personal rights, 2) that morals are developed both intrinsically and shaped by the societies we live in and 3) that human existence is dependent on interpersonal relationships. Individuals thus have a responsibility to act morally through their relationships with others, to respect individual rights, and to achieve justice and fairness (Chuwa, 2012; Gilligan, 1993; Kohlberg, 1958). The theory highlights that individuals' behaviours, decisions and personal circumstances are not only unique but layered with complexity. Due to these complexities, care for one individual, may not be the same for another (Noddings, 1986). Care is thus paying attention to a particular individual's context and situation and finding a moral resolution that fits into their reality. This individually tailored approach to care has been further supported by academics and health researchers (Fisher & Tronto, 1990; Held, 2005; Tronto, 1993). Care is also a physical action, not only an attitude. Caring is demonstrated through one's behaviour and action in response to another individual's concern or situation (Chuwa, 2012; Tronto, 1993). Ethics of Care fits in well with GC principles as it focuses on the relationship between two individuals, something that is valued within GC interactions (Schmerler, 2006).

6.5.1 Ethics of care & the caring institution

Tronto (2010) also extends the idea of care to institutions and healthcare services. She suggests that to truly evaluate whether a service is considered caring, one should consider alternative measures, other than the market- and consumer-related approaches (for example patient satisfaction surveys). Rather, attention to purpose, power and particularity is encouraged. This includes a focus on the *complexity* and *process* of care within healthcare institutions and patient-healthcare practitioner interactions. Here, the needs, actions and experiences of *both* patients and healthcare practitioners must be considered. Furthermore, to fully understand what 'care' is, Tronto (2010) suggests that attention to the following areas is needed: the goals of a

health service; inter-personal and broader institutional power relations, as well as the context and diversity of human individuality and experiences. To better understand these identified areas, researching clinical practices within institutions to make explicit the types of interactions and communicative processes is necessary. Placing a lens onto patient-healthcare practitioner 'talk' in interaction helps us to understand how the notion of care is being achieved, and what revisions are needed within institutions to meet the goals of healthcare practices, as well as the individual needs of patients and healthcare practitioners.

Tronto's idea of care is further supported by other health communication researchers. Thorne and Paterson (2001) argue that what is considered 'care and support' is not universal in all contexts for all patients. Furthermore, the supportive needs of a patient may also change over time. Relying on only one global theoretical approach regarding what is believed to be a kind, caring and supportive healthcare service or interaction is unrealistic. Thus, considering and admitting alternative ways of practice through reflexivity and paying attention to the daily realities of practice within a particular context becomes important in developing a caring institution or service. Tronto (2010) argues that through deliberate action, clear goals, and an awareness of context, healthcare institutions will be able to meet the needs of the people they intend to serve. This can be considered as true and authentic care.

6.5.2 Ethics of care: Ubuntu & understanding illness from a South African perspective

Ethics of care within a South African context can be thought of using the philosophy of '*Ubuntu*'. *Ubuntu* is an African philosophy that draws from the isiXhosa expression: "Umntu ngumntu ngabanye abantu" (Ntizimira et al., 2016, p. 277) which translates loosely to "I am who I am because you are who you are, and human beings are human because of the otherness of other human beings" (Chuwa, 2012, p.163). This philosophy of Ubuntu is rooted in an ethical code of conduct that one should act and live in a manner that is not only personally good but is considered good for those around us. It is thus about the power of relationships, respecting each other's dignity and achieving common wellbeing and harmony (Nussbaum, 2003).

GC risk discussions are based on the individual patient's health, as well as the health of extended family members since risk estimates in GC also take into consideration mendelian, empirical and pedigree data. Discussions on risk are thus not only aimed at the individual attending a GC consultation but often involve risks relating to extended family members,

children, and future pregnancies (Mendes et al., 2018). The 'narrow' personal and 'broader' family perspectives may be impacted by context and how an individual patient may view their ability to make decisions. In African healthcare services, individuals may view a decision as a family and community collective rather than in line with the western model that individuals should be able to make independent decisions (Berg, 2003; Ntizimira, et al., 2016; Sambala et al., 2020). Although *Ubuntu* respects individualism, decisions are framed in a way that benefits the self and benefits others. Hence, western ideas of individual rights, moral reasoning and care may not always be aligned in local African healthcare settings (Sambala et al., 2020). *Ubuntu* may thus assist in understanding patient decision-making and perceptions of care within GC risk discussions and decision-making.

In socially complex settings such as South Africa, the doctor-patient relationship may come with additional responsibility due to the power of the doctor and the vulnerability of the patient stemming from socio-economic and political variables. Here *Ubuntu* becomes important, to ensure positive outcomes, as well as to encourage a trusting partnership within the relationship (Green-Thompson et al., 2017). Tronto's (2010) idea that care is a relational concern, and that in caring for ourselves, we care for others, fits in well with the philosophy of *Ubuntu* (Chuwa, 2012).

Patient care in a South African GC context may thus require finding a balance between the western and African understanding of health and illness. Furthermore, the concept of cultural safety also needs to be foregrounded in South African GC practice. Being culturally safe is acknowledging that each patient is unique and that they should be cared for in a manner that aligns and supports their background, their values, their needs and what they stand for as individuals (Clear, 2008). Cultural safety also requires health practitioners to be aware of their own biases and contextual lenses, and how these may impact their interaction with patients whose backgrounds and values are different to their own (Penn & Watermeyer, 2018). Developing communicative approaches within local GC services that are sensitive to the psychosocial needs, cultural values and backgrounds of patients, together with the African philosophies followed in South Africa, is important and further exploration is needed.

6.6 Conclusion

In summary, the chosen theoretical framework is well suited to the focus of this study. By investigating the interactional processes and dynamics that occur between genetic specialists and patients whilst risk and uncertainty information is conveyed and negotiated is an important step in reviewing care within a South African GC context. By better understanding the goals, processes and challenges that occur, not only for patients but also for genetic specialists, practice and training guidelines can be enhanced to ensure that care remains a central focus within local GC services. Care in a South African context may look different to how western theories and guidelines may define care. Creating South African specific tailored GC approaches and theories are imperative to ensure that our institutions and services do indeed provide authentic care and support to the local patients and families who interact with them. Part of creating these tailored practice and training guidelines is to explore what happens within GC interactions, not only to rely on reported experiences. Verbal and non-verbal displays, even when subtle, may impact communication, and by extension care. If GC views itself as a caring service, the activities within GC consultations, including risk and uncertainty communication, need to show as much. Interactional research focusing on the real-time communicative processes may assist in providing such evidence and creating a base for discussion and review. Research is thus needed to understand how care is currently being provided, to enhance South African specific GC communication guidelines and practice.

CHAPTER 7: ESTABLISHING A NICHE FOCUS

7. 1 Introduction

The following chapter summarises some of the current gaps in genetic risk and uncertainty communication in socio-cultural diverse settings. Suggestions to address this paucity of knowledge are made, and an explanation of how this study may contribute to furthering knowledge in this area is offered.

7.2 The need for evidence-based genetic counselling practice guidelines using interactional research methods

There lacks a focus on actual GC consultations to understand how communication processes and features impact GC outcomes. Despite many of the models and theories positively contributing to the field of GC, what has remained largely problematic is that many of these models are informed by theory, rather than by empirical evidence. The heterogeneity of these GC models and principles has led to the GC process being described as an unknown 'black box' (Biesecker & Peters, 2001) with *the* ideal model for GC remaining elusive (Austin et al., 2014; Biesecker et al., 2019; Cragun & Zierhut, 2018; Veach et al., 2003; Weil, 2000). Despite outcome-based studies providing knowledge on whether GC in-put goals and ideals has been met or not, how these goals or ideals are achieved/not achieved in practice remains a poorly understood area (Paul et al., 2015).

There remains little consensus within the GC community on the goals of GC. GC is impacted and shaped by the various contexts in which it is now practised, as well as by the advancements in science and technology. These changes and advances may create tensions regarding historical definitions and outcomes of what GC is and how it should be conducted in the present day. The 'ideal' model may not be a one-size-fits-all for all contexts. Therefore, the goals, delivery and models of GC should be informed and adapted to the setting in which it is practised, as well as needs of the population it serves (Resta, 2019). The goals of GC thus shift according to where and when GC occurs, the patient, and their agendas. Hence, GC in actual everyday interactions may not provide/support the evidence-based guidance on how to achieve these goals in various settings as suggested by the traditional theoretical models used (Biesecker et al., 2019; McAllister et al., 2016; Redlinger-Grosse et al., 2017; Resta, 2019). This is not a new argument. Seymour Kessler (1999) , an American psychologist and Director

of the genetic counselling programme at the University of California, Berkley, argued as far back as the 1970s that "the paucity of process studies hinders us from finding ways to make the educational and counselling aspects of genetic counselling more effective" (p. 342). The genetic counsellor/genetic specialist's own goals and agendas may also impact the idea of 'success' in GC consultations outcomes. Thus, success in GC does not appear fixed or easily defined.

Part of understanding GC outcomes is to focus on the communicative approaches that are used in GC communication, including non-directiveness. Although the GC profession prides itself on being able to provide complex risk information in non-direct approaches, many researchers have debated whether non-directiveness is the best form of communication, or even whether it is possible in practice (Arribas-Ayllon & Sarangi, 2014; Bernhardt, 1997; Clarke, 1991; Wessels, 2014). Furthermore, some studies have indicated that patients who are used to paternalistic approaches within particular healthcare settings may not respond positively to non-directive strategies in GC (Bosk, 1992; Ellington et al., 2005; Yelei et al., 2013). More research is needed to further explore the impact of non-directive counselling in various GC settings, including South Africa.

Furthermore, most interactional health GC research has been performed in westernised settings (Babul-Hirji et al., 2010; Benkendorf et al., 2001; Lehtinen, 2013; Paul et al., 2015; Pilnick, 2002; Roberts & Sarangi, 2005), with few GC studies having been done in socio-cultural diverse settings (Pilnick & Zayts, 2014; 2019; Wessels, 2014; Wessels et al., 2015; Wessels & Koole, 2019; Yelei et al, 2013). Interactional studies by Pilnick and Zayts (2012; 2019), Wessels (2014) and Wessels et al., (2015, 2019) suggest that contextual features such as socioeconomic circumstances and language diversity have the potential to create challenges in patient decision-making and GC communication and that greater attention to interactional verbal and non-verbal features are important to understand how communication is achieved. More research is needed to better understand how contextual influences from diverse settings impact GC communication.

7.3 A lack of interactional research for risk communication & uncertainty in genetic counselling

A large corpus of risk communication in GC exists, using several different research approaches (Cicero et al., 2017; Eichmeyer et al., 2005; Ellington et al., 2005; Fransen et al., 2006; Henneman et al., 2008; Kinney et al., 2010; Michie et al., 2005; O' Doherty & Suthers, 2007; Schapira et al., 2001). However, there is a paucity of knowledge regarding methods that observe naturally occurring risk communication in GC practice. Calls for qualitative studies that focus on GC processes and the communicative dynamics that occur during interactions have been made, since a better understanding of process not only allows genetic specialists opportunities for reflexivity (Meiser et al., 2008; Paul et al., 2015) but also creates a deeper understanding of patient needs (Bylund et al., 2012). Austin (2010) argues that GC risk communication involves far more than providing and discussing numeric probability, thus only focusing on patient recall of risk information is a poor measure of what genetic counsellors actually do. These additional tasks and activities during the GC process are crucial to understand the impact of GC services. Appreciating proximal activities during a healthcare interaction such as conveying information, making decisions, addressing patient concerns, ensuring comprehension of information trust, rapport, and promoting patient autonomy are critical influence distal outcomes such as patient satisfaction, uptake of services, and patient decisions regarding treatment and clinical management (Street, 2003; Street et al., 2009). It is for these reasons that attention to communicative processes within healthcare consultations are important.

The importance of process and communication is shown in a recent qualitative study in the USA by Joseph et al. (2017) using audio-recorded consultation data, and post-counselling patient interviews. The study found that there was a mismatch between what information was conveyed during cancer risk information counselling and what was considered valuable amongst a group of low-income, ethnically- and linguistically diverse population. Further calls have been made for investigations into genetic risk communication within diverse cultural settings (Cheng et al., 2018; Gaff et al., 2007; Paul et al., 2015; Pilnick & Zayts, 2014; Sivell et al., 2008).

Besides risk, uncertainty has also been shown to be important when focusing on health interactions (Ahmed et al., 2012; Arieli & Tamir, 2018; Watermeyer et al., 2020a) and in GC

communication (Bernhardt et al., 2014; Bylund et al., 2012; Hammond et al., 2020; Lou et al., 2020a). Although research has captured the reflective accounts of uncertainty in GC, there has been little research focusing on how uncertainty shapes communication processes by studying actual interactions (Pilnick & Zayts, 2014; Sarangi & Clarke, 2002a; Steel et al., 2014, Stivers & Timmermans, 2016; Thomassen Hammerstad et al., 2020).

Uncertainty has become a particularly relevant and re-focused topic since the rapid advance of genetic testing and technology which has brought new data that we are yet to understand and interpret for clinical purposes. A deeper understanding of uncertainty as a relational concern and how it is managed in GC interactions is needed. An understanding of the contextual factors that influence uncertainty management and how uncertainty is introduced into a conversation is also important.

7.4 A need for further risk & uncertainty studies in a South African genetic counselling context

South Africa's broader public healthcare system poses challenges to communication due to the country's social, historical, and cultural context (Coovadia et al., 2009; Penn et al., 2010). One of the biggest communicative challenges during local healthcare interactions is the diversity of language, culture and context, amongst patients and healthcare practitioners (including genetic specialists). Interactions in the public healthcare system often involve patients who have poor literacy, numeracy as well as analytical skills barriers, making mutual understanding difficult (Coovadia et al., 2009; Schlemmer & Mash, 2006; Wasserman et al., 2010). Also, some healthcare practitioners struggle to use basic health education materials and to formulate language that is appropriate for the patient's level of understanding (Koch-Weser et al., 2010; Wasserman et al., 2010). The above-mentioned challenges have the potential to culminate into complex health interactions and discussions.

Several South African studies have indicated a need for GC communication and training revision to improve the effectiveness of GC amongst the country's diverse population (Gillham et al., 2015; Kromberg et al., 2013a; Shingwenyana et al., 2020; Solomon et al., 2012). Previous South African medical genetic studies have mainly focused on genetic testing for certain conditions, genotype-phenotype comparison studies, retrospective file reviews, service audits, post counselling patient perceptions, family communication, behaviour change, and knowledge

recall (Carzis et al., 2019; Erasmus, 2009; Feben et al., 2013; Jefferies, 2013; Kromberg et al., 2013b; Kruger, 2005; Macaulay, 2008; Morris, 2013; Schoeman, 2007; Sizer et al., 2012; Stoler, 2013; Wainstein, 2011).

Studies involving risk perceptions and communication have mainly involved White participants using patient file-based retrospective studies (Erasmus, 2009; Macaulay, 2008), telephonic interviews (Jefferies, 2013), or internet-based surveys (Stoler, 2013). These studies were also conducted across public- and private-based patient sample sets; thus, patients may have had different levels of knowledge about GC and scientific and health literacy.
Of the studies involving risk investigations across cultures, poor knowledge recall, a lack of screening uptake, and poor family risk communication have been reported (Gillham et al., 2015; Kruger, 2005; Loggenberg, 2006; Schoeman, 2007; Solomon et al., 2012). The latter studies all used post-counselling interview methods and demonstrated that intercultural risk communication is complex and that local patients may not be effectively utilising genetic information. Schoeman (2007) has suggested that risk communication be more closely assessed in local GC consultations together with the development of effective intervention strategies. Findings from Schoeman's study were based on interviews with parents who had received GC.

More research on the influence of context has also been previously suggested by South African GC researchers (Kruger, 2005; Loggenberg, 2006; Penn et al., 2010; Shingwenyana, 2020; Solomon et al., 2012). Kromberg et al. (2013a) also suggest investigation on how genetic counsellors adjust their communication and counselling style for individuals from other cultural groups. Part of this research focus may be acknowledging the uncertainties that cross-linguistic and cross-cultural communication brings and working towards implementing context-specific genetic specialist practices, rather than trying to implement standardised service guidelines on a global scale (Daniels & Swartz, 2007; Penn & Watermeyer, 2012b; Wessels, 2014).

Despite the calls for increased attention on South African GC communication, particularly risk communication, there is little existing South African GC interactional, or evidence-based research to support these calls (Perrott, 2010; Wessels 2014, Wessels et al., 2015, Wessels & Koole, 2019). Although both Perrott (2010)'s and Wessels' (2014) studies are useful in understanding the interactional components of GC communication in South Africa, Perrott did not fully explore the micro-interactional features of talk or non-verbal communication.

Wessels' (2014) study provides a richer account of micro-interactional features in local GC communication; however, risk and uncertainty were not the main focus. Both studies also only focus on a small area of GC services, namely GC for AMA referrals. There is thus, to my knowledge, limited information on how genetic risk is communicated for different types of genetic conditions and the subsequent impact of risk and uncertainty in public healthcare GC consultations in South Africa. How uncertainty is managed interactionally in South African GC consultations is also unclear. As part of enhancing and learning about what care in a South African GC context looks like, it is important to understand what the processes of communication are, including how uncertainty may weave itself into the interactional process and its effect on communication.

7.5 Conclusion

GC approaches, success, and goals continue to be guided by GC models and guidelines developed in the global north, which may be less applicable in other geographical settings (Speight, 2020). By exploring the micro interactional activities and details concerning risk and uncertainty, we may begin to see their impact on the overall outcome of a consultation. Furthermore, the impact of broader contextual influences on risk communication and uncertainty management can also be better appreciated. Further findings may enhance awareness of context and assist in developing new models that align with the on-the-ground needs of both South African patients and genetic counsellors, and that pay attention to context-specific nuances. Findings may also be useful as a guide for other GC services based in other globally socio-cultural diverse contexts.

Furthermore, gaps in international and local GC knowledge leave opportunities to provide evidence-based research that highlights the value of care in GC. Care in South African GC consultations may look different to care in other GC locations. To better understand what the actual determinants of service quality are, more attention needs to be paid to the GC process in various settings (McAllister et al., 2016). With the advancement of medical genetics and the many socio-cultural diverse settings and clients that the GC field now encounters, focusing on the process of GC interactions is an important step in enhancing and ensuring that care remains a core value within GC regardless of the setting in which it occurs. Further exploration of GC processes and communication in non-western contexts such as South Africa is thus imperative to enhance the field and outcomes of GC both on a local and global scale.

This study answers the call for further evidence-based research that focuses on communication processes and how these processes may influence risk communication, uncertainty, as well as care during GC consultations within a socio-cultural diverse setting.

CHAPTER 8: METHODS

8.1 Introduction

This chapter details the overall research methods: namely the research questions, aim, objectives, research design, the research setting, the study participants, inclusion and exclusion criteria, as well as the data collection, and analysis methods. Trustworthiness, together with ethical considerations and reflections are also discussed.

8.2 Research question

How is genetic risk communicated and uncertainty managed in South African genetic counselling consultations?

8.3 Aim & objectives

8.3.1. Aim

To explore how genetic risk is communicated and how uncertainty is managed during genetic counselling consultations in a South African public healthcare context.

8.3.2. Objectives

1. To identify the types of risks discussed during South African public genetic counselling consultations.
2. To identify genetic specialists' approaches when communicating risk to patients for different genetic conditions.
3. To explore the interactional processes that occur during discussions of risk and uncertainty in local GC consultations.
4. To identify contextual influences in South African public healthcare genetic counselling consultations that impact risk communication, uncertainty management, and care.

8.4 Research design

8.4.1 Qualitative research

The study adopted a qualitative research design. Qualitative research examines different phenomena within natural settings, in an attempt to make sense of how everyday routines or practices occur and how people bring meaning to them through subjective or social

interpretations (Denzin & Lincoln, 2011). This research approach is not only inductive, but also flexible. Having emerged from different disciplines and academic thinking, qualitative research has several theoretical paradigms, methods and focuses (Beeson, 1997). For this reason, this umbrella term can be difficult to singularly define. To give a simple definition, qualitative research can be understood as a practice of 'observing' the world around us and making 'visible' various phenomena in it. The 'observer' (either the researcher or participant) plays an important role in how the world is seen and is often part of the world, rather than separated from it (Denzin & Lincoln, 2011, p. 3).

In qualitative research, data is collected through various means. It can either be gathered from naturally occurring observations (e.g., video- and voice-recordings) or by researcher-provoked methods (e.g. interviews, focus groups, introspective journals, field observations, narratives and graphical material) (Denzin & Lincoln, 2011; Flick, 2014; Speer, 2002). These collections are then further interpreted using various analytical methods to explain evidential phenomena and/or to develop a theory (Denzin & Lincoln, 2011; Pope et al., 2000).

Studying the 'noise' of our social worlds is not easy. Qualitative data can be difficult to collect and challenging to interpret – both of which can be extremely time-consuming. Furthermore, many qualitative studies are context-specific; thus, the outcomes and recommendations may not be relevant in other settings. Despite the healthcare field's preference for quantitative research that is able to produce replicable, controlled, and globally comparable data (Swartz & Kilian, 2014), qualitative research brings a unique lens to healthcare as it can highlight the psychological, social, political and historical impact on health as well as conversational nuances that occur during healthcare service delivery (Beeson, 1997; Street, 2003; Waitzkin, 1991). The value of qualitative research has also previously been highlighted in genetic counselling (Biesecker, 2018; McAllister et al., 2016).

8.4.2. Choosing a suitable qualitative framework

To better understand communication in patient-health practitioner consultations, it is important to focus on both the interactional communication processes as well as details of the surrounding context (Drew et al., 2001; Street, 2003, Street et al., 2009). This type of approach may allow for rich and detailed descriptions of the phenomenon under investigation (Durrheim, 2006).

For my research, I have chosen to use a blended qualitative framework that uses different methods and draws predominantly from social constructivist and interpretive research paradigms. This blended approach pays attention to different micro and macro layers of communication.

Healthcare studies are particularly complex in nature due to the multi-dimensional aspects of healthcare settings, services and practice. Health researchers may thus need to select multiple methodologies that provide accurate reflections of these environments and the interactions that occur within them so as to provide a more holistic and realistic view of the research setting and its participants (Denzin & Lincoln, 2000; Foss & Ellefsen, 2002; Jones & Bugge, 2006; Street, 2003). Applying various research methods to better understand the complexities of healthcare communication and service delivery could thus be seen as a study strength and an important part of academic rigour. Previous South African healthcare communication studies have also approached local patient-health practitioner research in similar ways paying attention to interactional activities and context in different health consultations and settings (Evans, 2010; Penn & Watermeyer, 2018; Watermeyer, 2008; Wessels, 2014).

Finding a suitable methodological framework that encompasses different levels of attention was challenging and it took several months of deliberation to find the 'right fit'. Most methods do not focus on both the micro- and macro-levels of detail. After much thought, I decided that one type of data source and analytical method may be too narrow in its scope. I have thus chosen to use a *bricolage* inspired approach (Denzin & Lincoln, 2000; Kincheloe, 2011; Lèvi-Strauss, 1966).

8.4.2.1 Bricolage

Bricolage is an approach to research that involves borrowing from multiple, sometimes opposing, theoretical and methodological perspectives to engage with complex research questions and achieve meaning-making (Rogers, 2012). Although criticised by more traditional researchers, who oppose inter-disciplinary approaches due to the risk of superficial examination of phenomena, or the creation of chaos in the research process (Friedman, 1998; McLeod, 2000), researchers who support a bricolage approach argue that the world is complex with many different lenses. Thus, a singular research approach may be superficial (Kincheloe, 2011). Bricolage therefore encourages the researcher to consider different levels of inquiry, different theories, and different ways to collect and analyse data to gain a more realistic sense

of the world or of certain activities within it. Bricolage also highlights the impact of the researcher on the research process. The researcher's familiarity and connection with the research setting and participants, their socioeconomic background and their lived experiences play a significant role in how data is collected, analysed and reported. Thus, researcher self-reflection and awareness are a critical part of a Bricolage research approach (Kincheloe, 2011; Rogers, 2012).

In keeping with a Bricolage philosophy, this study's approach is by no means purist. Rather it draws and borrows from different analytical approaches and data sources. To better understand GC communication in a South African public-based healthcare setting, I have chosen to focus on observations of naturally occurring GC consultations as my main data source, together with interviews with individuals who provide GC services, and field observations of the GC clinics.

8.4.2.2 Paying attention to the micro aspect of communication: Collecting talk intrinsic data

My main interest for the study is the 'micro' use of language in interaction. Methods that pay attention to the use of language in interaction has been investigated by several different prominent researchers and different terms and methods have been described, predominantly in the fields of linguistics, psychology and sociology. These include discourse analysis (DA) (Silverman, 2011), conversation analysis (CA) (Sacks et al., 1974), linguistics (Chomsky, 1953) and ethnomethodology (Garfinkel, 1967).

Although having similarities and overlaps, these approaches also have different stances on how language and understanding is achieved during conversations. Some of these methods view language as a cognitive function i.e., conversations occur due to a speaker having an innate cognitive awareness of how to speak and the ability to understand what the other individual is saying (e.g., some linguistic and DA-based approaches). Alternatively, other methods view language production during conversations as a structured social phenomenon (e.g., other DA-based methods and CA). The latter methods, although acknowledging that cognitive awareness of grammar is important to the development of utterances, draw attention to the mundane, taken for granted activities such as turn taking, who speaks and when, and how turns are constructed, to demonstrate that the delivery of conversation occurs through unspoken rules that are highly patterned and organised. Language use in conversations between individuals is thus not a mere conduit of information exchange, but rather a collection of subtle social activities designed for various purposes (Wooffitt, 2005).

For this study, I chose to use a blended sociolinguistic approach that draws on 'principles' of CA (ten Have, 2007) and TODA (Roberts & Sarangi, 2005) - two distinct yet overlapping talk intrinsic or language-in-interaction based approaches. CA and some DA-approaches (such as TODA) can be considered to align with the concerns of ethnomethodology (Garfinkel, 1967). Ethnomethodology is an observation-centred approach that focuses on naturally occurring talk and how individuals make sense of everyday situations using tacit social resources within interactions. Everyday activities appear mundane and ordered at surface-level; yet, on a more intricate level, individuals orientate to specific unspoken social rules and assumptions that allow for meaning-making and orderliness (Wooffitt, 2005).

The organisation of institutionalised talk (e.g., healthcare settings) has become a popular area of ethnomethodological research (Heritage & Maynard, 2006). In addition to their focus on talk-in-interaction, DA methods such as TODA that are used with institutionalised settings are seldomly reduced to the level of speech only. Paying attention to surrounding contextual details such as space, non-verbal behaviours, and visual phenomena are also thought important (Roberts & Sarangi, 2005; Sarangi, 2010; Sarangi & Candlin, 2003). Thus, apart from data sources that capture naturally occurring interactions, additional data sources that highlight context and experience may also be necessary (Roberts & Sarangi, 2005).

8.4.2.3 Paying attention to the macro aspect of communication: Collecting talk extrinsic data

In addition to the interactional data, I also include voice-recorded participant interviews and observational researcher field notes (van Maanen, 1988) to gain a broader 'macro' understanding of the experiences and context of communicating genetic risk and managing uncertainty in South African GC consultations. Participant interviews and researcher field notes are considered 'talk extrinsic data' (Waring et al., 2012) that help one understand 'why' interactions occur in the way in which they do. Combining these methods with interactional language-focused methods can be valuable to understand complex medical contexts and consultations as demonstrated by previous cancer (Mahmoodi, 2015), pharmacy (Watermeyer, 2008) and genetic counselling (Wessels, 2014) studies.

For the interview data I use Thematic Analysis (Braun & Clarke, 2006), and for the observational field notes I have used principles of Content Analysis as suggested by Mays and Pope (1995). An in-depth explanation of the specific data sources, how CA and TODA have

been blended, as well as how additional data sources were collected and analysed is presented in the sections below.

To assist in describing this study's blended framework I use a metaphor of a patchwork quilt. The talk intrinsic data, on a micro level, can be viewed as the multiple threads that make up the components of talk within GC consultations, e.g., who speaks, when and how. The patches of the quilt represent the talk extrinsic data that provide a broader macro insight into the experiences of individuals who provide GC services, as well as the patient, social and institutional factors that influence the communication process in GC consultations e.g., the patient's lifeworld, the voice of medicine and the context of the clinic and the consultation.

Each thread and patch provides a unique lens to understand the communication of genetic risk and the management of uncertainty. However, to view the quilt in its entirety, it is necessary to look beyond the individual threads and patches to truly appreciate how the multiple threads and patches are interwoven and combined to create a much larger body of work. Similarly, using different sources and analytical approaches in this study aids in understanding the different intricate layers of genetic risk communication and uncertainty management in South African GC consultations on a more holistic level. Figure 8.1 provides a representation of the study's framework. An explanation of the specific data sources and analytical approaches is presented in more detail in the sections that follow below.

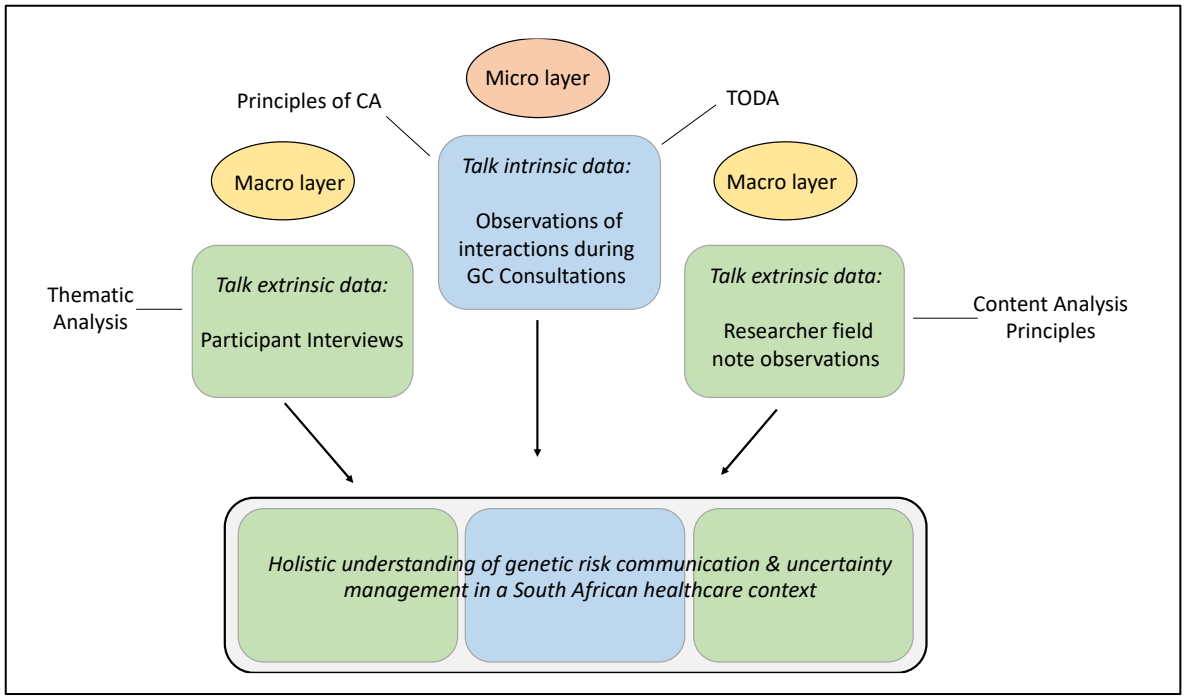


Figure 8.1 Representation of the study's blended research framework

8.5 The research setting

The study focused on GC clinics in South African public-based healthcare facilities. These facilities are the busiest and most established GC services in the country. Further, most genetic specialists providing GC and risk assessments in South Africa are based in public-based health institutions. The genetic clinics also offer a contextually rich environment for studying genetic risk and uncertainty communication as the patients who access the services are from diverse social, cultural and linguistic backgrounds.

Public genetic clinics are primarily situated at tertiary academic hospitals in Cape Town and Johannesburg. There are also limited public genetic services available in Bloemfontein, Pretoria and Durban. I approached the largest centres, namely those based in Johannesburg and Cape Town, to participate in the study. The genetic clinics based in Cape Town agreed to participate in the study, hence were chosen as the respective research sites. These genetic clinics are situated within three public tertiary academic hospitals. All three hospitals provide several healthcare services to large communities within Cape Town and surrounding areas. The hospitals also serve as teaching, training and research facilities for various healthcare students and practitioners.

Although the history of South African medical genetics and GC services has already been expanded upon in the literature review chapter, the provincial, city, hospital and clinic contexts of the research study, will now be elaborated on.

8.5.1 Western Cape Province & the City of Cape Town

The Western Cape is the fourth largest province in the country, compromising of 6.3 million (11.3%) of South Africa's 55.6 million individuals. Cape Town, situated in the southern peninsula of the province, is the provincial capital and economic hub. It is the country's oldest and second-largest financial centre and has a Mediterranean climate characterised by hot dry summers, and cold wet winters (Cummins, 2002; Municipalities of South Africa, 2017). In 2016, Cape Town's population was estimated at 4,004,793 (64% of the total number of individuals living in the Western Cape). The city's population compromises mainly Mixed Ancestry (42.4%) (the majority being of European-African-Malay descent); Black African (38.6%) (mostly isiXhosa rural-urban immigrants from the Eastern Cape); White (15.7%) and Indian/Asian (1.4%) individuals (Parker et al., 2012; Statistics South Africa, 2016; 2017). Although difficult to estimate accurate numbers, there are also individuals from other African countries, including Zimbabwe, Malawi, the Democratic Republic of Congo, Nigeria and Somalia - with 3.3% of the Western Cape's population thought to be non-South African citizens (Alfaro-Velcamp et al., 2017). Like all provinces in South Africa, multiple languages are spoken in the Western Cape, however Afrikaans (34.95%), isiXhosa (29.2%) and English (27.8%) are the dominant languages represented (Statistics South Africa, 2017).

Cape Town is a city of contradictions. On the one hand, it has become synonymous with the country's history and tourism industry, with its scenic ocean, mountain and wine-growing landscapes, considered some of the most beautiful in the world. Furthermore, it has the highest density of local multimillionaire's living on some of the most expensive real estate properties in the country. In comparison with other South African provinces and cities, Cape Town is perceived to have better standards of living due to lifestyle, public infrastructure, social services, and healthcare. Statistically, 90.5% of individuals living in the City are literate, over 90% have access to basic services, and Cape Town is thought to have some of the top local performing public healthcare facilities in the country. Not only do these facilities serve 74% of the Western Cape population, but they include two respected medical schools, as well as three large tertiary academic hospitals (the ones accessed in the study) (Cummins, 2002; Statistics South Africa, 2017; Urban et al., 2011; Western Cape Government Health, 2015).

Despite these positive aspects, the city has another side to it. Cape Town's wealth is starkly juxtaposed against extreme poverty. Race inequality, due to the legacy of Apartheid, still determines much of the socioeconomic challenges. Indeed, the majority of the informal settlements in the City disproportionately house poor Black African and Mixed Ancestry individuals, many of whose homes are flooded due to winter rainfall, and who do not have access to proper sanitation and electricity (Cummins, 2002). Current social concerns include a shortage of housing, a 39.3% unemployment rate (especially prevalent for individuals <35 years of age), a school drop-out rate of 39%, as well as individuals earning little to no salaries, with many living below the poverty line determined as earning less than R3500 per month (Statistics South Africa, 2017; Western Cape Government Provincial Treasury, 2014).

Furthermore, Cape Town, together with the broader Western Cape population, is burdened by national healthcare concerns, such as infectious diseases (TB/HIV); non-communicable diseases (diabetes, hypertension and cancer); as well as maternal and child health risks (including high rates of termination of pregnancy, maternal mortality and teenage pregnancy). Although spending a larger portion of governmental funding on healthcare compared to other provinces, the Western Cape's public healthcare facilities are still burdened by a lack of resources and staff unable to adequately serve population demand. Inter-personal violence, gangsterism, as well as drug and alcohol abuse are also common social problems. The Western Cape has become synonymous with some of the highest fetal alcohol syndrome (FAS) statistics, not just nationally, but globally (Cummins, 2002; Olivier et al., 2013; Western Cape Government Provincial Treasury, 2014). Alcohol use and FAS are thus common topics of discussion in the province's prenatal and paediatric healthcare services.

8.5.2 The hospitals & the genetic clinics

Although affiliated to different healthcare centres and tertiary academic institutions, the genetics clinics at the three chosen public hospitals run similar services. For purposes of anonymity, I have decided not to explicitly disclose the names of the hospitals. The reasons for this have been set out in Chapter One of this thesis. I however, acknowledge that maintaining complete anonymity of the research sites in this write up may be difficult.

The one chosen hospital is the largest healthcare facility in the Western Cape (Patel & van Niekerk, 2015). The hospital offers high-level, specialised tertiary patient care within the

public healthcare structures, annually produces a large number of graduating medical professionals and has received accolades for medical research and clinical advancement (Sanders & Kane-Berman, 2012; Western Cape Government, 2014). The hospital has several in-patient wards, sub-specialist services, out-patient clinics, and community outreach programmes. Furthermore, it serves a population catchment area of approximately 1.5 million individuals, with patients referred from various primary clinics and secondary healthcare facilities, both from in and around Cape Town as well as further afield (Myer et al., 2013; Patel & van Niekerk, 2015; Sanders & Kane-Berman, 2012). Most patients attending the hospital are individuals from lower socioeconomic strata (Myer et al., 2013).

The second chosen hospital until recently, was the only hospital in Southern Africa committed entirely to child healthcare. It manages approximately 260,000 national and international patient visits per annum with a third of patients being under the age of 1 year. Furthermore, it offers high-quality services, such as paediatric intensive care, organ transplants and cardiothoracic surgery. Although serving a range of patients, many are individuals from disadvantaged communities and families (Hospital Two, 2008). The hospital also serves as a teaching, training, and research facility for medical and allied healthcare professionals.

The third chosen hospital is considered to be the second-largest hospital in South Africa, servicing approximately 3.5 million people from areas including Cape Town's eastern and northern metro-districts, Khayelitsha, as well as rural regions beyond Cape Town, such as the West Coast, the Cape Winelands and Overberg (Hospital 3 Information Pamphlet, 2016; Hospital Three Overview, 2017; Schoeman et al., 2013). Patient demographics vary, but the hospital primarily provides services to poor, working individuals living mostly in informal settlements or in low-income formal housing communities (Li et al., 2010; Schoeman et al., 2013). The hospital has 4,441 dedicated staff, with annual patient figures equating to approximately 107,215 admissions and 492,670 outpatient visits. Hospital 3 offers several specialised health services as well as tertiary-level teaching, training and research (Bateman, 2015; Hospital 3 Information Pamphlet, 2016; Li et al., 2010).

Genetic services are predominantly funded by the Western Cape Department of Health or by the affiliated tertiary facility budgets. The majority of patients accessing GC services in the public healthcare system do not incur any personal fees. Genetic clinic staff comprise of medical geneticists, genetic counsellors, genetic nurses and medical scientists who offer full,

mainly outpatient, clinical and laboratory services to patients and families. District and regional outreach services and limited services for patients from the private healthcare sector are also provided at these clinics. In addition to clinical services, the genetic clinics also provide teaching, training and research opportunities for GC students, medical students, medical registrars and interns (Greenberg et al., 2012; Hospital Three Annual Report 2014, 2014).

Regular genetic clinics offered at the three hospitals include prenatal, paediatric, adult (which may **i**nclude oncology, syndromic and adult-onset disorders), as well as cardiac and neurology clinics. Additional services at these clinics also include stillbirth investigations and hospital-based call-outs (UCT, 2021b; Hospital 3 Annual Report, 2014).

8.6 Inclusion & exclusion criteria

8.6.1 Inclusion criteria

The study includes any GC consultation that took place at one of the three selected hospitals. The research was not limited to one genetic condition or a particular reason for referral, nor was it limited to a particular type of genetic clinic. Nearly all GC consultations contain some form of risk and uncertainty due to the typical nature of risk-focused assessments and discussions in GC consultations.

Reasons for choosing an open approach is due local genetic counsellors to be generalists, rather than only working with particular genetic conditions or referrals (Greenberg et al., 2012; Kromberg et al., 2013b; Ormond et al., 2018). Thus, there may be several types of GC referrals within the same clinic or at the same hospital. The clinics are also not prescriptive to whether the scheduled consults seen in a particular week are new consults or follow-up consults. Rather there is much heterogeneity is what is seen on a particular week at a given clinic. Limiting data collection to a particular type of condition, sub-set of individuals, or consultation would have failed to produce sufficient data within the given time frame of the project given the rarity of some genetic referrals. Although some clinics tend to see more common types of referrals e.g., prenatal clinics regularly have advanced maternal age (AMA) referrals, previous local interactional research by Wessels (2014), Wessels et al (2015, 2019) and Perrott (2010) had already been conducted on South African public-based AMA referrals specifically. Thus, I did not want to solely focus on public AMA referrals for this study. Although dedicated inherited cancer and cardiac genetic clinics are run at some of the research sites, again the number of

patient referrals at these condition-specific clinics are limited and the clinics occur less frequently.

Further motivation for my sampling choice was due to practical and logistical issues regarding data collection. I am not based in Cape Town, but in Johannesburg. Financial constraints and limited project funding were a major limiting factor for not being able to travel to Cape Town as frequently to collect data. The genetic clinics in Johannesburg were initially considered as sites to conduct the research. I am based in Johannesburg and am already familiar with these genetic clinics and staff due to my training. However, challenges were incurred due to the Johannesburg-based clinics busy work schedules and conflicts in supervision and faculty registrations at the University when considering these sites in the early research phase. In addition, due to the proximity of my professional and personal relationships with the staff at the Johannesburg-based genetic clinics (more so than with the staff at the Cape Town genetic clinics), there were ethical concerns about participant coercion and conflicts of interest. The genetic clinics in Cape Town were thus approached as alternative as data collection sites.

In saying that I used an open data collection, I was purposive in selecting consultations were patients and genetic specialists were comfortable for me to record them. Potentially complex cases where greater sensitivity was needed for example a severe fetal diagnosis in the prenatal clinics, or where patients were visibly distressed prior to their consults were not included for ethical reasons.

8.6.1.1. The participants

Participants in the study include patients and genetic specialists at the selected genetic clinics. Similar genetic clinics are offered to patients at all three hospitals included in the study. Individuals who may benefit from a referral to a clinical genetic consultation include:

- Those who have a family history, or who are affected by a genetic disorder.
- Are related (consanguineous) or are from a high-risk ancestral group for certain disorders (e.g., Indian, Ashkenazi Jewish or Afrikaner).
- Couples who have had multiple miscarriages, or who have/have had a child born with a birth defect or genetic syndrome.

- Pregnant women older than 40 years (considered of Advanced Maternal Age) (some hospitals consider this to be over 35 years of age).
- Pregnant women who have been exposed to teratogens (e.g., recreational drugs, alcohol, certain medication, or infections).
- Pregnant women who have had an abnormal fetal screening result (UCT, 2021b).

The patients selected for participation in the study were, thus, any individual, or caregiver of an individual, seeking or referred to a GC consultation for any of the above-mentioned reasons.

Due to the convenience sample approach, the research includes both first-time patient GC consultations, as well as follow-up GC consultations. Follow-up patients were either receiving genetic test results or were having follow-up clinical assessments. As described in the previous sections of this chapter, many patients accessing the hospitals and GC clinics included in the study are Mixed Ancestry or Black African individuals, (mostly Afrikaans or isiXhosa first language speakers), predominantly drawn from lower socioeconomic backgrounds. Rather than being self-referred, almost all patients at the genetic clinics were referred by other health practitioners or clinics from various facilities within the public healthcare system. All GC consultations that met the study inclusion criteria were selected regardless of the language in which the consultation was conducted.

For the genetic specialist participants, I realise that focusing only on fully qualified genetic counsellors with years of professional experience may have been more useful rather than using in-training or newly qualified genetic counsellors and other genetic specialists since experience may bring about comfort and confidence risk communication skills. However, due to the profession of genetic counselling being relatively young with few senior genetic counsellors available (at the time of data collection there were only two fully trained genetic counsellors with more than three years' experience working at the research sites), the sample size would have been too small. Given the limited number of available qualified genetic counsellors at the three research sites, I chose to include medical geneticists, as well as other individuals in their final stages of training, namely senior genetic counselling students, genetic counselling interns and medical genetic registrars in this study. Apart from the practical issue of few senior genetic counsellors being available at the research sites, part of my motivation is also that GC is not

synonymous with genetic counsellors only. Resta et al. (2006) highlight that the NSGC make an important distinction in their widely used definition of GC:

> Genetic counsellors have many roles… Although many of these activities relate directly to genetic counselling, they are distinct from the counselling process… Therefore, rather than defining what genetic counsellors **do**, we were charged with describing what genetic counseling **is**, i.e., the essence of the relationship between the client and counsellor when they interact. In defining genetic counselling practice, we do not stipulate who is qualified to provide it. (p. 78).

Although different levels of expertise, background training and years of experience do potentially bring differences in individual communication skills, all the chosen participants are trained in similar ways to convey genetic risk information in local GC consultations. In South Africa, medical geneticists and genetic nurses also receive GC training and practice GC, often working at the same professional facilities and clinics at which genetic counsellors train and work at (Greenberg et al., 2012; Kromberg et al., 2013a). This similar training made the selected participants suitable for the purposes of the study which was to describe (and not necessarily compare amongst) how genetic risk is communicated by local genetic specialists. The study is also not about evaluating genetic specialists' skills but exploring the process of GC risk and uncertainty communication and how this may impact patient care. All genetic specialists at the chosen genetic clinics were thus invited to participate in the study. Although there was a representation of various ages, races and gender, the majority of local genetic specialists included in the study are female, Caucasian, English and Afrikaans first-language speakers, whose ages range from mid-20s to 50 years of age.

8.6.2 Exclusion criteria

The study had no exclusion criteria regarding the types of genetic conditions or types of public genetic clinics. However, on the days during which data was collected, patients who were visibility upset or patients who had been referred for a very severe diagnosis which would involve complex information and extreme sensitivity (as reported by genetic specialists at the clinic) were not approached for inclusion in the study, to minimise any additional psychological distress.

Moreover, individuals who are still considered junior, first-year students were excluded from the study. First-year students are still mastering the skill of GC communication with many unable to provide independent counselling to patients. Having a senior staff member supervise a trainee's consultation with a patient whilst it is being video-recorded may impact how communication occurs and create additional pressures in the consultation. Interviewing junior, first-year students were also not deemed beneficial, as they did not yet have first-hand nor independent experience of communicating risk and uncertainty to patients in the local healthcare setting.

No patients were interviewed during the study (particular data sources are described later in this chapter). Although understanding patients' perspectives is important, the research focus for the study is more aligned to specialists' approaches to communicating risk and trying to understand their accounts of managing uncertainty. Evans (2010) and Wessels (2014) have noted that asking patients what constitutes 'good communication' in the South African healthcare context can be challenging since they often respond in passive ways or they provide what they feel may be a desired affirmative response. Also, although difficult to separate, asking a patient what they remember after a consultation may only assess knowledge recall and may not necessarily indicate patient understanding (Edwards & Elwyn, 1999). It may also not be ethically appropriate to interview a patient immediately after a GC consultation in which distressing news, or a severe medical diagnosis is conveyed to them. Therefore, it was decided not to conduct patient interviews as part of the study.

8.7 Data sources & data collection

For the study, the following data sources have been selected:

- Video-recorded genetic counselling consultations.
- Audio-recorded genetic specialist interviews.
- Researcher field notes.

I travelled between Cape Town and Johannesburg (where I am based) on four separate occasions to collect data at each of the three chosen hospitals included in the study. Table 8.1 includes details on the dates of these trips.

Table 8.1: Data collection process

Dates of site visits	**12-15 September 2016**	**9-11 November 2016; 14-16 November 2016**	**12-13 December 2016**	**15-16 February 2017**	**Total**
Number of genetic specialist-patient consultations recorded	3	6	6	3	**18**
Number of hours of video footage recorded	1 hour 22 min	2 hours 56 min	3 hours 33 min	2 hours 14 min	**10 hours 5 min**
Number of genetic specialist interviews conducted	4	5	2	1	**12**
Number of hours of interview data recorded	1 hour 38 min	2 hours 41 min	1 hour 34 min	33 min	**6 hours 26 min**
Length of time spent in a clinic in which field notes were recorded	4 & 4	4.5	5.5	4.5	**18.5 hours**

8.7.1 Genetic counselling consultations

For the study, it was decided that observations of naturally occurring GC consultations were the best-suited method to better understand the process of local genetic risk and uncertainty communication. Observation of every day patient-healthcare practitioner consultations has become one of the most reliable ways of gathering evidence in medical interactions and have been used both in local and international healthcare studies (Penn & Watermeyer, 2018; Pilnick & Zayts, 2014; Pino et al., 2017; Watermeyer, 2008; Wessels, 2014). Furthermore, observing every day, naturally occurring GC consultations allows one to delve into, what Biesecker and Peters (2001) have described as the unknown 'black box' of GC process, something that is still not well understood (Biesecker, 2020; Biesecker et al., 2019; Resta, 2019).

According to Marvasti (2014), observation research tries to understand how activities and events occur within particular contexts, not only from an outsider's perspective but also from the participants' perspectives. Merits of studying naturally-occurring talk include 1) minimal risk of the researcher influencing the setting through their own agenda (created through posed interview questions/vignettes or presumptions), 2) novel findings are revealed without any external prompting, and 3) providing an authentic and rich account of people in natural life situations and interactions (Potter, 2002). Such research methodology also sheds light on micro-interactional phenomena that occur beyond words, e.g., non-verbal gestures, the use of props and tools, as well as what occurs during moments of silence (Heath et al., 2010; Heritage & Maynard, 2006; Parry et al., 2016; ten Have, 2007; Toerien, 2014). Observations thus open a window into transparent, 'real' moment data, especially when the initial self-consciousness of participants dissipate, and they can ignore the video- or voice- recording equipment.

8.7.1.1 Collecting the genetic counselling consultation data

Video-recording equipment was used to collect the GC consultation data for the study when I attended the genetic clinics. Genetic specialist were invited to participate either on the day of the clinic if I had not yet met them, or they had been invited and given the patient information sheets earlier in the week at their offices. Genetic specialists were reminded that they were not obligated to participate in the study, and if they did, their anonymity would be protected.

Upon arrival at each genetic clinic on each visit, I was shown a list of patients who had been referred for the day and their reasons for referral to the genetic clinic. Together with input from the attending genetic specialists, I was able to discern which GC consultations might be appropriate for me to include in the study.

At the start of the clinic and before approaching any of the selected patients, I first discussed each consultation with the assigned genetic specialist. On receiving permission from the genetic specialist, I then approached the relevant patient(s) and invited them to participate in the study. I tried to ask patients at the start of the clinic if they would be interested to participate in the study and provided them with a relevant information sheet (Please see Appendix F) and then allow them some time to think about their decision whilst sitting in the patient waiting area before letting me know of their decision. This approach attempted to avoid patients making rushed decisions or feeling ambushed in the consultation room when about to commence their consultation. It also minimised the possibility of patients feeling coerced

which may have occurred if I had asked patient to participate in the presence of their managing genetic specialist. None of the genetic specialists were involved in inviting patients to participate, rather I did all informed consent procedures myself.

If agreeable, the patient then signed consent and continued to wait for the name to be called by the relevant genetic specialist for the consultation. When it was the patient's turn for their consult, I then set up the recording equipment in the relevant consultation room. I was not present in the consulting room while the consultations took place, rather relying on video- and voice-recordings to observe the consultation after the fact. The recording equipment in the room already had the potential to change the dynamics between the genetic specialist and the patient(s), thus I did not want to further compromise the research setting through my presence. Furthermore, because these clinics are housed within training hospitals, I also had to share the clinic space with genetic counselling students during some of my site visits. Two of the recorded GC consultations had a genetic counselling student sitting in the corner of the room observing the consultation. Although their presence could have impacted the dynamic of the session, the students were instructed to observe, but not provide any input. I also ensured that the students were not captured on the recording equipment. For each of the selected consultations, once the patient and the genetic specialist were ready to commence the consultation, I switched on the recording equipment and left the room. I similarly entered the room to switch off the recording equipment when the consultation had ended.

One patient and one genetic specialist declined participation in the study. The patient had been referred for advanced maternal age (AMA) counselling, and when invited to participate, cited that they did not have time to participate. The patient had been waiting at the clinic for some time and seemed to view participation in the research as another 'extra' in an already long day. The genetic specialist who declined participation cited their discomfort with the video-recorder and that it would make them feel anxious and did not want to be interviewed.

8.7.1.2 Overview of the collected genetic counselling consultation data

Although an attempt was made to create variety in the recordings (regarding the type of diagnosis and the individuals present), this was largely dependent upon timing and genetic specialists' availability during data collection. Thus, whilst the recorded GC consultations all involved different patient participants, some genetic specialists were recorded more than once across the different clinics. The average time of each GC consultation was 35 minutes, with the shortest consultation time being 19 minutes, and the longest 1 hour and 8 minutes. Time

differences depended on whether it was a first-time consultation or a follow-up consultation (follow-up consultations tended to be shorter), the complexity of the information conveyed and the patients' reason for referral. A total of 18 GC consultations were recorded and included in the study.

A summary of the types of recorded GC consultations can be found in Table 8.2. The demographics of the patients in the recorded consultations, their reasons for their referral and more specific details for each GC consultation can be found in Table 8.3. Table 8.4 provides basic details relating to the various genetic specialists included in the study.

Table 8.2: Summary of types of recorded genetic counselling consultations

Type of consultation	Type of session recorded	No of genetic specialists involved	Totals
Prenatal (9)	First-time consultation (8) Follow-up consultation (1)	7	
Paediatric (5)	First-time consultation (2) Follow-up consultation (3)	3	
Adult (4)	First-time consultation (2) Follow-up consultation (2)	3	
Subtotal of first-time consultations			12
Subtotal of follow up consultations			6
Total No of GC consultations recorded			**18**

Table 8.3: Patient participant details for the video-recorded genetic counselling consultations

Case no	**Type of GC consultation**	**Reason for referral**	**First-time consult or follow-up (FU) consult**	**Gender**	**Age of the affected individual**	**Ethnicity**	**Home language**	**Accompanying individuals**	**Language of consultation**	**Specialist involved**	**Length of consultation**
1	Paediatric	Treacher Collins Syndrome	First	Female	3	Mixed Ancestry	Afrikaans	Father & pregnant mother	English	S1	28 min
2	Adult	Family screening for chromosome translocation	FU	Male	62	Mixed Ancestry	Afrikaans	Daughter	English	S2	32 min
3	Adult	Predictive counselling for Inherited Breast & Ovarian Cancer (HBOC)	FU	Female	66	Mixed Ancestry	Afrikaans	Daughter	English	S2	22 min
4	Prenatal	Advanced maternal age (AMA) & previous child with Down syndrome	FU	Female	43	Mixed Ancestry	Afrikaans	None	English	S6	23 min
5	Prenatal	AMA	First	Female	42	Black	isiXhosa	None	English	S1	23 min
6	Prenatal	Osteogenesis imperfecta	First	Female	31	Mixed Ancestry	Afrikaans	None	Afrikaans	S7	48 min
7	Prenatal	Soft markers for chromosome abnormalities detected on ultra-sound screen	First	Female	21	Mixed Ancestry	Afrikaans	None	Afrikaans	S7	44 min
8	Prenatal	AMA & fetal abnormalities	First	Female	42	Mixed Ancestry	Afrikaans	None	Afrikaans	S8	19 min

		detected on ultrasound									
9	Prenatal	AMA & soft markers for chromosome abnormalities detected on ultrasound screen	First	Female	42	Mixed Ancestry	Afrikaans	None	Afrikaans	S8	19 min
10	Prenatal	Teratogen exposure: Maternal rubella infection	First	Female	24	Caucasian	Afrikaans	Mother	Afrikaans	S9	33 min
11	Prenatal	AMA & soft markers for chromosome abnormalities detected on ultrasound screen	First	Female	39	Mixed Ancestry	Afrikaans	None	Afrikaans	S10	34 min
12	Prenatal	AMA	First	Female	41	Black	isiXhosa	Male partner	English	S7	34 min
13	Paediatric	Hypophosphatasia	FU	Male	2	Black	isiXhosa	Father & mother	English	S2	31 min
14	Paediatric	Down syndrome	First	Female	2 weeks	Black	isiXhosa	Mother	isiXhosa (with an interpreter present in the consultation room)	S2	47 min
15	Paediatric	Gardner's syndrome	FU	Male	10	Caucasian	Afrikaans	Mother	English	S2	34 min
16	Paediatric	Query Prader-Willi syndrome	First	Male	10	Black	isiXhosa	Mother	isiXhosa (with an interpreter present via telephone)	S11	28 min
17	Adult	HBOC	First	Female	39	Mixed Ancestry	Afrikaans	None	English	S12	1 hour 08 min

18	Adult	HBOC	First	Male	32	Mixed Ancestry	Afrikaans	None	English	S12	38 min

Table 8.4: Genetic specialist participant details

Genetic specialist participant	**Genetic counsellor***	**Medical geneticist***	**Participation in the video-recorded GC consultations**	**Participation in the audio-recorded genetic specialist interviews**
S1	Genetic counsellor		Yes	Yes
S2	Genetic counsellor		Yes	Yes
S3	Genetic counsellor		No	Yes
S4		Medical geneticist	No	Yes
S5		Medical geneticist	No	Yes
S6	Genetic counsellor		Yes	Yes
S7		Medical geneticist	Yes	Yes
S8	Genetic counsellor		Yes	Yes
S9		Medical geneticist	Yes	Yes
S10	Genetic counsellor		Yes	Yes
S11		Medical geneticist	Yes	Yes
S12	Genetic counsellor		Yes	Yes
Sub total	**7**	**5**		
Total			**9**	**12**

* Socio-cultural details, professional training and years of experience for each specific genetic specialist has purposefully been omitted in Table 8.4 to maintain anonymity. To provide some context, although there was a representation of various ages, races and gender, the majority of local genetic specialists included in the study are female, Caucasian, English and Afrikaans first-language speakers, whose ages range from mid-20s to 50 years of age. The majority of genetic counselling participants had less than five years of professional experience, and the majority of medical geneticist participants had more than five years' professional experience.

8.7.2 Genetic specialist interviews

The second source of data incorporated in the study is semi-structured interviews with genetic specialists. Interviews are considered an interaction between a researcher and a participant, where the researcher guides the conversation by inquiring about a topic of interest (Babbie & Mouton, 2008). Combining interviews with research methods geared towards understanding communication processes, such as video-recordings of naturally occurring interactions, is thought to be a valuable knowledge-enhancing method to understand complex medical contexts and consultations, such as cancer, pharmacy and genetic counselling (Mahmoodi, 2015; Watermeyer, 2009a; Wessels, 2014).

Genetic specialists convey genetic risk and deal with uncertainty on regular basis, thus it was felt that they may provide further insight into the process, as well as details on context-specific challenges and facilitators. Although aware that only interviewing genetic specialists, and not patients, may lead to biased reporting, this method of data collection is considered valid if the researcher appreciates that it produces subjective data (Holstein & Gubrium, 2004). During the interview collection and analysis process, I followed recommendations made by previous researchers undertaking participant interviews. These recommendations include 1) practising reflexivity and minimising a predetermined agenda or viewpoint, 2) recognising that a researcher and the participants may have their self-interests and vulnerabilities, 3) placing participants' comments into context, 4) being aware of non-verbal participant cues, and 5) accepting that interviews are inherently difficult to perform well (Antaki et al., 2003; Gill et al., 2008; Potter & Hepburn, 2005).

8.7.2.1 Collecting the genetic specialist interviews

All genetic specialist interviews occurred once-off and were audio-recorded. Interviews took place in a private room at the genetic clinics, or in the genetic specialists' own offices. The length of the interviews varied with the average interview being 30 minutes, the shortest 17.22 minutes (the first interview conducted), and the longest 1 hour and 5 minutes (the second last interview conducted). Reasons some consultations took longer than others depended on the specialist's level of openness and possibly my role as a facilitator. Time constraints may have also added to the shorter length of some interviews as some of the interviews took place between specialist meetings, clinic attendance and other daily tasks.

8.7.2.2 The interview schedule

An interview schedule was used to facilitate discussion. Although this schedule provided a framework to help guide the interview, it was used in a flexibly. Thus, the order of questions varied, with some departure from the template, unscripted follow-on questions as well as additional comments made during different interviews. This flexibility allowed me to follow and appreciate the natural contingencies of conversation and interaction (Potter & Hepburn, 2005). The questions chosen for the interview schedule were informed by research interview guidelines (Gill et al., 2008) and designed to yield as much information about the research topic.

To refine the interview schedule, I conducted a pilot to assess its usefulness in the genetic specialist interviews. Piloting is helpful during the early stages of a study to ensure that an interview schedule is appropriate and covers the aims of the study (Gill et al., 2008). However, in some instances, piloting can be difficult, and even impossible, due to practical reasons, or because of limited participant availability (Chenail, 2011). During the study, piloting the interview schedule before data collection became a practical issue due to the proximity of the sites (I am based in Johannesburg and the sites are in Cape Town). I did not want to risk losing potentially vital information when testing the interview schedule, yet I was aware of the importance of ironing out potential problems early in the data collection process. I, therefore, used the first site visit and data collection trip to Cape Town on 12-15/09/2016 as a piloting opportunity for the interview schedule but included this pilot information in the study data. The pilot study showed that the interview schedule was largely suitable. Additional questions and probes were, however, added after doing the first few interviews to allow further in-depth exploration of the research topic. Details of the original and the adjusted interview schedules can be found in Appendix A.

8.7.3 Researcher field notes

The final 'talk-extrinsic' data used to complement the main participant observations are researcher field notes. Field notes are considered "short-hand reconstructions of events, observations and conversations that took place in the field" (van Maanen, 1988, pp. 223–224).

Observations of clinical activities are considered important since medical institutions are extensions of the broader social, cultural and political context of particular settings (Penn & Watermeyer, 2018; van der Geest & Finkler, 2004). Heath et al. (2010) further supports the

value of using field notes to complement the collection of video-data. Field notes may provide a deeper understanding of observations made within the video-data, especially where the participants orientate to features of a particular context in which the recordings were captured. Being a genetic counsellor myself and having previously worked in public genetic clinics (albeit in Johannesburg), many of the organisational routines and processes were familiar. However, prior to the study, I had never visited the genetic clinics in Cape Town. Thus, the hospital settings, the patients, some of the staff and the genetic clinic spaces in the study were new to me.

8.7.3.1 Collecting the researcher field notes

Taking cues from an ethnomethodologically inspired method (Garfinkel,1967), where one closely observes a setting and immerses oneself into the research participants' world and daily activities (Beeson, 1997; Mulhall, 2003), I chose to make observations whilst spending time at the genetic clinics.

A full ethnographic approach encourages a researcher to spend substantial time within a particular community. Wind (2008), however, highlights the challenges of doing full ethnographic approaches within specialised settings such as hospital and clinics. Since spending prolonged periods in such settings and blending in as 'one of them', i.e., a patient or a staff member, is practically difficult and many clinic and hospital activities occur within very structured timeframes and settings. Although I am a trained genetic counsellor, which did enhance my inclusion into "insider" conversations (Wind, 2008, p. 83), I was new to the research settings and many of the staff. Therefore, although having a good rapport with the clinic staff, I was still deemed an outsider. In the same way, although able to spend time with patients in the patient waiting-areas, my presence was noted as unfamiliar. Whilst observing clinic staff and patients, I had to be cautious of not making my observations as a researcher overly intrusive or that it interfered with work activities in the clinic. Furthermore, I was also cognisant of patients and staff feeling watched or judged. I noticed some security and medical staff as well as patients' discomfort when I sat in the patient waiting areas and when discussing my reasons for attending a hospital or clinic. Similar challenges have been noted by Wind (2008), where they describe the activity of hospital ethnography to be negotiated interactive observation, where the researcher observes the participants, but in the same way, participants observe the researcher and they do not fully allow the researcher to become part of their world.

To overcome the practical challenges of a full observational method, I rather decided to use a salient approach to my observations as suggested by Wolfinger (2002). A salient approach captures details and moments of interest but does not record all details in depth, hence is not considered an in-depth ethnographic approach. Mays and Pope (1995) indicate that the ability to capture every single detail through researcher observation is impossible, thus observation relies heavily on the researcher and what they choose to record. A salient approach is also subjective as to what gets noted, e.g., what is observed and how much is written. Furthermore, the notes may be influenced by whether the researcher is previously familiar with the space field or not (Wolfinger, 2002). Most of the observations of the research setting were done whilst sitting in the patient waiting-area or the staff-area of the genetic clinics. Although choosing to write about what clinical activities and contextual features stood out, I was also systematic in capturing certain features from each clinic. Being systematic is considered part of rigour in observational field notes (Mays & Pope, 1995). For each clinic I attempted to capture the following information: what the clinic looked like and its atmosphere, the activities of both patients and staff, clinical procedures and routines, coordination of patient referrals and administration activities. I also made notes on the participant recruitment process for each research site, as well as my emotional reactions and personal dilemmas encountered at each clinic. The details were kept in a field note diary that I took with me to each clinic, and I used the notes in this diary to then type up more detailed reflective pieces that were kept as electronic copies on a hard drive.

8.8 Data analysis

8.8.1 Analysing the genetic counselling consultations: A blended sociolinguistic approach utilising principles of CA & TODA

I believe that blending interactional qualitative methods such as CA and TODA are particularly well-suited to better understanding the process of GC and enhancing evidence-based practice. CA and other discourse methods dedicated to interaction have been critiqued for focusing on supposedly mundane details and activities, without addressing larger social issues. However, as Wooffitt (2005) argues, interaction and language are vital parts of our social worlds. Without grappling with how we manage communication activities and tasks within various settings, we fail to understand a key component of human behaviour.

The benefit of combining interactional methods has been previously reported (Potter & Hepburn, 2003; Roberts & Sarangi, 2005; Silverman, 1999; 2011; Wetherell et al., 2001; Wooffitt, 2005)**.** Whilst CA pays attention to the structures of talk and how interactions proceed on a very fine linguistic level, TODA focuses on how talk is constructed and used functionally (e.g., how turns of talk are formulated with particular goals in mind) and how it is situated within a broader context (Roberts & Sarangi, 2005; ten Have, 2007; Wooffitt, 2005). CA and TODA methods complement and overlap since they both: 1) look at the function and sense-making of language in interaction, 2) focus on analysing sequences of talk as social actions, rather than reflections of cognitive processes, 3) pay attention to the detailed features of talk, and 4) are open to new discoveries within the data without having prior assumptions.

In looking at the activity of talk 'under a microscope', we begin to understand the 'how' and 'why' of language in interaction (Roberts & Sarangi, 2002; 2005; Silverman, 1999). The addition of contextual-based data sources further enriches these insights.

8.8.1.1 Transcription & translation of the video-recorded GC consultation data

Several decisions need to be made at the start of the transcription phase of interactional data. Decisions may include whether to transcribe the entire recording or only certain segments of the recording, what features to focus on (verbal and/or non-verbal), what transcription conventions to use, and whether to include written records of the original language, if choosing to analyse translated text (Penn & Watermeyer, 2018).

For the study, rather than focusing on the entirety of each recorded GC consultation, I have chosen to focus only on segments of the GC consultations that deal specifically with risk communication and how uncertainty occurred or was managed in those moments. The reason for this is that the GC consultations include large volumes of information, are lengthy and involve different types of discussions on topics other than risk and uncertainty. Analysing all the phases and aspects of talk in the GC consultations from start to end would have proved laborious, time-consuming, and not within keeping with the aim of the study.

Based on the literature read as part of the study, the majority of challenges related to risk communication and uncertainty in GC consultations typically occur during the information giving and decision-making sections of a consult. In the information giving sections, sources of risk are identified, and risk information is conveyed using different formats (Perrott, 2010).

Risk is also further discussed during the decision-making phase of a GC consult and patients must make decisions with limited information (Wessels et al., 2015). These moments may further be exacerbated by contextual influences (Pilnick & Zayts, 2014). I thus used this background knowledge as a guide when deciding which segments of the recorded consultations in the study to pay further attention to, in line with the underlying research objectives.

Although not strictly adhering to either approach, basic CA and TODA transcription conventions were used, as suggested by Roberts and Sarangi (2005), Peräkylä (2007), ten Have (2007), and Toerien (2014), to transcribe and analyse the selected segments of the video-recorded GC data. The CA transcription notations used are those typically used in CA research and were developed by Gail Jefferson (Heritage & Maynard, 2006). The transcriptions aim to capture details of talk as they happen in 'real-time', which includes pauses, interruptions, laughter, hesitations, intonation, filler sounds, overlapping speech and lengthening of words. Non-verbal aspects have also been documented (Heath et al., 2010; Mondada, 2007). Transcribed details also include what activities precede and follow the chosen segments (Mazeland, 2006; Silverman, 2011). It is important to note that although researchers using Jefferson CA-conventions utilise similar symbols and notations, this system of transcription continues to evolve based on different research interests or needs (Heritage & Maynard, 2006), thus may have slight variations from researcher-to-researcher. A descriptive list of the Jefferson CA notations used in the study can be found in Appendix B.

