

Assessing patient recall, understanding, and vocabulary of genetic concepts, in a South African setting

Matsakale Barry Shingwenyana

A research report (in the format of a “submissible paper”) submitted to the Faculty of Health Sciences, University of the Witwatersrand, Johannesburg, in partial fulfilment of the requirements of the degree of Master of Science in Medicine (Genetic Counselling)

Johannesburg, 2020

Declaration

I, **Matsakale Barry Shingwenyana**, declare that this research report (in the format of a “submissible” paper), is my own, unaided work. It is being submitted for the degree of Master of Science in Medicine (Genetic Counselling) at the University of the Witwatersrand, Johannesburg. It has not been submitted before for any degree or examination at any other university.

27th day of February 2020, in Johannesburg

Contribution of the candidate to the paper

Declaration: Student's contribution to article and agreement of co-authors

I, Matsakale Barry Shingwenyana, student number 1859122, declare that this Research Report is my own work and that I contributed significantly towards research findings presented in the paper intended for publication below.

Signature of student _____

Date _____

Name of primary supervisor: Dr. Shelley Macaulay

Signature of primary supervisor _____

Date _____

Agreement by co-authors: By signing this declaration, the co-authors listed below agree to the use of the article by the student as part of his Research Report.

Article title: Assessing patient recall, understanding, and vocabulary of genetic concepts, in a South African setting

Author	Name	Signature	Date
Author 1	Matsakale Barry Shingwenyana		
Author 2	Merlyn Glass		
Author 3	Shelley Macaulay		

Dedication

This research report is dedicated to my wonderful, loving and supportive mother and grandmother, Tinyiko and N'wamadanchi Shingwenyana.

Presentations arising from this research paper

1. Southern African Society for Human Genetics Biennial Congress 2019, 3rd-6th August, Century City, Cape Town (Poster presentation).
2. School of Clinical Medicine Biennial Research Day, 23 October 2019, University of the Witwatersrand (Upcoming, oral presentation).
3. Division of Human Genetics Departmental Seminar, 30 October 2019, National Health Laboratory Service and the University of the Witwatersrand (Upcoming, oral presentation).

Abstract

Introduction: Effective communication between the patient and the genetic counsellor is necessary for a good outcome. However, for a multilingual nation like South Africa, English is the preferred language in the healthcare setting, often leading to miscommunication between a healthcare professional and a patient. Currently, no African vocabulary exists for genetic terminology which may impede patients' understanding during genetic counselling sessions.

Aims: This study aimed to assess counsellees' recall and understanding of genetic concepts discussed during a genetic counselling session for Down syndrome, and to develop a suitable South African-specific vernacular genetic vocabulary.

Methods: A quantitative, cross-sectional study was performed. Fourteen participants were recruited from the State Genetic Clinics of the National Health Laboratory Service and the University of the Witwatersrand, Johannesburg. Participants were interviewed immediately post-genetic counselling. Data were collected using a questionnaire that focused on demographics, genetic recall and understanding, and language usage. Illustrations of genetic concepts were used to assess recall and understanding. Participants were also asked to list clinical features of Down syndrome. The questionnaire was scored out of 29. In addition, participants were asked to provide terminology or analogies in their home languages for genetic terms. Data were analysed descriptively.

Results: Participants represented six ethno-linguistic groups (Northern Sotho, Sesotho, Setswana, Xitsonga, isiZulu, and isiXhosa). The majority of participants (11/14, 78.6%) were mothers of children with Down syndrome, followed by fathers (2/14, 14.3%), and a grandmother (1/14, 7.1%). The median (interquartile range) age of the participants was 39 (37.3 - 41.0) years. Mean (standard deviation) number of years of formal schooling was 12 ± 2.18 years and the majority of the participants (13/14, 92.9%) had studied biology or life sciences.

Participants obtained an average score of 22/29 (75.9%) for recall and understanding. The majority of them (12/14, 85.7%) could list two or more clinical features of the condition. Whilst some individuals could provide words to describe some of the genetic terms, others provided explanations or analogies. There were suggestions for all the genetic terms in isiZulu, for example

“*isicubu*” (cell), and “*ufuzo*” (inherited/inheritance), whereas there was only one suggestion for the term “*lee*” (egg) in Setswana.

Discussion: Most participants were able to recall information and demonstrated a good understanding, indicating the effectiveness of genetic counselling. The proposed words for genetic terms and explanation/analogies can be utilised by South African genetic counsellors to facilitate better understanding of these terms during a genetic counselling session for non-English speaking counselees.

Acknowledgements

Throughout my journey in conducting this research I have received great support. Firstly, I would like to extend my greatest gratitude to my supervisors, Ms. Merlyn Glass and Dr. Shelley Macaulay for their patience and invaluable input in formulating the research and the methodology thereof.

I would also like to thank the Genetic counsellors at the Division of Human Genetics, Clinical and Counselling Section, National Health Laboratory Service and the University of the Witwatersrand for their dedicated hard work and passion in conducting the genetic counselling sessions with the study participants.

I would also like to thank Dr. Macaulay again, for her support and commitment to the wellbeing of her students in the Division of Human Genetics.

Thank you Mr. Andrew May for your help with statistical analysis, your input is highly appreciated.

A special thank you to my fellow Genetic counselling students, Antonetta Malan and Jamey Thom for their consistent motivation, support and friendship over the past two years.

To my friends, thank you for your constant support and for always providing a happy distraction to rest my mind outside my academics.

In addition, I would like to thank my family for their continuous love, counsel, support and prayers. You are always there for me and thank you for allowing me to pursue my dreams.

This research was supported in part by the National Research Foundation of South Africa (Grant Reference: DAAD170709250911).

Finally, my greatest gratitude to the study participants without whom this research would not have been possible.

“For I know the plans I have for you” declares the Lord, “plans to prosper you and not to harm you, plans to give you hope and a future.” Jeremiah 29:11

Table of contents

Declaration	i
Contribution of the candidate to the paper	ii
Dedication	iii
Presentations arising from this research paper	iv
Abstract	v
Acknowledgements	vii
Table of contents	viii
List of figures	ix
List of tables	x
List of abbreviations	xi
Research report in the format of a submissible paper	1
Abstract for paper submission	3
Introduction	4
Methods	6
<i>Study participants and sampling</i>	6
<i>Data collection</i>	7
<i>Data analysis</i>	8
Results	8
Discussion	15
Acknowledgements	18
Declaration of interest	18
References	19
Appendices	24
<i>Appendix A: Author guidelines</i>	24
<i>Appendix B: Ethics certificate</i>	33
<i>Appendix C: Questionnaire</i>	34
<i>Appendix D: List of explanations/analogies</i>	51
<i>Appendix E: Turn-it-in report</i>	53
<i>Appendix F: Approved protocol</i>	55

List of figures

Figure 1 Pie charts illustrating the proportion of respondents who correctly/incorrectly recalled and described the diagrams/terms in the questionnaire (Appendix C)	11
---	----

List of tables

Table 1 Demographics of the study participants.....	9
Table 2 Participants' questionnaire scores	10
Table 3 Participant-recommended words for genetic terms.....	12
Table 4a Analogies/explanations used to describe genetic terminology for the Nguni-Tsonga languages.....	13
Table 4b Analogies/explanations used to describe genetic terminology for the Sotho-Makua-Venda languages	13
Table 5 Names/terms used to refer to people with Down syndrome in local communities.....	14

List of abbreviations

IQR	Interquartile range
n	Absolute number
REDCap	Research Electronic Data Capture
SD	Standard deviation
%	Percentage

Research report in the format of a submissible paper

This work has been written up as a paper for submission to the Journal of Health Communication. The author guidelines, including word count of abstract, and referencing style have been adhered to and can be found in Appendix A. However, tables and figures have been inserted in text for ease of reference.

Assessing patient recall, understanding, and vocabulary of genetic concepts, in a South African setting

Barry Shingwenyana¹ | Merlyn Glass¹ | Shelley Macaulay¹

¹ Division of Human Genetics, National Health Laboratory Service and School of Pathology, Faculty of Health Sciences, the University of the Witwatersrand, Johannesburg, South Africa

Corner Hospital and de Korte Street, Braamfontein, Johannesburg, 2001
PO Box 1038, Johannesburg, 2000

matsakale@gmail.com

Tel: +27 76 570 0547

Abstract for paper submission

No studies have been performed in a South African setting to assess patient recall and understanding of genetics. Currently, no African vocabulary exists for genetic terminology which may impede patients' understanding during genetic counselling sessions. This study aimed to assess counselees' recall and understanding of genetic concepts discussed during a genetic counselling session for Down syndrome, and to develop a suitable South African-specific genetic vocabulary.

A quantitative cross-sectional study was performed. Fourteen participants were recruited from State Genetic Clinics in Johannesburg. Participants were interviewed immediately post-genetic counselling. Recall and understanding were assessed through the use of diagrams illustrating genetic concepts that were shown in the session. Participants also provided suggestions in their home language for genetic terms such as "genetics" and "chromosome." Data were analysed descriptively.

The participants represented six ethno-linguistic groups. On average, they obtained 75.9% for genetic recall and understanding, proving that illustrations and analogies used to explain genetic concepts are effective. The proposed words and analogies can be utilised by South African genetic counsellors to facilitate better understanding during a genetic counselling session for non-English speaking counselees.

Word count: 180

Introduction

Effective communication between a healthcare provider and a patient is essential in facilitating a successful outcome. Effective communication can be described as thorough comprehension of medical information, facilitation of decision-making, and regulation of patients' emotions (Kaplan *et al.*, 1989; Ha *et al.*, 2010). However, language barriers often lead to miscommunication which may add to the patients' anxiety, uncertainty, emotional stress, and dissatisfaction with offered health services (Meuter *et al.*, 2015).

South Africa is a multiethnic and multilingual society, and as a result, miscommunication is bound to arise when the patient and the healthcare provider do not share the same first language. There are 11 official languages in South Africa, nine of which are major African languages divided into Nguni-Tsonga (SiSwati, Xitsonga, isiXhosa, isiNdebele and isiZulu) and Sotho-Makua-Venda languages (Setswana, Sesotho, Northern Sotho, and Tshivenda) (Statistics South Africa, 2019). English is the preferred language amongst healthcare providers, thus more than 80% of medical communication in South Africa occurs across language and cultural barriers (Penn, 2007).

Health literacy is described as having the motivation and skill to, gain access to, understand, and use information to promote and maintain good health (Nutbeam, 1998; Kickbusch, 2001). The entire healthcare system works with the assumption that all patients have an understanding of complex written and spoken information (Graham & Brookey, 2008). Wasserman *et al.* (2010) assessed the English literacy level of primary healthcare patients in Gauteng, South Africa, and reported that 67% of the participants had low literacy skills, described as the inability to read, write and use numbers effectively.

The concept of health literacy was further broadened into genetic literacy by the Institute of Medicine and the Agency for Healthcare Research and Quality in the United Kingdom. This refers to both adequate knowledge and understanding of genetic principles in order to make personal life-sustaining decisions (Bowling *et al.*, 2008). No studies have been performed in a South African setting to assess the level of patients', or the public's, genetic literacy, although such studies have been conducted in non-African countries (Molster *et al.*, 2009; Haga *et al.*, 2013).

A lack of basic understanding in genetics can have serious consequences such as distorted risk perception and unwillingness to pursue health services. One needs to understand genetics in order

to make informed health decisions as well as manage and prevent disease (Christensen *et al.*, 2010).

Christensen *et al.* (2010) examined the understanding of basic genetics in black and white women in the United States. The authors found that there were misunderstandings with regards to genetic concepts in 75% of the cases. Furthermore, Lanie *et al.* (2004) explored the American public's understanding of basic genetic concepts and reported there was hesitation around explaining the inheritance, function, and location of genes, regardless of the conversational familiarity with genetic terms. Another study found that individuals with lower health literacy levels had an overall lower recall of information and a low preference for active participation in decision-making after they had received written information about a given genomic test (Lillie *et al.*, 2007). It is therefore proposed that those with limited health literacy may understand less from written and oral communication regarding genomic information, and may engage less in discussing the information with healthcare providers (Kaphingst *et al.*, 2016).

There are limited studies that have been conducted to evaluate the recall of information after genetic counselling. Somer *et al.* (1988) reported that post-genetic counselling, 80% of counselees in Finland had adequate knowledge regarding the mode of inheritance, and 74% regarding the recurrence risk of the condition in their families.

Given the general misunderstanding of genetic concepts amongst the public, healthcare providers specialised in genetics, such as genetic counsellors, are needed to communicate, educate and explain genetic concepts, and information pertaining to genetic conditions, to individuals with varying levels of health and genetic literacy (Ormond *et al.*, 2018).

Genetic counselling is defined as “the process of helping people understand and adapt to medical, psychological and familial implications of genetic contributions to disease” (Resta *et al.*, 2006). Genetic counsellors help educate individuals about genetics in a meaningful way, assisting them in understanding the medical facts, hereditary nature, and risks of the condition. In addition, they provide emotional and psychological support to families that are either affected by, or at risk of, inherited conditions (Biesecker & Peters, 2001).

There are no African words that exist for genetic terminology which can impede patients' understanding of genetics. This challenge is not only unique to Africa, other countries experience

similar problems; the German, French and Spanish languages also lack words for genetic terms. A study was conducted to design a multilingual dictionary (in English, German, French and Spanish) for genetic terms to enhance communication internationally across these four languages (Lázaro *et al.*, 2014). Furthermore, a team of four bilingual Canadian genetic counsellors have created an online medical genetics translation tool called Lexigene®; this lexicon is aimed at improving communication of genetic information to patients by translating English and French terms relevant to genetic counselling (Cloutier *et al.*, 2019).

A common genetic condition seen by South African genetic counsellors is Down syndrome, also called Trisomy 21. This condition has an approximate birth prevalence of 1/500 live births in South Africa (Christianson *et al.*, 2005). It is the most common genetic cause of intellectual disability (Mazurek & Wyka, 2015). About 90-95% cases of Down syndrome are due to non-disjunction; failure of the two chromosome 21s to separate during spermatogenesis or oogenesis resulting in a total of 47 chromosomes after fertilisation (Mazurek & Wyka, 2015).

Parents of children with Down syndrome need to understand the cause of the condition, recurrence risk, and management requirements. For example, 50% of affected children have a congenital heart defect and a cardiac assessment is therefore essential. In addition, these children are at risk of hypothyroidism and so an annual thyroid function test is recommended. All children with Down syndrome have intellectual disability and need special schooling (Bull, 2011). It is therefore essential that parents of children with Down syndrome have a good understanding of the condition.

Considering that there are no African words that exist for genetic terms, South African genetic counsellors face several challenges when explaining these terms to counselees. The aims of this study were to assess the ability of counselees to recall and understand the genetic concepts that were discussed with them in a genetic counselling session for Down syndrome, and to develop a suitable South African-specific vernacular genetic vocabulary.

Methods

Study participants and sampling

The study population was comprised of parents/guardians who attended a genetic counselling session at the State Genetic Clinics of the National Health Laboratory Service and University of

the Witwatersrand, Johannesburg following a diagnosis of Down syndrome in their child or family member.

The parents/guardians of patients with non-disjunction type Down syndrome attended their scheduled genetic counselling appointments and those who met the inclusion criteria were then invited to participate in the study.

The study included black South African individuals, over the age of 18 years, whose first language was not English but who could understand and speak English. Participants had to have attended their first genetic counselling session which was conducted in English. Individuals who were not South African, and those who had received basic genetic counselling sessions in languages other than English, were excluded.

Eight genetic counsellors (excluding the researcher) were involved in providing the genetic counselling. Prior to the commencement of the study, the genetic counsellors were briefed by the researcher to ensure that consultations were standardised regarding the information discussed and diagrams used.

All participants gave written consent to participate. To ensure anonymity and confidentiality, the questionnaire did not require participants to reveal any personal identifying information and the generated data were captured into a Research Electronic Data Capture (REDCap) database (Harris *et al.*, 2009) with access granted only to those directly involved in the study. Ethical clearance was obtained from the Human Research Ethics Committee (Medical) at the University of Witwatersrand (Ethics clearance certificate: M190248) (Appendix B). Permission was also obtained from the individual tertiary hospitals.

Data collection

The study was quantitative and cross-sectional. Research participants answered a researcher-administered questionnaire (Appendix C). This questionnaire was divided into three main sections; demographics, genetic recall and understanding, and genetic vocabulary.

Recall and understanding were viewed as a single concept and were not mutually exclusive. The section on genetic recall and understanding comprised of five questions which involved diagrams

that were shown to the counselees during their genetic counselling sessions. For some of the questions, the participants were shown the diagrams and asked to “recall” what they were, and describe what they “understood” by them. The researcher had a list of possible answers/responses that he expected the participants to give. For each answer that the participant got correct, a score of one point was awarded. This section totalled 25 points. In addition, participants were also asked to list clinical features of Down syndrome. These features were divided into four domains; delayed milestones, facial features, cardiac abnormalities and intellectual disability, (four points in total; one point per domain). Therefore, the entire questionnaire was out of 29 points.

The section on genetic vocabulary was open ended. The researcher mentioned genetic words and terms (“Down syndrome,” “genetics,” “chromosome,” “gene,” “sperm,” “egg” and “inherited/inheritance”) to the participants and asked them what words and/or explanations/analogies they would use in their own home language to describe them. In addition, participants were asked what names existed for people with Down syndrome in their communities. Data collection occurred from the 24th of June to the 12th September 2019.

Data analysis

Statistical analyses were performed using XRealStats Microsoft Excel data package. Continuous variables that were normally distributed were reported as means and standard deviations, whilst those that were not normally distributed were reported as medians and interquartile ranges. Categorical variables were represented as percentages and frequencies.

Results

During the data collection period, a total of 33 new cases of Down syndrome were booked for genetic counselling at our State Genetic Clinics; 15/33 (45.5%) did not arrive for their appointments, 3/33 (9.1%) did not meet the study’s inclusion criteria, and 2/33 (6.1%) declined to participate in the study. Therefore, only 13/33 (39.4%) of the booked cases during the study period were included in the study. Two participants were a couple; husband and wife, who attended the genetic counselling session, with their child with Down syndrome, together. They were interviewed separately. Therefore, a total of 14 participants formed the final study sample.

Table 1 describes the characteristics of the study participants of which the majority (11/14, 78.6%) were mothers of children with Down syndrome. Most participants (13/14, 92.9%) had studied biology or life sciences at school and the most common spoken language was isiZulu (6/14, 42.9%). The majority of participants had completed school with the mean ($\pm$ standard deviation) number of years of formal education being 12 ± 2.18 years.

Table 1 Demographics of the study participants

	Median (IQR) or n (%) or mean $\pm$ SD
Characteristics	Total participants (N=14)
Parent/guardian age (years)	39 (37.3-41.0)
Relationship to child with Down syndrome	
Mother	11 (78.6%)
Father	2 (14.3%)
Grandmother	1 (7.1%)
Highest level of education	
Primary	1 (7.1%)
Secondary	8 (57.1%)
University	2 (14.3%)
Technical training	3 (21.4%)
Total number of years of formal education	12 ± 2.18
Home language	
isiZulu	6 (42.9%)
isiXhosa	3 (21.4%)
Sesotho	1 (7.1%)
Northern Sotho	2 (14.3%)
Setswana	1 (7.1%)
Xitsonga	1 (7.1%)
Household asset score (/11)*	5.5 (5-6)
Marital status	
Single	4 (28.6%)
Married	10 (71.4%)
Studied Biology or Life Sciences	
Yes	13 (92.9%)
No	1 (7.1%)
Age of child with Down syndrome (months)	9 (7-14)
Questionnaire score (/29)	22 ± 6.73

IQR: interquartile range; SD: standard deviation

*Household asset score: proxy for household socioeconomic status. Kept as a continuous variable; total number of household assets out of 11.

The mean score for recall and understanding was 75.9%, with the lowest score being 20.7% and the highest being 100%. All participants were able to list two or more features of Down syndrome (Table 2).

Table 2 Participants' questionnaire scores

Participant number	Relationship to patient	Home language	Score on clinical features /4 (%)	Questionnaire score /29 (%)
1	Mother	isiZulu	4/4 (100%)	18/29 (62.1%)
2	Mother	isiXhosa	4/4 (100%)	26/29 (89.7%)
3	Mother	isiXhosa	4/4 (100%)	12/29 (41.4%)
4	Grandmother	Sesotho	4/4 (100%)	24/29 (82.8%)
5	Mother	Northern Sotho	4/4 (100%)	27/29 (93.1%)
6	Mother	isiXhosa	4/4 (100%)	20/29 (69.0%)
7	Mother	isiZulu	4/4 (100%)	29/29 (100%)
8	Father	isiZulu	4/4 (100%)	27/29 (93.1%)
9	Mother	isiZulu	3/4 (75%)	25/29 (86.2%)
10	Mother	Northern Sotho	4/4 (100%)	17/29 (58.6%)
11	Mother	Setswana	2/4 (50%)	6/29 (20.7%)
12	Mother	Xitsonga	4/4 (100%)	29/29 (100%)
13	Father	isiZulu	4/4 (100%)	23/29 (79.3%)
14	Mother	isiZulu	4/4 (100%)	25/29 (86.2%)

The proportion of correct and incorrect answers for questions 1 – 5 of the questionnaire are illustrated in Figure 1. As mentioned in the methodology section, these questions involved diagrams from which participants had to state what they recalled and understood.

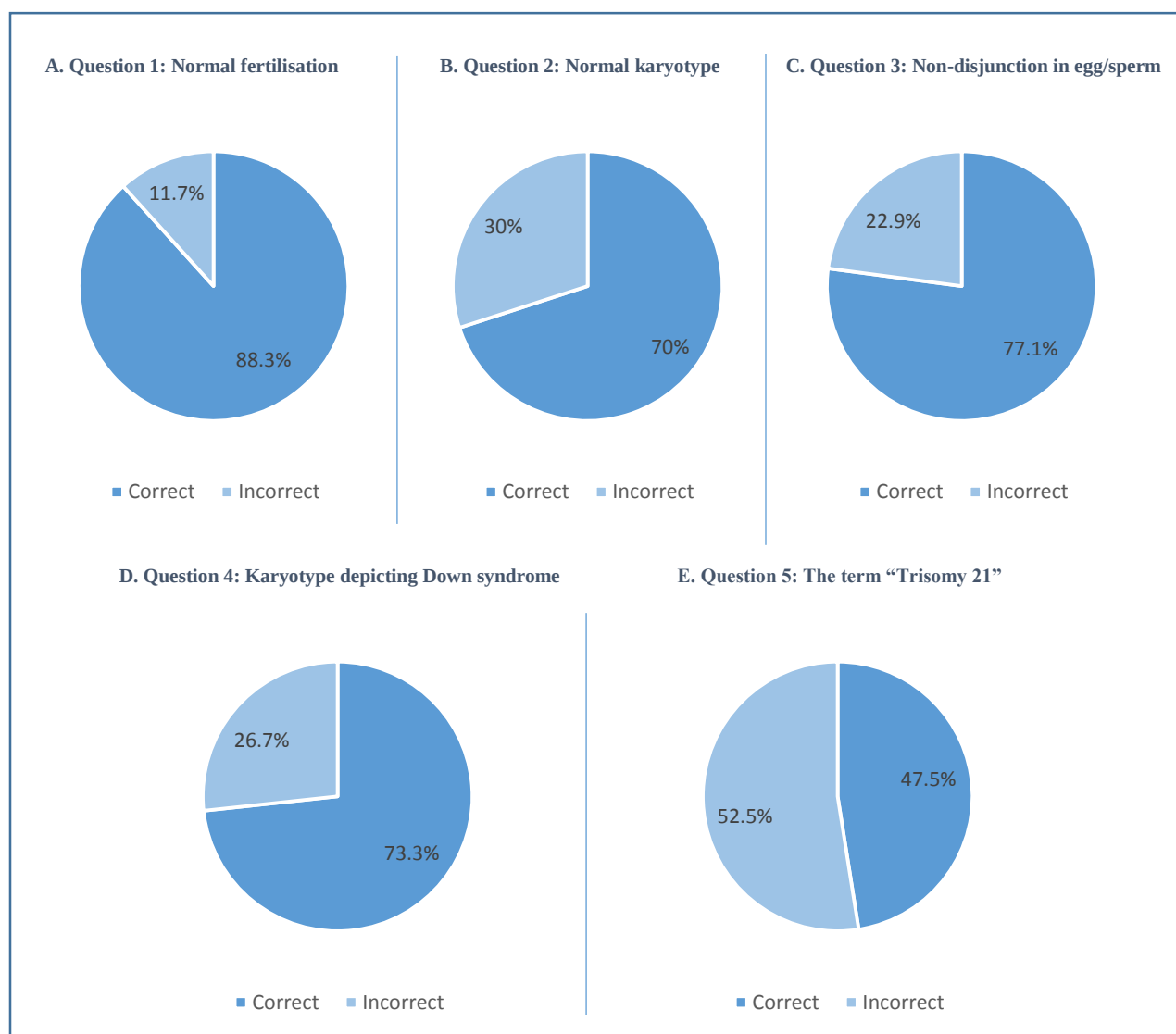


Figure 1 Pie charts illustrating the proportion of respondents who correctly/incorrectly recalled and described the diagrams/terms in the questionnaire (Appendix C)

Overall, participants scored fairly well. Question 1 (normal fertilisation) was the best answered question with an average score of 88.3% correct answers. Question 4 (Trisomy 21) was the poorest answered, with an average of 47.5% correct answers (Figure 1).

Table 3 lists the recommended words that participants would use in their own home languages to describe the selected genetic terms. There were suggestions for all seven genetic terms in isiZulu, but there was only one suggestion in Setswana.

Table 3 Participant-recommended words for genetic terms

	Recommended term					
English term	isiXhosa	isiZulu	Sotho	Northern Sotho	Setswana	Xitsonga
Genetics	<i>“Ingcambu”</i>	<i>“Amagenetics”</i>	-	-	-	-
Chromosome	-	<i>“Amachromosome”</i>	-	-	-	<i>“Tikromozoni”</i>
Gene	-	<i>“Izakha umzimba”/ “insosha”</i>	<i>“Peu”</i>	<i>“Taelo”</i>	-	-
Sperm	<i>“Mafinyela”</i>	<i>“Umqhikizo ka baba”</i>	<i>“Lero”</i>	<i>“Moroto o mogolo”</i>	-	<i>“Mbewu”</i>
Egg	<i>“Iqanda”</i>	<i>“Qanda”/ “Iqanda”</i>	<i>“Mahe”</i>	<i>“Mae”</i>	<i>“Lee”</i>	<i>“Tandza”</i>
Cell	<i>“Zicells”</i>	<i>“Isicubu”</i>	-	<i>“Ditho tsa mmele”</i>	-	-
Inherited/ inheritance	<i>“Lifa”</i>	<i>“Ifa”/ “lefa”/ “amafa”/ “ufuzo”</i>	-	<i>“Lefa”</i>	-	<i>“Ku tekela”</i>

Tables 4a and 4b provide a list of some of the explanations/analogies that participants suggested for some genetic terms (more explanations/analogies see Appendix D). Generally, as seen in Table 3, participants volunteered words for “egg” and “sperm” as opposed to explanations/analogies. As with the recommended genetic words (Table 3), explanations/analogies were suggested for all genetic terms in isiZulu, but for only one term in Setswana; genetics.

Table 4a Analogies/explanations used to describe genetic terminology for the Nguni-Tsonga languages

Nguni-Tsonga languages						
	IsiXhosa		IsiZulu		Xitsonga	
English term	Explanation/ analogy	English translation	Explanation/ analogy	English translation	Explanation/ analogy	English translation
Genetics	“Impumulo yakhe ifana ne ka makhulu”	Child’s nose resembles that of her grandmother	“Indlela omzimba usebenza ngawo”	This is how the body functions	“Swikulelwa/swik uriswa leswi endlaka leswaku munhu a vana na xivumbeko xo kari – ku fana no tekela swirho swa vatswari”	Things that help one grow and determine how a person will look. Like having some features looking like that of the parents
Gene	“Izinto ozi thata ku baba no mama, zidibene”	Combination of things from mother and father	“Yinto e ku chaza ubu wena”	Something that explains who you are	-	-
Chromosome	“Umuntu one Down syndrome uba ne chromosome ezininzi; ekwakheni komzimba wakhe”	A person with Down syndrome has more chromosomes which make up their body	“Amachromoso mes yiwo asi tshengisayo ukuthi umntwana/umuntu u phila ne Down syndrome no ma qha”	Chromosomes show us whether a person or a baby is living with a diagnosis of Down syndrome or not	“Swilo leswi vumbaka munhu”	Things that make up a person
Inherited/ Inheritance	“Fumaneka ebazalini”	Inherited from parents	“Ufuzo aluthole ku mzali wakhe”	Inheritance from their parent	“Ku tekela – utekele kokwana wakwe”	Looking like grandparents

Table 4b Analogies/explanations used to describe genetic terminology for the Sotho-Makua-Venda languages

Sotho-Makua-Venda languages						
	Sesotho		Setswana		Northern Sotho	
English term	Explanation/ analogy	English translation	Explanation/ analogy	English translation	Explanation/ analogy	English translation
Genetics	“Ntho e leng mmeleng wa rona, e re botsa gore nwana a hole joang”	Parts of the body which instruct the baby on how to grow	“Lebua ka bana ba sa phelang hantle”	Genetic condition	-	-
Gene	-	-	-	-	“Taelo ya mahlo, taelo ya letlalo”	Instruction for eyes, instruction for skin
Chromosome	-	-	-	-	“Seela sa mme le ntate ka nako ya thobalano dhlakane”	Parts of both parents that are involved in fertilisation
Inherited/ inheritance	“Di ntho tse a di fumanang ho batswadi ba hae”	Things that you get from your parents	-	-	“Otšia karolo ya mma le ntate, tša hlakana”	Inheriting the combined characteristics of the parents

Lastly, participants also volunteered names they had heard used in their communities to refer to people with Down syndrome (Table 5). Most of these names/terms were provided by isiZulu-speaking individuals, there was only one name/term in Sesotho, and most had negative connotations.

Table 5 Names/terms used to refer to people with Down syndrome in local communities

Names/terms used to describe Down syndrome in communities	Language	English translation
<i>“Idhomu”</i>	Xitsonga	A stupid child
<i>“Xa penga”</i>	Xitsonga	Crazy child
<i>“Wa gafa”</i>	Northern Sotho	Crazy child
<i>“Isidalwa”</i>	IsiZulu	Different child
<i>“Bo Takalani”</i>	isiZulu	Named after Takalani school for the intellectually disabled
<i>“Mashorty-shorty”</i>	Sesotho	Short child
<i>“Bana ba sa itakanelang”</i>	Northern Sotho	Abnormal child
<i>“Ukukhubazeka”</i>	isiZulu	Disabled child

Discussion

This was the first study to report on recall and understanding of genetic concepts in a South African setting. Overall, participants obtained a fairly high average score of 22/29 (75.9%) for the questionnaire, indicating good recall and understanding. This suggests that the genetic counselling sessions were informative and added to the participants' knowledge about their children's condition. Similarly, enhanced knowledge on genetics was reported by Seidenfeld *et al.* (1981) in mothers of children with Down syndrome who had received genetic counselling in Indiana.

Most participants (11/14, 78.6%) were mothers of children with Down syndrome, and only two fathers participated in the study (Table 2). This is common in our setting where most of our counselees are mothers of children with genetic conditions. Amato (1998) suggested that this might be because fathers are primarily viewed as financial providers and disciplinarians as opposed to primary caregivers attending clinic visits. In addition, the observed low attendance by fathers to genetic counselling clinics could be explained by the fact that South Africa has a high rate of absent fathers; with nearly 50% children living without daily contact with their fathers (Mavungu *et al.*, 2013).

Overall, most participants were able to answer questions correctly (Figure 1). However, it is important to take note that the study sample could be slightly biased in terms of educational attainment. Some participants (5/14, 35.7%) had attained a tertiary education (university and technical training), which is far higher than the 5.3% reported by Statistics South Africa for the Gauteng province (Statistics South Africa, 2019). One question that participants seemed to have some difficulty answering was Question 5 ("what does Trisomy 21 mean?") where an average score of 47.5% was obtained. This low score attests to the complexity of genetic terms. Language differences between genetic counsellors and counselees may be the reason for this, highlighting the need for African words to describe genetic terms.

Twelve of the participants (12/14, 85.7%) were able to name all clinical features of Down syndrome across the four domains (Table 2). A similar finding was reported by Morris *et al.* (2015), where most mothers of children with genetic conditions, including Down syndrome, were able to explain the basic clinical features post-genetic counselling. The ability of participants to explain clinical features could be because they are pointed out in a genetic counselling session,

allowing parents to recognise the diagnosis as a real and tangible entity, which enhances understanding.

The median (interquartile range) age of children with Down syndrome in the study was 9 (7-14) months, indicating that parents/guardians sought, or were referred to, genetic counselling in the early infancy period. This result suggests that literate people (as is the study sample) are more likely to seek healthcare services, a finding reported by others (Bowling *et al.*, 2008; Christensen *et al.*, 2010). The timing at which genetic counselling is provided is important; providing early genetic counselling may help alleviate the anxiety many parents face when receiving a genetic diagnosis in their child. Receiving genetic counselling soon after an affected child's birth, provides support to parents and addresses their immediate concerns. Furthermore, it may also allow parents to gain some sense of control (Morris *et al.*, 2015). This study has shown that providing early diagnosis and genetic counselling facilitates good recall and understanding of a given genetic condition.

The majority of participants identified as isiZulu-speaking (6/14, 42.9%), which is in keeping with the demographics of the Gauteng province; isiZulu is the most commonly spoken African language in Gauteng (Statistics South Africa, 2019). Participants were able to provide vernacular terms and/or explanations/analogies that they would use to describe some of the proposed genetic terms (Table 3, Table 4a, Table 4b and Appendix D). Supporting analogies have been reported to facilitate better understanding of complex genetic terms (Uhlmann *et al.*, 2009). Across all the languages, except Setswana, there were words to describe “egg” and “sperm”. In Xitsonga and isiZulu, there were words for “chromosome”, however, these were words borrowed from English; “*tikromozoni*” and “*amachromosome*”, respectively. The concept of borrowing words from other developed languages, such as English, has been documented to be common in indigenous South African languages (Mafela, 2010). The concept of word-borrowing is also seen in the suggested word for “genetics” in isiZulu, “*amagenetics*” and the suggested word for “cell”, “*zicells*” in isiXhosa (Table 3). This illustrates that language is not static; it evolves and people adopt and adapt words in their everyday life to allow for better communication (Mafela, 2010).