As there were no exclusion criteria regarding the language in which the GC consultations took place, GC consultations were recorded in English, Afrikaans as well as isiXhosa. Some of the genetic specialists in the study were first-language English speakers and some were first-language Afrikaans speakers. All were able to speak English, however not all were able to speak Afrikaans. None of the genetic specialists in the study were first-language isiXhosa speakers, nor were they able to speak isiXhosa. The language ability of the genetic specialist typically drove the decision in which language the consultation was conducted in, or whether an interpreter was needed. All genetic specialist interviews were conducted in English.

The majority (13/18) of the patient participants were first-language Afrikaans speakers and 5/18 were isiXhosa first-language speakers. Some of the Afrikaans and isiXhosa first-language patients, some were able to converse in English. However, two isiXhosa patients were unable to converse in English or Afrikaans and required an interpreter. In one consultation a formal

interpreter who works at the hospital in question was recruited before the start of the consultation as the staff at the genetic clinic had noticed that the patient was unable to converse in English or Afrikaans when she arrived at the clinic. In the other consultation that required an interpreter, it was noticed during the GC consultation that the patient was struggling to converse in English and could not speak Afrikaans. At this hospital there was no formal interpreter available, thus an off-site formal interpreter was arranged through a provincial-based telephonic interpreter service. Table 8.5 provides a summary of language use in the GC consultations.

Table: 8.5: Summary of languages spoken in the genetic counselling consultations

	English	Afrikaans	isiXhosa (in the presence of an interpreter)
No of GC consultations conducted in specific language	10	6	2

As all transcribed data was analysed in English for the study, the translation of the Afrikaans and isiXhosa consultations were necessary. For the translation process, rigour-based activities were conducted as suggested by Temple and Young (2004) and Penn and Watermeyer (2018). For each set of data needing to be translated, the following process was followed:

Translation process of the isiXhosa GC consultation data

I am personally unable to speak or understand isiXhosa. In instances where data was collected in isiXhosa, I employed two research assistants to aid with transcription and translation, both of whom are fluent in both English and isiXhosa. The two research assistants employed for the isiXhosa data specifically were postgraduate psychology students familiar in CA-based transcription conventions and interactional research.

I sat with the two research assistants and we watched the isiXhosa-based GC consultations together. Thereafter, appropriate segments were selected for translation and study as a collective. A discussion was regarding how best to approach the translation of selected segments to ensure consistency and rigour. A decision was made that 1) selected segments were listened to again independently, 2) selected segments were transcribed, and translated independently, taking note of interactional features, 3) translated data was then shared between the research assistants to cross-check each other's work independently, and, finally, 4) a meeting was held between the two research assistants and me to discuss findings and to resolve segments where incongruencies occurred.

Translation Process of the Afrikaans GC consultation data

As a native English and Afrikaans-speaker, I conducted all the segment selections and Afrikaans translations myself. To ensure rigour and the trustworthiness of my translations, I engaged a third assistant, namely an Afrikaans-speaking member of the HCRU who assisted in verifying my translations. This individual is also fluent in English and is also familiar with CA-based transcription conventions and interactional research methods. The Afrikaans HCRU member independently checked the translations after I had completed them, and we had a meeting to discuss the translated data and resolve any incongruencies. One of my supervisors, namely Dr Tina-Marie Wessels is a first-language Afrikaans speaker, thus was also able to cross-check my Afrikaans/English translations. Appendix C provides details of the research assistant non-disclosure agreements for the study.

It is important to highlight the complexity of translation in the study. Although avoiding bias in translation is important and the final analysed text should reflect the original spoken source, achieving neutral and exact translation can be difficult (Temple & Young, 2004). Cultural, linguistic and socio-political factors can further impact the interpretation and translation of information across languages as shown in South African healthcare studies (Kilian, 2013; Penn & Watermeyer, 2018; Solomon et al., 2012) where written and spoken versions of a language may differ, or where alternative words for certain English medical or scientific terms do not exist. I am thus cognisant that for the data collected and translated from Afrikaans or isiXhosa, personal interpretations and subjective meaning-making could have occurred, which may have altered the intended messages of the research participants. Involving research assistants to check and corroborate the translated data was thus an important step to help ensure accurate translations.

8.8.1.2 Analysis of the interactional data

To start the analytic process, I watched each video-recorded consultation in its entirety and generated rough summaries or content logs (Goodwin, 1994; Jordan & Henderson, 1995; ten Have, 2007). The content log for each consultation includes a family pedigree (a graphic representation of a patient and their family across several generations which shows various family relations and how a genetic trait may be shared between individuals), where the consultation took place, reasons for referral, who the participants are, interesting moments that stood out based on the research aims, contextual considerations, as well as recordings of the times and phases of the interaction. An example of a content log can be found in Appendix D.

I then used the content logs to select segments within each consultation that specifically dealt with risk and uncertainty information or conversations. Discussions on risk and displays of uncertainty occur most often during the information-giving and decision-making sections of the recorded consultations, as noted by Wessels et al. (2015).

Once all selected segments were transcribed (and translated where necessary), I read and re-read the selected sections. This initial reading can be seen as unmotivated exploring (Sacks, 1984; Schegloff, 1996). Psathas (1990) points out, however, that unmotivated exploring in CA-based research "is of course a contradiction or paradox since looking is motivated or there would be no looking being done in the first place" (p. 45). The idea of unmotivated exploring is thus to acknowledge one's intended focus but to be open to new phenomena. At this stage of the data analysis, I familiarised myself with the content of the selected segments, making initial notes regarding risk communication, moments of uncertainty and any areas in which context played a potential role.

From here I began to characterise the segments, paying careful attention to activities and interactional features including turn-taking, sequence organisation, repair and turn-design (ten Have, 2007) specific to risk communication and uncertainty. I also paid attention to TODA-focused themes such as framing, footing, hedging, rhetoric, neutrality, decision-making, face-work, and mitigation (Roberts & Sarangi, 2005).

To identify areas where risk is spoken about, I used the words 'risk' or 'chance' as guides on where in the consult to focus. In the consultation data, uncertainty is not overtly stated but is identified through associated words and speaker and responder turn-designs. To identify uncertainty in my consultation data, I have used previous interactional research focusing on uncertainty (Barr, 2001; Pilnick & Zayts, 2014; Sarangi & Clarke, 2002a; Steel et al., 2014, Stivers & Timmermans, 2016) as guidelines on how uncertainty is recognised in an interaction. These studies are described in Chapter Five but I re-highlight ways to identify uncertainty here. Words such as auxiliaries (e.g. could, might, maybe), adverbs (e.g. about, approximately) and filler words (e.g. uhm, uh, well) were searched for, as well as discourse strategies such as hedging, mitigation, and referal to other experts. Turn designs were also focused on particularly speech dysfluencies, re-starts, pauses and prosody (e.g. the rate, pitch as well as rising and falling intonation of a speaker's words and sentence construction). Collectively attention to these features in the consultations assist in identifying features of uncertain 'talk'. I also used

Wessels (2014) South African interactional GC study to assist in identifying different phases of the GC consultation (e.g. introduction, information-gathering, information giving, decision-making) to assist in grouping the different types of risk discussions had in each consultation.

Once looking at the various interactional features, I then curated collections. These collections are made up of either individual consultations or moments across several consultations that have similar interactional features (Drew, 2003; Toerien, 2014). Cross-comparisons within the collections have been used to demonstrate similarities, differences or unique consultations. The result is a compilation of patterns across the data. These patterns are further interpreted and elaborated to show that conversations do not happen randomly, but rather, are jointly constructed and influenced by surrounding conversational features (Curl, 2006).

8.8.2 Analysis of the genetic specialist interviews: Thematic analysis

Genetic Specialist interviews formed part of the talk-extrinsic data collected in addition to the interactional data from the GC consultations. The interview data provided a better understanding of the broader research context, i.e., the experience of communicating genetic risk and uncertainty in a South African healthcare context as told from the perspectives of the healthcare service providers in the field.

Thematic analysis (TA) was chosen to analyse the genetic specialist interviews. TA aims to find patterns of meaning across data sets to elucidate participants' experiences of a particular research topic (Braun & Clarke, 2012). Typical TA analytical phases include 1) familiarisation of the data, 2) development of codes (identifying and labelling areas of interest-based on the research aim and objectives), 3) creation of themes by collating and comparing identified codes, 4) reviewing the themes in relation to each other and whether they represent the data collected, 5) refining and naming the themes, and 6) writing up the findings. Results typically include data extracts or participant quotes to allow transparency, as well as existing literature to support or contextualise the results (Braun & Clarke, 2006). It is important to note that these suggested steps are not necessarily linear and that the development of codes and themes is an active process that evolves during data analysis (Braun & Clarke, 2006, 2012). Reflective practice and critical questioning are thus important aspects of the TA research process (Braun & Clarke, 2020).

TA has been used in many different areas of qualitative and quantitative research using different methodologies, epistemological standpoints and paradigms. TA can be used as both a deductive (a top-down approach which has preconceived theories about the data) and an inductive method (bottom-up approach in which the data is observed in an open manner with no prior assumption of what will be found) (Braun & Clarke, 2012, 2014). Due to its wide application in different areas of research, TA has been critiqued by some researchers in terms of rigour, coherence, and reliability, as well as failing to engage with philosophies and theories, especially when used in qualitative research (Levitt et al., 2018; Sparkes & Smith, 2009). More recent TA methodological papers have since aimed to reflect upon some of these critiques and to highlight the assumptions of TA as a method to create improved understanding (Braun & Clarke, 2019, 2020; Braun et al., 2019). Whilst most TA researchers aim to find patterns within data and follow similar steps as suggested in Braun and Clarke (2006), it is important to note that different versions of TA exist depending on what type of research paradigm one is working within, namely 'coding reliability' TA, 'reflective' TA and 'codebook' TA (Braun & Clarke, 2020). These three versions of TA come with different understandings of the data analysis approach and how it develops.

Coding reliability TA is typically used in deductive, quantitative research where stringent and accurate coding is necessary. After data familiarisation, early theme development occurs, and a structured and fixed codebook is created. This codebook is then used by multiple independently working individuals who then provide consensus on the identified codes as a form of inter-rater reliability. Reflective TA takes on a full qualitative approach where the subjectivity of the researcher is acknowledged, and a broader research team is not involved in comparing codes. Rather than being bias, or a threat to credibility, the researcher's own context and knowledge is seen as important to the data analysis. Themes develop over time as the researcher engages in the research process.

Codebook TA falls somewhere in-between the two previously mentioned TA approaches and makes use of elements of each. Researchers using codebook TA tend to develop early themes, a codebook or a framework to analyse the data and make use of some sort of coding reliability approach such as asking members of the research team for input and consensus. However, codebook TA acknowledges research subjectivity and the contextual nature of knowledge production. Codebook TA may also take on a more pragmatic approach than some pure qualitative approaches. For example, it may take into consideration predetermined information

needs (themes can be developed based on the responses to predetermined interview questions), the reality of time pressures to collect data, and the involvement of team members to meet research project deadlines (Braun & Clarke, 2020).

For the study, a codebook TA approach was utilised. I initially familiarised myself with the audio-recorded genetic specialist interview data by listening to the recordings and making initial preliminary summaries. The notes included length of the audio-recordings, date, place and who was present. I transcribed all audio-recorded data verbatim (since the functional purpose of language was not the focus here, a fine level of transcription was not necessary) and read and re-read it several times. Unlike the GC consultation data, all interviews were conducted in English, thus no translations were necessary. For each recording, I labelled areas and segments of interest into codes. These codes were collated to form themes and sub-themes. Themes were compared across all the interviews, pulling together perspectives or comments that are similar in nature, and making note of those that were unique. Unique findings were not overlooked but rather categorised into their own theme. I further compared and re-studied collected themes to eliminate overlap and to ensure that all ideas had been considered. Relevant quotes from the finalised themes and sub-themes are included in the results chapters to provide transparency to the 'voices' of the participants. Supporting literature is also included to contextualise the findings. Figure 8.2 gives an example of the coding process followed for the interviews. The figure is not an exhaustive representation of the codes, but rather an example of how codes were created.

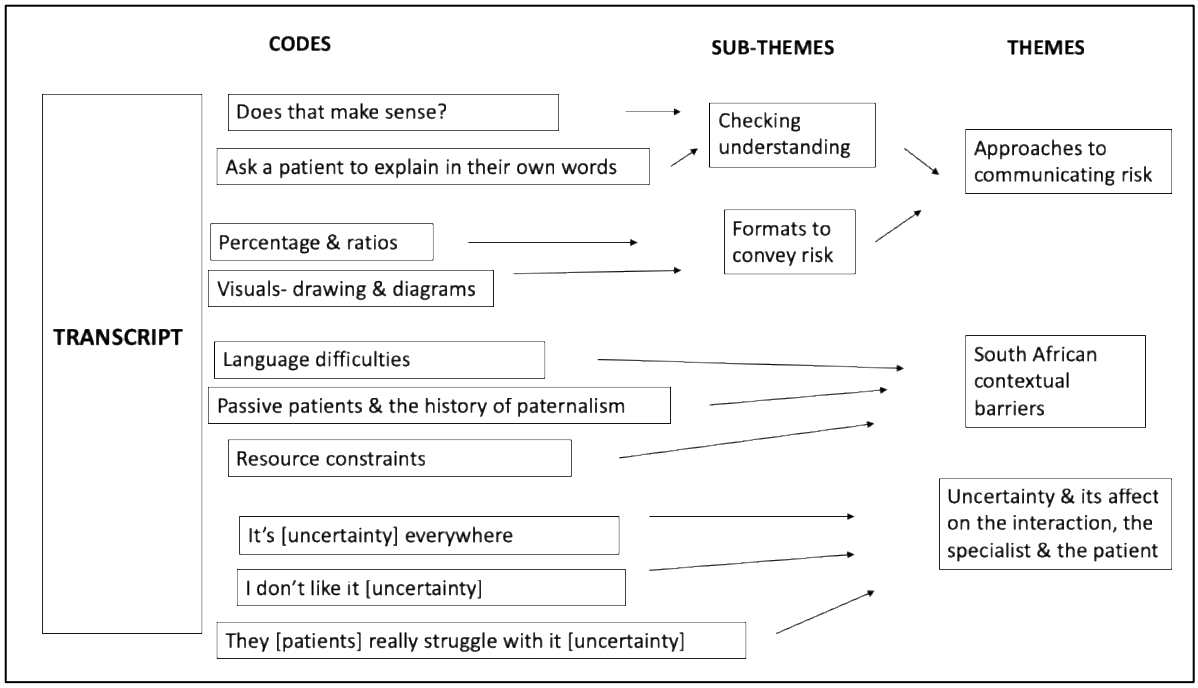


Figure 8.2: Example of the data analysis process for the interview data

8.8.3 Analysis of the researcher field notes: Content analysis principles

I wrote observational field notes whilst attending the research sites which include prenatal, paediatric and adult-focused genetic clinics. For each clinic, I made shorthand notes on-site and then wrote more comprehensive reflective notes after I had left the clinic. For each clinic I wrote all the reflective notes within 24 hours of attending the clinic, so that the recollection of the activities was still 'fresh' in my mind.

The field notes provide insight into the 'atmosphere' (Langewitz, 2007) of the research setting and activities including the hospital, the clinic, the staff, the patients, as well as the informed consent and video-recording procedures. During the period that I made field notes, I sat in the genetic clinic patient waiting-area making observations of my surroundings as well as the clinic- and ancillary- activities. I also used this time to reflect on my experience entering the hospital and clinic space on the day as well as my first impressions. Spatial, sensory, and verbal aspects, attention-grabbing moments, patient and staff responses towards my presence, as well as participant recruitment experiences were documented. Summaries include thoughts about the informed consent processes as well as information pertaining to recorded GC consultations (e.g., where the consultation took place, patient demographic information, the reason for referral, and which genetic specialist was involved). An example of field notes taken at one of the research sites can be found in Appendix E.

I have borrowed suggestions made by Mays and Pope (1995) to analyse the field notes. Although very detailed, descriptions alone do not necessarily provide explanations, Thus, some sort of analysis is necessary. However, in addition to the activities captured from the research sites, field notes also entail personal or emotive details about the researcher. Analysing detailed descriptions captured through observational field notes can thus be challenging, since much of the details are subjective. Mays and Pope (1995) suggests looking for patterns across different sites or cases, similar to a content analysis process, and making notes of differences or similarities. Through this analytic process, the researcher can start making sense of what has been observed.

For the study, thick descriptions of the research sites, the participants and activities were conducted using a somewhat systematic process regarding what I decided to take note of. All notes were carefully written down and expanded on in my reflective pieces for each clinic. I carefully read all my notes, taking into consideration similarities and differences from each clinic. I used my understanding of the atmosphere and context from each clinic as a backdrop to better understand the data that I had collected in the GC consultations and the genetic specialist interviews. The field notes were thus a useful source for triangulation of data in the study. Specific notes related to clinic protocols or particular participants are included in the results sections of the thesis, where relevant. I have also incorporated my field notes into reflective and ethical pieces, which have further informed the analysis of the data sources. The field notes assisted me in both emotionally and professionally navigating the many 'hats' I have worn during this research process, which includes researcher, genetic counsellor, mother and patient. I include the reflective and ethical pieces in my discussion section of the thesis.

8.9 Triangulating the data

Due to the complex nature of the study, as well as the use of a blended research approach, triangulating the data sets and findings was an important process to tie the analysis and interpretation of the data together. Triangulating data sources and findings is especially useful in complex healthcare studies, where some sources highlight the micro-details of a setting (e.g., video-recording of interactions), whilst others pay attention to macro contextual nuances and a greater understanding of personal perspectives and experiences (e.g., researcher observations and interviews). Several international (Hindmarsh & Pilnick, 2002; Lambert & Loiselle, 2008;

Mahmoodi, 2015) and local (Etheredge et al., 2017; Penn & Watermeyer, 2018; Wessels, 2014) healthcare studies have made use of and supported, triangulation of 'micro' and 'macro' data sources.

I am aware that using different data sets and analysis approaches may create tensions in epistemic stance and the 'lenses' in which the results from the data sets are viewed (Fielding & Fielding, 1986; Kuper et al., 2008; Reeves et al., 2008; Sim & Sharp, 1998). To deal with these tensions, researchers may create methodological hierarchies in which one method dominates over another (Razum & Gerhardus, 1999). Ranking one method over another has, however, been challenged since methods (e.g., quantitative and qualitative) should be viewed as different, rather than competitive (Patton, 1999). Thus, in studies where different data sources are used, triangulation should not be seen as a method of validation, but should rather be considered as a way of combining the strengths of different sources to create a deeper understanding of a phenomena (Denzin & Lincoln, 2011; Lambert & Loiselle, 2008; Patton, 1999). I thus use triangulation of different resources and methods in the study as a way of providing a holistic sense of genetic risk communication and uncertainty in a South African GC context. The talk-extrinsic data, namely the genetic specialist interviews and researcher field notes, are there to complement and provide further richness to the consultation data collected.

Whilst I have analysed the GC consultations, the genetic specialist interviews and researcher field notes separately, all data sets are jointly considered when deliberating the results and arguments in the study. Figure 8.3 and Figure 8.4 illustrate how the various methods and data sources have been brought together through triangulation.

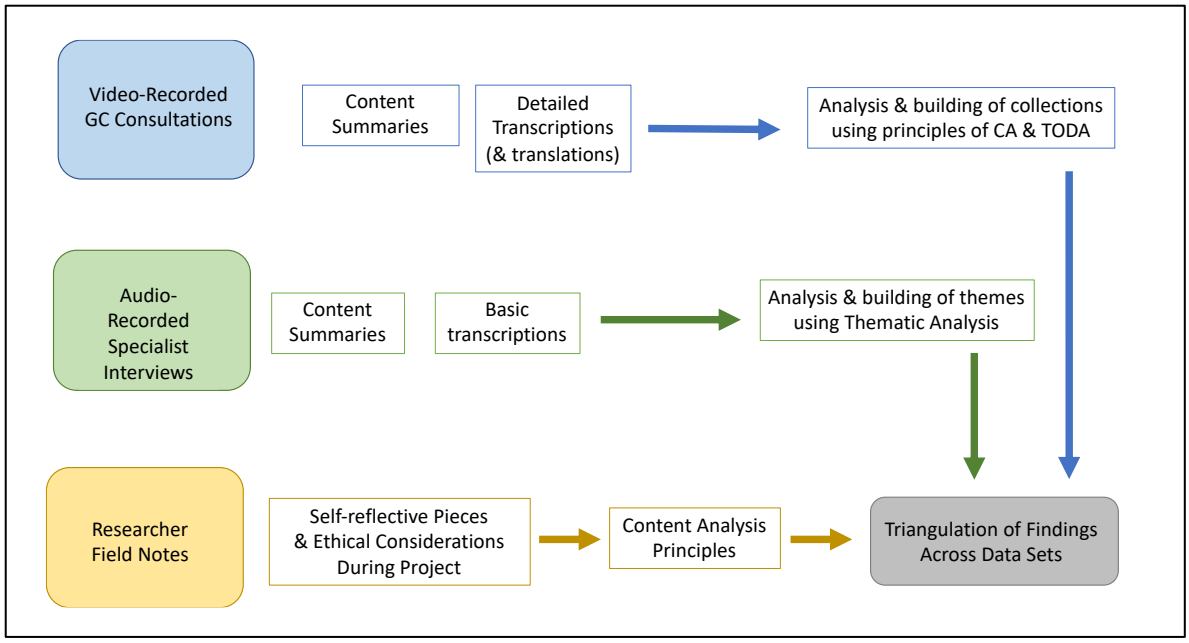


Figure: 8.3: Analytical framework of the study

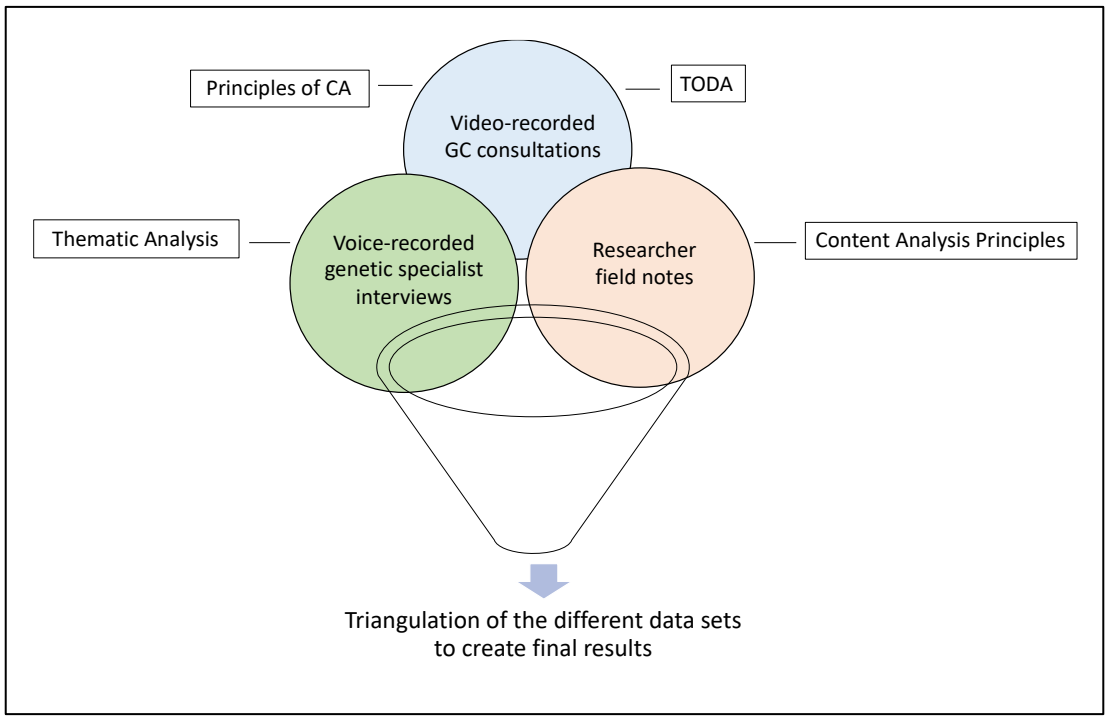


Figure 8.4.: Sources of data, analytical approaches applied to the data & how results have been triangulated

8.10 Data saturation & sample size

Data saturation considers the richness and depth of gathered information. One of the biggest challenges of qualitative research is knowing when an analysis is complete and how much of the analysis to include within a write-up that comprehensively represents and supports the

arguments made. Difficulties in deciding when data saturation has been reached may also occur due to qualitative research being a circular, rather than a linear, process which requires revisiting of data (Jørgensen & Phillips, 2002). In addition, data in qualitative studies can vary in source and size, further complicating data saturation decision-making (Fusch & Ness, 2015; O'Reilly & Parker, 2013). Data saturation in qualitative research is thus not guided by numbers, but rather is dependent upon whether or not the research question has been sufficiently addressed, whether or not available resources have become exhausted, what is already known about the topic, whether or not the phenomenon has been addressed from different perspectives, and how complex the phenomenon under investigation is (Fusch & Ness, 2015; Morse, 2015; O'Reilly & Parker, 2013).

For the consultation data, 18 video-recorded GC consultations were collected and included in the study. Video-recordings of human interaction can capture a multitude of complex visual, acoustic and activity-based features, e.g., speech, gestures, facial expressions, environmental descriptions, activities and related tools. Making it one of the most effective, yet complex, social science methods (Knoblauch et al., 2006). Even a small sample can produce a rich source of data for various research interests and questions (Heath et al., 2010), on both a micro- and macro-interactional scale. The recorded GC consultations in the study have many visual and verbal features and capture a range of different types of genetic conditions, risk discussions and moments of uncertainty. When watching and analysing the GC consultation data and transcripts after my fourth site visit, I realised that I had enough variation and detail within the recorded interactions to identify several different patterns across the data set.

For the audio-recorded genetic specialist interviews, I only had access to local genetic specialists based at the selected research sites. The number of interviews conducted was thus guided by the number of willing participants. At the time of data collection, there were 15 available genetic specialists across the three research sites. 12 genetic specialists were included as part of the study. Of the 12 genetic specialists included in the study, 9/12 participated in the recorded GC consultations, whilst all 12 participated in the interviews. One of these genetic specialist's agreed to be interviewed but did not want to participate in the video-recorded consultations, citing discomfort with the video-recorder. The other two additional genetic specialists who were interviewed agreed to be video-recorded but were not included in the consultation data as they were not available at the clinics on the days that the video-recorded

data was collected. Analysis of the genetic specialist participant interviews shows overlaps and repetition of themes and findings, suggest that data saturation has been achieved.

Observational field notes were also collected from each genetic clinic that I attended. Some genetic clinics I only attended once (e.g., the adult clinic), whilst others were attended more than once (e.g., some prenatal and paediatric clinics). This was coincidental and dependent on the dates that I happened to attend at each research site. For clinics attended more than once, I did not make notes for each visit. Reasons for choosing to only record notes from one visit was to respect the staff at the clinic and not to be too intrusive whilst they conducted their work activities.

8.11 Trustworthiness

Part of a researcher's responsibility is to explain their thinking throughout the research process. This step ensures a transparent audit of the research, it enhances readers' confidence in the researcher's ability, and creates greater dependability on the study findings and claims (Guba & Lincoln, 1994; Jones & Bugge, 2006). Rolfe (2006) describes this process as a shift of judgement of quality from the researcher to the reader. Even though the researcher is the producer of knowledge, the research and knowledge are only considered worthy if the reader, as the consumer, says it to be. Describing, documenting, and verifying the research process of any study is thus crucial.

To gauge whether research has been done 'well', a study is expected to meet a certain set of standards or follow particular guidelines. These standards, however, differ greatly depending on the chosen methods (e.g., quantitative versus qualitative) as well as the epistemological outlook (e.g., positivism versus relativism) (Jørgensen & Phillips, 2002; Rolfe, 2006) of the researcher. Furthermore, several terms are used within research such as validity, truth, rigour, replicability, triangulation, saturation, and credibility, and may each have different interpretations depending upon the methods and phenomena of study (Koch & Harrington, 1998; Nixon & Power, 2007; Rolfe, 2006).

Qualitative research is complicated and sub-divided by the wide range of theories, research focuses, data collection methods and analytical approaches. Despite sharing particular epistemological and ontological stances, qualitative researchers have found it particularly

difficult to reach agreement on what is considered 'good research' (Rolfe, 2006). Many qualitative studies have used the notion of 'trustworthiness' described by Guba and Lincoln (1989; 1994), which is thought to include credibility, transferability, dependability, confirmability and authenticity.

Credibility is thought of as a form of internal validity (Schwandt et al., 2007) where continuous research engagement as well as reflexivity, peer debriefing and member-checking ensures that the researcher is mindful of their own biases and that they provide a truthful representation and interpretation of the participants, their activities, and their experiences (Cope, 2014; Polit & Beck, 2012). *Transferability* can be considered a form of external validity (Schwandt et al., 2007) where detailed documentation of the research process, participants and findings allows for study replication. However, Cope (2014) highlights that many qualitative studies are context-specific, thus findings may not be generalisable to other research settings. Clear documentation of the research process and thick descriptions (Guba & Lincoln, 1982), however remain important parts of qualitative research. *Dependability* overlaps with credibility and transferability in that the researcher needs to ensure that the results and study processes are consistent and transparent so that the study can be reproduced using similar research parameters or conditions (Koch, 2006; Polit & Beck, 2012).

I have provided clear descriptions and documentation for the motivation for the study, the aims and objectives, the research design, the different data phases and processes, as well as my findings, arguments and conclusion. All stages and processes of the study have been thoroughly discussed with my research supervisors on a continuous basis. All transcripts in the study have been carefully created since the quality, uniformity and detail of transcripts can greatly affect the reliability of data (Silverman, 2004). The transcripts have been checked by my research supervisors as well as international experts in the field who visited the HCRU during the time of my PhD. Data that has required translation has been translated independently (by me or one of the research assistants) and cross-checked by other individuals to ensure uniformity.

Confirmability is the assurance that the results represent the participants findings or views and not the researcher's own biases (Cope, 2014; Polit & Beck, 2012). How data is presented e.g., participant quotes and explanations on how interpretations and conclusions are drawn can assist in providing surety that findings are not solely the researcher's bias opinion (Cope, 2014; Schwandt et al., 2007). *Authenticity* refers to the way participant experiences, or activities are

represented by the researcher and whether this is done faithfully and fairly (Cope, 2014; Polit & Beck, 2012; Schwandt et al., 2007). Further guiding principles such as openness, careful data collection, thorough data analysis, consideration of all data and theoretical frameworks in the development of individual ideas, as well as reflexivity also considered key (Burns & Grove, 2001; Smith et al., 2009).

Although independent data analysis was conducted as part of the study, colleagues in the field and my supervisors have been consulted regularly during the research process to assist in validating or critiquing the findings. Some colleagues independently coded sections of the interactional and interview data to further ensure the trustworthiness of the interpretation of the data, as suggested by Seale and Silverman (1997). Experts in the field of CA, applied CA, and TODA have also been consulted, as necessary. I also made extensive use of valuable feedback received from local and international conferences and workshops, where preliminary data from the study was shared.

Furthermore, extracts from my field notes, self-reflections, interactional transcripts, and the interview data are included in this write-up to reveal 'authentic' participant moments and to provide transparency, as suggested by Mays and Pope (1995) in their description of field note analysis and validity. Self-reflective pieces, together with regular discussions with my supervisors has also helped me to be aware of my own biases. My supervisors are from two different academic fields, one being a health communication expert based in Humanities, whilst the other is a genetic counsellor based in Health Sciences. Having access to their two different perspectives when discussing challenges in the research process and the interpretation of the study's results has ensured that I do not view my research through a singular lens.

8.12 Reflexivity

Reflexivity and self-reflection have formed an important part of the research process. Being reflexive can be thought of as turning the attention on oneself, particularly to notions of moral values, background, social and political standpoints, personal beliefs as well as describing difficulties and decisions regarding data collection, analysis, and inclusion (Ellis, 2009; Finlay, 2002; Mertens, 2014; Parker & O'Reilly, 2013). According to Rolfe (2006), this allows for self-critique and appraisal throughout the research process, as well for readers to understand

dilemmas and decisions deliberated during the research process when viewing self-reflections in a researcher's write-up.

During the study, I became particularly aware of my position as a researcher, a genetic counsellor**,** and more recently, the additional positions of being a parent and a GC patient myself. I reflect further on these positions in the discussion section of this thesis in Chapter Twelve. I have also reflected on the atmosphere of the research sites, my feelings and reactions when observing the details and activities of a particular space, the challenges of using video-recordings as a research method (particularly in a South African GC context), how participant recruitment occurred and what some of the challenges were during data collection and analysis. Previous research has noted that conducting research within formal institutions, such as hospitals can highlight both good and bad aspects of service provision, as well as social and moral dilemmas. Observations of both positive and negative aspects of a research space may challenge a researcher in their responsibility for what/whom they choose to write about, and how they choose to represent these elements in the write-up (Bantjes & Swartz, 2018; Kouritzin, 2002; Penn & Watermeyer, 2018; Wolfinger, 2002). Self-reflection has helped me gain perspective on these challenges throughout the research process for the study.

As a GC who had previously worked in similar clinical environments in Johannesburg, many of my observations at the research sites and in my data were familiar to me. However, viewing GC and the genetic clinics through a 'researcher' lens has given me a heightened sense of awareness, sensitivity, and appreciation of the daily activities in these types of settings. Although a practice typically related to social science research, reflection is important for all medical-related research, both qualitative and quantitative, in which sensitive data is commonplace. Self-reflection may be particularly important for health researchers using video-recordings (Lomax & Casey, 1998), and has certainly played a significant and constructive part in this research process.

8.13 Ethical considerations

8.13.1 Ethics clearance

Ethics clearance for the study was obtained from the relevant genetic clinics, the three hospitals, and two tertiary academic human research ethics committees (HRECs) (University of the Witwatersrand Ref: M160318, University of Cape Town Ref: 440/2016). Appendix F details the approvals that were required from various institutions, clinics and committees, the

relevant ethical clearance certificates, and permission letters, as well as the participant informed consent documents and information sheets.

8.13.2 The use of video-recorders in research

Video recordings are not without risk. Previous literature has reported on the risks of using video recording for research purposes. Risks include the challenge of ensuring anonymity for participants (and research sites) (Parry et al., 2016), the potential of recording sensitive data and whether participants can comprehend the associated risks when consenting to participate (Block et al., 1985; Butler, 2018; Hargreaves, & Peppiatt, 2001), as well as the problem of data protection and how to present research results in an appropriate manner (O'Reilly et al., 2011). Using video-recorders in healthcare settings can be particularly invasive, especially for patients placed in vulnerable positions. This has been highlighted in studies including gynaecological discussions (Fatigante & Orletti, 2014), palliative care consultations (Pino et al., 2017), emergency resuscitations of neonates (Gelbart et al., 2009), or when participants share personal stories of abuse and poor social circumstances (Butler, 2018). Healthcare practitioners may also become concerned when their practice is placed under scrutiny by video-recording their conduct, which may result in negative career or legal consequences (Parry et al., 2016). Furthermore, video-recordings may create psychological distress for researchers when viewing sensitive data (Kiyimba & O'Reilly, 2016).

The use of video-recorders in certain social contexts can also raise concerns. In countries such as South Africa, where there is a complex history of social inequality, abuse of power, racism and disease-related stigma, patient-participants (Penn & Watermeyer, 2018), and healthcare staff (Evans, 2010), may be particularly wary of researchers wanting to use video-recordings. These concerns may make governing ethics and institutional committees less willing to allow the conduct of video-based health research (Iedema et al., 2006; Liu et al., 2015). Although these reasons may be strong justifications for not using video-recordings, researchers such as Wiles et al. (2011) and Lomax (2015) have in contrast highlighted that video-based research is necessary. Video recordings in institutionalised settings encourage important debate regarding service improvement, especially for marginalised individuals, who are typically excluded from research for the very reason that they are considered vulnerable. Avoiding marginalised or vulnerable individuals may be considered unethical.

Researchers using video-based research methods may find themselves facing ethical dilemmas in practice and in varying contexts. For example, how does one ensure that no harm will occur when unable to fully predict what will occur during a video-recording? The challenge of pre-empting ethical concerns during the planning phase of a study has been previously reported (Beauchamp & Childress, 1994; Boden et al., 2015; Mertens, 2014). There is also continued discussion about what comprises informed consent, since its interpretation may differ across healthcare settings. How consent is designed (e.g., timing, amount of information discussed, documentation, and how a person's ability to consent is determined) (Grady, 2015; Parry et al., 2016) as well as if, and how, contextual factors (e.g., language barriers, literacy, as well as cultural and social determinants) are considered (Grady, 2015; Penn & Evans, 2010; Watermeyer & Penn, 2008) remains important. Researchers using video-recorders need to be particularly cognisant of these matters during participant informed consent procedures.

8.13.3 Participant recruitment & the informed consent process

During my application to the respective Ethics Committees for the conduct of the study, I explained that I was interested in the non-verbal and verbal communicative aspects that occur in routine practice. I thus motivated for the use of video-recorders to observe GC consultations. I explained that patient and genetic specialist participants were not expected to perform any specific tasks, but to conduct the consultation as it would typically occur in everyday practice. Thus, the informed consent documents that were created pertained only to the use of a video-recorder during a particular consultation, not the actual clinical information that was to be discussed. Moreover, since consultations conducted in languages other than English was anticipated, I ensured the use of an interpreter in such circumstances to help facilitate and check understanding during informed consent. Ethical issues pertaining to the voluntary nature of the study, protection of raw data, confidentiality and anonymity was also addressed and approved. The details of how these listed concerns are addressed are described in the participant informed consent documents and information sheets (available in Appendix F).

Despite the lengthy procedure of obtaining clearance (due to the multiple sites and governing bodies involved), at the time of acceptance, the only concerns raised were that I obtain written permission from each of the research sites before collecting data and that I indicate what would happen to the video-recorded data after study completion. No concerns were raised by the two HRECs regarding informed consent procedural challenges, the potential for obtaining sensitive data, or the intrusive nature of video-recording. These ethical concerns and tensions have,

however, been realised during and after data collection, which has called for considerable reflection on my part as the researcher. I reflect on the ethical dilemmas of the study and its methodologies in more detail in the general discussion in Chapter Twelve. Some of the ethical reflections are also included in a recently published paper:

Scott, M., Watermeyer, J., & Wessels, T-M. (2020). Video-recording complex health interactions in a diverse setting: Ethical dilemmas, reflections and recommendations. *Developing World Bioethics, 20, 16-26.* https://doi.org/10.1111/dewb.12226 [ISI accredited].

For the reasons described above, one area that was particularly challenging in the study was the informed consent process during patient participant recruitment. Especially for patients who were attending the genetic clinic for the first time and who had very little expectation of what GC was or what was going to be discussed in their consultation. During the analysis process of recorded interactions, I felt that some patient participants only fully comprehended the seriousness of their referral during the recorded GC consultation when they received unanticipated and/or upsetting news. As I did not conduct retrospective consent in addition to obtaining consent before video recording, there may have been a possibility that participants felt some degree of regret regarding being video-recorded. As the researcher I felt concerned about this possibility, but since I had not obtained contact details for the participants, I had no way of contacting them to offer retrospective consent. Although participants were told they could withdraw from the study at any time and given my contact details, none contacted me after data collection to decline participation. It remains unclear whether some patient participants would have chosen to participate in the study had they known what was about to transpire in their consultation. Although anonymising the data mitigates this matter to an extent, these complexities remain an issue within video research that captures naturally occurring activities in sensitive healthcare settings.

During the recruitment process, before approaching any patient, I would discuss the patients' reasons for referral with the genetic counselling team and would also assess the patients' visible state (I did not approach any patients who were visibly distressed before their consultations). Collectively, we would decide whether it was appropriate for me to approach a patient before their GC consult to ask if they would be interested in participating in the research. When approaching potential patient participants, I did not indicate what their GC consultation would be about or what details would be discussed. I only asked to record their consultation and

confirmed that there was no expectation of them or the genetic specialist in the consult to do anything different to what they would have normally done in the absence of a video-recorder.

Drawing on my prior experience as a genetic counsellor, I knew that some of the impending consultations would likely be challenging and sensitive for patient-participants. Even though I could not predict all the upcoming conversational details. To respect counselling procedures at the clinic (e.g., the suitable time and space to break bad news and the qualified assigned person to do so) as well as patient-specialist confidentiality, it would not be appropriate to divulge or probe the patient about any clinical or personal information before their consultation. Especially not when done in a hospital corridor or outside of a formal consultation. Despite this, I wanted to ensure informed consent and did not want the participants to feel unprepared for the upcoming recorded consultation.

The difficulties that can occur in decision-making when an act is in part considered morally right, but also morally wrong has been previously described (Beauchamp & Childress, 1994). Furthermore, many ethical guidelines tend to focus on informed consent as a process that occurs in a vacuum between the researcher and the participant, without fully acknowledging how they and their interaction may be shaped by surrounding context, individuals or systems (Fisher, 2013). In contexts where participants are deemed vulnerable due to their inability to understand research information, who may feel unable to decline participation, or who feel that they have no control over what happens to their information, researchers need to be particularly aware of issues of power (Gray et al., 2017). Although many ethical guidelines promote autonomy, and equal power between a researcher and a potential participant during informed consent, prior evidence (Grady, 2015; Gray et al., 2017), as well as the study suggests that this aim often falls short in practice.

Such ethical dilemmas were particularly encountered during the study in the prenatal genetic clinics for first-time consultations. In these clinics, only complex and high-risk pregnancy cases are seen. Patients are often given devastating news about a previously assumed healthy pregnancy, to which they now must make risk-based decisions under uncertainty. Some of which involves the offer of termination of pregnancy. Owing to these difficulties, prenatal GC research, especially psychosocial or interactional studies, is known to be an under-studied area (Pilnick & Zayts, 2014). Due to this, I was apprehensive about receiving informed consent to video-record these prenatal consultations. This feeling of unease at the point of informed consent has been described in previous qualitative health research (Mattingly, 2005).

Furthermore, considerations regarding the benefits of conducting the research and whether it outweighed the unintended consequences that emerged because of participation also had to be considered.

This unease also arose in one of the paediatric clinics where, for example, the mother of a new-born baby was unaware before her first-time consultation at the genetic clinic that her child had a diagnosis of Down syndrome. Although the referring healthcare practitioner was aware of the diagnosis, this had not been explained to the mother. When inviting the mother to be a participant, I was unaware of how little she knew about her daughter's diagnosis at the time. What made this session even more difficult, was that the mother was an isiXhosa woman, who was unable to speak English. Despite the recruitment and presence of an interpreter during the informed consent procedure, and during the consultation, there was a sense, whilst observing the recorded video, that the genetic specialist was unable to sufficiently provide psychological support to the mother due to the existing language barrier. For the same reasons, the mother could not discuss her concerns in depth. This, together with the lack of forewarning from the referring healthcare practitioner felt unfair and upsetting to watch. Upon reflection, I felt that perhaps I had committed the very same injustice by not appropriately preparing the mother or other pregnant mothers in the prenatal clinics.

Possibly excluding data from the study, where patients became upset during the consultation, was discussed with the research supervisors and I spent several weeks deliberating the ethics of including such data in the final cohort. One prenatal consultation was extremely upsetting to watch and I at times felt like a voyeur. Even if participants have provided consent, a researcher must decide against video-recording if they feel that any harm may occur to the participant through doing so (Block et al., 1985). After much reflection, I chose to include these sensitive consultations in the data. What happened during the consultation reflects the South African context and how interactions may play out in local genetic clinics, making it invaluable for research purposes.

Although not an entirely neutral medium of research, I do not believe that video-recording necessarily added further distress to the consultations and that the events that occurred would have happened anyway in its absence. As distressing as it may be to observe and analyse these difficult areas of GC, understanding how genetic specialists adjust their information giving and pay attention to patients' psychosocial concerns in these highly sensitive conversations is

critical for understanding how care is achieved through communication. Using this research approach for this study could itself be contributing to the study's framework of care. Even though collecting sensitive data from vulnerable individuals may be ethically questionable, not allowing their stories to be heard may further disempower or marginalise participant groups (Liamputtong, 2007; Lomax, 2015; Pino et al., 2017; Smith, 2008).

8.14 Conclusion

In conclusion to this chapter, I re-highlight the study's use of a bricolage-inspired research framework that makes use of a blended analytical approach that draw from different micro- and macro-level data sources and analytical methods. The different sources are necessary to understand the multiple threads and patches that make up the complex and rich quilt that represents a holistic view of the communication of genetic risk and the management of uncertainty in a South African healthcare context. The chapter provides a detailed overview of the research design, reasons for choosing approaches, details on the research sites, inclusion and exclusion criteria, the participants, as well as the data collection and analysis processes. The chapter also describes ways in which authenticity and academic rigour has been upheld and highlights the ethical considerations in this study.

CHAPTER 9A: INTRODUCTION TO RESULTS & DISCUSSION

The findings of this study reveal the complex nature of GC risk communication and uncertainty management within a South African healthcare context. Data suggests that genetic risk information is challenging to discuss since there are several types of risks that need to be identified and conveyed during GC consultations. Furthermore, there are different formats used to convey risk information in attempts to assist patient comprehension. Results shows that this communication process is complex and is impacted by differing patient needs and genetic specialist and patient agendas. The study results further show that genetic specialists may struggle to balance their roles of remaining neutral and imparting risk information.

The study results show that there are several moments of uncertainty that occur when discussing genetic risk information that has to be interactionally managed. Some of the uncertainty is owed to the incomplete nature of genetic risk knowledge such as universal and personal uncertainties, whilst GC context and GC professional guidelines are seen to further impact interactional uncertainty. Individual patient characteristics e.g., provision of unclear personal or family information, poor patient health literacy, language mismatches and passive patient behaviour particularly create interactional uncertainties. The genetic specialist interviews provide further insight into how the local genetic specialists experience uncertainty during GC consultations and how they attempt to manage it.

Figure 9.1 provides an overall summary of the factors thought to influence South African genetic risk and uncertainty communication, according to the findings from the consultation data, the interview data and observational field notes used in this study.

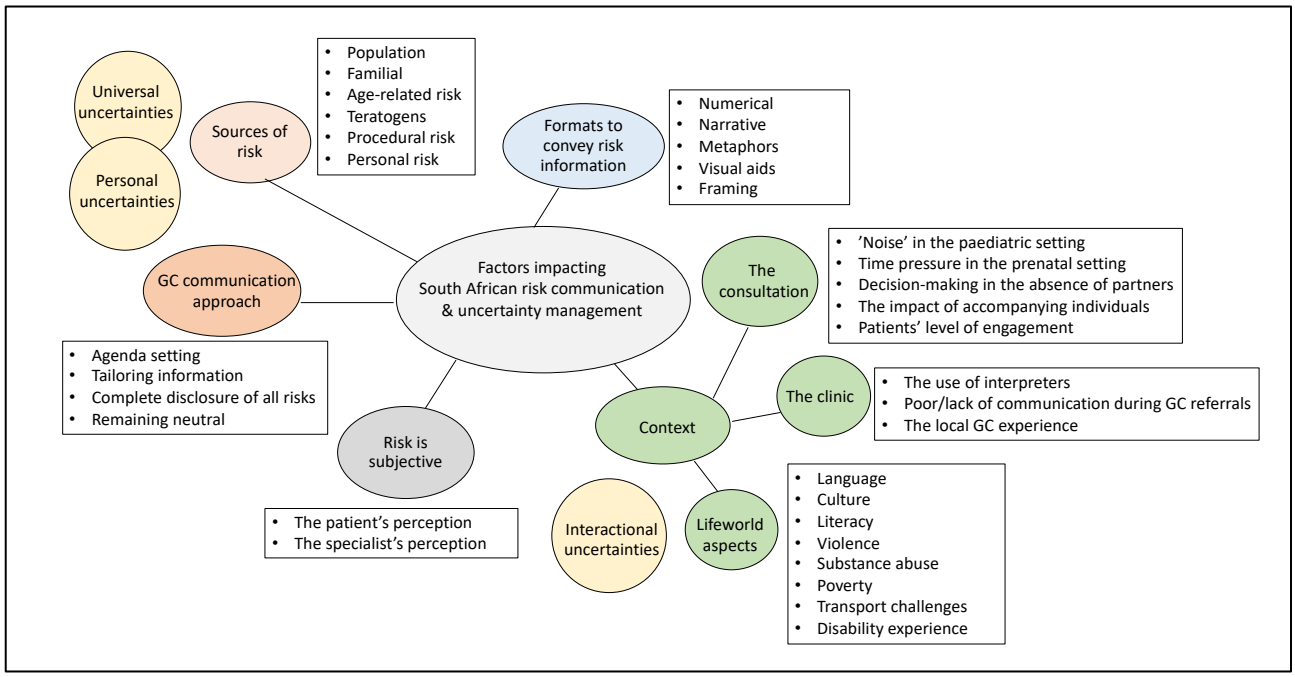


Figure 9.1: Factors thought to impact South African risk & uncertainty communication

The results and discussion chapters that follow present and discuss the identified features of local genetic risk communication and how moments of uncertainty are dealt with interactionally. My results are presented in three chapters and are accompanied by discussions and relevant supportive literature:

The remainder of *Chapter Nine* is largely descriptive, and centres on the risk content in GC consultations. The descriptions and examples used here aim to 'set the scene' to show what types of risk are discussed in local GC consultations, as well as what formats local genetic specialists deploy to convey risk information to patients. The results here demonstrate not only the volume and variety of risk information that could potentially be discussed in a GC consultation but also the many approaches that are considered. The examples presented in *Chapter Nine* have not been analysed in-depth using interactional methods, as this is not the intended purpose of the chapter. Some examples in the chapter include 'lite' interactional analysis to support some of the arguments made in this chapter. A more thorough interactional analysis is reserved for the data presented in *Chapter Ten* and *Chapter Eleven.*

Chapter Ten presents the different layers of uncertainty identified in the study. Layers of uncertainty include universal uncertainties, personal uncertainties and interactional uncertainties. The chapter also provides key examples of contextual factors that impact risk

communication and how these impact interactional uncertainty. The chapter also provides evidence from the genetic specialist interviews on the communicative impact of diagnostic uncertainty, and how genetic specialists reportedly experience interactional uncertainty.

Chapter Eleven highlights the practical challenges of achieving GC professional outcomes and how this creates challenges in managing uncertainty. I present two challenges observed in the data. The first focuses on the challenge of balancing agendas, tailoring risk information according to an individual patient's needs, and ensuring that a patient is informed. The second challenge deals with GC's central tenet of non-directiveness, and how differing patient and genetic specialist risk perceptions may impact neutrality during a GC consultation.

For the results and discussion chapters the reader is encouraged to note the following:

1. The study comprises a large volume of data from various sources. Not all of the captured details are presented in the results chapters (for example those shown in Figure 9.1), as many of the details are beyond the scope of this study. I have chosen to present and focus on the main risk and uncertainty findings from this study, paying particular attention to the video-recorded GC consultation data. I use genetic specialist interview quotes and field note observations to further support my arguments made, together with relevant literature.

2. The data is presented in an interwoven manner, rather than as separate sections for each source used. The presentation supports the triangulated approach I have followed to gain a holistic sense of South African genetic risk and uncertainty communication using different data sources.

3. For the transcripts from the video-recorded GC consultation data:
    1. Since only the sections on risk and uncertainty were transcribed in detail, rather than full consultations, the ordering of line numbers in the transcripts do not present the entire consultation from the first utterance to last. Rather the line numbers present different sections that were selected and transcribed separately. In instances where two examples are presented from the same consultation, the transcript line numbers are not necessarily sequential.

2. For GC consultations conducted in another language, each transcript excerpt shows the original language (either Afrikaans or isiXhosa) in the first line of each numbered line, and the translated English (written in italics) in the second line of each numbered line. The translated line does not provide a literal or exact syntactic translation. Rather each line has been translated in such a way that allows it to be understood in English.
3. Genders, ages and ethnicity of the specialists have been concealed to protect their identity. For the genetic specialists, I use the pronoun 'they' in the data analysis discussions/interpretations to conceal their gender.

4. The transcript excerpts and interview quotes that I have chosen to present in the next three chapters provide either typical examples of the patterns and themes identified in this study or findings that were unique. In the results, I differentiate which examples are typical and which are unique.

5. When referring to the word 'agenda' in the thesis I mean the expectations, goals and concerns of an individual (either the specialist or the patient) for the GC consultation.

CHAPTER 9B: THE TYPES OF RISK & THE FORMATS USED TO CONVEY RISK INFORMATION

9.1 Introduction

The following section pays attention to the type of risk information that is discussed during GC consultations, as well as what formats genetic specialists use to convey risk information to a patient.

9.2 Types of risk

Several types of risk are considered and spoken about during GC consultations. These either relate to *genetic*-based risk (e.g. population, familial, teratogen and personal-related risk) or *procedural*-risk associated with diagnosing and managing a potential genetic concern (e.g. the risks associated with performing an amniocentesis).

During the observed consultations either all or some, of the risks, were discussed with the patient. The types of risk discussed are dependent on the context of the consultation, the patient's agenda and the genetic specialist's discretion. Multiple risk considerations often led to lengthy and information-dense consultations, some of which lasted almost an hour. The types of risk identified in the consultation data are shown in Table 9.1. Each type of risk is further described and discussed below.

Table 9.1: Types of risk identified in the genetic counselling consultation data

Types of risk	**Type & no. of consultations in which pattern was noted**
Population	• 1/5 paediatric consults • 3/4 adult consults (all are HBOC referrals) • 0/9 prenatal consults
Familial	• 3/5 paediatric consults • 4/4 adult consults • 1/9 prenatal consults
Age-related risk	• 7/9 prenatal consults (the majority are AMA patients) • 0/4 adult consults • 0/5 paediatric consults
Teratogens	• 3/9 prenatal consults • 0/4 adult consults • 0/5 paediatric consults
Personal risk	• 18/18 consults
Procedural risk	• 9/9 prenatal cases • 2/4 adult consults (adult breast cancer-related referrals) • 1/5 paediatric consults

9.2.1 Population risk

Population risk is considered to be the number of people in the general population who are affected by a particular disorder, or who carry a certain genetic trait (National Cancer Institute, 2019). From the observations made, population risk was mentioned in one of the paediatric consultations and three adult cancer consultations. Example 9.1 and Example 9.2 are typical examples of the pattern observed in the consultations where population risk is mentioned. In both examples, the patients provide minimal responses to the conveyed population risk information.

Example 9.1

Case overview: ***Case 1- Treacher Collins syndrome (TCS)***

The case involves a Mixed-Ancestry couple who attended a follow-up consultation at a paediatric genetics clinic together with their three-year-old daughter who had a confirmed diagnosis of TCS. The couple had returned to the clinic as they were expecting another child and were concerned about possible recurrence risks. The mother was 11 weeks pregnant at the time of the consultation.

S = Specialist; F = Father; M = Mother

56	S:	So Treacher Collins what we (.) they found it in
57		(0.5)
58	S:	.Hhh well↑ because it's so variable (.) they found it in-between
59		(0.2) hhh ((starts to laugh at the child who is knocking over toys on the desk))
60		(0.5)
61	S:	**one in ten and fifty thousand people (0.2) ok**
62	M:	°Yes°
63	S:	**Worldwide (.) it happens across the world (.) All sor- (.) all**
64		**places (.) all (.) countries (.) everywhere (.) one in- (.) one in**
65		**ten thousand is highest↑ number we got (.) one in fifty thousand is the**
66		**lowest↑ number we got (.) so its very difficult to tell exactly↑ how**
67		**many people are born with it**
68	F:	((small head nod))
69	M:	((small head nod))

Example 9.2

Case overview: ***Case 3- Predictive counselling for inherited breast & ovarian cancer (HBOC)***

This case involved a 66-year-old female patient and her 45-year-old daughter. Both were of Mixed Ancestry and had no history of cancer. However, they had been referred to GC as they had a family history of breast cancer and a *BRCA2* gene mutation had been identifed in a close family member. This placed the patient and her daughter at risk to potentially have the same mutation. Carriers of this genetic mutation have a higher risk than the general population to develop certain cancers such as breast (61%) and ovarian cancer (17%) (Kuchenbaecker et al., 2017). The patient had undergone genetic testing and was attending the consultation to receive her result.

77	S:	Okay (0.2) So we know↑ that **cancer is actually really↑ common in**
78		**our population**
79	P:	((small head nod))
80	S:	The stats (.) for any↑ person at (.) uhm (.) over their lifetime
81		is **about 1 in every 3 people will develop cancer** (0.4.)
82	D:	((head nod))
83	S:	So it's quite↑ common
84	P:	Mm ((head nod))

In one genetic specialist interview, a genetic specialist indicates that they do not find population risk information helpful unless the patient has a higher literacy level (Example 9.3).

Example 9.3
S12: "I don't often give population risks to be honest. I don't find them very useful, except when the patient is very uhm… of a higher educational background."

Although population risk is often used as a comparison to patient's own risk in GC consultations (especially cancer GC consultations) (Hallowell et al., 1997), previous research has indicated that population risks may not be beneficial to patients. A survey conducted with 308 British women who were at higher risk for familial breast cancer showed that only 16% recalled the correct lifetime population risk (Evans et al., 1994), suggesting that risk information is more likely to be recalled if it is personally relevant or useful.

It is difficult to tell from the interactional data in which population risk is mentioned whether it is helpful to patients or not. However, it does not appear to be a popular source of risk to discuss with a patient as it is only referred to in 4/18 of the recorded GC consultations. Of the four consultations that mention population risk, three of the consultations were for HBOC referrals where population risk was used to contextualise a particular patient's risk for developing cancer when compared to the general population. The finding supports previous research which indicates that healthcare practitioners may convey population risks to contextualise a patient's personal risk (Ahmed et al., 2012; Edwards et al., 2013; Hunt, et al., 2006).

9.2.2 Familial risk

Familial risk is commonly discussed in GC consultations, particularly in the paediatric and adult genetic clinics. Familial, or hereditary risk is the risk conferred onto an individual by virtue of them sharing similar genetic make-up to other family members. Genetic traits are passed on from generation to generation between blood relatives, thus if one close family

member has been identified to be at risk, or been diagnosed with a genetic condition, these risks may be relevant to other family members too. Hence genetic risk is considered something that affects 'self' as well as 'family', including future children (Mendes et al., 2018). The inclusion of familial risk in genetic risk assessments are demonstrated in the genetic specialists' responses when asked what the term 'genetic risk' means (Examples 9.4 and 9.5).

Example 9.4
S2: "I suppose it's the risk that a person has, to having inherited a genetic mutation that's maybe running in the family that will predispose them to a condition…ja the same goes if a couple comes to you and they want to know the recurrence risk for them... their offspring."

Example 9.5
S3: "Genetic risk is like, the first word is the 'genetic' so it's a genetic marker, inherited and passed to the family and in which it can cause risk for others… So, the 'genetic risk' it's a term itself that we explore for that individual, for progeny, for family members to be at risk for a certain condition."

Interpreting familial risk is included in the NSGC's definition of GC practice which is: "*Genetic counselling is the process of helping people understand and adapt to the medical, psychological and familial implications of the genetic contributions to disease"* (Resta et al., 2006, p. 79). Risk in GC can include discussions about self, immediate family members, intergenerational and future family members (Gaff et al., 2007; Mendes et al., 2018).

In the data, the familial risk was mentioned in 3/5 paediatric consults, all (4/4) of the adult consults and one (1/9). Familial risks are highlighted by genetic specialists (Example 9.6) and also foregrounded by patients (Example 9.7).

Example 9.6

Case overview: ***Case 18- Predictive counselling for inherited breast & ovarian cancer (HBOC)***
This case involved a 31-year-old man of Mixed Ancestry who had been referred for GC as his younger sister had recently been diagnosed with breast cancer, as well as a *BRCA2* genetic mutation at the age of 29. The patient had no history of cancer and was the father of twin girls. He had been referred to the genetic clinic to discuss his risk of carrying a similar *BRCA2* mutation identified in his sister (his risk was 50%). If he carried a *BRCA2* mutation, it placed him at an increased risk of developing several cancers. The session was conducted in English, the patient's second language.

S= Specialist; P= Patient

14	S:	Because your dad's side of the family is really↑ big (.) he's got
15		lots of brothers and sisters and none of them have cancer (.) but
16		there is a family history on your mom's side
17	P:	Alright

		((head nod))
18	S:	So↑
19		(0.5)
20	S:	We think that that possibly came from your mom's side of the family
21		**(0.4) now↑ (0.4) every time↑ that you have children (0.2) I said**
22		**you get 50 percent from mom (.) and 50 percent from dad hey**
23	**P:**	**((head nod))**
24	**S:**	**So if your sister looks like this (0.2) she got this from your ((holding up palms of hands))**
25		**mom or your dad (0.2) we know↑ that your mom or your dad looks**
26		**like this (0.3) now every time that they had you guys**
27		**(0.5)**
28	**S:**	**it's fifty-fifty (0.3) which one they give ((pushing each palm away from their body separately to demonstrate allele inheritance))**
29	**P:**	**Mm**
30	**S:**	**Okay (0.3) so for each one of your siblings (0.4) flip of a coin ((making palm into a fist and flicking thumb up and down to demonstrate flipping of a coin))**
31		**(0.3) okay (.) did it land heads or tails**
32	**P:**	**Mm**
33	**S:**	**So that's why we're talking to all of you guys (0.2) because all↑**
34		**of you have a chance to carry the same↑ change that Anne* [has]**
35	P:	[.hhhh] ((widens eyes and leans back))
36		(0.3) even my prostate thingy?
37	S:	(0.4) °Even your prostate°
38	**P:**	**.hhh wha:at! ((widens eyes in shock and leans back in chair))**
39		**(1.0)**
40	**S:**	**You say "wha:at" (0.2) what do you mean by that? ((demonstrates how patient leant back during his response))**
41	**P:**	**What (.) like like quite a shock ((his arms are folded across his chest and he rubs his one hand over his face))**
42	**S:**	**It's a shock for you to hear [that]**
43	**P:**	**[Ja (.) Ja (0.3) I know↑ that (.)**
44		**like men can**
45	**S:**	**Ja**
46	**P:**	**But to know that I can that whole uh (0.3) error message (.) hh ((forming fists with his hands and placing his them together))**
47		**hhh**
48		(0.5)

In Example 9.6 above taken from the middle of the consultation, the specialist indicates the hereditaty cancer risk that the patient is exposed by virtue of his family members (in this instance his sister) having a history of cancer.

Evidence for the patient's unease with the newly imparted risk information is seen in lines 38-47 where he demonstrates discomfort through fidgeting and self-touching (Street & Buller, 1987), laughter (Haakana, 2002; Sacks, 1992) and direct verbal confirmation that the information (i.e. having a potential 'error message' in his DNA that may cause cancer) is shocking to him. The example highlights familial risk implications and how the risk

information may be both upsetting and novel to patients since they may not have considered *themselves* to be at direct risk for a disease that has affected a different family member. Familial risk information is thus relevant to patients.

In Example 9.7 below, I refer again to the same consultation shown in Example 9.2. The example shows how the potential risk to other family members may be a motivator for some patients to undergo genetic testing. Some patients risk-related decisions may thus be socially motivated, with their risk concerns extending beyond only themselves.

Example 9.7

Case overview: ***Case 3- Predictive Counselling for HBOC***

This case involved a 66 year-old patient and her 45-year-old daughter. Both were of Mixed Ancestry and had no history of cancer. However, they had been referred to GC as they had a family history of breast cancer and a *BRCA2* gene mutation had been identified in a close family member. Confirmation of a *BRCA2* mutation in the family confirms a diagnosis of HBOC in the family. The patient and her daughter are at risk to have the same mutation. Carriers of a BRCA2 or an HBOC-related genetic mutation have a higher risk than the general population to develop certain cancers such as breast (61%) and ovarian cancer (17%) (Kuchenbaecker et al., 2017). The recorded consultation was a follow-up consult where the patient had previously undergone GC and genetic testing and was attending the follow-up consultation to receive her result.

S = Specialist, P = Patient, D = Daughter

218	**S:**	**So today↑ we have $good news for you$ (0.2) hhh hhh** **((looking at mother and starts to smile))**
219	P:	$Okay↑!$ HHH HHhh [hhhh] ((big smile and hearty laughter))
220	D:	[Hhh hh] ((looks away))
221	S:	It means (.) it means
222	P:	Mm ((head nod))
223	S:	It means you inherited this↑ one (.) and you didn't inherit this
224		one ((using their hands to demonstrate allele inheritance))
225	P:	Okay ((big head nod))
226	D:	((head nod))
227	S:	So you're not at (0.2) you're norma:al↑ population risk (.) to
228		get breast cancer [(.)] and any other cancer↑
229	P:	[Mm] Yes:s ((head nod))
230	D:	((head nod))
231	**S:**	**But it's not associated with the family**
232		**(0.5)**
233	**P:**	**$My children is safe!$** **((smiling and looking at her daughter))**
234	**D:**	**Hhh hh[hhh hh]**
235	**S:**	**[Hh hhh]**
236	**P:**	**$My children is safe!$ Hhh hhh**

		((big smile))

In Example 9.7, the genetic specialist conveys the patient's negative test results. The patient is not a carrier of a *BRCA2* pathogenic variant, meaning that her personal risk is reduced, and by extension, her children's risks are also reduced. The patient's response to her negative test results are not only jubilation for herself, but she repeatedly mentions that her children are 'safe' (lines 233 and 236). The patient's interactional gestures demonstrate that genetic risk has implications not only for the patient but also for broader family members and that it brings about strong emotions for patients. The relief felt by the patient and their prior uncertainty about whether they carry a pathogenic *BRCA2* mutation or not, is demonstrated both by verbal indications (lines 233 and 236) and also their non-verbal behaviour (laughter, smiley voice and eye gaze towards their daughter) in lines 219, 233 and 236.

Similar patient behaviours to uncertain outcomes and potentially upsetting health results have been demonstrated in a recent paper by Watermeyer et al. (2020a) focusing on results giving for hearing status in local audiology consultations. Their study found that patients appear to come to the clinic with a set goal in mind, namely to receive their test results. Additional information provided during their consult may thus be deemed superfluous by the patient. When considering where in the consult the patient's negative test result was conveyed in the example above, the genetic specialist conveys the results in the middle (minute 11) of a 22-minute consultation. Before delivering the result, the specialist provides a summary of the condition, the risk implications and the genetic testing.

During the agenda-setting, the specialist offers the patient the choice of first receiving the test result and then having the genetic specialist summarise the information, or to do it the other way around. When the specialist receives minimal input from the patient and her daughter during the agenda-setting of the consult, the specialist makes the decision based on what they think the sequence of information should be. They indicate their decision to first summarise the information and then provide the result. Based on the patient's reaction to the results provided halfway through the consult, it suggests that this was the patient's main agenda for attending the consult and what they were really interested in hearing. Providing the result earlier in the consult may have been more useful to the patient. The remainder of the consult includes discussions on what the result means both from a risk perspective and emotionally, with the patient again reiterating that her children are safe. The patient's daughter also confirms

that she no longer requires genetic testing now that she is aware of her mother's negative results. Knowing whether genetic testing was necessary for the patient's daughter is a further agenda for the patient and her daughter which is only addressed at a later stage of the consultation, rather than upfront.

Research focusing on inherited cardiac conditions (Erskine et al., 2014), as well as inherited cancer syndromes such as Lynch syndrome (Esplen et al., 2007), Inherited Breast and Ovarian Cancer (HBOC) (Clark et al., 2000) and Li-Fraumeni syndrome (Lammens et al., 2010) all indicate that alleviating fears over the potential risks for children and other family members is a reason for individuals to pursue genetic testing.

The patient's overt emotional and verbal reaction in Example 9.7 above demonstrates that reducing uncertainty regarding the risk for children and other family members is also an important motivator for GC patients. It appears that the patient' primary agenda for the consultation was to receive their personal result and by extension to gain certainty on whether their children were at risk or not. Familial risk implications, particularly in cancer consultations may be important drivers for GC and genetic testing. Establishing patients' family-related agendas early on in local cancer GC consultations and responding to it in a patient-centred manner thus seems important to ensure effective communication and patient-centred care.

Although prenatal genetic consults typically involve pregnant women, hence discussions on the current and future health of offspring, broader familial discussions were only held in one (1/9) of the prenatal consultations. The reason broader family discussions were held in this particular prenatal case is that the pregnant patient had been referred as her husband and his children, from a previous relationship, were affected with osteogenesis imperfecta (OI). The diagnosis of OI in her husband placed their unborn baby at risk for OI. The prenatal case was unique in comparison to the rest of the collected prenatal genetic clinic data since most referred pregnant patients had no concerning family history and had been referred for a personal risk (e.g., AMA), or because a genetic risk had been identified in their unborn baby (e.g. a soft marker for a chromosome abnormality) during their pregnancies.

9.2.3 Age-related risk

A woman who falls pregnant over the age of 35 years is of advanced maternal age (AMA) and is at a higher risk to give birth to a baby with a chromosome abnormality (Lean et al., 2017). In South African GC services, pregnant AMA patients are typically referred during their second trimester to discuss their AMA risks and testing options which includes the offer of invasive testing (usually an amniocentesis). In this study, 6/9 of the prenatal consultations were referred because of AMA. Of the six consultations, two were referred for AMA risks alone, three were referred for AMA risks plus additional risks that were noted during fetal ultrasound screening, and one was referred for AMA and having had a previous baby born with Down syndrome (a syndrome caused by a chromosome abnormality). The AMA patients in the study were all counselled about the genetic risks involved which were dependent on their specific ages. Below is a typical example (Example 9.8) of one of the recorded AMA consultations.