Generally, isiZulu speakers provided descriptive words for all the genetic terms and volunteered explanations/analogies to describe them compared to the other ethno-linguistic groups (Table 3, Table 4a and Table 4b). It may be that isiZulu is a more flexible and evolving language. However,

it is important to note that 6/14 (42.9%) of the study sample were isiZulu speakers which may have biased the results. As English is the second most spoken language after isiZulu in Gauteng, it is likely that the two languages influence each other (Kadt, 2009; Statistics South Africa, 2019).

Overall, the terms listed by participants to describe Down syndrome had negative connotations; “*idhomu*” translating into “stupid child”, and “*wa gafa*” which translates into “crazy child” (Table 5). The held beliefs that genetic conditions are due to punishment from ancestors or a result from witchcraft practices, as well as the noticeable intellectual disability in people with Down syndrome, may account for the negative associations (Phatoli *et al.*, 2015). Given the misperceptions of the condition, it can be challenging when parents of the affected child have to explain the condition to other family members, as well as their community. Ensuring a good understanding of the cause of the condition and having the vocabulary to explain it to others is essential. Therefore, genetic counsellors should instill confidence in their counselees and provide them with accurate information so that they are able educate others about Down syndrome. In addition, it is very important that genetic counsellors in South Africa, together with African communities, work towards raising awareness around genetic conditions.

The results from this novel study add to the literature on genetic literacy amongst African individuals. The newly acquired language concepts can be circulated to the South African genetic counselling community to assist in their sessions with counselees. Both counselees and genetic counsellors can work together to further advance the genetic vocabulary.

One of the main limitations to this study was the small sample size ($n = 14$), and as a result only six ethno-linguistic groups were included. Furthermore, due to the small sample size, linear and logistic regression analyses could not be performed and as a result it could not be determined if there was any association between demographic variables and the questionnaire score. In addition, the questionnaire used to collect data has not yet been validated. Since the research was conducted immediately post-genetic counselling, the study could also be affected by recall bias (Althubaiti, 2016). Therefore, future studies could evaluate recall and understanding after a longer period of time post-genetic counselling. In addition, assessing participants’ knowledge prior to and after genetic counselling, would be another interesting study.

The results of this study could be used to start developing an extensive South African-specific multilingual dictionary or translation tool that can be used by genetic counsellors which could facilitate a better understanding of genetic concepts amongst African patients.

Acknowledgements

We would like to acknowledge the counsellees who participated in the study. Special thanks to the genetic counsellors at the Division of Human Genetics, National Health Laboratory Service and the University of the Witwatersrand for conducting the genetic counselling sessions. Lastly, to the National Research Foundation (Grant Reference: DAAD170709250911) and Faculty Research Committee individual grant for funding.

Declaration of interest

The authors declare no conflicts of interest.

References

- Africa, S. S. (n.d.-a). Local Municipality | Statistics South Africa. Retrieved October 10, 2019, from http://www.statssa.gov.za/?page_id=993
- Althubaiti, A. (2016). Information bias in health research: Definition, pitfalls, and adjustment methods. *Journal of Multidisciplinary Healthcare*, 9, 211–217.
<https://doi.org/10.2147/JMDH.S104807>
- Amato, P. R. (1998). More than money? Men's contributions to their children's lives. In *Men in families: When do they get involved? What difference does it make?* (pp. 241–278). Mahwah, NJ, US: Lawrence Erlbaum Associates Publishers.
- Biesecker, B. B., & Peters, K. F. (2001). Process studies in genetic counseling: Peering into the black box. *American Journal of Medical Genetics*, 106(3), 191–198.
<https://doi.org/10.1002/ajmg.10004>
- Bowling, B. V., Acra, E. E., Wang, L., Myers, M. F., Dean, G. E., Markle, G. C., ... Huether, C. A. (2008). Development and Evaluation of a Genetics Literacy Assessment Instrument for Undergraduates. *Genetics*, 178(1), 15. <https://doi.org/10.1534/genetics.107.079533>
- Bull, M. J. (2011). Health Supervision for Children With Down Syndrome. *Pediatrics*, 128(2), 393.
<https://doi.org/10.1542/peds.2011-1605>
- Christensen, K. D., Jayaratne, T. E., Roberts, J. S., Kardia, S. L. R., & Petty, E. M. (2010). Understandings of Basic Genetics in the United States: Results from a National Survey of Black and White Men and Women. *Public Health Genomics*, 13(7–8), 467–476.
<https://doi.org/10.1159/000293287>
- Christianson, A., Howson, C. P., & Modell, B. (2005). *March of Dimes: Global report on birth defects, the hidden toll of dying and disabled children*. White Plains: March of Dimes Birth Defects Foundation. CABDirect.

- Cloutier, M., Hathaway, J., Vanneste, R., Hodges, P. D., & Sillon, G. (2019). Lexigene®: An online medical genetics translation tool to facilitate communication. *Journal of Genetic Counseling*, 28(3), 717–721. <https://doi.org/10.1002/jgc4.1099>
- Graham, S., & Brookey, J. (2008). Do patients understand? *The Permanente Journal*, 12(3), 67–69. <https://doi.org/10.7812/tpp/07-144>
- Ha, J. F., & Longnecker, N. (2010). Doctor-patient communication: a review. *The Ochsner journal*, 10(1), 38–43.
- Haga, S. B., Barry, W. T., Mills, R., Ginsburg, G. S., Svetkey, L., Sullivan, J., & Willard, H. F. (2013). Public Knowledge of and Attitudes Toward Genetics and Genetic Testing. *Genetic Testing and Molecular Biomarkers*, 17(4), 327–335. <https://doi.org/10.1089/gtmb.2012.0350>
- Harris, P. A., Taylor, R., Thielke, R., Payne, J., Gonzalez, N., & Conde, J. G. (2009). Research electronic data capture (REDCap)—A metadata-driven methodology and workflow process for providing translational research informatics support. *Journal of Biomedical Informatics*, 42(2), 377–381. <https://doi.org/10.1016/j.jbi.2008.08.010>
- Kadt, E. de. (2005). English, language shift and identities: A comparison between “Zulu-dominant” and “multicultural” students on a South African university campus. *Southern African Linguistics and Applied Language Studies*, 23(1), 19–37. <https://doi.org/10.2989/16073610509486372>
- Kaphingst, K. A., Blanchard, M., Milam, L., Pokharel, M., Elrick, A., & Goodman, M. S. (2016). Relationships Between Health Literacy and Genomics-Related Knowledge, Self-Efficacy, Perceived Importance, and Communication in a Medically Underserved Population. *Journal of Health Communication*, 21(sup1), 58–68. <https://doi.org/10.1080/10810730.2016.1144661>

- Kaplan, S. H., Greenfield, S., & Ware, J. E. (1989). Assessing the Effects of Physician-Patient Interactions on the Outcomes of Chronic Disease. *Medical Care*, 27(3), S110–S127. Retrieved from JSTOR.
- Kickbusch, I. S. (2001). Health literacy: Addressing the health and education divide. *Health Promotion International*, 16(3), 289–297. <https://doi.org/10.1093/heapro/16.3.289>
- Lanie, A. D., Jayaratne, T. E., Sheldon, J. P., Kardia, S. L. R., Anderson, E. S., Feldbaum, M., & Petty, E. M. (2004). Exploring the Public Understanding of Basic Genetic Concepts. *Journal of Genetic Counseling*, 13(4), 305–320. <https://doi.org/10.1023/B:JOGC.0000035524.66944.6d>
- Lázaro, C. C., Lapuerta, A. M. M., & Martínez, A. C. (2015). Designing a multilingual dictionary of genetic terms (English, French, German and Spanish) for the European Portal Eurogene and the International Scientific Community. *Journal of Human Sport and Exercise*, 9(4), 773–781. <https://doi.org/10.14198/jhse.2014.94.04>
- Lillie, S. E., Brewer, N. T., O'Neill, S. C., Morrill, E. F., Dees, E. C., Carey, L. A., & Rimer, B. K. (2007). Retention and Use of Breast Cancer Recurrence Risk Information from Genomic Tests: The Role of Health Literacy. *Cancer Epidemiology Biomarkers & Prevention*, 16(2), 249. <https://doi.org/10.1158/1055-9965.EPI-06-0525>
- Mafela, M. J. (2010). Borrowing and Dictionary Compilation: The Case of the Indigenous South African Languages. *Lexikos*, 20(1), 691-699–699. <https://doi.org/10.5788/20-0-162>
- Mavungu, E. M., Thompson-de Boor, H., & Mphaka, K. (2013). So we are ATM fathers’’: A study of absent fathers in Johannesburg. *South Africa: Centre for Social Development in Africa, University of Johannesburg*.
- Mazurek, D., & Wyka, J. (2015). Down syndrome-genetic and nutritional aspects of accompanying disorders. *Roczniki Państwowego Zakładu Higieny*, 66(3).

- Meuter, R. F. I., Gallois, C., Segalowitz, N. S., Ryder, A. G., & Hocking, J. (2015). Overcoming language barriers in healthcare: A protocol for investigating safe and effective communication when patients or clinicians use a second language. *BMC Health Services Research*, 15(1), 371. <https://doi.org/10.1186/s12913-015-1024-8>
- Molster, C., Charles, T., Samanek, A., & O'Leary, P. (2009). Australian Study on Public Knowledge of Human Genetics and Health. *Public Health Genomics*, 12(2), 84–91. <https://doi.org/10.1159/000164684>
- Morris, M., Glass, M., Wessels, T.-M., & Kromberg, J. G. R. (2015). Mothers' experiences of genetic counselling in Johannesburg, South Africa. *Journal of Genetic Counseling*, 24(1), 158–168. <https://doi.org/10.1007/s10897-014-9748-x>
- Nutbeam, D. (1998). Health promotion glossary (1998). *Health Promotion International*, 13(4), 349–364. <https://doi.org/10.1093/heapro/13.4.349>
- Ormond, K. E., Laurino, M. Y., Barlow-Stewart, K., Wessels, T.-M., Macaulay, S., Austin, J., & Middleton, A. (2018). Genetic counseling globally: Where are we now? *American Journal of Medical Genetics Part C: Seminars in Medical Genetics*, 178(1), 98–107. <https://doi.org/10.1002/ajmg.c.31607>
- Penn, C. (2007). Factors affecting the success of mediated medical interviews in South Africa. *Current Allergy & Clinical Immunology*, 20(2), 66–72.
- Phatoli, R., Bila, N., & Ross, E. (2015). Being black in a white skin: Beliefs and stereotypes around albinism at a South African university. *African Journal of Disability*, 4(1), 10. <https://doi.org/10.4102/ajod.v4i1.106>
- Resta, R., Biesecker, B. B., Bennett, R. L., Blum, S., Estabrooks Hahn, S., Strecker, M. N., & Williams, J. L. (2006). A New Definition of Genetic Counseling: National Society of Genetic

Counselors' Task Force Report. *Journal of Genetic Counseling*, 15(2), 77–83.

<https://doi.org/10.1007/s10897-005-9014-3>

Seidenfeld, M. J., Antley, R. M., & Opitz, J. M. (1981). Genetic counseling: A comparison of counselee's genetic knowledge before and after (part III). *American Journal of Medical Genetics*, 10(2), 107–112. <https://doi.org/10.1002/ajmg.1320100203>

Somer, M., Mustonen, H., & Norio, R. (1988). Evaluation of genetic counselling: Recall of information, post-counselling reproduction, and attitude of the counselees. *Clinical Genetics*, 34(6), 352–365. <https://doi.org/10.1111/j.1399-0004.1988.tb02893.x>

Uhlmann, W. R., Schuette, J. L., & Yashar, B. (2011). *A Guide to Genetic Counseling*. John Wiley & Sons.

Wasserman, Z., Wright, S. C. D., & Maja, T. M. (2010). Assessment of the English literacy level of patients in primary health care services in Tshwane, Gauteng province: Part 2. *Health SA Gesondheid*, 15(1), 6. <https://doi.org/10.4102/hsag.v15i1.469>

Appendices

Appendix A: Author guidelines

Instructions for authors

Thank you for choosing to submit your paper to us. These instructions will ensure we have everything required so your paper can move through peer review, production and publication smoothly. Please take the time to read and follow them as closely as possible, as doing so will ensure your paper matches the journal's requirements.



For general guidance on every stage of the publication process, please visit our Author Services website.



For editing support, including translation and language polishing, explore our Editing Services website

SCHOLARONE MANUSCRIPTS™

This journal uses ScholarOne Manuscripts (previously Manuscript Central) to peer review manuscript submissions. Please read the guide for ScholarOne authors before making a submission. Complete guidelines for preparing and submitting your manuscript to this journal are provided below.

Contents

- About the Journal
- Peer Review and Ethics
- Preparing Your Paper
- Structure
- Word Limits

- Style Guidelines
- Formatting and Templates
- References
- Editing Services
- Checklist
- Using Third-Party Material
- Submitting Your Paper
- Data Sharing Policy
- Publication Charges
- Copyright Options
- Complying with Funding Agencies
- Open Access
- My Authored Works
- Reprints

About the Journal

Journal of Health Communication is an international, peer-reviewed journal publishing high-quality, original research. Please see the journal's Aims & Scope for information about its focus and peer-review policy.

Please note that this journal only publishes manuscripts in English.

Journal of Health Communication accepts the following types of article: original articles.

© 2017 Thomson Reuters, 2016 Journal Citations Report® ranks *Journal of Health Communication* 21st out of 79 in the Communication (social science) and 29th out of 85 in Information Science & Library Science (social science) categories with an Impact Factor of 1.614. 2015 Five-Year Impact Factor: 2.440 All figures ©2017 Thomson Reuters, 2016 Journal Citation Reports® *Journal of Health Communication: International Perspectives* is the leading journal covering the full breadth of a field that focuses on the communication of health information globally. Articles feature research on: • Developments in the field of health communication; • New media, m-health and interactive health communication; • Health Literacy; • Social marketing; •

Global Health; • Shared decision making and ethics; • Interpersonal and mass media communication; • Advances in health diplomacy, psychology, government, policy and education; • Government, civil society and multi-stakeholder initiatives; • Public Private partnerships and • Public Health campaigns. Global in scope, the journal seeks to advance a synergistic relationship between research and practical information. With a focus on promoting the health literacy of the individual, caregiver, provider, community, and those in the health policy, the journal presents research, progress in areas of technology and public health, ethics, politics and policy, and the application of health communication principles. The journal is selective with the highest quality social scientific research including qualitative and quantitative studies. Peer Review Policy: All articles in the journal have undergone rigorous screening, based on both editorial and double-blind peer review.

Peer Review and Ethics

Taylor & Francis is committed to peer-review integrity and upholding the highest standards of review. Once your paper has been assessed for suitability by the editor, it will then be double blind peer reviewed by independent, anonymous expert referees. Find out more about what to expect during peer review and read our guidance on publishing ethics.

Preparing Your Paper

Structure

Your paper should be compiled in the following order: title page; abstract; keywords; main text introduction, materials and methods, results, discussion; acknowledgments; declaration of interest statement; references; appendices (as appropriate); table(s) with caption(s) (on individual pages); figures; figure captions (as a list).

Word Limits

Please include a word count for your paper.

A typical paper for this journal should be no more than 5000 words.

Style Guidelines

Please refer to these quick style guidelines when preparing your paper, rather than any published articles or a sample copy.

Please use American spelling style consistently throughout your manuscript.

Please use double quotation marks, except where “a quotation is ‘within’ a quotation”. Please note that long quotations should be indented without quotation marks.

Formatting and Templates

Papers may be submitted in Word format. Figures should be saved separately from the text. To assist you in preparing your paper, we provide formatting template(s).

Word templates are available for this journal. Please save the template to your hard drive, ready for use.

If you are not able to use the template via the links (or if you have any other template queries) please contact us [here](#).

References

Please use this reference guide when preparing your paper.

Taylor & Francis Editing Services

To help you improve your manuscript and prepare it for submission, Taylor & Francis provides a range of editing services. Choose from options such as English Language Editing, which will ensure that your article is free of spelling and grammar errors, Translation, and Artwork Preparation. For more information, including pricing, visit [this website](#).

Checklist: What to Include

1. **Author details.** All authors of a manuscript should include their full name and affiliation on the cover page of the manuscript. Where available, please also include ORCiDs and social media handles (Facebook, Twitter or LinkedIn). One author will need to be identified as the

corresponding author, with their email address normally displayed in the article PDF (depending on the journal) and the online article. Authors' affiliations are the affiliations where the research was conducted. If any of the named co-authors moves affiliation during the peer-review process, the new affiliation can be given as a footnote. Please note that no changes to affiliation can be made after your paper is accepted. Read more on authorship.

2. Should contain an unstructured abstract of 200 words.
3. You can opt to include a **video abstract** with your article. Find out how these can help your work reach a wider audience, and what to think about when filming.
4. Do not include **keywords**. Read making your article more discoverable, including information on choosing a title and search engine optimization.
5. **Funding details.** Please supply all details required by your funding and grant-awarding bodies as follows:

For single agency grants

This work was supported by the [Funding Agency] under Grant [number xxxx].

For multiple agency grants

This work was supported by the [Funding Agency #1] under Grant [number xxxx]; [Funding Agency #2] under Grant [number xxxx]; and [Funding Agency #3] under Grant [number xxxx].