Example 9.8

Case Overview: ***Case 5- Advanced Maternal Age AMA)***

The case involved a 42-year-old isiXhosa woman who was 15 weeks pregnant and referred for AMA GC. No soft markers were detected during her ultrasound scan prior to attending the consultation, nor did the patient have an significant family history to suggest other types of genetic risk.

S = Specialist, P = Patient

1	S:	So so as Megan said my name is Susan* (0.3)
2	P:	°hello Susan*° ((head nod))
3	S:	uhm and I'm from genetics
4		(0.5)
5	**S:**	**Did they say to you why↑ you needed to come see us?**
6	**P:**	**>The:ey told me< that they want to:o:o (0.4) see that there are**
7		**no:o abnormalities in the baby**
8	S:	((head nod))
9	**P:**	**since I'm over forty years.**
10	**S:**	**So basically they told you to come see us because you are older ((rapid head nod)) ((smiles))**
11	**P:**	**.hhh .hhh**
12	S:	Uh ok (.) the abnormalities (0.2) did you understand what they
13		meant↑ by that?
14	P:	Uh (0.2) maybe chi:ild has disabilities or=
15	S:	=ok. (0.4) So they didn't mention any specific disabilities or
16		anything like that?
17	P:	mm mm. ((head nod))
18	**S:**	**Okay. So basically ja (.) we're going to speak to you about (.)**
19		**because you're older↑ (.) there's more of a cha:ance that your**
20		**baby has got certain conditions ok.**
21	P:	Okay.

In the example above, the pregnant patient seems to have some understanding of why she is referred (she is older), but after some probing from the specialist, it becomes apparent that she does not know the specific details, or the risks (that being older places her baby at risk for having a genetic problem). A lack of patient awareness on reasons for referral appeared to be a typical pattern in the collected GC consultation data in this study, not only for the AMA consultations but for several other consultations. Example 9.9 and Example 9.10 from two genetic specialist interviews further supports the observation that patients in the public (state) healthcare system may not understand their reasons for referral to GC and are referred by other health practitioners.

Example 9.9
S1: "Sometimes the state patients are just told to go to genetic counselling, whereas the private patients tend to know why they are there."

Example 9.10
S12: "I think that it would be uh… in the great minority the ones that actually know why they have been referred. And I think that most people, I think most people just say: "You have to go to [hospital name] they will, they will tell you why."

Based on previous literature (Morris, 2013; Wessels, 2014; Wessels et al., 2015; Wessels & Koole, 2019), many locally referred GC patients either have no or limited, knowledge of what to expect in a GC consult and do not actively seek it out. Rather they are referred by other health practitioners who recognise the need for GC. AMA patients in the public health care system are typically referred by nurses, sonographers, fetal medicine specialists and obstetricians to prenatal GC clinics (Wessels et al., 2015). Furthermore, local AMA GC services are limited to tertiary facilities in Johannesburg and Cape Town (Bee, 2005; Wessels, 2014), which may impede the amount of GC AMA referrals due to geographic restrictions and may be reasons why many patients are unaware of the service and the genetic risks associated with AMA and conception.

Four of the genetic specialists reported in their interviews that AMA risk alone in the absence of additional risks e.g., fetal ultrasound findings or soft markers can sometimes be a problematic GC referral as it has the potential to create heightened levels of patient anxiety and uncertainty (Examples 9.11, 9.12 and 9.13). AMA counselling makes up a large portion of South African GC services. The interview examples further demonstrate how risk perceptions may vary between an AMA-referred patient and a specialist.

Example 9.11
S3: "If you look at the context here, a AMA lady with no soft markers, nothing that can tell any risk so far and the risk is even reduced because of the absence of uh all these markers and then counsel about: "You could also be at risk for Down syndrome" or other whatever other aneuploidies. I find it sometimes really challenging raising anxiety unnecessarily."

Example 9.12
S7: "Ja… so uhm I think… uh… it depends because at this clinic we see obviously severe fetal anomalies… and then it ranges from severe fetal anomalies to a woman who is 40 and she is here because she is AMA and the ultrasound doesn't show any abnormalities, would she like prenatal invasive testing? And I think patients sometimes might find that harder because there's just uh as soon as you bring them into a room like this and talk to them something has to be wrong, otherwise why are you there?"

Example 9.13
S10: "For example an AMA, I sometimes wonder if we don't stress them out more than we actually help. By telling them, there could this something, but I can't actually tell you, and you don't want to test. So now just be on your way and worry about it for the rest of your pregnancy."

Prenatal medical screening including genetic diagnosis and testing has previously been critiqued by researchers such as Lippman (1991), Katz Rothman (1988; 1993), and Rapp (2000) who argue that pregnancies have been reduced to scientific lenses that focus on assessing and eliminating risk, rather than acknowledging the emotional and psychosocial aspects of pregnancy. Other researchers share these sentiments, highlighting that modern-day pregnancies are especially scrutinised from a risk perspective and that minor uncertainties and further test offerings may be overtly highlighted by healthcare practitioners to avoid potential legal repercussions should a child be born with a genetic condition (Hashiloni-Dolev et al., 2019; Marchant & Lindor, 2013). These findings highlight that the 'voice of medicine' in AMA GC referrals may be prioritised above the patient's psychological and 'lifeworld' concerns.

In this study, some AMA-referred patients did not seem overly distressed by the AMA-risk information, whilst others did. In one AMA consultation where the findings are not in keeping with the other AMA consults, tensions rise between the patient and a genetic specialist during decision-making for invasive testing, as the patient believes that their AMA genetic risk is high, thus warranting invasive testing. The genetic specialist demonstrates interactionally that they disagree with the patient's risk assessment and that invasive testing is not warranted. An example of the unique AMA consultation is demonstrated in Example 11.6 in *Chapter Eleven* which deals with interactional tensions due to risk perceptions and the challenge for genetic specialists to remain neutral in the interaction. The AMA consultations in the study suggest that balancing risk perceptions, disclosure of risk and uncertainties, as well as being sensitive to patient's emotional responses to risk information are important communicative strategies in local GC practice.

9.2.4 Teratogen exposure risk

Discussions around teratogens, particularly in the prenatal genetic clinic consultations, were another layer of risk spoken about. Teratogens are environmental agents, substances or processes that can cause developmental harm to the fetus or embryo (Oxford Concise Medical Dictionary, 2007). These may include exposure to radiation; maternal infections, certain medications; and social or illicit drug use (Oxford Concise Medical Dictionary, 2007; The Merck Manual, 2006). Discussions around teratogen exposure occurred in most of the prenatal consultations when obtaining a pregnant patient's medical history.

Both alcohol and tobacco use in pregnancy are considered teratogenic as they can cause harm to the unborn baby (The Merck Manual, 2006). They are thus important risk topics during prenatal GC consultations. In a GC consultation, if a patient indicates that they have consumed alcohol or smoked cigarettes during their pregnancy, further probing from the specialist typically ensues to establish the amount, and gestational timing of the use. Probing questions are used to establish the potential extent of the teratogenic risk to the baby. An interactional example of questioning around teratogenic substance such as alcohol and tobacco use from one of the consultations is shown below in Example 9.14.

The example also demonstrates how establishing a patient's risk can be clouded in uncertainty due to the patient's vague recollection of details. The patient's lack of clarity may be forgetfulness or a strategy by the patient to mitigate their actions since there is social stigma related to the act of using alcoholic and tobacco-based substances during pregnancy.

Example 9.14

Case overview: ***Case 6: Osteogenesis imperfecta (OI)***

This case involved a 20-week pregnant, 31-year-old woman of Mixed Ancestry descent whose husband was diagnosed with osteogenesis imperfecta. The patient had been referred to the clinic to discuss her husband's condition and the inheritance risk to their unborn child. The session was conducted in Afrikaans, the patient's first language. In the transcript below the first line of each numbered line shows the original Afrikaans language used in the consultation (shown in italics) and the second line shows the translated English.

S = Specialist, P = Patient

10	**S:**	*Ok (0.3) en uhm (.) rook↑ jy of drink↑ jy?*
		Ok (0.3) and uhm (.) do you smoke↑ or drink↑?
11		(0.5)
12	**P:**	*Ja dokter*

		((nods head))
		Yes doctor
		((nods head))
13	**S:**	*Watter een?*
		Which one?
14	**P:**	*°Rook en drink°*
		°Smoke and drink°
15	**S:**	*Rook en drink↑?*
		Smoke and drink↑?
16	**P:**	**((head nod and slight smile))**
17	**S:**	*Uhm (0.2) Ok↑ (.) en hoeveel (0.4) uh*
		Uhm (0.2) Ok↑ (.) and how much (0.4) uh
18		(0.5)
19	**S:**	*Hoeveel drink jy?*
		How much do you drink?
20		**(2.0)**
21	**P:**	*Ek drink nie eintlik baie nie*
		I don't actually drink a lot
22		**(1.0)**
23	**P:**	*Ek sal nou dokter se (.) die waarheid dis miskien nou op 'n (.)*
24		*Saterdaggie (.) dan sal ek net ene dalk net ses (0.4) maar die*
25		*weekend was dit 'n family gathering (.) so nou nogal verder (.)*
26		*ek het (0.3) toe was dit nou te veel↑ weer*
		I'll tell you now doctor (.) the truth it's maybe on a (.)
		Saturday (.) then I'll maybe just one maybe just six (0.4) but
		this weekend it was a family gathering (.) so now quite a bit
		more (.) I (0.3) then it was too much↑ again
27	**S:**	**Mm**
28		**(0.5)**
29	**S:**	*En drink [jy elke]*
		And do you drink [every]=
30	**P:**	*=[Maar dis] nie baie nie*
		=[But its] not a lot
31	**S:**	*Nie baie nie?*
		Not a lot?
32	**P:**	**Mmhhmm**
33	**S:**	En drink jy elke↑ naweek?
		And do you drink every↑ weekend?
34	**P:**	*Nee dokter*
		((frowns))
		No doctor
		((frowns))
35	**S:**	*Nee?*
		No?
36	**P:**	**Mmhmm**
		((shakes her head))
37	**S:**	*En soos (.) soos by (.) by hierdie family gathering wat jy van*
38		*gepraat het*
		And like (.) like at (.) at this family gathering that you spoke
		about
39	**P:**	**Mm**
40	**S:**	*Hoeveel sal jy (.) hoeveel (0.2) uhm (0.4) wat het jy gedrink?*
		((placing hands in air))
41		*Wyn↑? Of bier↑? Of*
		How much will you (.) how much (0.2) uhm (0.4) what did you
		((placing their hands in the air))
		drink? Wine↑? Or beer↑? Or
42	***P:***	*.Hhh ja dis bier wat ons gedrink het (0.2) en een six-pack*
		.Hhh yes it's beer that we drank (0.2) and one six-pack
43	**S:**	*Ok (.) en hoe (.) hoe (.) hoe veel van dit sou jy self gedrink*
44		*het van daai?*
		Ok (.) and how (.) how (.) how much of that would you have drunk
		yourself?

45		**(2.0)**
46	**P:**	*As ek gedrink het (.) het ek net seker een gedrink vir die dag*
47		*(.) 'n six-pack of ses biertjies hhh hhhh ((laughs nervously and places her hand over her mouth))*
		If I drank (.) I probably only drank one for the day (.) 'n
		six-pack or $six little beers$ hhh hhhh ((smiley voice and laughs nervously, places her hand over her mouth))
48	**S:**	**Ok (.) ok**
49	**P:**	*As ek alleen gewees het (.) maar nou 'n tjommie daarby hhh*
		If I was alone (.) but now 'n buddy was with hhh hhhh
50	**S:**	*Ok (.) so dan (.) so divided by two?*
		Ok (.) so then (.) so divided by two?
51	**P:**	**((head nod and smile))**
52	**S:**	*Ok uhm (0.3) en het hulle vir jou enige iets gese↑ toe jy geboek*
53		*het oor (0.2) uh alkohol gebruik in swangerskap?*
		Ok uhm (0.3) and did they say↑ anything to you when you booked
		in (0.2) uh about alcohol use in pregnancy?
54	**P:**	*Yes:s (.) hulle het (.) wat kan↑ gebeur in die swangerskap ((nods head))*
		Yes:s (.) they did (.) what can↑ happen in the pregnancy ((nods head))
55	**S:**	*En wat het hulle vir jou daar gese?*
		And what did they tell you there?
56	**P:**	*Die kind kan (.) problems he (.) hart (.) longe (.) neusie (.)*
57		*oortjies of (0.3) daai's die was die suster vir my gese↑*
		The child can (.) have problems (.) heart (.) lungs (.) little
		nose (.) little ears or (0.3) that's what the sister told↑ me
58	**S:**	*Ok (0.2) so as jy alkohol drink as jy swanger is (.) is (.) uh*
59		*kan dit die baba se (0.2) wel al die organe af (.) affekteer*
		Ok (0.2) so if you drink alcohol when you're pregnant (.) it (.)
		uh it can affect the baby's (0.2) well it affects (.) all organs
60	**P:**	**((head nod))**
61	**S:**	*Maar meestal die brein↑*
		But mostly the brain↑
62	**P:**	**((head nod))**
63	**S:**	*So (.) dan is daar kans dat die baba leer↑ probleme he (0.3)*
64		*dis hoekom ons se (0.2) uh dis beter om glad nie te drink as*
65		*jy swanger is nie↑*
		So (.) then there is risk that the baby can have learning↑
		problems (0.3) thats why we say (0.2) uh it's better to not
		drink at all↑ when you are pregnant
66	P:	*Mm*

The patient's attempt to downplay their own potentially problematic behaviour of alcohol and tobacco use in pregnancy is seen in Example 9.14 above, specifically in lines 19-28 when the specialist asks about the quantity of alcohol consumption after the patient acknowledges that they have consumed alcohol during their pregnancy.

Previous research indicates that questions about a person's lifestyle and habits can be seen as a value-judgement, especially in healthcare interactions where discussions can take on a "moral tone" (Waitzinn & Britt, 1993 as cited in Sorjonen et al., 2006 p. 341). Alcohol and tobacco use, especially during pregnancy, can be frowned upon due to the risks involved to an unborn

baby. Patient self-reporting on these activities may thus be unreliable (Czeizel et al., 2004). Patients may also orientate their responses to a healthcare practitioner's questions about their habits in specific ways, to make the behaviour non-problematic as seen in the example above using mitigative language. Such patient responses have been reported in interactional studies focusing on healthcare provider's questions on alcohol use, sugar consumption, smoking and exercise (Sorjonen et al., 2006).

9.2.5 Personal risk

Personal risk assessments and discussions are a core part of GC and the reason most patients attend consultations. During GC consultations patients may be made aware of risks they have never previously considered or been aware of. They may also suddenly have to make decisions where several risks have to be taken into account. I again return to the consultation shown in Example 9.6 and use it in Example 9.15 to highlight how a patient may face multiple personal risks and how they are made aware of new risk information during a GC consultation.

Example 9.15

Case Overview: ***Case 18- Predictive Counselling for Inherited Breast & Ovarian Cancer (HBOC)***
This case involved a 31-year-old man of Mixed Ancestry who had been referred for GC because his younger sister had recently been diagnosed with breast cancer, as well as a *BRCA2* genetic mutation. The finding of a *BRCA2* mutation confirms HBOC in the patient's sister & places himself and other family members at risk for also having the same genetic mutation and developing HBOC.

In this consultation, namely Case 18, the following risk information is discussed with the patient:

- The *familial* risk to which the patient is exposed.
- The *personal* risk to which the patient may be exposed in the form of different cancers.
- The general *population risks* to develop cancer.
- *Procedural risk* information for cancer-related risk-reducing surgeries.

Not all the personal risks mentioned above are presented in Example 9.15 as the section is particularly long. I have thus chosen to present only the personal risk discussed during the consultation. The example further shows how this risk information can be novel, and potentially distressing to a patient.

S= Specialist; P= Patient

1	S:	So (.) I think to summarise (0.4) you have a 50/50 risk
2	P:	Ja ((head nod))
3	S:	to get this genetic change↑
4		(0.5)
5	**S:**	**If you DO carry this change (0.2) <you don't have cancer↑> (0.2)**
6		**but you have an increased↑ RISK↑ for cancer (0.4) and the ones**
7		**we worry about are (.) prostate for you↑ (.) and male↑ breast**
8		**cancer=**
9	**P:**	**=can you speak about that?**
10	**S:**	**Male breast cancer? (0.3) So↑ (.) men have less breast tissue than**
11		**women okay**
12	**P:**	**Mhmm** **((head nod))**
13	**S:**	**So (0.2) the risk for male breast cancer is 10 percent (0.3) okay (0.3)**
14		if you think about (0.2) 100 percent (.) you have a 10 percent risk for
15		**male breast cancer (0.2) so you need to make sure (0.3) you know for**
16		**women they say: "You must feel your breasts" (0.4) so you must** **((placing their hands on their chest))**
17		**still >feel your muscles< in your (.) in your chest and see if**
18		**there're any lumps**
19	**P:**	**((places both hands on his chest))**
20	S:	Okay (.) but we don't do any (.) check-ups necessarily for you↑
21		(0.2) because it's quite easy for you to feel (.) if you feel ((touching their own chest))
22		anything abnormal in your chest cavity
23	P:	Mmhmm
24	S:	Because there's not much too↑ feel
25	P:	Mmhmm
26	S:	Ok
27	P:	Mmhmm
28	**S:**	**OK (.) so you feel for yourself for male breast cancer (.) 10%**
29		**risk I said (0.2) and then prostate cancer risk is about 20 percent** **((wobbles palm of hand to demonstrate approximate figure))**
30		**(0.4) so that's the more (.) the one we worry about a bit more**
31		**(0.2) okay**
32		**(1.0)**
33	**S:**	**Then there are other↑ cancers** but there's no specific numbers that
34		I can give it (0.3) okay (0.2) so if you have (0.2) like a freckle ((rolls up the sleeve of their shirt and points to their forearm))
35	**P:**	**Mmm**
36	S:	that changes↑ (0.2) or that goes very dark (0.2 or gets bigger
37	**P:**	**((head nod))**
38	**S:**	**You need to °keep an eye on that° (0.4) there's a risk for melanoma**
39		**(.) a (.) a specific type of skin cancer**
40	**P:**	**Mmhh** **((head nod))**
41	S:	Okay (.) which can be quite dangerous okay (0.4) If (.) if you
42		have spots on you (.) just keep an eye on them okay
43		(0.5)
44	**S:**	**And then to just (.) in general↑ (0.3) if you're not feeling right**
45		**(.) go to the doctor and check it out sooner rather than later**
46	**P:**	**Mmhmm** **((head nod))**
47	**S:**	**Okay (0.2) because there are other↑ risks for other cancers (0.2)**
48		**like the pancreas (.) or**
49	**P:**	**Mm**
50	S:	things like that (0.4) ok (0.2) which we can't↑ test for (0.2)
51		and do surveillance for (.) but you must just (.) pick up symptoms

52		early (0.3) okay (.) but (.) but the ones that we worry about the
53		MOST (.) the highest risk ones (.) we can put checks in place like
54		[the prostate]
55	**P:**	**[so for me will] (.) they'll also cut my boobies?**
56	**S:**	**IF they found a breast cancer (.) yes:s**
57		**(0.5)**
58	**P:**	**No more boobies** **((touches his chest))**
59	**S:**	**Okay hh hhh** **((smiling))**
60	**P:**	**That's fine (.) no more boobies.**
61	**S:**	**So (0.2) they probably wouldn't cut your boobies unless you had a**
62		**cancer**
63	**P:**	**Ja I know! Hhh hhh**
64	**S:**	**Hhh okay**
65		**(1.5)**

In the example above the personal risk of developing prostate and male breast cancer is mentioned by the specialist in lines 5-8. The reason the patient is at risk to develop these cancers is due to a hereditary genetic *change* (i.e., a genetic mutation) (line 5) that has been identified in his sister. As siblings share DNA passed on to them from their parents, the patient and his sister may have similarities in their genetic make-up. Hence the patient may carry the same genetic change that has caused his sister to develop cancer.

Whilst mentioning the personal cancer risks to the patient, the patient interrupts the genetic specialist in line 9 requesting that the genetic specialist elaborate on the cancer risks mentioned. The rushed manner (indicated by = symbols in lines 8 and 9) in which the patient interrupts the speaker could indicate that this is novel information that has caused some uncertainty for him. The specialist goes on to discuss the risks of male breast cancer and the importance to do self-breast/chest examinations. In lines 28-48 the specialist discusses the other cancer risks associated with the cancer syndrome including breast, prostate, melanoma, and pancreatic cancer.

9.2.6 Procedural risk

The last source of risk that I will focus on is procedural risk. Procedural risk is considered the clinical or physical risk that a patient may be exposed to whilst detecting a genetic condition e.g., invasive prenatal genetic testing, or whilst managing a genetic condition e.g., performing risk-reducing surgery or invasive screening for an individual diagnosed with a genetic condition. In the consultation data, all (9/9) prenatal consultations included discussions on procedural risk, i.e., the risk of miscarriage due to an amniocentesis procedure. An amniocentesis was offered to pregnant women who were at risk for having a baby with a

genetic condition based on either their age, findings from a routine screening procedure (e.g., a prenatal ultrasound scan), their family or personal history, or a combination of these factors.

Of the adult and paediatric consultations, the procedural risk was discussed in 2/4 of the adult consults, and 1/5 of the paediatric consults. These consultations all had patients who had been referred for hereditary cancer (the adult consultations were for HBOC, and the paediatric consult was for Gardner's syndrome), where cancer risk-reducing surgery or preventative cancer screening was discussed.

Example 9.16 shows a typical example of a procedural risk discussion in a prenatal GC consultation.

Example 9.16

Case overview: ***Case 4- AMA & a previous child with Down syndrome***

This was a follow-up consultation involving a 41-year-old Mixed Ancestry woman, who was 20 weeks pregnant. During her previous consultation she had accepted the offer of an amniocentesis to detect whether her unborn baby potentially has a congenital condition, based on her age-related risk. However, the sample could not be processed by the laboratory as not enough fetal cells were present in the collected amniotic fluid. Thus, the fetal DNA concentration was too small to be able to produce a result. In the recorded follow-up consultation, the patient was offered a second amniocentesis procedure. The consultation was conducted in English, the patient's second language.

S = Specialist, P = Patient

182	**S:**	**So (0.3) with the amnio (.) comes the miscarriage risk**
183	P:	Yes
184	**S:**	**which we've spoken about hey (0.2) about one in five hundred (.) is**
185		**the chance to have a miscarriage .hh °ok°.**
186		(0.5)
187	S:	Would you be willing to take that cha:ance↑ (.) again?
188		(0.5)
189	S:	to get that result?
190		(1.5)
191	P:	I will ye:es:s↑.(.) because I don't (.) I'm positive as I've
192		said ne. (0.2) So I don't think there will be any miscarriages
193		(.) after the amnio. (0.2) Because there is only negative
194		feeling now in this process. ((uses hand in circular motion to demonstrate this session?))
195		(1.0)

Although several non-invasive screening tests exist such as first-trimester biochemistry testing, ultrasounds and more recently cell-free fetal DNA testing, invasive procedures such as amniocentesis and chorionic villus sampling (CVS) are still the most commonly used

diagnostic tests used to confirm the suspected presence of a congenital abnormality in an unborn baby. These procedures carry additional risks such as a miscarriage. Reasons for the pregnancy loss includes various independent maternal factors as well as operator experience (Salomon et al., 2019).

In South Africa, women of AMA are usually offered a second-trimester amniocentesis procedure (if available at the treating facility) (Bee et al., 2013; Constantatos et al., 2014). In Example 9.16 above, the patient has been offered a second amniocentesis procedure at 20 weeks' gestation to perform fetal genetic testing (after the initial procedure failed to produce a result). The specialist proceeds cautiously, foregrounding the risk of miscarriage.

Although not discussed in this consultation, it is also important to be aware of context and the additional risks that exist for some South African women such as maternal-fetal HIV transmission during an amniocentesis procedure. Although a patient's HIV status was never explicitly discussed in any of the collected consultation data, I am aware from my observational field notes and discussions with genetic specialists at the genetic clinics, that some of the pregnant patient participants in the study were HIV positive. South Africa is considered to have one of the highest HIV rates in the world with approximately 20.4% (7.7 million) of the general population living with HIV (United Nations Programme on HIV/AIDS [UNAIDS], 2020). Although HIV maternal-fetal transmission risk with an amniocentesis is greatly reduced if a HIV positive pregnant mother is receiving highly active antiretroviral therapy (HAART) (Constantatos et al., 2014), previous local GC research has indicated that a pregnant patient's HIV status further complicates decision-making and risk management in local GC consultations (Bee et al., 2013). These additional procedural risk discussions are important to consider when communicating risk and uncertainty in South African GC consultations.

9.3 Formats used to convey risk information

The results reveal the types of risks spoken about and also how risk information is presented to patients. The following section provides brief descriptions of the various verbal and non-verbal formats that genetic specialists used to convey risk information. This includes numerical information, narratives, metaphors, visual aids and framing. Table 9.2 provides a summary of the risk formats mentioned in the genetic specialist interviews, and Table 9.3 provides an overview of the formats identified in the consultation data.

Table 9.2: Formats to convey risk information as described in the specialist interviews

Themes	**Sub-themes**
Formats to communicate Risk	Numerical
	Narratives
	Metaphors
	Visual aids
	Framing

Table 9.3: Formats used to convey risk information as observed in the genetic counselling consultation data

Formats to convey risk	**Type & number of consultations in which pattern was noted**
Numerical	All (9/9) prenatal consults All (4/4) adult consults All (5/5) paediatric consults
Narratives	5/9 prenatal consults 0/4 adult consults 0/5 paediatric consults
Metaphors	7/9 prenatal consults 4/4 adult consults 3/5 paediatric consults
Visual aids	7/9 prenatal consults 4/4 adult consults 3/5 paediatric consults
Framing	6/9 prenatal consults 0/4 adult consults (0/5 paediatric consults

9.3.1 Numerical formats

The data shows that numerical formats are the most common way to present risk information with percentages and frequencies (ratios) typically used.

During the interviews, some specialists mentioned that when they use numerical formats, they prefer using percentages (probabilities) over ratios (frequencies). One specialist reported that lay individuals find percentages easier to understand (Example 9.17), and another specialist believed that ratios, rather than percentages, have a greater potential to create anxiety for patients (Example 9.18).

Example 9.17
S10: "I like percentages simply because I think, I feel that everyone knows that 100% is good.
Researcher: Ok. It's certain?
S10: Ja. So, if I say: "We are 99% sure this baby doesn't have Down syndrome". They at least know that is quite high."

Example 9.18
S12: "I had a patient last week and she got presented the risk of 1 in 16 for Down syndrome. And then when I put that in the positive and I turned it into a percentage… she was like: "What the hell was I worried about?" So just the different formatting of it made her think about it so differently. Whereas before she was a nervous wreck for 2 weeks."

The quotes above support previous risk literature that indicate that individuals may find frequency representations (percentages) of risk easier to interpret than probabilities (ratios) (Reimer et al., 2015). Furthermore, as demonstrated in Example 9.18, although the mathematical calculations of the risk information may not change across formats, the way individuals cognitively process and react to different formats may differ (Reimer et al., 2015). Two other genetic specialists indicated that they do not like using specific numerical figures (Example 9.19 and 9.20), rather choosing to use broad figures. The specialist felt that exact numbers are not helpful considering a patient's broader concerns.

Example 9.19
Researcher: "Any formats that you think are not so helpful?"
S9: "Mmm."
Researcher: "That you tend to stay away from?"
S9: "Well I think I would just be cautious about talking… just numbers. You know I think if one restricts that, you know, I'd be cautious. I think people often have a fairly limited kind of educational background. I don't know if, what their concept of number exactly is. So, I mean if I, I do use numbers, I don't, I avoid using very like very specific numbers, so if I get a number like 1 in 45, I will round it off to a kind of fairly round number and put it into some kind of real-world context basically."

Example 9.20
S5: "The hardest thing is I personally am not a huge fan of the very specific numbers particularly in the prenatal setting because I don't think it always really helps you. I think it gives you a perspective, but it doesn't really help you. Uhm in the prenatal setting I will often just talk in broad groups…
Researcher: Ok
S5: because I think if you say: "13% have a good outcome and 27% have an intermediate outcome…" you know I usually say that you know: "A SMALL number will do really, have serious complications, the MAJORITY…."
Researcher: Ok"

Other genetic specialists reported that they do not use numerical formats at all, as they personally do not find it useful (Example 9.21) or that patients do not find it useful (Example 9.22).

Example 9.21
S8: "So I'm not a numbers person, so because I'm not a numbers person, I try and think as non-number people."

Example 9.22
Researcher: "What do you think does not work? Are there any methods that you feel that you wouldn't use? Or you think there are methods available in the curriculum, but you find it quite [challenging]?
S4: I don't use, I don't use numbers in general."

To comprehend numerical risk presentations, it has been suggested that greater cognitive demands are necessary, including analytical and numeracy skills (Fagerlin et al., 2011; Stacey et al., 2017; Trevena et al., 2013). Previous studies have demonstrated that combining numerical risk formats with other formats that add context, e.g., narrative explanations, metaphors, analogies, as well as visual and graphical displays, allows numeric information to

become more tangible, without losing the accuracy that numerical evidence provides (O'Doherty & Suthers, 2007; Smerecnik et al., 2009; Trevena et al., 2013).

Numerical formats are used in all the recorded prenatal, paediatric and adult GC consultations. Also, across several consultations it is noted that both numerical formats e.g., a percentage and a ratio are used to convey the same risk information and that these numerical formats are typically combined with other risk formats (e.g., graphical displays or narrative explanations) when conveying risk information to a patient. Example 9.23 provides evidence of how numeric risk information is presented in two different ways in local GC consultations.

Example 9.23

Case overview: ***Case 10- Teratogen exposure (rubella infection)***

This was a prenatal case involving a 24-year-old Caucasian woman (labelled as the patient in the transcript) who attended a follow-up consultation at one of the prenatal genetic clinics at 19 weeks' gestation. She was accompanied by her mother (labelled as the patient's mother in the transcript). The patient was referred because at 16 week's gestation, she developed a rubella infection. Most rubella infections present with a mild rash in adults and children, with some affected individuals showing no symptoms. Rubella infections in early stages of pregnancy are considered high risk, as the infection can cause miscarriages, still births as well as fetal congenital abnormalities such as hearing impairment, cataract and cardiac complications. The risk for rubella embryopathy is highest in the first trimester of pregnancy, with the 2nd and 3rd trimesters having a lower risk, and less severe outcomes. Risk predications for an unborn baby can become difficult as the baby may become infected with rubella, but may not necessarily be affected i.e., develop complications from the infection. Alternatively, they may be infected, and severely affected. Testing or assessment for associated medical complications are not possible during pregnancy and can only be confirmed after birth (Motaze et al., 2020). The session was conducted in Afrikaans, the patient's first language. In the transcript below the first line of each numbered line shows the original Afrikaans language used in the consultation (shown in italics) and the second line shows the translated English.

S= Specialist; P = Patient; M = Patient's Mother

78	**S:**	*So as 'n mens uh (.) rubella kry baie↑ vroeg in die swangerskap*
79		*(.) so nou praat jy van (0.2) 8 weke van swangerskap (0.3) dan*
80		*is daar 'n baie hoer↑ kans miskien (.) 90 persent kans dat baba*
81		*uh (0.2) effekte sal kry en dat daardie effekte erg↑ is.*
		So if a person uh (.) gets rubella very↑ early in the pregnancy
		there is a very high↑ chance maybe (.) 90 percent chance that
		baby uh (0.2) will get effects and that those effects are
		severe↑
82		(0.5)
83	S:	*Maar so later dit word in die swangerskap (0.2) hoe minder is*
84		*die kans↑ (0.2) en ook hoe minder↑ is die effekte*
		But the later it gets in the pregnancy (0.2) the smaller the
		chance↑ is (0.2) and the lesser the effects will be
85	**P:**	**((slow head nod))**

86	**S:**	*Ok (0.3) uh op (.) 16 weke swangerskap (.) is dit min of meer*
87		*(0.2) 20 persent kans (.) of 1 uit 5 kans dat uh (0.2) dat baba aangetas*
88		*sal word*
		Ok (0.3) uh at (.) 16 weeks pregnancy (.) it is more or less
		(0.2) 20 percent chance (.) or one out of five chance that uh (0.2)
		that baby will be affected
89	**M:**	**((head nod))**

The interactional example above shows how risk information is presented in two different numerical formats by the specialist. The first format presents the information as a percentage (lines 79 and 87). In line 87 the named percentage (20%) is reformulated into a second numerical format, namely a ratio (1/5). In this example, even though both numerical formats were used, it is difficult to say whether the patient and her mother understood the information, or what their risk format preferences were as they remained largely passive in their response to the risk information. The specialist continued to use different numerical formats for the remainder of the consult and combined numerical formats with other types of risk formats.

9.3.2 Narratives

Narrative format is another identified way to convey risk information to patients. In the consultation data of this study, narrative format was used mainly in the prenatal consultations (5/9) where it was typically combined with ratio/frequency statements (e.g. 1/200) to contextualise the numerical figures given. The use of narrative format often occurred when discussing the risk of having a child with a congenital abnormality (e.g. Down syndrome), or when explaining procedural-related miscarriage risk for an amniocentesis. Narrative formats were not identified in the paediatric or adult consultation data.

Using narrative formats to contextualise frequency information was also mentioned in three of the genetic specialist interviews. Two of the specialist interviews mentioned using this format in AMA consultations to assist a patient in comprehending numerical information (Example 9.24). The other genetic specialist interviewed did not specify in which type of GC consultation they use a narrative format, but mentioned that they use narrative format to make numerical information 'real' to a patient (Example 9.25).

Example 9.24

S8: "I mean in the AMA scenario I'll say: "Uh if you're risk is 1 in 10, if we put 10 women in this little room and I stand on this table and I say to you: "1 of you 10 women that are standing in this room will have a baby with Down syndrome but the other 9 won't."

Example 9.25

S9: "So often I might say you know: "If you've got a 1 in 50 risk uhm that would be like saying well you know, that's 50 people, maybe a busload of people, so you've got a full bus of people, all in the same situation as you…" and uhm you know and you sort of try, try to make it real in some sort of way like that."

Example 9.26 demonstrates how a narrative format is used to contextualise numerical information that is provided as a ratio.

Example 9.26

Case overview *Case 12- AMA*

This case involved a 41-year-old isiXhosa woman who was 21 weeks pregnant who was referred for AMA counselling. No abnormalities had been detected on ultrasound, nor did she have any significant family history to suggest further genetic risk. She was accompanied to the consultation by her partner and they were counselled in English, the couple's second language.

S= Specialist; M= Mother (patient); F = Father (partner)

97	**S:**	**The chance that your↑ baby has Down syndrome is one out of forty**
98	M:	[Mmm]
99	F:	[((head nod))]
100	**S:**	**°Okay° (0.3) do you know what that means? (0.4) or not?**
101		**(1.5)**
102	**S:**	**If we say one out of forty?**
103		**(2.0)**
104	**F:**	**Not exactly hhh ((looking at specialist and then at mother))**
105		(0.5)
106	S:	Do you know what it means? ((asking mother))
107	**M**	**one out of forty? [(0.2)] It's not much:h (0.3) maybe it's 1 percent? ((shrugs shoulders))**
108	S:	[Mmm?]
109		(0.5)
110	**S:**	**Okay it's a bit more↑ than that**
111	M:	Is it?
112	S:	But (.) but ja (.) you're on the right (.) the right track.
113	M:	Mmm
114	**S:**	**It means (0.4) if we took forty women**
115	M:	[Mmm]
116	F:	[okay]
117	**S:**	**who were your sisters [(.) your] twin sisters (0.2) and we put**
118	**M:**	**[Mmm]**
119	**S:**	**them in this r[oom (0.3)] and they were all the same age (0.3)**
120	M:	[Mmm]
121	F:	[okay]
122	**S:**	**And they were all just as far pregnant as you**
123	M:	[Mmm ja ja]
124	F:	[((head nod))]
125		(1.0)
126	**S:**	**Then based on (.) based on only your age [(0.4)] one of them will**
127	M:	[Mmm]
128		**have a child with Down syndrome (0.4) and**
129	M:	[Mmm]
130	F:	[okay] ((head nod))

131	S:	And then we say forty minus one↑ [(0.3) the other thirty nine] they
132	F:	[is 39]
133	M:	[is 33 said]
134	S:	won't (.) their babies won't have Down syndrome
135	M:	[Ja:a okay]
136	F:	[Ok uh uh]
137	S:	that's what that means
138	F:	[Okay]
139	M:	[Mmm mmm] ((nods head))

In the example above a narrative format is used to aid in explaining and contextualising numerical risk information for a patient and her partner. The specialist initially presents the risk for the mother to have a baby with Down syndrome as a ratio (1 out of 40) in line 97. After providing the ratio, the specialist enquires whether the patient and her partner understand what the risk means (lines 100 and 102). The patient and her partner respond to the question with lengthy pauses (lines 101 and 103) which could indicate uncertainty in understanding. The father thereafter explicitly admits he doesn't understand the risk information (line 104). The specialist then asks the mother if she understands (line 106), to which the mother responds (line 107) by attempting to explain the numerical risk information provided by the specialist. Evidence that the mother's interpretation is incorrect is provided in line 109 and 110 where the specialist corrects the mother's interpretation by saying '*it's a bit more than that'* after an interactional pause.

The incorrect risk interpretation prompts the specialist to change gear and re-explain the risk information in a narrative format in lines 114-134. The narrative used involves a story where 40 pregnant women who are the same age as the patient are placed in a room together. Of the 40 women, one will have a baby with Down syndrome whilst the other 39 women won't have a baby with Down syndrome. This information is a reformulation of the one out of forty ratio risk information provided in line 97.

The findings from the consultation and the interview data, together with the previous literature findings indicate that narratives may be a useful format to explain risk information particularly when patients struggle to comprehend numerical figures in isolation.

9.3.3 Metaphors

Figurative language is another common way to convey risk information in GC consultations. For the purpose of this study, the term 'metaphor' refers to both similes, metaphors and any other forms of figurative language.

In 14/18 of the consultations, including 7/9 prenatal consultations, 4/4 adult consultations and 3/5 paediatric consultations, genetic specialists use metaphors as a way to explain risk. Genetic specialists incorporate metaphors into their discussions with patients either to explain molecular or procedural-related risk information or to compare feelings of uncertainty. In the interview data, six genetic specialists reported using metaphors to explain concepts of risk. Example 9.27 and Example 9.28 provides evidence from the genetic specialist interview data on how metaphors are used.

Example 9.27
S6: "Uhm also sort of to explain uhm chromosomes just you know packets of information or our body's instructions manuals or building plans. So things like that."

Example 9.28
S12: "I will always use a metaphor, especially when I do a 1 in 100 risk. I will sort of describe women in a room, or I'll say marbles in a bag."

The consultation data also shows that patients use metaphors, as seen in three of the consultations. The data shows that patients use metaphors to discuss emotion, clarify biological terminology and function, and to assist in describing physical symptoms. Unlike the metaphors used by specialists, none of the metaphors used by the patients in this study were directly related to risk or uncertainty.

Based on my own experience and training as a GC, metaphors are typically encouraged as a format by GC educators. Some figures of speech may be passed on through learnt behaviour such as junior students observing senior staff (T-M. Wessels, personal communication, April 2020) whilst others may be developed personally. It is difficult to establish whether metaphors are helpful to patients based on observations in this study given the patients' limited responses to these formats. However, researchers have reported metaphors to be helpful to explain complex concepts, especially to lay individuals. These include areas such as medical genetics (Sarangi, 2000; Ashworth et al., 2018), cancer studies (Krieger et al., 2010; Semino et al., 2017); mental health messaging (Lazard et al., 2016), infertility treatment (Palmer-Wackerly

& Krieger, 2015), general practice (GP) consultations (Skelton et al., 2002); as well as HIV and TB research (Penn & Watermeyer, 2018).

Despite their value, the dangers of using metaphors in health communication have also been reported (Loftus, 2011; Penn & Watermeyer; 2018; Reisfield & Wilson, 2004). An example in which potential patient confusion may have occurred during a GC consultation due to the use of an overly complex metaphor is shown in Example 9.29. The consultation used in this example is the same paediatric consultation shown in Example 9.1. In the example, a metaphor representing weather patterns is used to explain uncertainty.

Example 9.29

Case overview: ***Case 1- TCS***

The case involves a Mixed-Ancestry couple who attended a follow-up consultation at a paediatric genetics clinic together with their three-year-old daughter who had a confirmed diagnosis of TCS. The couple had returned to the clinic as they were expecting another child and were concerned about possible recurrence risks. The wife was 11 weeks pregnant at the time of the consultation.

The example below is taken from a section in the conversation where the specialist is attempting to use a metaphor to explain the uncertainty of whether both parents are unaffected (meaning neither of them have TCS) or whether one of them may have TCS, but that the features may be so mild that it is unnoticeable (due to variable penetrance). Depending on the scenario, the risk implications for a pregnancy are significantly different (either <1% risk if both parents are unaffected, or a 50% risk is one parents is affected but shows very mild features).

Although it seemed that both parents are unaffected (both parents were clinically examined by a genetic specialist prior to the consultation), the genetic specialist in the consultation provides both risk scenarios to the parents.

S = Specialist; F = Father; M = Mother

1	S:	So when Dr Smith* looked at you (0.2) she thinks that you don’t have
2		the condition (0.2) so she thinks she’s in the 60 percent (.) that
3		happened for the first time in her
4		(1.0)
5		in which case the chance of having another child like this is very
6		small↓
7	F:	((nods head))
8	M:	((nods head))
9	S:	Oka:ay (.) .hhhh but there a:are↑
10		(1.0)
11	S:	40 percent where it’s inherited from someone in the family (0.2) You
12		don’t have a family history:y:y↑ and you- she can’t see any ca- any
13		likelihood <but↑ we can never say never>.
14		(1.0)
15	S:	you know.

16		(1.0)
17	**S:**	**It's a bit like (0.3) it's a bit like looking out of the window and**
18		**saying: "Is it raining today or not?"**
19		**(1.0)**
20	**S:**	**And we can look outside today and there's blue skies and we can be**
21		**a:almo:ost↑↑ 100 percent certain that it's not raining and you know**
22		**(0.2) we've all been outside and it looks like it's not raining (0.3)**
23		**and it's (.) it's raining (0.2) Oka:ay↑?**
24		**(1.0)**
25	**S:**	**Uhm and then you can go (.) and you can look outside and say: "Woah↑**
26		**there's very grey skies! (0.4) It probably it raining!" (0.2)**
27		**and then you go outside and it's not.**
28		**(0.5)**
29	**S:**	**It's the same thing (0.3) We can't↑ be 100 percent certain that neither**
30		**of you don't have the condition (0.2) because it is so vari:iable↑**
31		**(1.0)**
32	**S:**	**and because it can be mild enough that you actually can't pick it up**
33		**without doing a genetic test [(0.3) [°which unfort]unately isn't**
34	**F:**	**[((small head nod))]**
35	**S:**	**available at this point°**
36		**(1.0)**
37	**F:**	**((nods head))**
38	**S:**	**.Hhh oka:ay $(0.3) So what am I saying now?$ ((starts to smile))**
39		**(0.5)**
40	**S:**	**I've given you this $who:o:ole↑ story about!$ Hhhh ((smiling))**
41	**M:**	**[((produces an awkward smile))]**
42	**F:**	**[((biting lip and looks at specialist, no reciprocal smiling produced))]**
43	**S:**	**.Hhhh (0.2) U:uhm we don't↑ know**
44		**(1.0)**
45	S:	We (.) we're pretty↑↑ su:ure you're not affected (0.2) we're
46		pretty sure it's blue skies and it's not raining outside ((point towards window in consultation room))
47	F:	((nods head))
48		(0.5)
49	S:	Oka:ay? (0.2) But there's always that small chance that you might
50		have it (0.3) [you might]
51	M:	[yes:s]
52	F:	((nods head))

In the example above, the specialist explains that the patient's condition could either be an event where a genetic mutation occurred for the first time (called a *de novo* event), or that a faulty gene may have been passed on by one of her parents and that one of the parents may also be affected (however the parent may have very mild features that may go unnoticed) (lines 1-16). The parents provide little or no response during the specialist explanations. In lines 17-37, the specialist attempts to explain the risk scenario using a metaphor. An example of the weather is used and how an individual may look out of a window to determine the weather. An individual may think that it may, or may not rain, based on their observations of either blue skies or grey clouds; but may realise that their prediction was incorrect when they actually go outside. The metaphor used in this instance is lengthy and somewhat confusing when explaining genetic risk and uncertainty. In lines 38-44 there is evidence that the patients in this

consultation may not find this metaphor helpful through their pauses, awkward non-verbal gestures and minimal/no verbal responses. The specialist also provides evidence that interactional uncertainty has occurred because of the metaphor and they may now doubt their use of the metaphor by saying: '*So what am I saying now?*' which is accompanied by a smile.

9.3.4 Visual aids

Visual aids were commonly used when presenting risk information to patients. Visual displays were seen to typically accompany verbal explanations of risk and included using pen and paper to draw concepts, as well as using diagrams of numerical, anatomical or molecular-related risk information. Observations from the consultation data show that 14/18 GC consultations use visual aids whilst explaining risk information. The break down of the 14/18 consultations includes 3/5 paediatric consultations, 4/4 adult consultations and 7/9 prenatal consultations.

In the prenatal consultations, particularly for the AMA-referred patients, genetic specialists in these consultations use a comparative maternal age-related risk table to assist in contextualising and explaining a particular pregnant AMA patient's risk. The AMA-risk table used in the consultations shows how a woman's risk to have a baby with Down syndrome (trisomy 21) or any chromosome abnormality increases exponentially with each increasing year (particularly after 35 years of age). An example of the AMA risk table used in the consultations can be seen in Table 9.4 below.

Table 9.4: The risk & birth prevalence of Down syndrome & other chromosome abnormalities according to maternal age.

The Risk & birth prevalence of Down syndrome & other chromosome abnormalities according to maternal age		
Maternal age	**Risk of Down syndrome**	**Risk for all chromosome abnormalities**
20	1/1528	1/526
25	1/1351	1/476
30	1/909	1/385
35	1/384	1/192
40	1/112	1/66
45	1/28	1/21
46	1/21	1/16
47	1/15	1/13
48	1/11	1/10
49	1/8	1/8

This table represents a typical diagram that is used in South African GC consultations for AMA patients. The data in this table is sourced from Palomaki and Haddow (1987) as well as Morris et al. (2002).

Example 9.30 below provides a typical example of a recorded AMA consultation in which a comparative AMA table is used to contextualise a patient's risk by comparing the patient's age-related risk to age-related risks of other women at similar, or different ages.

Example 9.30

Case Overview: ***Case 5- AMA***

The case involved a 42-year-old isiXhosa woman who was 15 weeks pregnant. She was referred for advanced maternal age counselling and no soft markers were detected during her ultrasound scan.

S= Specialist; P= Patient

1	**S:**	So (.) basically these are pictures of people with Down syndrome ((showing patient diagram in her counselling file)
2		**(0.2) basically I'm going to show you this table (.) ((pointing to an age-related risk table))**
3	**P:**	**Mm ((attention focused on table))**
4	**S:**	**they (.) they looked at women of all different ages (.) from twenty to**
5		**forty six (.) And they looked at how many women of those ages have**
6		**babies with Down syndrome. Okay?**
7	**P:**	**((head nod and looking at diagram))**
8	**S:**	**And you can see from the gra:aph↑ that it goes up and up and up**
9		**as you get older.**
10	**P:**	**Mm ((eye gaze and attention still focused on the diagram))**
11	**S:**	**So if you had (0.2) your first (.) your older daughter at thirteen (.)**
12		**you're about (0.2)**
13	**P:**	**twenty eight**
14	**S:**	**Twenty eight? Make it thirty okay.**
15	P:	Mm
16	**S:**	**The chance there of having a baby with Down syndrome is about one**
17		**in nine hundred. Okay?**
18	**P:**	**((head nod))**
19	S:	Close to one in a thousand. Around there
20	P:	Mm
21	S:	So that means (.) every nine hundred women who are thirty years old and
22		are pregnant (0.3) one of them will have a baby with Down syndrome (.)
23		the other eight hundred and ninety nine children won't have Down
24		syndrome
25	P:	((small head nod))
26	**S:**	But we can't say that they won't have anything else (.) but we
27		know that they won't have Down syndrome because just from all
28		those numbers (0.3) **And you are now forty two?**
29	**P:**	**((head nod)) yes:s**
30	**S:**	**Okay (.) So the cha:ance now has gone up to one in sixty five (0.4) So ((pointing at the table))**
31		**you see it starts going up quite steeply as you get over 40.**
32	**P:**	**Hmm ((head nod))**

In the example above, the specialist compares the particular patient's age in lines 14, 16-17, 28 and 30-31 by juxtaposing her age-related risk when she was 30 (risk is given as 1 in 900) to

her current age of 42 (risk is given as 1 in 65). In producing these figures, the specialist also uses additional formats to convey the risk such as narrative (lines 21-24).

In the interview data, visual aids were mentioned as being useful in six specialist interviews. Examples 9.31 and 9.32 provide quotes from the interviews in which visual aids are reported to be beneficial.

Example 9.31
S5: "Something visual I think is helpful for people."

Example 9.32
S10: "…Uhm other things, for example, recessive conditions I always end up drawing pictures, you know drawing the 4 options and explaining 1 out of 4 would be this, and that would be a 25%."

Previous literature has described the usefulness of visual aids, diagrams and props in improving information recall as it enhances the understanding of conveyed verbal and numerical information (Dowse, 2020; Garcia-Retamero & Hoffrage, 2013; Houts et al., 2006; Tait et al., 2010; Watermeyer & Penn, 2009a).

Instances in which the use of visual aids became interactionally problematic were noted in one prenatal and one paediatric consultation. In the prenatal consultation, a patient verbalises confusion when a genetic specialist draws visual representations of numerical information. Similarly, in the paediatric consultation, the specialist becomes engrossed in drawing several complex inheritance patterns and risk outcomes to a couple, who demonstrate disinterest and lack of engagement with the visual risk information. They have also previously indicated that they are not interested in the information which the specialist decides to convey and draw anyway. An example of this consultation is discussed in more detail in Example 11.4 in Chapter Eleven.

The potential for diagrams to cause negative reactions, or patient confusion has been demonstrated in South African genetic counselling (Wessels, 2014); pharmacy (Dowse & Ehlers, 2001), and audiology (Penn & Watermeyer, 2018) research. In Wessels' (2014) study on AMA GC consultations in Johannesburg, a picture of an amniocentesis procedure is shown by a genetic counsellor to a high-risk AMA patient who is unfamiliar with the procedure. The picture is shown by the specialist in an attempt to facilitate patient decision-making regarding whether they would want to proceed with an amniocentesis or not. Wessels' (2014) results

show how the patient becomes visibly distressed by the diagram due to the size of the amniocentesis needle and it's proximity to the unborn baby during the procedure. Despite the specialist attempting to reassure the patient that the needle will not 'touch' the baby, the patient declines the offer. Previous literature, together with this study thus shows that although commonly used in GC consultations and reported to be useful by genetic specialists, sensitivity to context and patient interests are necessary when using visual risk information.

9.3.5 Framing

In the consultation data, the framing of risk is noted in the prenatal (6/9) consultations, but not in the paediatric or adult consultations. Framing involves the description of a feature or an item in which the positive or the negative outcome or risk is emphasised (Ahmed et al., 2012). In the six prenatal consultations in which framing is noted, five of the consults are AMA referrals (some of which had additional soft markers noted on ultrasound), whilst one referral is for soft markers noted on an ultrasound. All involve risk discussions on having a baby with Down syndrome. Framing is also mentioned in one of the genetic specialist interviews with an emphasis on how it has the potential to impact patient risk perception and emotional responses to risk information.

Four (4/9) of the prenatal consultations show how risk is first framed in a negative manner (the chance of having a baby with Down syndrome), and then thereafter a positive manner (the chance of not having a baby with Down syndrome). Much of the conversation that follows this way of presenting risk typically focuses on the negative risk frame (the chance of having a baby with Down syndrome). All four consultations discuss risk in the formats of a ratio (e.g., 1/30).

In the other two consultations where framing is used, the emphasis is placed on the positive frame of risk (the chance of not having a baby with Down syndrome), rather than on the negative. These two cases are considered unique in comparison to some of the other patterns noted in the consultation data where negative framing is typically emphasised. In one of the consultations, a ratio is used to present the risk information with the specialist first presenting the positive frame (the chance to not have a baby with Down syndrome), and thereafter the negative frame (the chance of having a baby with Down syndrome). In the other consult, the specialist converts risk information initially presented in a ratio, into a percentage and frames

the percentage figure given positively, emphasising the chance to have a baby that would not be affected by Down syndrome.

Evidence from one of the unique prenatal consultations is shown below (Example 9.33: Part 1 and Example 9.33: Part 2) where the genetic specialist focuses on positive framing. The consultation further highlights the impact of personal risk perception and how it may influence how risk information is presented and understood.

Example 9.33

Case overview: ***Case 9 -AMA & a soft marker for Down syndrome***

This consultation involved a 42-year-old Mixed Ancestry woman who presented at one of the prenatal clinics at 21 weeks' gestation. She had been referred for GC due to her advanced maternal age, and because on ultrasound, her baby was seen to have a flat nasal profile, an indicator for possible Down syndrome. In the examples below, the specialist conveys the patient's risk of having a baby with Down syndrome. The specialist uses two sources of information: The adjusted-risk based on the patient's prenatal ultrasound findings and the patient's age-related risk. The session was conducted in Afrikaans, the patient's first language. In the transcript below the first line of each numbered line shows the original Afrikaans language used in the consultation (shown in italics) and the second line shows the translated English.

In Example 9.33: Part 1, when asked why she had been referred for GC, the patient indicates that she had been referred because of her advanced maternal age and that the doctors had been concerned about the flat nasal profile of the baby, detected as a soft marker during the patient's ultrasound check-up. The patient reports that she is not concerned about the baby's nasal profile as she believes that the baby has inherited her partner's nasal profile, whom she says also has a 'flat' nose. The patient is thus reportedly not concerned about the soft marker as an indication of Down syndrome risk. This section of the consultation provides evidence of the impact of context, particularly personal risk perception. The section also provides evidence of the genetic specialist's manner of framing risk information and how this can have an impact on the patient's emotional responses.

Example 9.33: Part 1

S= Specialist; P = Patient

19	***S:***	*Okay (0.2) so vertel nou eers vir my (0.2) uhm (0.3) wat het jy*
20		*verstaan oor hoekom jy vandag hierna toe moes kom?*
		Okay (0.2) so first tell me (0.2) uhm (0.3) what did you
		understand about why you had to come here today?
21		(1.5)
22	***P:***	*Omdat ek twee en veertig is*

		Because I am forty two
23	S:	Okay↑
24	***P:***	*Dat ek 'n high risk is*
		That I'm a high risk
25	**S:**	*Okay↑ (0.4) so uh (.) jy's reg (.) ons sien almal hierso wat 37*
26		*jaar en ouer↑ is (.) omdat (.) 'n vrou soos wat sy ouer word is*
27		*daar 'n groter kans vir (.) abnormaliteite↑ (0.3) soos Down sindroom*
28		*(0.3) maar ook vir Ma se (.) Ma se gesondheidsprobleme (0.3) so jy is*
29		*(maar net n beitjie me:e:er hoe risiko (0.2) high risk (0.3) maar↑*
30		*daar is ALTYD↑ 'n groter kans vir 'n norma:ale baba (.) al selfs is*
31		*mens 42.*
		Okay↑ (0.4) so uh (.) you're right (.)we see everyone here who
		is 37 and older↑ (.) because (.) a woman as she gets older there
		is a greater chance for (.) abnormalities↑ (0.3) like Down syndrome
		(0.3) but also for Mother's (.) Mother's health problems (0.3) so you
		are just a little bit mo:o:ore high risk (0.2) high risk (0.3) but↑
		there is ALWAYS↑ a greater chance for a normal baby (.) even if a
		person is 42.
32	P:	((head nod))
33	**S:**	Okay↑? (0.4) dis maar net dat jy hier n baie goeie↑ scan↑ kry (.) en
34		dat (.) en dat jy OPSIES↑ het as jy n bietjie ouer is (0.2) dit se nie
35		dat daar (0.3) definitief woud is nie (0.2) ne?
		Okay↑? (0.4) it's just that here you get a very good↑ scan↑ (.) and
		and that (.) and that you have OPTIONS↑ when you are a little bit
		older (0.2) is doesn't say that there is (0.3) definite fault (0.2) hey?
36	**P:**	**((head nod))**
37	S:	*Okay↑ (.) so dit is hoekom jy hier↑ is (0.3) en wat? (.) jy't al*
38		*klaar jou sonar gehad ne?*
		Okay↑ (.) so that is why you are here↑ (0.3) and what? (.) you've
		already had your sonar hey?
39	*P:*	*((head nod))*
40	***S:***	*en wat het hulle vir jou gese↑?*
		and what did they tell↑ you?
41		(1.5)
42	**P:**	*Hulle het vir my gese dat*
		They told me that
43		**(0.5)**
44	**P:**	*babatjie se (0.3) neus bientjie 'n bietjie*
		baby's (0.3) nose bone is a little bit
45		***(3.5)*** ***((the patient places her fingers on her nose))***
46	**P:**	*plat is*
		flat
47	S:	*Okay*
48	**P:**	*Maar toe se ek: "Maar dis die pa s'n"*
		But then I said: "But it's the father's"
49	**S:**	*Dis die pa↑ s'n?*
		It's the father↑'s?
50	**P:**	**Hhh** **((smiling))**
51	**S:**	*Okay hhh (0.2) dis 'n belangrike ding (0.3) want dit is een* *((head nod))*
52		*van die goeitjies waarna ons kyk↑ (0.3) want ons weet babatjies*
53		*met Down sindroom het baie keer (.) n plat neus beentjie (0.2)*
54		*maar↑ ons weet (.) ons lyk almal verskillend↑*
		Okay hhh (0.2) that's an important thing (0.3) because it is **((head nod))**
		one of the things that we look at↑ (0.3) because we know babies
		with Down syndrome often have (.) a flat nose bone (0.2) but↑ we
		know (.) we all look different↑
55	**P:**	**((nods head and smiles))**

56	**S:**	*Party van ons het GROOT↑↑ neus bene (.) en party van ons het*
57		*PLAT↑ neus bene*
		Some of us have BIG↑↑ nose bones (.) and some of us have FLAT↑
		nose bones
58	**P:**	**((nods head and smiles))**
59	**S:**	*En ons (0.2) die (.) rekenaar ook wat ons gebruik om die risiko*
60		*uit te werk↑ (.) en daai sonar↑ gaan nie in AG neem (.) hoe die*
61		*ma lyk (.) of hoe die pa lyk nie↑*
		And we (0.2) the (.) computer also that we use to work↑ out the risk
		(.) and that sonar↑ is not↑ going to take into CONSIDERATION (.) how
		the mom (.) or the dad looks↑
62	**P:**	**((nods head and smiles))**
63	**S:**	*Ons sien↑ nou nie eers die pa nie (0.2) maar dit is 'n belangrike ((shaking head))*
64		*ding om in ag te neem (.) want jou neus↑ moet van iewers af kom. ((smiling))*
		We don't even see↑ the dad (0.2) but it is an important thing to take ((shaking head))
		into consideration (.) because your nose↑ must come from somewhere. ((smiling))
65	**P:**	**((smiling))**
66	S:	*Van jou ma↑ of jou pa↑ af kom*
		Come from your mom↑ or your dad↑
67	P:	**((nods head and smiles))**
68	**S:**	*So uhm (0.3) dit is iets wat <u>absoluut↑</u> 100 persent norma:al kan wees↑*
		So uhm (0.3) so it's something that can be↑ <u>absolutely</u>↑ 100 percent
69		normal
70	P:	((head nod))
71	**S:**	*Maar (.) ongelukkig↑ (.) omdat jy twee en veertig is (.) en daar's nou*
72		*n' (.) 'n merker↑ gesien wat ons weet kan ook Down sindroom beteken maak*
73		*dit nou (0.2) jou risiko hoer*
		But (.) unfortunately (.) because you are forty two (.) and there is now
		a (.) a marker↑ seen that we know can also mean Down syndrome it now
		makes (0.2) your risk higher
74	**P:**	**((head nod))**
75	**S:**	*Ons gaan nou rustig daaroor gesels*
		We are now going to calmly talk about it
76	**P:**	**((head nod))**
77	**S:**	**Okay?**
78	**P:**	**((head nod))**

In Example 9.33: Part 1 above, the patient seems to respond positively to the way the specialist has framed her reason for being referred for GC. Although the specialist acknowledges that the patient's age is a concern and places her at risk for conditions such as Down syndrome (lines 25-27), the specialist frames this risk information in a positive manner by stating that despite the patient being 42, the patient has a much higher chance of having a healthy baby, even at her age (line 29-31). The specialist further decreases the level of risk by saying that the patient is *'just a little bit more high risk'* (line 29). By decreasing the patient's level of risk, the specialist creates more certainty for the patient that maybe this referral is not something to be as concerned about. The specialist reassures the patient that although she has been referred to GC, it does not mean that there is necessarily a problem with her baby (lines 33-35). The patient

responds to this information with minimal head nods. Similar use of mitigative language by healthcare practitioners to place patients at ease during risk-based conversations has been observed in South African audiology interactions (Watermeyer et al., 2020a).

In lines 40-78, the discussion turns specifically to the issue of the baby having a flat nasal profile where the patient continues to attempt to justify the baby's 'flat nose'. The specialist justifies and supports the patient's reasoning by indicating that it may be a valid argument (lines 51-54, 56-57, 63-65). The specialist further says that the technology used to screen the patient's baby does not take into consideration contextual aspects such as the parent's features in lines 59-61 and that a flat nasal profile can be *'absolutely normal'* (lines 68-69). The extreme formulation in line 68 greatly reduced the uncertainty of this potentially 'risky' finding in the baby, framing the situation and outcome in a positive manner. The specialist's supportive responses seem well received by the patient, indicated through head nods and smiling (lines 55, 58, 62 and 65).

In Example 9.33: Part 2 below, the use of framing and how a specialist chooses to present the order of information is shown.

Example 9.33: Part 2

S= Specialist; P = Patient

257	**S:**	*So (.) omdat ons die neus beentjie sien wat 'n bietjie kleiner*
258		*is () het die kans gegaan van een uit dertig na een uit nege↑*
		So (.) because we see the nose bone that is a little smaller (.)
		the chance has gone from one out of thirty to one out of nine↑
259	P:	((head nod))
260	S:	Okay?
261	P:	((head nod))
262	**S:**	*So dit beteken↑ (.) dat daar is agt uit nege kans (.) dat die babatjie*
263		*nie↑ Down sindroom het nie↑ (0.2) maar daar is 'n een uit nege kans*
264		*dat dit wel (.) Down sindroom [is i:s] (.) okay? (0.3)*
		So that means↑ (.) that there is an eight out of nine chance (.) that
		the baby won't↑ have Down syndrome ().2) but there is well a one out
		of nine chance that it (.) is i:s Down [syndrome] (.) okay? (0.3)
265	**P:**	*[sindroom is]*
		[syndrome]
266	S:	*en niemand het p. (.) [pa se neus been] nie*
		and no-one has d. (.) [dad's nose bone].
267	**P:**	[(nods head and smiles))]
268		(0.5)

In Example 9.33: Part 2 above, the specialist presents the patient's age-related risk (presented as 1 in 30) against the patient's now adjusted risk based on their ultrasound findings (presented

as 1 in 9) in line 258. Thereafter, instead of focusing on the negative frame just presented (1 in 9) and interpreting it further, the specialist re-frames the 1 in 9 ratio provided into a positive frame of *'8 out of 9 chance that the baby won't have Down syndrome'* in lines 262-264. In line 266 the specialist refers to the baby's flat nasal profile, indicating that it may be a sign of Down syndrome and not simply a representation of the *'dad's nose bone'*. The patient provides a head nod and a smile in response in line 267.

Subjective risk perception in decision-making and communication plays a large part in risk communication. In the consultation example above, the patient's risk perception plays an important role in how she responds to the risk given to her. The finding in the consultation is supported by an interview quote from another genetic specialist (Example 9.34) that suggests that despite the potential for framing to sway patient decision-making, a patient's risk perception may have a larger impact on their decision-making.

Example 9.34
S9: "…And then another one, I try to kind of say: "Well that means that 1 out of that [50], you know, those people, would be likely to have the condition, we don't know in advance whether you would or wouldn't be that person," You know of course that also means, because I think one must also obviously you know, flip it around and say: "Well 49 people of the 50 wouldn't" and you know, I think you know, some people see the 1 and some people see the 49."

The patient's risk perception in the consultation example above seemed to have impacted how the genetic specialist framed the risk information.

Patient perceptions are not the only factor in risk communication. Genetic specialist risk perceptions also appear important. Kirklin (2007) discusses how healthcare practitioners have a choice over how they choose to frame information given to a patient. The act of information giving may be influenced by a healthcare practitioner's personal values and experiences, as well as contextual factors such as their training and the attitudes of the healthcare system in which they practice. Presenting risk information both in the positive and negative frame is thought to be an honest approach which allows a patient to have a better perspective over a particular risk (Paling, 2003). Presenting information in only one frame is considered a persuasive rather than a neutral act. Individuals are more likely to choose a risk-avoiding option when the information is negatively presented to them, whilst riskier behaviour is adopted when positive outcomes are emphasised (Edwards et al., 2001; Tversky & Kahneman, 1974).

Although the specialist presents both the positive and negative frames in Example 9.33: Part 2, indicating a neutral and objective risk communication approach, they are more direct in choosing only one frame (namely a positive one) in the prior section of the conversation shown in Example 9.33: Part 1. I argue that the specialist's use of only one frame is based on the patient's justification of their ultrasound findings and their risk perception that the baby's flat nose is not an indication of Down syndrome, but rather a normal physical feature attributed to the baby's father. In the other video-recorded consultations in the study's data where framing is noted, the specialists tend to convey risk information by presenting both frames, however, the negative frame is typically presented first and then focused on for the duration of the consult. The order of the positive and negative frame could indicate a subtle persuasive technique from the genetic specialists for patients to act on the risk presented in a particular manner.

Previous research has indicated that despite obligations to remain non-directive, genetic counsellors in prenatal settings may communicate in ways that highlight the negative aspects of continuing a pregnancy where an unborn child is known to have a disability (Asch, 2018). Specialists may thus promote the uptake of invasive testing in subtle ways when there is a related risk indication (Wessels et al., 2015; Wessels & Koole, 2019). In Example 9.33, the opposite seemed to have occurred however. Although the specialist uses one frame, it seems that they are trying to dissuade the patient from opting to do invasive testing and highlighting the chance of having a healthy baby, rather than focusing on the chance of having a baby with a disability. At the end of the consultation, the patient decides not to undergo any further investigation or genetic testing (not shown in the transcript in the example above) as they believe that all would be well. The baby's nose can be explained by the father's similar facial features. The GC consultation outcome may have had to do with the format in which risk information is presented and framed during the consultation by the specialist, together with the patient's risk perception.

Based on the findings presented in Example 9.33 and findings from the other data in this study, I argue that the choice of approach when communicating risk is influenced by contextual factors including patient and genetic specialist risk perceptions. Genetic specialists may find it difficult, if not impossible to remain non-directive or neutral in GC practice since whichever way a specialist chooses to present risk information will have an impact on how a patient perceives that information.

9.4 Conclusion

In conclusion, this chapter has provided evidence on the content (the *what*) and the formats (the *how*) of risk communication in a South African GC setting. Genetic specialists refer to several sources of risk and use different approaches to convey risk information to patients. The most common formats to convey risk in local GC consultations appear to be numerical formats, metaphors and visual aids. Numerical formats being the most commonly used risk format is in keeping with local (Perrott, 2010) and international (Fagerlin et al., 2011; Stacey et al., 2017; Trevena et al., 2013) studies focusing on risk communication. Metaphors (Kendall-Taylor et al., 2013) and visual aids (Dowse & Ehlers, 2001; Watermeyer, 2008; Watermeyer & Penn, 2009a) have both been described as beneficial in explaining complex information. Also, that metaphors and visual aids are typically combined with numerical formats in this study further affirms previous research that numerical representations of risk are best understood when presented together with more tangible ways of presenting risk such as narratives, metaphors, as well as visual aids (O'Doherty & Suthers, 2007; Smerecnik et al., 2009; Trevena et al., 2013). Caution must be exercised though to the use of formats and the type of risk information presented since context shapes how the risk information and formats are responded to and received by patients. Awareness of context is thus important for genetic specialists when discerning what information and format to use in a GC consultation.

The study results demonstrate that providing risk information is challenging, and that patient understanding and the value of various risk formats may be difficult to determine. Several of the consultations show reserved patient behaviour with minimal responses or active engagement in conversation. Reserved or passive patient behaviours may create additional uncertainties through the potential of information being misunderstood and patients not requesting clarification. Even in instances where checking of understanding is conducted by genetic specialists, patients may still provide minimal responses.