6. **Disclosure statement.** This is to acknowledge any financial interest or benefit that has arisen from the direct applications of your research. Further guidance on what is a conflict of interest and how to disclose it.
7. **Data availability statement.** If there is a data set associated with the paper, please provide information about where the data supporting the results or analyses presented in the paper can be found. Where applicable, this should include the hyperlink, DOI or other persistent identifier associated with the data set(s). Templates are also available to support authors.
8. **Data deposition.** If you choose to share or make the data underlying the study open, please deposit your data in a recognized data repository prior to or at the time of submission. You will be asked to provide the DOI, pre-reserved DOI, or other persistent identifier for the data set.
9. **Supplemental online material.** Supplemental material can be a video, dataset, fileset, sound file or anything which supports (and is pertinent to) your paper. We publish supplemental material online via Figshare. Find out more about supplemental material and how to submit it with your article.

10. **Figures.** Figures should be high quality (1200 dpi for line art, 600 dpi for grayscale and 300 dpi for color, at the correct size). Figures should be supplied in one of our preferred file formats: EPS, PDF, PS, JPEG, TIFF, or Microsoft Word (DOC or DOCX) files are acceptable for figures that have been drawn in Word. For information relating to other file types, please consult our Submission of electronic artwork document.
11. **Tables.** Tables should present new information rather than duplicating what is in the text. Readers should be able to interpret the table without reference to the text. Please supply editable files.
12. **Equations.** If you are submitting your manuscript as a Word document, please ensure that equations are editable. More information about mathematical symbols and equations.
13. **Units.** Please use SI units (non-italicized).

Using Third-Party Material in your Paper

You must obtain the necessary permission to reuse third-party material in your article. The use of short extracts of text and some other types of material is usually permitted, on a limited basis, for the purposes of criticism and review without securing formal permission. If you wish to include any material in your paper for which you do not hold copyright, and which is not covered by this informal agreement, you will need to obtain written permission from the copyright owner prior to submission. More information on requesting permission to reproduce work(s) under copyright.

Submitting Your Paper

This journal uses ScholarOne Manuscripts to manage the peer-review process. If you haven't submitted a paper to this journal before, you will need to create an account in ScholarOne. Please read the guidelines above and then submit your paper in the relevant Author Center, where you will find user guides and a helpdesk.

Please note that *Journal of Health Communication* uses Crossref™ to screen papers for unoriginal material. By submitting your paper to *Journal of Health Communication* you are agreeing to originality checks during the peer-review and production processes.

On acceptance, we recommend that you keep a copy of your Accepted Manuscript. Find out more about sharing your work.

Data Sharing Policy

This journal applies the Taylor & Francis Basic Data Sharing Policy. Authors are encouraged to share or make open the data supporting the results or analyses presented in their paper where this does not violate the protection of human subjects or other valid privacy or security concerns.

Authors are encouraged to deposit the dataset(s) in a recognized data repository that can mint a persistent digital identifier, preferably a digital object identifier (DOI) and recognizes a long-term preservation plan. If you are uncertain about where to deposit your data, please see this information regarding repositories.

Authors are further encouraged to cite any data sets referenced in the article and provide a Data Availability Statement.

At the point of submission, you will be asked if there is a data set associated with the paper. If you reply yes, you will be asked to provide the DOI, pre-registered DOI, hyperlink, or other persistent identifier associated with the data set(s). If you have selected to provide a pre-registered DOI, please be prepared to share the reviewer URL associated with your data deposit, upon request by reviewers.

Where one or multiple data sets are associated with a manuscript, these are not formally peer reviewed as a part of the journal submission process. It is the author's responsibility to ensure the soundness of data. Any errors in the data rest solely with the producers of the data set(s).

Publication Charges

There are no submission fees, publication fees or page charges for this journal.

Color figures will be reproduced in color in your online article free of charge. If it is necessary for the figures to be reproduced in color in the print version, a charge will apply.

Charges for color figures in print are \$400 per figure (£300; \$500 Australian Dollars; €350). For more than 4 color figures, figures 5 and above will be charged at \$75 per figure (£50; \$100 Australian Dollars; €65). Depending on your location, these charges may be subject to local taxes.

Copyright Options

Copyright allows you to protect your original material, and stop others from using your work without your permission. Taylor & Francis offers a number of different license and reuse options, including Creative Commons licenses when publishing open access. Read more on publishing agreements.

Complying with Funding Agencies

We will deposit all National Institutes of Health or Wellcome Trust-funded papers into PubMedCentral on behalf of authors, meeting the requirements of their respective open access policies. If this applies to you, please tell our production team when you receive your article proofs, so we can do this for you. Check funders' open access policy mandates [here](#). Find out more about sharing your work.

Open Access

This journal gives authors the option to publish open access via our Open Select publishing program, making it free to access online immediately on publication. Many funders mandate publishing your research open access; you can check open access funder policies and mandates [here](#).

Taylor & Francis Open Select gives you, your institution or funder the option of paying an article publishing charge (APC) to make an article open access. Please contact openaccess@tandf.co.uk if you would like to find out more, or go to our Author Services website.

For more information on license options, embargo periods and APCs for this journal please go [here](#).

My Authored Works

On publication, you will be able to view, download and check your article's metrics (downloads, citations and Altmetric data) via My Authored Works on Taylor & Francis Online. This is where

you can access every article you have published with us, as well as your free eprints link, so you can quickly and easily share your work with friends and colleagues.

We are committed to promoting and increasing the visibility of your article. Here are some tips and ideas on how you can work with us to promote your research.

Article Reprints

You will be sent a link to order article reprints via your account in our production system. For enquiries about reprints, please contact Taylor & Francis at reprints@taylorandfrancis.com. You can also order print copies of the journal issue in which your article appears.

Queries

Should you have any queries, please visit our Author Services website or contact us [here](#).

Updated 13-02-2019

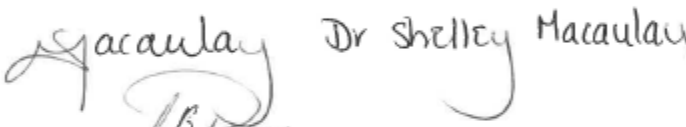
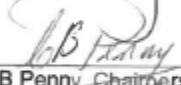
Appendix B: Ethics certificate



R14/49 Barry Shingwenyana

HUMAN RESEARCH ETHICS COMMITTEE (MEDICAL)

CLEARANCE CERTIFICATE NO. M190248

NAME: Barry Shingwenyana
(Principal Investigator)
DEPARTMENT: Human Genetics
Chris Hani Baragwanath Academic Hospital
Charlotte Maxeke Johannesburg Academic Hospital
PROJECT TITLE: Assessing patient recall, understanding, and vocabulary of genetic concepts, in a South African setting
DATE CONSIDERED: 22/02/2019
DECISION: Approved Unconditionally
CONDITIONS:
SUPERVISOR:  Dr Shelley Macaulay
APPROVED BY: 
Doctor CB Penny, Chairperson, HREC (Medical)
DATE OF APPROVAL: 19/06/2019

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 on the Third Floor, Faculty of Health Sciences, Phillip Tobias Building, 29 Princess of Wales Terrace, Parktown, 2193, University of the Witwatersrand. I/we fully understand the conditions under which I am/we are authorized 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 the application 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 reviewed in February and will therefore be due in the month of February each year. Unreported changes to the application may invalidate the clearance given by the HREC (Medical).


Principal Investigator Signature

26/06/2019
Date

PLEASE QUOTE THE PROTOCOL NUMBER IN ALL ENQUIRIES

Assessing patient recall, understanding, and vocabulary of genetic concepts, in a South African setting

Questionnaire

Visit date: _____

SECTION 1

Demography

1.1 Age of participant (years):

1.2 Relationship to child with Down syndrome:

Mother Father

Other _____

1.3 Marital status:

Single Widowed

Married/Cohabiting ☐

Separated/Divorced ☐

1.4 Total number of years of formal education:

1.5 Highest level of education attended:

No school attended ☐

Professional/technical training ☐

Primary ☐

University ☐

Secondary ☐

1.6 Did the participant study biology/life sciences?

Yes ☐

No ☐

1.6.1 **If Yes, Where?** _____

1.7 What is the participant's occupation?

1.8 Does the participant's household have or own any of the following: *(tick all that apply)*

Electricity		Cell phone		Bicycle	
Radio		Personal computer		Motorcycle	
Television		Farm animals		Car/truck/tractor	
Refrigerator		Agricultural land			

1.9 What is the participant's first language?

Tsonga		Sotho		Venda	
Ndebele		Swati		Xhosa	
Zulu		Tswana		Northern Sotho	

Other: _____

SECTION 2

Genetic recall and understanding

2.1 Referring to Picture A, did you see this during the genetic counselling session?

Yes ☐ No ☐

2.1.1 **If yes**, explain what you understood about picture A?

<u>Expected answers</u>	<u>Tick if mentioned</u>	<u>Score</u>
Egg	☐	[1]
Sperm	☐	[1]
23 chromosomes from mother	☐	[1]
23 chromosomes from father	☐	[1]
Normal fertilisation	☐	[1]
46 chromosomes in baby	☐	[1]

Notes: _____

Question 2.1 score

/6

2.2 Referring to Picture B, did you see this during the genetic counselling session?

Yes ☐ No ☐

2.2.1 **If yes**, what is it?

<u>Expected answer</u>	<u>Tick if mentioned</u>	<u>Score</u>
------------------------	--------------------------	--------------

A picture of chromosomes/genetic information	☐	<i>[1]</i>
---	--------------------------	------------

2.2.2 How many of these structures (chromosomes) are we meant to have?

<u>Expected answer</u>	<u>Tick if mentioned</u>	<u>Score</u>
------------------------	--------------------------	--------------

46	☐	<i>[1]</i>
-----------	--------------------------	------------

2.2.3 Where do these structures (chromosomes) come from?

<u>Expected answer</u>	<u>Tick if mentioned</u>	<u>Score</u>
------------------------	--------------------------	--------------

From our father and mother	☐	<i>[1]</i>
-----------------------------------	--------------------------	------------

2.2.4 How many of these structures (chromosomes) do we get from each parent?

<u>Expected answer</u>	<u>Tick if mentioned</u>	<u>Score</u>
------------------------	--------------------------	--------------

23 from each parent/half from each parent	☐	<i>[1]</i>
--	--------------------------	------------

2.2.5 What do these structures (chromosomes) do?

Expected answer

Tick if mentioned Score

They instruct our bodies how to look and function ☐ *[1]*

Notes: _____

Question 2.2 score

/5

2.3 Referring to Picture C, did you see this during the genetic counselling session?

Yes ☐ No ☐

2.3.1 If yes, what do you understand about the picture?

<u>Expected answers</u>	<u>Tick if mentioned</u>	<u>Score</u>
Egg	☐	<i>[1]</i>
Sperm	☐	<i>[1]</i>
Extra chromosome/information in the egg	☐	<i>[1]</i>
23 chromosomes from father	☐	<i>[1]</i>
24 chromosomes from mother	☐	<i>[1]</i>
Fertilisation	☐	<i>[1]</i>
Baby with 47 chromosomes	☐	<i>[1]</i>

Notes: _____

Question 2.3 score

/7

2.4 Referring to Picture D, did you see this during the genetic counselling session?

Yes ☐ No ☐

2.4.1 **If yes**, what is different about this picture compared Picture B?

<u>Expected answers</u>	<u>Tick if mentioned</u>	<u>Score</u>
-------------------------	--------------------------	--------------

Extra chromosome 21	☐	<i>[1]</i>
----------------------------	--------------------------	------------

47 chromosomes in total	☐	<i>[1]</i>
--------------------------------	--------------------------	------------

2.4.2 What does this cause?

<u>Expected answer</u>	<u>Tick if mentioned</u>	<u>Score</u>
------------------------	--------------------------	--------------

Down syndrome	☐	<i>[1]</i>
----------------------	--------------------------	------------

Notes: _____

Question 2.4 score

/3

2.5 Referring to Picture E, did you see/hear this word during the genetic counselling session?

Yes

☐

No

☐

2.5.1 **If yes**, what does Trisomy 21 mean?

<u>Expected answers</u>	<u>Tick if mentioned</u>	<u>Score</u>
Tri = 3 copies	☐	[1]
Somy = chromosomes	☐	[1]
21 = chromosome number 21	☐	[1]

2.5.2 What is another name for Trisomy 21?

<u>Expected answer</u>	<u>Tick if mentioned</u>	<u>Score</u>
Down syndrome	☐	[1]

Notes: _____

Question 2.5 score

/4

2.6 What does Down syndrome cause?

(Write down what clinical symptoms or features the participant lists)

Delayed milestones

[1]

Facial features

[1]

Cardiac problems

[1]

Intellectual disability

[1]

Notes:

Question 2.6 score

/4

43

SECTION 3

Genetic vocabulary

3.1 In your own home language, what words would you use for the following genetic words:

3.1.1 Genetics

3.1.2 Chromosomes

3.1.3 Gene

3.1.4 Sperm

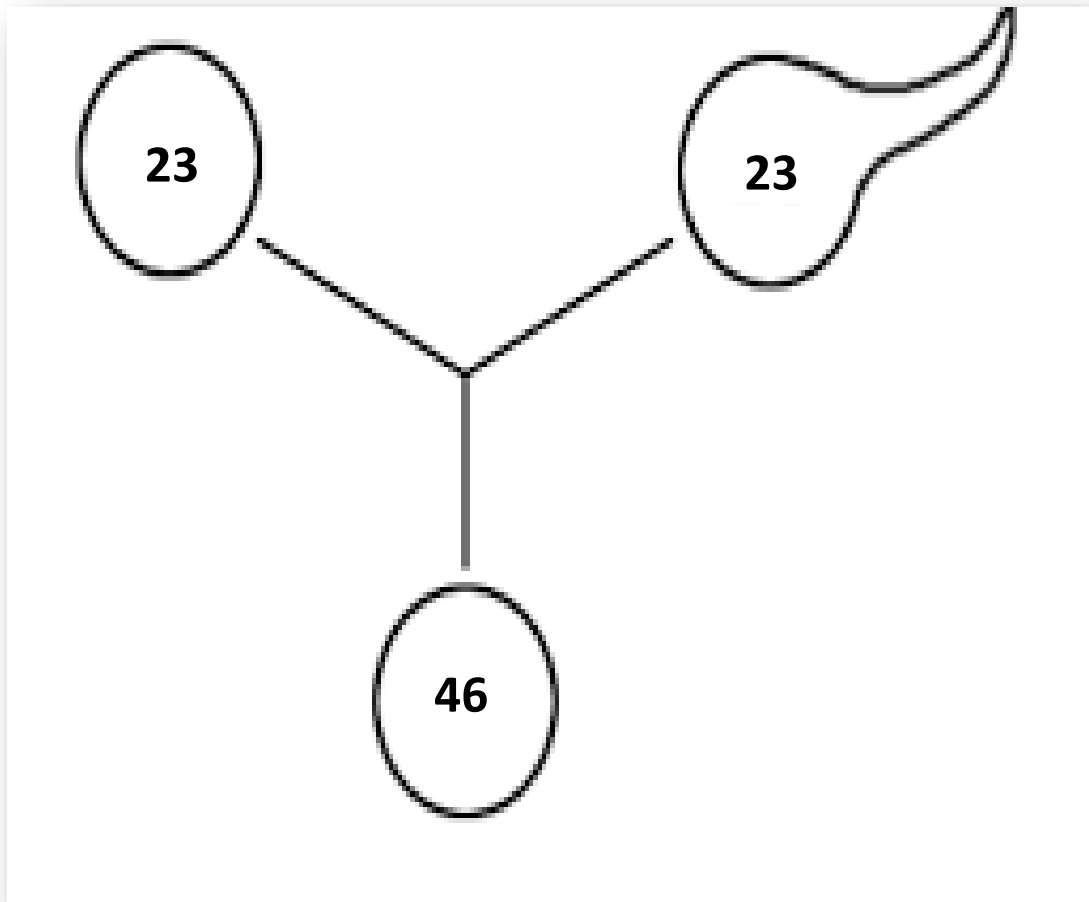
3.1.5 Egg

3.1.6 Cell

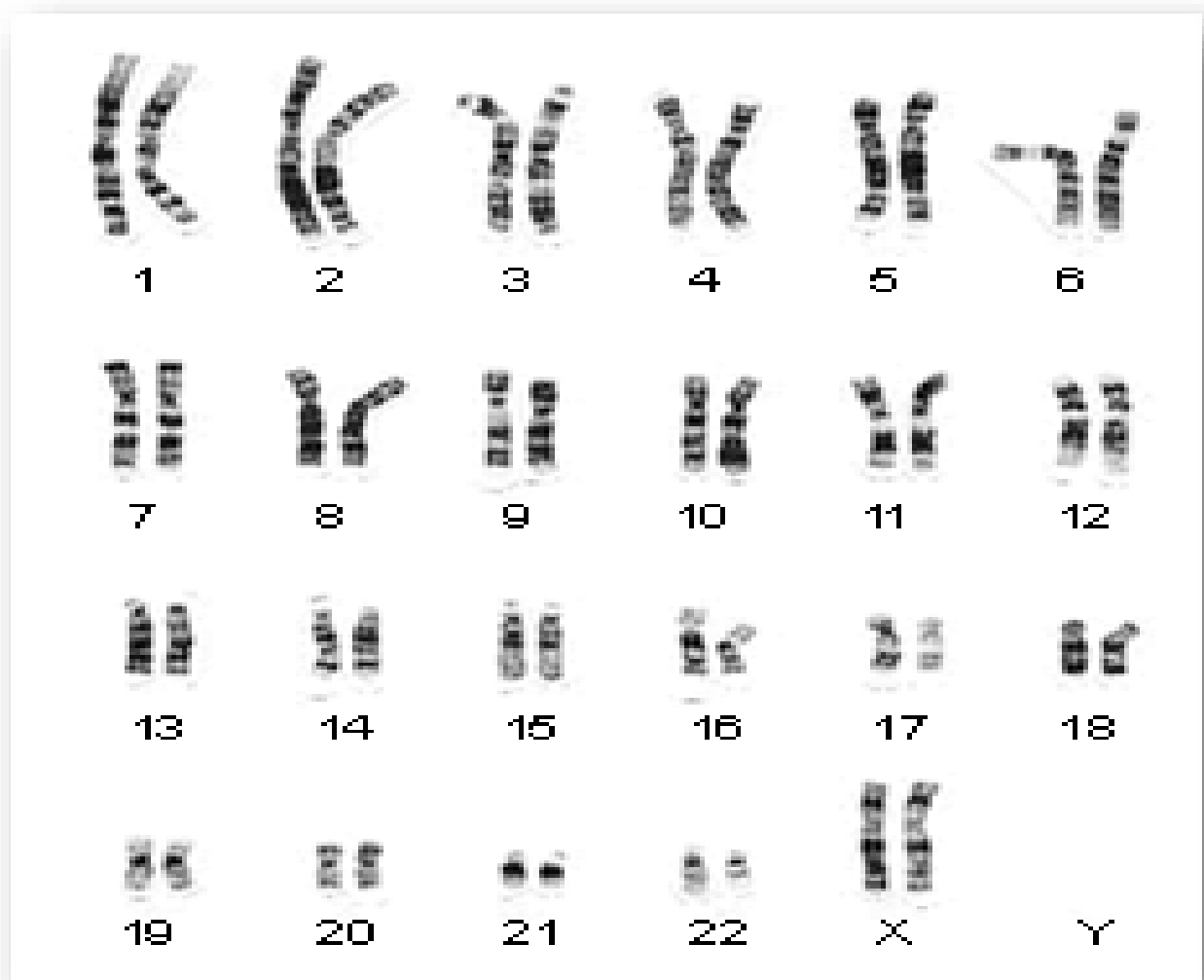
3.1.7 Inherited/inheritance

3.2 What names do your community use for people with Down syndrome?

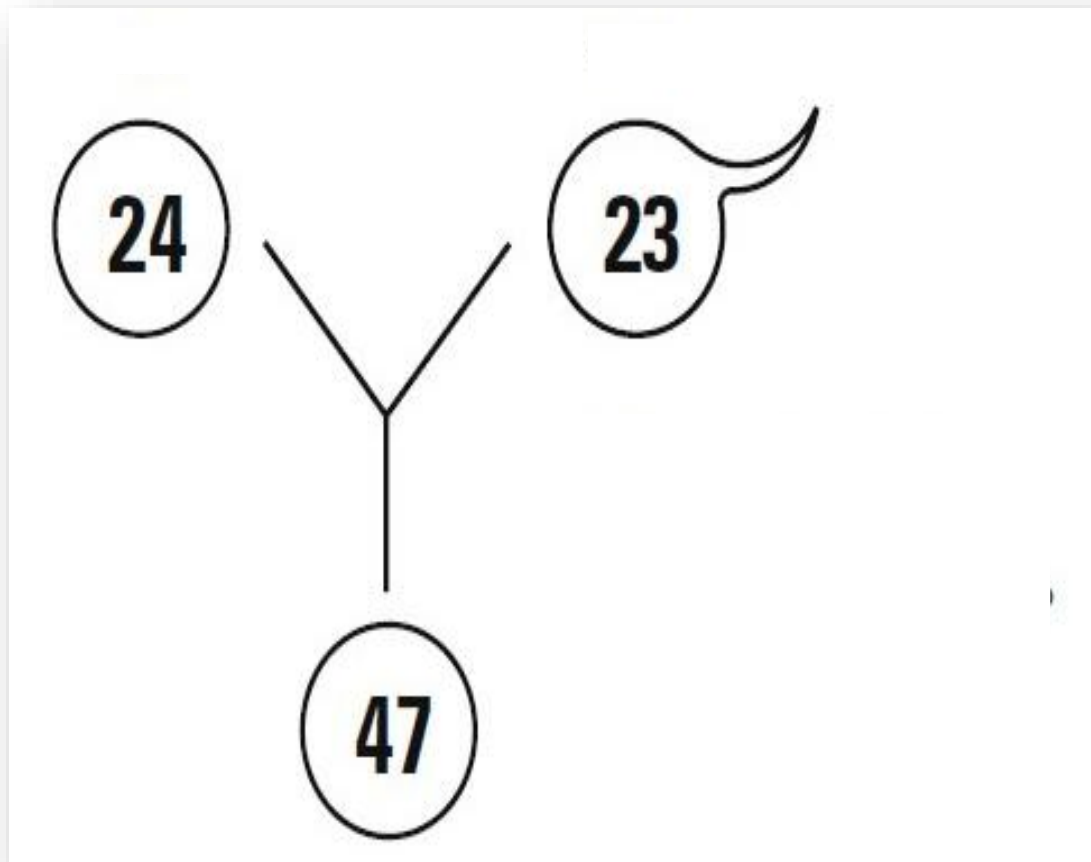
Picture A



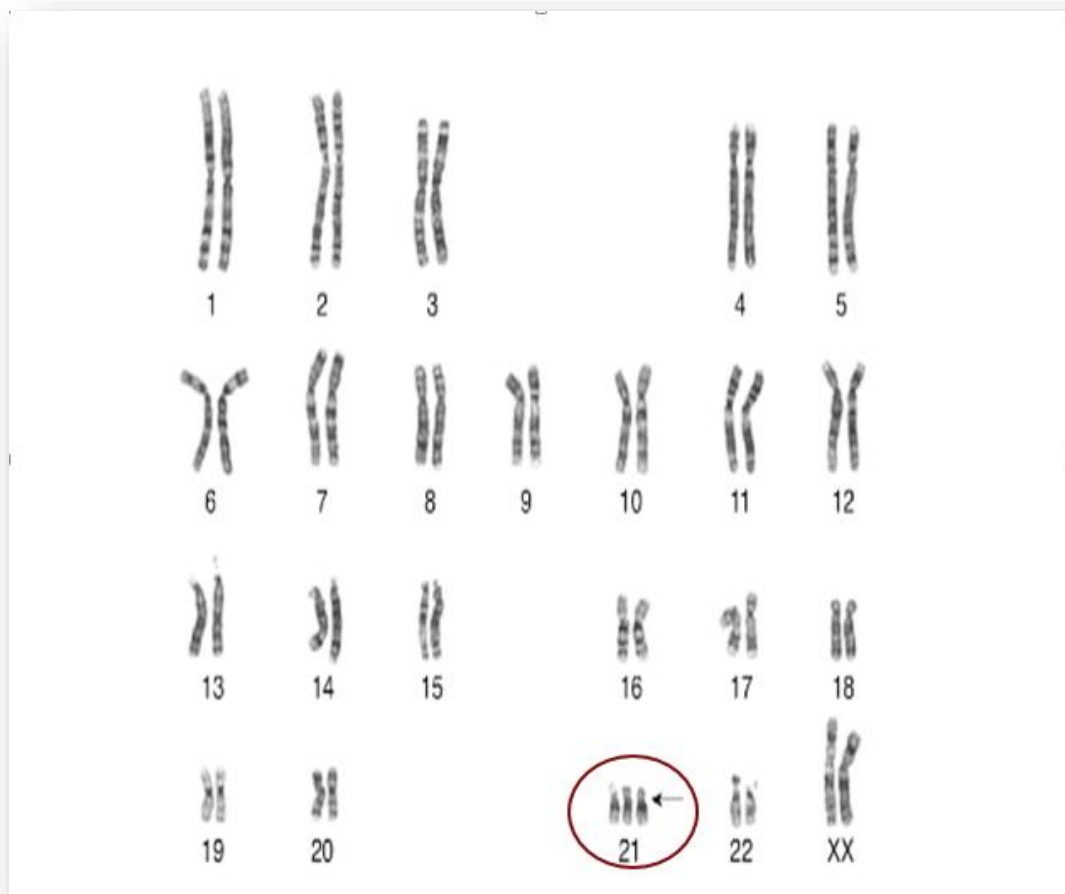
Picture



Picture C



Picture D



Picture E

Trisomy 21

Appendix D: List of explanations/analogies

Appendix B

English genetic term	isiXhosa analogies	English translation
Genetics	“Impumulo yakhe ifana ne ka makhulu” “Yinto e hlanganisa amaqanda ka mama no baba ma ni senza umntwana”	Child’s nose resembles that of her grandmother Directs the combination of the egg and sperm during fertilisation
Chromosome	“Umuntu one Down syndrome uba ne chromosome ezininzi; ekwakheni komzimba wakhe”	A person with Down syndrome has more chromosomes which make up their body
Gene	“Izinto ozi thata ku baba no mama, zidibene” “Kufanele afane na bani umntwana? Ingabe umntwana une colour ka mama kanye e ka tata?”	Combination of things from mother and father Who should the child look like? Does the child have mother or father’s complexion?
Sperm	“Lento eyenza ukuthi umama a khulelwe”	Something which impregnates a woman
Inherited/inheritance	“Fumaneka ebazalini”	Inherited from parents

English genetic term	isiZulu analogies	English translation
Genetics	“Indalo yo kwakhiwa komzimba womuntu ngokwe mvelo yakhe ukusuka ku bazali” “Indlela umzimba osebenza ngawo” “Indlela izitho zomzimba ezisebenza ngayo, uma umuntu esakhula/esakheka” “Indlela yokwakheka kwezitho zomuntu, kusukela eseyihlule, kuze kube aphelele” “Amagenetics a sebenza ngo kuthi a lawule umzimba u kuthi usebenze kanjani”	How the human body is formed, directed by parental origin This is how the body functions This is how parts of the body work together from formation and tells the body how to grow How a person is made, and how they grow up to be a whole being Controls how the body should work
Chromosome	“Kulapho ku hlangana khona amachromosome abazali bobabili ukuthi zakhe amachromosome okwakha umntwana” “Lezi zinto noma lezizicubu ezenza umuntu ziningi kanti zihlukene, ukuhlukana kwazo ku bizwa ngo kuthi amachromosomes” “Amachromosomes yiwo asi tshengisayo ukuthi umntwana/umuntu u phila ne Down syndrome no ma qha”	Parents give their chromosomes and these chromosomes make up the baby These are different things that make up a person and there is a number of them, they are called chromosomes Chromosomes show us whether a person or a baby is living with a diagnosis of Down syndrome or not
Gene	“Leso sitho (ufuzo) esitshela umzimba ukuthi usebenza kanjani nokuthi ube njani” “Yinto e ku chaza ubu wena”	Part of the body which instructs it on how to work and look Something that explains who you are
Sperm	“Isidoda sikababa esenza kube khona umtwana kumama”	Part of a man responsible for making a baby

	<i>“Yinto evela ku baba yiya eqandeni lika mama yiyo eyenza umtwana”</i>	Something that comes from the father and goes into mother’s egg to make a baby
Egg	<i>“Iqanda likamama lokwakha umtwana”</i> <i>“Iqanda yinto eyenza umama abe no mntwana”</i>	Mother’s egg involved in the formation of a baby Mother’s egg involved in the formation of a baby
Cell	<i>“Izinhlavana ezakha umzimba”</i> <i>“Izitho ezingaphakathi komzimba”</i> <i>“Yinto esinayo emzimbeni wethu”</i>	Small particles that build a body Things found inside the body Constitutes the body
Inherited/inheritance	<i>“Ufuzo aluthole ku mzali wakhe”</i> <i>“Ukuba nentsoosha ephuma kubazali bobabili”</i> <i>“Ukuhlengana kwe sperm sika baba neqanda lika mama”</i>	Inheritance from their parent Resembling both parents When the sperm and egg come together

English genetic term	Xitsonga analogies	English translation
Genetics	<i>“Swikulelwa/swikuriswa leswi endlaka leswaku munhu a vana na xivumbeko xo kari – ku fana no tekela swirho swa vatswari”</i>	Things that help one grow and determine how a person will look. Like having some features looking like that of the parents
Chromosome	<i>“Swilo leswi vumbaka munhu”</i>	Things that make up a person
Inherited/inheritance	<i>“Ku tekela – utekele kokwana wakwe”</i>	Looking like grandparents

English genetic term	Setswana analogies	English translation
Genetics	<i>“Lebua ka bana ba sa phelang hantle”</i>	Genetic condition
Sperm	<i>“Meetsi/moroto o dirang ngwana”</i>	It is like water that is involved in making a baby

English genetic term	Sesotho analogies	English translation
Genetics	<i>“Ntho e leng mmeleng wa rona, e re botsa gore nwana a hole joang”</i>	Parts of the body which instruct the baby on how to grow
Cell	<i>“Misifa e kopanyang marapo le nama”</i>	Like muscle which connects the bones and skin
Inherited/inheritance	<i>“Di ntho tse a di fumanang ho batswadi ba hae”</i>	Things that you get from your parents

English genetic term	Northern Sotho analogies	English translation
Chromosome	<i>“Seela sa mme le ntate ka nako ya thobalano dihlakane”</i>	Parts of both parents that are involved in fertilisation
Gene	<i>“Taelo ya mahlo, taelo ya letlalo”</i>	Instruction for eyes, instruction for skin
Inherited/inheritance	<i>“Otšia karolo ya mma le ntate, tša hlakana”</i>	Inheriting the combined characteristics of the parents

Appendix E: Turn-it-in report

MSC (MED) GENETIC COUNSELLING | PLAGIARISM DECLARATION

DIVISION OF HUMAN GENETICS, SCHOOL OF PATHOLOGY, FACULTY OF HEALTH SCIENCES

UNIVERSITY OF THE WITWATERSRAND



WITS
UNIVERSITY

I NATOMILE BARRY JHINQWENYAMA (Student number: 1859122), am a student registered for the degree of MSc(Med) G.C in the academic year 2019.

I hereby declare the following:

- I am aware that plagiarism (the use of someone else's work without their permission and/or without acknowledging the original source) is wrong.
- I confirm that the work submitted for assessment herein is my own, unaided work except where I have explicitly indicated otherwise.
- I have followed the required conventions in referencing the thoughts and ideas of others.
- I understand that the University of the Witwatersrand may take disciplinary action against me if there is a belief that this is not my own unaided work or that I have failed to acknowledge the source of the ideas or words in my writing.

ASSIGNMENT TYPE: RESEARCH REPORT

ASSIGNMENT TITLE: ASSESSING PATIENT RECALL, UNDERSTANDING, AND VOCABULARY OF GENETIC CONCEPTS, IN A SOUTH AFRICAN SETTING

DATE: 11/10/2019.

SIGNATURE: 

ORIGINALITY REPORT

2%

SIMILARITY INDEX

0%

INTERNET SOURCES

2%

PUBLICATIONS

0%

STUDENT PAPERS

PRIMARY SOURCES

1

Morris, Megan, Merlyn Glass, Tina-Marié Wessels, and Jennifer G. R. Kromberg. "Mothers' Experiences of Genetic Counselling in Johannesburg, South Africa", Journal of Genetic Counseling, 2015.

Publication

2%

Exclude quotes On

Exclude bibliography On

Exclude matches < 2%

Appendix F: Approved protocol

 UNIVERSITY OF THE WITWATERSRAND, JOHANNESBURG		 FACULTY OF HEALTH SCIENCES	
CANDIDATE'S SURNAME: Shingwenyana [Please print]		FIRST NAME/S: Matsakale Barry	
STUDENT NUMBER: 1859122			
CURRENT QUALIFICATIONS: BSc Medical Sciences			
TEL: 011 489 9223		CELL: 076 5700 547	
E-MAIL: matsakale@gmail.com		FAX:	
DEGREE FOR WHICH PROTOCOL IS BEING SUBMITTED: MSc (Med) Genetic Counselling			
PART-TIME OR FULL-TIME: Full-time			
FIRST REGISTERED FOR THIS DEGREE:		TERM : 1	
YEAR: 2018			
DEPARTMENT: Human Genetics			
TITLE OF PROPOSED RESEARCH: Assessing patient recall, understanding, and vocabulary of genetic concepts, in a South African setting			
CANDIDATE'S SIGNATURE: <i>[Signature]</i>			DATE: 28-01-2019
SUPERVISOR 1 (NAME & SURNAME): Ms. Merlyn Glass			% Supervision: 50%
SUPERVISOR'S QUALIFICATIONS: RN, RM, Dip Paeds, MSc			
SUPERVISOR'S DEPARTMENT: Human Genetics			
SUPERVISOR'S ADDRESS / TEL / E-MAIL: Room 4, First Floor, Jack Metz Building, Cnr Hospital & De Korte Str, Braamfontein/ Tel: 011-489-9335/ Email: merlyn.glass@nhls.ac.za			
SUPERVISOR 2 (NAME & SURNAME): Dr. Shelley Macaulay			% Supervision: 50%
SUPERVISOR'S QUALIFICATIONS: BSc, BSc(Hons), MSc (Med), MSc (Med) Genetic Counselling, PhD			
SUPERVISOR'S DEPARTMENT: Human Genetics			
SUPERVISOR'S ADDRESS / TEL / E-MAIL: Room 15, First Floor, Jack Metz Building, Cnr Hospital & De Korte Str, Braamfontein/ Tel: 011-489-9243/ Email: shelly.macaulay@wits.ac.za			
<u>SYNOPSIS OF RESEARCH:</u> PTO (see separate page)			
ETHICS PENDING: ☒ Y ETHICS APPROVED: ☐ Y (circle appropriate symbol)*		IF Y SUPPLY ETHICS CLEARANCE No:	
*Please note human ethics clearance/waiver certificate or animal ethics certificate must be in the student's name			
As supervisor, I confirm that I have read the protocol which has been submitted for assessment.			
SIGNATURE OF SUPERVISOR/S: <i>[Signature]</i>		SIGNATURE PG OFFICE STAFF: <i>[Signature]</i>	
REGISTERED YES...✓ NO....		STAMP 	

Assessing patient recall, understanding, and vocabulary of genetic concepts, in a South African setting

Barry Shingwenyana

1859122

MSc (Med) Genetic Counselling

Supervisor 1:

Ms. Merlyn Glass

RN, RM, Dip Paeds, MSc, Genetic Nurse Counsellor,

Division of Human Genetics, National Health Laboratory Service,

University of the Witwatersrand

Supervisor 2:

Dr. Shelley Macaulay

MSc (Med) Genetic Counselling, PhD, Genetic Counselling Manager and Lecturer,

Division of Human Genetics, National Health Laboratory Service,

University of the Witwatersrand

1. Literature Review

1.1 The importance of language in healthcare communication

Effective communication is required to facilitate successful outcomes between a patient with chronic diseases and a healthcare provider (Kaplan et al., 1989). Language barrier can lead to miscommunication which may subsequently add to the patients' anxiety, uncertainty, emotional stress, and dissatisfaction about offered health services. Inequities inherent to social dynamics between the patient and healthcare provider can also lead to miscommunication (Meuter et al., 2015).

The South African population is a mixture of different ethnic groups, thus a major barrier towards effective healthcare communication is bound to arise when the patient and the healthcare provider do not share the same first language. As English is the preferred language of healthcare providers, more than 80% of medical communication in South Africa (SA) occurs across language and cultural barriers (Penn, 2007).

1.2. Health literacy

Health literacy is described as having the motivation and skill to gain access to, understand, and use information in ways that promote and maintain good health (World Health Organization, 1998; Kickbusch, 2001). The entire healthcare system works with the assumption that all patients have an understanding of complex written and spoken information (Graham and Broome, 2008).

1.2.1 Genetic literacy

The Institute of Medicine and the Agency for Healthcare Research and Quality in the United Kingdom have further broadened the concept of health literacy into Genetic Literacy. This refers to both sufficient knowledge and understanding of genetic principles in order for one to make personal life-sustaining decisions as well as allow effective participation in social decisions on genetic issues (Bowling et al., 2008). No studies have been performed in a South African setting that assess the level of patients' or public genetic literacy, although there have been such studies conducted in non-African countries (Molster et al., 2009; Haga et al., 2013).

A study by Christensen et al. (2010) examining the understanding of basic genetics in black and white women in the United States, highlighted a high prevalence of approximately 75% of misunderstanding with regards to genetics. A quantitative study conducted to explore the public

understanding of basic genetic concepts amongst Americans, reported that although there is conversational familiarity with genetic terms amongst many Americans, there were frustrations or hesitations when they had to explain the inheritance, function, and location of genes (Lanie et al., 2004).

Lillie et al. (2007) found that individuals with lower health literacy levels had an overall lower recall of information and a low preference for active participation to make decisions after they had read written information about a given genomic test. It is therefore suggested that those with limited health literacy may understand less from written and oral communication regarding genomic information, and may engage less in discussing the information with healthcare providers (Kaphingst et al., 2016).

There are limited studies that have been performed to evaluate recall of information after genetic counselling. A study by Somer et al. (1988), reported that 80% of consellees had an adequate knowledge regarding the mode of inheritance and 74% regarding the recurrence risk of the condition in their families.

Given the public misunderstanding of genetic concepts and low literacy levels, healthcare providers specialised in genetics, such as genetic counsellors, are needed to communicate, educate and explain genetic information regarding health and disease to individuals with varying levels of health and genetic literacy (Ormond et al., 2018).

1.3. Genetic counselling

Genetic counsellors are registered medical professionals trained at a Masters degree level, who specialise in the field of clinical genetics (Kromberg et al., 2013).

The Genetic Counselling Definition Task Force of the National Society of Genetic Counsellors define genetic counselling as “the process of helping people understand and adapt to medical, psychological and familial implications of genetic contributions to disease” (Resta et al., 2006). The role of genetic counsellors is to help educate individuals about genetics in a meaningful way, to assist them in understanding the medical facts, hereditary nature and risks of the condition. In addition, genetic counsellors provide emotional and psychological support to families that are either affected by, or at risk of, inherited conditions so that they may adapt to the condition (Biesecker and Peters, 2001).

1.3.1. Universal words used in genetic counselling

There are common words that are used during genetic counselling sessions which include “cell”, “chromosomes”, “DNA”, “gene”, “genetics”, “sperm”, “egg”, “karyotype”, “inheritance” or “heredity, and “non-disjunction”. Supporting analogies, drawings and pictures are often used to describe these terms as there is a lack of African words for them (Uhlmann et al., 2009).

Loebner (2013), argues that one cannot understand what a word means when they either hear or see that word for the first time. This is the case with many African patients as there are no words that exist for genetic concepts in African languages which can impede their understanding of genetics and may be worsened if one’s level of education and literacy is low. Other countries experience similar problems; the German, French and Spanish languages lack words for genetic terms. A study was therefore conducted to design a multilingual dictionary for genetic terms (Cuéllar Lázaro et al., 2014).

Lack of basic understanding in genetics can have important consequences such as distorting an individual’s risk perception and willingness to pursue services like genetic counselling and seeking genetic testing (Christensen et al., 2010). The authors argue that an individual needs to understand genetics in order to make informed health decisions, manage and prevent disease.

Genetic counsellors counsel on a wide range of conditions including but not limited to, single-gene disorders (for example, Huntington disease, cystic fibrosis, neurofibromatosis), multifactorial conditions, and chromosomal disorders (for example, Trisomy 13, Trisomy 18 and Down syndrome) (Skirton et al., 2015).

1.4. Down syndrome

Down syndrome (DS), also called Trisomy 21, is the most common chromosomal disorder in South Africa and has an approximate birth prevalence of 1/500 live births (Christianson et al., 2005). Down syndrome is the most common genetic cause of intellectual disability (Mazurek and Wyka, 2015).

Down syndrome is therefore a frequent condition that a genetic counsellor encounters in genetic clinics (Sheets et al., 2011). In 2018, the Division of Human Genetics; Clinical and Counselling Section at the National Health Laboratory Service (NHLS) & the University of the Witwatersrand (WITS), Johannesburg, saw 104 patients with DS from January to December at their clinics at two

academic hospitals; Chris Hani Baragwanath Academic Hospital (CHBAH) and Charlotte Maxeke Johannesburg Academic Hospital (CMJAH).

1.4.1 Genetics of Down syndrome

Down syndrome results from an overexpression of chromosome 21 in cells; there are three copies of the chromosome instead of the normal complement of two (Mazurek and Wyka, 2015). The high birth prevalence of DS in developing countries can be ascribed to high proportion of women who are of advanced maternal age (more than 35 years) when falling pregnant (Christianson et al., 2005), although there are many other younger women also at risk of having children with DS (Agopian et al., 2012).

Non-disjunction trisomy 21 accounts for 90-95% cases of DS; it results from failure of chromosome 21 to separate during spermatogenesis or oogenesis. In 95% of the cases, it occurs in the oocytes resulting in a total of 47 chromosomes after fertilisation (Mazurek and Wyka, 2015).

1.4.2 Clinical features of Down syndrome

Down syndrome individuals have variable phenotypes, there are some features that are common; congenital hypotonia, epicanthic folds, flattened nasal bridge, upward slanting palpebral fissures, small ears, excessive skin at the back of the neck, single transverse palmar creases and low set ears. All children with DS have some degree of intellectual disability ranging from mild (IQ of 50-70) to severe (IQ of 20-30) (Bull, 2011).

1.4.3 Genetic counselling for Down syndrome.

Genetic counselling is indicated for families of an individual with DS (Micheletto et al., 2013). The models in which the process of genetic counselling is delivered varies around the world, however, they are all based on standard practices and principles (Micheletto et al., 2013).

Genetic counselling for DS in the Division of Human Genetics, Clinical and Counselling section, at the NHLS & WITS would typically involve the following:

- Taking a family history from the counsellee (usually the parents or a parent of the child).
- Taking medical and pregnancy history and also enquiring about development of the child.
- Information-giving about DS. The counsellor explains how DS may have arisen in the child. This involves the counsellor demonstrating fertilisation using illustrations (drawing the egg and sperm cell with the extra chromosome 21 in either cell) as he/she explains the

cause to the counselee (Appendix A). It also involves the use of both the normal karyotype and DS karyotype to help the counselee visualise the extra chromosome 21 (Appendices B & C). The counsellor will also provide information about the phenotype of the condition.

- Risk analysis. Discussing with the counselee about the recurrent risk, management and prenatal testing of the condition.
- Referrals to all the clinics indicated for management of DS including ophthalmology, cardiology, audiology, speech therapy and occupational therapy/physiotherapy. The counselee is also referred to the DS support group.

2. Motivation for research

There are no studies that have been performed to assess genetic recall and understanding of the public or patients in a South African setting. There are also no African words that exist for genetic terms and concepts thus making it difficult when genetic counsellors explain these to counselees in a genetic counselling session.

3. Study aims and objectives

3.1 Aims

The aims of this study are to assess the ability of counselees to recall and understand the genetic concepts discussed with them in a genetic counselling session for DS, and to develop a suitable South African-specific genetic vocabulary.

3.2 Objectives

1. To assess counselees' recall and understanding of genetic language and symbols used during genetic counselling sessions for DS.
2. To determine what words counselees would use in their own home languages to describe the genetic concepts discussed in a genetic counselling session.

4. Methodology

4.1 Research design and tool

This will be a quantitative, cross sectional study, which will involve participants answering a researcher-administered questionnaire. Due to a lack of similar studies to this particular one, the researcher has developed and designed his own questionnaire. To ensure that this questionnaire is reliable, it will be piloted on some of the staff members at the Division of Human Genetics, at the

NHLS & WITS before commencement of the study. The questionnaire focuses on three main sections namely; demographics, genetic recall and understanding, and genetic vocabulary (Appendix D). Prior the commencement of the study, the researcher will meet with the nine genetic counsellors at the NHLS & WITS to standardise and ensure that their consults for DS with counselees are similar regarding the genetic concepts and illustrations used.

For this particular project, recall and understanding are viewed as a single concept. They are not mutually exclusive. The section on genetic recall and understanding has six questions involving pictures and other concepts that are shown to the counselees during their genetic counselling sessions. For some of the questions the participants will be shown the pictures to “**recall**” what they are and describe what they “**understand**” by them. The researcher has a list of likely answers/responses that he expects the participants to give; for each answer that the participant gets correct a score of 1 point will be awarded to the participant. This section has a total of 29 points. As a genetic counselling student, the researcher has been fully trained to be self-aware on how patients may pick up on non-verbal cues, facial expressions, and tone of voice. Therefore, the researcher will maintain a stance of neutrality when interviewing the participants and hearing their responses.

The section on genetic vocabulary is open ended, the researcher will mention the genetic words and terms (Down syndrome, genetics, chromosome, gene, sperm, egg and inherited/inheritance) to the participants and ask them what words they would use in their own home language to describe them. These words will be compiled into a list which will be shared with genetic counsellors in SA for utilization in their genetic counselling sessions.

4.2 Study participants and sampling

The study population will comprise of parents/guardians attending genetic counselling at the Genetic Clinics of CHBAH and CMJAH, due to a diagnosis of DS in their children or family member. The Division of Human Genetics, Clinical and Counselling section, at the NHLS & WITS offers genetic counselling services every two weeks at these two state hospitals.

The parent/guardians of patients with DS will attend their scheduled routine genetic counselling appointments and will then be invited to participate in the study thereafter. The researcher will approach the individuals after their genetic counselling sessions to explain the study to them and to give the information sheet (Appendix E). If the participants agree, they will be asked to sign the

consent form (Appendix F) and will be taken to a separate room where the interview will be conducted. Due to participants having to stay in the hospital for longer than they normally would, refreshments will be provided.

4.2.1 Inclusion criteria and Exclusion criteria

The following inclusion and exclusion criteria will be used for this study. Those who meet the inclusion criteria will be invited to take part in the study.

Inclusion criteria	Exclusion criteria
1. Black South African individuals	1. Non-South Africans (due to local language concerns).
2. Participants who can converse fluently in English	2. Participants with children who have other genetic diagnoses other than Down syndrome.
3. Participants with a child or relative with Down syndrome (the non-disjunction type) who have received genetic counselling	3. Patients who have been counselled in languages other than English
4. Participants that are aged 18 years and older.	4. Patients who have received very basic genetic counselling session where genetic concepts were not described
5. New patients who are receiving genetic counselling for the first time (not those who are seen for follow-up sessions).	5. Participants with children or relatives with translocation or mosaic type of Down syndrome

4.3 Sample size

The Division of Human Genetics, Clinical and Counselling section, NHLS & WITS saw 104 DS cases in the year 2018 at CHBAH and CMJAH genetic clinics, on average this amounts to approximately nine cases per month. Data collection for this study will be performed over three months, this study will therefore aim to recruit between 25 and 30 participants.

4.5 Data Analysis

This study will involve basic descriptive statistics, data will be analysed using STATA version 15. Normally distributed continuous variables (e.g age and total number of years of formal studying) will be represented as means and standard deviations (SD) whilst those that are not normally distributed will be represented as medians and interquartile ranges (IQR). Categorical variables (e.g marital status, first languages, and occupation) will be represented as percentages and frequencies. The questionnaire score on genetic recall and understanding section will be the outcome variable. It will be kept continuous, thus a linear regression model will be performed to analyse whether there is an association between participants' recall and understanding of genetic concepts (the questionnaire score) and their demographics. The questionnaire score will also be categorised into tertiles (represented as percentages and frequencies), therefore a logistic regression model will be performed to assess if there are any associations between the groups and participants' demographics. Each question will be scored to see how many participants got them right. The age, sex, education, and occupation of the participant may be confounding variables.

5. Ethics

Participation in the study will be voluntary, and withdrawal from the study will be allowed with no impact on participants' or their children's treatment and management. Consent forms will be signed by eligible participants.

Confidentiality, which refers to nondisclosure of information except to another authorised person, will be maintained throughout all the stages of this research by ensuring that only the researchers have access to the identifying information and no names will be disclosed. To maintain anonymity codes for participants will be used.

This research protocol will be submitted to Human Research Ethics Committee (Medical) at the University of the Witwatersrand on the 7th of February 2019 for ethical approval and clearance.

Should the researcher feel that the participants will benefit from another genetic counselling session a follow-up appointment will be arranged for them where the researcher, a genetic counselling student will conduct the session in the participant's home language as he is fluent in four African languages (Xitsonga, Sotho, Sepedi and isiZulu). In a case where the researcher also feels that there is distress in any of the participants, referral to senior genetic counsellors will be made.

6. Timing

This study will commence as soon as ethical approval and clearance is obtained.

	Dec 2018	Jan 2019	Feb 2019	Mar 2019	Apr 2019	May 2019	Jun 2019	Jul 2019
Literature review								
Protocol planning and design								
Internal Protocol Assessment								
Application to Ethics and Approval								
Protocol Assessment								
Data Collection								
Data Analysis								
Write up and submission								

7. Funding

The following funding has been obtained which will cover expenses.

- National Research Foundation (NRF) student funding = R80 000
- I will apply for the FRC grant.

7.1 Predicted Budget

Expenses	Estimated Costs
Printing • Questionnaires • Information sheets • Consent forms	39c X 395 pages =R154.05
Refreshments • Biscuits and fruit	(R4.29 X 30) (R30 X 2) =R188.70

Total	= R342.75
--------------	-----------

8. Potential limitations

- This study will exclude patients who have been counselled in other languages (only those who have been counselled in English will be invited to part in the study).
- The study will have a small sample size due to time constraints.
- This study will use a newly developed tool that has not been validated.