CHAPTER 10: LAYERS OF UNCERTAINTY: UNIVERSAL UNCERTAINTIES, PERSONAL UNCERTAINTIES & INTERACTIONAL UNCERTAINTIES

10.1 Introduction

The chapter provides evidence of the different layers of uncertainty identified in the study. Layers of uncertainty identified include universal uncertainties, personal uncertainties and interactional uncertainties.

Universal uncertainty can be described as limitations in scientific knowledge (i.e. nobody knows this information, or at least not yet), whilst personal uncertainties are caused by one's knowledge limitations or lack of understanding (Fox, 1957; Gigerenzer, 2003). In this study, universal and personal uncertainties were mostly linked to dealing with an unknown diagnosis or outcome and attempting to interpret uncertain test results. Genetic specialists report that universal uncertainties are common in practice and create pressures in providing patients with answers. Genetic specialists further report on personal uncertainties and how they attempt to manage these.

In this chapter, I highlight how additional layers of uncertainty caused by contextual features can cause tensions in patient-genetic specialist communication during GC consultations. Contextual features seen to particularly create interactional uncertainty in local GC risk communication include patients who are unable to provide accurate background information whilst genetic specialists attempt to establish risk, patients who have limited literacy, as well as the presence of a language barrier between a patient and a specialist.

10.2 Universal & personal uncertainties

Table 10.1 provides a summary of the areas and types of universal and personal uncertainties identified in the consultation and interview data.

Table 10.1: Areas & types of universal & personal uncertainties identified

Major areas identified	**Examples of universal & personal uncertainties**
The impact of uncertainty in GC consultations	Uncertainty as a core part of GC consultations.
	Uncertainties in prenatal GC consultations
	Uncertainties in paediatric GC consultations
	Uncertainties in adult GC consultations
Genetic specialists' response to feelings of uncertainty	Genetic specialists' personal feelings of uncertainty
	Patients' expectations to know the answer
	Genetic specialists' perceived pressure from other healthcare practitioners to know the 'diagnostic answer' for a patient
Genetic specialists' approaches to managing uncertainty in GC consultations	Being honest with patients
	Accepting the presence of uncertainty
	Asking colleagues for guidance
	Altering the way risk information is provided
	An unhelpful approach: Being verbose

10.2.1 The impact of uncertainty in genetic counselling consultations

Nine (9/12) genetic specialists spoke about how uncertainty is a core part of GC (Example 10.1 and 10.2).

Example 10.1
S1: "I think it's [uncertainty] always there in genetic interactions. I think it's not, it's like just about a whole part of it. Everything."

Example 10.2
S8: "So, I think uncertainty is a very part of… it's very nice if you can counsel with certainty but we often can't. So, it is part, it's part and parcel of what we do."

Previous researchers have described the omnipresence of uncertainty in GC. Much of the uncertainty is owed to the complexity of risk information and the uncertainties relating to inheritance, diagnosis, disease management, treatment and prevention (Bernhardt et al., 2014; Bylund et al., 2012; Henneman, 2008). Furthermore, this study supports previous research which highlights the difficulty of conveying unclear information to patients, who have to use this limited information to make decisions (Sarangi & Clarke, 2002a).

10.2.1.1 <u>Uncertainties in prenatal genetic counselling consultations</u>

When discussing the types of clinics, 6/12 genetic specialists mentioned that universal uncertainty in prenatal consultations is particularly challenging. The presence of soft markers during prenatal ultrasound scans were mentioned as sources of uncertainty, particularly in terms of their outcome. In prenatal GC consultations, specialists have to provide risk assessments and outcome predictions with limited information. Although ultrasound findings

are meant to provide an indication of an unborn baby's health, soft markers identified in an unborn baby, may either mean a severe risk outcome or may mean a low-risk outcome. Facilitating and supporting a pregnant patient's decision-making during these types of circumstances is reportedly difficult for genetic specialists. Example 10.3 and Example 10.4 provide evidence of specialist's quotes regarding ultrasound soft markers.

Example 10.3
S5: "I think for me probably the most challenging still remain these issues of these complex neurological phenotypes in the prenatal setting, you know, you can have a really dreadful outcome and you can have a very mild outcome and trying to figure out how on earth you can try negotiate with people how they make those decisions...when you really can't make predictions".

Example 10.4
S8: "If we think in the prenatal context uhm if you see uhm, a baby uhm agenesis of the corpus collosum for example, uhm there is SO much uncertainty. Because it can be so fine, but it can be so not fine. It can be part of a big syndrome, it can cause developmental delay, it can cause all sorts of psychiatric issues."

The presence of uncertainty during prenatal consultations has been previously reported. Much of the uncertainty is due to the inability to interpret the risk implications of either ultrasound soft markers (Åhman et al., 2010; Richards et al., 2015b) or genetic test results (Hashiloni-Dolev et al., 2019; Lou et al., 2020a; Richardson & Ormond, 2018). These universal and personal uncertainties are difficult to address in prenatal GC consultations.

The GC consultation data also shows several examples where establishing risk or knowing risk outcomes in prenatal consultations is challenging. The inability to conduct certain medical tests for unborn babies further complicates the establishment of risk. In Example 10.5 I return to the teratogen example from Example 9.23. The example shows how a genetic specialist may struggle to determine the risk and outcome of a patient's pregnancy due to limitations in medical testing. In the example, the pregnant mother has been exposed to a teratogen that has placed her unborn baby at risk for having congenital complications. I demonstrate in the example, how the specialist's turn design and choice of wording introduce and foreground uncertainty into the interaction.

Example 10.5

Case overview: ***Case 10 -Teratogen exposure (maternal rubella infection)***

This was a prenatal case involving a 24-year-old Caucasian woman (labelled as the patient in the transcript) who attended a follow-up consultation at one of the prenatal genetic clinics at 19 weeks' gestation. She was accompanied by her mother (labelled as the mother). The pregnant patient was referred for GC as she developed a rubella infection at 16 week's gestation.

During pregnancy, the only possible test that can be offered to confirm infection in a pregnant woman and her unborn baby is a rubella immunoglobulin M (IgM) test or a rubella-specific RNA test that can be performed on the pregnant woman's blood, or alternatively using amniotic fluid or cord blood from the unborn baby (Motaze et al., 2020; Tang et al., 2003). The session was conducted in Afrikaans, the patient's first language. In the transcript below the first line of each numbered line shows the original Afrikaans language used in the consultation (shown in italics) and the second line shows the translated English.

S: Specialist; P = Patient; M= Patient's Mother

78	**S:**	*So as 'n mens uh (.) rubella kry baie↑ vroeg in die swangerskap*
79		*(.) so nou praat jy van (0.2) 8 weke van swangerskap (0.3) dan*
80		*is daar 'n baie baie hoer↑ kans miskien (.) 90 persent kans dat baba*
81		*uh (0.2) effekte sal kry en dat daardie effekte erg↑ is.*
		So if a person uh (.) gets rubella very↑ early in the pregnancy
		there is a very very higher↑ chance maybe (.) 90 percent chance that
		baby uh (0.2) will get effects and that those effects are
		severe↑
82		(0.5)
83	**S:**	*Maar so later dit word in die swangerskap (0.2) hoe minder is*
84		*die kans↑ (0.2) en ook hoe minder↑ is die effekte*
		But the later it gets in the pregnancy (0.2) the smaller the
		chance↑ is (0.2) and the lesser the effects will be
85	P:	((slow head nod))
86	**S:**	*Ok (0.3) uh op (.) 16 weke swangerskap (.) is dit min of meer*
87		*(0.2) 20 persent kans (.) of 1 uit 5 kans dat uh (0.2) dat baba aangetas*
88		*sal word*
		Ok (0.3) uh at (.) 16 weeks pregnancy (.) **it is more or less**
		(0.2) 20 percent chance (.) or 1 out of 5 chance that uh (0.2) that
		baby will be affected
89	M:	((head nod))
…	…	…
111	**S:**	*Ja ok (0.3) n. nou waar dit nou raak 'n bietjie meer*
112		*gekompliseerd (0.2) is wat ons kan doen is 'n toets om te sien*
113		*of babatjie (.) babatjie die infeksie gekry het↑ (0.2) ok (.)*
114		*met ander woorde (.) ons sien of die rubella versprei het van*
		Yes ok (0.3) n. now where **it now becomes a little bit more**
		complicated (0.2) is what we can do is a test to see if baby (.)
		baby got↑ the infection (0.2) ok (.) in other words (.) we see
		if the rubella spread from Mom's bloodstream over the placenta
115	P:	((head nod))
116	**S:**	*En uhm (0.3) ja:a maar (.) maar dit (0.2) uhm kannie vir ons 'n*
117		*100 persent verseker se (.) dat baba die hoor probleem sal kry (.) ok* ((pointing at own ears))
		And uhm (0.3) yes:s but (.) but **it (0.2) uhm can't tell us 100 percent**
		for certain (.) that baby will get the hearing problem (.) ok ((pointing at own ears))
118	P:	((head nod, frowning and pursing lips in worried manner))

119	S:	*So dit is moontlik dat die baba 'n infeksie kan kry sonder dat*
120		*die baba eintlik geaffekteer is deur die infeksie (.) so dis*
121		*'n bietjie meer compleks*
		So it's possible that the baby can get the infection without
		the baby actually being affected by the infection (.) so it's
		uh a bit more complicated
122	P:	((no verbal or non-verbal expression)
123	M:	((nods head))
124	**S:**	*Maar (0.2) miskien uhm (0.2) ja (.) en dis nie eintlik moontlik*
125		*vir ons om 100 persent vir seker te se nie (.) dat die (0.2) baba*
126		*eintlik aangetas sal word (0.2) so dis die naaste wat ons kan*
127		*kom is om te se (.) ok die babatjie het uhm (.) die infeksie*
128		*gekry en daarom vergroot dit die kans (.) so dis nou meer as 20 persent*
129		*(.) miskien is dit 50 of 50 persent (.) kans* *((head nod))*
		But (0.2) maybe uhm (0.2) yes (.) and **it's not actually possible**
		for us to say with 100 percent certainty (.) **that the (0.2) baby**
		actually will be affected (0.2) so it's the closest that we can
		get to say (.) ok the baby did (.) get the infection and thus
		it increases the chance (.) so it's now more than 20 percent (.) **maybe**
		it's 50 or 50 percent (.) chance ((head nod))
130	M:	((smile))
131	P:	((small head nod))

In the example above the specialist displays uncertainty in establishing an accurate risk for the patient's unborn baby through their formulation of words and how they design their turn. Evidence that the specialist is more confident about the risks for a rubella infection during the first trimester is shown by their choice of words in line 79: *'There is a very very higher chance maybe 90 percent chance'.* The repetition of 'very' further provides evidence of the specialist's certainty and their attempt to convince the patient of the high risk. Repetition of a single word or phrase in a speaker's single turn constructional unit be a way of convincing the listener about the information proposed (Sacks et al., 1974; Stivers, 2004). Although the specialist claims that the impact of the rubella infection is less severe during the second trimester in lines 83-84, evidence that the specialist is not as sure about the second-trimester risk assessment is provided in line 86-87 when they state, *'it is more or less 20 percent chance'.*

In lines 90-110 (not shown) the specialist discusses that the risks stated (90 percent and 20 percent) are specifically for hearing loss and that there may be risks for the baby to have other complications. The specialist goes on to explain that the baby may have hearing loss that is present from birth, or that they may initially be able to hear, but may experience progressive hearing loss as they get older. The specialist discusses that a baby's hearing problems can be confirmed by doing a hearing test on the baby after birth and again when they are older. During the section of the conversation, the patient and her mother provide no verbal or non-verbal responses.

In lines 111-131 the specialist explains to the patient and her mother that although an unborn baby may also be infected by rubella, they may not necessarily develop adverse effects from the infection. Hence, the outcome of a prenatal rubella infection may be severe (i.e. cause permanent hearing loss) or may be negligible (i.e. the baby's health and development is not impacted). Indirect evidence of the specialist's uncertainty on providing an accurate risk assessment is displayed by the specialist's use of hedging: '*It now becomes a little bit more complicated'* (lines 111-112), *'maybe it's 50 or 50% chance'* (lines 128-129). By hedging information, an individual can exercise caution and avoid full commitment to a claim made as there may be variation in the outcome of their claim (Staples et al., 2020; Stivers & Timmermans, 2016). Further evidence for uncertainty is shown by the specialist's use of collective, rather than personal, pronouns: 'it uhm can't tell *us*' (line 116), *'it's not actually possible for us to say'* (lines 124-125); *'it's the closest that we can get to say'* (lines 126-127). The use of a pronoun is a mitigative device that allows the speaker to categorise themselves as part of a particular group, or it enables them to point to other people (Caffi, 1999) to whom the problem may be relevant. The mitigative device also allows the speaker to create metaphorical distance between what they say and how the listener may perceive the speaker's personal image (Haverkate, 1992; Weiste et al., 2018). Sarangi and Clarke (2002a) show how genetic specialists use mitigative strategies such as these to manage uncertainty in GC by demarcating the boundaries of expertise. In the example above, by using collective pronouns the specialist frames their personal position, i.e., the uncertainty in the situation is a universal uncertainty, rather than a personal uncertainty on the part of the specialist.

10.2.1.2 Uncertainties in paediatric genetic counselling consultations

Paediatric GC consultations also have several universal and personal uncertainties. Uncertainties stem from dealing with an unknown diagnosis, or uncertainties concerning the future wellbeing or life expectancy of a child diagnosed with a genetic condition.

Diagnostic uncertainty exists as many children referred to GC for an assessment may remain undiagnosed (Example 10.6 and Example 10.7). Despite efforts from genetic specialists, some patients remain without a definitive answer to the cause of their condition due to universal uncertainties.

Example 10.6
S4: "…The problem in our field is that more than 60, more than 60- or 70% cases we usually don't have a diagnosis."

Example 10.7
S6: "…Uhm a lot of the genetic conditions that we do see at the paediatric clinic though, there's a lot of weird and wonderfuls we don't have testing for, so we're unable to give a definite diagnosis and therefore give appropriate genetic counselling."

Research on families with children with unknown diagnosis indicate that these families experience heightened feelings of anxiety and vulnerability (Lewis et al., 2010; Zurynski et al., 2017) and genetic specialists may struggle to communicate effectively with families when a diagnosis cannot be reached (Steel et al., 2014).

Paediatric GC consultations where a specialist and parent(s) are unable to predict the well-being or future life expectancy of an affected child's life further creates personal and universal uncertainty. In 2/5 recorded GC consultations, the parents of an affected child became concerned with their child's future outcomes. In one consult, a parent became concerned whether their young daughter, who has a diagnosis of Treacher Collins syndrome will be able to attend a 'normal' school one day. In another consultation, the parents of a child with hypophosphatasia, ask a genetic specialist about their son's life expectancy.

In these two consultations, it became evident that genetic specialists can alter the level of uncertainty for a parent through their own interpretations of universal knowledge, their verbal expressions, and how they design their communication approach in response to a parent's question. Previous literature has shown that genetic specialists are not merely detached vessels that convey information to patients. Rather, they remodel unclear knowledge in subtle ways when addressing patients to redefine the boundaries and of uncertainty as shown by a study by Stivers and Timmermans (2016). In this study, 23 video-recordings of medical geneticist-parent interactions revealed that when delivering genomic test results to parents of a child in which a variant of unknown significance (VUS) was found, medical geneticists altered the boundaries of uncertainty, by providing their interpretation on whether the VUS was indeed causal or not causal. The provision of personal interpretations not only altered the boundary of uncertainty in the consultation but created a care-relationship between medical geneticists and parents that assisted in addressing parental anxieties around their child's diagnosis and the uncertainty thereof. Even when a clear diagnosis was not available for a child, the empathetic way uncertain information was conveyed by the specialists was welcomed by the parents.

The use of uncertainty as a tool in healthcare communication, depending on whether its outcomes are a threat or an opportunity, has been described in Mishel's theory of uncertainty in illness (Mishel, 1990). In situations where uncertainty represents a dreaded outcome, healthcare practitioners highlight strategies to reduce it. Alternatively, if uncertainty infers a possible optimistic outcome, healthcare specialists may work to maintain its presence to promote hope (Brashers, 2001; Mishel, 1990; Paul et al., 2015; Peräkylä, 1991; Pilnick, 2002). Peräkylä (1991) describes this activity as "hope work" (p. 407) where medical practitioners use uncertain situations to create psychological comfort for patients.

In the two paediatric consultations where parents have questions about the uncertain future of their child, the genetic specialists used uncertainty as a positive 'tool' to create hope for parents. I demonstrate the activity of 'hope work' in an uncertain situation from one of these paediatric consultations in Example 10.8.

Example 10.8

Case overview: ***Case 13- Hypophosphatasia***

This paediatric consultation involves an isiXhosa couple and their two-year-old son who has a diagnosis of hypophosphatasia. Hypophosphatasia is a rare genetic condition characterised by the abnormal development of bones and teeth due to poor mineralisation. This leads to soft bones that are prone to breakages as well as loss of dentition. There are several sub-types of the condition, with childhood sub-types tending to be more severe with respiratory complications, craniosynostosis, developmental delay and low life expectancy **(**Zurutuza et al., 1999). There is no genetic testing available in South Africa and the diagnosis is typically made on a clinical basis. The couple attend the genetic clinic for a follow-up consultation for their son to assess his general wellbeing and to review the related genetic risk information. The session is conducted in English, the couple's second language.

The example below shows how the father queries the future wellbeing of his son, to which the specialist must provide an answer. The specialist responds by saying that the child is not going to be cured of the condition. The specialist also uses the father's concern to address another difficult topic about the future, namely the son's life expectancy, which is limited. The specialist positively frames uncertainty about the future and adds their interpretation of the situation: There might be future research that may be able to aid in the son's treatment, and although the condition has a low life expectancy, no-one knows if, or when he will pass away. This specialist strategy could be seen as doing hope work (Peräkylä, 1991) as a way of being

sensitive to the parent's anxieties and their psychosocial well-being within an uncertain situation.

The two examples below (Example 10.8: Part 1 and Example 10.8: Part 2) are related, however, occur at different sections of the interaction, hence the transcript numbering differs, and they do not follow on sequentially from one another.

Example 10.8: Part 1

S= Specialist; M = Mother; F = Father

189	**S:**	**Do:o you have any questions for me↑?**
190		(3.0)
191	**F:**	**The only question that I wanted to ask**
192		(1.0)
193	**F:**	**about growing up with bones that was still (.) fragile ne? (0.3) Is he ((indicating fragility of bones by hitting his hand on his wrist))**
194		**going to gro:w gro:w? (0.2) maybe to uh?**
195	S:	°Mmm mmm° ((nods head))
196	M:	((looking at specialist))
197		(3.0)
198	**S:**	**So we know that**
199		**(0.5)**
200	**S:**	**there is no medication or anything that we can give him (0.2) °to take**
201		**this away° ((points at inheritance risk diagrams that have been drawn during the consultation))**
202	M:	((nods head))
203	S:	We know that (0.2) this is what he was born with (.) It was when he was
204		first conceived the egg and the sperm came together (.) to ma:ake him
205		(.) and that↑ (.) those instructions were there from the beginning and
206		then↑ he grew
207		(1.0)
208	M:	((nods head))
209	F:	((sitting with arms folded. Focuses gaze between specialist and son who is sitting on the floor at his feet))
210	S:	So:o for him we can never↑ take that↑ instruction awa:ay↑ (.) we can
211		never fi:ix↑ the instruction either.
212	M:	((closes eyes in an introspective manner))
213		(1.0)
214	S:	So↑ to answer your question↑ he is never
215		(0.5)
216		at some stage in his life suddenly gro:ow and his (.) uhm bones and all
217		of that will be fixed.
218	F:	((small head nod))
219		(0.5)
220	S:	So we know that this is how he↑ has been bo:orn↑ and it will be like this
221		all↑ of his li:ife (.) so we can't↑ change that al. all.
222		(0.5)
223	**S:**	**We can hope for the fu:utu:ure↑ and as more and more medicine and research**
224		**is going on they can try↑ to find therapies↑ to [manage the symptoms]**
225	M:	[((head nod))]
226		(0.5)
227	**S:**	**But we'll °never be able to take it away°**
228		(6.0)

		((Father looking at son and seems to be thinking and taking in the information). Son is babbling and saying: "Dada Dada")
229	F:	Because you can see he's bright that's why I'm asking ((looks at specialist is sceptical smile))
230	S:	Mmm that's very true↑ (.) and uh I (.) I think that's ha:ard↑ ((nods head))
231		for parents as well (.) because (.) for him (.) this↑ condition doesn't
232		affect the brain at all ((frowns and shakes head))
233	M:	Mmm ((shakes head))
234	S:	It's not at all↑
235	M:	Yes:s
236	F:	Ja:a

In line 189, the specialist encourages the parents to reflect on any concerns that they may have. After an extended pause in line 190, the father responds in line 191 and 193-194 that he wants to know whether his son's bones will always remain fragile as he grows up. This response indicates the father's agenda for the follow-up consultation. The specialist replies in lines 198, 200 and 201 that the son's condition is permanent and cannot be cured with medication. They are tentative in providing this certain information through pauses and softly spoken words. They also mitigate themselves by using the pronoun *'we'* in line 200. They then go on to emphasise the permanent state of their son's condition (lines 203-221).

In lines 223-224, as if sensing the father's disappointment, the specialist introduces hope for the future by indicating that ongoing research may be able to provide improved management for their son's condition. The specialist hedges their previous statement by indicating the permanent state of the son's condition in line 227, making them less accountable to their statement made in lines 223-224 regarding possible future treatment, which may or may not happen in the future.

Example 10.8: Part 2

S= Specialist; M = Mother; F = Father

1	**S:**	**So the other↑ thing that (.) obviously you were asking about**
2		**(0.2) him (.) if it- (.) if his bones would ever get better? (.)**
3		**we know that with this condition (.) it does mean that some of**
4		**the children (.) they don't always live for very long (.)**
5		**[but for him] he's doing very↑ well for his age (.) we're**
6	M:	[((rolls eyes and breaks eye contact with specialist))]
7	**S:**	**actually very surprised↑ how well he is doing**
8	M:	((arms folded over her chest and looking at specialist))
9	S:	And he will only be able to (.) tell his↑ story (.) we will never↑ be
10		able to say to him this is what his future is going to look like (.)
11		but we do know because↑ of the condition (0.3) sometimes the children
12		don't always live for very very long
13		(1.0)

14	M:	((just looking at specialist))
15	S:	So I kno:ow it's hard↑ [that's hard] (0.4) I don't know if any of the
16	M:	[((rolls eyes and looks away))]
17	S:	doctors have ever said something?
18	M:	They told↑ us about that (0.2) they're saying 9 months (0.4) but Tsepho* ((waves hand in dismissive manner))
19		is now
20		(0.5)
21	M:	so:o bi:ig
22	S:	Bu:ut that's what we're saying↑↑ (0.4) and we can never↑ (0.2) we will ((frowns))
23		never↑ be able to tell [you this is ho:ow long] because he is going to
24	M:	[((frowns))]
25	S:	tell his own story (.) even many of the doctors are surprised↑ that they
26		(.) they're saying 9 months and he's 2↑ years old and he's doing very
27		very well.
28	M:	I love you too baby boy ((no longer looking at specialist but at her son who is babbling))
29	S:	Hhh hhh ((looking at son))
30		(2.0)
31	S:	I know for parents' that's also a hard↑ thing (.) but (.) like I said↑
32		(.) let's ho:ope↑ that in future↑ (.) they'll be able to come up with
33		mo:ore treatments↑ and therapies that maybe can help (.) Tshepo*.
34	M:	((nods head))
35		(2.0)
36	F:	((standing so cannot see facial expression as his face not within camera shot))

In Example 10.8: Part 2 the specialist addresses the topic of life expectancy with the parents. In lines 1-5 the specialist broaches the subject of life expectancy, indicating that individuals with hypophosphatasia have limited life expectancies. The specialist however carefully formulates their turn in lines 3-4 by mitigating and hedging their talk with the use of the words *'we know', 'some of the children they don't always live for very very long'.* Although delivering bad news that their son has a limited life expectancy, the way the specialist has formulated their turn leaves some uncertainty on the life expectancy of the child since no actual timeline has been indicated. The repetition of the word '*very*' could be a sort of mitigation tactic to leave room for uncertainty, yet still providing hope to the parents. This way of presenting information could perhaps be likened to the South African informal use of the words 'now now' and 'just now'. Both 'now now' and 'just now' indicates that something will occur at some point in the future, but they do not provide a definitive or clear timeline (Watermeyer, 1996).

With conveying this potentially upsetting information, the specialist adds hope and positive re-enforcement to the son's context in lines 5 and 7, by stating 'we're actually very surprised how well he's doing'. The word 'surprised' is accompanied by a rising intonation. The specialist may have formulated the sentence in this manner to emphasise the unexpected progress of the

child. After the mother indicates in lines 18-19 and 21 that the doctors predicted that their son would only live to 9 months, but is now *'so big'*, the specialist further emphasises the positive uncertainty of the situation in lines 22-23 and 25-27, by indicating that they will never know how long he is going to live because he is going to *'tell his own story.'* In this way, the specialist still supports the prediction provided by the patient's other treating healthcare practitioners (that the son has a limited lifespan) but designs their turn of talk in a manner that provides comfort for the parents too. Dwelling on prognostic uncertainty and future outcomes may be seen as a form of hope for some patients (Peräkylä, 1991; Smith et al., 2013). Previous research has also indicated that healthcare practitioners tend to positively frame patients' lifespans when faced with dire situations. In an American study of 504 terminally ill patients and 365 doctors, researchers found that doctors over-estimated patients survival rates in 63% of cases when providing information to them (Christakis & Lamont, 2000).

Even though the specialist's approach of emphasising the positive side of the situation is meant to provide comfort to the parents, it does not seem to be well received. Throughout the segment, the father does not respond to the specialist. The mother's responses also display an eye roll in line 16, a frown in line 24, and no response when the specialist tries to reassure the parents in lines 22-23 and 25-27. Instead, the mother chooses to pay attention to her son in line 28. The parents' interactional displays (or lack thereof) could indicate that the specialist's attempt towards hope work, and thus uncertainty management in the interaction, may have not been successful, as the parents may have wanted more definitive answers on their son's life expectancy.

Evidence that the parents' responses are problematic to the specialist after attempting hope work is displayed by the specialist's awkward laughter in line 29 and the two second silence in line 30. The specialist also tries to mitigate themselves from the uncertainty by stating in lines 31-33 *'let's hope that in future that <u>they'll</u> be able to come up with more treatment and therapies that <u>maybe</u> can help Tshepo*'*. Referral to other expertise, as in Example 10.8: Part 2, is a strategy used by the specialist to demarcate their boundary of expertise (Sarangi & Clarke, 2002a), and to highlight that the uncertainty in the situation is based on a universal lack of knowledge, rather than their personal lack of knowledge.

10.2.1.3 Uncertainties in adult genetic counselling consultations

Advanced genetic tests (for example microarray testing and genomic sequencing), although having the potential to provide comprehensive information, also has the potential to create further universal or personal uncertainties when they produce difficult to decipher test results, particularly VUS results. Although advanced genetic tests can be utilised in all areas of GC, most are offered in paediatric or adult settings. Although genomic testing can be more informative than traditional methods (e.g., karyotyping) in prenatal settings, it also has the potential to create complexity in pregnancy decision-making, as well as raise parent anxiety (Clarke & Wallgren-Pettersson, 2019; Richardson & Ormond, 2018). In South Africa, advanced genetic testing is typically reserved for diagnosis of complex paediatric or adult genetic syndromes, including inherited cancers.

Example 10.9 and Example 10.10 provides an example of the potential for genetic test results to cause uncertainty.

Example 10.9
S4: "…Uhm I think new technology, I think there are a lot of new testing that that uh uh offered in genetics now, new technologies. But as you say many of them of the testing will come with a lot, a lot of uncertainties."

Example 10.10
S9: "…Uhm and then I think also with the new kinds of testing, that you're kind of going to have new levels of uncertainty and variants of unknown significance is something that we've sort of, more and more going to think about."

South Africa has limited local resources for advanced genetic testing. Local patients undergoing or seeking these types of genetic tests must have the test performed by an international laboratory which can be expensive, and local medical insurance companies often do not cover these costs. Alternatively, local patients can opt for available local testing and accept the limitations thereof. Local testing is typically paid for by the government for patients in the public healthcare system, or for private patients, paid for personally or through their medical insurance cover.

Although advanced genetic testing is largely inaccessible for underprivileged patients in the public healthcare system, one genetic specialist (Example 10.11) mentioned that some patients in the public health system may be offered advanced genetic testing on a case-by-case basis. Thus, local genetic specialists working in the public healthcare system also potentially face the challenge of interpreting uncertain test results for a patient.

Example 10.11
Researcher: "And the, the, how often are those types of tests available to patients in state [public]?"
S9: "Often actually more available than in private."
Researcher: "Oh ok, that's good."
S9: "So mmm because in, the medical aids are only now starting to take on board, or well to fund you know… so you need mmm ja. So even if you've got a medical aid, you can't get a, get an expensive genetic test."

Despite the potential for advanced genetic testing options for some patients, one genetic specialist mentioned that there were limitations of local genetic testing options for many patients in the public health system. For patients who could not afford further advanced testing or who were not deemed eligible for more advanced testing through the government's merit criteria (as mentioned in Example 10.11), another layer of uncertainty was created due to the inability to offer any further diagnostic genetic testing. The example used by the particular specialist (in Example 10.12) refers to HBOC genetic testing for adult patients.

Example 10.12
S6: "I think the really difficult part of it is sort of, for example, breast cancer again where, uhm you know, we don't offer full sequencing to all of the patients, and obviously depends on family history and criteria and the rest and, and so I tend not to mention that we do have another option, uhm…that they are not afforded it, in order to avoid having to, having to…let them down by saying that we can't offer it to them."

HBOC testing for ethnically diverse populations such as those found in South Africa remains complex since most globally used HBOC testing is based on investigations from European populations (Schoeman et al., 2013; Seymour et al., 2016). Additionally, although advanced genomic testing (which can detect many genetic mutations at once) is available for HBOC in South Africa, local costs range between R9000-R25000 per test (Seymour et al., 2016) which is typically not feasible for patients in the public health system. An alternative for HBOC high risk patients is a more limited state-funded genetic test which can test seven common HBOC mutations. This limited panel-based test is more suited to patients who have either Jewish, Afrikaner or isiXhosa ancestry since most of the mutations tested for are considered 'founder mutations', meaning that they appear more frequently within particular populations groups (Schoeman et al., 2013). For these reasons, and as demonstrated in Example 10.12 above, diagnostic uncertainties may remain for patients identified to be high-risk for carrying HBOC-related genetic mutations but who are excluded from testing based on cost and access constraints, or who are not from particular population groups.

10.2.2 Genetic specialists' personal uncertainties

All the genetic specialists (12/12) mentioned feeling uncertain during GC sessions (Example 10.13) with most admitting that they did not like feeling uncertain (Example 10.14, Example 10.15 and Example 10.16).

Example 10.13
Researcher: "Do you ever feel uncertain?"
S1: "Oh ja, all the time! (laughs)"

Example 10.14
Researcher: "How do you deal with feelings of uncertainty? As a professional?"
S3:"I mean that it is extremely complex. Uh because uhm I think every time that you as a professional uh if you are uncertain, you know you are uncertain uh when it comes to dealing with something that is also uncertain for the patient, it becomes very very difficult. Uh, it's always easy to work when you are confident, you are 100% confident about what, about the information you are providing to that patient."

Example 10.15
S8: "I don't like it obviously, but I think it's a very big reality when you work in genetics. That there will be uncertainty."

Example 10.16
S9: "Ja I think look… uncomfortable I suppose."

Two genetic specialists also reported feeling personally responsible and pressurised to have a diagnostic answer for patients and their families, as well as for referring healthcare practitioners (Example 10.17 and Example 10.18). A specialist's inability to provide a diagnosis and having to deal with personal feelings of uncertainty made them feel like they were letting patients and other healthcare practitioners down**.**

Example 10.17
Researcher: "What about it do you not like?
S7: Not having an answer.
Researcher: OK
S7: "Because uhm I mean traditionally about 50% of patients with a genetic condition, we just don't have an answer, especially in the kind of special disability kind of field. Uhm most of our children have a special disability. Uhm ja just that feeling of you know the patients come back to you recurrently, they coming, they've been referred, and you know that the child has a genetic condition but you don't know what it is... and in terms of just not being able to give the family an answer."

Example 10.18
S11: "I think we all feel that drive from ourselves, from your peers, the other doctors, from the patients, to have this answer. To have this magical answer. And you are now the last stop. You know. We've been many places and you are now, now the person that needs to fix this…And that's a lot of pressure! And the fact of the matter is and then we obviously feel disappointed and we feel that we disappoint other people, because we, we, we don't make this earth-shattering diagnosis that is now everybody's expectation."

The genetic specialists' quotes above support previous literature that finding a diagnosis for rare diseases can be challenging for healthcare practitioners (Elliot & Zurynski, 2015; Knight

& Senior, 2006; Zurynski et al., 2017). An Australian study focusing on 462 children with rare diseases reported that 38% of the study cohort had consulted with more than six doctors in a quest to find a diagnosis, with 69% of the children's families reporting that the healthcare practitioners lack of knowledge contributes to the delay in diagnosis. Of the 462 children included in the study, 7% remained without a definitive diagnosis (Zurynski et al., 2017).

10.2.3 Genetic specialists' approaches to managing universal & personal uncertainties in genetic counselling consultations

Genetic specialists reportedly have different approaches to managing uncertainty in GC consultations. The most reported approach is to be honest with patients and to acknowledge feelings of uncertainty when discussing risk. 7/12 specialists reported in their interviews that admitting that they don't know the extent, or outcome, of a risk to a patient, was a useful way of dealing with feelings of uncertainty. Examples 10.19, 10.20 and 10.21 provide evidence of specialists' quotes honesty approach to managing uncertainty.

Example 10.19
S7: "I think ja I think if you're if you're honest with the patient and you explain. I think if the patient can see that you're looking, and you know, you've referred for investigations or you've referred to other specialists, you've done database searches, and you've explained what you've done and what you're looking for and that you can't find it. I think that helps."

Example 10.20
S8: "I think something with time, I've also learnt to be OK with is to say, to be honest about it. To be honest when there is uncertainty. Uhm to be honest about the information that you can give and give as accurate and as much information as you can. But if there's uncertainty say it."

Example 10.21
Researcher: "Uhm do you ever... acknowledge to a patient when they want you to give them certainty, but you can't?
S12: Yes, ja. Uhm I sometimes do say: "I wish I could give you answers but I can't." type of thing ja.
Researcher: And how do they respond to that?
S12: Generally speaking, quite well. I think they appreciate the fact that you are being human and that you can't always give answers."

The idea of openly acknowledging uncertainty to patients as a part of managing and tolerating the inescapable presence of uncertainty in healthcare has become increasingly more popular in the literature. Recent studies have emphasised the importance of acknowledging and discussing uncertainty and the limitations of knowledge with patients to enhance communication, and that medical student training needs to incorporate teachings on how to cope and manage with the presence of uncertainty in healthcare (Newson et al., 2016; Simpkin & Schwartzstein, 2016; Tonelli & Upshur, 2019). Strout et al. (2018) conducted a literature review of 67 studies

focusing on uncertainty tolerance in healthcare, concluding that uncertainty tolerance in healthcare is important for patient and health practitioner outcomes. Health practitioners who have a higher tolerance for uncertainty experience less psychological distress, burnout, and can cope better within complex medical environments. The same was seen with patients. Patients who can tolerate uncertainty also experience lower levels of psychological distress, have improved quality of life, and are less likely to orientate to avoidance behaviours such as circumventing talking or thinking about their medical concern. An Israeli study interviewing 20 patients on their experiences and expectations for doctors' visits found that even when a diagnostic answer was unavailable, how a doctor spoke to the patient creates a form of reassurance and comfort (Arieli & Tamir, 2018). The findings in this study are in keeping with literature that supports the idea of uncertainty being accepted and discussed with patients (Example 10.22 and Example 10.23).

Example 10.22
S3: "I think one of the tools that I personally usually use is the, I try to explain. To give the true and correct information to the patient.
Researcher: Ok
S3: I try my best to tell them: "Listen this is what we do, this is how we do this thing, this is the limitation of this."
Researcher: Ok
S3: "And this limitation is not only in South Africa, it's everywhere in the world."

Example 10.23
S10: "If I can ask someone, I do that. But otherwise, I just tell them. I don't know, and nobody knows. Nobody will ever know, unfortunately.
Researcher: And how do patients respond?
S10: "(sighs). I think that as bad as it is, they do at some point accept it. Because you know, it is what it is."

Other genetic specialists mentioned that to manage uncertainty, one must become personally comfortable with its presence (Example 10.24 and 10.25).

Example 10.24
S5: "Ja. No, I think, I think I mean, I think you do, you do get comfortable with uncertainty, you have to get comfortable with uncertainty. It's never troubled me to say: "I don't know" about something ok… (Laughs) it really doesn't worry me in the slightest. Uhm I'll offer to see if I can find out more information or I'll see if I can find somebody else or something better I'll send you there, but uhm I'll, for me it's not been a major concern, I don't know, ever actually. So maybe that reflects a kind of personality thing or whatever it is that uhm ja, I think that that's the benefit of experience, is that you feel more comfortable in your uncertainty, and you're not having all the answers and you know also probably a benefit of age! (Laughs)"

Example 10.25
Researcher: "How, how do you deal with feelings of uncertainty?
S11: I think probably… occasionally just as bad as everyone else. But I think it's part of being in genetics. One kind of needs to learn to live with it… because it happens to us so often. I am certainly much better at it."

One specialist also spoke about how uncertainty impacts communication with patients, but how it has the potential to take a toll on them regarding their religious beliefs and moral standings (Example 10.26). The specialist felt that even though at times they questioned their role in the situation, it was important to feel comfortable with moments of uncertainty, especially when discussing difficult topics such as termination of pregnancy and facilitating patients to make decisions with limited information. The moral dilemmas that GCs face, particular in the prenatal setting, have been described previously (Woltanski et al., 2009; Hashiloni-Dolev et al., 2019). An online survey focusing on 709 genetic counsellors' attitudes on termination of pregnancy, revealed that although genetic counsellors agree that termination should be available to all individuals, some are more conservative in their personal views on abortion than in their professional views. Much of the conservative views were based on religious and psychosocial reasons (Woltanski et al., 2009). Although no local study has focused on genetic specialists' personal or religious dilemmas when discussing and offering the choice of termination of pregnancy to a patient, a study by Harries et al. (2009) focusing on attitudes towards termination of pregnancy highlights that patients may experience barriers to termination of pregnancy (even though it is a legal right to all women in South Africa) due to local healthcare staff's personal views on abortion, and exercising their right to conscientious objection.

Example 10.26
S8: "But it's, it's hard also, I mentioned yesterday that sometimes you feel like the 'angel of death' you know, you. Its, it's also important for me as a genetic counsellor, as a Christian, as a mother, as a person, that I feel uhm… OK with the information that I give, with the options that I provide and that with, and with the justification for the options I give parents."

When feeling uncertain about a patient's risk and outcomes, three genetic specialists reported that seeking advice from colleagues was another way to manage their uncertainties. Example 10.27 and Example 10.28 provide quotes from two of the specialists.

Example 10.27
S3: "I spent like quite a long time to discuss with my colleagues and also to discuss myself to make up my mind how to give them [the patients] the information."

Example 10.28
S11: "So, I think that uh for myself, if I'm unsure, I would ask others' opinions."

The benefit of seeking advice and support from colleagues has been previously reported in nursing (Bedward & Daniels, 2005), physician practices (Hu et al., 2012) and medical genetics (Koole et al., 2017). Observational notes from this study further support the genetic specialists' quotes, as I noted several occasions where genetic specialists had formal and informal meetings

amongst each other to discuss patient-related risk assessments, management, and testing. I noted that formal clinic review meetings were held straight after two of the genetic clinics that I attended. I also noted that informal conversations were done amongst the genetic specialists during the clinics if one of them had a particular query that required advise or discussion.

Despite the useful strategies mentioned above, one genetic specialist mentioned that their response to managing uncertainty was not useful to patients. The specialist reported that feelings of uncertainty made them verbose during an interaction and that it was not helpful for a patient (Example 10.29). This was the only specialist to mention that they provided too much information when feeling uncertain.

Example 10.29
Researcher: "How do you manage feelings of uncertainty? When you feel uncertain?
S2: Ja. I struggle (laughs). When I am uncertain of something, I tend to talk a lot because I don't want to show that, well ja, that I'm uncertain or make the patient feel like I don't... which is probably an error on my part but I'm also very cautious with what I give."

Despite verbosity not being a useful strategy, the specialist has the self-awareness to know that their approach may need to be altered. The importance of self-reflection and self-awareness in GC practice is highlighted in GC textbooks (Biesecker et al., 2019; Veach et al., 2003). The genetic specialist quote above further supports what was seen interactionally in some of the consultations. In one consultation, another specialist became particularly convoluted in their communication when discussing topics that involved uncertainty. Part of this problematic communication in that consultation is presented in Example 9.29 in Chapter Six in which the specialist provides a confusing metaphor, likening uncertainty to weather patterns.

10.3 Interactional uncertainties

The next layer of uncertainty that I noted in the data was interactional uncertainty caused by contextual features. These contextual features include patient characteristics, and the way risk information is gathered, provided and responded to between genetic specialists and particular patients. Table 10.2 provides an overview of the interactional uncertainties identified across the GC consultation data.

Table 10.2: Interactional uncertainties identified during patient-genetic specialist communication that stem from contextual features

Major areas identified	**Contextual causes of interactional uncertainty**
Establishing patient risk	When patients are unable to provide accurate family information
	Guarded patient responses when discussing certain lifestyle behaviours
Conveying risk information	Limited patient literacy
	Lost in translation: Language barriers

10.3.1 Uncertainty in establishing patient risk

During the consultations, I observed that interactional uncertainty may occur when a genetic specialist attempts to establish a patient's personal and family genetic risk, and the patient is unable to provide clear information. Establishing a patient's genetic risk using their personal and family clinical history is an important part of GC, to determine and convey accurate risk outcomes (Resta et al., 2006). If a genetic specialist is unable to establish accurate patient information, accurate risk outcomes cannot be predicted.

The prenatal consultations in my data set further show that genetic specialists are unable to provide accurate risk assessments as patients are unable to provide clear information on the cause of death for different family members, unable to explain or provide a name for a family member's reported medical concern, or are unclear on personal alcohol consumption during pregnancy. Certain personal questions are also seen to evoke emotive or guarded patient responses when a specialist attempts to establish risk. In the data, the topic of alcohol consumption during pregnancy is seen to cause some patients to respond in guarded manners. When patients are unable to provide accurate risk information, not only can it lead to unclear risk assessments, but a breakdown in trust.

Two examples (Example 10.30 and Example 10.31) of interactional uncertainty during the establishment of patient risk are provided below. Both examples are taken from the information gathering phase (as described by Wessels, 2014) of two GC consultations. In this phase, a patient's personal and clinical information, as well as information about their family's health, is gathered by a genetic specialist to establish risk and to enable them to decide what risk information is important to convey to patients.

In Example 10.30, I return to the prenatal consultation that is described earlier in Example 9.33 in Chapter Nine. The example is used to demonstrate the interactional uncertainty that arises when a patient is unable to provide accurate clinical information about the causes of death of their family members.

Example 10.30

Case overview: ***Case 9 -AMA & a soft marker for Down syndrome***

This consultation involved a 42-year-old Mixed Ancestry woman who presented at one of the prenatal clinics at 21 weeks' gestation. She had been referred for GC due to her advanced maternal age, and because on an ultrasound, her baby was seen to have a flat nasal profile, an indicator for possible Down syndrome. In the examples below, the specialist conveys the patient's risk of having a baby with Down syndrome. The specialist uses two sources of information: The adjusted-risk based on the patient's prenatal ultrasound findings and the patient's age-related risk. The session was conducted in Afrikaans, the patient's first language. In the transcript below the first line of each numbered line shows the original Afrikaans language t used in the consultation (shown in italics) and the second line shows the translated English.

S: Specialist; P = Patient; Jamil* = Pseudonym for the patient's partner

15	S:	*°okay° (0.2) so daar's niks aan jou kant van die familie nie?*
		°okay° (0.2) so there is nothing on your side of the family?
16	P:	°No° ((shakes head))
		°Nee° ((shakes head))
17	S:	*Buiten gewoond (0.3) en jou ouers↑? (0.2) leef hulle*
18		*nog?*
		Out of the ordinary (0.3) and your parents↑? (0.2) are they still
		alive?
19	P:	*Nee (.) altwee al (0.2) dood*
		No (.) both are (0.2) dead
20	S:	*Albei al oorlede*
		Both passed already
21	P:	((head nod))
22	**S:**	*En waarvan is hulle oorlede?*
		And what did they pass from?
23		**(2.0)**
24	**P:**	*Asma*
		Asthma
25		**(0.5)**
26	**P:**	**((head nod))**
27	**S:**	*Asma albei?*
		Asthma both?
28		**(0.5)**
29	**P:**	**((head nod))**
30	**S:**	*Okay (.) jonk? oud?*
		Okay (.) young? old?
31		**(1.5)**
32	**P:**	*My pa was 43 uh 40=*
		My father was 43 uh 40=
33	**S:**	*=Dis maar jonk?* *((writing down information as part of family pedigree))*
		=that is young? **((writing down information as part of family pedigree))**

34	P:	*(0.3) en my ma was is*
		(0.3) and my mother was is
35		(1.5)
36	P:	62 of [(0.3) so]
		62 or [(0.3) so]
37	**S:**	*[okay °okay°] (0.3) dis jonk vir jou pa ne?*
		[okay °okay°] (0.3) that is young for your father hey?
38		**(1.5)**
39	**S:**	*Het hy komplikasies gehad as gevolg van die asma? Of het hy long*
40		*onsteking gekry? Wat het gebeur? Weet jy?*
		Did he have complications as a result of the asthma? Or did he
		get pneumonia? What happened? (.) Do you know?
41	**P:**	*(0.4) Ek sal nou nie weet nie maar ek weet hy het asma gehet.*
		(0.4) I won't now know but I know that he had asthma
42	S:	*°Okay° (0.4) en Jamil* se kant van die familie↑ (.) het hy ook*
43		*broers en susters?*
		°okay° (0.4) and Jamil*'s side of the familie↑ (.) does he also
		have brothers and sisters?
…	…	
135	S:	Weet jy of Jamil* se ma enige miskrame gehad het?
		Do you know whether Jamil's mother had any miscarriages?
136	P:	*Ek kannie se nie*
		I can't say
137	S:	*Okay (0.2) leef (.) uh (.) sy's al oorlede het jy gese?*
		Okay (0.2) alive (.) uh (.) she's already passed you said?
138	P:	((head nod))
139	**S:**	*En waarvan is sy↑ oorlede? Weet jy?*
		And what did she↑ pass from? Do you know?
140	**P:**	*Asma*
		Asthma
141	**S:**	*Ook asma?* *((writing down notes in family pedigree))*
		Also asthma? **((writing down notes in family pedigree))**
142		(0.5)
143	**S:**	*En sy pa↑? Leef sy pa nog?*
		And his father↑? Is his father still alive?
144	**P:**	*Sy pa is ook oorlede*
		His dad has also passed
145	**S:**	*En weet jy wat het met hom gebeur↑?*
		And do you know what happened↑ to him?
146	**P:**	*(0.3) Hy't ook asma gehad* *((shifts in chair and bites lower lip))*
		(0.3) He also had asthma **((shifts in chair and bites her lower lip))**
147		**(0.5)** ((specialist writing down notes in family pedigree))
148	**S:**	**Okay**
149		**(1.0)**
150	**S:**	*Klink vir my vreemd↑↑ 'n bietjie almal wat hierdie asma (0.3)*
151		*[gaan hhh hhh(0.3) uhm maar in elk geval hhh hhh okay↑]* *((giving wry smile and turns to look at GC intern who is sitting in corner of the room and observing the consultation as part of their training))*
		Sounds strange to me↑↑ a bit everyone with this asthma (0.3)
		[gone hhh hhh (0.3) uhm but anyway hhh hhh okay↑] **((giving wry smile and turns to look at GC intern who is sitting in corner of the room and observing the consultation as part of their training))**
152	**P:**	**[hhh hhhh hhhh hhhh]** **((rocking back in chair, smiling and looks at specialist and GC intern))**
153	S:	*Goed (.) so (0.3) uhm*

		Good (.) So (0.3) uhm
154		(1.5)

In the extract above, the start of the problematic sequence occurs from line 20 when the patient indicates that both of her parents have passed away. When the specialist enquires the reason for their death (line 22), the patient replies that they both passed away from asthma (line 24). The patient's verbal response occurs after a two-second pause (line 23) indicating some hesitancy in her response. The specialist's surprise at the patient's description of the cause of death is indicated in line 27 when the specialist designs their turn as a question. Asking the patient to re-confirm that both parents died of asthma could show hesitance to believe the patient's account. The patient's turn design and the response after a half-second pause (line 28) and a head nod (line 29) further indicates uncertainty in the interaction. When the patient indicates that her father was between 43 and 40 when he passed away in line 32, the specialist provides an immediate response (indicated by = symbols) in line 33 saying that the father's age of death is young. The specialist designs her turn in a surprised and questioning manner. The specialist further re-iterates her dismay in line 37 when re-emphasising her surprise that the patient's father passed away at a young age from asthma. When the patient does not respond to the specialist's dismay in line 38 (rather a 1.5-second pause occurs), the specialist probes further by enquiring whether the patient's father had a co-morbidity that may have resulted in his death (lines 39-40).

Asthma is a chronic medical condition caused by several complex environmental and genetic factors. Although asthma can lead to death, it does not typically show a strong pattern of familial inheritance with mortality appearing more frequently in elderly individuals, young children, or in association with other co-morbidities such as respiratory infections and hypertension (D'Amato et al., 2016; Lisspers et al., 2018). A lack of these additional factors in the patient's family history as well as continual probing from the specialist could indicate that the specialist does not trust the patient's responses and explanations of the cause of death for their family members.

The specialist continues to ask about Jamil's various family members (lines 44-134) (not shown in the example). Further interactional uncertainty occurs when the specialist enquires about the cause of Jamil's mother's death (line 139). When the patient indicates that her mother-in-law also passed away from asthma (line 140), the specialist responds in a surprised

manner as displayed by their turn design in line 141 where they ask the patient to re-confirm their statement in line 140. Further interactional uncertainty occurs when the patient does not respond to the specialist's request for confirmation, but rather a pause ensues (line 142). When asking about Jamil's father's cause of death (line 145), the patient responds that he too died of asthma (line 146). The pause before answering that he has died of asthma, accompanied by shifting their body and biting their lip in line 146, provides evidence that the patient has realised that her responses to the cause of deaths have become problematic. The specialist further supports the presence of interactional uncertainty by providing two lengthy pauses in lines 147 and 149 and through their verbal and non-verbal responses in lines 150-151. By saying *'sounds strange to me a bit everyone with this asthma'* and *'um but anyway okay'* accompanied with laughter and looking at their colleague (a GC intern who is sitting in the corner of the room, observing the consultation) provides clear evidence that the specialist does not trust the patient's account. The mistrust in information leads to uncertainty and an inability for the genetic specialist to provide an accurate risk assessment for the patient. The patient provides laughter particles in line 152, further supporting the problematic moment in the consultation. Laughter in medical conversations when no moment of jest has occurred could indicate that there is interactional trouble or misalignment between participants (Haakana, 2002; Sacks, 1992).

In Example 10.31 below I show a typical example from the data of how interactional uncertainty occurs in a prenatal consultation when a patient provides unclear and guarded responses to a genetic specialist's questions relating to their lifestyle behaviours. Pregnant patients' lifestyle behaviour surrounding tobacco and alcohol use during pregnancy created uncertainty in some of the interactions.

Example 10.31

Case Overview: *Case 12- AMA*

This case involved a 41-year-old isiXhosa woman who was 21 weeks pregnant. No abnormalities had been detected on ultrasound and she was referred for general AMA counselling. She was accompanied to the consultation by her partner and they were counselled in English, the couple's second language.

The example below is taken during the 'information gathering' phase (Wessels, 2014) of the consultation where the specialist enquires about the mother's personal and pregnancy health.

Here, teratogen risks are discussed in relation to maternal infections, as well as tobacco and alcohol use.

S = specialist, M = Mother; F = Father

1	S:	**And during pregnancy uh at (.) at the beginning especially↑ did**
2		**you have any fevers↑ (.) or skins rashes like chickenpox or**
3		**German measles↑?**
4	M:	Mmhhmm (0.4) no:othi:ing ((shakes head))
5	S:	°Ok°
6		(0.5)
7	**S:**	**Do you smoke↑? Or do you drink↑?=**
8	**M:**	**=No** **((breaks eye contact with specialist and looks at video recorder))**
9	**F:**	**Hh hhh** **((smiles))**
10	**M:**	**I did↑ drink before (0.3) but (0.2) occasionally** **((shrugs shoulders))**
11	S:	Mm ((nods head))
12	**M:**	**But no:ow that (0.2) I'm (0.3) expecting this↑ baby (.) I don't**
13	**S:**	**Ok (.) well (0.2) that's actually a very smart decision**
14	F:	Yes:s ((nods head))
15	M:	°Mm° ((nods head))
16	**S:**	**Uhm (0.4) until you said befo:ore↑ you were expecting you did**
17		**drink occasionally↑**
18	**F:**	**((head nod))**
19	**S:**	**Uhm when (.) when did you stop↑?**
20		(0.5)
21	M:	[((turns and looks at partner))]
22	F:	[((looks at mother))]
23	**S:**	**Using alcohol?**
24		(0.5)
25	**S:**	**Or when? (.) Ja**
26	F:	Befo:ore when °you get pregnant or when°? ((looking at mother))
27		(1.5)
28	**M:**	**Ah uh (0.3) I can't be sure**
29	**F:**	**Hhh hhh** **((rubs hand on face))**
30	**M:**	**Because I wasn't sure when (.) I can't↑ say I get pregnant (0.3)**
31		**which month↑** **((smiles awkwardly, seems uncertain and raises hand in air))**
32	S:	Ja:a ((nods head))
33	M:	You understand?
34	S:	Ja
35	M:	Because of the cycle
36	**S:**	**Ja you weren't sure**
37	M:	Ja it (.) something you see
38	S:	Mm
39	**M:**	**Otherwise (0.2) uh (.) as soon as I found out that I (0.2) I**
40		**just [stop]**
41	F:	[stop]
42	**S:**	**Well that's good (0.3) that's really good** **((nodding head rapidly))**
43	M:	Mm
44	F:	((smiles))
45	**S:**	**Uhm because we know that alcohol can affect the baby's brain**

46	**M:**	**[((big head nod))]**
47	**F:**	**[Ja:a]**
48	**S:**	**So it's good what you're doing**
49		**((head nod))**
50	**S:**	**Just (0.2) it's better not to drink when you are pregnant**
51	**F:**	**[Hhh hhh]** **((smiling))**
52	**M:**	**[°Ja°]** **((head nod))**
53	**S:**	**Good choice↑**
54	F:	Hhh hhh ((smiling))
55	M:	((nods head and smiles))

In the example above, the specialist starts by enquiring about any possible maternal infections (lines 1-3) to which the patient gives a negative response in line 4. This patient's response is accompanied by a 0.4-second pause, indicating that she is giving the question some thought; however, this question does not produce any problematic response from the couple.

However, in line 7, when the specialist asks the mother whether she drinks or smokes, the mother gives an immediate 'no' response in line 8 and looks directly at the video-recorder. The immediacy of her response is indicated by the latching between the turn constructional units in lines 7 and 8. The specialist has hardly completed her turn of talk, and the mother immediately responds, leaving no room for a pause. This rushed response could indicate that this is an uncomfortable topic for the mother. Significantly shorter gaps between turn-taking occur during troublesome conversations, e.g., arguments or when details of an embarrassing nature are discussed (Heldner & Edlund, 2010). Alcohol and tobacco consumption during pregnancy is not only considered a medical problem, but also a social concern likened to child abuse and immoral behaviour (Bell et al., 2009; Hammer & Inglin, 2014). Stigma (placing a negative label onto something or someone who deviates from the social norm (Link & Phelan, 2001)) may thus exist when a pregnant woman consumes alcohol or admits to doing so. The father's unprompted laughter in line 9, which is not reciprocated by the mother, or specialist, further indicates that this line of enquiry may be awkward. In conversation, laughing alone is considered problematic or an indication of misalignment between individuals since this activity is usually reciprocated amongst individuals (Sacks, 1992; Haakana, 2002).

The mother goes on to admit in line 10 that she did drink alcohol, but carefully mitigates her response by adding the word *'occasionally'*. She further mitigates her use of alcohol in line 12, by stating that she has stopped drinking since expecting the baby and provides further justification for her actions in lines 28 and 30. Here she creates uncertainty by stating that she

may have drunk whilst pregnant but unknowingly because she was not sure of the timing of when she fell pregnant. Her partner laughs awkwardly in line 29, indicating possible trouble with her response. She goes on to explain that she had an irregular menstrual cycle, thus may have been unaware that she was pregnant at the time. This lengthy response from the mother is a 'face-saving' strategy in which she attempts to maintain a respectful view of self in front of the specialist (Goffman, 1955; 1959). Her use of alcohol could pose a risk to her unborn baby, and by extension categorise her as a 'bad mother'.

A patient's mitigative behaviour to avoid personal responsibility has been previously described in studies involving interactions between clinicians and psychiatric patients (Weiste et al., 2018) and non-compliant diabetic patients (Helme & Harrington, 2004) when potentially contentious information is discussed. Sorjonen et al. (2006) note that when a healthcare practitioner formulates a question regarding a potentially problematic lifestyle behaviour, patients provide expanded answers to not only justify their behaviour but to mitigate and allow the behaviour to be non-problematic. In a similar way this is seen in the patient's expanded responses in the example above.

The specialist shows their uncertainty in response to the patient's statements in lines 13, 16, 19, 23 and 25 by the filler word 'uhm' and pauses. The specialist further contributes to the potential 'bad' versus 'good' mother image of the patient through their choice of responses in lines 42, 48 and 53 where the specialist attempts to maintain a positive face and relationship with the patient by applauding her for stopping alcohol use when she found out she was pregnant. The specialist also goes on to convey the associated risk of alcohol use during pregnancy in lines 45 and 50 highlighting the negative consequences of maternal alcohol consumption whilst pregnant.

Alcohol and tobacco use, and their perception of risk are shaped through cultural and societal norms. Hammer and Inglin (2014) conducted interviews with 50 Caucasian educated pregnant women in Switzerland and found that their perceptions varied to medical recommendations on smoking and alcohol cessation during pregnancy. Although smoking was considered risky, moderate alcohol consumption during pregnancy was considered acceptable. However, the women acknowledged that these acts were considered morally contentious since mothers are expected to be responsible and protect their child from any potential harm. A South African study involving 24 Black and Mixed Ancestry pregnant or one-year post-partum women from

an impoverished community in Cape Town similarly indicated that although they were aware of health messages warning against the dangers of alcohol consumption during pregnancy, these were ignored as drinking was considered 'normal' in their community. However, several of the women felt ambivalent in their behaviour and admitted feeling judged and having to defend their actions (Watt et al., 2016). Sorjonen et al. (2006) also note that questions on alcohol consumption in medical interactions are gendered. Healthcare practitioners often assume that men drink and ask questions such as "How much do you drink?" (p. 347), assuming that they drink. When asking women, however, healthcare practitioners tend to first ask whether the behaviour exists at all and if so, they ask for specifics. These societal views on gender may further exacerbate the image of a 'bad mother' for women who not only drink, but worse who drink whilst pregnant. These studies, together with the findings in this research, indicates that particular patient lifestyle behaviours and risk-related questions from healthcare practitioners regarding these behaviours can create tension and uncertainties in the interaction. The uncertainty displayed in Example 10.30 and Example 10.31 above can also be linked to the concepts of trust and mistrust. Trust can be defined as a belief in the veracity of a person or object, whilst distrust is the opposite. Mistrust can be considered a pause position between trust and distrust, where an individual is still contemplating their decision on whether the information received is to be trusted or not. Evaluating the level of trust in the information conveyed by an individual is dependent on the receiver's prior knowledge on the topic, their emotive state, their self-interest, as well as how that information is produced (Breakwell, 2020). Arribas-Ayllon and Sarangi (2014) interviewed 25 health professionals (including genetic counsellors and medical geneticists) in the United Kingdom and found that trust and distrust are constantly negotiated in GC consultations to understand patients' motivations, expectations, and goals for genetic testing. They highlight that trust and mistrust occurs on a sliding scale during GC consultations. Although their study pays particular attention to the negotiation of trust during patient decision-making in GC, the findings in this study demonstrate that trust is assessed and negotiated during other parts of GC consultations too, such as the information gathering and risk assessment stages.

An additional study by Thomassen and Sarangi (2012) focusing on GC interactions in Norway highlights how patients and genetic counsellors assess evidence and the certainty of claims by providing verbal and non-verbal cues. How information is provided, who is providing it, and the timing of when the information is provided are all thought to impact the listener's trust and level of reliability on the information stated. Hence, what information is provided and how it

is provided is assessed differently (Antaki, 1994). Listeners and speakers thus provide direct and indirect evidence of their orientations to certainty or uncertainty within interactions when providing and responding to information. When considering the uncertainty in both consultations in Example 10.30 and Example 10.31, both the genetic specialists provide interactional gestures that indicate that they are assessing the certainty and uncertainty of the claims made by the patients.

10.3.2 Conveying risk in the context of limited patient literacy

In some of the consultations, the formats used to convey risk information create moments of uncertainty for both the specialist and the patient. The uncertainty appears to be caused by limitations in the patient's literacy (including medical, science and maths literacy), which is then further compounded by the specialist trying to find a suitable format and level at which to pitch the related risk information. In Example 10.32, I return to the example of the pregnant woman whose husband has OI.

Example 10.32

Case Overview: ***Case 6- OI***

This case involved a 20-week pregnant, 31-year-old woman of Mixed Ancestry descent whose husband was diagnosed with OI. OI is a type of genetic condition that results in brittle bones, which can result in multiple fractures in-utero and in childhood. This can lead to life-long complications for affected individuals. There are several types, each with a different form of severity, and inheritance patterns. Depending on the type, symptoms may also include dental abnormalities and short stature (Winship, 2003). The patient had been referred to the clinic to discuss her husband's condition and the inheritance risk to their unborn child. This was their third pregnancy, with their two other children being healthy. The husband had two affected children from previous relationships. The session was conducted in Afrikaans, the patient's first language. In the transcript below the first line of each numbered line shows the original Afrikaans language used in the consultation (shown in italics) and the second line shows the translated English.

In the example below, whilst explaining risk information to the patient, the patient indicates that she does not understand the information being conveyed to her when the specialist pauses to check her understanding. The uncertainty for the patient occurs specifically whilst the specialist is using a numerical format to convey risk information. After realising that there is misalignment, the specialist attempts to reformulate the information into another format using figurative language, which seems to be more beneficial to the patient in facilitating understanding. Please note that the excerpt below is lengthy because the uncertainty occurs

across several turns in the conversation whilst the specialist attempts to use different formats to explain risk information and facilitate patient understanding.

S = Specialist; P- Patient ; Julian* = Pseudonym for patient's partner

99	**S:**	*So dis dan 'n twee↑ uit vier↑ kans in elke swangerskap*
		((drawing ratio on blank paper))
		So it's then a two↑ out of four↑ chance in every pregnancy
		((drawing ratios on blank paper whilst talking))
100	**P:**	***((head nod))***
101	**S:**	*Ok↑ (.) en dis die selfe as een uit twee*
		Ok↑ (.) and that's the same as one out of two
102	**P:**	**Mm**
		((head nod)
103		(0.5)
104	**S:**	*Maak daai nommer sin↑ vir jou?*
		Does that number make sense↑ to you?
105		**(1.5)**
106	**P:**	**((looking at diagram. Produces several small head nods and a smile))**
107		**(1.0)**
108	**S:**	*Of nie?*
		Or not?
109		**(7.0)**
110	**P:**	*°Ek weet nie°*
		°I don't know°
111	S:	So↑ (.) it's (0.2) so it's like (0.2) in every pregnancy it's
112		like cha:ance↑ (0.2) it's like tossing a coin↑
		So↑ (.) dis (0.2) so it's like (0.2) met elke swangerskap it's
		like cha:ance↑ (0.2) it's like tossing a coin↑
113	P:	Mm
		((head nod))
114	S:	So (0.4) uhm
115		(0.5)
116	S:	*As jy (.) so ja (0.3) elke keer is daar 'n (0.4) is daar vier*
117		*verskillende moontlikehede*
		If you (.) so ja (0.3) every time there is a (0.4) there is four
		different possibilities
118	P:	((head nod))
119	S:	*En twee van daardie moontlikhede is dat 'n babatjie osteogenesis*
120		*imperfecta kan he*
		And two of those possibilities is that a little baby can have
		osteogenesis imperfecta
121	P:	*Ja:a dokter*
		Yes:s doctor
122		(1.0)
123	S:	*En twee uit vier is die selfde as een uit twee*
		And two out of four is the same as one out of two
124	P:	((Head nod but looks unsure again. Frowning))
125	S:	*So met elke swangerskap dat jy het↑*
		So with every pregnancy that you have↑
126	P:	((head nod))
127	S:	Is daar 'n 50 persent kans↑ [(0.2)] of een uit twee kans↑ dat die
		There is a 50 percent risk↑ [(0.2)] or one out of two risk↑ that the
128	P:	*[kans]*
		[chance]
129	S:	*babatjie sal die selfde kondisise he as sy pa*
		little baby will have the same condition as his dad
130	P:	((head nod))
131	S:	((head nod))
132		(0.5)
133	**S:**	*Maak dit sin vir jou?*

		Does that make sense to you?
134		**(2.0)**
135	**P:**	*Ek verstaan nie eintlik nie hhh hh* *((places her hand on her head in puzzled manner))*
		I don't really understand hhh hh **((places her hand on her head in puzzled manner))**
136	**S:**	*Nee? (0.3) Verstaan jy nie↑?*
		No? (0.3) Do you not understand↑?
137	**P:**	**Mm mm** **((smiles awkwardly))**
138	**S:**	*Ok (0.3) watter (.) watter gedeelte verstaan jy nie↑?*
		Ok (0.3) what (.) what part don't↑ you understand?
139		**(2.5)**
140	**P:**	*Soos ek nou (.) ek het twee van uh (0.2) so so (0.3) soos dokter*
141		*dit nou vir my verduidelik nou (.) maar nou* *((rubbing chin))*
		Like I now (.) I have two of uh (0.2) so so (0.3) like doctor
		has now explained it to me (.) but now **((rubbing chin))**
142		**(0.5)**
143	**P:**	*Ek het nou al twee↑*
		I now have two↑
144	S:	*Ja:a* *((head nod))*
		Yes:s ((head nod))
145		(0.5)
146	S:	*So↑ (0.2) met ELKE↑ swangerskap (0.4) het jy hierdie kans↑*
		So↑ (0.2) with EVERY↑ pregnancy (0.4) you have this risk↑
147		(0.5)
148	P:	Mm ((head nod))
149	S:	*En met elke swangerska[p het] jy hierdie kans weer van die begin*
		And with every pregnan[cy you] have this risk again from the start
150	P:	*[het]*
		[have]
151	S:	*af*
		again
152	P:	*Yes:s*
153	S:	*So met (0.4) uh (0.3) met (.) met elke↑ swangerskap met Julian**
		So with (0.4) uh (0.3) with (.) with every↑ pregnancy with Julian*
154	P:	((head nod))
		Het jy (0.3) 'n 50 persent KANS↑ gehad
155	S:	You had (0.3) a 50 percent CHANCE↑
156	P:	Mm ((head nod))
157	S:	*Om 'n kind te he met osteogenesis imperfecta*
		To have a child with osteogenesis imperfecta
158	P:	((head nod))
159	S:	*So met jou twee↑ seu[ns wie] ouer is*
		So with your two↑ son[s who] are older
160	P:	[Yes:s] ((head nod))
161		(0.5)
162	S:	*Uhm (0.3) het jy elke keer (0.3) of hierdie instruksies (.) of* *((going back and pointing at drawn allele combination diagrams that were drawn on blank paper))*
163		*daai instruksies*
		Uhm (0.3) you had every time (0.3) either these instructions (.) or ((going back and pointing at drawn allele combination diagrams that were drawn on blank paper))
		those instructions
164	P:	((head nod and looking at diagram))

165	S:	*Want hulle het dit nie↑*
		Because they don't↑ have it
166	P:	Yes ((head nod))
167	S:	*MAAR↑ (0.2) daar was ook 'n kans dat dit kon of hierdie of daai*
168		*gewees het [(0.3)] maar dit was↑ nie so nie* *((pointing at allele combinations that have mutated gene))*
		BUT↑ (0.2) there was also a risk that it could've been this or
		that [(0.3)] but it wasn't like that ((pointing at allele combinations that have mutated gene))
169	P:	[Yes] yes ((head nod))
170	S:	((head nod))
171	P:	((head nod))
172	S:	So it's kind↑ of like playing the lottery↑
173	P:	>Yes:s< ((head nod))
174		(0.5)
175	S:	Of (.) or uh (0.4) you're tossing (0.3) you're tossing a coin↑
		Or (.) or uh (0.4) you're tossing (0.3) you're tossing a coin↑
176	P:	((head nod))
177	S:	*En jy weet nie watter kant (0.2) which side it's going to land*
		And you don't know which side (0.2) which side it's going to land
178	P:	Yes:s
179	S:	*Ja:a*
		Yes:s
180	P:	((head nod))
181	S:	*En (.) in elke swangerskap↑ (0.2) doen jy dit weer↑ van die begin af*
		And (.) in every pregnancy↑ (0.2) you do it again↑ from the start
182	P:	Mm ((head nod))
183	S:	You're tossing a coin from the start
184	P:	Mm mm ((head nod))
185	S:	*Dan sien jy (0.2) maar (0.4) is dit hierdie↑ een↑ (.) of daai (0.3)* *((pointing at allele combinations hand drawn on blank paper))*
186		*is dit hierdie↑ een↑ (0.3) of gaan di[t daai ee]n wees?*
		Then you see (0.2) but (0.4) is it this↑ one↑ (.) or that (0.3) ((pointing at allele combinations hand drawn on blank paper))
		is it this↑ one↑ (0.3) or is i[t going to be] that one?
187	P:	*[daai wees]* *((head nod))*
		[that one] ((head nod))
188	**S:**	*En met elke babatjie (0.2) is dit weer↑ die kans van die begin [af]*
		And in every baby (0.2) it is the risk again↑ from the [beginning]
189	**P:**	*[van]*
190		*die begin (0.4) yes:s (0.2) ek verstaan dit nou [(.) want]* *((head nod))*
		[from]
		the beginning (0.4) yes:s (0.2) I understand it now [(.) because **((head nod))**
191	**S:**	*[Maak]*
192		*dit meer↑ sin?*
		[Does]
		it make more↑ sense?
193	**P:**	*Dit maak meer sin nou*
		It makes more sense now
194		**(0.5)**
195	**S:**	**Okay**

In lines 99 and 101, the specialist explains risk information for autosomal dominant inheritance using ratios, during which the patient provides minimal acknowledgement tokens in lines 100 and 102. Her facial expression changes in line 102 where she seems to look confused, and there is a pause in line 103. The specialist then responds in line 104 asking if the risk information makes sense. The lines thereafter show a 1.5-second pause (line 105), a head nod and smile from the patient (line 106) and a further one-second pause (line 107). When receiving unclear responses from the patient, the specialist requests further confirmation of understanding by reformatting the question in line 108. After a significantly long seven-second pause in line 109, the patient eventually replies in line 110 by saying in a soft tone: "*I don't know*", indicating that she is uncertain whether she understands the information.