References

- Agopian, A.J., Marengo, L.K., Mitchell, L.E., 2012. Predictors of trisomy 21 in the offspring of older and younger women. *Birt. Defects Res. A. Clin. Mol. Teratol.* 94, 31–35. <https://doi.org/10.1002/bdra.22870>
- Biesecker, B.B., Peters, K.F., 2001. Process studies in genetic counseling: peering into the black box. *Am. J. Med. Genet.* 106, 191–198. <https://doi.org/10.1002/ajmg.10004>
- Bowling, B.V., Acra, E.E., Wang, L., Myers, M.F., Dean, G.E., Markle, G.C., Moskalik, C.L., Huether, C.A., 2008. Development and Evaluation of a Genetics Literacy Assessment Instrument for Undergraduates. *Genetics* 178, 15–22. <https://doi.org/10.1534/genetics.107.079533>
- Bull, M.J., 2011. Health supervision for children with Down syndrome. *Pediatrics* 128, 393–406. <https://doi.org/10.1542/peds.2011-1605>
- Christensen, K.D., Jayaratne, T.E., Roberts, J.S., Kardia, S.L.R., Petty, E.M., 2010. Understandings of Basic Genetics in the United States: Results from a National Survey of Black and White Men and Women. *Public Health Genomics* 13, 467. <https://doi.org/10.1159/000293287>
- Christianson, A., Howson, C.P., Modell, B., 2005. March of Dimes: global report on birth defects, the hidden toll of dying and disabled children. *March Dimes Glob. Rep. Birth Defects Hidden Toll Dying Disabl. Child.*
- Cuéllar Lázaro, C., Mallo Lapuerta, A.M., Córdova Martínez, A., 2014. Designing a multilingual dictionary of genetic terms (English, French, German and Spanish) for the European Portal Eurogene and the International Scientific Community. <https://doi.org/10.14198/jhse.2014.94.04>
- Graham, S., Brookey, J., 2008. Do Patients Understand? *Perm. J.* 12, 67–69.
- Haga, S.B., Barry, W.T., Mills, R., Ginsburg, G.S., Svetkey, L., Sullivan, J., Willard, H.F., 2013. Public Knowledge of and Attitudes Toward Genetics and Genetic Testing. *Genet. Test. Mol. Biomark.* 17, 327–335. <https://doi.org/10.1089/gtmb.2012.0350>
- Kaphingst, K.A., Blanchard, M., Milam, L., Pokharel, M., Elrick, A., Goodman, M.S., 2016. Relationships Between Health Literacy and Genomics-Related Knowledge, Self-Efficacy, Perceived Importance, and Communication in a Medically Underserved Population. *J. Health Commun.* 21, 58–68. <https://doi.org/10.1080/10810730.2016.1144661>

- Kaplan, S.H., Greenfield, S., Ware, J.E., 1989. Assessing the effects of physician-patient interactions on the outcomes of chronic disease. *Med. Care* 27, S110-127.
- Kickbusch, I.S., 2001. Health literacy: addressing the health and education divide. *Health Promot. Int.* 16, 289–297. <https://doi.org/10.1093/heapro/16.3.289>
- Kromberg, J.G.R., Wessels, T.-M., Krause, A., 2013. Roles of genetic counselors in South Africa. *J. Genet. Couns.* 22, 753–761. <https://doi.org/10.1007/s10897-013-9606-2>
- Lanie, A.D., Jayaratne, T.E., Sheldon, J.P., Kardia, S.L.R., Anderson, E.S., Feldbaum, M., Petty, E.M., 2004. Exploring the public understanding of basic genetic concepts. *J. Genet. Couns.* 13, 305–320.
- Lillie, S.E., Brewer, N.T., O'Neill, S.C., Morrill, E.F., Dees, E.C., Carey, L.A., Rimer, B.K., 2007. Retention and use of breast cancer recurrence risk information from genomic tests: the role of health literacy. *Cancer Epidemiol. Biomark. Prev. Publ. Am. Assoc. Cancer Res. Cosponsored Am. Soc. Prev. Oncol.* 16, 249–255. <https://doi.org/10.1158/1055-9965.EPI-06-0525>
- Loebner, S., 2013. *Understanding Semantics*, 2 edition. ed. Routledge, New York, NY.
- Mazurek, D., Wyka, J., 2015. Down syndrome--genetic and nutritional aspects of accompanying disorders. *Rocz. Panstw. Zakl. Hig.* 66, 189–194.
- Meuter, R.F.I., Gallois, C., Segalowitz, N.S., Ryder, A.G., Hocking, J., 2015. Overcoming language barriers in healthcare: A protocol for investigating safe and effective communication when patients or clinicians use a second language. *BMC Health Serv. Res.* 15, 371. <https://doi.org/10.1186/s12913-015-1024-8>
- Micheletto, M.R.D., Valerio, N.I., Fett-Conte, A.C., 2013. Effects of a genetic counseling model on mothers of children with down syndrome: a Brazilian pilot study. *J. Genet. Couns.* 22, 784–794. <https://doi.org/10.1007/s10897-013-9619-x>
- Molster, C., Charles, T., Samanek, A., O'Leary, P., 2009. Australian study on public knowledge of human genetics and health. *Public Health Genomics* 12, 84–91. <https://doi.org/10.1159/000164684>

Ormond, K.E., Laurino, M.Y., Barlow-Stewart, K., Wessels, T.-M., Macaulay, S., Austin, J., Middleton, A., 2018. Genetic counseling globally: Where are we now? *Am. J. Med. Genet. C Semin. Med. Genet.* 178, 98–107. <https://doi.org/10.1002/ajmg.c.31607>

Penn, C., 2007. Factors affecting the success of mediated medical interviews in South Africa : review article. *Curr. Allergy Clin. Immunol.* 20, 66–72.

Resta, R., Biesecker, B.B., Bennett, R.L., Blum, S., Estabrooks Hahn, S., Strecker, M.N., Williams, J.L., 2006. A New Definition of Genetic Counseling: National Society of Genetic Counselors' Task Force Report. *J. Genet. Couns.* 15, 77–83. <https://doi.org/10.1007/s10897-005-9014-3>

Sheets, K.B., Crissman, B.G., Feist, C.D., Sell, S.L., Johnson, L.R., Donahue, K.C., Masser-Frye, D., Brookshire, G.S., Carre, A.M., Lagrave, D., Brasington, C.K., 2011. Practice guidelines for communicating a prenatal or postnatal diagnosis of Down syndrome: recommendations of the national society of genetic counselors. *J. Genet. Couns.* 20, 432–441. <https://doi.org/10.1007/s10897-011-9375-8>

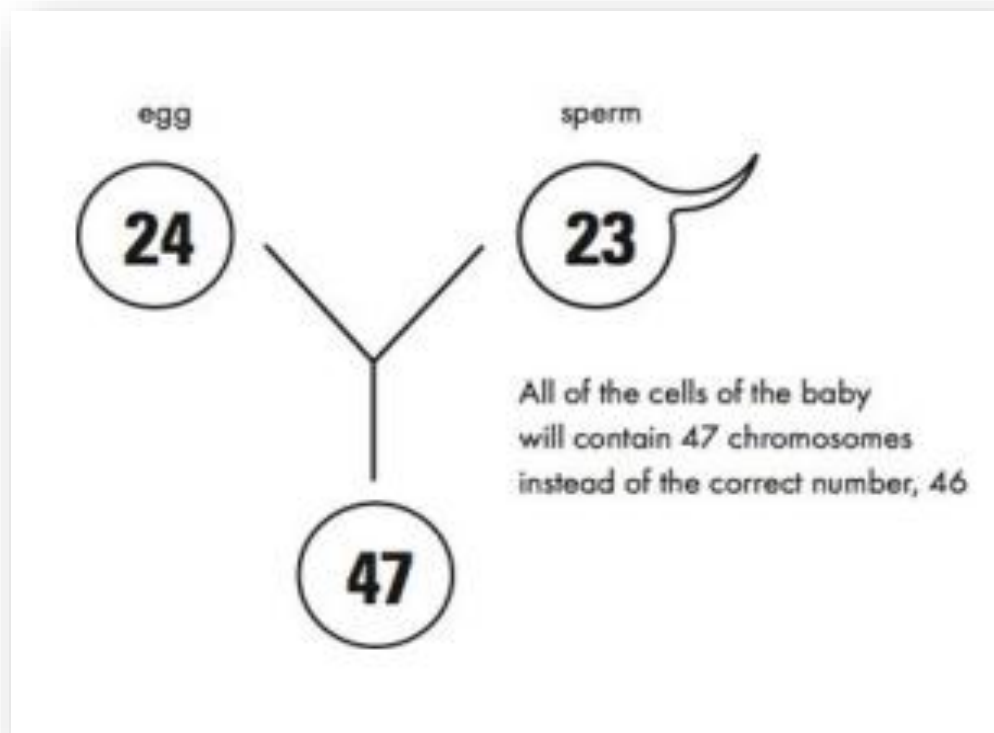
Skirton, H., Cordier, C., Ingvaldstad, C., Taris, N., Benjamin, C., 2015. The role of the genetic counsellor: a systematic review of research evidence. *Eur. J. Hum. Genet. EJHG* 23, 452–458. <https://doi.org/10.1038/ejhg.2014.116>

Somer, M., Mustonen, H., Norio, R., 1988. Evaluation of genetic counselling: recall of information, post-counselling reproduction, and attitude of the counselees. *Clin. Genet.* 34, 352–365.

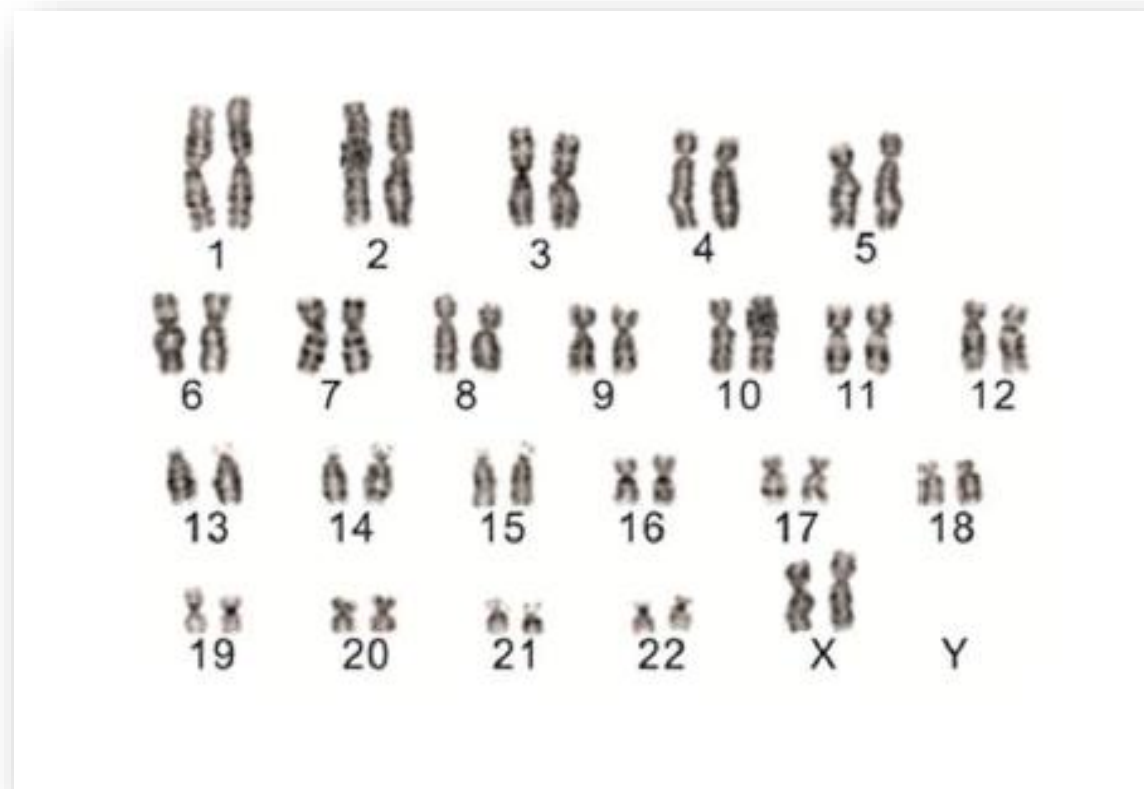
Uhlmann, W.R., Schuette, J.L., Yashar, B., 2011. *A Guide to Genetic Counseling*. John Wiley & Sons.

World Health Organization, 1998. Health promotion glossary.

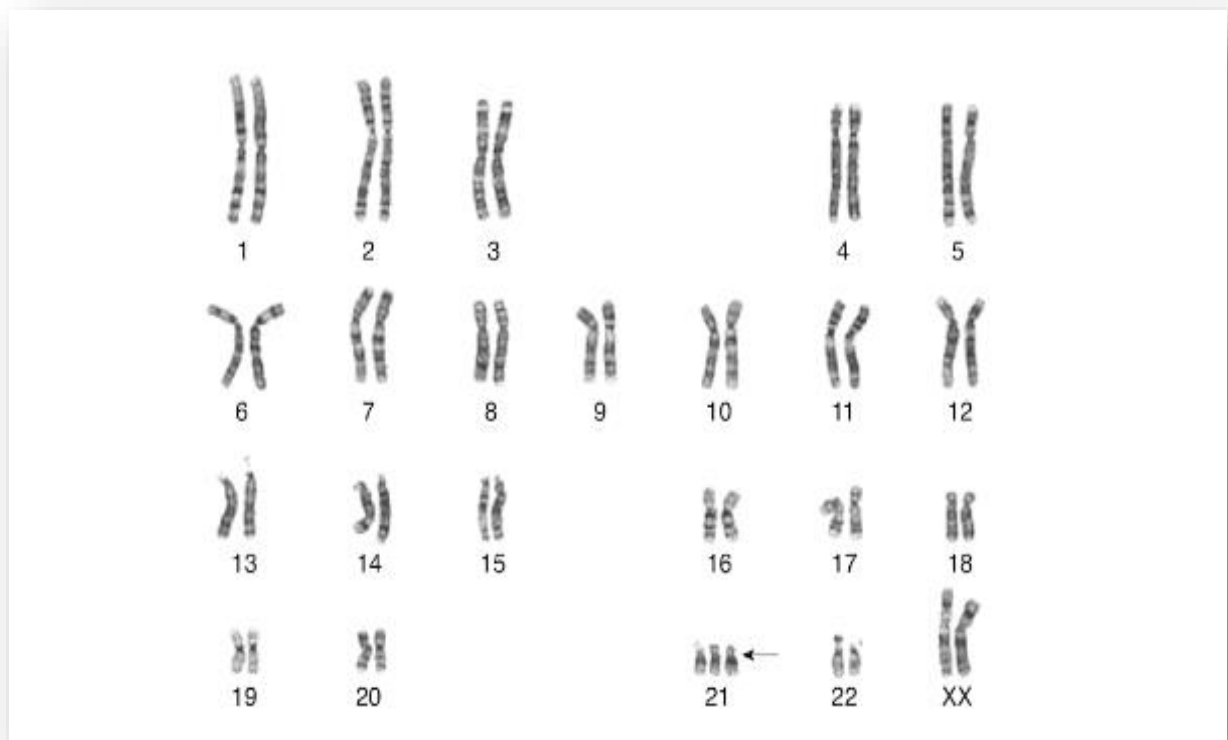
Appendix A



Appendix B



Appendix C



Assessing patient recall, understanding, and vocabulary of genetic concepts, in a South African setting

Questionnaire

Visit date: _____

SECTION 1

Demography

1.1 Age of participant (years):

1.2 Relationship to child with Down syndrome:

Mother Father

Other _____

1.3 Marital status:

Single Widowed

Married/Cohabiting Separated/Divorced

1.4 Total number of years of formal education:

1.5 Highest level of education attended:

No school attended	☐	Professional/technical training	☐
Primary	☐	University	☐
Secondary	☐		

1.6 Did the participant study biology/life sciences?

Yes ☐ No ☐

1.6.1 If Yes, Where? _____

1.7 What is the participant's occupation?

1.8 Does the participant's household have or own any of the following: *(tick all that apply)*

Electricity	☐	Cell phone	☐	Bicycle	☐
Radio	☐	Personal computer	☐	Motorcycle	☐
Television	☐	Farm animals	☐	Car/truck/tractor	☐
Refrigerator	☐	Agricultural land	☐		

1.9 What is the participant's first language?

Tsonga	☐	Sotho	☐	Venda	☐
Ndebele	☐	Swati	☐	Xhosa	☐

Zulu

Tswana

Northern Sotho

Other: _____

SECTION 2

Genetic recall and understanding

2.1 Referring to Picture A, did you see this during the genetic counselling session?

Yes ☐ No ☐

2.1.1 If **yes**, explain what you understood about picture A?

<u>Expected answers</u>	<u>Tick if mentioned</u>	<u>Score</u>
Egg	☐	[1]
Sperm	☐	[1]
23 chromosomes from mother	☐	[1]
23 chromosomes from father	☐	[1]
Normal fertilisation	☐	[1]
46 chromosomes in baby	☐	[1]

Notes: _____

Question 2.1 score

/6

2.2 Referring to Picture B, did you see this during the genetic counselling session?

Yes

☐

No

☐

2.2.1 **If yes**, what is it?

Expected answer

Tick if mentioned

Score

A picture of chromosomes/genetic information

☐

[1]

2.2.2 How many of these structures (chromosomes) are we meant to have?

Expected answer

Tick if mentioned

Score

48

☐

[1]

2.2.3 Where do these structures (chromosomes) come from?

Expected answer

Tick if mentioned

Score

From our father and mother

☐

[1]

2.2.4 How many of these structures (chromosomes) do we get from each parent?

Expected answer

Tick if mentioned

Score

23 from each parent/half from each parent

☐

[1]

2.2.5 What do these structures (chromosomes) do?

Expected answer

Tick if mentioned Score

They instruct our bodies how to look and function ☐ [1]

Notes: _____

Question 2.2 score

/5

2.3 Referring to Picture C, did you see this during the genetic counselling session?

Yes ☐ No ☐

2.3.1 **If yes**, what do you understand about the picture?

<u>Expected answers</u>	<u>Tick if mentioned</u>	<u>Score</u>
Egg	☐	<i>[1]</i>
Sperm	☐	<i>[1]</i>
Extra chromosome/information in the egg	☐	<i>[1]</i>
23 chromosomes from father	☐	<i>[1]</i>
24 chromosomes from mother	☐	<i>[1]</i>
Fertilisation	☐	<i>[1]</i>
Baby with 47 chromosomes	☐	<i>[1]</i>

Notes: _____

Question 2.3 score

/7

2.4 Referring to Picture D, did you see this during the genetic counselling session?

Yes

☐

No

☐

2.4.1 **If yes**, what is different about this picture compared Picture B?

Expected answers

Tick if mentioned

Score

Extra chromosome 21

☐

[1]

49 chromosomes in total

☐

[1]

2.4.2 What does this cause?

Expected answer

Tick if mentioned

Score

Down syndrome

☐

[1]

Notes: _____

Question 2.4 score

/3

2.5 Referring to Picture E, did you see/hear this word during the genetic counselling session?

Yes ☐ No ☐

2.5.1 **If yes**, what does Trisomy 21 mean?

<u>Expected answers</u>	<u>Tick if mentioned</u>	<u>Score</u>
Tri = 3 copies	☐	<i>[1]</i>
Somy = chromosomes	☐	<i>[1]</i>
21 = chromosome number 21	☐	<i>[1]</i>

2.5.2 What is another name for Trisomy 21?

<u>Expected answer</u>	<u>Tick if mentioned</u>	<u>Score</u>
Down syndrome	☐	<i>[1]</i>

Notes: _____

Question 2.5 score

/4

2.6 What does Down syndrome cause?

(Write down what clinical symptoms or features the participant lists)

Delayed milestones

[1]

Facial features

[1]

Cardiac problems

[1]

Intellectual disability

[1]

Notes:

Question 2.6 score

/4

SECTION 3

Genetic vocabulary

3.3 In your own home language, what words would you use for the following genetic words:

3.3.1 Genetics

3.3.2 Chromosomes

3.3.3 Gene

3.3.4 Sperm

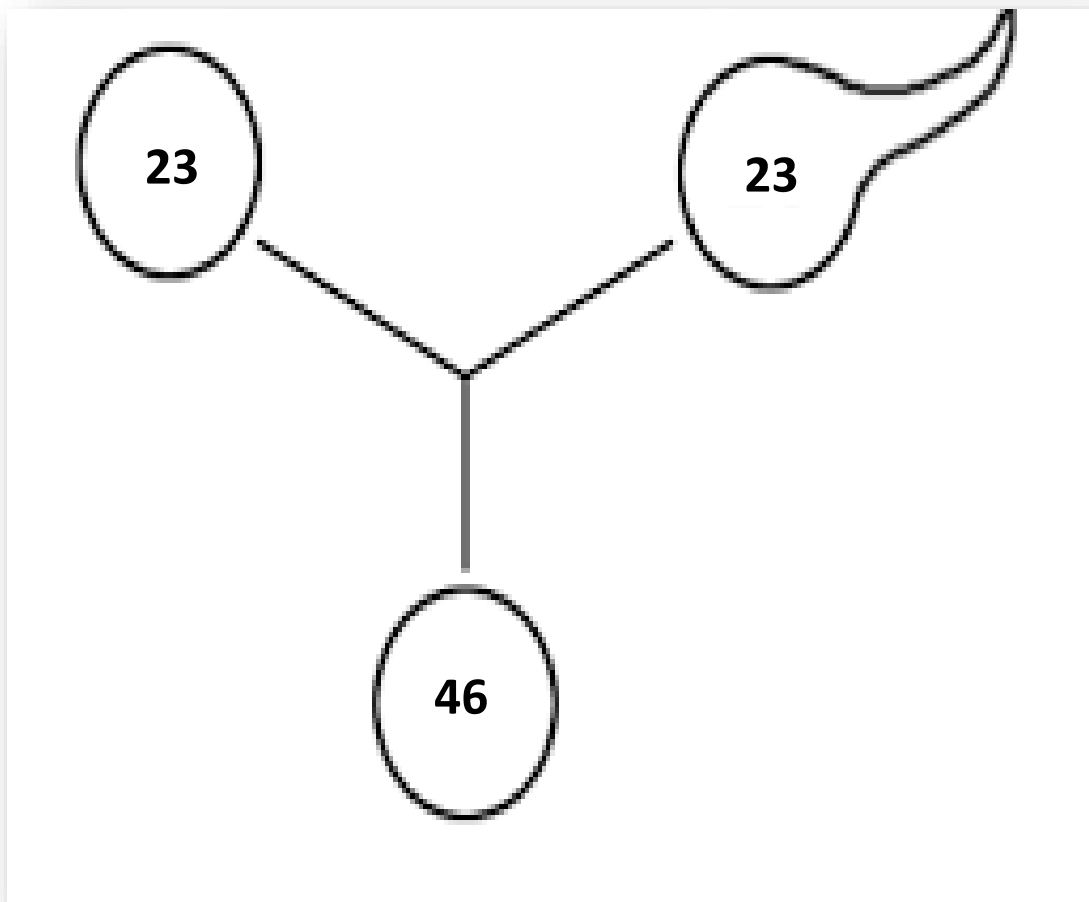
3.3.5 Egg

3.3.6 Cell

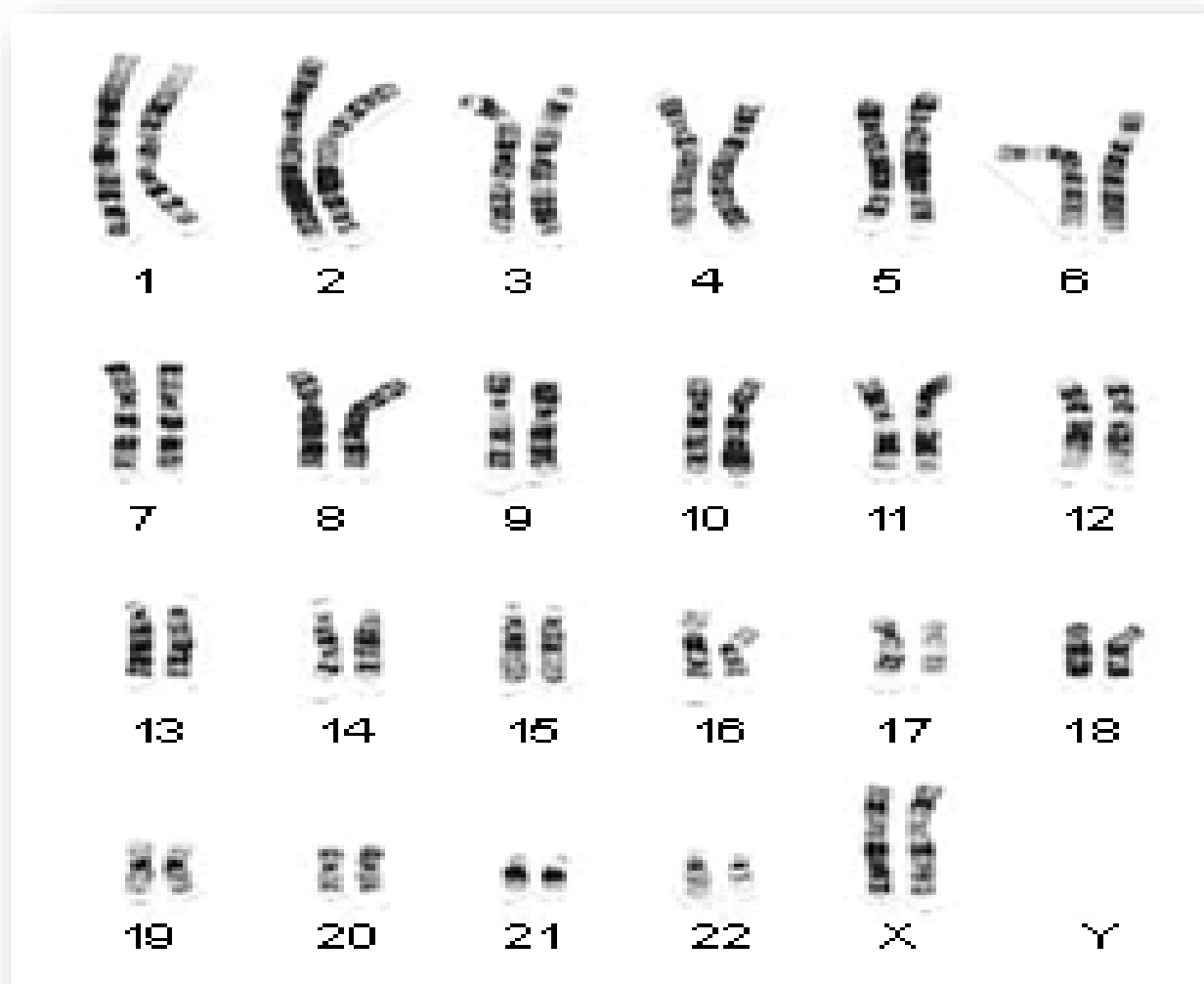
3.3.7 Inherited/inheritance

3.4 What names do your community use for people with Down syndrome?

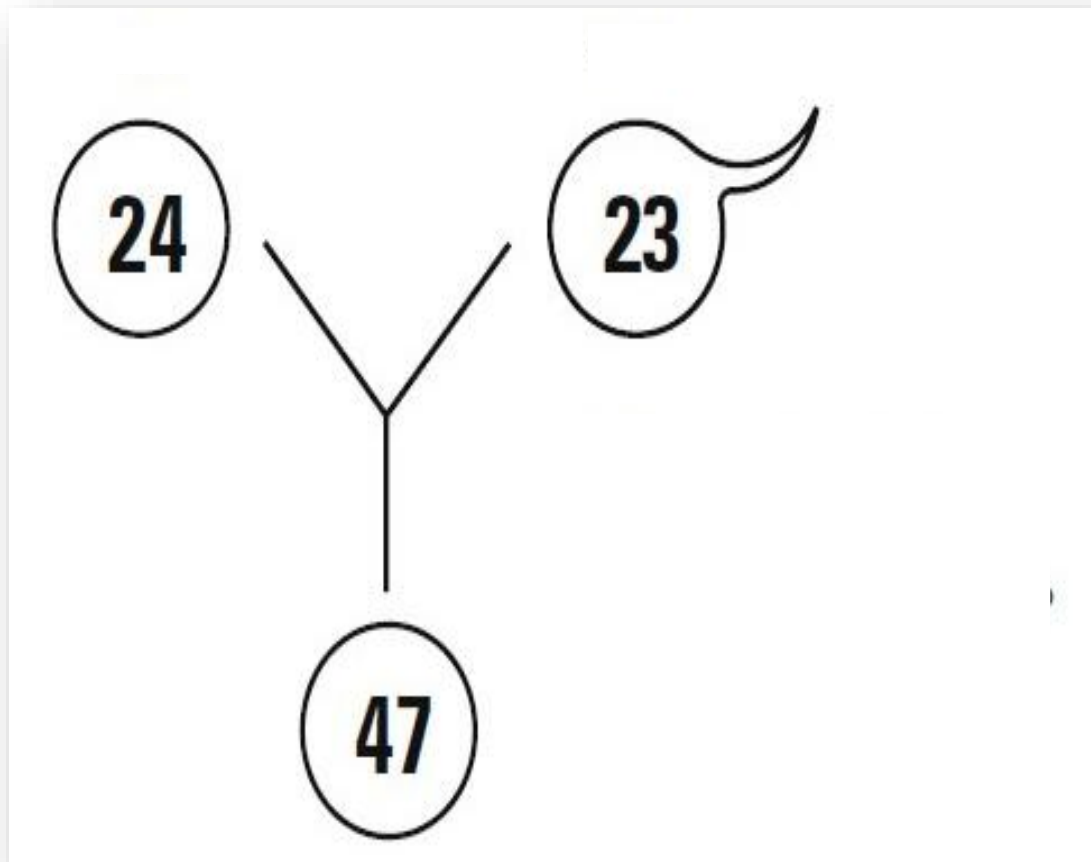
Picture A



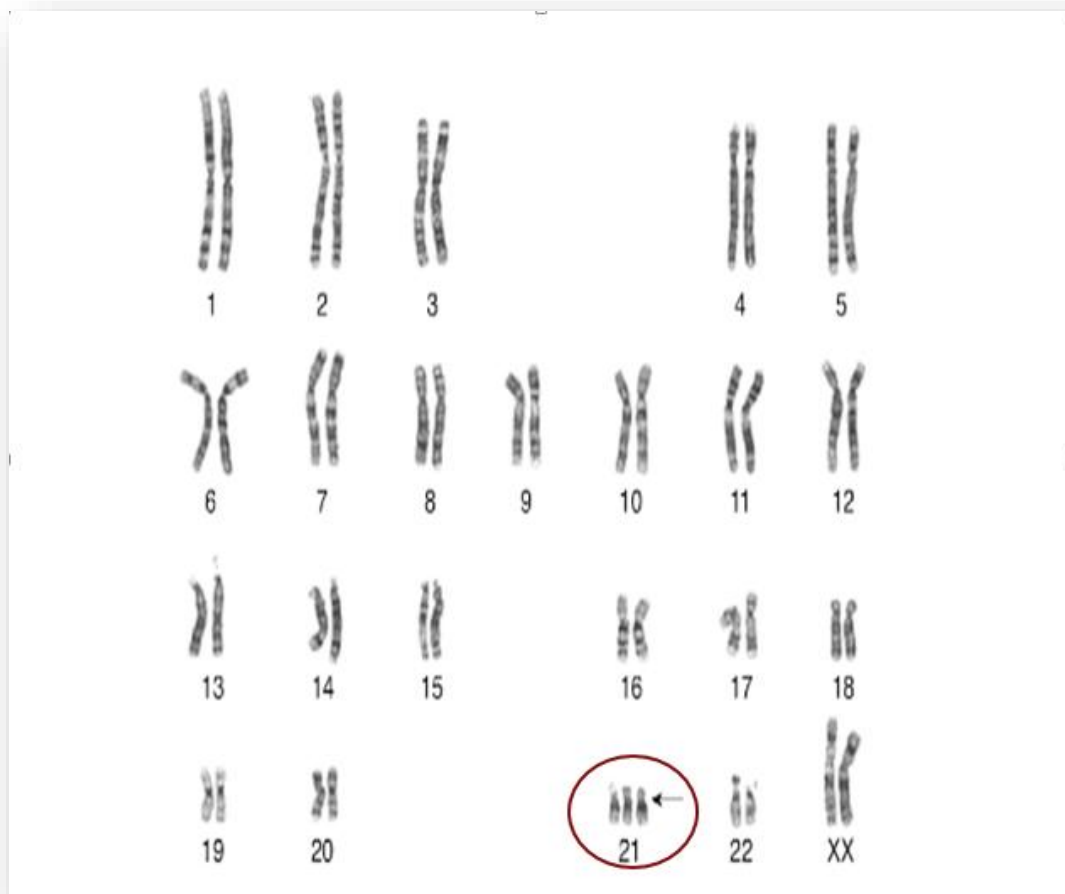
Picture B



Picture C



Picture D



Picture E

Trisomy 21

Appendix E

INFORMATION SHEET

Study title: Assessing patient recall, understanding, and vocabulary of genetic concepts, in a South African setting.

Good day,

My name is Barry Shingwenyana and I am an MSc (Med) Genetic Counselling student in the Division of Human Genetics, at the National Health Laboratory Service (NHLS) and the University of the Witwatersrand (WITS). As part of my degree I am conducting a research project to assess how well patients attending genetic counselling recall and understand the genetic concepts discussed with them in a session. I also want to ask these patients what words they would use in their own home languages for some of the genetic terms used in the sessions. The results of this study will be used to develop a suitable South African specific genetic vocabulary which could be used by South African genetic counsellors which may assist in a better understanding of genetics for patients.

I invite you to participate in the study. This study involves a researcher-administered questionnaire; I will conduct the interview. There are no risks posed to you and there will be no direct benefits. Your information will remain anonymous. Participation is voluntary, and should you choose not to participate this will have no negative impact on you or your child's treatment and management.

Please feel free to contact us if you require further information.

Contact details of researcher:

Researcher: Mr. Barry Shingwenyana | 011 489 9216 | barry.shingwenyana@nhls.ac.za

Supervisor 1: Ms. Merlyn Glass | 011 489 9335 | merlyn.glass@nhls.ac.za

Supervisor 2: Dr. Shelley Macaulay | 011 489 9243 | shelley.macaulay@nhls.ac.za

Contact details of research ethics committee:

Chair: Prof Clem Penny | 011 717 2301 | Clement.Penny@wits.ac.za

Administrator: Ms. Zanele Ndlovu | 011 717 1252/2700/1234/2656 | zanele.ndlovu@wits.ac.za

Appendix F

INFORMED CONSENT

With regard to the research study entitled “**Assessing patient recall, understanding, and vocabulary of genetic concepts, in a South African setting**” I (Name), _____ agree to participate by answering a questionnaire and understand that the results will be analysed by the researcher and his supervisors.

- I agree to the use of the data collected from the questionnaire to be used for the aforementioned project.
- I acknowledge that I have read and understood the information document.
- I understand that findings from this study will not be reported back to me and that results may be published within accredited journals.
- I understand that my information will remain anonymous unless required by law.

Participant Signature _____ Date _____

Witness Signature _____ Date _____

Record ID