The extended seven-second pause in line 109 in the interaction could serve more than one purpose. It could indicate trouble in intersubjectivity, and by extension uncertainty in comprehension from the patient since it occurs directly after a request from the specialist to confirm understanding. Pauses (especially pauses that are longer than one second) from a recipient, after a speaker requests confirmation, could indicate trouble in mutual understanding (Jefferson, 1989; Pomerantz, 1984; Robles, 2017). Alternatively, the pause could be a management strategy from the specialist to encourage patient reflection. The latter explanation is supported by data in the specialist interviews (Example 10.33).

Example 10.33
S11: "And I think the one thing that I've learnt is to be a bit more comfortable with putting a silence... in there. So, it kind of forces the situation into something."

According to the specialist in the quote above, silence is thought to make patients reflect more slowly and deeply about their concerns and their decision-making. Silences before a turn of speech may thus indicate that a response is required that encompasses greater, and more carefully considered, cognitive input (Cappella, 1979).

In lines 111-132, the specialist attempts to re-explain the information. They use the analogy of 'tossing a coin' in every pregnancy (lines 111-112) to explain chance and try to re-explain the inheritance options using the same ratio format. They also compare the ratio of 2/4 as 50% in line 127. The reformulation could be an attempt to use another format to present the information, even though it remains a statistical format.

When requesting confirmation of understanding again in line 133, a two-second pause ensues in line 134, followed by the patient responding with awkward laughter and that she still does not understand the risk information in line 135. The awkward laughter not only signifies uncertainty but that there is trouble in the interaction (Haakana, 2002; Sacks, 1992). These lines are followed by a further confirmation request from the specialist line 136, to which the patient provides a minimal acknowledgement that she does not understand the information in line 137. This segment in the interaction provides evidence that the initial seven-second pause in line 109 is, in fact, indicative of trouble in intersubjectivity and mutual understanding, rather than the patient giving the information any deeper reflective thought.

When asked by the specialist in line 138 what part they do not understand, the patient attempts to relay her understanding back to the specialist in lines 140-141, and 143. The patient's attempt is preceded with a 2.5-second pause in line 139, together with a 0.5-second pause in line 142. The hesitancies provide further proof of uncertainty from the patient.

Upon realising that the patient does not understand the information conveyed to her, the specialist decides to use another strategy to explain the risk. They explain that the risk is present in every pregnancy in lines 146 and 149. Instead of re-using the statistical format that they previously relayed to the patient, they use the analogies of 'playing the lottery' (line 172) and 'tossing a coin' (line 175). These analogies are used to explain that in every pregnancy there is a chance to have a baby with OI and a chance to have a healthy baby. One cannot predict what the outcome is going to be because *'you don't know which side [of the coin] (0.2) which side it's going to land'* (lines 177). During this strategy of explanation, the patient seems more attentive and provides a series of head nods and 'yes' responses, possibly indicating understanding.

Some understanding from the patient seems to occur in line 189-190 when she repeats part of what the specialist says in lines 189 and says, '*I understand it now.*' in line 190. The patient's response prompts the specialist to request further confirmation of understanding in line 191-192, to which the patient replies that it makes more sense in line 193. The patient's claim of understanding seems to be accepted by the specialist with a minimal *'okay'* acknowledgement token (line 195).

The case demonstrates not only the interactional uncertainty that is created when explaining and understanding risk information but how using multiple formats to explain risk can cause confusion in some instances. The example is an unique example since it is one of only two consultations in the data set where the patient openly admits to not understanding the risk information provided to them. This overt admission of misunderstanding by the patient is in itself an important finding as it indicates that patients may not openly admit misunderstanding of the risk information conveyed to them during GC consultations. The uncertainty caused by passive patient behavior in a South African GC context is further highlighted by some of the genetic specialists in their interviews (Example 10.34 and 10.35).

Example 10.34
S2: "Some patients are difficult to gauge because like you're saying they might just give yes or no answers and they're not really communicative and in that sense, you can't get much out of them."

Example 10.35
Researcher: "Anything that patients do that's not helpful?
S12: Just sitting there (laughs)
Researcher: And not saying anything?
S12: And not saying anything (laughing) just staring blankly! (laughs)
Researcher: OK (laughs)
S12: and if you say: "How do you feel about that?" "Mmm" and then you don't know what you're dealing with! Cos you, if they understand that or not, you don't know (laughs)"

The quotes in the example above and findings from the consultation data support previous literature findings, that in local health care interactions South African patients tend towards passive deference to the health professionals' authority (Keikelame & Swartz, 2013; Patel & Dowse, 2015; Penn & Watermeyer, 2018; Wessels, 2014). Although the interactional data shows that there are attempts by specialists to encourage patient participation in the conversation and decision-making process, there is limited evidence to confirm whether most patients in the data corpus truly understood the risk information conveyed in their consultations. Most patients in this study provided either minimal head nods or 'yes' responses, or alternatively offered no verbal or non-verbal gestures when probed by genetic specialists about their understanding of risk information and/or their feelings towards the information conveyed.

When considering the rest of the consultation in Example 10.32, the patient indicated at the start of the consultation that her highest schooling qualification was Standard 4 (Grade 6 equivalent of primary school). Being privy to this knowledge early in the consultation, the

specialist should have an awareness that using complex formats and diagrams to convey risk information may have been a step too far for this patient. The approaches used by the specialist are confusing to the patient, which she confirms verbally. Evidence from the interaction indicates that risk information is initially not tailored to the needs of the patient. Providing simplistic information to explain risk from the start may have been more suited to ensuring patient comprehension, and allowing informed decision-making for this particular patient.

The value of simplistic risk information versus providing detailed risk information was emphasised in the specialist interviews (Example 10.36 and 10.37).

Example 10.36
S7: "Medical terms, like you use the absolute basics. I have, I have seen, most people are kind of in the middle. So, I've seen some people who come from quite rural areas and don't have, have very little schooling and I'd seen a few who'd told me they don't know about an egg and sperm cell… which I find very hard to believe. OK. And then there's a few where when you say: "chromosomes, DNA" like then they know what you're talking about which is really great... but most have some understanding but not in terms of what we need to explain often. So, then you kind of simplify things to a point to try make it understandable but not uh nonsensical because it's so simple… but also not too much so that they're overwhelmed. So, try and find a line."

Example 10.37
S10:"Uhm… I can't really think of a way that it went really bad. Uhm…. Because I, usually when I see… I mean it definitely has happened where I try this and try the percentages and try the numbers and I can absolutely see this person is not understanding at all what I'm talking about. Very often with, with the Xhosa speaking patients, where English is a problem.
Researcher: OK
S10: Uhm and I sort of say (laughs): "OK forget EVERYTHING I've spoken about" (laughs). And then I go: "You have a high risk of having a baby with Down syndrome. There's a big chance." Then I go, I sort of leave it at that.
Researcher: OK.
S10: Go from there. Try and keep it absolutely simple."

Genetic specialist participant S7 in Example 10.36 is the same specialist involved in the consultation example described above in Example 10.32. Although they express the importance of keeping information simple and assessing the patient's ability to understand the information in their interview, the specialist's actions and communication style in an actual patient consultation demonstrates that this may be more difficult in reality than in theory. In the consultation, despite being aware of the patient's limited maths, medical literacy and medical knowledge, the specialist explains the risk information using several high-level formats.

10.3.3 Lost in translation: The risk of misunderstandings across languages

The final contextual factor that I would like to highlight is the potential for misunderstandings and interactional uncertainty when risk is conveyed across languages. South Africa's language

and socio-cultural diversity is well known and documented, with the country having 11 official languages. Additional languages are also spoken, particularly from immigrant populations. Thus, hearing several languages at a South African hospital or healthcare facility is common. In the Western Cape Province (where all the research sites for this study was based) Afrikaans (46,6%), isiXhosa (31,1%) and English (19,6%) are the main spoken languages (Statistics South Africa, 2016). Although English and Afrikaans remain common conversational languages in healthcare consultations, these are often not the first language of many patients (Penn & Watermeyer, 2018; Shingwenyana, 2020).

Despite isiXhosa being a commonly spoken language in the Western Cape, most of the genetic specialists in this study were unable to converse in it. This finding aligns with a previous GC study in the Western Cape (Perrott, 2010). Penn (2007) reports that few local English first language speakers can converse in an African language. Furthermore, even when individuals are linguistically matched, English often becomes the conversational language in healthcare interactions due to the nature of the settings, the materials used, as well as hierarchical power dynamics between the healthcare practitioner and the patient.

Previous South African research in family medicine (Schlemmer & Mash, 2006), mental health services (Kilian et al., 2015; Swartz & Kilian, 2014), health communication (Penn, 2007; Penn & Watermeyer, 2018) and GC (Kromberg et al., 2013b; Morris, 2013; Shingwenyana, 2020; Solomon et al., 2012; Wessels et al., 2015) have indicated that language mismatches are a major barrier to local healthcare practice. Language barriers have the potential to create a break down in communication adverse health outcomes, ethical dilemmas, mistrust, patient misunderstanding, healthcare practitioner frustration on whether information is being understood, as well as a lack of rapport-building. Collectively, these can lead to feelings of uncertainty and mistrust both on the part of the healthcare practitioner as well as the patient.

In this study, all patients included in the consultation data were South African nationals. Most patients were able to understand and communicate in English or Afrikaans, even if either of these were not their first language. Two consultations involving isiXhosa patients required the assistance of an interpreter. Conveying risk information and genetic concepts may become particularly problematic when there are language mismatches present in a consultation, especially when an interpreter is not available to assist. Lack of available interpreters is typical

in many South African healthcare contexts (Watermeyer, 2020). Examples 10.38, 10.39, 10.40, and 10.41 demonstrate these language dilemmas in the genetic clinics.

Example 10.38
S1: "…language-wise I always really struggle if I know that I'm not counselling in their first language… like if I'm not speaking in their first language then it worries me that they are not getting everything that I'm trying to say to them."

Example 10.39
S7: "So, unfortunately, none of us speaks Xhosa, that would be such a bonus."

Example 10.40
S10: "…Uhm obviously for the Xhosa patients there is always the… possibility of a language barrier which doesn't make explaining a concept like genetic risk ANY easier. And where someplace like (hospital name) has a translator on hand, we don't.
Researcher: OK
S10: So, we often have to struggle with making do in English."

Example 10.41
S12: "I can only counsel in English and Afrikaans unfortunately."

Specialists also noted that at times foreign nationals would attend genetic counselling consultations and language barriers would be anticipated by the patient in advance. These patients would thus ensure that they were accompanied by an individual who could act as an interpreter on their behalf (Example 10.42).

Example 10.42
S11: "Occasionally now we get African patients speaking French…but often I see them bringing a partner because they know that it's a language not readily accessible in the service. So, we usually find that there's a partner or companion."

Conveying genetic concepts and risk information can be challenging, even when individuals are linguistically matched. In GC consultations, words such as 'DNA', 'gene', 'chromosome', 'non-disjunction', 'hereditary' and 'inheritance' may be used frequently. Previous research has indicated that there are often no known words for these concepts in African languages or no straightforward translations or equivalent terms and that low literacy and language barriers may further impede understanding (Christianson & Kromberg, 1996; Gillham et al., 2015; Morris, 2013; Penn et al., 2010; Shingwenyana, 2020; Solomon et al., 2012; Wessels et al., 2015).

Professionally trained interpreters in the South African healthcare settings are either scarce in some local healthcare settings or non-existent in others, with most interpreters assisting on an informal ad hoc basis (Beighton et al., 2012; Penn & Watermeyer, 2018; Schlemmer & Mash, 2006; Swartz & Kilian, 2014). Learning that the research sites included in this study had formal

interpreter services (Examples 10.43 and 10.44) came as something of a surprise. The specialists reported that two of the hospitals in the study had dedicated hospital interpreters, whilst the third hospital made use of a telephonic interpreting service provided by the Western Cape Department of Health. These interpreters are general hospital interpreters, thus they are not employed soley in the GC clinics nor are they specifically trained to interpret genetic terminology.

Example 10.43
Interviewer: "Are there systems in place in the clinic that can facilitate better understanding if a language barrier does arise?"
S1: "We do have translators."
Interviewer: "Ok."
S1: "And we have one translator… who always comes up to the Pregnancy Clinic for us. So, I think she could do the counselling on her own! (laughs)."

Example 10.44
S12: "…we try make use of a translator with the Xhosa patients, particularly in our pregnancy counselling situation, we have easy access to a translator, which is quite nice."

Apart from facilitating conversations and understanding of information, research has shown that interpreters also take on socio-cultural roles that extend beyond simply the translation of words. Additional roles may include cultural broker, counsellor, investigative assistant and patient advocate (Penn & Watermeyer, 2012b; 2014; 2018; Schouten et al., 2012; Sleptsova, et al., 2014). These additional roles are highlighted in a clinic experience related by participant S11 in Example 10.45 below. The story highlights how an interpreter made a patient feel 'heard' for the first time by allowing them to tell their story. The specialist viewed this as a successful consult, even if the focus was not entirely about genetic risk information.

Example 10.45
S11: "…initially when I started here there was a dedicated translator that we could call…. and we did. But unfortunately, that person got ill…long-term that wasn't renewed. And then since then, they've got a service… where we have a line that we can phone. And then we have a telephone with two handsets and there are various uhm languages that you can choose from. And to be honest, typically it works really, initially, I was really sceptical. Because uhm thinking about a counselling scenario, you want to be able to communicate with the person themselves or have somebody present uhm…
Researcher: I suppose sort of the non-verbal features...
S11: Exactly!
Researcher: that you don't see.
S11: Uhm…. however, I have since realised that for a patient who have never been able to communicate and ask all the questions that they want to ask, and for you to be able to get information…
Researcher: Mmm
S11: that uhm… you would not have had otherwise because of the nuances of the language uhm THOSE benefits have completely outweighed any of the other. And I think we have had instances where after the session, some of the mothers have been crying saying it's been the first time that they've been able to speak to somebody. And occasionally you have these long monologues (laughs) between the patient and uh the translator… and in the beginning, I was like: "oh what are they saying?" But it's also good for the patient to have somebody who she

feels that she can speak to. Uhm even if that's not me. So, in the end, I've actually had a lot of experiences where… I've seen that it's been a great benefit for the family and even if they would talk about other issues. Maybe you know that are not directly related to the information that I wanted to give through, or the counselling, or whatever. In the end, it's somebody being heard for the first time….and that I think can be quite powerful."

Although initially expressing concern about the telephonic service, the ability for a patient to express themselves and have the information explained to them in their preferred language was regarded as highly beneficial for the genetic specialist participant in the example above.

In highlighting the value of interpreters, it is also important to note that interpreters can create additional communicative complexities and uncertainties too. Evidence for additional complexities due to the presence of an interpreter is provided in Example 10.46. Analysis of the consultation suggests that uncertainty is created in the interaction due to the interpreter misunderstanding the risk information she is asked to convey to the patient.

Example 10.46

Case overview: ***Case 14- Down syndrome***

This case involved a 38-year-old isiXhosa mother and her 2-week-old daughter who had a diagnosis of Down syndrome. Prior to the consultation, the mother was unaware of her daughter's diagnosis. She reportedly only understood the instruction from the referring doctor to attend a genetic counselling consultation where further information would be explained to her about her daughter's medical 'concern.' As the mother was also unable to converse in English, an interpreter was asked to assist during the consultation. The session was conducted in English and isiXhosa. In the transcript below, for the translated sections, the first line of each numbered line shows the original isiXhosa (in italics) and the second line shows the translated English.

The transcript below focuses specifically on the section of the consultation where the genetic specialist enquires whether the mother is interested in having more children. The specialist asks the interpreter to explain two different types of risks to the patient:

1. One risk is about the mother's age. The mother is 38 years old and is of advanced maternal age (AMA). For AMA women, the risk to have a child with Down syndrome will increase with each subsequent year, independent of her previous children's medical history (Morris et al., 2002).

2. The other risk is about the patient already having a child with Down syndrome. The recurrence risk to have another baby with Down syndrome in a future pregnancy is dependent on the molecular mechanism of how the genetic fault has been acquired. The

one mechanism is known as a chromosome non-disjunction in which one of the parental chromosome copies fail to separate, resulting in trisomy 21. An alternative molecular cause for Down syndrome is a chromosome translocation in which there is a higher recurrence risk in future pregnancies. For this consultation, genetic testing confirmed that the cause for the baby's diagnosis of Down syndrome was due to chromosomal non-disjunction. The recurrence risk for non-disjunction to occur again in a future pregnancy is approximately 1%.

The interpreter's uncertainty and potential misunderstanding of these two conveyed risks are evident in the transcript below. Furthermore, due to the existing language barrier, the specialist does not realise the misunderstanding from the interpreter, nor does the interpreter request clarity from the specialist to ensure that the correct risk information is conveyed to the patient.

S = Specialist, I = Interpreter, M = Mother

4	S:	Are you wanting some more children?
5	I:	*Usafuna abanye abantwana?*
		Do you still want more children?
6	M:	*Ha-a* *((sitting with baby asleep on her lap. Baby is wrapped in a thick blanket))*
		No ((sitting with baby asleep on her lap. Baby is wrapped in a thick blanket))
7	**S:**	**But if you did want more children (.) the chances of this happening**
8		**again are related to your age.**
9	I:	*Abathuba axhomekeka kwiminyaka yakho (0.2) neh?*
		That chances are dependent on your age (0.2) alright?
10	M:	((small head nod. Looking at interpreter))
11	S:	So as you get older and older and older (.) the chance for this to
12		happen (0.2) incre:eases:s.
13	I:	*Ja, ayanyuka amathuba okuba mayenzeke, xa usowukhula ukhula ukhula*
		Yes, the chances for it to happen increase, as you grow older you grow
14	M:	Okay
15	**S:**	**So then, depending↑ on your age we can then give you a RISK↑ for this**
16		**to happen again (0.4) but we know for you now at this age↑ and because**
17		**you've already had a child with down syndrome (0.3) the chances for it**
18		**to happen again are probably about 1 percent.**
19	**I:**	*Umhlawumbe ke amathuba akamaninzanga ke kuba usowunaye umntwana*
20		*Onjeya, then amathuba akazuphinda abemaninzi, uba uphinde xa*
21		*ufumana onye umntwana kowuphinda kube nje.*
		Maybe your chances are less because you already had a child like this
		then your chances are few if you want another child it **might** be like
		this.
22	**M:**	**.hhh** **(worried facial expression and looks away from interpreter and gazes at the floor)**
23	S:	And if you do fall pregnant again (0.3) then you can phone us and we
24		can book you into our clinic so they can scan to see if the baby has a
25		higher or lower chance to have Down syndrome. ((places hand up near her head to indicate 'high' and then down at her knees to indicate 'low')

26	I:	*Kuyimfuneko ukuba ukuba uyafuna (.) Kufuneka bahlole u-amathuba*
27		*aphezulu okanye aphantsi ukuba unomntwana onje ((places hand against her stomach- indicating an ultrasound scan procedure))*
		So it is necessary that if you want to (.) they have to check the
		chances are they high or low such that you have a child like this ((places hand against her stomach- indicating an ultrasound scan procedure))
28		(0.5)
29	**I:**	*So kufuneka ke xa ufuna ukuphinda ukuba mawube ujongwe ukuba amthuba*
30		*Anjani (.) makhulu na nokuba mancinci ukuba mawuphinde ube nombtwana*
31		*onje (0.2.) Uyaqonda?*
		So, if you want to repeat that, you have to look at it. **The chances**
		are big (.) big and small for a child like that (0.2) **Do you**
		understand?
32	**M:**	**Mm** **((small head nod))**
33	I:	*Ja, ufowunele kulaclinic, bazakunika eza-numbers ufowune xa*
34		*Unophinda ufune ukuba nomnye umntana*
		Yes, you must call that clinic, they will give you the numbers to
		call if ever you would want to have another child=
35	M:	*=°Hayi (.) andifuni mntwana°* *(shifts uncomfortably in chair and looks at interpreter))*
		=°No (.) I don't want a child°. (shifts uncomfortably in chair and looks at interpreter))
36	I:	She doesn't want ((shakes hands and looks at specialist))
37	S:	She's not? ((looking at interpreter)
38		(1.0)
39	S:	Okay ((looks at patient))

In lines 7-8, the specialist indicates that the chance of having another baby with Down syndrome is dependent on the patient's age, which the interpreter translates for the patient in line 9. In line 11-12, the specialist explains that as the woman gets older the chance of having a child with Down syndrome increases. When the interpreter conveys this information in lines 13, she substitutes the specialist's word 'older' for the word 'grow'. This shows how the choice of words across languages can be altered, having the potential to alter the meaning of the intended message. Although this could have potentially caused misinterpretation, the patient seems to understand the message, indicated by a confirmatory 'okay' in line 14. Translation and interpretation across languages are complex. South African GC studies have shown that the translation of genetic risk information can be particularly complex in local GC interactions where words such as 'genes' and 'chromosomes' do not exist in indigenous languages (Solomon et al., 2012).

In lines 15-18, the specialist relays slightly different risk information, now referring to recurrence risks in future pregnancies based on the mother's history of already having a child with Down syndrome. Here, the specialist does not clarify to the interpreter that this is a

different risk (recurrence risk owing to chromosome non-disjunction) to the former risk spoken about (which relates to the mother's age). Evidence for the interpreter's uncertainty about the risk information is provided by the use of uncertainty markers *'maybe'* in line 19 and '*might*' in line 20. These are used by the interpreter to mitigate her interpretation of the specialist's message due to her uncertainty of whether she is conveying the correct information or not. The use of mitigative language is an interactional technique used by a speaker to manage the risk of being challenged, proved wrong, and to save face, amongst other functions (Caffi, 1999; Thomassen & Sarangi, 2012). The patient's in-breath and worried facial expression indicate that the information received about her personal risk is not comforting nor reassuring.

After the specialist indicates that the patient has the option to have a risk assessment if she falls pregnant in future (lines 23-25), the interpreter conveys this information in lines 26-27. In lines 29-30, the interpreter reformulates what she had just conveyed and repeats the message. The interpreter, however, uses the confusing phrase '*the chances are big (.) Big and small for a child like that.*" This contradictory statement has the potential for patient confusion, which is supported by the interpreter's question of "*Do you understand*?" in lines 30-31. Here it seems the interpreter is uncertain about whether she has understood the information from the specialist and is also concerned as to whether her explanation has made sense for the patient. The patient's minimal response token of '*Mm*' in line 32 provides little reassurance to the interpreter as to whether the patient has understood the risk information.

During this time, the specialist is not involved in the conversation and relies on the interpreter to understand and convey the correct message. The specialist is only invited back into the conversation in line 36 when the interpreter indicates that the mother doesn't want more children. At that moment, the interpreter does not relay the patient's uncertainty, nor does she create an opportunity to correct the information by asking for clarification from the specialist. The specialist is also seemingly oblivious to what has just transpired and that there has been potential misunderstanding and uncertainty regarding the accuracy of the interpretation of the risk information conveyed. In the example above, the interpreter does not admit her uncertainty around the interpretation of the risk information, nor does she request clarity from the GC. Hence, in essence, the interpreter contributes to interactional uncertainty.

The interpreter may not have been comfortable to admit her uncertainty to the genetic specialist as it would imply that she is not confident or competent in her role. Research focusing on

interactional repair attempts during Korean interpreter-mediated witness examinations in Australian courts demonstrates that facework may be an important part of interpreters' actions and responses when providing interpretation across languages (Lee, 2009; 2013). In moments of actual or potential misunderstanding, an interpreter may choose to either try to repair miscommunication by requesting clarification, or alternatively may choose not to and to rather move on with the conversation to maintain a positive face with the other individuals, or audience (Lee, 2009). The interpreted interaction therefore strongly hinged on trust. Trust that the correct message is conveyed, and trust that the interpreter will indicate when they are uncertain and request clarification to ensure that the correct message is conveyed. Example 10.46 above supports the claim that conveying risk information across languages, even in the presence of an interpreter, adds additional layers of risk and uncertainty to what is already a complex task.

10.4 Conclusion

This chapter demonstrates the different layers of uncertainty in local GC consultations. Genetic specialists acknowledge the inherent presence of uncertainty in medical genetics and that those feelings of uncertainty impact the communication process within GC consultations. Uncertainties can be divided into universal and personal uncertainties and interactional uncertainties influenced by contextual factors. A patients' personal characteristics such as an inability to provide accurate personal or family information, literacy challenges and language barriers are particularly noted in local GC consultations. Genetic specialists also report on their personal reactions to uncertainty and provide insight into how they manage moments of uncertainty in interactions. The findings in the chapter highlight the complexities of conveying uncertain risk information and how contextual factors may add further uncertainties and risks to an already challenging task.

CHAPTER 11: GENETIC COUNSELLING GOALS: EXPECTATIONS VERSUS REALITY

11.1 Introduction

This final results chapter focuses on GC professional goals. I argue that the South African setting may make it difficult to achieve certain GC goals in practice. I have chosen to highlight some of the goals of GC: Firstly I focus on the tasks of establishing a patient's agenda, tailoring risk information according to the individual patient's needs, and simultaneously ensuring that a patient is informed. Secondly I focus on the GC goal of remaining neutral when imparting risk information and how differing patient and genetic specialist risk perceptions may influence this goal. The data from the study indicates that achieving balance and addressing all these goals simultaneously in a GC consult can be difficult and that several interactional tensions and uncertainties can occur.

The chapter starts by presenting the genetic specialist's challenge of attempting to balance the patient's agenda with their own, tailoring risk information for a patient, and ensuring that the patient is informed of all the relevant risks. The chosen examples demonstrate the difficulty of this task, and how misalignment in communication occurs. The section also provides genetic specialist interview examples and highlights the value of interactional research and its ability to highlight differences in what is 'said' and what is 'done' in practice.

The chapter further presents data on the issue of neutrality and non-directiveness. I argue that achieving neutrality whilst imparting risk information is particularly challenging for genetic specialists and that personal risk perceptions play an influential role in how risk is spoken about in GC consultations. I also touch on patient's fatalistic views and how this may impact their response to genetic risk information. Even when not explicitly conveyed, evidence from the data shows how genetic specialists may implicitly display preferences for patient actions based on their own perception of a particular risk. Preference of action is particularly noted in the prenatal consultations when genetic specialists offer patients the option of invasive genetic testing.

11.2 Balancing agendas, tailoring risk information & ensuring that the patient is informed

In the consultation data, the majority of the consultations are dominated by genetic specialist talk, with specialists appearing to guide what risk information is spoken about and the volume thereof. Almost all of the consultations demonstrate a typical pattern of talk: An introduction and clarification for a patient's reason for referral, establishing a mutual agenda, collecting background information about the patient and their family to assess and establish risk, providing risk information and offering genetic testing (if appropriate), decision-making and reflecting on the risk information, referring to further management and ending the consultation. I will not elaborate further on the different phases and general types of discussions in South African GC consultations, as these are described extensively by Wessels (2014) who focused on the communication process in AMA-referred GC consultations in Johannesburg.

For this study, I have focused on different types of risk discussions. However, I highlight the genetic specialist and patient agenda (or lack thereof) as it provides an understanding of how genetic specialists may tailor risk information according to patient's informational needs or psychosocial concerns. The reader is reminded that when using the term agenda in this thesis, I mean the expectations of an individual for the GC consultation.

In the data, genetic specialists typically verbalise clear agendas and what they want to discuss with a patient. For example, gathering personal and family clinical information, and relaying related risk information to a patient seems an important part of the genetic specialist's agenda. Regarding the patient's agenda, the data shows the following different patterns:

- Attempts are made to explore a patient's full agenda (Example 11.1)
- A partial patient agenda is established (Example 11.2).
- The patient provides an agenda at the start of a consultation, but the specialist's agenda takes precedence (Example 11.3).
- A patient does not provide an agenda even when solicited by the specialist (Example 11.4).
- A patient's agenda becomes apparent during, rather than at the start, of a consultation (Example 11.5).

Table 11.1 provides an overview of agenda setting patterns observed in the GC consultations.

Table 11.1: Agenda setting patterns observed in the consultation data

Type of consultation	**Prenatal (9 consultations)**	**Paediatric (5 consultations)**	**Adult (4 consultations)**
Genetic specialist indicates own agenda	All	All	All
Genetic specialist directly enquires about patient's agenda/expectations/concerns	1/9	0/5	2/4
Genetic specialist enquires about 'why' the patient has been referred to GC.	5/9	3/5	2/4
No enquiry about the patient's agenda and only the specialist agenda is stated.	3/9	2/5 (both cases involve an interpreter)	0/4

11.2.1 Exploring the patient's full agenda

Example 11.1

Case overview: ***Case 17- Predictive Counselling for HBOC***

This case involves a 39-year-old woman of Mixed Ancestry who had been referred for GC as her younger sister had recently been diagnosed with breast cancer. Further investigation on her sister's DNA confirmed a *BRCA2* mutation, which is associated with an increased risk for HBOC. The confirmation of a *BRCA2* mutation in the patient's sister, together with an extensive family history of cancer, places the patient at an increased risk to carry the same genetic change in her own DNA, and by extension a risk to develop cancer. The patient was asymptomatic as the time of attending GC and was offered genetic testing based on the presence of a *BRCA2* mutation in a close family member. The session was conducted in English, the patient's second language.

I show in the example below how the specialist establishes the patient's agenda at the start of the consultation by enquiring what her concerns are and what specifically she would like to discuss. Together they then establish a mutual agenda for the consultation.

Although the patient's agenda is well established by the specialist, I argue that this is somewhat of an unusual case since the patient already had a good understanding of what to expect in the GC. The patient in the consultation example is in a unique position in comparison to the majority of patients attending a first time GC consultation as she is already aware of her cancer risks, the chance of being a *BRCA2* carrier because of her family history, and the option to have genetic testing prior to coming to GC. She, together with other at-risk family members, had an

in-depth discussion with the referring healthcare practitioner regarding her cancer risk, genetic risks and genetic testing options.

S = Specialist; P = patient

65	**S:**	**So what have you:u read? (0.2) or what questions are you coming**
66		**with today >or what do you↑ want to discuss today<**
67		(0.5)
68	P:	uhm
69		(0.2)
70	P:	Actually (0.4) to be honest ((frowns, leans back and folds arms over chest))
71		(3.5)
72	**P:**	**How long does this take?**
73		**(0.5)**
74	**S:**	**The tests?**
75	**P:**	**Yes**
76	**S:**	**Oka:ay (.) that's a good question↑**
77		**(0.5)**
78	**P:**	**How long will it take possibly to get an answer**
79	S:	Ja ((head nod))
80		(0.5)
81	S:	[A:and?]
82	P:	[Uhm (.)] like the doctors said (0.2) or the counsellor said at
83		the
84		(0.5)
85	P:	uhm (0.3) at the [clinic↑] ((frowns and points at window))
86	S:	[At L33] (.) ja ((head nod0)
87	P:	Yes (0.2) they said (0.3) uhm (0.4) that there's a 50 percent chance that
88		we DO↑ have it
89	S:	Mmm ((head nod))
90	P:	Because it's so wide spread in our family↑
91	S:	Mmm
92		(1.0)
93	P:	Uhm
94		(2.5)
95	P:	They told me about (0.4) say now I'm (.) I'm just saying↑↑ ((looking up at ceiling & seems to be thinking))
96	S:	Yes
97	P:	I don't (0.3) I'm not looking that wa:ay↑ ((closing eyes and using hands to demonstrate that she's not going a certain direction))
98	S:	Yes (0.2) [BUT]
99	P:	[I just] want to make sure
100	S:	Ja
101	**P:**	**SAY now I DO have it** **((places both hands on her chest))**
102		**(0.5)**
103	**S:**	**Yes**
104		**(0.5)**
105	**P:**	**What's gonna happen then?** **((produces awkward smile))**
106	**S:**	**Oka:ay↑ (0.2) that's a very important question that I have to**
107		**answer for you [today]**
108	P:	[like th]ey said you have to take out your ovaries
109		(0.2) well I don't have a problem with it
110	S:	Ja
112	P:	I don't want any more kids! Hhh hh hhh

		((slaps her hands on her legs))
113	S:	°Ja° ((head nod))
114	P:	That's not (0.4) uhm
115	S:	Mmm
116	P:	Uhm
117		(1.5)
118	**P:**	**that's about it**
119	**S:**	**Oka:ay**
120	P:	And I belie. (.) I mean honestly↑ (0.2) I TRUST you guys
121	S:	Mmm (0.4) okay ((nodding head))
122	P:	I really trust you guys
123	S:	[°really appreciate that°]
124	P:	[I know you (.) I really] do↑ trust you guys
125	S:	°okay°
126	P:	<Don't TRUST so much the internet> you know trust ((frowns))
127	S:	hhh hhh [okay hhh]
128	P:	[badly so] information may be better
129	S:	okay
130	P:	I DID read on↑
131	S:	See the internet gives you a lot of information↑ (.) but it doesn't
132		give you the CONTEXT↑
133	P:	Yes:s! ((raises hands in agreement)
134	**S:**	**So:o (0.3) we're here to give you the context today**
135	P:	°Mm° ((head nod))
136	**S:**	**Okay (0.2) So I'll answer↑ those questions for you about (0.2)**
137		**the testing↑ (.) and the process↑ (.) and the timing↑ (.) of**
138		**results and that**
139	P:	((nods head, arms folded across chest & looking at floor))
140	**S:**	**And then also↑ we can discuss (0.2) what does a result like that**
141		**mean↑ (.) and (.) and how would that be for you:u (.) depending**
142		**on (0.2) what that result is**
143	P:	Mmm ((head nod))
144		(1.0)
145	S:	And then of course we are here to support you:u↑ (0.3) through
146		this process
147	P:	°ok°
148	S:	°ok?°
149	P:	((head nod))
150		(1.0)
151	**S:**	**Okay so is that sort of what we can discuss today? [(0.2) Is]**
152	**P:**	**[Yes:s] ((frowns))**
153	**S:**	**there anything else you want to discuss today?**
154	**P:**	**You know what (0.2) I have so (.) a lot of questions**
155	S:	Mmm
156	P:	but when I walked in here (0.2) I was just calm and say
157		(0.5)
158	P:	.hhh uhm (0.3) I have faith (0.2) I prayed about this
159	S:	Mm
160	P:	I have been praying since I found out my sister had cancer
161	S:	Mm
162	P:	About this
163	S:	((head nod))
164	**P:**	**So all the questions↑ (0.3) tha:at b. (0.2) s:s:stu= ((waving both hands in front of face))**
165	**S:**	**=build up**

166	P:	Yes:s. Is just gone
167	S:	All gone

In the example above at the start of the consultation, the specialist enquires about the patient's agenda by asking her what questions she has and *'what do you want to discuss today?'* (lines 65-66). This line of questioning is different from the other consultations in this study where the specialists typically ask patients why they have been referred, rather than what their personal concerns are (examples of this can be seen in the examples that follow in this sub-section of the chapter).

The patient then proceeds to indicate her own agenda by stating that she wants to know more specifically about the genetic test and how long a result would take (lines 74 and line 78), and if she was a carrier for the *BRCA2* mutation what would that mean for her and what would happen thereafter (lines 101-105). The specialist's acknowledgement of the patient's agenda is displayed in line 76 and lines 106-107 where they say the patient's questions are *'good'* and *'important'*. The specialist further indicates that they personally have to answer the patient's questions, displaying that they have the expertise and authority to do so and can alleviate some of the patient's uncertainty on the matter. After some further thought, the patient states *'that's about it'* in line 118, suggesting the previously stated questions are her main concerns and agenda for the consultation.

After some discussion about the patient having read some information on the internet (in addition to the information already received by the referring healthcare practitioner), the specialist indicates their agenda by saying that although the patient already has information, the specialist aims to contextualise the risk information for the patient (line 134). They then provide their agenda for the session in lines 136-142, but do so in a manner that aligns with the patient's agenda. The specialist checks with the patient that their stated agenda and plans for the consult is agreeable by stating: *'Is that sort of what we can discuss today?'* (line 151) and if *'there anything else you want to discuss today?'* (line 153). The repeated question about further patient agenda in line 153 and the use of the hedging word *'sort of'* in line 151 demonstrates that the specialist may be uncertain that they have correctly captured the patient's agenda. The patient indicates that they may have had additional questions, but no longer do, which the specialist then accepts and proceeds to the remainder of the consultation.

This example demonstrates a thorough attempt by the specialist to establish the patient's true agenda, which has been successful. The relative ease for establishing the patient's agenda is the patient's willingness to engage in dialogue, and to ask questions. During the consult (transcript of this section is not shown), the patient verbalises and discusses how she *'trusts'* the genetic specialist and the genetic clinic. I claim that the patient's ease at discussing their agenda is linked to her trust in the genetic specialist. A Canadian GC study shows that when there is a level of trust between a patient and a genetic counsellor (and by extension other genetic specialists), the patient is more willing to engage in the dialogue of the consultation and share their concerns more openly (Thain et al., 2019). The patient's level of assertiveness and comfort may also be present because the patient has already heard this GC-related information and thus has more knowledge about GC and the content of the discussion than what a typical patient referred for GC in the local context would have. A patient with no background information may not be as assertive or able to produce an agenda, as I will show in the latter examples in this sub-section.

11.2.2 Establishing a partial patient agenda

In Example 11.2 (Part 1, Part 2, and Part 3) I return to a previously shown consultation. I demonstrate that this is a typical example of how patients are asked a partial agenda, since the patient is only asked why they have been referred to GC, and not what their concerns may be. The example further shows how the specialist provides a clear agenda for what their expectations are for the consultation. Although asking why patients have been referred may seem like an attempt to establish a patient's agenda, the questions do not explore patient concerns, but rather prompt the patient simply to convey the instruction they were given for GC referral from another healthcare practitioner.

Example 11.2:

Case overview: ***Case 7- Soft markers for Down syndrome***

The case involves a 21-year-old pregnant woman of Mixed Ancestry who presented at one of the prenatal clinics at 20 weeks' gestation. This was her first pregnancy and she had been referred for GC to discuss the option of diagnostic invasive testing after the baby was found to have several markers for Down syndrome.

Prior to attending the GC consultation, the patient had watched an educational video on Down syndrome which is routinely offered to all AMA pregnant women, and pregnant women where soft markers have been identified on ultrasound, at one of the prenatal genetic clinics. The video included details of the condition, as well as screening and invasive prenatal tests (including their pros and cons) that can be offered to detect Down syndrome in utero.

After hearing about her unborn baby's risks and being offered an invasive test during the GC consultation, the patient decided that she did not want to go ahead with prenatal genetic testing. The session was conducted in Afrikaans, the patient's first language. In the transcript below the first line of each numbered line shows the original Afrikaans language used in the consultation (shown in italics) and the second line shows the translated English.

Example 11.2: Part 1

S= Specialist; P= Patient

1	S:	*Ok so (.) soos ek gese het (0.2) my naam is Lily*↑ (0.2) ek is een*
2		*van die genetiese spesiaaliste* wat werk hier by die sonar departement*
		Ok so (.) like I said (0.2) my name is Lily*↑ (0.2) I'm one of the
		genetic specialists* who works here at the sonar department
3	P:	((head nod))
4	S:	Ok ((head nod))
5		(0.5)
6	**S:**	*.Hhh (0.2) uhm (.) so ek will eerste sins vir jou vrae*↑ *(0.3) weet jy*
7		*hoekom jy hier*↑ *is vandag?*
		.Hhh (0.2) uhm (.) so I want to firstly ask you↑ (0.3) do you know why
		you are here today?
8	**P:**	*°ja°* *((shy smile))*
		°Yes° **((shy smile))**
9	**S:**	*Hoekom is jy verwys?*
		Why were you referred?
10		**(0.5)**
11	**P:**	*Vir die baba se niertjies*
		For the baby's kidneys
12	**S:**	**((head nod))**
13	**P:**	*Om net seker te maak dat alles ok is*
		Just to make sure that everything is ok
14	S:	°Ok° ((head nod))

In Example 11.2: Part 1 above, the specialist introduces themselves at the start of the consultation (lines 1-2). The specialist asks the patient whether they know why they are at the GC consultation (lines 6-7), and why they have been referred (line 9). The questions demonstrate that the patient has not attended the consultation on their own accord, but rather that they have been told to be there by another healthcare practitioner. The patient explains that they have been referred to GC because of their unborn baby's kidneys and to check that everything is ok (line 11 and line 13).

In lines 15-32 (not shown) the specialist enquires where the patient has been referred from, and when they had a scan at the clinic. The patient replies that they have been referred from a smaller clinic within the Cape Town metropolitan and that they had had a scan at the clinic three days ago.

Example 11.2: Part 2

S= Specialist; P= Patient

33	**S:**	*Ok (0.3) uh (.) wat het (0.2) wat het hulle vir jou gese aan daai kant*
34		*van die niere?*
		Ok (0.3) uh (.) what did (0.2) what did they tell you on that
		side about the kidneys?
35	**P:**	*Dat die niertjies 'n bietjie te groot↑ is*
		That the kidneys are a little bit too big↑
36	S:	*Ok↑ (0.3) en=*
		Ok↑ (0.3) and=
37	P:	*=en*
		=and
38	S:	*=j:a:a?↑*
		=ye:e:s?
39	**P:**	*En dat die kind (0.2) en dat hulle my verwys net om SEKER↑ te maak*
40		*(0.2) ek is nie seker nie (.) maar dat daar niks verkeerd is met die*
41		*baba nie*
		And that the child (0.2) and that they are referring me just to make
		SURE↑ (0.2) I'm not sure (.) but that there is nothing wrong with the
		baby
42	S:	*Ok (0.4) so ja:a (0.2) die dokters wat hierdie scans doen is baie goed*
		Ok (0.4) so ye:es (0.2) the doctors that do these scans are very good
43	P:	((head nod))
44	S:	*En hulle het babatjie mooi↑ deur gekam↑ (0.2) na baba (.) alles (.)*
45		*alles van babatjie gekyk om te sien of hulle enige iets anders daar*
46		*dalk optel (0.4) wat miskien↑ (0.2) uh bekommerwekkend kan wees*
		And they combed↑ baby thoroughly↑ (0.2) with baby (.) everything (.)
		looked at everything of the baby to see if there was anything else
		that they might detect (0.4) that maybe↑ (0.2) uh is worrying
47	P:	((head nod))
48	**S:**	*En wat het hulle vir jou gese van die scan vandag?*
		And what did they tell you about the scan today?
49	**P:**	*Dat (.) dat dis nie net die niertjies nie (.) dat daar nog iets op die*
50		*brein is* *((touching her head))*
		That (.) that it's not just the kidneys (.) that there is also
		something on the brain

		((touching her head))
51	S:	Mmm ((head nod))

In Example 11.2: Part 2 above, the specialist continues to probe the patient about their reasons for the referral and what she has been told about her unborn baby's health. When asked for more specific details about the baby's kidneys by the specialist (lines 33-34) the patient replies that the baby's kidneys are '*a little bit too big'* (line 35) and that she has been referred to the hospital's pregnancy clinic for a more detailed assessment (lines 39-41). The use of the words *'little bit'* demonstrates that the patient is uncertain as to what the ultrasound findings mean.

In lines 42-47, the specialist highlights the expertise of the doctors at the hospital's pregnancy clinic and that when they performed an ultrasound assessment for the patient and her baby, they did a thorough examination of the baby. In line 48, the specialist asks the patient what she has been told about the results of the ultrasound scan at the hospital, to which the patient replies that an additional problem has been noted in the baby's brain (lines 49-50).

In lines 52-86 (not shown) the specialist goes through the patient's ultrasound report (that they have a copy of), describing the various problems and soft markers that have been identified in the baby. In addition to the kidney and brain problems, soft markers have also been detected in the baby's heart. The patient is aware of the report which has been briefly discussed with her by the fetal medicine specialist before the GC consultation. The specialist explains that these markers that have been identified in the baby could indicate an underlying genetic problem such as Down syndrome, but that they could also mean nothing, and that the baby is healthy. There is thus uncertainty in what these ultrasound findings mean.

Example 11.2: Part 3

S= Specialist; P= Patient

87	**S:**	daar is dan 'n moontlikheid dat hierdie babatjie Down sindroom KAN he
		there is then a chance that this baby COULD have Down syndrome
88	P:	((small head nod))
89	S:	*Ons se nie↑ dit IS Down sindroom nie (0.2) maar dit bedoel dat die*
90		*kanse is groter↑ (.) as die wat nie↑ hierdie (0.2) gesien het*
91		*nie*
		We are not↑ saying that it IS Down syndrome (0.2) but it means that
		the chance is bigger↑ (.) in comparison to those that this was not↑
		seen
92	P:	((small head nod))
93	S:	Maak dit sin vir jou?
		Does that make sense to you?
94	P:	*°Ja°* *((head nod))*

		°Yes° ((head nod))
95	**S:**	*Ok (0.4) so↑ voor ons in hierdie detail in ga:a:an (0.4) uh:hm (0.2) ((pointing at the ultrasound report in the patient's file which is on the table))*
96		*ons vrae altyd (.) uit oor die familie geskiedenis en die swangerskap*
97		*(0.3) kan het eers daai↑ inligting van jou af kry?*
		Ok (0.4) so↑ before we go:o↑ into this detail (0.4) uh:hm (0.2) ((pointing at the ultrasound report in the patient's file which is on the table))
		we always ask (.) about the family history and the pregnancy (0.3) can
		I first have that↑ information from you?
98	**P:**	**((Head nod))**
99	**S:**	*En dan (0.2) en dan sal ons praat oor wat hulle gesien het op die*
100		*[scan↑] (0.2) en die vide[o↑ (0.4)] en wat (.) wat hierdie (0.2) kan*
		And then (0.2) and then we will speak about what they saw on the [scan
		(0.2) and the v[ideo↑] (0.4) and what (.) what this can
101	**P:**	**[((head nod))] [((head nod))]**
102	**S:**	*beteken (0.3) wat is die kans dat hierdie babatjie Down sindroom kan*
103		*he*
		mean (0.3) what is the chance that this baby has Down
		syndrome
104	**P:**	**((Head nod))**
105	**S:**	*Is dit reg so?*
		Is that alright?
106	**P:**	*Ja* *((head nod))*
		Yes **((head nod))**
107	S:	Ok ((starts focusing on the patient's file and has a pen in their hand to start writing down further information))
108		(1.0)
109	S:	*So ek wil net check is dit jou kontak nommer? (0.2) 086******* ((looking at information on the patient's file))*
		So I just want to check is this your contact number? (0.2) 086******* *((looking at information on the patient's file))*

In Example 11.2: Part 3 above, in lines 87-94 the specialist reiterates that the findings could mean that the baby has Down syndrome, but it may also not. The specialist confirms whether the patient understands this uncertainty (line 93), which the patient confirms that she does understand (line 94).

The specialist's agenda becomes apparent when they explain what they intend to do (ask the patient about family and personal clinical history) (lines 95-97), before explaining further what the ultrasound findings mean in terms of Down syndrome risk, and why the information provided in the video shown to the patient before attending the GC consultation is relevant (lines 99-103). The specialist confirms whether the patient agrees with this agenda for the consultation (line 105), to which the patient agrees (line 106).

GC guidelines describe the importance of agenda-setting at the start of a GC consultation. Guidelines indicate that genetic specialists should outline their agenda for the session, and should establish a patient's reasons for attending GC, what their goals are, and should encourage the patient to explain their understanding of what will happen in a GC consultation (Biesecker et al., 2019; Cragun & Zierhut, 2018; Veach et al., 2003). During this initial phase of the consultation in Example 11.1 above, although the patient is asked why she has been referred, she is not asked what her specific concerns or expectations for the GC consultation are. In addition, although the specialist outlines their agenda for the session, they simply confirm whether the patient agrees with this, rather than asking the patient to add onto the agenda for the consultation. Thus, the two agendas are not jointly established for the session, nor is the patient's concerns or agenda truly elucidated.

11.2.3 The patient provides an agenda at the start of a consultation, but the specialist's agenda takes precedence

In Example 11.3 (Part 1 and Part 2) below, I show how the patient provides an agenda at the start of a consultation, but that the specialist's agenda takes precedence over the patient's agenda regarding what type of risk information is spoken about. This is a typical example seen in the consultation data.

Example 11.3

Case overview: Case 11: AMA & soft markers for Down syndrome

This is a 39-year-old Mixed Ancestry patient who presented at the clinic at 13 weeks' gestation. She had been referred for GC as she was of advanced maternal age, and because soft markers for chromosome abnormalities, particularly Down syndrome, were detected including an increased nuchal translucency, and an increased ductus venosus pulsatility index.

Prior to attending the GC consultation, the patient had watched an educational video on Down syndrome (the same video as described in Example 11.1 above) The patient stated at the start of her consultation that she did not want to place her pregnancy at any risk, thus was not interested in doing an amniocentesis. The specialist, rather than tailoring their agenda and information giving to the patient's agenda, provides additional genetic risk information during the consultation and re-offers invasive testing to the patient based on her age and the ultrasound findings. The session was conducted in Afrikaans, the patient's first language. In the transcript below the first line of each numbered line shows the original Afrikaans language used in the consultation (shown in italics) and the second line shows the translated English.

Example 11.3 : Part 1

S= Specialist; P= Patient; Nina* = genetic specialist pseudonym

1	S:	*Goed (.) soos sy gese het (.) my naam is Nina↑**
		Right (.) so like she said (.) my name is Nina↑*
2	P:	((head nod))
3	S:	*Ek is een van die genetiese spesiaaliste wat hier werk*
		I am one of the genetic specialists who works here
4	P:	Ok ((head nod))
5	S:	*Mevrou is Afrikaans ne↑?*
		Ma'am is Afrikaans hey↑?
6	P:	*Ja*
		Yes
7	S:	*Ek het nou net sommer aangeneem op die (.) op die (.) naam (.) maar*
8		*eintlik moet ek $maar check$* *((said in smiley voice))*
		I just assumed now on the (.) on the (.) name (.) but actually I
		should $check$ ((said in smiley voice))
9	P:	((smiles & nods head))
10	**S:**	*Uhm (0.2) so ek gaan maar net 'n bietjie verder met Mevrou gesels*
11		*van die scan wat hulle gedoen het*
		Uhm (0.2) so I'm just going to talk a little bit further with Ma'am
		about the scan that they did
12	**P:**	**((Nods head))**
13	**S:**	*En wat ons nou verder (.) kan doen (0.2) en wil doen*
		And what we can now do (.) further (0.2) and want to do
14	**P:**	**((Head nod))**
15	**S:**	*Maar se↑ vir my n bietjie wat het hulle vir Mevrou gese so ver↑?*
16		*(0.4) oor hoekom jy met my moet gesels↑?*
		But first, tell↑ me a little bit what did they tell Ma'am so far↑?
		(0.4) about why you must talk to me?
17		**(1.0)**
18	**P:**	*Uh (.) die dokter het vir my gese (0.3) uh sy (.) ga:a:an no:o:u*
19		*(0.2) n bietjie meer op die rekenaar gaan kyk oor (.) Down sindroom*
		Uh (.) the doctor told me (0.3) uh she (.) they are no:ow go:oing
		(0.2) to look a bit more on the computer about (.) Down syndrome
20	**S:**	**Mmmhmm** **((long head nod))**
21	**P:**	*En dan gaan hulle vir my 'n video wys en dan gaan iemand met my kom*
22		*gesels (.) uh* *((shifts in seat, frowns and starts to look a bit unsure))*
		And then they are going to show me a video and then someone is going
		to come talk to me (.) uh **((shifts in seat, frowns and starts to look a bit unsure)))**
23	**S:**	*gesels (.) ok (0.4) e:e:n↑ het sy enige sins gese hoekom ons (.)*
24		*hoekom ons die video vir Mevrou wys? (.) hoekom dink ons aan Down*
25		*sindroom?*
		talk (.) ok (0.4) a:and↑ did she say at all why we (.) why we are
		are showing Ma'am the video? (.) why we are thinking about Down
		syndrome?
26	**P:**	*Nee*
		No
27	**S:**	*Nee (.) ok*
		No (.) ok
28	**P:**	*Nee sy het nie vir my verder nie*
		No she did not further
29	**S:**	*Nee ok (0.3) goed (.) dan weet ek maar net waar↑ ek moet begin*
30		*gesels*
		No ok (0.3) right (.) then I just know where↑ I must start
		discussing
31	P:	Mmm (head nod))

32		(1.0)

In the example above, the introductory phase of the consultation involves the specialist introducing themselves and checking the patient's preferred language (lines 1-9). In lines 10-13, the specialist states their agenda for the session which is to provide further information to the patient regarding the findings on the ultrasound scan. The specialist also uses directive language by saying they are going to talk about what '*we can do'* and '*want to do'* (line 13), indicating that they seem to have certain expected outcomes for the GC consultation.

In lines 15-16, the specialist enquires what the patient has been told about GC and why she has been referred. Asking the patient about their reasons for attending GC is *part* of the agenda-setting for a patient (Veach et al., 2003). After a one-second pause in line 17, the patient explains that she has been told by the fetal medicine specialist that the fetal medicine specialist is going to '*look a bit more on the computer about Down syndrome'* (line 19), meaning the ultrasound machine, that the patient is going to watch a video on Down syndrome and that 'someone is going to come talk to me', meaning the genetic specialist (lines 20-21). When the genetic specialist enquires whether the patient has been told why she must watch a video on Down syndrome (lines 23-25), the patient replies that she has not been told the reason why she has been asked to watch the video (line 26). The specialist provides evidence in lines 29-30 that the woman's prior (or lack of prior) knowledge about Down syndrome is used as a basis to develop the specialist's education and risk information agenda for the consult by saying *'ok right then I just know where I must start discussing.*'

The Down syndrome educational video is shown to all high-risk women (e.g., AMA pregnant women, and pregnant women where soft markers have been identified on ultrasound) at one of the prenatal genetic clinics. During my time at the clinic, I was shown the video, and from discussions with the genetic specialists, I was told that the video has been developed by the genetic counselling team at the hospital and is based on information from one of the genetic specialists' post-graduate research and the Western Cape Down Syndrome Association. Patients can choose to watch the short video in either English or Afrikaans and the purpose of the video is to provide medical and genetic information about Down syndrome, the pregnancy risks and ultrasound findings associated with the condition, and what options are available to pregnant women where a risk for Down syndrome has been identified.

The video further details invasive prenatal testing options, the associated risk and the outcome of each option and gives examples from different couples and pregnant women on what they decided to do when faced with similar situations. Outcomes include accepting invasive testing and then either choosing to terminate, or not terminate the pregnancy, if a positive diagnosis is confirmed; or alternatively declining invasive testing and choosing to continue with the pregnancy. In addition to its goal of educating the patient, the video is meant to serve as a decision-making aid to better prepare women and couples for GC where they are provided with more contextualised risk information and an offer of invasive testing by a genetic specialist.

In lines 33-58 (not shown), the specialist explains that the soft markers identified in the patient's unborn baby increase the risk for Down syndrome, but that they do not necessarily mean that the baby has a diagnosis of Down syndrome. The specialist also explains that apart from the soft markers identified, the women's age has placed her at a higher risk to have a baby with Down syndrome.

Example 11.3: Part 2

S= Specialist; P= Patient

59	**S:**	*Se vir my gou (.) wat het Mevrou gedink↑ van die video?*
		Tell me quick (.) what did Ma'am think↑ about the video?
60		**(1.5)**
61	**P:**	*Ek het die video interessant gevind (0.2) maar*
		I found the video interesting (0.2) but
62		**(0.5)**
63	**P:**	*ek het (0.2) tot 'n besluit gekom*
		I have (0.2) come to a decision
64	**S:**	*°is dit°*
		°is that so°
65	**P:**	*ja:a:a*
		Yes:s:s
66	**S:**	**((head nod))**
67	**P:**	*As hierdie baba dan (.) Down sindroom het (0.4) dan aanvaar ek dit*
68		*maar (0.2) maar ek gaan nie die risiko loop om (0.2) toets te laat*
69		*doen en dan die baba te verloor nie*
		If this baby then (.) has Down syndrome (0.4) then I accept it
		but (0.2) but I am not going to run the risk to (0.2) do the test
		and then I lose the baby
70	**S:**	*Ok (0.2) goed*
		Ok (0.2) right
71		(0.5)
72	S:	*So↑ dit is hoekom ons die video vir mense wys (.) want dit gee mense*
73		*baie meer inligting as wat mens gewoonlik het*
		So↑ that is why we show the video to people (.) because it gives
		people a lot more information than what they usually have
74	P:	Mmm ((head nod))
75	S:	*en dit help ook tog om (.) om by daardie besluit uit te kom*
		and it also helps to (.) to get to that decision
76	P:	((head nod))
77	**S:**	*Ek gaan nou vir Mevrou (0.2) NOG↑ n bietjie inligting daar by gee↑*

		I am now going to give↑ Ma'am (0.2) a bit MORE information
78	P:	((head nod))
79	**S:**	*Maar dis goed dat jy al klaar 'n beitjie gedink het wat (0.2) wat*
80		*(.) en hoe*
		But its good that you have already thought a bit what (0.2) what
		(.) and how
81	P:	((head nod))
82	**S:**	*Voor↑ dat ons nou verder gesels oor die scan↑ en (0.2) die besluite*
83		*(0.4) gaan ek net Mevrou 'n paar vrae oor die familie↑*
		Before↑ that we now chat further about the scan↑ and (0.2) the
		decisions (0.4) I am just going to ask Ma'am a few questions about
		the family
84	P:	((head nod))
85	**S:**	*Dis maar nou net om seker te maak dat daar nie iets anders is wat*
86		*ons oor moet weet (.) en ook aan dink nou behalwe die (.) die*
87		*ouderdom en so*
		It's just to make sure that there is nothing else that we need to
		know about (.) and think about besides about the (.) the age and
		that
88	**P:**	**((head nod))**
89	**S:**	**Ok** **((smiles and starts looking down at patient's hospital file that is on their lap))**
90		**(0.5)**
91	**S:**	*So↑ as ek reg is (.) hierdie is Mevrou se (0.3) vierde↑ swangerskap?* *((looking at notes in the patient's hospital file))*
		So↑ if I'm correct (.) this is Ma'am's fourth↑ pregnancy? ((looking at notes in the patient's hospital file))
92	P:	*Ja:a* *(head nod))*
		Yes:s ((nods head))

In the example above, the specialist explores the patient agenda further by asking her what she thought about the video in line 59. After an extended pause in line 60, the patient says they found the video 'interesting' (line 61) but she has decided that after watching the video she does not want invasive testing as she is concerned about the procedural-related miscarriage risk, and will accept a baby with Down syndrome (lines 62-69). The specialist confirms that they have heard the patient's decision in line 70, and further highlights that the video assists individuals to make risk-related decisions (lines 71-75).

Rather than further exploring the patient's motivations behind her decision, or tailoring their agenda to fit in with this new knowledge conveyed by the patient, evidence that the specialist's agenda takes precedence over the patient's agenda is seen in line 77, where the specialist says '*I am now going to give↑Ma'am (0.2) a bit MORE information'* and in lines 82-83 where they indicate that they want to gather additional family information from the patient and to chat further about the findings on the patient's ultrasound. Although the specialist shows acknowledgement and awareness of the patient's decision in lines 79-80, they proceed to state

their agenda, and the remainder of the consultation is driven by the specialist's agenda which includes revision of the information provided in the video, as well as an offer of invasive testing, which the patient declines. The level of risk information detail conveyed in the consultation and offer of invasive testing is not necessary if the patient's stated agenda had been actively considered.

Although tailoring information according to a patient's goals, background and desired amount of detail are important GC outcomes (Cragun & Zierhut, 2018), GC studies from the Netherlands (Pieterse et al., 2006), Australia (Lobb et al., 2002) and South Africa (Wessels, 2014) show that genetic specialists may struggle to adapt their own agenda to a patient's agenda. An Australian study focusing on 158 audio-recorded cancer GC consultations showed that genetic counsellors state their agendas in 86% of the consultations, whilst patients were asked about their agendas in only 69% of the consultations. Findings in this study support these previous literature findings and demonstrate that even when a patient's agenda is known, a genetic specialist may guide the risk discussions in the GC consultations according to their agenda preferences.

11.2.4 A patient does not provide an agenda even when solicited by the specialist

In the example below, Example 11.4, I show how two patients are asked about their expectations for a GC consult, but they respond passively and do not provide their own agenda. The specialist then continues to provide their agenda which the patients accept. This is a typical example seen across the GC consultation data.

Example 11.4

Case Overview: ***Case 2- Chromosome translocation***

This follow-up case (the attending specilist did not see the patients for their initial consultation) involved a Mixed Ancestry family with three daughters. The family had been referred to the genetic clinic because the youngest daughter had several physical and intellectual disabilities. After medical investigations, which included a karyotyping genetic test, the youngest daughter was found to have an unbalanced chromosome translocation, which was the cause for her physical and intellectual disabilities. Although her parents and other siblings were phenotypically normal, genetic testing had subsequently been offered to them to determine the risk of them being carriers of this particular translocation.

The follow up GC consultation was attended by the 62-year-old father, and the middle daughter, who was 21-years-old. The father had already received his results which indicated that he was a balanced carrier for the particular chromosome translocation. Thus there were no health implications for him, however there were risks for his children. The second daughter, who received her results in the consultation, was found to have a normal chromosome profile, hence she was not at any health risk, and there were no implications for her future children. The eldest daughter was unable to attend the consultation on the day due to work commitments. The consultation was conducted in English, the patients' second language.

The genetic specialist who conducted the follow-up consult with the father and middle daughter is not the genetic specialist who had counselled the family at their initial GC consult, thus they were not acquainted with one another.

S= Specialist; D= Daughter; F= Father, C= Child

23	S:	So↑ I'm a genetic specialist*
24	D:	((head nod))
25	**S:**	**And the reason for today's consultation is really for you (.) and your**
26		**sister** **((looking at daughter))**
27		**(0.5)**
28	**S:**	**Remember the last time we took the blood** **(rubbing hand across arm to indicate blood collection))**
29	D:	((head nod))
30	F:	((head nod))
31	**S:**	**So its to get (0.2) to give you your results**
32	D:	((head nod))
33	S:	(0.3) uhm and then (.) through the session we can obviously look at
34		the whole↑ family and we can deal with your younger daughter (.) and ((looks at father))
35		deal with all the issues step-by-step (0.4) **but (.) the main (0.3) uhm**
36		**(.) reason for today is to give you those results** ((looking at daughter))
37	D:	(head nod))
38	**S:**	**Is there any other expectation that you had coming to this clinic?**
39		**(0.2) Or anything else you wanted me to address? (.) Other than just**
40		**giving you your results?**
41		(1.0)
42	D:	(0.5) ((daughter looks across at father in a hesitant manner. Father looking at genetic specialist but provides no verbals or non-verbals. Arms are folded across his chest))

43	**D:**	**No it's $fi:i:ne$** **((smiles and looks at specialist and waves hands in front of her body))**
44	S:	Ok (0.4) ok ((nods head)
45		(0.5)
46	S:	So (.) let me just first (0.4) because I haven't seen you before (.) I
47		actually just want to actually understand what you heard the last time
48		(0.2 and what you remember because sometimes (.) gene:etics (0.) can
49		be quite complicated (0.2) and sometimes people don't alwa:ays
50		understand (0.2) or remember everything from this FULL information
51		session
52	D:	((head nod))

In the example above, in lines 1-22 (not shown) the father is discussing the youngest daughter (who is not in the follow-up consult but who is the initial proband and reason the family have all attended GC) and how she is struggling in her current mainstream school. The discussion about the daughter has been prompted by the specialist's question on how the family are doing. The question is asked whilst the father and his middle daughter are settling into their seats in the consultation room.

In lines 23-31, the specialist re-introduces themselves and indicates their agenda for the consult, which is to provide the middle daughter (and what was meant to also be the eldest daughter) with her genetic test result. The specialist acknowledges the youngest daughter's problems in lines 33-34 but reiterates that the agenda for the consult is to provide genetic test results (lines 35-36). The middle daughter (lines 29, 32 and 37) and father (line 30) both provide head nods in acknowledgement of the agenda.

In line 38-40, the specialist attempts to establish the father and middle daughter's agenda for the consultation by asking what their expectations are and if they had anything further that they want to address during the consult. After a one-second silence, neither the daughter nor the father provides an answer. Evidence that this is a transition relevant place and that conversational norms expect an answer to a question, is provided in line 42 where the daughter provides a hesitant non-verbal gesture by looking across at her father in anticipation that he provides an answer. The father provides no verbal or non-verbal cues, and the daughter then looks back at the specialist and responds on behalf of both herself and her father by saying *'no it's fi:ine'* in line 43. The daughter's turn is accompanied by a smiley voice and she rapidly waves her hands in front of her body in a hurried, almost embarrassed manner.

The specialist responds to the daughter's reply with the acknowledgement token *'ok'*, a pause and repetition of *'ok'* in line 44. An 'ok' combined with a pause(s) is commonly displayed in dispreferred responses, whereas an 'ok' without any hesitation or pausing is seen more frequently in preferred responses (see Schegloff, 2007). The repeated acknowledgement tokens and pause produced by the specialist could indicate difficulty in accepting the patient's response and their lack of agenda. The specialist however does not probe further and continues with the remainder of the consultation using their agenda as a guide.

Although establishing patient agendas may be theoretically important, empirical evidence from both international (Bernhardt et al., 2000) and South African (Morris, 2013; Wessels, 2014) GC studies show that patients may have few, or no, goals for a GC consultation, since they may not know what to expect. South African patients attending GC consultations also typically do not attend on their accord but are rather referred by other healthcare practitioners (Wessels, 2014). Despite the lack of clear expectations, patients may still have concerns about their health, their family's health, or the health of their child. In Example 11.3 above, asking the patients what their agenda is may have come as a surprise, as previous research in South African healthcare has shown that many patients in the public healthcare system lack agency and do not demonstrate assertiveness during conversations with healthcare practitioners (Penn & Watermeyer, 2018). Hence, establishing true patient concerns in South African GC consultations may be challenging for genetic specialists in reality, even though GC guidelines assume that patients come with questions and expectations (Veach et al., 2003).

11.2.5 A patient's agenda becomes apparent during, rather than at the start of a consultation

In the final example for this sub-section, Example 11.5, taken from a paediatric consultation, I provide typical evidence of how a patient's or parent's agenda may only become apparent during, rather than at the start of, a GC consultation. The example further shows how a genetic specialist may struggle to adapt to a patient's or parent's agenda when it is revealed and how they may continue to orient to their own script-based agenda that has predetermined at the start of the consultation.

Example: 11.5

Case overview: ***Case 13- Hypophosphatasia***

This paediatric consultation involves an isiXhosa couple and their two-year-old son who has a diagnosis of hypophosphatasia. Hypophosphatasia is a rare genetic condition characterised by the abnormal development of bones and teeth due to poor mineralisation. There is no genetic testing available in South Africa and the diagnosis is typically made on a clinical basis. The couple attend the genetic clinic for a follow-up consultation for their son to assess his general wellbeing and to review the related genetic risk information. The session is conducted in English, the couple's second language.

During the consultation, the parents' son is very boisterous and vocal. During the information giving section of the consultation where risk information is being conveyed, the mother indicates that she and her husband do not want more children. These are two important factors in this consultation that require consideration. The genetic specialist, although being aware of the noisy environment and the parent's intentions, continues to convey large amounts of detailed pregnancy-related risk information using diagrams and numerical formats. Whilst the genetic specialist is providing the information, the parents appear distracted whilst trying to attend to their noisy toddler.

In the example below I include several lines to provide evidence of the misalignment between the genetic specialist's and patients' agendas and how the genetic specialist fails to pay attention to interactional and contextual gestures that indicate that their information is not appropriate or relevant for the patients at the time.

S= Specialist; M= Mother; F= Father, C= Child

1	S:	So:o what do you remember about (0.1) uhm (0.4) for
2		[the both of you (0.2) if you were to have more]
3		children (0.2) for this to happen again?
4	C:	[tjee tjee (0.4) tthhh:hhh] (sitting on floor babbling))
5		[(1.0)]
6	F:	[((no response, arms folded across chest))]
7	M:	[((frowns and looks at specialist))]
8	S:	Did [anyone ever tell you °that°?]
9	C:	[tjee TJEE TJEE] (sitting on floor babbling loudly))
10		(1.5)
11	F:	Hhh Hhh ((awkward smile)
12	M:	Sorry↑? ((frowning. Looks at father and then back at specialist with confused facial expression.))
13	S:	Di:id anyone↑ ever tell you the cha:ances for (0.3) if you were to
14		have (0.1) another [chi:ild?]
15	F:	[Ja:a] ((smiling and rubbing his cheek in pensive manner))

16	**S:**	**And (0.2) I $don't know if you (0.1) w:aant$?=**
		(smiling and looking at father))
17	**M:**	**=We don't↑↑ want!**
		((leaning back in chair))
18	**S:**	**You don't want?=**
		((looking at mother))
19	**M:**	**[=No]**
		((raising eyebrows and looking back at specialist))
20	**F:**	**[Ja] but they told us that yes. (0.2) They say**
21		**that it is possible to get a child like this.**
22		**(0.5)**
23	S:	°Yes°
		((nodding head and looking at father))
24	M:	So we must (0.2) come to
25		(0.5)
26	M:	the:e:em? (0.2) to (0.2) [tell us (0.2) what we can do?]
27	C:	[eh eh eh (0.2) eh eh eh]
		((sitting on floor and looking at his mother))
28	S:	Ja:a
29	M:	[A:and all that stuff]
30	C:	[eh eh eh (.) eh eh eh]
31	**S:**	**So:o let me (0.2) I know that there (0.2) [So you're saying you**
32	**C:**	**[eh eh eh (.) eh eh eh]**
33	**S:**	**don't↑ want any more children?]**
34	**F:**	**[((smiles and shakes head))]**
35	**M:**	**[((smiles, shakes head and waves hands across chest in 'no go' manner))]**
36	**S:**	**But sometimes .hhh (0.2) somethings things happen (0.2) sometimes**
37		**it's $out of our contro:ol$**
		((smiles))
38	**F:**	**[No no uh uh hhh hhh!]**
		[((shakes head))]
39	**M:**	**[Hh hhh]**
		[((shakes head and looks awkwardly at floor))]
40	**C:**	**[eh eh eh (.) eh eh eh (0.2) eh eh eh]**
41	**S:**	**Hhh hhh**
		((smiles))
42		**(1.0)**
43	**S:**	**Let's just say [(0.3) hypothetically you did want more children (0.1)**
44		**[or for example you fell pregnant even↑] if you weren't planning to]**
45	**C:**	**[EH EH EH (0.1) EH EH EH (0.2) EH EH!]**
		((sitting on floor between parents and babbling loudly))
46		**(1.0)**
		((mother and father provide no verbal or non-verbals. Both staring blankly at specialist. Father has hands folded across chest & mother has hand on chin leaning forward))
47	**S:**	**We know that there is a cha:ance↑ for this to [happen again (0.1) and**
48		**I'll explain to you why]**
49	**C:**	**[na na (.) noh noh]**
		((sitting on floor and looking around room))
50		(0.5)
		((mother and father provide no verbal or non-verbals. Both staring blankly at specialist. Father has hands folded across chest & mother has hand on chin leaning forward))
51	S:	.Hhh so↑ [remember I was telling you about the DNA with the
52		instructions that make our bones strong?]
53	C:	[eh eh (.) ah ah (0.3) oh]
		((looking up at specialist))
54		(1.2)
55	S	And in all↑ of us we actually have two↑ copies of those instructions↑.
56		(0.1) because we got one from our mo:om and one from our da:ad (.) So
57		[we have double copies of them.]
58	M:	((small frown and looks away from specialist))
59	C:	[D:daa:ada!]
60	F:	Mmm?
		((father looks at son, not paying attention to specialist))

61	C:	DA:DDA!
62	F:	((head nod and looks at son who is sitting on the floor))
63	S:	[So for exa:ample this is you over here↑ (0.3) you have two↑ copies (.)] ((drawing diagram of gene alleles with pen and paper. Paper is resting on specialist's lap and angled in a way that the parents can see what is being drawn))
64	C:	[Eee! (0.4) Ooo!]
65	S:	one from your mom and one from your dad (0.2) so this copy must tell ((looks at mother))
66		your body to add↑ the minerals to the bones to make them strong (0.3)
67		[a:and you have two copies okay↑?] ((still looking at mother))
68	C:	[Eee Ssss (0.2) Da. DADDA!] ((leans over and hits father on the leg to get his attention)
69	F:	((looks down at child is not focusing on genetic specialist))
70	M:	(1.0) ((mother looking at the drawings done by the specialist))
71	C:	Dadda? (0.2) Dadda! ((looking at father))
72	S:	And the:en Siya* you also have [two (.) one from your mom and one ((turns attention to father and continues to draw diagrams))
73	C:	[thhhh (0.2) Dad (0.3) Dadda]
74	S:	from your dad]
75		(2.0)
76	M:	((small head nod when specialist looks at her))
77	S:	A:and when you have children↑ (0.3) our children must now get one from
78		mom and one from dad (0.4) So:o you have two (.) so you give one (.) ((draws alleles and then looks up at mother))
79		one or the other to your child.
80	M:	((mother looks bored. No verbal or facial expression))
81	S:	And [you also have two] so you can give one ((turns attention to father))
82	C:	[Eeee (.) oh oh] ((sitting on floor and rocking back and forth))
83	S:	or the other
84	F:	((also not showing interest in information nor looking at diagram))
85	S:	And it's completely↑ ra:andom how our bodies give (.) that to our
86		children (0.2) is that we know (.) that in our parents↑ (.) in both of
87		the parents (.) one↑ of their copies is not working↑
88	C:	((shuffling across floor and making grunting noises))
89		(2.0) ((specialist drawing mutated allele on the diagram))
90	S:	So for example for yo:ou (0.2) one is [not working (.) a:and for yo:ou] ((looks at mother)) ((looks at father))
91	C:	[Eeee (0.2) Ma (.) Ma (0.2) Ma]
92	S:	one is not working
93	F:	((small head nod))
94	S:	But both of yo:ou (.) we can see you don't have any bone problems (.) ((shakes head))
95		[you (.) you don't have the condition↑]
96	C:	[Mamma! (0.2) ma (.) uhh uhhh (0.2) Mamma!]
97	M:	((small head nod and long eye blink))
98	S:	[You don't have hypophosphatasia]
99	C:	[Neh neh (0.2) neh neh!] ((shuffling across floor))
100	M:	((shakes head and looking at specialist))
101	S:	And that's because you have a back-up copy (.) so although one is not
102		working (.) you have one that can do that job (0.2) Does tha:at make
103		sense? ((looks at mother))
104	M:	°°Yes°° ((slight head nod))
105	F:	((no verbal or non-verbal display))

106	S:	But (.) so for yo:ou↑ both it doesn't have any concer.- (.) uh problems
107		for you (.) medical problems (.) because you have one that is working↑
108		(.) but when you have children↑ (.) there is a cha:ance that
109	C:	((child has now shuffled across floor and sitting at specialist's feet looking at their shoes))
110		(1.5)
111	S:	So like I said↑ (.) when you have a chi:ild (.) you must give one of ((looks at mother))
112		yours (.) and you must give one of yours (.) so your chi:ild↑ has ((looks at father))
113		two (.) °one from mom and one from dad°
114	C:	((making grunting noises))
115	M:	((blinks eyes and gives small head nod when specialist looks at her))
116		(2.0) ((specialist drawing out inheritance options with allele combinations))
117	S:	So you can give thi:s↑ one and you can give thi:is one (0.2) and ((pointing at drawn alleles))
118		that child will be fine↑ (.) no problems (0.2) you can give this
119		o:one and you can give this o:one
120		(1.2) ((specialist drawing out allele combination))
121	S:	and this child will also↑ be fi:ine↑ (.) they will be like one of
122		you where we (0.2) call them (0.3) CARRIERS (.) [they carry↑ one]
123	M:	[((frowns))] ((looking at specialist))
124	S:	that is not working
125	C:	((playing with draw of the desk that specialist and mother are sitting next to))
126	S:	But you don't have↑ the condition because you have one that is working
127	F:	((small head nod))
128		(1.0)
129	S:	[So you can pass on this one and you can pass on this one] ((continuing to draw allele inheritance options and focusing on diagram))
130	C:	[Oya?] (looks at mother))
131	M:	((looks at child, at door of the room and then back at child. Shrugs shoulders))
132		(2.0)
133	C:	BYE? Ooooh! ((places hands over mouth and looks around room))
134	S:	And again↑ you can see
135		(2.0)
136	S:	This person is also↑ a carrier↑
137		(0.5)
138	S:	But you can pass on this one (.) and tha:at↑ child will have two
139		that's NOT↑ working
140		(0.5)
141	M:	((head nod and looks at specialist))
142	F:	(sitting with arms folded, face in neutral expression))
143	C:	((making grunting noises and again playing with drawer of desk))
144	S:	The body doesn't have any↑ instructions to add that min- (0.2) those
145		minerals to our bones (.) and that's why you have weak bones (.) like
146		your son (.) ((looks at father and mother))
147	M:	((staring vacantly across room, not paying attention to specialist))
148	S:	so this↑ is °your son° ((pointing to affected allele combination on drawn diagram))
149	M:	Mmm ((stops staring, blinks and looks at specialist))
150	F:	((father sitting at a distance with arms folded. Neutral facial expression))
151	S:	But so (.) every↑ ti:ime you have a chi:ild↑ you have a one↑ out of
152		four (.) cha:ance (0.2) that your child (.) can have this condition

153		°okay° (.) you can have a two↑ out of four (.) or a 50 percent↑ cha:ance
154		that they will be just↑ like you↑ (.) carriers↑ (0.2) but they don't
155		have the condition at all
156	M:	((slight head nod, appears bored))
157	S:	But then there is also a one↑ out of four (.) 25 percent cha:ance that
158		they have (0.2) [they don't↑ have the condition and] they don't have it
159	C:	[Daaaii eh meeeh (0.2) EH EH] ((stops playing with draw and turns attention to specialist))
160		(1.0)
161	**S:**	**[Does that make a little bit of sense?]**
162	**C:**	**[EH (0.2) Eeehh]**
163	**F:**	**[((nods head))]**
164	**M:**	**[Yes]**
165		**(1.0)**
166	**S:**	**So how do you fe:el about this:s?**
167		**[(3.0)]**
168	**C:**	**[Ehh (0.3) Daaaa (0.2) DADDA! (.) DA:AD:DA:A:A]** **((looking at specialist but calling father))**
169	**M:**	**((shrugs shoulders))**
170	**F:**	**((no verbal or non-verbal response))**
171		**(2.0)**
172	**S:**	**°You don't know?°**
173	**M:**	**No:o:o it's alright (.) °ja (.) no°** **((frowns and shrugs shoulders))**
174		**(1.5)**
175	**S:**	**Has it been (0.2) quite hard for you?** **((looking at mother))**
176	**C:**	**Da:a (0.2) DADDA!** **((trying to get father's attention))**
177		**(1.0)**
178	**M:**	**No↑↑ (.) [there is nothing that's hard now (.) it was hard (.) before]**
179	**C:**	**[Daaa! (0.3) DADDA! (.) DA! (.) DAD:DA:A?]** **((looking at father))**
180	**F:**	**[((not making eye contact with son, looking at mother))]**
181	**M:**	**(0.2) but now** **((shakes head))**
182	**S:**	**°Okay°**
183	**C:**	**Da! (0.2) DADDA!**
184	**F:**	**Yea** **((turns attention to son and makes eye contact with him))**
185		**(1.0)**
186	**S:**	**And I suppose (0.2) for both↑ of you sa:aying you don't↑ want any**
187		**more children**
188	**F:**	**((shakes head))**
189	**M:**	**((frowns and looks at specialist))**
190	**C:**	**Dadda!** **((trying to get father's attention again))**
191	**S:**	**This maybe isn't a RISK for you (.) at all** **((looks at mother))**
192	**M:**	**[((nods head))]**
193	**C:**	**[Dadda:a! (.) Dad:da!]** **(trying to get father's attention))**
194	**S:**	**[Because (0.2) there's no cha:ance to pass it on** **((turns to look at father))**
195	**C:**	**[DA! (.) DADDA! (.) DADDA↑!)** **((child starting to get desperate and his voice trembles as if going to start crying))**
196	**F:**	**Ja?** **((looks at child and makes eye contact with him))**
197		**(2.0)**
198	**F:**	**((unfolds arms and reaches arms out to his son and smiles gently at him))**
199	**C:**	**((seems to calm down and looks at father))**
200		**(0.5)**

201	**M:**	**((looking at her son))**
202	S:	Uh (0.2) but why↑ we say this (.) is a (.) genetic↑ condition uh (0.2)
203		is ((pointing with pen at the drawn diagram))
204		(0.5)
205	S:	You can se:e:e (0.2) you got this (.) both↑ of you have [it (0.4) and]
206		remember everyone↑ in the population (0.3) is a carrier↑ for
207		something
208	F:	[((folds arms across chest again and reverts to neutral facial expression))]
209	M:	((looking at specialist and fiddling with her shoe))
210		(0.5)
211	S:	So we all↑ carry between 10 (.) and 20 different things in our bodies
212		(.) that can cause diseases
213	M:	((head nod))
214	S:	But what happens [when two carriers come together (.) and they have] ((making hands into fists and bringing them together to emphasise point))
215	C:	[aaaahh aaaa (0.3) waaa waaa]
216	S:	the [same↑ one that can cause a disease (0.4) then it can result in]
217	C:	[aaaa (0.2) wa wa (.) aaa (0.2) wa]
218	S:	this situation ((pointing at diagram))
219	M:	((small head nod))

In lines 1-10, the specialist tries to confirm that the parents are aware that their son's condition can occur again in a future pregnancy. The specialist's question produces no verbal response from the father (line 6) and the mother seems confused by the statement, which she demonstrates via a frown (line 7). When the specialist probes further to establish whether the parents are aware of this information the mother demonstrates misunderstanding by saying 'Sorry?' in line 12. This is done in a question format, which requires a repair from the specialist. The specialist again repeats the question in lines 13-14, to which the father acknowledges that he has heard that there is a risk for future pregnancies in line 15.

A critical part of the interaction occurs in lines 16-17 when the specialist enquires whether the parents would like to have more children. The specialist has hardly finished asking the question when the mother swiftly interjects that they do not want more children. Her quick response is demonstrated through latching between lines 16 and 17 (indicated with = symbols) and again by further latching and a swift response in line 19 when the specialist repeats the question in line 18. This emphatic response from the mother demonstrates her lack of interest in having future children. The father, although not verbalising his feelings about future children, provides further knowledge that they are aware of the risk of having further affected children in lines 20-22. In lines 31 and 33, the specialist again confirms with the parents whether they don't want further children. The mother and father both provide non-verbal confirmation responses in lines 34 and 35. During this time the child is sitting on the floor and babbling loudly (line 32).

Despite this re-confirmation, the specialist demonstrates their own agenda in lines 36, 37, 43, 44, 47 and 48 by verbalising that although the parents do not want more children, sometimes the future may be *'out of our control'* (said in a smiley voice). The specialist further indicates their agenda by stating *'let's just say hypothetically you did want more children'* and '*there is a chance for this to happen again and I'll explain to you why.'* During the specialist's confirmation of their own agenda, both parents provide awkward laughter and head shaking in lines 34, 35 and 39. The father also provides a double negative response *'no no'* in line 38.

When the specialist makes it clear that their agenda is to provide further information about pregnancy-related risks and future children, the parents give no verbal response to this offer but rather a one-second pause ensues (line 46). During these lines of talk, the specialist and parents have to compete with the child's loud babbling when communicating with each other (lines 32, 40, 45, 49). Rather than the specialist prioritising the parents' stated intention not to have more children and just briefly mentioning to them that there is a risk for them to have another child with hypophosphatasia and they should return for further GC should they change their minds in future, the specialist displays their intention to cover risk information related to pregnancy and future children. The specialist thus prioritises their own agenda above the parents' clearly stated agenda. What follows is that the parents receive risk information that is not relevant or useful right now, since the specialist has deemed it important.

The specialist's desire to cover additional risk information causes a lengthy expansion of sequences from lines 51-160 in which the specialist uses a hand-drawn diagram to explain complex molecular and autosomal recessive inheritance risk information. Despite attempts from the specialist to engage with both parents (by making eye contact with both of them and encouraging them to look at the drawn diagrams), the parents seem disengaged and uninterested in the risk information. Evidence for this includes the mother's bored expression, frowning (which indicates a negative emotion and may reveal a person's inner psychological state such as disagreement or confusion (Kaukomaa et al., 2014), and vacant staring across the room. Furthermore, the father seems to have an uneasy non-verbal pose with his arms folded across his chest and leans back in his chair and sits some distance away from the specialist, which would make it difficult for him to engage or see the diagrams being drawn by the specialist. When an individual has their arms folded tightly across their chest it could indicate unease and a defensive, rather than an open, stance (Davis et al., 2000).

During lines 51-160 there are also several pauses at transition relevant places where the parents could give an indication of understanding or interest in the information. However, at these transition relevant places, the mother and father either provide no or minimal cues. These minimal responses from the parents are not expanded on, nor does the specialist stop to check their understanding. The child also continues to be disruptive and makes loud babbling sounds and grunts, shuffles on the floor, plays with the drawer of the desk, and tries to get his parents attention by calling *'Dadda'* or *'Mamma*' several times. The specialist seems to ignore these communicative challenges and continues to provide information and talks over the child's babbling. The parents also attempt to ignore the child's verbal and non-verbal behaviour but eventually concede by acknowledging the child (lines 60, 62, 69 and 131), which further takes their attention away from the specialist.

When the specialist finally checks the parents' understanding in line 161, they do so cautiously and in a hedged manner by saying: *'Does that make a little bit of sense?'*. The specialist's word choice *'little bit'* could show interactional uncertainty from the specialist on the usefulness of the information conveyed during lines 51-161. Although the mother and father both provide a head nod (father) and a verbal *'yes'* (mother) in lines 163-164, microanalysis of lines 51-160 shows that there is misalignment in agendas and several moments of communication breakdown.

Further evidence for trouble is seen in lines 166-182. In line 166, the specialist enquires how the parents *'feel'* about the risk information conveyed to them. This is followed by a lengthy three-second pause in line 167, a shoulder shrug from the mother in line 169 and a lack of verbal or non-verbal response from the father in line 170. After a further two-second pause in line 171, the specialist's uncertainty about the usefulness of the information is confirmed when they prompt a response from the parents by asking: '*You don't know?'* in line 172. The specialist asks the question in a quiet, almost defeated tone. The child is shouting: *'Dadda! Dadda!'* throughout this section of the conversation. A lack of response from the parents and the lengthy pauses indicates trouble in an interaction, especially when this position requires a response from an individual (Jefferson, 1989). The parents lack of engagement with the specialist about the risk information seems to indicate that this is not information that they wanted to hear or are interested in.

In response to the specialist's questioning in line 172, the mother responds by saying: '*No:ooo it's alright (.) ja (.) no'*, accompanied by a frown and shoulder shrug in line 173. The mother's non-verbal cues are misaligned with her verbal response. When the father provides no further response (rather paying attention to his son), and the mother goes on to talk about how they feel about their son's diagnosis, rather than the inheritance risk just conveyed (lines 178-185), the specialist realises that the lengthy explanation of future pregnancy risks may not have been useful to the parents. The parents may perhaps have benefited more if the genetic specialist had followed a psychological counselling agenda (Kessler, 1997b) by discussing their reasons for not wanting children, rather than the specialist's risk information and educational focused agenda. Evidence for the specialist's realisation that the educational section and additional risk information has not been useful to the parents is demonstrated in lines 186-194 when the specialist says: *'and I suppose for both of you saying you don't want any more children', 'this maybe isn't a risk for you at all'*. The use of '*suppose'* and *'maybe'* indicates the specialist's uncertainty whether this information has been helpful to the parents, and in doing so may realise that they have not met the parents' needs for the session. Sarangi and Clarke (2002a) have previously shown how genetic counsellors manage uncertainty by using discourse strategies such as hedging or referrals to experts who have more knowledge of a particular topic (which in Example 11.4 are the parents since they already knew and indicated that this risk information was not relevant to their context or future).

In the framework of relational or psychosocial focused counselling, establishing a mutual agenda should involve negotiation and flexibility between a patient and a genetic specialist (Biesecker et al., 2019). However, the findings of my study, together with previous research (Biesecker et al., 2019; Butow & Lobb, 2004; Joseph et al., 2017; Kessler & Antley, 1981; Wessels, 2014) indicate that genetic specialists may drive a consultation through their own agenda, focusing on content-heavy and educational based risk counselling. Despite the importance of tailoring information and avoiding a step-by-step recipe approach provided in GC practice guidelines (Veach et al., 2003), a study by Joseph et al. (2017) focusing on 170 audio-recorded GC consultations and 49 patient interviews, found that the information provided to patients, and the information patients desired, was mismatched. Despite patients' differing needs and contexts, genetic specialists struggle to adapt their agenda and tailor information to meet the needs or interests of an individual patient. The specialist in this consultation not only misread the 'atmosphere' (Langewitz, 2007) of the consultation but failed to tailor their information giving to suit the needs of the parents. Being attuned to the parents'

agenda and being more flexible in their counselling approach may have assisted the genetic specialist in avoiding the breakdown in communication seen in this consultation.

This example also reveals the power of interactional research. In the interview data, despite several specialists indicating that too much information is not helpful and that tailoring information is important, the example provides an opportunity to compare what is *'said'* in theory (Examples 11.6, 11.7, 11.8) versus what is *'done'* in practice (Example 11.5) which appear to be at odds.

Example 11.6
S6: "And tailoring it for each patient, you know not every patient is going to be the same. Their education, their background, their previous understanding, whether they were on Google five minutes before and probably know all the terms you were just talking about. So obviously getting a feel for what the patient understands. That's the first step. Because before you run off in one direction and lose them or leave them behind."

Example 11.7
S11: "...because if you give, give some um patients too much information, I feel, and I think initially when I started, I almost felt like I needed a genetic tut for everybody! So that they understand OK fine you've got an autosomal dominant condition, but you need to understand it in the context of genetics. Or this is a multifactorial, but you need to understand in the context of, there are other genetic, and then, in the end, I think it might confuse quite a lot of people. To get SO much information but then they have to now suddenly know that they have to hone in on that very important part that you now stress. But now they still trying to grapple with all this other information."

Example 11.8
S11: "So, I certainly, in the beginning, gave too much information. And I could see that by giving less, it seemed that people respond better to that. Or they or they retain it more. Maybe. Uhm… so that I think at least think for many patients. Sometimes maybe taking things in steps, you know doing a specific thing now, especially when there is loads of information. We can always go with X information and sometimes ask them: "What's important for you to know now?" And then focus on that now."

The quotes above, as well as the evidence in the consultation in Example 11.4 where the specialist realises their mistake of providing irrelevant risk information, indicates that genetic specialists are aware of the dangers of providing too much information to patients. This danger includes confusing and overwhelming patients, as well as providing information that is not relevant 'right now'.

In the quotes above the specialists also allude that they made the mistake of providing too much information early in their careers e.g. *'I think initially when I started, I almost felt like I needed a genetic tut for everybody'* and *'I certainly, in the beginning, gave too much information'.* The specialist in Example 11.4 was a newly qualified genetic specialist with only a few years of experience, whereas the specialists in Examples 11.6 and 11.7 were more senior genetic specialists. The differing responses may indicate that practical experience and intuitive

expertise (Gobet & Chassy, 2008) may increase genetic specialists' confidence and comfort to be more flexible in their approach and to pay greater attention to individual patient needs, rather than relying on script-like approaches.

11.3 Risk perceptions

Personal risk perception appears to be a powerful influencer of risk communication in GC consultations. In the following section, I describe local genetic specialists' accounts of risk perceptions, and how this creates challenges in risk communication. Differing risk perceptions between a patient and a gentic specialist can impact how decisions are made about a particular risk and can create challenges for genetic specialsits to remain neutral when conveying risk information. Furthermore, genetic specialists report that local patients in the public healthcare system may also respond towards risk in fatalistic manners, suggesting that their decision-making may also be more passive, in addition to their noted communication styles.

11.3.1 Differing risk perceptions

Risk can be understood from either objective or subjective viewpoints (Anscombe & Aumann, 1963; Bradbury, 1989; Holton, 2004). Genetic risk can thus hold a different meaning for different people, and people may respond towards risks in emotive ways. Individual patient risk perceptions are thought to skew accurate interpretations of risk according to GC guidelines, such as those written by Veach et al. (2003). Part of GC's goals is thus to enhance comprehension of complex genetic risk information and to ensure that patients have an accurate understanding of their risk. Despite two systematic reviews indicating that GC may improve accuracies in patients' risk perceptions (Butow et al., 2003; Smerecnik et al., 2009), quotes from this study suggest that GC may not necessarily alter patient's perceptions of risk. In one quote, Example 11.9, a genetic specialist indicates the many ways different individuals may view the same risk. In Example 11.10 another genetic specialist highlights that despite attempts to alter a patient's risk perceptions through information giving in GC, some patients may view risk in a dichotomised or binary manner, and thus will base their genetic testing decision on the mere presence of risk regardless of how big or small the risk actually is.

Example 11.9

S9: "I think each family is different and what they do with that risk or information is different, so when you give actually the exact same, uh say 1 in 100 AMA risk to ten different people or whatever, there will still be ten different counselling sessions."

Example 11.10

S12: "I don't know, some people I just find… sometimes I don't present a risk number because that person just doesn't take any meaning from the risk. They just say: "There is a risk. And if there is a risk, then I must do something about it.""

The quotes above indicate that patients react towards risk in personalized and emotive ways. Previous research highlights that patients often make emotionally driven decisions that focus on the presence or absence of risk, rather than focusing on the specific details of the risk such as its scale or severity (Lippman-Hand & Fraser, 1979; Lloyd et al., 2001; Paling, 2003; Ropeik & Gray, 2002). A South African GC study by Wessels and Koole (2019), focusing on AMA consultations, provides further support that local patients may respond to genetic risk in dichotomised ways.

11.3.2 Passive patient responses to risk & fatalism

In the interviews, genetic specialists report that local patients in the public healthcare system may respond towards risk in fatalistic manners. Fatalism, or passive acceptance, is the belief that all events are predetermined and that individuals are powerless to change them (Merriam-Webster, 2021). Understanding fatalistic views in relation to risk perception is important as it impacts how individuals may respond towards risk, especially risk avoidance (Ngueutsa & Kouabenan, 2017). Rather than attempting to actively manage a given risk through the various options that may be offered to a patient in a GC consult, genetic specialists in this study reported that some local patients may choose to do nothing about a risk and leave the outcome to fate. This view may be different from genetic specialists' or healthcare practitioners' views who may believe that patients may be interested in genetic testing (e.g. an amniocentesis or for hereditary cancer) to confirm the presence or absence of a potential genetic condition so that management options can be put in place to deal with or prepare for the outcome.

Example 11.11, Example 11.12 and Example 11.13 provide evidence for these reported passive and fatalistic patient risk responses. Both latter examples indicate that part of local patients' reported fatalistic views is related to their religious beliefs. The outcome of a risk-based situation is left to divine intervention, or God's will, rather than something that the patient believes they have personal control over.

Example 11.11
S10: "I'm much more used to, almost not underplaying the risk, but not to go into detail about it because our patients usually don't ask very much."

Example 11.2
S7: "…we see patients who I think are more, I don't know if their fatalistic, or more accepting of whatever happens in their pregnancy….so if their child is born, or if their child has multiple anomalies uhm… ja that that's OK and ja… it'll go away, or it won't go away and that what God wants."

Example 11.13
S8: "It's a different population group [public healthcare-based patients], it's much more accepting, generally much more fatalistic. Religion, plays a much more major role. Not that people out there are not religious, but the values are different, and what they tolerate and what they accept."

The quotes above suggests that in addition to patients' noted passive communication styles seen in the interactional data, they may also be passive in their decision-making towards a given risk. Fatalistic patient views have been previously described in local (Kromberg & Jenkins, 1997; Todd et al., 2010; Wessels, 2014) and international (Abad et al., 2014) GC studies. A South African retrospective study by Todd et al. (2010), focusing on 171 women counselled for severe fetal abnormalties between 2002-2006 at genetic clinics in Johannesburg, found that many women, particularly Black women, were less likely to accept an offer of termination of pregnancy, and rather left the outcome of their pregnancy to fate. These collective findings have implications for autonomous decision-making and what patients do with the health or risk information that they receive. Attention to patients' concerns, backgrounds and beliefs are thus important in GC as patients' responses to risk information and risk decision-making are contextually and socio-culturally framed.

Fatalistic patient views are also noted in the interactional data of the study when patients are offered genetic testing. Patients cite leaving the outcome of an uncertain situation up to 'God' as reasons for declining the offer of testing. Below, I show an example of this type of patient view in Example 11.14 (Part 1 and Part 2) in which a patient at risk for inheriting a *BRCA2* cancer causing mutation believes that she is not at risk because she has faith in God. The example further shows how a patient may feel obliged to take up the offer of testing due to pressure from family members or healthcare practitioners, even if the patient has a passive stance on their health outcomes. Genetic specialists may further communicate in directive ways regarding the uptake of genetic testing.

Example 11.14

Case overview: *Case 7- HBOC*

This case involves a 39-year-old woman of Mixed Ancestry who had been referred for GC as her younger sister had recently been diagnosed with breast cancer. Further investigation on her sister's DNA confirmed a *BRCA2* mutation, which is associated with an increased risk for HBOC. The confirmation of a *BRCA2* mutation in the patient's sister, together with an extensive family history of cancer, places the patient at an increased risk to carry the same genetic change in her own DNA, and by extension a risk to develop cancer. The patient was asymptomatic as the time of attending GC and was offered genetic testing based on the presence of a *BRCA2* mutation in a close family member. The session was conducted in English, the patient's second language.

As mentioned in Example 11.1. the patient in this consultation is already aware of her cancer and genetic inheritance risks. She is also aware that genetic testing is an option for her as she had an overview discussion with the referring healthcare practitioner regarding her risks because of her sister's diagnosis of HBOC. The patient is referred to GC to go over the risk and genetic testing information again and to decide on genetic testing.

Example 11.14: Part 1

S = specialist, P = Patient

13	**P:**	**And honestly I didn't want to come**
14	S:	Mm
15	**P:**	**Because I said I don't like to worry (.) I'm free**
16	S:	Mm
17	**P:**	**And she said how do you know and I said: "Because I'm a believer"**
18		**(0.2) I'm cancer free↑ (.) there's no cells↑ (.) there's nothing**
19		**wrong** **((sitting with arms folded across chest and closed eyes))**
20		(1.0)
21	**P:**	**And she said: "How do you know?" and I said: "I believe in God"**
22	S:	Mmm
23	**P:**	**I believe that I don't have it (.) so I'm gonna waste the**
24		**people's time↑ (0.3) I'm gonna waste the Government's resources**
25		**(0.2) wasting money up (.) It's unnecessary**
26	S:	Mm °okay°
27	P:	And then she laughed at me and said: "You must go still!" hhh hhh
28		hhh hhh
29	S:	Hhh hhh
30		(0.5)
31	**S:**	**I understand- (.) you know it's so important to have that FAITH**
32		**that you have (0.2) and that belief↑ (0.3) it's so:o↑ important**
33		**(0.4) But scientifically↑↑ I can't↑ say you're in the clear**
34		[and that's] the reason why you have to waste our time! (0.2) ((smiling))
35	**P:**	**[I understand]**
36	S:	it's not a waste ok! ((places her hand on patient's leg in an empathetic manner))
37	P:	Hhhh hhhh hhh
38	**S:**	**Hhh hhh (0.4) and I know you feel that you are a believer (.)**
39		**but we can't↑ promise you (.) that you don't have this genetic**

40		**change** **((pointing at diagram))**
41	**P:**	**°I believe°**
42		**(1.5)**
43	**S:**	**I ALSO↑ want to (.) believe with you**
44		(0.5)
45	S:	okay? ((smiles))
46	P:	((smiling))

In lines 13, 15, 17-19, 21, 23-25 the patient relays her hesitance in attending a GC consultation. Although the patient's sister (referred to as 'her' in the transcript above) has insisted that the patient attend GC to better understand her risks of inheriting the same genetic mutation that her sister has and to do genetic testing, the patient feels that it is a waste of time and government resources because she *'believes in God'* (line 21). Due to her religious beliefs, the patient is convinced that she is not at risk for developing cancer and that the information relayed to her by her sister, the referring healthcare practitioner, and now also the genetic specialist, is thus irrelevant.

The patient's perception of risk is delicately managed by the specialist who, although acknowledging the patient's faith, counters this perception in lines 31-33, 38-40. Here the specialist carefully hedges their response, by acknowledging the patients belief and the importance of having faith, however scientifically the patient remains at risk. When the patient says she understands in line 35, however again states that *'I believe'* in line 41, the specialist further attempts to save-face in line 43 by stating *'I also want to believe with you'*. Reasons for face-saving strategy is that the specialist does not want to down-play the patient's value of religion, however, wants to fulfil their professional duty of informing the patient of her genetic risk. In acknowledging the importance of religious (subjective) and scientific (objective) understandings of risk, the specialist tries to find a balance in their communication approach.

In Example 11.14: Part 2 below, I show how despite the patient indicating she is not concerned about her risk for getting cancer, the specialist highlights the benefit of genetic testing to manage the patient's future health. Although the genetic specialist is largely neutral throughout the consultation, the example demonstrates that a genetic specialist may oscillate between different levels of neutrality and directiveness in their communication with a patient. Part of this oscillation may have to do with addressing patients' concerns, especially when patients demonstrate uncertainty.

Example 11.14: Part 2

S= Specialist; P= Patient

460	**S:**	**But how do you↑ feel about these risks?**
461		(0.5)
462	**S:**	**I mean (.) let me first ask (.) how do you (.) how do you feel**
463		**(.) I know you feel that you don't↑ have this genetic change**
464		**hey?**
465	P:	Mmhm
466	S:	Okay
467	**P:**	**Yes (0.4) °I don't have it°** **((frowns with hand on chin))**
468	**S:**	**You don't think you have it?**
469	**P:**	**((shakes head and frowns))**
470		**(0.5)**
471	**S:**	**If you (0.2) If↑ I test you**
472	**P:**	**Mm**
473	**S:**	**and you DO have it (0.4) °How're you're gonna feel?°**
474	**P:**	**I'll be fi:ine**
475		(0.5)
476	S:	°You'll be fine?°
477	P:	I'm gonna be fine↑ really
478	**S:**	**You're not gonna feel bad?**
479	**P:**	**I'm not gonna feel bad (.) I will (.) I'm gonna be fine↑** **((shakes head))**
480		(0.5)
481	S:	Do you think you'll feel shocked?
482	P:	No (0.4) I'm gonna be fine I promise↑ you↑ (0.2) I've not even ((shakes head))
483		(.) yesterday there (.) there will be my [___] yes (.) But I'm
484		**gonna be (.) I will be able to handle it**
485	S:	Mm
486		(2.0)
487	P:	You know what (0.4) uhm
488		(1.0)
489	P:	uhm (0.3) I went for >a whole lots of stuff already all these
490		years ((making counting gestures using her fingers))
491		(0.5)
492	P:	And I think this will (.) be (0.4) ((shrugs shoulders))
493	S:	It's nothing
494	**P:**	**It's nothing (.) serious** **((shakes head))**
495	**S:**	**°ok° (0.4) So=**
496	**P:**	**=AND**
497		**(1.5)**
498	**S:**	**there is something that we can do↑**
499	**P:**	**There is something! Yes!** **((nods head))**
500	**S:**	**It's [not like we're giving you a death sentence]**
501	**P:**	**[there there (0.2) you're not telling me] (0.4) you have this**
502		**change (.) and you're gonna die↑! (0.3) you're telling me (0.2)**
503		**you're telling me there's options↑ (.) there's stuff we can do↑ for**
504		**you**
505	**S:**	**Ja** **((head nod))**
506	P:	You're gonna LIVE
507	S:	((small head nod))
508	**P:**	**As long as you tell me this (.) you're gonna LIVE**
509		(0.5)
510	S:	((head nod))

511	**P:**	**You're still gonna see (.) your grandchildren↑ (.) you're gonna**
512		**see your son matriculate**
513		(0.5)
514	P:	then I'm happy ((slaps hands on top of thighs))
515		(0.5)
516	**S:**	**See that's the point of these tests (0.2) or well (.) that we**
517		**offer↑ these tests (0.3) we call it a predictive↑ test okay**
518		**(0.2) because we're not saying that you have cancer (.) but**
519		**there's a possibil[ity (.) a hi]gh↑ chance that you COULD get**
520	P:	[possibility]
521	S:	ca[ncer]
522	P:	[yes]
523	**S:**	**but in order to make the chance as low↑ as possible (.) we do**
524		**one of those three things (0.2) okay (0.3) so we monitor you to**
525		**prevent↑ a cancer (0.2) or to (.) >catch it early< okay**
526	P:	I'm fine ((head nod and smiles))
527	**S:**	**Versus ok (.) not doing anything↑**
528	P:	((smiles))
529	**S:**	**And then you get a cancer and then it's too late to do anything**
530	**P:**	**Ok (0.3) that's what my sister said so that's why I'm here ((smiling))**
531	S:	Alright

In the consultation above, the specialist initiates patient reflection in line 460 by asking the patient how she feels about the risk information provided (the preceding lines involve a discussion on various types of cancers the patient could potentially be at risk for). After a half second pause in line 461, the specialist provides a self-repair in lines 462-464 by asking again how the patient feels and acknowledges that the patient has previously indicated earlier in their conversation that she does not believe that she is a carrier of a *BRCA2* mutation (as seen in Example 11.14: Part 1 she cites having faith and trust in God as reasons for not harboring a genetic fault in her DNA and that she won't develop cancer). The patient confirms the specialist's assessment in line 467 by saying: *"yes I don't have it"*.

The specialist again prompts further patient reflection by enquiring how the patient would feel if she did have genetic testing and it confirmed that she carries the mutation (lines 473), to which the patient responds that they would feel fine (line 474). The patient then provides a justification for her reasons for feeling fine about having a *BRCA2* mutation in lines 477-494 stating that she will be able to handle it (having a mutation and by extension the potential of developing cancer) (line 484) as she has gone through several hardships over the years. The patient indicates that being told she has a *BRCA2* mutation will be *'nothing serious'* (line 494). The patient's responses and reflection about being at risk for a *BRCA2* mutation and potentially cancer, and not being concerned about it because of her faith, orientates towards a fatalistic view on her risk and future health and that she has no control over it. Thus, although she has

not directly declined the offer of genetic testing, her lack of concern about the risk and faith that she doesn't have the genetic mutation could imply that she is not interested in genetic testing. Her indication that she is wasting everyone's time and the government's resources (as indicated earlier in the consultation shown in Example 11.14: Part 1) is further evidence that the patient may not want to be at the GC consult, nor have genetic testing.

In Example 11.14: Part 2, in lines 495-529, the specialist challenges the patient's perceived lack of autonomy and passive stance by taking on a more direct communicative approach to indicate that the patient's risk can be managed rather than leaving it up to fate or God. This is demonstrated by the *'so'* (in line 495), and *'there is something that we can do'* (in line 498) (i.e., a genetic test can be done to confirm whether the patient is a carrier of the genetic mutation or not, and subsequent risk management can be done according to the result).

The specialist's use of *'so'* in line 495 is important. The use of 'so' at the start of a turn has been shown to have several functions. One of these functions is to display that the upcoming action is a pending agenda for the speaker during the interaction and that there is an expected action from the recipient in relation to what is about to be said (Bolden, 2009; Raymond, 2004). 'So' also indicates that the action is either in relation to a preceding topic (Sacks, 1987) or the introduction of a new one (Schiffrin, 1987). In this example, the use of 'so' by the specialist is in relation to the patient's passive stance regarding her response to her risk, and 'so' further indicates that the specialist has an agenda, i.e. to highlight the value of accepting genetic testing so that the patient can actively manage and potentially avoid developing cancer. Risk avoidance and risk management is thus something that the specialist deems important for the patient rather than a stance of not responding to the risk at all. Further evidence that the specialist supports uptake of genetic testing to manage the patient's cancer risk is shown through their words: *'so that's the point of these tests'* (line 516), '*in order to make the chance as low as possible'* (line 523) *'we monitor you to prevent a cancer'* (lines 524-525), *'catch it early'* (line 525), *'versus not doing anything'* (line 527), *'and then you get a cancer and then it's too late to do anything'* (line 529).

In line 530, the patient responds to the specialist's positive framing of the test by confirming that her sister had mentioned the same risk management strategy that the specialist has just conveyed, hence why she is at the GC consultation. The patient's choice of words and the way they design their turn is further evidence that they are not at GC, nor are they doing genetic

testing, out of their own choice. Rather they have attended the consult at the wish or concern of other individuals.

Although the specialist has communicated in a direct manner in the latter lines of Example 11.14: Part 2, I argue that this may have been done to manage some of the patient's sudden verbalised concerns produced *after* her claim of being fatalistic. Although the patient indicates a fatalistic stance in lines 467-494, the patient suddenly changes her stance in lines 499-514 and verbalises her concerns about being told that she is going to die, and that she is grateful to know that she has options, and that the genetic specialist may be able to assist her in managing her risk. She also indicates that she wants to *'live'* (line 508*)* and that she wants to see her grandchildren grow up and her son finish school (lines 511-512). This sudden disclosure of psychosocial and 'lifeworld' concerns could explain why the specialist uses the genetic test to manage the feelings of uncertainty in the interaction by indicating that something can be done about the patient's risk which will aid in her having a healthy future. The specialist's positive framing of the uptake of genetic testing could also be a form of hope work (Peräkylä, 1991).

Although the specialist in this consultation has been direct in discussing the uptake of genetic testing, the direct approach is done in relation to addressing the patient's psychosocial concerns which the patient seems to value and appreciate. The specialist has thus enacted a caring relationship by making the patient despite them being more direct. The patient's change from verbalising a fatalistic viewpoint, to her sudden concern about her options to manage her future cancer risk is somewhat confusing, however. At the end of the consultation the patient decides to proceed with genetic testing for HBOC. As the patient was not interviewed after the consult, her true feelings regarding her risk perception remain unclear.

11.4 The challenge of remaining neutral when conveying risk information

Genetic specialists in the study further indicate that their risk perceptions may impact how they provide risk information in GC consultations. Differing risk perceptions between genetic specialists and patients are seen to be especially problematic when it comes to remaining neutral in their communication towards a patient. Evidence is provided in Examples 11.15, 11.16 and 11.17.

Example 11.15
S1: "When I think to myself: "Oh my word 5% that's nothing," but uh, it's sort of an internal feeling and I'm hoping that it doesn't, that I don't portray it to them, I may in some way. Um there are, very occasionally a slip like: 'Its only 1%." Not releasing that maybe, they think only 1% is a very high risk. So, you know, that kind of thing trying not to put my perceptions out there is quite difficult."

Example 11.16
S2: "Ja. And I always try and get the overall bigger perspective for example if you have an AMA uhm counselling session and the woman has a 1 in 100 risk and there is nothing found on ultrasound and she's crying her eyes out saying she's so worried… sometimes you're not dir. I'm not directive but I'll give uhm the overall like perspective, put it into context and say: "look there is a 99% chance that this baby's fine, only 1%."

Example 11.17
S6: "…. I think we all have our preconceived uhm, well you know, opposite perceptions of risk. You know, a 1 in 5000 risk is VERY small, it's impossible, it's definitely not going to happen. You know. But you can never use the words definitely, you can never put it to the patient like that, you HAVE to remain, uhm, non-directive and unbiased when you are giving risks…Uhm and I think it's impossible to do it perfectly every time."

A quote from a genetic specialist (Example 11.18) suggests that genetic specialists in the local context are discouraged from using descriptive terms such as 'big' or 'small' to describe a risk. However, communication between a genetic specialist and a patient could influence each other's perceptions and feelings of uncertainty based on their personal characteristics and what each of them 'brings' to a consultation.

Example: 11.18
Researcher: "Do you feel like that [risk perception] has an effect on uncertainty communication in genetic counselling or genetic consultations?
S12: (long pause) I've come to learn with time that you can present risks objectively without sort of any emotion attached to it when you have a specific risk figure. OK. But at the same time, I feel that as counsellors you assign a meaning to that based on your interaction with the patient anyway.
Researcher: Mmm
S12: "So, depends on what the patient is bringing a lot of the time, you might frame it and say: "There's a small risk of something." Instead of just using the figures. And I feel like as a student, I was very careful not to say small or use any kind of description of a risk figure. And now as I get more into my counselling and I'm more established, I find myself slipping and using those kinds of uhm adjectives. Which I don't know if they're always as appropriate or not, but I think I make the call depending on the situation."

The quotes given in the examples above highlight how specialists may grapple with feelings of uncertainty about whether they have 'done the right thing' when attempting to remain neutral whilst simultaneously trying to alleviate the patient's fears and perceptions about a risk. Rather than engaging in paternalistic practices where the healthcare practitioner decides on behalf of the patient, GC promotes patient autonomy and patient-centred decision-making (Chima, 2013; Sarangi & Clarke, 2002a). Local guidelines for GC also prescribe non-directiveness (HPCSA, 2013, 2019). Neutral or non-direct counselling approaches are thus favoured in GC.

A recent Dutch study by van der Steen (2019), focusing on the level of information provided, patient-centeredness and directiveness by clinicians when providing GC to pregnant couples, found that the clinicians' counselling styles and the way that they communicated risk influenced patients' risk perceptions, even when attempts at neutrality were made. Genetic specialists in this study admitted that although they attempt to remain non-directive in their communication, they may struggle with balancing internal feelings and external actions when discussing risk and may thus unintentionally encourage patients into a particular decision-making direction. For example, spending more time discussing one test over another, or genetic specialists' word choices e.g., the risk is *only* 1/1000 may be regarded as subtle prompting of a specific decision.

The challenge for genetic specialists to remain neutral when imparting risk information to patients is noted throughout the interactional data. In the following section, I focus specifically on evidence from the consultations, where issues of neutrality occur when genetic specialists offer patients the option of genetic testing. In the consultations where a genetic test is offered, careful analysis of the way the specialists formulate and design their turns of speech during patient decision-making around a given risk reveals that genetic specialists may struggle to conceal their personal risk perceptions or their preferred patient responses. When a patient chooses a decision that is not aligned with the specialist's preference or when patients have different risk perceptions, a specialist may provide subtle or overt directive displays.

Of the eighteen video-recorded GC consultations in the study, ten of the consultations (eight prenatal and two hereditary cancer consultations) involve an offer of a test. In these consultations, testing in the prenatal consultation data involves an amniocentesis to collect amniotic fluid to assess the baby's chromosome make-up to determine the presence of a congenital condition, most commonly Down syndrome. For the two hereditary cancer consultations, *BRCA2* genetic testing is offered to two asymptomatic siblings whose sister has breast cancer and has been confirmed to carry a *BRCA2* mutation (each sibling was counselled in a separate consultation). The remaining eight consultations in the data do not involve the offer of a genetic test, either because they are follow-up consultations where a genetic test has already been performed, or there is no available genetic test for the condition concerned.

In the following examples below I argue that genetic specialists use *perspective display sequences (PDS)* (described by Maynard, 1989) when discussing genetic testing. These PDSs inadvertently introduce direct communication that indicates implicit or explicit preferences about a patient's decision-making regarding a test.

Maynard (1989) explains that when a speaker offers an opinion upfront, it may have potential relational consequences if the listener disagrees with the speaker's assessment. Thus, to cautiously navigate the request for an assessment or an opinion, a speaker may display a three-part PDS in the following order:

1. The speaker invites the listener's opinion.
2. The listener responds to the request by producing their own opinion.
3. The initial speaker then produces their own opinion about the topic thereafter.

The production of the initial speaker's own opinion in part three can be done in different ways: Firstly, via *extreme case formulation* (described by Pomerantz, 1986) that explicitly displays misalignment with the listener's given opinion, or in an implicit manner where alignment may be implied through for example a 'yeah' or 'mm' token, but the speaker has not necessarily produced a real opinion or assessment (Maynard, 1989).

In explicit displays of opinion, a speaker's use of extreme case formulations could be an attempt to convince the listener to act in a particular way. Pomerantz (1986) shows that when a speaker claims something in a conversation, they may use *extreme case formulations* together with descriptive details to justify and support their claim to convince the listener that their claim is legitimate. Furthermore, when healthcare practitioners try to encourage patient reflections on coping with risks and feelings of uncertainty, they may use a PDS and extreme case formulations to introduce *hypothetical framing* (Peräkylä, 1995) (encouraging reflection on contrasting outcomes), or *contra-indicative statements* (Candlin & Lucas, 1986) (statements that emphasise the negative outcomes of a particular decision). A GC study by Sarangi and Clarke (2002b) demonstrates how genetic counsellors and patients may use various theoretical outcomes and contrasting scenarios as rhetorical devices to display and negotiate their preferred choice. Their study further highlights how a genetic counsellor may use these linguistic devices on a micro-interactional level to display aligned choice, whilst still

attempting to remain within their professional boundary of non-directiveness on a macro-social level.

A speaker's implicit approach in part three of a PDS may be used to avoid conflict in an interaction, particularly when preferred responses are misaligned. Research on informal conversations (Maynard, 1986; 1989) as well as healthcare interactions (Heritage & Maynard, 2006; Maynard, 1991) indicates that subtle responses in PDS's are typically used in conversations to maintain rapport cautiously and navigate delicate topics between unacquainted individuals. I argue that the findings in this study show that genetic specialists use PDS's and subtle responses to a patient's choice to display support, or implied support (even when their risk perceptions may be misaligned).

Table 11.2 provides a comparative summary of implicit and explicit displays of direct communication by specialists when they disagree with the patient's decision regarding genetic testing.

Table 11.2: Summary of implicit & explicit displays of direct communication

Perspective display sequence (PDS) pattern	***Implicit* displays of direct communication by the genetic specialist showing *misalignment***	***Explicit* displays of direct communication by the genetic specialist showing *misalignment***
1) The speaker invites the listener's opinion	Specialist invites the patient to reflect on risk presented through initiating a PDS	Specialist invites the patient to reflect on risk presented through initiating a PDS
2) The listener responds to the request by producing their own opinion	Patient provides own risk perception together with an account to justify their reasoning behind their decision.	Patient provides own risk perception together with an account to justify their reasoning behind their decision.
	Patient's response is treated as a misaligned response by the specialist in accordance with their own risk perceptions	Patient's response is treated as a misaligned response by the specialist in accordance with their own risk perception.
3) The initial speaker then produces their own opinion about the topic	Misalignment is displayed implicitly by the specialist.	Misalignment is displayed explicitly by the specialist.
	Specialist indicates apparent support of patient's choice due to the following direct communicative displays:	Interactional trouble is overtly displayed by the specialist through: • Pauses • Minimal acknowledgement tokens • Repetition of questioning

	• Specialist provides several agreement tokens in response to patient's choice. • Specialist emphasises positive aspects of the patient's choice.	• Directly challenging the patient's decision. • Use of extreme case formulations to assist in creating uncertainty for the patient as to whether the right choice was made.
	Misalignment of the patient's choice becomes evident through the following actions: • Genetic test is offered/reoffered regardless of patient's indication of not wanting or being interested in testing. • Genetic risk is re-emphasised.	• If a specialist's (multiple) attempts to dissuade patient's decision fails, the specialist unwillingly concedes. • The conversation moves on in a somewhat awkward and clumsy manner, indicating unresolved tension.

I have chosen three consultations from the data to demonstrate implicit and explicit displays of direct communication.

- The first two example shows *implicit* displays of direct communication where specialist and patient choices regarding the genetic test *misalign.*

- The third example provides evidence of explicit forms of direct communication from the specialist when the specialist's and the patient's choices regarding the genetic test misalign.

11.4.1 Implicit displays of direct communication when specialist & patient choices misalign

This section demonstrates the subtle interactional pattern observed when there is a misalignment between the specialist's and the patient's risk perceptions and when a patient orientates towards a decision that the specialist does not agree with.

In Example 11.19, I return to case 5, a prenatal consultation to demonstrate implicit misalignment. In the consultation, the patient declines an offer of an amniocentesis, indicating that they are not concerned about the risks conveyed to them by the genetic specialist. The specialist, however, displays that the test and risk information should be considered. Although displaying misalignment with the patient's choice, the example shows how a specialist may not necessarily overtly challenge the patient, rather showing their misalignment in more subtle ways. The finding in this example supports Maynard's (1991) and Heritage and Maynard's

(2006) descriptions of the use of PDS in patient-physician interactions and how they assist in navigating potentially contentious medical information.

Example 11.19

Case Overview: ***Case 5- Advanced Maternal Age***

This case involved a 42-year-old isiXhosa woman who was 15 weeks pregnant who was referred for AMA counselling, which included the option of invasive testing (namely an amniocentesis) in order to confirm the possible presence of a fetal chromosome abnormality. The patient had undergone an ultrasound examination prior to her GC consultation in which no soft markers were detected. These soft markers are used as risk determinants for a chromosome abnormality. It is important to note that the soft markers are considered screening tools and are not diagnostic. The patient had four healthy children and no history of a genetic concern. The patient's risk was thus determined based on her age alone. The session was conducted in English, the patient's second language, and the specialist's first language.

S= Specialist; P= Patient; N = Nurse

29	S:	Okay (.) So the cha:ance now has gone up to one in sixty five (0.4) So ((pointing at table))
30		you see it starts going up quite steeply as you get over forty.
31	P:	((head nod)) Hmm
32	S:	So sixty five forty two year old women (.) all we know is that you are
33		forty two↑ and pregnant (0.2) one of those women will have a baby with
34		Down syndrome↑ (.) and the other sixty four wont (.) Okay?
35	P:	((small head nod))
36	S:	So that means you cha:ance is abo:out one in
37	N:	(0.5) ((knocking at door and session interrupted as nurse comes into the room and starts looking for a file))
38	S:	Oh hey Sheryl* (0.4) one in (.) about uhm- ((looking at nurse))
39		(1.5) ((specialist and patient both looking at nurse until she leaves room with the file))
40	S:	about uhm 1.5 percent [okay]?
41	P:	[((head nod))] Hmm. (0.4)
42	S:	1.5 percent. That's about 98.5 percent chance that child doesn't have
43		Down syndrome (0.3) Okay?
44	P:	(head nod))
45	**S:**	Does that? (0.2) **How does that fe:el?**
46		**(0.5)**
47	**P:**	**.Hhh .hhh** **((looks away))**
48		**(2.0)**
49	**P:**	**Aai I feel positive [uh because there is no-one in the family↑]**
50	**S:**	**[((rapid head nod))]**
51	**P:**	**and my aunt had a child at (0.3) uh what was it? (0.3) uh I think**
52		**at forty three.**
53	**S:**	**Ja (0.2) Mmm (.) ok.** **((head nod))**
54	**P:**	**And my sister also had a child now (0.4) at forty one or forty two**
55	**S:**	**And they've all been fine?=**
56	**P:**	**=yes** **((smiling))**
57	**S:**	**So your experience tells you that your chance is not (.) not**
58		**big [to have a child] with Down syndrome?**

59	**P:**	**[yes] (0.3) yes:s ((smiling))**
60	**S:**	**°Okay°**
61		(1.0)
62	S:	And I think the question is would you want to know anyway?
63		(0.5)
64	**S:**	**So we can offer you a test to see if the baby has got Down**
65		**syndrome? (0.4) Would you want to know?**
66	**P:**	**.hhh .hhh .hhh**
67		**(0.5)**
68	**P:**	**What test is that? ((produces a wry smile))**
69	**S:**	**(0.2) Okay it's the needle↑ test (0.4) The amniocentesis ((pointing at her own stomach))**
70		**(1.5)**
71	**P:**	**Is it?**
72		**(1.0)**
73	**P::**	**Mmm (0.2) .hhh .hhh Is it (0.2) sore?**
74		**(1.5)**
75	**P:**	**[.hhh.hhh]**
76	**S:**	**[I'll show you] (0.2)** Appa↑rently (.) I've never had it so I ((smiling & takes out a diagram of an amniocentesis from a file and places it on lap))]
77	S:	can't tell you if its sore or not
78		(2.0) ((flipping the page over in the counselling file to show patient))
79	**S:**	**Uhm (.) but basically what happens is that they do a scan↑**
80	**P:**	**Mm**

In the example above the genetic specialist informs the patient about her risk to have a baby with Down syndrome based on her age (lines 29-43). The specialist uses both ratios (presented as one in sixty-five), percentages (presented as 1.5% and 98.5%) and framing (presented as 1.5% chance to have a baby with Down syndrome and a 98.5% chance to have a healthy baby) to convey the risk information to the patient.

After a head nod from the patient in line 44, the specialist initiates a PDS in line 45 by asking the patient how they 'feel' about the risk information. The questions could also be the specialist exploring the patient's psychosocial concerns. After some awkward behaviour displayed by the patient through pauses and unsolicited laughter in lines 46-48, the patient responds that she does not perceive the risk for Down syndrome to be high. She accounts for her risk assessment by explaining that her aunt and sister both had healthy pregnancy outcomes when they were of advanced maternal age in lines 49, 51-52 and 54. Accounts are often offered by individuals during an interaction to justify their thought process and to establish their expertise on a topic (Heritage, 1984). Pregnant AMA women using other family members who had healthy babies when they were older as a justification for not being concerned about their pregnancy risks has been previously described in another South African GC study (Wessels, 2014).

After confirming in line 55 that the patient's children are all healthy, and summarising the patient's risk perception in lines 57-58, the specialist appears to show alignment with the patient's choice and seemingly accepts their risk perception. Apparent alignment is displayed by the specialist with an *'okay'* in line 60, as well as head nodding in line 50 and '*ja (yes), mm and okay'* in line 53.

Indication for possible misalignment in risk perceptions and the specialist's goal of offering the patient a genetic test based on her AMA risk becomes apparent from line 62. Rather than accepting the patient's risk perception where she has stated that she is not concerned about the 1.5% risk that her child could have Down syndrome, the specialist proceeds to offer the patient an amniocentesis regardless of the patient's verbalised stance on the matter. In line 62, the specialist encourages further patient reflection by asking whether the patient would want to know whether her unborn child has Down syndrome or not. Before allowing the patient to answer, the specialist offers an invasive test and again encourages patient reflection by asking if they would want to know the baby's health status in lines 64-65. The repeated questioning from the specialist about whether the patient would want to know, and the offer of an invasive genetic test, even when the patient has indicated that she is not concerned about the risk, could be an attempt by the specialist to highlight that the patient's risk should be of concern to her. The patient response to this offer is provided through an uncertain and hesitant manner, indicating that the offer of a test is problematic. The patient's hesitancy is displayed through non-verbal behaviour including pauses (lines 63, 67, 70, 72, 74), unprompted laughter (lines 66, 73,75) and a wry smile (line 68). The patient's displays could indicate that there is trouble in the interaction and that the patient knows that her disinterest in pursuing testing is problematic to the specialist. The specialist either doesn't notice or chooses to ignore these nonverbal patient cues and moves on to explain the amniocentesis procedure in further detail.

It is usually standard practice in local genetic clinics to offer invasive testing to referred AMA patients, most commonly an amniocentesis. Globally, and in South Africa, AMA is one of the main indications for invasive testing to detect associated risks for fetal chromosome abnormalities (Beekhuis et al., 1994; Wessels, 2014). Age categories for AMA vary, with AMA being considered from the age of 35 years in South Africa (Wessels, 2014). In Example 11.19 above where the specialist chooses to explain and offer an amniocentesis to the patient, not only does the specialist's misaligned risk perception become apparent, but their agenda and goal to offer an amniocentesis to the patient regardless of her risk perception is highlighted.

After explaining the test procedure, the patient sticks with her original decision, which is accepted by the specialist and the conversation moves on to another topic.

Although it is not possible to know the motivations behind the specialist's behaviour in these lines, given that the offer of genetic testing for AMA women is standard practice in local GC consultations, the specialist may feel that they need to default offering the test regardless of the patient's indication that they are not interested in knowing more about the test. The example supports evidence seen in other consultations (e.g., Example 11.4) in the study that genetic specialists may revert to script-like agendas during local GC consultations and provide additional risk information to patients even when patients indicate that they are not interested in the information.

11.4.2 Explicit displays of direct communication when specialist & patient choices misalign.

Focusing on the explicit pattern in Table 11.2, the following example, Example 11.20 (Part 1 and Part 2), taken from a prenatal consultation, demonstrates direct displays of genetic specialist communication when they overtly indicate their preferred patient choice regarding an offer of a genetic test. The overt display is shown when own risk perceptions do not align with that of the patient. In this example, I further demonstrate the interactional dilemma when a specialist is asked for advice by a pregnant patient when deciding on the uptake of an invasive genetic test.

This consultation is an example that is not in keeping with the other study findings as it is the only consultation where the patient has already previously undergone an amniocentesis in her current pregnancy, and who also terminated a baby diagnosed with Down syndrome in her previous pregnancy (the diagnosis at the time was also confirmed through an amniocentesis). The patient thus had personal experience of undergoing an amniocentesis procedure and having a termination for a baby with Down syndrome which may have significantly impacted her perception of both the procedural risks associated with an amniocentesis, and the risk of having a baby with Down syndrome. Although some genetic specialists mentioned in their interviews that they had previously encountered patient requests for advice, this consultation is also the only interactional example in the data set where a direct patient request for advice was observed.

Example 11.20

Case overview*: Case 4- AMA & a previous baby with Down syndrome*

Case Overview: This is a follow-up consultation involving a 43-year-old Mixed Ancestry woman who is a nurse at one of the research site hospitals. The patient had initially been referred to the prenatal GC clinic due to her age, which placed her at a higher risk for having a baby with a chromosome abnormality. In addition, she had terminated a previous pregnancy because the baby had Down syndrome.

During her initial GC consultation for the current pregnancy, she decided to have an amniocentesis which was performed on the day. During her follow-up GC consultation, she was 20 weeks pregnant and had also received a detailed fetal anomaly ultrasound scan prior to the consult. No soft markers for any chromosome abnormalities were detected at the time, thus her risk has been reduced.

The patient was attending the GC follow-up consult to discuss these adjusted risks as well as her amniocentesis results. During the consultation, the specialist conveys that the amniocentesis has failed to produce a result owing to a poor-quality amniotic sample, thus they were unable to provide the patient with a diagnostic answer. After discussing the failed amniocentesis and the patient's reduced risk based on her most recent ultrasound findings, the option of re-doing the amniocentesis is offered. The patient decides that she wants to go ahead with a second amniocentesis to clarify her risk. The patient's decision creates tension in the interaction, due to what I will argue are differences in risk perception and preferred choice between the patient and the specialist. In the example, it appears that the genetic specialist thinks the risk of Down syndrome is low and the risk for miscarriage is high, whilst the patient thinks the risk for Down syndrome is high and the risk for miscarriage is low. The session was conducted in English, the patient's second language.

In the excerpt below, Example 11.20: Part 1 the specialist is relaying the patient's adjusted risk based on the patient's ultrasound findings. The analysis shows displays of direct communication. Interactional tension is created when the patient chooses to have a second amniocentesis, whilst the specialist works hard to convince her otherwise.

Example 11.20: Part 1

S= Specialist; P= Patient

91	**S:**	… because they didn't see anything↑ (.) has made
92		your risk **very very very small** (0.2) to have a baby with
93	P:	°mm°
94	S:	Down syndrome. So it's about one in five thousand now.
95		(2.5)
96	P:	Yes I understand
97	S:	You understand?
98	P:	Yes
99	**S:**	**How does that make you feel?**
100	P:	More positive.

101	S:	↑MORE positive?
102	P:	More positive °yes°
103	S:	You seem like a very positive person [(.) you definitely] seeing
104	**P:**	**[Hhhh hhh yes]**
105	S:	the positive in this
106	**P:**	**Yes, I uh am**
107	**S:**	**Ok so I mean the question now is whether then you still want**
108		**to go (.) ahead and do another amnio↑ (0.2) or if you'd like**
109		**to leave it?**
110		(0.5)
111	**P:**	**No:o I will do↑ the amnio**
112	**S:**	**You want↑ to do the amnio↑?=**
113	**P:**	**= yes:s**
114		(0.5)
…	…	…
143	**S:**	**So:o:o (.) .hh the question then is (.) whether (0.3) WHY↑ you (placing ultrasound report back on desk))**
144		**want to do the amnio?**
145		(1.0)
146	**S:**	**WHAT's is your (0.3) >what is your< ne:ed for this °amnio°?**
147		(0.3) <Will↑ it mean that>
148	**P:**	**. hhh she mos (.) the way that the doctor explained it to**
149		**me now is=**
150	S:	Ja=
151	**P:**	**=because I asked her**
152		(1.0)
153	S:	Ja
154	**P:**	**Do I NE:ED↑↑ to do the amnio (0.2) again today? (.) Because of**
155		**the failure last (.)**
156	S:	°Ja°
157	**P:**	**The last amnio.**
158		(0.5)
159	**P:**	**So I AM↑ going to do it** (0.3) Cos (.) the only thing that they
160		going to check is for Down syndrome again↓? ((shrugs shoulders and emphasising point with hand in air))
161	S:	Ja
162	P:	And then they will see if there's st. still any abnormalities
163	S:	Ja
164	P:	Ne?
165	S:	Mhmm
166	P:	>That's is how I understand it< (0.3). So that is the only
167		reason why (.) I will still just (0.2) ((flings hands away from body to emphasise point))
168	**S:**	**You want to do it? (0.3)**
169	**P:**	**Yes I will do it.**
170		(0.5)
171	**S:**	**.hhh And the fact that your risk is very very low now↑?**
172		(3.0)
173	**S:**	**That doesn't <change anything> for your (.) in your mind?**
174	P:	No
175	**S:**	**About the chances?**
176		**(0.5)**
177	**P:**	**°No°**
178	S:	Do you think your chances are still high↑ to have a [baby with]
179		Down syndrome?
180	P:	[No I'm not]
181		**(1.5)**
182	**S:**	**So (0.3) with the amnio (.) comes the miscarriage risk**
183	P:	Yes
184	**S:**	**which we've spoken about hey (0.2) about one in five hundred (.)**
185		**is the chance to have a miscarriage .hh °ok°.**
186		(0.5)

187	**S:**	**Would you be willing to take that cha:ance↑ (.) again?**
188		(0.5)
189	**S:**	**to get that result?**
190		(1.5)
191	**P:**	**I will ye:es:s**

In the example above, the specialist initially discusses and compares the patient's risk given to her during her initial GC consultation (1/600) (not shown in the example) to her new adjusted risk in the current follow-up consultation (1/5000) based on her most recent ultrasound screen (lines 91-94). In line 92, the specialist frames this new risk figure as *'very very very small'*, increasing the certainty of the situation that Down syndrome is extremely unlikely to be present in the baby (and by extension that a second amniocentesis is not necessary). The use of repetition of a single word or phrase e.g. 'no-no', or 'right now, right now', in a single turn constructional unit can be seen as a strong suggestion for the broader course of action proposed by a speaker (Sacks et al., 1974; Stivers, 2004). This manner of presenting the information is thus a subtle but direct strategy from the specialist to convince the patient not to accept an amniocentesis. After a two and a half-second pause in line 95, the patient verbally confirms their understanding in line 96, and the specialist repeats the patient's statement in line 97, leading the patient to produce a verbal *'yes'* in line 98.

In line 99, the specialist initiates a PDS by asking the patient how the presented risk makes them '*feel'*. The patient replies in line 100 that they feel *'more positive'* to which the specialist responds by exaggeratedly repeating the patient's answer using a rising intonation. Although the patient seems to refer to feeling more positive about the risk situation specifically, the specialist goes on to extrapolate the patient's positivity to her general disposition and personality in lines 103 and 105. By stating *'you seem like a very positive person. You're definitively seeing the positive in this'* and using extreme case formulations *'very'* and *'definitely'*, the specialist displays her intention of encouraging the patient not to proceed with a second amniocentesis.

All individuals in a conversation speak with a goal. This goal may be to convince, to convey information, to impress, to agree, to disagree, to gain sympathy etc. Speakers formulate their turns of speech and choose their words with these goals in mind and adapt them according to their audience. By making verbal inferences about their audience's character traits, they attempt to obtain confirmation from the audience whether their goal is being successful or not. Furthermore, the speaker gains insight into the audience's understanding, beliefs and affective

state, so that they can enhance and adapt their communicative strategies to achieve their goal (Kraut et al., 1982). The patient responds to the specialist's inference that they are a positive person who is seeing the positive aspect in the risk situation, by confirming that they are indeed feeling positive (lines 104 and 106). The inclusion of laughter particles by the patient in lines 104, which is not prompted or reciprocated by the specialist, could, however, be a sign of trouble, as previously reported in medical consultations (Haakana, 2001; Jefferson et al., 1987).

After highlighting that the patient is a positive person, the specialist enquires whether the patient wants to have a second amniocentesis procedure in lines 107-109. The patient responds in line 111 that she does. The specialist requests clarification by repeating the patient's response in line 112. The specialist's repetition of the patient's response in line 112 is said in a somewhat surprised manner with rising intonation, indicating that the specialist did not anticipate this response from the patient.

Repetition of a patient's statement by physicians is thought to display receipt of information and to encourage elaboration of the information supplied (Beckman & Frankel, 1984). Furthermore, the intonation in which the other speaker responds to a statement becomes important in understanding the purpose of the repetition. A study by Svennevig (2004) of Norwegian conversations found three different forms of repetition, each having different sequential implications depending on how the repetition was presented. If produced with falling intonation, the repetition typically displayed hearing, whilst a repeat accompanied by a response particle such as 'ja' ('yes') indicated understanding. If repetition were produced with a rising intonation it often would indicate emotion such as surprise, approval or interest in the information presented or claim made by the previous speaker. The specialist's repetition of the patient's statement in the example above demonstrates that she wants to proceed with the amniocentesis indicates surprise from the specialist.

In lines 115-142 (not shown in the transcript), the patient justifies her reasons for wanting to pursue the procedure by providing an account that she is concerned about the baby's growth and wants to confirm that there are not any genetic problems. This does not seem like an acceptable justification to the specialist and the specialist probes further into the patient's reasoning for wanting an amniocentesis in lines 143-144 and 146. The patient then explains in lines 147-167 that she wants to redo the amniocentesis because the last one failed. She uses a discussion with the doctor (a fetal medicine specialist at the pregnancy clinic) who performed

her most recent ultrasound as a further justification for a second amniocentesis. Although the patient does not provide insight into what the doctor has told her specifically, she suggests that the doctor may have encouraged her to perform the second amniocentesis. This is evident in the patient's choice of words: '*The way the doctor explained it to me now'* (lines 148-149); '*because I asked her*' (line 151), '*Do I NEED to do the amnio again today?*' (line 154) and '*So I AM↑ going to do it'* (line 159). In using her discussion with the doctor as a motivator for her decision to redo the amniocentesis, the patient defers to medical hierarchy, implying that the doctor's opinion has the power to override the genetic specialist's preference that the patient does not perform a second amniocentesis. The patient also infers that the doctor may have been more direct in their advice-giving suggesting that she does do a second amniocentesis. The doctor's actual level of directiveness in the prior discussion with the patient is however unknown.

This patient is in a unique position in this GC consultation as she is a nurse at the hospital where the consultation took place. Healthcare practitioners tend to have a greater source of power than patients in the context of healthcare interactions by virtue of their association with medical institutions (Nimmon & Stenfors-Hayes, 2016). The patient uses her professional identity to further highlight her authority and having an equal position of power to the genetic specialist (who in this instance is a genetic counsellor). Her formal nurse's uniform that she wears may add a further layer of authority to the atmosphere (Langewitz, 2007) of the consultation. Genetic counsellors are considered allied healthcare professionals. Doctors are considered to have more status and authority in decision-making and patient management than allied healthcare professionals and nurses (Apker et al., 2005; Braithwaite et al., 2013; Nugus et al., 2010). The patient could thus use her discussion with the doctor and their reported encouragement to do the amniocentesis as an interactional resource to not only justify her decision-making, but to re-enforce the doctor's authority and expertise on the risk. The nurse may feel in a more powerful position to challenge the genetic specialist, a medical colleague, compared to other layperson patients.

The patient is in a further unique position in that she has already undergone an amniocentesis, thus has an enhanced level of understanding on what to expect, not only from her professional insight but from personal experience. The specialist seems not to accept this account from the patient and again requests clarification in line 168 on whether the patient wants to go ahead

with an amniocentesis. This repeated questioning by the specialist after the patient has already indicated on more than one occasion that they do want to go ahead with the test, demonstrates the misalignment between patient's and specialist's preferred outcomes and the specialist's rejection of the patient's decision.

When the patient insists that she wants to do the second amniocentesis in line 169, the specialist tries to change tactics in lines 171, 173, 175, and 178 to dissuade the patient by emphasising and framing the risk as '*very very low now'* and asks whether this changes the patient's perception. The patient rejects the specialist's statement by giving short *'no'* responses in lines 174, 177 and 180. The positioning of the 0.5-second pause in line 176 before the softly-spoken 'no' response from the patient in line 176 indicates that the patient realises that this is a response that the specialist does not want to hear. Schegloff (2007) highlights that in the case of misaligned responses, a pause often occurs between a speaker's turn in the first pair-turn part and a recipient's response in the second pair part. Furthermore, the pause in line 181 indicates trouble for the specialist, who then adjusts their strategy further in lines 182, 184-185, 187 and 189 by foregrounding the associated negative consequences of going ahead with the procedure, namely the risk for procedural-related miscarriage. The emphasis on the negative consequences of a particular decision is a previously described interactional strategy deployed by healthcare practitioners when discussing risk with patients (Candlin & Lucas, 1986).

Highlighting the negative consequences of the patient's decision proves an unsuccessful dissuasive strategy for the specialist and the patient confirms that she will proceed with the second amniocentesis procedure by replying *'I will yes'* in line 191 in response to the specialist's question: *'Will you be willing to take that chance again?'* in line 187. The segment continues with several further attempts by the specialist to convince the patient not to go ahead with the procedure, with further active displays of resistance from the patient.

The patient's adamant goal of proceeding with the amniocentesis could be an indication that her perception of potentially having another baby with Down syndrome may be too much for her. She needs to know whether her unborn baby in her current pregnancy is affected or not. The patient's need to know thus outweighs the risk of potentially losing her baby due to the amniocentesis procedure. Therefore, she is willing to take the risk of performing a second amniocentesis where: *'the only thing that they going to check is for Down syndrome again'* (line 159).

The second part of the example, Example 11.20: Part 2, shows the moment in the interaction where the patient overtly requests direct advice from the specialist. The patient's request for advice creates a sudden shift in the specialist's communication style from being direct, to displaying a deliberately neutral position.

Example 11.20: Part 2

S= Specialist; P= Patient, Ben*= The baby's father & patient's partner

35	S:	Ok (0.4) Cos that's really now (.) the only thing really now
36		is that (.) is that you want to do this test now (.) that (.)
37	P:	Mmm
38	**S:**	to think about what would happen then (0.2) **how you would feel**
39		**about it and how Ben* would feel about it if (.) you did have**
40		(.) **you were that one in five hundred**
41	P:	Ja
42	**S:**	You did have this miscarriage (0.2) **I mean your risk is (.) is**
43		**very low in terms of (.) to have a baby with Down syndrome.**
44	**P:**	**Mm**
45		**(0.5)**
46	**P:**	**So if I can ask YOU a question (0.2) If you were in**
47		**MY shoes now (0.4) WHAT would YOU do now?** **((points at specialist and then at herself))**
48	**S:**	**It's a very difficult thing to say. Because [I'm not in your**
49	**P:**	**[hhhh hhhh**
50		**shoes]. (0.3) I know it (.) that sometimes we want↑ to ask**
51		**for advice (0.2) and to ask for (.) for help (.) [hey?]**
52	P:	[Ja:a]
53	**S:**	**These decisions are very difficult to make (.) It's not fair**
54		**that you have to make these decisions.**
55	P:	Ja:a
56	**S:**	**But (0.2) but I think you can make this decision (.) what's**
57		**best for you.**
58		**(2.0)**
59	**S:**	**What do you think it best?**
60		**(0.5)**
61	**S:**	**And perhaps Ben* also (0.2) What does he think about all this?**
62		**(3.0)**
63	**S:**	**Is he meant to help you make these decisions?**
64		**(2.0)**

After several attempts to dissuade the patient from going ahead with a second amniocentesis, the specialist seems to become lost for words and appears to concede that they have failed to discourage the patient from their decision. The specialist's hesitancy is evident by several pauses, hitches and muddled sentence construction in lines 35-43. In these lines, the specialist seems to employ a further strategy to encourage reflection via hypothetical framing to point out the potential psychosocial consequences for the patient and her partner, Ben*, if she was that '*one in five hundred'* (line 40) to have a miscarriage. The specialist's personal risk perception and lack of neutrality are further highlighted when they state: *'I mean your risk is very low in terms of having a baby with Down syndrome'* in lines 42-43, displaying that the patient's decision to have the test is misaligned with the specialist's perception of risk.

The direct utterance from the specialist in lines 42-43 appears to create uncertainty for the patient regarding her decision-making. The patient displays uncertainty by an '*mm*' response in line 44 and a pause in line 45 as if she is considering the specialist's prior utterance. The patient's uncertainty is explicitly highlighted in lines 46-47 when she decides to seek advice from the specialist. The patient starts her turn with the discourse marker '*So*'. Again, the marker 'so' may display a speaker's agenda for the next turn of phrase, and that there is an expectation for a course of action (Bolden, 2009; Raymond, 2004). The patient's use of the marker *'so'* indicates that her agenda is to get the specialist to provide an opinion of whether the patient should do an amniocentesis or not. Even though interactional analysis reveals that the specialist's risk perception of the situation is low and that their preferred choice is for the patient to decline the offer of an amniocentesis, the specialist never overtly verbalises their preference.

With the explicit request for direct advice, the specialist immediately reverts to a neutral position in lines 48 and 50-51. Overt displays of neutrality from genetic counsellors after the offering of invasive testing to a pregnant patient have previously been described in a South African study (Wessels et al., 2015). The specialist orientates to a neutral position by acknowledging the patient's difficult decision, but they are however not in the patient's shoes and cannot make the decision for them. The neutral response from the specialist is reacted to by laughter from the patient in line 47, indicating that the specialist's response is problematic. Unprompted and unreciprocated laughter is thought to be a sign of trouble in an interaction (Haakana, 2001; Jefferson et al., 1987). The specialist's sudden overtly neutral stance may be confusing for the patient since the specialist, even though never directly saying *'I don't think you should have an amniocentesis'*, has openly challenged the patient's decision-making and risk perception in the preceding sections of the consultation. After the specialist re-enforces her neutral position in lines 56-57 by expressing that the patient has the autonomy and ability to make the best decision for herself, the patient responds with a two-second silence in line 58. When the specialist poses the question asking the patient what she thinks is best for herself (line 59) and her partner (lines 61), the question is met with further prolonged silence from the patient (lines 60, 62 and 64). These lengthy silences provide additional interactional evidence for trouble in communication. The non-committal response provided by the specialist is not what the patient wants to hear.

Refraining from providing advice, even when patients seek it, has been previously described in GC research (Zayts & Schnurr, 2014). Genetic specialists in this study reported that perhaps the most challenging aspect of remaining neutral in GC consultations is when patients ask for direct advice regarding decision-making on conveyed risk information as shown in Examples 11.21 and 11.22.

Example 11.21
S7: "And then they have all these options and then I think that, to pick the right one and uhm… I have, I've been asked numerous times I mean what would I do? Like, what decision would I make? If I was in their shoes? And you say: 'Well I can't do that because I'm NOT you and I have a different frame of reference…"

Example 11.22
S6: "And the difficult thing is, I've actually had it twice in like the last week is people saying: "what would you do?" And asking for that advice.
Researcher: "How do you respond?"
S6: "Also no right or wrong way to do that! (laughs). Uhm…. We were taught in a way how to say that and uhm I've sort of moved it around to make it work for me in that setting. Uhm… I generally say: "I understand that this is a very difficult decision for you to make, uhm and often we do want to ask advice from people to ask what someone else would do… but I'm not in your shoes right now." And this, you sort of trying to empower the patient to, to be able to answer the question themselves..."
Researcher: "OK"

The genetic specialist in Example 11.22 is the same genetic specialist that experiences the interactional dilemma of advice-giving observed in the consultation above (Example 11.20). As stated in their interview, the interactional data shows how the specialist reverts to a neutral position in the consultation when the patient asks them for advice, using the described strategy of acknowledging the patient's difficult decision and reminding them that they are 'not in their shoes'. However, further analysis of the surrounding talk involving the offer of the test and the discussion provides evidence that the specialist has not been neutral in the consultation, rather displaying a preferred choice and challenging the patient directly on her decision. These findings support the argument that communication in GC practice is more complex than is suggested in theory.

Being asked for direct advice from a patient can place a genetic specialist in a dilemma regarding how best to support the patient yet remain true to the principles of GC (Weil, 2003; Wessels et al., 2015). Avoiding patient influence may be particularly difficult when society views healthcare professions as experts in their fields (Cockerham, 1992). An interactional study focusing on over 150 recorded GC consultations in Hong Kong demonstrates that healthcare practitioners can struggle in their roles, especially when they view themselves as information providers, but patients view them as advice-givers and co-decision-makers (Zayts & Schnurr, 2014). Further research has indicated that some patients find non-direct

communication unhelpful. When receiving no advice from a healthcare practitioner when directly requested to do so by a patient may threaten the therapeutic relationship (Wyatt, 2001; Yip, 2020). Example 11.23 highlights that local genetic specialists also grapple with feeling responsible for helping patients navigate uncertain decision-making.

Example 11.23
S6: "And I think it's fair you know for people to ask for advice. You know, when we are sitting in the hospital, and YOU'RE the healthcare professional, YOU'RE the expert, you're the person who SHOULD know, you know."

Sarangi (2000) discusses the complex roles that genetic specialists have to negotiate carefully due to the hybrid nature of GC. Although considered an *information*- rather than the *advice*-giving profession, much of the communication in GC is about conveying risk. The very nature of conveying risk information may be seen by the patient as a warning, or by extension advice-giving, even if not intended by the specialist. Arribas-Ayllon and Sarangi (2014) argue that to view GC as a neutral profession devoid of influence and authority is over-simplistic. Hence, genetic specialists must balance carefully the 'tight rope' of meeting patient expectations, while simultaneously not overstepping ethical and professional boundaries.

Patients' requests for advice in GC could be linked to a lack of patient agency in the South African healthcare context. One genetic specialist reported that some local patients do not view themselves as being able to make their own healthcare decisions (Example 11.24).

Example 11.24
Researcher: "Have you had any incidents where patients have found the non-directiveness challenging or new?
S10: I think so. I mean there is always some patients who ask what they should do. Or who say: "No they'll do whatever they…" You know, "You can just tell me what to do." Those types. Or ja, patients who say: "No they're here because this doctor told them to." Or they did that because this 'doctor told them to.'
Researcher: OK
S10: Or other. Not exactly how it should have happened or.
Researcher: Mmm
S10: Uhm and it's also not necessarily true that it happened because of the doctor telling them so, but that's obviously how they think about it. You know, they had an amnio because the doctor told them to."

The lack of patient agency has been previously reported by fetal medicine specialists who argue that patients in the South African healthcare context are more accustomed to direct forms of communication, thus may find non-direct forms problematic, especially in stressful situations such as prenatal counselling, as it may be perceived as abandonment or indifference by the specialist (Adam & Lindeque, 2017). Penn and Watermeyer (2018) have also highlighted the lack of agency and patient assertiveness in a South African healthcare context. Some of the reasons for a lack of patient agency in healthcare interactions may be attributed to broader

societal issues such as socio-economic problems, gender and race relations, poverty, politics, and historical power imbalances (Farmer, 2004; van der Geest & Finkler, 2004). Many of these broader issues are well described in a South African setting, and they may create tensions in local health interactions that impede theoretical healthcare goals, for example, those described in GC textbooks.

The reported patient view from Example 11.20: Part 2 above may have stemmed from their personal experience (and potentially also their professional experience as a nurse) with other local healthcare services where the healthcare practitioner typically offers advice and guides decision-making. Instruction giving approaches have been described in previous South African healthcare communication practices (Watermeyer & Penn, 2009b). Evidence that other areas of healthcare follow more direct forms of communication than GC consultations, is provided by a quote from another genetic specialist in Example 11.25.

Example 11.25
S7: "…And I think other areas of medicine, I mean I haven't worked in, I've worked in paediatrics, but I haven't worked in other areas of medicine for a long time, so I don't know, but I think it, it is possibly is more paternalistic."

Findings in this study, together with findings from local GC (Wessels et al., 2015; Wessels & Koole, 2019) and other South African health research (Adam & Lindeque, 2017; Penn & Watermeyer, 2018) highlight the lack of awareness amongst local patients about their rights to autonomy and being active participants in their own health decision-making. The results show that patients do not reflect, or deliberate on a particular risk in detail, as they may presume that the responsibility of assessing risk and decision-making rests with the genetic specialist, rather than themselves. Other international studies have also highlighted that patients accept and expect healthcare practitioners in state-funded facilities to be more directive and authoritative in their communication approach (Pilnick & Zayts, 2012).

Besides *knowing* their rights to be autonomous decision-makers, patients also require the ability to *see* themselves as autonomous decision-makers. The latter may be shaped by their background and social circumstances (Walker, 2009). Social challenges may cause patients to lack confidence or assertiveness. A large-scale study focusing on Nigerian and Namibian women's decision-making and autonomy in reproductive health showed that factors such as education levels, employment and wealth-status impacted autonomy. Those in poorer socioeconomic positions viewed themselves as being in less powerful positions to make

decisions on their health (Bamiwuye et al., 2013). A lack of agency can be challenging for local genetic specialists to convince patients that they have a right to be involved in their health decision-making, especially when it comes to making decisions regarding risk. Encouraging patients to make decisions in healthcare consultations where there is limited risk information that have uncertain outcomes may be an even more challenging task for genetic specialists.

11.5 Conclusion

In summary, the chapter provides evidence of the tensions that exist regarding expected GC outcomes versus the realities of practice. I have highlighted how genetic specialists struggle to balance patient agendas with their own agendas for a consultation, to tailor information according to individual patients, and ensure that patients are informed of their risks.

Evidence from the data suggests that South African genetic specialists have educational-focused agendas which may result in script-like communicative approaches in GC consultations. Patient-centred care may require greater flexibility in communicative approaches, and greater attention to the therapeutic relationship of GC, which prioritises individual patient goals and affective needs.

The chapter also shows the impact of personal risk perception and how it may influence how genetic specialists respond to risk information as well as their communicative approaches with a patient. Genetic specialists struggle with managing their own risk perceptions and remaining neutral in their interactions with patients. Furthermore, genetic specialists display preferred courses of patient action when offering genetic testing. The preferred patient choice is oriented to via implicit and explicit displays of direct communication from the genetic specialist. Direct displays of communication particularly place tension on GC's theoretical goal of remaining neutral when discussing risk information and facilitating patient decision-making.

The findings further show how patient expectations of the genetic specialist's role in decision-making and advice-giving may complicate the communicative process. A patient's expectation for advice may arise from local attitudes and practices which lead to patients becoming accustomed to more direct forms of communication in other public health services, or the patients' lack of confidence in making their own decisions due to social and institutional asymmetries. Collectively patient expectations impact not only rapport and trust in the

interaction but also bring about questions regarding patient care and how best to support patients whilst remaining within the professional boundaries of GC practice.

CHAPTER 12: GENERAL DISCUSSION

12.1 Introduction

The findings in this study point to the challenges of risk communication and the management of uncertainty in South African GC consultations due to the tension between the ideals and the realities of practice. The study shows that actual GC practice occurs in a complex interactional space. Factors that contribute to the complexity of practice include the many sources of risk that have to be considered for individual patients, decisions on which risk communication format is most suited to ensure patient comprehension and how much information to convey, an awareness of individual patient characteristics, agenda and concerns to tailor information, as well as ensuring that communication occurs in a non-direct manner. Differing patient and genetic specialist risk perceptions further perpetuate complexities in risk communication. Collectively, these complex interactions leave room for interactional uncertainties which place additional pressures on communication and establishing caring patient relationships. Findings from this study suggest that attempts to achieve multiple GC goals simultaneously may impede, rather than promote, patient care in a South African context.

Reflecting on the study's methodology, the approach of using different data sources and a blended qualitative approach to analysis has proved challenging. However, I believe that the methodological approach has also been critical in achieving the aims and objectives of the study. The interactional data reveals the *what* and *how* of risk communication, whilst the genetic specialist interviews provide a more personal perspective i.e. the *why* of risk communication, including what formats genetic specialists prefer using, what formats are helpful/unhelpful, as well specialists' perceptions of genetic risk. In addition, the interview data provides insight into the broader contextual challenges of providing GC in a South African setting. Finally, the researcher field notes have aided in understanding the research setting and have created a base to consider ethical dilemmas encountered during the project, as well as reflect on my position as a researcher investigating my professional field.

In the chapter below, I consider the findings from the study in relation to GC's goals and patient care. I also focus on the research methodology and some of the ethical dilemmas and practical challenges encountered during the research process.

12.2 Study findings

12.2.1 Challenges in achieving genetic counselling goals: Teaching, counselling & tailoring information

One of the biggest dilemmas facing the GC profession is the lack of agreement on what GC goals should be and how best to achieve them. These dilemmas are not only internally felt within the GC community, but the practice is coming under increasing pressure from other healthcare professions and medical management to provide evidence of the added value of GC and how GC improves patient care (Austin, 2010; Cragun & Zierhut, 2018; Hooker et al., 2017; McAllister et al., 2016; Resta et al., 2019).

This study's results show that genetic specialists struggle to balance and simultaneously achieve differing GC goals when communicating risk information to patients in a South African context. These goals include (but are not limited to) educating patients to ensure informed decision-making, establishing a patient's agenda, tailoring information to meet individual patient's needs, and remaining neutral. In the discussions that follow I consider these goals, what the findings in this study reveal about these goals, and the impact on patient care. I use existing literature to support my argument, referring to international and local GC practice and training guidelines and how attempts to simultaneously achieve multiple goals may be impeding GC practice and patient care in South Africa.

Educating and ensuring that patients can make informed decisions are important goals of GC as described by the NSGC (Resta et al., 2006); international GC practice manuals (Biesecker et al., 2019; Uhlmann et al., 2009; Veach et al., 2003), as well as local GC practice and training guidelines (HPCSA, 2013, 2019). Complete disclosure of relevant risk information is thus considered an important aspect of GC communication (Uhlmann et al., 2009). Findings in this study show that genetic specialists may convey large volumes of risk information in one GC consultation. This information incorporates multiple sources of risk that may include population risk, personal risk, familial risk, age-related risks, teratogens, and procedural risks. The study further shows that genetic specialists utilise several formats to present risk information including numerical formats, narratives, framing, metaphors and visual aids. These formats are typically combined in local GC consultations, rather than used individually.

To achieve the GC goals of providing all risk information and promoting informed decision-making, Sarangi (2000) describes the occurrence of 'hyper-explanations' in GC consultations where large amounts of information are provided to patients. Although different sources of risk are important to consider when providing accurate risk information to a patient, and different formats to explain the same risk information may increase patient understanding (O'Doherty & Suthers, 2007; Smerecnik et al., 2009; Trevena et al., 2013), evidence from the study suggests that when too much information is provided and patients are unable to find meaning in this information, this approach to risk communication may not actually be helpful to patients.

Conveying large volumes of information and using several formats to do so within one consultation has been described as an informative approach to risk communication (Bodemer & Gaissmaier, 2012). Informative risk communication assumes that a patient can make their own informed decisions based on their preferences and context (Bodemer & Gaissmaier, 2012). To promote patient choice, this approach to risk communication may include detailed information that discusses all risks as well as all risk management options and their respective outcomes. Several formats may also be used to explain the same information (Ahmed et al., 2012; Bodemer & Gaissmaier, 2012; O'Doherty & Suthers, 2007). However, to ensure that patients have been informed of all their options, large amounts of information are conveyed to patients, as is evidenced in this study. These large amounts of information may achieve the opposite outcome than what is intended and findings in this study show that patients may become confused or detached from the conversation as the volume of information may be overwhelming. Previous studies have found similar findings (Fagerlin et al., 2011; Zikmund-Fisher et al., 2010; Zikmund-Fisher et al., 2011).

Informative risk communication can also be considered a defensive risk communication approach. Defensive risk communication is typically used by health practitioners to advocate outcome options which may not necessarily be beneficial to a patient, but that legally protect the health practitioner (Bodemer & Gaissmaier, 2012). An example is doctors promoting prostate-specific antigen (PSA) screening to patients, even if such screening does not decrease prostate cancer mortality rates (Djulbegovic et al., 2010; Sandblom et al., 2011). Even if a practitioner would themselves not consider PSA-screening beneficial, because of the fear of legal action if a patient does not undergo such screening and subsequently develops prostate cancer, they would rather recommend that patients undergo such screening (Bodemer & Gaissmaier, 2012). Although GC does not provide recommendations for patient decisions or

actions, the provision of all risk information, together with various management and testing options could be defensive risk communication strategies by genetic specialists to ensure that they have achieved the GC goals of educating and informing patients in a non-direct manner. Providing all risk information could also be an effort by the genetic specialist to avoid instances of non-disclosure which may lead to litigation. Therefore, genetic specialists would rather say too much than too little.

Considering the specialists' goal of educating the patient, interactional evidence from the study shows that specialists may miss non-verbal and verbal cues from patients about what their agendas and real concerns are. The importance of establishing a patient (or client)-led agenda and addressing their concerns is described in local and international GC literature (Biesecker et al., 2019; HPCSA, 2013; Pilnick, 2002; Uhlmann et al., 2009; Veach et al., 2003). The interactional data in this study shows that much of the time involved in GC consultations is allocated towards assessing risk and conveying genetic risk information, rather than towards addressing patients' psychosocial concerns. Genetic specialists also provide clear education-focused agendas for themselves at the start of GC consultations.

Establishing patient agendas is a complex task in practice. Although it was noted that most genetic specialists enquire about patient agendas at the start of a GC consultation, most consultations involved genetic specialists asking patients why they have been referred to GC rather than focusing on personal concerns. In instances where patients' agendas were stated upfront at the start of the consultation, or during the consult, some genetic specialists continued to frame the content and activities of the GC consult according to their own, rather than the patient's agenda. True patient agendas or expectations may thus not have been fully explored or solicited in the GC consultations in this study. The findings in this study are supported by previous literature that indicates that agenda setting in GC may become asymmetrical between genetic specialists and patients with the potential for genetic specialists to become overly focused on their own agenda, rather than the patients (Butow et al., 1995; Ellington et al., 2005; Kessler, 1992; Pilnick, 2002).

In the South African public healthcare system, what makes agenda setting particularly challenging is that many patients are referred by other healthcare practitioners, rather than independently choosing to attend GC of their own accord. Although some patients in this study did indicate an agenda when asked, the majority of the patients had little understanding of why

they have been referred, thus had little to no expectations for GC. GC guidelines assume that patients come with at least some information, or expectation for GC, hence are able to indicate an agenda (Clarke et al., 1996; Pilnick, 2002; Veach et al., 2003). However, finding in this study together with previous South African GC literature (Morris, 2013; Perrott, 2010; Wessels, 2014), and international GC studies (Babul-Hirji et al., 2010; Pilnick, 2002; Redlinger-Grosse et al., 2020) show that some patients may not necessarily know what to expect in GC, or why they have been referred. In the study, in instances where specialists asked specifically about a patient's concerns or if they had specific questions they wanted answered, most patients provided no, or minimal, responses. These types of patient responses make it difficult for specialists to establish the patient's agenda when attempts are made. A UK-based interactional GC study highlights that when patients are unable or unwilling to provide an agenda for a consultation when prompted, genetic specialists go ahead and outline the topics and agenda for the consultation since a patient-led agenda becomes interactionally unachievable (Pilnick, 2002). Pilnick (2002) further argues that GC is specialised and complex field, thus although some patients may understand the basics of risk and genetics, most patients do not possess prior expertise or high-level knowledge to know what to discuss or what may be relevant. Patient agenda setting in reality is thus not a straightforward task and minimal patient engagement creates challenges in achieving GC goals. In contrast, however, if *counselling* (Kessler, 1997b; Pilnick, 2002) and *listening* (Clarke, 1994, Clarke et al., 1996) are integral parts of GC, then establishing a patient agenda should occur and finding ways to achieve this needs to be further explored.

Passive patient behaviours create further additional risks through the potential for information to be misunderstood, patients not requesting clarification, or patients finding the information or service unhelpful as the information may not seem personally relevant. Previous international (Meeuwesen et al., 2007; Menendez et al., 2017; Otten et al., 2015) and local (Keikelame & Swartz, 2013; Patel & Dowse, 2015; Penn & Watermeyer, 2018; Watermeyer, 2020; Wessels, 2014) literature suggests that patients, especially those who are marginalised, do not actively engage in medical consultations. Reasons include patients' perceived lack of power and their orientation to healthcare practitioners being in positions of authority (Keikelame & Swartz, 2013; Menendez et al., 2017; Patel & Dowse, 2015), feeling embarrassed to admit misunderstanding (Harmsen, 2003; Otten et al., 2015) or having poor literacy (Patel & Dowse, 2015). Language barriers (Penn & Watermeyer, 2018) may further hamper patient participation in health consultations.

Due to genetic specialists' determination to educate patients, together with the complexities in establishing patients' agendas, the study further notes that tailoring information for individual patients according to their concerns, their needs and their personal backgrounds may be hampered in a local context. The findings in this study support previous studies that GC interactions may be more aligned to biomedical teaching models (Biesecker et al., 2019; Meiser et al., 2008; Skirton, 2018). Although teaching and educating patients on their genetic risk are goals of GC (Biesecker et al., 2019; HPCSA, 2013, 2019; Resta et al., 2006; Veach et al., 2003), the psychosocial goals and the ideal of providing patient-centred care, as promoted through GC's counselling model (Biesecker et al., 2017; 2019; Kessler, 1992; Kessler & Antley, 1982; Redlinger-Grosse et al., 2017), needs to be more strongly prioritised in the South African context.

The tensions observed in this study regarding the task of educating patients, establishing patient agendas, and tailoring information, supports previous interactional GC research. A Canadian study by Babul-Hirji et al. (2010) found that when genetic counsellors foreground their own education-based agendas in GC consultations, they dominated the amount of talk, and consultations occurred in a structured manner that followed a pre-set plan of how the GC consultation should be conducted and what information should be conveyed. Outcomes of these types of GC consultations created missed opportunities to determine the patient's true agenda and there was a lack of flexibility to accommodate the patient's voice. The researchers argue that the volume of complex genetic risk and scientific information provided in education-heavy GC consultations may be intimidating for patients and that their lack of familiarity with the information and dominating behaviour of the genetic counsellors may cause patients to orientate towards passive behaviours, where they provide minimal verbal engagement, and they avoid initiating new topics or asking questions. Alternatively, the same study found that when GC consultations occurred in a less rigid manner where patients were given opportunities to have an open dialogue about topics that concerned them and more attention was given to building rapport and addressing patients' emotions and the psychosocial aspects of the consultation, greater symmetry was achieved in the GC consult and the "human voice" (Babul-Hirji et al., 2010, p.45) of the patient emerged more strongly. Similar results on genetic specialist dominance of talk and greater attention to medical, rather than psychosocial, concerns have been found by a review on communication in genetic consultations by Paul et al (2015). In a study assessing recordings of general practice consultations in the UK (Barry et al., 2001) found that when health practitioners paid attention to the lifeworld of the patient,

patients were seen to discuss their agendas and psychological concerns more openly which resulted in better rapport and patient outcomes. The poorest communication and patient outcomes occurred when patients made attempts to discuss their lifeworld and psychological concerns, but these were either ignored or overruled by the health practitioner's agenda and voice of medicine (Barry et al., 2001). The importance of psychological (Kessler, 1992; Weil, 2000) or relational (Biesecker et al., 2019) GC is thus highlighted.

Although the South African context may lend itself to patients behaving in passive manners due to historical and socio-economic disparities and power dynamics between healthcare practitioners and patients in the broader healthcare system, I argue that the lack of flexibility in genetic specialist agendas, and their structured approaches to risk information giving may further suppress the patient voice in South African GC consultations. Despite local genetic specialists having good intentions to educate patients on risk and genetic information, patients who may already be in vulnerable positions due to language and literacy barriers and socio-economic circumstances, may be further excluded from engaging in GC interactions when overloaded with risk information. In this case, GC then fails at helping individuals and families adapt and make meaning of genetic risk, which at its core is what GC is all about (Biesecker et al., 2019; Joseph et al., 2017). If GC is to be a patient-centred health service that prioritises listening to patients as suggested by Clarke (1994) and Clarke et al. (1996), then understanding patients and prioritising what information is important to them right now, is a critical step in promoting patient care. Much of South Africa's GC training and practice is based on western models and biomedical beliefs. South Africa is a socio-cultural diverse setting where individuals may differ greatly in terms of ideas around risk, health, communication approaches and psychosocial needs. Current South African GC training and practice approaches need to be reconsidered to better align with local patients' needs and context.

The importance of addressing patients' psychological concerns and tailoring information to make GC information personally relevant has been previously highlighted (Austin et al., 2014; Biesecker et al., 2019; Cragun & Zierhut, 2018; Henneman et al., 2008; HPCSA, 2013; 2019; Kessler, 1979; 1997a; 1997b; Redlinger-Grosse et al., 2017; Skirton, 2018; Smets et al., 2007; Veach et al., 2003; 2007). This study further supports the importance of patients' needs and findings suggest that tailored, client-focused practices are not being adequately achieved in South Africa. van Dulmen (2011) describes that when communicating with a patient, it is important to acknowledge that a patient has two needs: "The need to feel known and

understood, as well as the need to know and understand" (p. 381). I argue that paying attention to a patient's context, as well as their emotional and informational needs, and then adjusting GC communication approaches, accordingly, is imperative for achieving true patient care.

The ethic(s) of care theory prioritises mindful and purposive practice that is attuned to the individual needs of patients (Tronto, 2010). These needs may change over time, within different contexts, and with different patients. Providing care within institutions and health practices requires four key phases: Recognition for the need for care, commitment to providing care, attempting to meet that need, and lastly reflecting on the care that has been provided and whether it has been adequate (Fisher & Tronto, 1990). The results of this study have been useful to understand the process of care within the context of risk communication and uncertainty management in South African GC consultations and identify where challenges occur in reality that impedes patient care. Fisher (1990) discusses care as something 'extra' that can be difficult to describe, but that is considered essential in maintaining wellbeing and harmony. Tronto (2010) claims that caring health services incorporate this something 'extra' into their routine practices, rather than the 'extra' being something that only occurs occasionally.

For GC, this 'extra' in care involves promoting trust, active listening and acknowledging a patient's individual concerns as suggested by a PCC approach. Trust is a particularly important part of PCC. A breakdown of trust can occur when a healthcare practitioner does not listen to the patient's concerns or tailor information according to the patient needs, or when the healthcare practitioner is not convinced by a patient's reported symptoms or whether a patient will return to the clinic for further management. If a healthcare practitioner feels uncertain about a patient's reported symptoms, diagnosis, or whether the patient will return to the clinic, they may 'over-treat' the patient by ordering multiple tests during a single consultation to eliminate any room for uncertainty by covering all bases. This can be thought of as a strategy to avoid litigation, as well as a means for healthcare practitioners to enact a 'saving-face' strategy to enhance trust and appear to be paying attention to all possible health-risk scenarios (Fritz & Holton, 2019). Similarly, in South African GC consultations, genetic specialists, in addition to their already pre-planned agenda of conveying all related risk information, may feel uncertain about the accuracy of reported patient information and may be concerned about whether the patient will be able to return to GC for a follow-up consult due to the geographic, social or financial constraints in a local context. Hence, they may convey a large amount of

risk-related information in a single consultation in attempts to manage the uncertainty of the situation. I argue that despite well-intended, this strategy may have the opposite effect where a patient may feel overwhelmed and a breakdown in trust in the relationship may occur as the information is not necessarily tailored to a patient. Although patients should be informed and provided with information, greater symmetry is needed in the agenda-setting between patients and genetic specialists, so that a patient's voice is not only heard and acknowledged but that the patient can also guide what is spoken about in the consultation, rather than the discussion being led only the genetic specialist and their agenda.

By promoting the patients' voice and thus highlighting the voice of the lifeworld (Mishler, 1984), risk information provided to patients in GC consultations may become more personally relevant, patients may better cope with uncertain information and ultimately adjust to the presence of genetic risk or condition in their or their family member's life. A patient's concerns and agenda may be produced more subtly in local GC consultations, therefore requiring active listening and reflexive practice by genetic specialists. Active listening needs to occur not only at the start but throughout the consultation. Genetic specialists also need to reflect during different moments in the consultation on whether the information that they are about to deliver to a patient is aligned with the patient's current context and needs at that moment in time. Flexibility in a genetic specialist's communicative approach for different patients is thus crucial in ensuring patient care, not only at the start of a consultation but also during the consultation depending on what information the patient provides and how they respond to risk information and uncertainty. Attention to the patient's needs and promoting patient participation in GC interactions further allows for opportunities to identify personal and broader contextual factors that may create interactional uncertainty during communication (Brashers, 2007; Gudykunst, 1995). Enhanced trust and communication between patients and genetic specialists will further improve the management of uncertainty in GC consultations, which is another aspect of ensuring care.

A study by Wessels (2014) previously reported that South African GC consultations follow a typical pattern with the way information is presented, both in content and the order of information. These findings suggest that South African GC practice may be more standardised and genetic specialist-led than what theoretical goals and outcomes intended. A script-like approach is encouraged in healthcare, particularly when assessing communication skills and competencies. Research in the fields of pharmacy (Ford et al., 2019) and medical education

(Setyonugroho et al., 2015) highlight the use of rubrics to assess healthcare providers' communication success with patients. However, the use of script-like approaches in health communication and a lack of tailoring for individual patients has been shown to create breakdowns in communication as described in South African audiology consultations (Watermeyer et al., 2020a). Script-like approaches also exist within GC practice and guidelines. Veach et al. (2003) describe the use of a checklist to assist genetic specialists to know what information to cover in a consultation, and that patients may value evidence of a written checklist as it indicates professionalism and thoroughness. Other international GC research have also indicated that some GC consultations are highly standardised (Lobb et al., 2005), and that risk communication checklists (Fransen et al., 2006); standard communication protocols (Bjorvatn et al., 2007) and GC scripts (Codori et al., 2005) are used or suggested.

The details of the risk formats, communication approaches and GC techniques are not elaborated in the local GC HPCSA training and practice guidelines (HPCSA, 2013, 2019). However, based on personal experience as a South African trained and practising GC, together with discussions with my supervisor, who is a GC training programme convenor at UCT (T-M. Wessels, personal communication, 16 October 2020), much of the local GC training and guidelines are based on international GC literature and guidelines (Evans, 2006; Gerard, 2002; Harper, 2010; LeRoy et al., 2010; Ulhmann et al., 2009; Veach et al., 2003; Weil, 2000). These guidelines may subsequently promote the use of structured and script-like approaches to GC, rather than flexibility.

Local GC training is also based on written exams (50%) clinical assessments and continuous evaluation of practical skills (50%) (T-M Wessels, personal communication, December 2020). Continuous observation and evaluation of students at clinics by genetic specialist educators and trainers may cause students to become accustomed to a particular way of providing information to patients and orienting to script-like approaches to provide information the 'right' way. The utilisation of script-like approaches may continue after students have graduated and are practising independently as evidenced in this study's interactional data. A script-like or checklist approach may feel safer and more structured, enabling the young graduate to feel that they are ensuring that all relevant risk information is provided. Stewart (2001) highlights that to provide all information about a healthcare topic is a misunderstanding of what it means to be patient-centred. Rather, patient-centred care involves taking into consideration a patient's

desire for information and adjusting the medical consultation accordingly. Caution is thus necessary when using script-like approaches in local GC consultations.

An additional reason why all risk information may be discussed and why local GC may be more medical rather than psychosocial focused is that much of GC practice in the South African public healthcare system occurs within broader healthcare teams. These teams often oversee collective management of patients at a particular clinic, thus they regularly have patient review and management meetings as witnessed when visiting the research sites. For example, in the prenatal clinics, meetings are typically attended by fetal medicine specialists, sonographers, nurses, medical geneticists, genetic counsellors, students and interns. Each patient's clinical history, management and what information has been individually conveyed to them is reported and discussed amongst the team members at the meetings. Genetic specialists and respective students may thus feel pressure to orientate to the voice of medicine in their GC consultation with a patient, since the healthcare practitioners who are part of the broader clinic team may have a different understanding of what patient care means. Prioritising a patient's psychosocial concerns and agenda above the need to educate a patient about their risks and clinical management may not be encouraged by the broader healthcare team. Findings in this study are supported by previous UK-based research (Pilnick, 2002) that indicate that the "genetic *counselling*" (p. 79) aspect of GC may conflict with routine practice, behaviours and expectations that occur within medical consultations and settings. Genetic counsellors (and other genetic specialists) may thus struggle to adequately address patient's psychosocial concerns and needs in practice as expected in theoretical GC guidelines since their attention is often focused on establishing and conveying risk, delivering a diagnosis and medical management concerns. Broader contextual constraints such as staff shortages, long patient ques, as well as time and space pressure within a busy clinic (which is often the case in South African public health facilities) may be further justifications by broader healthcare teams or institutions to dissuade prolonged attention to a patient's lifeworld stories or psychosocial concerns (Penn & Watermeyer, 2018).

Tailoring information for patients and allowing patients to discuss their psychosocial concerns during individual GC consultations not only takes confidence as a genetic specialist but by extension permission is needed from GC and medical colleagues, training and health institutes and professional bodies that oversee practice. Ogden et al. (2017) similarly highlights that for

PCC to occur, broad system changes and agreement is required across healthcare groups and organisations.

12.2.2 Managing uncertainty in genetic counselling interactions

The findings of this study show that different layers of uncertainty exist in local GC consultations including universal uncertainty, personal uncertainty, and interactional uncertainty. Universal uncertainties related to unclear knowledge about a patient's prognosis or test outcome creates difficulties in GC interactions, particularly when discussing decision-making or questions related to what to expect for the future. Genetic specialists may also experience personal uncertainties when they feel that patients and other healthcare practitioners place expectations on them to provide clear answers to eliminate clinical or genetic uncertainties. The study results support previous literature that highlights the communicative challenges of universal and personal uncertainties in GC consultations. Many of these uncertainties are caused by the complexity of genetic risk information, unclear diagnosis', and challenges in interpreting genetic test results (Arribas-Ayllon & Sarangi, 2014; Clarke & Wallgren-Pettersson, 2019; Hashiloni-Dolev et al., 2019; Lou et al., 2020a; Pilnick & Zayts, 2014; Richardson & Ormond, 2018; Steel et al., 2014; Stivers & Timmermans, 2016).

The study also highlights an additional layer of uncertainty that occurs in GC interactions, namely interactional uncertainty due to contextual factors. Interactional uncertainty in GC has been reported previously (Pilnick & Zayts, 2014; Sarangi & Clarke, 2002a; Thomassen & Sarangi, 2012), but not to the same extent as other types of uncertainty, nor has it been addressed in a South African GC setting. Contextual factors in this study which impact on interactional uncertainty include the inability to provide accurate risk information when patients provide unclear personal or family background information, when patients are guarded in their responses to the types of risk assessment questions, limited patient literacy and genetic specialist challenges in gauging comprehension, as well as language barriers.

Many of these interactional uncertainties seem underpinned by issues of trust and mistrust. Arribas-Ayllon and Sarangi (2014) have focused on how genetic counsellors negotiate the level of trust and mistrust during patients' decision-making in GC interactions to gauge whether a patient has considered all their risk options. The findings in this study further demonstrate that trust is also negotiated in other phases of the GC consultation such as the information gathering phase where patients provide background information that is used to establish risk. In this phase

of the GC consult the study shows that trust becomes an interactional concern either because a patient is unable to provide the information that the genetic specialist requests or the genetic specialist does not trust the information that is provided by the patient. The findings support previous literature (Fritz & Holton, 2019) which indicates that uncertainty may occur because of a health practitioner's mistrust in a patient's reported medical or family information.

The study also shows that trust needs to be established during the information giving phase of the GC consultation. Moments of uncertainty occur when a patient has limited literacy and is unable to comprehend the information provided, even when a genetic specialist tries to use several formats to convey information. In these moments of conveying and attempting to explain risk information, the specialist must trust that the patient will indicate understanding or misunderstanding through verbal or non-verbal confirmations during the interaction. The patient also must feel comfortable enough with the genetic specialist to admit when they misunderstand the information, or when they would prefer an alternative format for the information to be explained in. In this study, patients typically provided little verbal engagement during risk discussions and provision of information, with most of the talking being done by the genetic specialist. This asymmetrical dialogue supports a more traditional form of health communication, where the healthcare practitioner steers the conversation authoritatively and the patient 'follows' with minimal disruption or questioning (Meeuwesen, 2003; Meeuwesen et al., 2007). These passive patient responses and power asymmetries have been described in international (Menendez et al., 2017; Schouten et al., 2007) and local (Watermeyer & Penn, 2009b; Watermeyer et al., 2020a; 2020b; Wessels, 2014) health and GC literature.

For patients to become active participants in GC interactions, genetic specialists must provide interactional spaces that allow for a trusting relationship where patients can openly discuss their personal and family backgrounds and where they feel they can ask questions and clarify information that may not make sense to them. Development of trust in GC consultations is particularly important in contexts such as South Africa where many patients come from impoverished and difficult socio-economic backgrounds, and who may typically orientate to passive healthcare behaviours. When trust is established, it promotes increased patient dialogue in the conversation, which in turn allows genetic specialists to understand patients concerns and tailor information accordingly (Smets et al., 2008; Thain et al., 2019). Listening to patients will also improve the management of interactional uncertainty in local GC consultations.

Genetic specialists need to develop strategies that encourage patient participation when discussing risk and uncertainty related information in GC consultations. A local study by Watermeyer and Penn (2009b), looking at how pharmacists check patients understanding of dosage instructions, suggests that strategies such as using specific questioning, careful monitoring of patients verbal and non-verbal cues, and interactional resources such as response solicitations may assist in encouraging patient engagement in health consultations and understanding of complex information. Similarly, GC literature suggests using teach-back principles when discussing information with patients as a strategy to check understanding and to foster participation in a health conversation (Biesecker et al., 2019; Fagerlin et al., 2011; Joseph et al., 2017). Awareness of cultural nuances and socio-economic status is important in developing strategies since they may impact on communication (Meeuwesen et al., 2007; Watermeyer & Penn, 2009a; 2009b; Williams et al., 2016).

The study shows that language barriers create moments of interactional uncertainty in local GC consultations, and are reported by genetic specialists as a major concern in communicating effectively with patients. In consultations where language barriers exist, trust has to be instilled and negotiated across the whole GC consultation, not only between a genetic specialist and a patient but also with an interpreter when available. Access to professionally trained medical interpreters in GC services in the Western Cape seems to be better than other healthcare facilities or GC services elsewhere in the country. Genetic specialists in the study highlight the value of interpreters at their clinics, not only in relaying information but in allowing the patient's voice to be heard, where it would otherwise not have been due to language limitations. Despite these positives, the study also shows the problems that may arise with the presence of an interpreter such as the interpreter misunderstanding the information that has to be conveyed, and not admitting to the misunderstanding. In addition to in-person interpreting, the specialists in the study also make use of telephonic interpreter services. Although better than having no interpreter, telephonic services may miss some of the non-verbal and atmosphere (Langewitz, 2007) cues that in-person interpreting can detect. The value and scarcity of professionally trained interpreters in the South African healthcare context have been previously reported (Kilian et al, 2015; Ndachi, 2014; Penn & Watermeyer, 2012b; 2014; 2018; Schlemmer & Mash, 2006). The findings in this study suggest that a greater focus is needed to understand the roles of interpreters in local GC services to ensure that these roles are contextually tailored to assist both the patient and the healthcare practitioner. To form trusting relationships and to

better manage interactional uncertainty in GC consultation where language barriers exist, dedicated interpreters are needed at local GC clinics need to be trained in genetic knowledge and GC principles. GC-specific risk information training and understanding the principles of GC practice, for example, non-directiveness, may enhance the trust that patients are receiving appropriate information. I further support suggestions made by Watermeyer (2020) that trusting relationships between healthcare practitioners and interpreters need to be developed that allow interpreters flexibility in their interpreting style and permission to gauge what information needs a patient may have. Establishing good working relationships with trained interpreters in local GC clinics, encouraging flexibility and being in tune with a patient's needs may assist in practising patient-centred care in interpreted GC consultations.

Brashers' (2001; 2003; 2007) uncertainty management theory (UMT) is useful to understand uncertainty in local GC consultations and how it may be of relevance within a framework of care. One of the key aspects of managing uncertainty in healthcare and improving communication is to identify the sources of uncertainty (Babrow & Kline, 2000; Bylund et al., 2012). The study has shown that South African GC consultations have different layers and sources of uncertainty as described above. If GC is concerned with the communication and the relational aspects within consultations (Biesecker et al., 2019; Kessler, 1997a; 1997b; Veach et al., 2003), it should by extension be concerned with managing uncertainty. Genetic specialists and services need to aim to identify those factors that contribute to the presence of uncertainty in a South African setting. Helping patients to become comfortable and to tolerate uncertainty has been previously reported in previous GC studies (Bylund et al., 2012; Hitch et al., 2014; Stivers & Timmermans, 2016).

I extend the arguments made in UMT to suggest that for genetic specialists to assist patients to become comfortable with uncertainty, they first need to become comfortable with uncertainty themselves. Genetic specialists in the study report the necessity to accept uncertainty in GC for what it is and to learn to cope with it. In becoming comfortable themselves, local genetic specialists will be able to provide the interactional space for patients to cope and adjust to the presence of uncertainty in GC consultations, and by extension promote patient-centred care. For genetic specialists, becoming comfortable with uncertainty not only relates to the inherently uncertain nature of genetic risk information but also becoming comfortable with their own practice, particularly about using flexible communication approaches for different patients.

12.2.3 Neutrality in genetic counselling: Tensions between theoretical ideals & the realities of practice

When considering personal risk perceptions and non-directiveness in local GC consultations, this study shows that genetic specialists are, in theory, placed in positions where they must address patients' risk concerns whilst refraining from providing personal views on a particular risk. Evidence from the study shows, however, that a non-directive stance from the genetic specialist may lead to moments of greater uncertainty and trouble within the interaction. A genetic specialist may oscillate between direct and non-direct forms of communication to balance playing a supportive role to the patient, managing their own risk perception, and remaining within the ideological boundaries of their profession. The findings from this study, together with those from previous research (Arribas-Ayllon & Sarangi, 2014; Benkendorf et al., 2001; Biesecker et al., 2019; Clarke, 1997; 2017; Pilnick, 2008; Pilnick & Zayts, 2012; Sarangi, 2010; Weil et al., 2006; Wessels & Koole, 2019; Wessels et al., 2015; Wyatt, 2001) suggest that GC practice is more directive than what is suggested in theory.

Some of the consultations in this study showed more implicit forms of direct communication, whilst other consultations revealed more explicit displays of direct communication when genetic specialists discuss the offer of genetic testing. Many of the interactional trouble occurred in the prenatal consultations when patients were offered an amniocentesis. In instances where patients directly asked for advice on their test decision, advice-seeking created particularly heightened levels of interactional trouble. In these instances, GC principles such as remaining neutral, become juxtaposed against patients' needs with assistance with their decision-making. Complexities in prenatal decision-making and neutrally providing information have been previously reported in the local (Wessels & Koole, 2019; Wessels et al., 2015) and international genetic counselling literature (Biesecker et al., 2019; Pilnick & Zayts, 2012). Even though a specialist may assume that they are remaining neutral, analysis of the interactions from this study suggests otherwise. Genetic specialists in this study do seem to be aware of the difficulty to remain neutral however and report that they try to be mindful of how they frame risk information. However, how to manage neutrality in actual practice remains a challenge as is evident in the interactional data.

Genetic specialists may also feel obliged to offer invasive testing to patients who are referred for GC as seen in the cancer and prenatal data of this study. Part of genetic specialists'

obligation to offer testing is informing patients of their options in risky situations but may also be done to offer the patient some sort of management option. When patients decline the offer of a test, genetic specialists may subsequently struggle to remain within the boundaries of their profession and may act in directive ways to encourage the uptake of a test as evidenced in the study. In saying this, however, one consultation from the study demonstrates that the uptake of a repeat genetic test may also not be what a genetic specialist wants, especially when a genetic specialist's perception of risk is misaligned with a patient's and the specialist believes that a genetic test is not warranted. This interactional finding shows the power of risk perceptions, and how genetic specialists may struggle to accept patients' risk perceptions when they misalign with their own. However, this case seemed to be unique when compared to the other data in this study, as well as other local GC research that suggests that genetic specialists prefer patients to accept genetic testing offers (Wessels et al., 2015; Wessels & Koole, 2019).

Although well-received in westernised communities, non-directiveness may not be an effective or appropriate approach for individuals from all cultures or social backgrounds. GC guidelines from the Global North may assume that patients are aware that they have autonomy, and that patients and healthcare practitioners come from homogenised backgrounds in terms of socioeconomic status, education, culture, language, and ethnicity. In countries with high rates of poverty, illiteracy and inequality, and where overarching healthcare systems do not allow for patients to be fully in charge of their healthcare management or referrals, non-directive approaches may be poorly received and confusing. Findings in the study also suggest that some patients in the local health system may also have differing views on whether to act upon a risk, or not. Some patients may rather leave the outcome of an uncertain situation to fate or in 'God's hands'. This passive view may be different to western ideas of risk assessment and management.

A patient's desire to be self-directive, or else to rely on a healthcare practitioner's opinion during decision-making, is thought to be based on individuality and cultural differences (Meeuwesen et al., 2007; van der Steen et al., 2019; Yip, 2020). Although some patients may value being offered a choice, they may want the healthcare practitioner to choose for them (Barnett et al., 2008; Ford et al., 2003; Toerien et al., 2018). Patient choice and the offer thereof are thus far more multi-dimensional and interactionally complex than what is suggested by policies and practise guidelines.

Perhaps one of the fundamental problems with the notions of directiveness and non-directiveness is that there is no consensus on definitions for the two terms, despite the terms having been used for several decades (Arribas-Ayllon & Sarangi, 2014; Kessler, 1992; van der Steen et al., 2019). Kessler (1992; 1997a) has argued that being non-direct is not the absence of persuasion, nor the opposite of being direct. Rather non-directiveness is a task of 'engaged neutrality' where counselling skills are used to ensure patient autonomy and self-actualisation. Thus, complete passive behaviour or avoidance of being direct by a healthcare practitioner is not only unhelpful but is a misunderstanding of the concept of non-directiveness. The term non-directiveness has thus become somewhat distorted. The findings in this study not only show moments of direct communication and genetic specialist preferences of how the patient should decide but that when asked for advice, genetic specialists revert to completely passive behaviours, which are also unhelpful to a patient.

Rather than focusing on the negative aspects of advice-giving and how GC does not promote this activity, Arribas-Ayllon and Sarangi (2014) suggest that non-directiveness should be seen in a more positive lens where the genetic specialists' potential to influence patient outcomes is acknowledged. Weil (2003) further suggests that the notion of non-directiveness should be strived for rather than seen as an absolute goal. The inability to achieve complete non-directiveness relates to issues of "inadvertent directiveness" and "inevitable directiveness" (Weil, 2003, p. 202). The former relates to a genetic specialist's inability to completely mask their personal values (and risk perceptions), and the latter relates to how the choice of what information to include and the format used to communicate this information can subtly influence a patient's perception and decision-making.

Genetic specialists need to acknowledge that their communication with a patient may have inescapably direct interactional moments. The study findings show that it is impossible to be completely neutral. Even in GC consultations where a genetic specialist may seem to be aligning with a patient's choice, a closer look at the interactional details reveals instead that genetic specialists may not be aligning with the patient but rather subtly steering the patient towards a particular decision. Despite an inability to be neutral in their communication, I believe that genetic specialists, when attuned to a particular patient's needs and context, can align and tailor their risk information giving to ensure that the information, and how it is conveyed, remains patient-centred. This idea has been raised by other researchers, that genetic specialists may need to adopt different levels of non-directiveness in response to patients'

individual needs (Bernhardt, 1997; van der Steen et al., 2019). Complete neutrality in GC may negatively impact on patient-centred care since it limits the counsellor's ability to fully engage with the patient (Benkendorf et al., 2001; Weil et al., 2006).

An interactional study by Benkendorf et al. (2001) shows that when genetic specialists revert to neutral positions after using more direct forms of communication within the same GC consultation, and when genetic specialists use indirect forms of speech, e.g., describing what 'they' or 'other people' do rather than referring to the actual patient, it creates patient confusion, and missed opportunities to explore individual patient needs. Wyatt (2001) further argues that withholding personal risk perceptions or opinions may cause distrust and affect the patient-counsellor relationship, especially when a patient seeks advice. Being non-directive in situations where patients are highly anxious (e.g., during discussions on abortion and procreation), a health practitioner's (including genetic specialists) decision to avoid disclosing their own risk perceptions could be viewed as uncaring, since patients often seek guidance from healthcare practitioners, not just facts. Local and international studies show that some individuals do not feel comfortable making autonomous decisions and genetic specialists may be more direct in their risk communication style with individuals whom they feel cannot make independent decisions (Eichmeyer et al., 2005; Pilnick & Zayts 2012; Rantanen et al., 2008; Yelei et al., 2013; Wessels et al., 2015). These studies, as well as findings from this research, indicate that levels of neutrality must be considered when assisting patients to make meaning of risk and how it may impact their life.

12.3 Reflecting on the research project

In parallel with the complexities of my topic of focus, this project proved far more complex than I anticipated. As a researcher and a genetic counsellor, I found myself grappling with several ethical dilemmas during the research process, influenced by my unique position as a researcher investigating my own profession, my choice of methods, as well as the setting of the study. The following section focuses on the risk and uncertainties that occurred as a result of the project itself. Although perhaps considered 'additional findings', I thought it important to return to the idea of wearing 'different hats' during this project, particularly that of researcher and a South African trained and practising genetic counsellor. The challenges of undertaking a study that utilised video-recordings within a sensitive and contextually challenging healthcare

setting also needs to be addressed. The sections below expand on these ethical dilemmas, drawing substantially from my published article (Scott et al., 2020).

Although initially seeing the position of researching my professional field as advantageous, I soon realised the challenges thereof, with prior experience not necessarily making me better equipped for this study. Despite efforts to remain neutral, researchers are undeniably influenced by their prior experiences in the field, and their perceptions of their ability to cope with data collection (Bahn & Weatherill, 2013; Boden et al., 2015; Jackson et al., 2013; Mattingly, 2005). Having an existing relationship with the staff at the clinic was thought useful, as it would facilitate the research process, and allow for the development of trust and rapport (Liu et al., 2015). Whilst this was helpful, it also created larger unanticipated ethical challenges too. Aware of potential coercion and power dynamics has been important during the project (Fisher, 2013). Although not an immediate colleague or in a senior position to the recruited GC participants, one of my research supervisors is, which may have unduly influenced individuals' decisions to participate. Furthermore, potential GC participants may have felt obligated to participate, given the limited number of genetic specialist participants available for the study. This dilemma has been previously reported when conducting research in small fields (Ford & Reutter, 1990). The voluntary nature of the study had to thus be emphasised, and I had to try to remain neutral during the recruitment phase of the research. As a genetic counsellor invested in the well-being of a very small South African GC community, I have also had to be aware of my personal biases during the data analysis. Although some positive aspects are noted, my study reveals some communicative variables that are not in keeping with the theoretical ideals of GC. My study results thus challenge current local practice. Reporting on these findings made me worry about the possible impact this would have on my personal and professional relationships in the local GC field. Tietze (2012) has described problems of objectivity due to the proximity of the researcher-researched relationship, and how insider knowledge and continued connections after a project has ended can create ethical complexities. The overall experience has brought about an important lesson: Researching one's own field and practice is far more difficult than assumed. However, being an 'insider' may assist in providing feedback to my field and encourage change within the practice and student training.

The presence of a video-recorder can raise different concerns for research participants. For patients, their personal information and stories are captured in minute detail, creating the danger of exploitation; whilst for healthcare practitioners, their actions and skills are exposed

and recorded, making them vulnerable to professional scrutiny (Evans, 2010; Høstgaard & Bertelsen, 2012; Iedema et al., 2006; Parry et al., 2016). However, recordings may also be valued by participants, as they may support their potential to improve services as described in a palliative care study by Hargreaves and Peppiatt (2001). During this study, although the majority of invited genetic specialists agreed to participate in the recorded consultations, I sensed a level of apprehension when approaching some individuals during participant recruitment. One genetic specialist declined to participate stating that the presence of a video-recorder would make them feel stressed, which would then affect their attention and quality of service towards the patient. They also declined to be interviewed. Another genetic specialist agreed to be interviewed but declined to have their consultations with patients' video-recorded stating their anxiety about having their communication skills assessed. One genetic specialist who agreed to participate also became visibly flustered and started to laugh nervously mid-consultation. They justified their behaviour to the patient by pointing at the video-recorder and stating how its presence made them feel uncomfortable. Patient participants seemed less concerned about having their consultation video-recorded with several providing altruistic reasons for participation and assisting in service improvement. Perhaps this broader positive response to participation in the study is due to an awareness that the GC clinics are situated within public hospitals where tertiary teaching and research frequently occurs. Acceptance that scrutiny of practice is part of the services offered in such a context may be reasons for patient participation in this study. It is, however, important to note the risk of coercion and power imbalances in a South African context, especially for patients considered vulnerable due to their psychological/physical well-being, their poor socio-economic context, or who view healthcare practitioners in high regard (Fisher, 2013; Penn & Watermeyer, 2018; Pino et al., 2017; Wessels, 2014). These factors may make patients in the public South African healthcare system less likely to decline participation in research.

Another ethical challenge is the issue of anonymity during video-based research, especially for genetic specialist participants. Professionals within the South African clinical genetics field all tend to know each other, even those that work at different public and private facilities. The difficulty of ensuring anonymity of individuals and organisations during video-based research (Heath et al., 2010; Parry et al., 2016) medical genetics studies (Bickford & Nisker, 2015) and in small specialized local medical fields (Etheredge, 2015) has been previously described. This dilemma created tension in the research process in trying to balance the ethical considerations of anonymity against the expectations and nature of thick descriptions required in qualitative

research. During the data collection and analysis phases, anonymising the participants from the research team was impossible. Being able to clearly visualise the verbal and non-verbal features of interaction are vital for researchers in health communication to understand communicative phenomena (Parry et al., 2016). How best to manage this concern in this study is challenging. The study does not aim to compare communication styles between e.g., ages, genders, professional groups or clinical sites; thus, this particular information is not considered necessary and has been kept deliberately vague. I use the broad label of 'genetic specialist' rather than stating exact professions and roles and anonymised all participant information. The language spoken during the interaction and the level of genetic specialists' professional experience seemed important to report on in some instances however given the possible effect on genetic risk and uncertainty health communication. The level of professional experience is only reported on if significant to the interactional features in the consultation. For publications and presentations, I do not show any video-recorded data, but rather use written and anonymised transcripts. Transcripts provide a transparent reflection of the consultations detailing both the verbal and non-verbal phenomena without the need to refer to the raw data (Heath et al., 2010; Heritage & Maynard, 2006; Penn & Watermeyer, 2018).

The last point of discussion concerns the ethical challenges surrounding informed consent and the collection of sensitive data. Sensitive data can be described as private experiences, or data in which there is a threat (emotionally or physically) either to the participant, or the researcher (Cowles, 1988; Lee, 1993). This harm may occur during data collection or post hoc activities such as analysis and transcription (Dickson-Swift et al., 2009; Kiyimba & O'Reilly, 2016). Thus, ethical decisions do not only occur during data collection but also need to be reflected on once the data has been collected. For the study, further critical reflection was required especially for recorded cases in which unexpected intense emotion or overtly sensitive data was disclosed. Participants in hindsight, may not have wanted to participate had they been fully aware of the sensitive information that was going to be discussed in their GC consultations. Retrospective consent would have been helpful for participants in this study, especially for patient-participants. However, I had not collected contact details from the patient participants thus was unable to re-contact them. Consideration of retrospective consent for participants is something that I have become acutely aware of during this project, and something that I would recommend for future studies, especially those video-recording naturally occurring clinic activities or sensitive health topics. It is also important to discuss the potential for distress and secondary trauma, not only to the research participants but also to the research team when

viewing video-recorded data. Distress can occur during data collection or post hoc activities such as transcription and analysis. Although assumed an unemotional, mundane task, transcription, and analysis of data (either first-hand or via secondary analysis) can invoke negative emotional responses (Dickson-Swift et al., 2009; Kiyimba & O'Reilly, 2016; Lapadat & Lindsay, 1999). The video-recorded consultations in this study captured sensitive contextual and personal stories. These included accounts of domestic violence and assault, infant death, poverty, unemployment, alcohol and drug abuse, depression, loss and grief, as well as failures of support by local healthcare services and larger social systems. Furthermore, intense patient participant emotions, the devastation of receiving a life-changing diagnosis or risk information, and the anxiety around making a difficult decision under uncertainty was challenging to view. These emotions, together with factors that created challenges in the communication process such as literacy and language barriers elicited reactions such as sadness, anger, frustration and anxiety for myself, my supervisors, and the research assistants. Regular debriefing sessions and self-reflection (Fitzpatrick & Olson, 2015; Shaw et al., 2019) were done amongst the research team to offer support, and to help make sense of the emotions felt. Collectively, these experiences during the data collection and analysis highlight the challenges and risks of doing video-based research focused on healthcare services that contain complex and sensitive patient-health practitioner discussions.

12.4 Conclusion

In summary, the findings from this study suggest that risk communication and managing uncertainty in South African GC consultations is inherently difficult. The challenges in addressing these activities in GC impact care and patient-focused outcomes. More research is needed into understanding the impact of local GC risk communication and uncertainty on patients, and how the task of communicating complex risk information can be made more comfortable and manageable for genetic specialists. Tailoring information, paying attention to patient agendas and context, and learning to become comfortable with the presence of uncertainty and being flexible in communication approaches are some of the ways to improve communication and care in a local setting. The study also shows the power of interactional research that focuses on everyday communication practices. Caution is however needed when studying health interactions focused on sensitive topics, especially when video-recording routine clinical practice, since data collection and data analysis may involve unanticipated ethical and emotional dilemmas that must be navigated.

CHAPTER 13: CONCLUSION

13.1 Introduction

The aim of the study was to explore how genetic risk is communicated and how uncertainty is managed in genetic counselling consultations in a public healthcare context in South Africa, particularly in the Western Cape province. The research is also so much more. It addresses issues of care, directiveness, neutrality, patient-centredness and the tensions of theoretical ideals of GC and the realities of practice. It also highlights the complexity, yet critical value, of interactional research and the importance of focusing on GC process, not only theories and outcomes. The following chapter discusses the implications of the study findings, training and practice recommendations, methodological and research considerations, as well as the study's strengths and limitations.

13.2 Implications

The ideals of GC are not easily achieved in reality and some of the guidelines may impede, rather than facilitate patient-centred care. My experience of personally being exposed to genetic risk has further confirmed the complexity of genetic risk and uncertainty communication. Despite our attempts as counsellors to be objective and impartial during medical consultations, we remain human, and the emotive side of medicine plays a large part in clinical encounters. Emotions and motivations drive much of our relationships, our decision-making and how we communicate with others. Here, ethical notions of *'what is good care?'* become important. Care for one patient may look different to care for another patient. Reflecting on, and researching, the communication and interactive processes of GC interactions remains critical to understanding some of the complexities and finding ways to make this difficult task a little easier for both patients and genetic specialists. Being a genetic counsellor myself and having been a patient, thus seeing both sides of this research topic, has made this project even more imperative and meaningful to me.

Medical genetic knowledge and testing is becoming increasingly accessible and utilised by other health professionals and the uniqueness and value of GC has thus come under review (Austin, 2010; Cragun & Zierhut, 2018; Hooker et al., 2017; McAllister et al., 2016; Resta et al., 2019; Smets et al., 2007). To answer the internal and external calls for GC to prove its worth as a profession, I propose that GC's power as a profession lies in its *equal* prioritisation of a patient's psychological needs to that of educating. Even though PCC and care are

considered a universal medical priority, medical genetics is a unique medical area that requires even greater attention to patient concerns. Decision-making relating to medical genetic risk can be complex, morally contentious, emotionally charged and has consequences for several intergenerational family members, rather than a single individual. GC provides an appropriate and suitable platform to navigate these conversations and decisions due to its clear prioritisation of psychosocial and lifeworld concerns within its practice and training guidelines and frameworks. The missing part however it how to ensure that these psychosocial patient concerns and agendas are continuously prioritised and achieved in practice and how to make them contextually attuned.

Existing GC guidelines proposed several years ago are coming increasingly under review, especially with the expansion of GC services into new healthcare areas and socio-cultural diverse settings. Due to the differing needs and backgrounds of patients, having only one universal GC model or approach is unrealistic (Biesecker et al., 2019). Lewis (2002) has further called for empirical research for GC in culturally diverse settings to guide locally relevant models of practice, noting that these models may differ to the traditionally implemented models used in western contexts. Process-related empirical research in non-western settings remains scarce nearly 20 years on from Lewis' call, with only a handful of researchers focusing on this area of research to my knowledge (Pilnick & Zayts, 2012; 2014; 2019; Wessels, 2014; Wessels et al., 2015; Wessels & Koole, 2019). Advances in GC guidelines need to allow flexibility to adapt to the patient, the information and the setting in which GC occurs. Conducting research in various contexts that focus on GC process is therefore imperative to gain a broad and rich understanding of GC practice to guide and enhance GC theoretical frameworks. This is a critical step in ensuring care in modern GC practice, and a step this study has contributed towards through its research findings. The interactional and 'real-time' nature of this study provides powerful insight into what some of the everyday GC communicative challenges are in a South African setting, and how we (as practitioners and researchers) can perhaps alleviate some of the theoretical and practical tensions identified.

13.3 Training & practice recommendations

13.3.1 Tailoring information & prioritising the patient's agenda

Evidence from this study suggests that despite there being several sources and formats to assess and convey risk information in local GC consultations, providing large volumes of risk

information and using specialist-led educational agendas are problematic. These approaches are not in keeping with a patient-centred approach. Care in a GC sense may involve providing context-specific, tailored information rather than providing all-risk related genetic risk information stringently.

To offer care, a genetic specialist needs to allow the consultation and patient agenda to flow naturally, rather than impeding it through stringent, script-like boundaries. This skill may include less talking, more listening, intuition, flexibility, reflexivity in the consultation as moment-by-moment activities or topics occurs, and the confidence to modify the information towards the needs of a particular patient. Genetic specialists further have to be able to read the atmosphere (Langewitz, 2007) of a consultation to decide what information and concerns are important right now for a particular patient in a particular context. This ability has been described as 'intuitive expertise' (Gobet & Chassy, 2008) or the 'hidden curriculum' (Browning et al., 2007; Inui, 2003; Watermeyer et al., 2020a).

Tailoring information requires advanced communication skills that are thought to come with maturity and experience. Senior genetic specialists may have more confidence in tailoring information for individual patients and being more flexible in the way they provide GC, as evidenced by quotes from three senior genetic specialists in this study. Younger or inexperienced genetic specialists alternatively, may automatically orientate to teaching and education-based GC models or goals that pay attention to content and checklists. Checklists may provide a safety harness that can be clung onto and that ensure predictable and anticipated conversations. Focusing on patients' psychological concerns or contextual needs may bring about difficult topics and intense emotions that a younger or inexperienced counsellor may not feel equipped to deal with.

By encouraging information tailoring, I am not suggesting that patient education or informing patients of their risks should be abandoned. Providing risk information and educating a patient should always remain an integral part of GC. However, I argue that for information to be meaningful and for GC services to be of value, genetic specialists must meet patients halfway when establishing agendas for and during the consultation. Patients need to feel heard and supported, and specialists need to have the insight to know what risk information is imperative to a patient in a particular moment, and what may not be.

Skills such as tailoring information and being flexible are not as easily taught, and although a student may be aware of the need to do so, they may not know how to do so. One cannot expect new and inexperienced students to know what to talk about or how to tailor information with no foundational guidelines or suggestions on how to approach a GC consultation. Thus, although students may be taught a 'standardised' way of doing things in a typical GC consult, flexibility rather than stringent frameworks need to be emphasised to students. Flexibility in GC consultations needs to be actively taught, mentored and demonstrated during GC training so that students not only learn how to be flexible, but they see the value of tailoring information for different patients in different settings. Listening skills and thinking carefully about the patient's point-of-view and understanding what they want from a GC consultation is also important. If, for example, their main goal for attending a consultation is to receive their test results, then this part of the GC consultation should be addressed at the start of the consultation, rather than at the end; then additional risk information or discussions on other related topics such as diagnosis or management options can be pursued.

Communication skills need to be taught actively and explicitly in curricula, and not seen as soft skills that will be picked up vicariously. Teaching and learning medical and scientific-based information are often prioritised over communication skills training in medical professions. Recent international calls have been made for further research and prioritisation of communication skills training for professionals who provide GC (Medendorp et al., 2021). Although counselling skills and attention to psychosocial needs are taught in local GC training and encouraged in practice (HPCSA, 2013; 2019) greater prioritisation is necessary. Evidence from the HCRU in areas of emergency medicine (Penn et al., 2017), audiology (Watermeyer et al., 2020a), as well as TB and HIV (Penn & Evans, 2009; 2010; Scott et al, 2019) highlights the value and success of communication skills training and workshops for healthcare practitioners and students. Although local genetic specialists receive counselling skills training, I believe that these can be modified and enhanced through additional communication skills training and workshops. Content of the workshops or curricula could include showing video-recorded interactional examples taken from actual consultations where communication breakdown occurs and finding ways to improve it. Roleplay learning and student video-reflexivity are already used in local GC curriculums as a way of teaching students about their own practice (T-M. Wessels, personal communication, November 2020), however, these learning opportunities could be further enhanced by deploying CA and TODA principles to analyse the interactions and the communication processes that occur within them even further.

These interactional methods may highlight the complexities in seemingly taken for granted communicative moments in everyday practice. Lessons learnt from and during real everyday messy healthcare encounters provide better insight into a patient's needs rather than relying solely on healthcare practitioner's well-intended, yet sometimes poorly considered, theoretical or classroom-based thoughts of what is important or relevant in a patient interaction (Browning et al., 2007; Gobet & Chassy, 2008; Watermeyer et al., 2020a). International and local researchers have shown the value of interactional methods in communication training for police crisis negotiators in the UK (Sikveland & Stokoe, 2020); secondary school teaching practices in Norway (Sikveland et al., 2021); South African emergency call operators (Penn et al., 2017); and UK medical professionals communication skills training (Pilnick et al., 2018). Researchers in Hong Kong (Zayts-Spence et al., 2021) have also made calls for using transcripts from authentic real-time GC consultations for GC communication training purposes, especially for GC practitioners that serve socio-cultural diverse patient populations. I further advocate for the incorporation of interactional methods in local GC communication skills training as they have important implications for developing South African based GC guidelines and practices.

These suggested changes will require broader institutional adjustments, including buy-in from GC training institutes, genetic specialists in practice, broader healthcare teams, as well as professional bodies and committees who oversee GC practice. These changes and deliberations are critical to establishing patient-centred approaches and caring institutions.

13.3.2 The importance of follow-up genetic counselling consultations

A study by Penn (2015), focusing on discussions of HIV status disclosure in a paediatric clinic between a doctor, a counsellor and a grandmother of an HIV positive adolescent child, describes the heaviness of the discussion in terms of information, emotions, responsibility, and decision-making. This heaviness is felt by all participants in the interaction when discussing a difficult topic that impacts an individual's health, psychological wellbeing, relationships with others, and social adjustments. The participants aptly sum up their discussion as "too much for one day" (Penn, 2015, p. 578) acknowledging the challenge of dealing with such a complex topic and decision and the weight of the information conveyed. Similarly, GC consultations often too have a sense of heaviness in terms of the sensitive topics discussed, the volume and complexity of risk information imparted, the responsibility of making difficult decisions in the context of uncertainty, and the potential for intense patient emotions to arise.

I propose that the notion of GC follow-up consultations be considered more strongly. Rather than conveying large amounts of risk information in a single consultation or trying to address all concerns, genetic specialists need to pay attention to what information/concerns require direct attention and what information/concerns may be more suited to be expanded on at a later follow-up GC consultation. Follow-up consults will give genetic specialists the ability to spread the volume and load of GC informational, decision-making and psychosocial focuses across sessions which may aid in dealing with the heaviness of GC. Additionally, a patient's informational and the psychosocial needs may change over time, and follow-up GC consultations would make ideal opportunities to address these needs.

It is thus important for genetic specialists to welcome patients to contact them and return to GC by providing the relevant clinic or genetic specialists' contact details. Genetic specialists could also use their discretion and directly arrange a follow-up appointment at the end of an initial GC consult if they strongly feel that a patient may benefit from further consultation and do not want to wait for the patient to contact them. Although genetic specialists may feel concerned for the potential that a patient may not contact the genetic clinic or genetic specialist after their initial GC consult, and thus may not have received all the necessary risk information at their initial GC consult, if a patient feels heard and care for and their personal concerns are addressed at an initial consultation, they will then be more likely to return to the clinic. This idea is supported by findings by Berg (2003) from a community Parent-Infant Mental Health Clinic based in an impoverished area of Cape Town. Berg (2003) argues that although the clinic in her study is not necessarily able to offer significant health or social interventions, many patients return to the clinic as the healthcare practitioners make a point of paying attention to the patients' lifeworld concerns and patients feel heard and cared for. In the same way, I believe that GC can offer opportunities for patient's facing genetic-related risks and concerns to feel heard, which may in turn facilitate improved communication and the establishment of valuable and respectful relationships between patients and genetic specialists at genetic clinics.

Although contextual factors such as geographic, social, and financial constraints may place pressure on some patients as to whether they will be able to return for a follow-up GC consultation or not, many patients with genetic conditions e.g., paediatric and adult patients, need long-term health management and care at various clinics. Most of these clinics are available at the same tertiary public hospitals where the GC clinics are based. Hence, there are existing opportunities for genetic specialists to coordinate with a particular patient and the staff

at other specialist clinics to plan for these patients to be seen again. GC appointments could be arranged on the same day as a patient's other clinic appointments to avoid multiple hospital visits. Buy-in and joint consideration from the genetic clinics, other specialist clinics and broader hospital management will however be critical for the success of follow-up GC consultations since limited clinic space and genetic specialist staff availability at the three hospitals will have to be carefully managed.

Regarding pregnant patients, although certain genetic tests are only available during particular gestational periods, these patients may also have additional informational or psychosocial needs that can be addressed at a follow-up GC consultation. Many high-risk pregnant women would typically continue regular prenatal check-ups at their referred tertiary level hospital where GC clinics are based, hence returning to the GC clinic would be accessible. Although some pregnant women, such as AMA-referred pregnant women who have no additional identified risks, may return to their primary or secondary level clinics after their initial GC consult or fetal assessment, providing opportunities for these women to return to GC would also be important. If face-to-face follow-up consultations are not feasible for these (or other) patients, alternative GC platforms such as tele-counselling could be arranged. Recent developments in this regard, because of the global COVID-19 pandemic and the South African national lockdown, have shown that tele-counselling may be an option for local patients, although the value of tele-counselling has not been fully explored in a local context.

13.3.3 Development of contextually attuned South African genetic counselling models & guidelines

Redlinger-Grosse et al. (2017, p.1386) highlights that existing GC curriculums and training institutions need to "make room" for the inclusion of new ideas and evidence-based research suggestions. This study contributes to this important area of context-specific, empirical research that highlights *process* rather than only outcome. These findings may be of importance for South African GC training and practice.

The study supports the argument made by Biesecker et al. (2019) that a singular GC definition and practice model is not recommended, since GC goals differ according to individual patient needs, as well as context. Findings from this study suggest that locally relevant GC models and guidelines are needed and that these may look different from the GC models and guidelines suggested in other global settings. The local model would need to take into consideration

contextual factors such as patient lifeworld views, personal beliefs, language mismatches, literacy challenges, a lack of patient awareness of GC services, as well as passive patient behaviours, which this study has drawn attention to. Results from the study suggest that South African patients withdraw during conversations when information is not tailored to their contextual needs or when they do not feel heard.

To assist patients to navigate the complex risk information presented to them in GC interactions, and to truly offer care, genetic specialists are required to have good counselling and listening skills and are encouraged to establish active and trusting genetic specialist-patient relationships during a consultation (Austin et al., 2014; Biesecker et al., 2019; Redlinger-Grosse et al., 2017; Skirton, 2018). Part of establishing an active genetic specialist-patient relationship should be: What do patients *want* to hear? Rather than assuming what the patient *should* hear. A paper by Steel et al. (2014), focusing on communication patterns in Australian paediatric genetic consultations, has indicated that getting medical geneticists to ask parents at the start of a consultation "what would you like to get out of the appointment?", rather than indicating their own agenda and stating 'is that ok with you?', is far more beneficial in ensuring patient participation during the consultation, and by extension allowing for a more patient-focused session (p. 2832). I agree with these communication strategies mentioned by Steel et al. (2014) to encourage patient dialogue and establish the patient' agenda. However, in the South African context, where many patients may not know much about GC or why they have been referred, I would suggest using an alternative question such as: "What are your concerns?" rather than "What would you like to get out of the appointment?", as many patients may not know how to answer the latter. This type of question may also assist in developing rapport in the genetic specialist-patient relationship that promotes trust, respect and mutual acknowledgement.

Using a multimodal approach to information giving is also recommended. This approach could include not only spoken information, but also written or graphical information that could be utilised by the patient at a later stage. For example, should there be too much information to explain to a patient in a short period of time, or should a patient become overwhelmed or distressed during a GC consult, a short information sheet, letter or diagram can be provided by the genetic specialist for the patient to take home and use at a later stage. This multimodal approach could also serve as an opportunity to share information with partners and family

members. It is important however that this additional information is useful and tailored to individual patient need. Thus sensitivity to language, literacy and comprehension is essential.

Guidelines and training for interpreters at local GC clinics are imperative to consider in the future. Becoming aware of genetic knowledge, GC goals and communicative practices would be important so that interpreters are better prepared before patient consultations, and genetic specialists feel comfortable in their abilities to convey genetic risk information. GC training for interpreters would allow for a more trusting working relationship with genetic specialists, which subsequently would result in improved patient care. Treating interpreters as equal partners in the communication relationship and permitting them to use flexible interpreting styles to get health messages across and to be patient advocates is also thought important in improving patient care as suggested by Watermeyer (2020). Although using a limited sample within a small field, this study adds to the continued discussion on the need for professionally trained interpreters within South African healthcare facilities and adds to the corpus of local interpreter knowledge by providing real-time evidence of language barriers and interpreter practice.

13.3.4 Accepting the presence of uncertainty in genetic counselling

Genetic specialists need to accept the presence of uncertainty in GC consultations and need to be prepared to navigate complex communicative moments with patients. Similar calls have been made for the acceptance and management of uncertainty in GC, rather than attempting to reduce it (Armstrong, 2018; Barlow-Stewart, 2018; Newson et al., 2016; Thomassen Hammerstad et al., 2020). Acceptance of uncertainty in clinical practice, and being comfortable with the limits of knowledge, may lead to more caring relationships, especially when communicating with patients who remain without a diagnosis. By accepting one's own uncertainty, healthcare practitioners may assist patients to become comfortable with their own feelings towards uncertainty (Rogers & Walker, 2016).

Uncertainty management training for genetic counsellors (and by extension other genetic specialists), as suggested by Clarke and Wallgren-Pettersson (2019), Newson et al. (2016) and Simpkin and Schwartzstein (2016) may be an important step in ensuring effective communication in GC consultations. Training needs to involve identification of the medical, personal and social (Brashers, 2003) sources of uncertainty in South African GC consultations and how this may impact communication and patient outcomes. Knowing what factors

contribute to uncertainty, genetic specialists may be able to better anticipate the complexities of communicating uncertainty in GC consultations. Having honest conversations with patients and admitting uncertainty is also important. Although professionalism and expertise enhance trust, authenticity and allowing patients to see the 'imperfect and *not* all-knowing' side of a genetic specialist may also enhance trust.

Uncertainty training will assist genetic specialists to feel more comfortable with the presence of uncertainty and give them the confidence to have candid conversations with patients. Being more comfortable with uncertainty may also give genetic specialists the confidence to be more flexible in their communication with a patient, rather than relying on a structured checklist. Previous researchers suggest that for healthcare practitioners to become more comfortable with uncertainty, the deep-rooted idea of uncertainty elimination in medicine needs to be reconsidered, not only in practice but also in training of students. For change to occur within healthcare institutions, health curricula need to acknowledge that some situations may have more than one answer, that human health is not necessarily categorised into neat linear compartments, and that in some health instances certainty is not the end goal (Armstrong, 2018; Simpkin & Schwartzstein, 2016). In GC training and practice the acknowledgement of uncertainty seems particularly important. Introducing philosophical teachings of uncertainty in healthcare student curriculums may assist students to understand what creates uncertainty in healthcare, and why healthcare practitioners and patients may respond to uncertain situations in different ways (Tonelli & Upshur, 2019). I believe that these types of teachings would be useful in GC curriculums and may assist in creating more meaningful, and patient-centred communication during uncertain situations and decision-making.

13.3.5 The issue of non-directiveness

The study has highlighted the value of non-directiveness in theory, but also the difficulties of achieving non-directiveness in practice. Much of this challenge is around managing uncertainty and risk perceptions. Whilst fully agreeing that patients' experiences, values, and autonomy should always be respected, I argue whether remaining completely objective as a genetic specialist is helpful to a patient, or even possible. This study supports Kessler's (1992) previous claim that non-directiveness is contextually situated. How information is provided and perceived is dependent on who is providing the information, whom the information is provided to, what their concern is and the setting in which the consultation occurs. The level of direct and non-direct speech in GC consultations is thus not static but rather occurs along a continuum

that is shaped by a specific context. Claiming that specialists practice complete objectivity and leave their personal risk perceptions at the door when addressing patients also seems unrealistic.

Even though withholding information and making recommendations can be considered as being directive (Benkendorf et al., 2001; Brunger & Lippman, 1995; Michie et al., 1997), evidence in this study suggests that complete neutrality and providing all risk information, in as many different formats as possible, is unhelpful and may not be assisting patients to make their own informed decisions as theoretically intended. Although the ethos of non-directive counselling may have been created to assist patients to make educated, autonomous decisions, and allow specialists to take comfort in being unaccountable during difficult decisions, e.g. termination of pregnancy, these actions in practice seem to create further uncertainties and anxieties for both patients and genetic specialists.

A further consideration is needed in local GC guidelines and training curriculums regarding what is meant by 'non-directiveness' in a South African context. Finding ways to adapt local GC practice that ensures patient autonomy and respect whilst simultaneously allowing patients to feel supported in their decision-making is important. Some of these forms of support may challenge normative expectations and definitions of non-directiveness provided by current international GC guidelines. Genetic specialists need to consider the patient's individual needs and characteristics and adjust the level of directive/non-directiveness accordingly. This type of approach may result in a better form of care and may enhance patient-centred outcomes.

Genetic specialists, especially those in contexts of social, historical, and political inequality such as South Africa, are reminded of the potential for patient vulnerability and the inherent power dynamics that exist in any encounter. Thus, ethical conduct and regular reflection on the communication process is critical when enacting various levels of non-directive and directive communication. Although these actions will need to be carefully balanced along an ethical tightrope, I believe that patient care and patient-centredness can occur regardless of direct or non-directive talk, so long as the talk is tailored to the needs and concerns of the patient. Although talk in GC interactions may be directive, the GC consult as a whole does not have to be if a genetic specialist is prepared to actively engage with the patient in a meaningful way.

Additionally, if genetic specialists acknowledge their own risk perceptions and emotions during GC consultations with patients, it may alleviate the fear of being too direct. Genetic specialists' fear of being direct has shown to lead to patterns of information dumping when it comes to risk communication, and poor handling of uncertain interactional moments. Being more comfortable with their own practice and personal biases may facilitate counselling that will better assist patients to navigate difficult medical situations and decision-making.

13.4 Methodological & research considerations

Process studies have been proposed to be valuable learning opportunities for various healthcare fields. In a reverse approach, they demonstrate what is happening in practice, which in turn allows the development of recommendations and guidelines, rather than the other way around (Mendick et al., 2011). Research that pays attention to process, rather than proposed actions or measured outcomes, can also be understood as what Adams et al. (2014) term 'slow research'. Slow research considers research questions from a bottom-up approach and pays attention to everyday social practices and how they may differ in different settings. Slow research requires researchers to grapple with ideas, spend time understanding the research setting and the phenomena under investigation. Slow research also acknowledges the power of small scale, non-experimental research to address larger global problems. In "thinking globally and acting locally" slow research has the potential to address large-scale problems through "transforming many small successes into a global mosaic of health" (Adams et al., 2014; p. 194). I conclude that the methodological approach used in this study contributes to both national and international GC communication literature by providing insight into the everyday complexities of risk and uncertainty communication in local GC consultations, and may provide context-specific solutions that promote patient care and help to redefine more realistic GC goals.

Further qualitative research that focuses on healthcare communication processes are needed in South Africa. To my knowledge, the work by Wessels (2014) and Perrott (2010) are the only other studies that have focused on the interactional complexities and communicative processes within local GC consultations. Understanding GC service provision and communication within socio-cultural diverse contexts using evidence-based methods is important to ensure ongoing patient care that is contextually attuned. Uncertainty and risk will always inherently be a part of healthcare and genetic disease, however understanding and improving communication processes in GC may be one way in alleviating additional uncertainties and pressures placed

upon the service created by contextual and interactional challenges. The findings in this study have both national and international value for GC as well as for other areas of healthcare where risk and uncertainty are frequently conveyed.

The study demonstrates the value of video-based research to understand naturally occurring interactions and communicative processes. Recording patient-healthcare consultations offers particularly important and exciting opportunities for health communication research. However, this method has the potential to be fraught with ethical dilemmas. Not only is it privy to narratives that may be deeply personal, emotive, and traumatic, but it also has a proclivity to comment on people's behaviours in detail (Kiyimba & O'Reilly, 2016; Liamputtong, 2007).

Careful planning, consideration and reflection are required by the researcher, especially the recruitment of participants and acquisition of informed consent since potentially sensitive health interactions can be captured. Retrospective consent is important in video-based research. The researcher must also be aware of potential secondary trauma to themselves and the research team when capturing and analysing sensitive data. Video-based research thus requires a greater sense of responsibility from the researcher that goes beyond governing ethics committees. Fitzpatrick and Olson (2015) and Shaw et al. (2019), encourage self-reflection in research and openly grappling with your emotions since it makes one better equipped to deal with ethical dilemmas in the research process. I agree and have found self-reflection a particularly beneficial activity during this study.

Using interactional research methodologies that borrow from principles of CA and TODA calls for detailed attention to transcription, as well as identifying verbal and non-verbal features. The translation of data may also be required when data is captured in another language. This research approach can thus be laborious and time-consuming to manage and analyse, especially when using video-recordings that capture large amounts of verbal and non-verbal data. Caution for future researchers is recommended along with an awareness of the enormity of such a task, especially when recording lengthy GC consultations within a socio-cultural diverse setting.

Triangulating methods and data sources that pay attention to the macro and micro features of communication and context, has been beneficial but has also come with added complexities. Particular challenges have been the tensions of varying epistemic stances and lenses in which the results from the data sets are viewed (Fielding & Fielding, 1986; Kuper et al., 2008; Reeves,

et al., 2008; Sim & Sharp, 1998). Awareness of pitfalls and added pressures in managing and analysing data in this type of research approach is required. Consultation with colleagues and experts in the chosen analytical fields, as well as peer debriefing, may assist in recognising blind spots in the research process.

Lastly, researching one's own professional field and the communication processes of colleagues comes with added ethical and personal dilemmas, especially in small fields with few practising professionals. Issues may include coercion, the potential to create tensions in professional relationships due to skills being evaluated, and researcher biases. Reflexivity is a critical part of undertaking research of this nature. Reflexive practice not only creates an awareness of personal, professional, and ethical dilemmas encountered, but it assists in ensuring that research is done in an authentic, open, and non-defensive manner (Bantjes & Swartz, 2018; Finlay, 2002; Parker & O'Reilly, 2013). I encourage those undertaking future research of a similar nature not only to practice reflexivity during their research process, but to include some of the deliberations in their write-up to provide a transparent account of some of the challenges incurred.

13.5 Study strengths

Although a large body of empirical research exists on communication-orientated research, little is known how South Africa's unique socio-cultural contexts with its unique set of social determinants of health impact risk and uncertainty communication in public-based genetic counselling consultations. The study has attempted to address this gap in knowledge and demonstrates the power of interactional research methods in understanding healthcare services in real-time. Focusing on language during patient-healthcare practitioner interactions highlights the complexities and tensions that occur when the voice of medicine and the voice of the lifeworld come together. Paying attention to language and communication can elucidate what the barriers to care may be. The study further reminds us that healthcare is not only a science but is largely a social act. Healthcare is shaped by the surrounding context, the activities that occur within healthcare systems and the communication between individuals (Penn, 2015; Tronto, 2010). Paying attention to these determinants to healthcare services is thus important.

The GC consultations, the genetic specialist interviews, and the observational field notes have provided important sources for researching risk and uncertainty communication in public South African GC consultations, particularly those in the Western Cape Province. Collectively they

have created a rich understanding of the research context than if I had only chosen one data source and one analytical method. The study's bricolage-inspired approach and blended socio-linguistic and talk-extrinsic methodologies have been a critical part of this study that have allowed the different sources and aspects of risk and uncertainty communication in a local GC setting to be captured in a rich and holistic way.

Although challenging at times, I also believe that my unique position as a researcher, genetic counsellor, and patient has further allowed me to see this study and its findings through different lenses. These different perspectives have helped me to appreciate the complexity of genetic risk communication and uncertainty management in GC on a deeper level.

The study includes 12 genetic specialists. Although the goal of qualitative research is not to generalise findings, this study is unique by qualitative research standards as it includes the majority of GCs working in the South African public GC services at the time of data collection. The study also includes nearly 50% of the medical geneticist complement in South Africa. Although findings from this study remain contextualised to public GC consultations in the Western Cape, this unique opportunity has assisted in understanding how a large portion of local GC practice occurs. Collectively, this unique position strengthens some of the arguments made in this study and provides strong implications for South African GC training, curriculum and public-based GC practice.

13.6 Study limitations

The study focused on the GC clinics in Cape Town, in the Western Cape Province. Although Wessels (2014) has conducted interactional research on public AMA GC consultations in Johannesburg in Gauteng Province, and Perrott (2010) on AMA consults in Cape Town, Western Cape, insight into other types of GC consultations in public GC services in Gauteng and other areas of South Africa remains unknown. South Africa's healthcare is divided into public and private services. This study did not include genetic specialist experiences or consultations conducted in the private healthcare sector, thus understanding of risk and uncertainty communication in this sector remains unknown.

Despite having insight from genetic specialists, the study does not include local patient-participants interviews. Insight into the their personal understandings of risk, uncertainty, the

contextual factors that impact GC communication, or the value of various risk formats and information provided to them in GC consultations remains unclear. Previous South African GC and health communication research by Wessels (2014) and the HCRU have shown that patients interviewed on a one-on-one basis after their GC or medical consultations can be challenging as patients may be more reserved, even when linguistically matched. Group interviews may thus be more useful when interviewing patients in a South African context (Penn et al., 2010; Scott et al., 2019). GC consultations however have the potential to include sensitive information and complex decision-making that some patients may not feel comfortable to discuss in a group setting, especially after their GC consultation. Careful consideration of how to achieve appropriate GC patient interviews for research purposes is thus needed for future studies.

Due to the small professional field, attention and caution in concealing genetic specialist participant details have been practised in this thesis write-up. Hence, genetic counsellors, genetic counselling students, and interns, as well as medical geneticists and medical registrars in the study have collectively been given the label of genetic specialists. Background information such as qualifications, years of experience, age, gender, and ethnicity has also been concealed to the best of my ability. The particular names of the hospitals and genetic clinics included in the study are also not explicitly mentioned. Qualitative research calls for in-depth details of the research site, participants, and data collection observations and processes. I have attempted to balance ethical obligations to my participants, whilst ensuring richness to my results and descriptions of the research process. This has not been an easy task.

The study adopted a convenience sampling approach with no real exclusion criteria. Data collection was thus guided by what conditions, genetic specialists and patients were available to me at the time that I was present at the different research sites. Although this approach has provided variety in the data, I am aware that this unfocused approach may have created difficulty in being able to provide in-depth analysis of a specific GC clinic, referral type (e.g. prenatal, paediatric or adult based GC), risk discussion, contextual factor or interactional detail. The data collected in this study is also largely prenatal data (9/18 consultations), thus other GC risk areas or conditions may not have been fully explored or represented.

Lastly, the blended methodological approach, together with using multiple data resources (including various types of GC consultations, video-recordings, and interviews) produced a

large volume of data. The volume of data at times has been difficult to analyse and manage, especially being a novice at the chosen qualitative methods. The dual focus on risk and uncertainty has also proved challenging. Although risk and uncertainty are intertwined and linked topics, they remain separate constructs. Narrowing down the research topic may have allowed for a more focused research goal.

13.7 Final reflections

Spending the last five years grappling with the many questions and uncertainties that GC risk communication brings, together with juggling the hats of professional, researcher and patient has made this project even more imperative and meaningful to me. I have grown on a personal level and have further learned that it is impossible to understand the world through a single lens. The world we live in is far more complicated. Here, qualitative research can assist the field, since genetic advances only become useful to people when we can understand the value of the services we provide, together with the needs of patients and families.

Understanding health communication practices is imperative to ensure good patient care. I believe that this research contributes to the understanding of genetic risk and uncertainty in South African GC consultations, and adds to a growing body of literature on GC communication and interactional research in socio-cultural diverse settings. To the reader, I hope that this thesis will not only allow you to better understand risk and uncertainty communication within South African GC consultations, but that it will spark constructive debate on risk information and how it is communicated, how uncertainty is managed, and what constitutes good patient care. These deliberations need to feed into the current GC curriculum and the way we teach up and coming genetic specialists, as well as encourage critical reflection by current practising genetic specialists. Although there may never be the perfect GC consultation, research on communication practices, and grappling with the complexities of everyday service provision remains important.

Findings from the study show that western theoretical assumptions that frame GC principles do not necessarily align with local contextual needs, for both genetic specialists and patients. Theoretical ideals and practice guidelines attempt to simplify healthcare consultations and provide suggestions on how to best approach this activity during GC training and practice. However, the true qualities of care in patient-provider health practices are often only made visible through evidence-based practice and qualitative research methods that are well suited

in capturing the complexities of communication (Stewart, 2001). Interactional research and methods such as CA and TODA provide novel opportunities to better understanding the communicative complexities and tensions within GC consultations in different settings.

South Africa is a country of contradiction and diversity, making it a rich setting in which to study healthcare communication processes. Providing complex risk information in South Africa is not easy, often creating several moments of uncertainty in terms of what the information means, and how to effectively convey it. The additional influence and impact of context cannot be ignored, and greater consideration is needed in the development and practice of GC communication in local healthcare settings to enhance patient care. Tailoring information for patients and giving genetic specialists the confidence (and permission) to be flexible in their communication to patients may be one way to work towards ensuring that individual patients' needs are being met in a contextually appropriate manner.

This research has important implications for future GC training and practice. The methodologies utilised highlight the messiness of practice, which subsequently forms an important source for debate and reflection. If we continue to be reflexive and are willing to adapt and learn in our roles as genetic specialists, I believe that it will ensure that conscientious and patient-centred practice remains a hallmark of GC. Findings from this study have both national and international value, for GC and other areas of healthcare where risk and uncertainty are frequently conveyed.

Genetic risk information and uncertainty is complex. It is human nature to seek orderliness in our own lives and the world around us so that things do not happen by accident, but rather by intentional processes and actions (Kahneman, 2011). It is thus understandable how uncertainty and risk can become contentious and uncomfortable concepts for many. These concepts have the potential to cause significant distress, yet, they also has the ability to empower patients and their families, if addressed appropriately. In a world where the only certain thing is uncertainty, learning to navigate the tension between what we can, and cannot know and embracing ambiguity is necessary. Paying attention to the lifeworld needs of patients, the contextual settings in which GC practices occur, and creating confidence in genetic specialists through their communication skills is imperative. A greater focus on these aspects of GC training and practice will ensure that patients are able to better adjust to their situations, their concerns are

validated, and patients feel supported whilst they navigate uncertain and difficult situations. This, I believe is where the true value and essence of the GC profession lies.

I end with a quote from Lowen (1970) (as cited in Kelly, 1977, p.88):

> One gains knowledge when information is broken down and assimilated into the personality. Until this is done, information is like a tool which is useless because the person does not know how to handle it. Learning is not simply a matter of acquiring information. The learned person knows how to apply this information to life, especially to his own life. He has related it to his feelings and has integrated it with his experiences.

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APPENDICES

Appendix A: Original & adjusted semi-structured interview schedules

Original semi-structured interview schedule

1. What do you understand about the term ‘genetic risk’?

2. Tell me about conveying genetic risk information patients and/or their family in the South African state healthcare system.

3. What makes it difficult?

4. What makes it easy?

5. What formats/strategies do you use when conveying genetic risk?

6. What formats/strategies do you think facilitate genetic risk communication?

7. What formats/strategies do you think hinder genetic risk communication?

8. How do you deal with feelings of uncertainty?

Adjusted semi-structured interview schedule

1. Demographics
    a. Position?
    b. Years working in the field?
    c. Years working at the genetic clinics at the hospitals?

2. What do you understand about the term ‘genetic risk’?

3. Tell me about conveying genetic risk information patients and/or their family in the South African state healthcare system.
    a. What it is like?
    b. What type of risk information do you talk about?
    c. What types of contextual factors influence the communication of risk? (e.g. language, culture, gender, literacy). Can you elaborate?

4. What makes it difficult?

5. What makes it easy?

6. Do you think that risk perceptions are influenced by various contextual factors in South Africa? Can you elaborate?

7. What formats/strategies do you use when conveying genetic risk?

8. What formats/strategies do you find particularly helpful?

9. What formats/strategies do you think are not helpful?

10. What do patients do that helps you to know that they have understood the risk information?

11. Do you adjust your approach depending on the patient or clinic? How?
    a. How do you know that the patient, or the patient's parents have understood the information provided to them in clinic?

12. What types of uncertainties arise in the consultations?

13. How do you deal with feelings of uncertainty?
    a. For the patient?
    b. For yourself?

14. Do you think uncertainty can affect the relationship between a specialist and a patient? If so, how?
15. Does a non-directive approach affect feeling of uncertainty in any way?

16. Are there any aspects of communicating uncertainty that you find particularly difficult?
    a. Does it differ for different genetic conditions or timing of consultation (e.g., prenatal consultations, pediatric consultations & adult consultations)?
    b. How do you think it influences the interaction or relationship with the patient?
    c. What helps?

17. Have there been any particular cases that you have found to be particularly challenging because of the presence of uncertainty?
    a. Could you tell me more about that experience?

18. Is there anything else that you would like to add?

Appendix B: Jefferson CA transcription notations

Jeffersonian transcription notations

Symbol	Name	Use
Text [text] [text]	Brackets	Start and end points of overlapping speech.
=	Equal sign	No hearable pause between utterances.
(# of seconds) (0.0)	Timed pause	A number in parentheses indicates the time, in seconds, of a pause in speech.
(.)	Micropause	A brief pause, usually less than 0.2 seconds.
. or ↓	Period or down arrow	Falling pitch utterance-final or internal (respectively).
? or ↑	Question mark or up arrow	Rising pitch utterance-final or internal (respectively).
–	Hyphen	An abrupt halt, cut-off or interruption in utterance.
>text<	Greater than / less than symbols	Enclosed speech was delivered more rapidly than usual for the speaker.
<text>	Less than / greater than symbols	Enclosed speech was delivered more slowly than usual for the speaker.
°	Degree symbol	Whisper or reduced volume speech.
TEXT	Capitalized text	Shouted or increased volume speech.
text	Underlined text	Emphasizing or stressing the speech.
:::	Colon(s)	Prolongation of an utterance.
(hhh)	H in parentheses	Audible exhalation, laugh particles, breathiness.
(.hhh)	.H in parentheses	Audible inhalation.
(text)	Parentheses	Speech which is unclear or in doubt in the transcript.
((italic text))	Double parentheses	Annotation of non-verbal activity such as smiling, laughing, pointing, etc.

Figure A: Jefferson CA transcription notations (Jefferson 1984, as cited in Robles, 2017)

Appendix C: Research assistant non-disclosure agreement



APPENDIX E

NON-DISCLOSURE AGREEMENT FOR RESEARCH ASSISTANTS

Research Study: ***Communicating Genetic Risk & Uncertainty in a South African Healthcare Context***

I_____, confirm that:

1. The research has been explained to me and that I understand the purpose of the study.
2. I am participating in the study voluntarily.
3. I confirm that I have been sufficient opportunity to ask questions.
4. I understand that my personal information will be kept confidential.
5. I agree to keep all participant information confidential at all times.
6. I will not disclose, distribute or copy any study, or participant information.
7. I agree that all data collected or analysed, will be immediately handed over to the principal researcher.

Signed on this date:_____ at this time:_____

Research Assistant: Name:_____ Signature:_____

Witness: Name:_____ Signature:_____

Researcher: Name:_____ Signature:_____

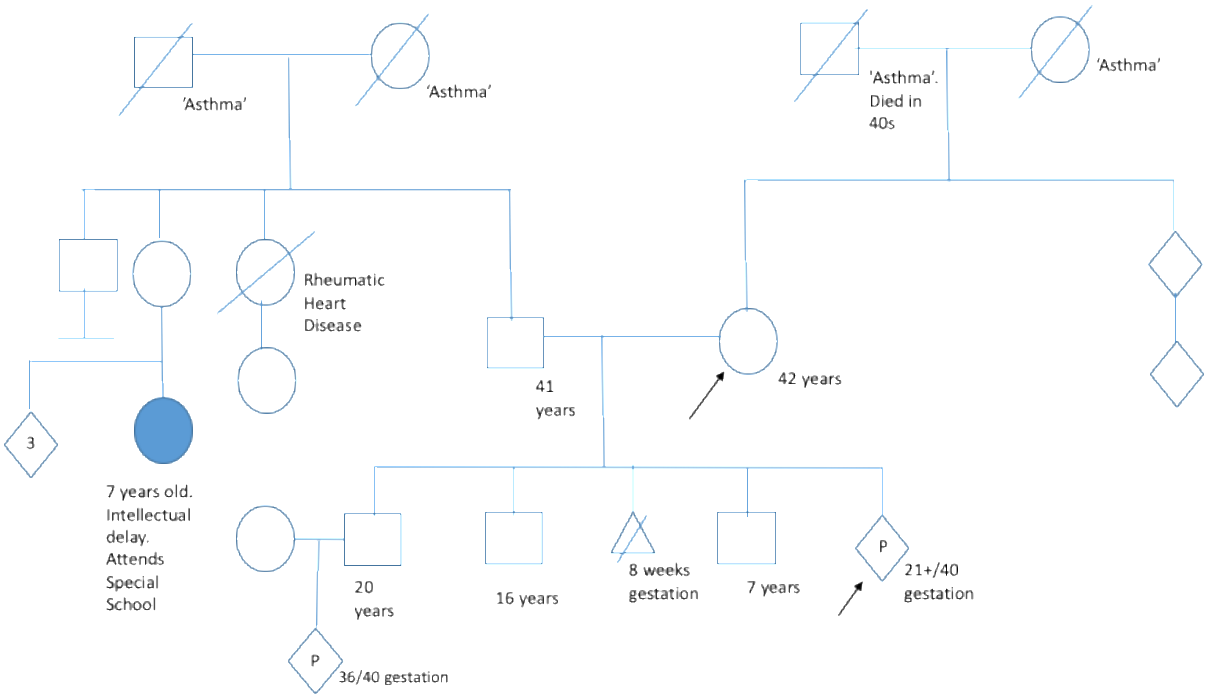
The Health Communication Research Unit, Emthomjeni Centre, 1 Jan Smuts Avenue, Johannesburg
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Appendix D: Example of a content log

Case 9- Advanced maternal age (AMA) & a soft marker for Down syndrome (DS)

Case No: 9	**Date of Recording:** 15 November 2016
Duration of Recording (minutes) **Video:** 18:48 **Audio:** 19:08	**Venue:** Pregnancy Counselling Clinic, Tygerberg Hospital
Date of start of transcription: December 2016	**Name of Transcriber:** Megan Scott
Key to Acronyms Used: • S = Specialist • P = Patient	**Pseudonyms Used:** • Leah* = Specialist • Jamil* = patient's husband
Individuals Present: Genetic specialist, pregnant patient, intern observing session.	
Comments: Consultation was conducted in Afrikaans. Patient: 42 years of age, Mixed Ancestry, Afrikaans 1st language speaker. Specialist: Female, Afrikaans 1st language speaker. Pregnancy gestation: 21+ weeks. Finding on ultrasound: flat nasal profile Adjusted risk: 1:9 for Down syndrome	

Family Pedigree



Case Overview

Introduction & agenda setting (0:00-2:21)

- (0:00-0:08) Specialist asks patient if it's okay that a student sits in & observes the session. Patient agrees. Patient looks worried & specialist asks if she is ok? She smiles awkwardly & hesitates before shaking her head.

- (0:12-1:05) Specialist introduces herself & explains that she is a GC. When asking the patient, her understanding of why she has been referred, patient responds: 'Omdat ek 42 is, dat ek high risk is.' Specialist says this is correct, all women older than 37 years of age are referred for a more detailed scan & GC because the risk is higher for older women to have a baby with abnormalities & for older women to have health complications. However, specialists emphasises that the chance to have a healthy baby is still higher, & patient's referral does not mean that there is a problem, it's just to offer the patient with some options.
- (1:06-2:21) When probing about patient's understanding of her scan result, the patient says that she was told the baby's nasal bone is flat. But she says the baby's father has a flat nasal profile. Some light laughter when specialist explains that this can be the case, however because the mother is 42 years old, seeing a flat nasal bone on the baby is something to be concerned about. But is can also mean nothing at all & the baby can be healthy.

Information gathering: Family & pregnancy history (2:22-9:50)

- (2:22-4:38) Patient has 3 sons- all healthy. The eldest is expecting a child, so patient is going to be a grandmother. Discussion on also being pregnant herself- this was not a planned pregnancy, but patient & her husband have accepted it & are now excited about it.
- (4:39-5:00) When asking about the baby's father, patient explains that he is the father of all her children, and they have been together for over 20 years. Specialist is surprised by this & remarks that this is uncommonly seen at the clinic. Commends patient on this.
- (05:01-5:38) Husband & patient both healthy. Patient has arthritis only. Patient has had a healthy pregnancy thus far.
- (5:39-6:42): Specialist asks about patient's family. Patient's family seem healthy, but specialist does not seem convinced when patient explains that her father died of 'asthma' at 42 years of age. Patient cannot give further elaboration on why or how he died.
- (6:43-8:26) Specialist asks about husband's family. Patient explains that her husband's niece is intellectually & physically delayed but cannot explain type of problem. Says *'Sy lyk soos 'n Mongooltjie'* but when probed if the child looks like the children with DS on the video, the patient seems unsure.
- (8:27-9:23) Further asking about husband's siblings & their children. Specialist trying to establish risk based on whether there are other affected family members. She mentions that she is not too worried anymore if there are no other affected individuals.
- (9:23-9:50) When asking about husband's parents, patient explains that they both passed away from 'asthma'. Specialist is not convinced- demonstrates in more proving, awkward laughter & body language.

Information giving: (09:51-13:47)

- (09:51-10:23) Specialist explains that the purpose of today's scan was to see that the baby is developing normally & to identify any major concerns. Regarding all the major things, including the baby's heart & brain, everything looks normal.
- (10:24-11:31) What is a concern though is the baby's nose. The profile looks flatter than expected. However, specialist does explain that this can be a normal occurrence if one of the parents (in this case the father) has a flat nose. Specialist explains that this can be more common in the Mixed Ancestry & Black population.
- (11:32-13:26) However because patient is 42 years old, a flat nasal profile may indicate something like DS, as DS associated with AMA. The chance to have a baby with DS now bigger. Other features of DS can also be seen on ultrasound; however, they were not seen in this case. This is good; however, they can also be missed on an ultrasound. Risks given: Age-related risk = 1 in 30. But adjusted risk now 1:9.
- (13:27-13:47) Reason patient was referred to watch DS information video and referred for GC. Amniocentesis discussed & offered. Explained that it is the only way to say with certainty is baby has DS or not. However, amniocentesis has cons, such as the risk of miscarriage.

Decision-making & psychosocial aspects (13:48-16:23)

- (13:48-14:33) Specialist asks patient about her feelings about possibility having a child with DS. Patient says that her & her husband will love & accept the child, in the same way the family loves & accepts her husband's niece. Specialist asks if husband would feel the same? Patient says he would.
- (14:41-15:37) Specialist asks how she feels about the test? Patient responds that she doesn't want the test as she would accept the baby as it is. Specialist says that if the patient will accept the child, then no need to take the test, as the amnio has a miscarriage risk & no need to take that risk then. The specialist says that the patient will then continue with the pregnancy, and at birth, if there is something worrying, then blood can be taken from the baby, without there being any risks, to test for DS.
- (15:38-16:23) Specialist says that patient can go home now without needing to worry. As it her last pregnancy, specialist encourages her to enjoy her pregnancy without worrying about DS, and *'Ons loop oor daai bruggie as dit nodig is.'*
- Specialist asks if patient has any questions? None.

Psychosocial (16:24-17:07)

- (16:21-16:48) Specialist asks if husband will be excited that they are expecting another son? Yes, patient says. Some light laughter & chatting about having a 'rugby team' of boys & that the family not having a daughter. Specialist wishes patient all the best & mentions how their son will be good friends with her eldest son's expectant baby.

Ending of consultation (17:08-18:48)

- (17:08-17:15) Specialist mentions that she wants to write some notes in the patient's clinical file & asks patient to wait for her in the waiting room. Patient gets up & leaves.

- (17:16-17:30) Specialist & intern debate on whether to switch off the recorder- they decide not to. Both leave room.
- (17:31-18:48) Room is empty until I come & switch off the recorder.

Areas/times where risk is discussed:

- (0:12-1:05)
- (11:32-13:26)
- (13:27-13:47)
- (14:41-15:37)

Areas/time where uncertainty is discussed:

- (1:06-2:21)
- (5:39-9:50)
- (10:24-11:00)

Appendix E: Example of research field notes

Site: One of the prenatal genetic counselling clinics

Today is warm and blustery with a strong bergwind blowing from early this morning. I meet some of the genetic team members at their offices and we walk across the road to the main entrance at the hospital. As we walk through the large front doors, I am stopped by a female security guard when she notices the recording equipment and tripod I am carrying. She is polite but firm. Who am I? And what am I planning to do at the hospital? I explain my purpose at the hospital and show her copies of the permission letters that I have received from the relevant university ethics committees, hospital and clinic managers. Once satisfied, she lets me through but mutters in Afrikaans that she is just doing her job and doesn't want to get into trouble with her boss. She watches me suspiciously as I walk up the stairs to the clinic.

At the clinic everything seems calm and organised. It is still before 8:00 and patients are slowly trickling in. Bright fluorescent lights burn above the clinic corridors. Again, like most of the clinics I have been too, I notice how hot and stuffy the clinic is. This is exacerbated in the prenatal clinic due to several large ultrasound machines that are being used that seem to radiate heat. The walls of the clinic are painted cream, with large wooden benches arranged along the sides of a long corridor. The ultrasound consultation rooms are on the right, the genetic counselling (GC) consultation rooms and clinic administration rooms on the left.

I notice that there are two nurses, several obstetricians and gynaecologists (some who are fetal medicine specialists) at the clinic, all looking busy and walking between rooms with purpose. The rest of the genetic clinic team soon arrives too including genetic counsellors, genetic counselling students, medical geneticists and medical registrars. It is a full clinic! One of the nurses gives the sense that she is in charge and seems rather stern. She is sorting patient files into piles on a desk. One pile has a slip of paper on top stating: "counselling." These cases have already had their sonar scan (either this morning or during the previous week) and have now been referred to the genetics team for further information and counselling. From experience, I know that these referrals usually mean difficult decisions for the patient, and in most cases invasive testing and/or termination of pregnancy has most likely been offered. It is up to the genetic team to explain what this means and what their options are. Cases range from fetal structural abnormalities, soft markers (genetic risk indicators) noted on ultrasound, and women

of advanced maternal age (≥35 years) who have a higher chance of having a baby with a chromosome abnormality. Pregnancies range in gestation, but many are 18 weeks or above. 20 weeks' gestation is usually when a full anomaly scan is offered, as it is the best time to visualise the baby's development on ultrasound.

The nurses are wearing white uniforms, with navy jerseys. They are both of Mixed Ancestry and are both wearing large red brooches with writing that says: "No means NO!!!" in bold print. I ask what it means, and they say it is to raise awareness against sexual abuse and assault. They seem like proud ambassadors. The less stern looking nurse has bright purple dyed hair cut into a short-cropped style. It is obviously a new hairstyle as the other nurse teases her and says it's quite a shock from her previously, I assume, conservative look. The specialists comment too as they walk past. The purple-haired nurse giggles. The staff all seem to all have an easy, comfortable rapport with each other, although the doctors have an air of authority and there is a definite noticeable hierarchy amongst the nurses, as well as the junior and senior specialists by the way that they communicate with each other.

Patients (mostly pregnant women) sit along long benches against the clinic corridor. Some are accompanied by partners, family members and young children. Some look anxious, others tired. Others chat quietly and politely. A man walks into the clinic and pushes a food cart down the corridor offering snacks to purchase. There is drinking yogurt, popcorn, chips, chocolate, sweets and fizzy cooldrink. Not the healthiest options. Some patients get up and buy some things. Patients often have to wait long hours before being seen and I think this a nice service to be offered. It fills a gap in a hungry tummy, and I guess it helps pass the time too. I notice a large woman sitting on a bench and I think to myself that it's rather early to be eating NikNaks as I watch her munching the luminous orange chips out of a big packet, her fingers coated in sticky, MSG laden crumbs. Then I think: I suppose she is pregnant and has cravings… Other thoughts cross my mind. I start thinking about how long it took her to get to the clinic this morning? What time did she have to get up? Is this her first meal for the day? Does she know why she is at this tertiary level specialist clinic? What is the diagnosis of her unborn baby? I chide myself. These women are here because of a complexity in their pregnancy, some unaware of the seriousness of such a referral. My attitude quickly changes. This clinic is not like your average community clinic, or secondary hospital prenatal clinics. These fetal medicine clinics only see complex cases- be it a previous family history of a genetic condition, an issue with the current pregnancy/baby's development, or a maternal concern that could affect the baby

(e.g. chronic disease such as uncontrolled diabetes or hypertension; teratogen exposure for example certain epileptic medications or recreational drugs, or the mother is of advanced maternal age). Most of the cases are complex and involve difficult discussions and decision-making.

During the morning whilst I wait for the video-recorded data to be collected, I spend most of my time sitting in the waiting area together with the patients and the nurses. A typical consultation room set-up is illustrated in Figure 7.4.

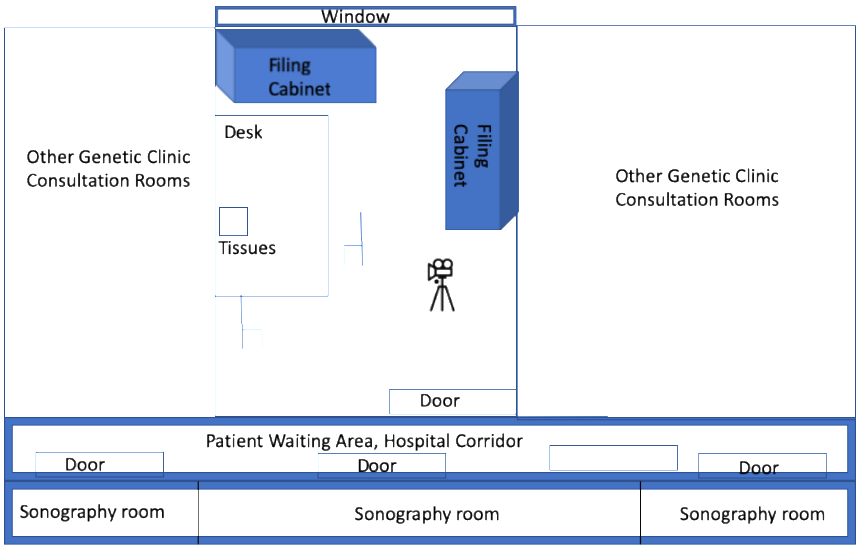


Figure B: Example of a consultation room set-up at one of the prenatal genetic counselling clinics

As the morning continues the clinic gets busier and busier. All the staff are on the go, doors are slamming, staff are walking with purpose and everyone seems to be serious and in 'work mode.' I sit quietly at the nurses' station in the waiting area, observing the happenings around me. My data collection and selection of potential participants goes as follows: As a doctor finishes an ultrasound consultation and places the patient file on the 'counselling' pile on the desk at the nursing station, I glance over the first page to see the indication for referral. I then discuss the case with the relevant genetic team member, and we make decisions on whether it is an appropriate case for me to record.

I ask one of the genetic specialists if I can record one of their referred consultations. After looking at the file they hesitate, glance at the already upset couple, and then shake their head

softly and say: “I’m sorry” as they walk towards the consultation room. A few minutes later I ask another genetic specialist if I can record their case. It is a follow-up case with a couple whose baby passed away soon after birth due to fetal abnormalities. Again, I am met with: “I’m sorry but I would prefer not to have this video-recorded.” I cringe and feel embarrassed that I even asked when I glance at the sombre couple as they walk into the consultation room. The husband placing his hand in a comforting manner on the wife’s lower back as she steps into the room. These cases are emotionally charged and sensitive, and between the genetic team and myself, we have to decide the suitability of cases for me to include in my research data. This proves to be challenging and we have to balance my need for wanting data, over being sensitive to the delicate situation at the clinic.

The longer I sit in the waiting area, the more I notice how the atmosphere shifts as the morning proceeds. What started out as a relatively relaxed and calm feeling amongst staff and patients, has taken a more solemn and heavy turn. The patients, either because they themselves have been seen for an ultrasound scan and given upsetting information, or they have watched other women experience this and are anticipating their same fate, all sit together waiting to see a doctor or a genetic specialist. Staff also seem more gloomy and tired as the morning wears on. One gynaecologist tells me in passing that although the clinic always sees tragic cases, today’s cases and diagnoses have been particularly devastating. Three spina bifida cases (which is rare), two sacral agenesis cases (which is even rarer) and several cases in which severe and gross structural abnormalities have been detected, many with poor outcomes. I get the sense that they have been ‘bearers of bad news’ all morning and it’s taking its toll on them.

I notice one woman come out of the consultation room with the fetal medicine specialists and she joins the counselling queue in the waiting area. She is a tall, slim, Black African woman in her early 30s and looks visibly in shock. She is crying softly. I see two other older Black African pregnant women still waiting to be seen, look at her sympathetically. One offers her a tissue, which she takes slightly embarrassed. Trying not to seem obvious, I subtly glance over at her file on the desk…. She has just been told her baby has severe spina bifida. I decide I am not going to ask to record her session. I watch her and for a few minutes she says nothing and just stares blankly at the wall in front of her, as if in a trance. She seems stoic, yet tears stream silently down her cheeks. She seems to snap back into reality when the two women shift closer to her, one gently touching her arm. They chat quietly in isiXhosa trying to comfort her. A few minutes later, a genetic specialist picks up her file and calls her name. They make eye contact

and the counsellor smiles softly in an empathetic manner asking her to please follow her to the consultation room. The woman stands up, straightens her clothing, wipes her tears and walks to the consultation room door holding her belly protectively. She does this as if sensing that she is going to be offered a termination. I feel horrible as I watch her leave.

After the clinic has ended, I am invited to attend a review meeting with the clinic staff. There seems to be a sense of relief that the difficult morning is over, and lunch is provided for everyone. Whilst eating cheese sandwiches, we all gather into a meeting room and all cases seen that day are discussed (showing related ultrasound images on a projector). The team then collectively agree/disagree with the management plan. In the meeting I sense shock and empathy from some of the staff as the cases are read out. Others however take on a more scientific, matter-of-fact approach. One specialist seems annoyed when discussing one case in which the patient has not attended any prenatal clinics until 33 weeks' gestation and has now been referred to the clinic because of fetal abnormalities. My knee-jerk reaction is to get upset with specialist for being so desensitised to the human and psychosocial factors at play for these patients. But then I think again... If I have found this experience so upsetting today, then how must they feel doing this on a weekly basis? At some point these specialists have to find ways to protect themselves emotionally from the trauma they see every-day. Perhaps pretending that these patients are numbers on a list rather than people within difficult circumstances is one way to do this. Is this right? And who should I be concerned for? The patients? Or the specialists? I am not sure I have a perfect answer to these deliberations. However, seeing the perspectives from all stakeholders involved seems so important here.

Appendix F: Ethical clearance certificates

R14/49 Ms Megan Scott

HUMAN RESEARCH ETHICS COMMITTEE (MEDICAL)

CLEARANCE CERTIFICATE NO. M160318

NAME: (Principal Investigator)	Ms Megan Scott
DEPARTMENT:	Speech Pathology and Audiology University of the Witwatersrand Health Communication Research Unit
PROJECT TITLE:	Communicating Genetic Risk & Uncertainty in a South African Healthcare Context
DATE CONSIDERED:	01/04/2016
DECISION:	Approved unconditionally
CONDITIONS:	Clearance of research must be sought from Cape Town and Stellenbosch University Data may only be collected from an institution when that body has given permission for the study at the site.
SUPERVISOR:	Prof C. Penn, Prof J. Watermeyer and Dr T. Wessels
APPROVED BY:	Professor P. Cleaton-Jones, Chairperson, HREC (Medical)
DATE OF APPROVAL:	09/05/2016

This clearance certificate is valid for 5 years from date of approval. Extension may be applied for.

DECLARATION OF INVESTIGATORS

To be completed in duplicate and **ONE COPY** returned to the Research Office Secretary in Room 10004,10th floor, Senate House/2nd floor, Phillip Tobias Building, Parktown, University of the Witwatersrand. I/We fully understand the the conditions under which I am/we are authorised to carry out the above-mentioned research and I/we undertake to ensure compliance with these conditions. Should any departure be contemplated,from the research protocol as approved, I/we undertake to resubmit to the Committee. **I agree to submit a yearly progress report**. The date for annual re-certification will be one year after the date of convened meeting where the study was initially reviewed. in this case, the study was initially review in March and will therefore be due in the month of March each year.

Principal Investigator Signature

Date 17/05/2016

PLEASE QUOTE THE PROTOCOL NUMBER IN ALL ENQUIRIES

UNIVERSITY OF CAPE TOWN
Faculty of Health Sciences
Human Research Ethics Committee

Room E52-46 Old Main Building
Groote Schuur Hospital
Observatory 7925
Telephone [021] 406 6492
Email: sumayah.ariefdien@uct.ac.za
Website: www.health.uct.ac.za/fhs/research/humanethics/forms

28 June 2016

HREC REF: 440/2016

Prof C Penn
Department of Speech Pathology & Audiology
Umthombo Building
East Campus, Wits University
1 Jan Smuts Avenue-JHB

Dear Prof Penn

PROJECT TITLE: COMMUNICATING GENETIC RISK AND UNCERTAINTY IN A SOUTH AFRICAN HEALTHCARE CONTEXT-(PhD-candidate-M Scott)

Thank you for submitting your study to the Faculty of Health Sciences Human Research Ethics Committee (HREC) for review.

It is a pleasure to inform you that the HREC has **formally approved** the above-mentioned study.

Approval is granted for one year until the 30 June 2017.

Please submit a progress form, using the standardised Annual Report Form if the study continues beyond the approval period. Please submit a Standard Closure form if the study is completed within the approval period.
(Forms can be found on our website: www.health.uct.ac.za/fhs/research/humanethics/forms)

Please quote the HREC REF in all your correspondence.

We acknowledge that the student, Megan Scott will also be involved in this study.

Please note that the ongoing ethical conduct of the study remains the responsibility of the principal investigator.

Please note that for all studies approved by the HREC, the principal investigator **must** obtain appropriate institutional approval before the research may occur.

Yours sincerely

PROFESSOR M BLOCKMAN
CHAIRPERSON, FHS HUMAN RESEARCH ETHICS COMMITTEE

Federal Wide Assurance Number: FWA00001637.
Institutional Review Board (IRB) number: IRB00001938

Participant information sheets & informed consent documents

UNIVERSITY OF THE WITWATERSRAND, JOHANNESBURG | Health Communication Research Unit HCRU

INFORMATION SHEET FOR GENETIC SPECIALISTS

Research Study: ***Communicating Genetic Risk & Uncertainty in a South African Healthcare Context***

Good Day,

My name is Megan Scott. I am a genetic counsellor, and am doing a PhD research project through the Health Communication Research Unit (HCRU) at the University of the Witwatersrand (Wits). For my project, I would like to look at the process of communication during genetic specialists' (genetic counsellors, medical geneticists and genetic nurses) interactions with patients, and family members, who have, or may be at risk for a genetic condition or birth defect. I would like to particularly focus on how information pertaining to genetic risk and uncertainty is talked about and negotiated.

I would like to invite you to take part in this study. Data will be collected through video-recorded genetic specialist-patient interactions, as well as audio-recorded genetic specialist interviews. If you agree to participate, then your consultations with patients will be video-recorded. Video-recording your consultations will be important for this research study as it will help me to understand not only what information is conveyed, but will help me to understand the non-verbal communication too. This would include things like gestures, body language, and the use of educational material (e.g. pictures, drawings or graphs) to help explain genetic risk information. This study is not aimed at assessing your ability as a healthcare professional, but is focused on the process of communication. For the consultations, you will not be expected to do anything other than talk to the patient as you normally would. I will not be present in the consultation, but will use the video-recording to obtain information about the communication that occurred.

I would also like to have an audio-recorded interview with you to help me understand what the task of communicating genetic risk is like in the South African healthcare system. This interview would not take longer than 45 minutes and would be done in a private space, at a place and time of your convenience.

Please note that the field of medical genetics in South Africa is small and services are only available at particular tertiary hospitals that are linked to specific academic institutions. For this reason it may be easier for participants in this study to be identified. However, every effort will be made to ensure that participants' identities are protected through the use of pseudonyms and excluding any personal information. However total anonymity may not be possible.

During the research process, your privacy will be respected at all times. Only my supervisors and I, as well a carefully chosen research assistant who has signed a non-disclosure agreement will have access to the data. All data will be kept safe in a secure locked room, or on a password protected computer and folder, to which only I will have access to. After completion of this study, data, including tape recordings will be stored for a minimum of 2 years after publication

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or 6 years in the absence of publication. Thereafter it will be destroyed. The same secure storage and confidential research practices will be upheld during this time.

The results of the study will be written in a report that will be submitted to Wits University at the end of the research. The results may also be included in academic publications or in a conference presentation. Your identifiable information and name will never be included.

Please note that participation in this study is voluntary. If you choose not to participate, this will not affect your work environment in any way. You may also withdraw from the study at any stage.

Should you have any questions regarding the research please contact myself, or any of the people listed below.

Ms. Megan Scott- Researcher 011 717 4585 or 0824438349
Professor Claire Penn- Supervisor 011 717 4579
Professor Jennifer Watermeyer- Supervisor 011 717 4578
Dr. Tina-Marie Wessels- Co-supervisor 021 406 6373
Professor Cleaton-Jones- Chairman of the Wits Human Research Ethics Committee (Medical) - 011 717 2301/2635

Thank you for taking the time to read this. If you agree to participate, please can you read and sign the consent form that is given to you.

INFORMED CONSENT FORM FOR GENETIC SPECIALISTS

Research Study: ***Communicating Genetic Risk & Uncertainty in a South African Healthcare Context***

Consent to Participate in Research

I_____, confirm that:

1. The research has been explained to me and that I understand the purpose of the study.
2. I understand that this research may not benefit me directly as a study participant, but that it will help researchers to gain a better understanding about genetic risk and uncertainty communication in South Africa.
3. I am participating in the study voluntarily.
4. I confirm that I have been given sufficient opportunity to ask questions
5. I understand that my personal information will be kept confidential.
6. I understand that I can withdraw at any time without having any effect on my work environment.

Signed on this date:_____ at this time:_____

Genetic Specialist: Name:_____ Signature:_____

Witness: Name:_____ Signature:_____

Researcher: Name:_____ Signature:_____

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INFORMED CONSENT FORM FOR GENETIC SPECIALISTS

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Consent to Audio-Record Semi-Structured Interview

I_____, confirm that:

1. I am aware that the study will make use of an audio-recorder and that my interview with the researcher will be audio-recorded.
2. I understand that the audio-recordings will be kept safe and only the researcher and authorised individuals will have access to these.
3. I understand that the recordings will never be copied or distributed.
4. I understand that my personal identity and information will be kept anonymous, and no identifiable information will be included in the write-up of this study.

Signed on this date:_____ at this time:_____

Genetic Specialist: Name:_____ Signature:_____

Witness: Name:_____ Signature:_____

Researcher: Name:_____ Signature:_____

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INFORMED CONSENT FORM FOR GENETIC SPECIALISTS

Research Study: ***Communicating Genetic Risk & Uncertainty in a South African Healthcare Context***

<u>Consent to Video-Record Healthcare Consultation</u>

I_____, confirm that:

1. I am aware that the study will make use of a video-camera and that my consultation with a patient will be video-recorded.
2. I understand that the video-recordings will be kept safe and only the researcher and authorised individuals will have access to these.
3. I understand that the recordings will never be copied or distributed.
4. I understand that personal identity and information will be kept anonymous, and no identifiable information will be included in the write-up of this study.

Signed on this date:_____ at this time:_____

Genetic Specialist: Name:_____ Signature:_____

Witness: Name:_____ Signature:_____

Researcher: Name:_____ Signature:_____

INFORMATION SHEET FOR PATIENTS

Research Study: ***Communicating Genetic Risk & Uncertainty in a South African Healthcare Context***

Good Day,

My name is Megan Scott. I am doing a PhD research project through the Health Communication Research Unit (HCRU) at the University of the Witwatersrand (Wits). Research is the process to learn the answer to a question. For this study, we would like to learn more about the way genetic specialists (including genetic counsellors, medical geneticists, and genetic nurses) and patients talk to each other about genetic risk and uncertainty during a consultation at the hospital.

I would like to invite you to take part in this study. If you agree to participate, then your consultation with a genetic specialist will be video-recorded. Video-recording your consultation will be important for this research study as it will help me to understand not only what words are spoken, but will help me to see what non-verbal gestures are used and how/if the genetic specialist uses tools such as pictures, drawings or graphs to help explain genetic risk information. For the video-recording, you will not be expected to do anything else but be in the consultation and to communicate with the genetic specialist as you normally would. I will not be present during the consultation, but will use the video-recording to get information about the type of communication that took place. The genetic specialist who will be seeing you is aware of my study.

You might be worried about who will be able to watch, or listen to the video-recording. Please know that your and your family members' information will be kept private and no-one at the hospital or clinic will be able to see it. Only authorised individuals such as my supervisors, a carefully chosen research assistant who has signed a non-disclosure agreement, and I will see the video-recording. Once the information has been collected from the video-recording, it will be kept safe in a locked room, or on a password protected computer and folder. Only I will be able to access to this. After completion of this study, data, including tape recordings will be stored for a minimum of 2 years after publication or 6 years in the absence of publication. Thereafter it will be destroyed. The same secure storage and confidential research practices will be upheld during this time.

At the end of the study, the results will be written in a report that will be given to Wits University. I may also publish some of the results in an academic journal or present it at a conference. Your and your family's identifiable information will never be included in any write up or presentation and your privacy will be respected at all times.

This study is completely voluntary and you do not have to take part if you do not want to. If you agree, and at a later stage would like to withdraw from the study, then that is OK too. By not taking

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part in the study, or withdrawing from it, will not affect any of your, or your family's present or future treatment at the clinic or at the hospital.

Please feel free to contact myself, or any of the people listed below should you have any questions about the study.

Ms. Megan Scott- Researcher 011 717 4585 or 0824438349
Professor Claire Penn- Supervisor 011 717 4579
Professor Jennifer Watermeyer- Supervisor 011 717 4578
Dr. Tina-Marie Wessels- Co-supervisor 021 406 6373
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UNIVERSITY OF THE WITWATERSRAND, JOHANNESBURG

Health Communication Research Unit HCRU

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5. I understand that my personal information will be kept confidential.
6. I understand that I can withdraw at any time without having any effect on my, or my family's treatment.

Signed on this date:_____ at this time:_____

Patient: Name:_____ Signature:_____

Witness: Name:_____ Signature:_____

Researcher: Name:_____ Signature:_____

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1. I am aware that the study will make use of a video-camera and that my consultation with a genetic specialist (a genetic counsellor, a medical geneticist or a genetic nurse) will be video-recorded.

2. I understand that the video-recordings will be kept safe and only the researcher and authorised individuals will have access to these.

3. I understand that the recordings will never be copied or distributed.

4. I understand that my and my family members' personal identity and information will be kept anonymous, and no identifiable information will be included in the write-up of this study.

Signed on this date:_____ at this time:_____

Patient: Name:_____ Signature:_____

Witness: Name:_____ Signature:_____

Researcher: Name:_____ Signature:_____

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