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JOHANNESBURG

# The Utilisation of Genetic Counselling Services Amongst Prenatal Healthcare Providers in Gauteng, South Africa

by

Megan Duvenhage

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 for the degree of Master of Science in Medicine (Genetic Counselling)

Johannesburg, 2024

# Contents

## Declaration

I, Megan Duvenhage, declare 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 at the University of the Witwatersrand, Johannesburg. It has not been submitted before for any degree or examination at any other University.

I, Megan Duvenhage, as principal investigator, had full access to the original data generated through this study and take responsibility for the integrity of the data. All authors contributed to the interpretation of the data and take responsibility for the accuracy of the data analysis.

Megan Duvenhage

26 February 2024, in Johannesburg

# Contribution of the candidate to this paper

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

I, Megan Duvenhage, student number 1076441, 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: 26 February 2024

Name of Supervisor:

Ms Monica Araujo

Signature of Supervisor

Date: 26 February 2024

Name of Co-supervisor:

Mrs Katryn Fourie

Signature of Co-supervisor

Date: 26 February 2024

**Agreement by Co-Authors:**

By signing this declaration, the co-authors listed below agree to the use of the article(s) by the student as part of her Research Project.

**Article Title:** The Utilisation of Genetic Counselling Services Amongst Prenatal Healthcare Providers in Gauteng, South Africa

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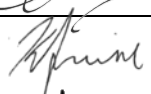
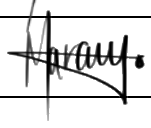
| 2nd Author | Katryn Fourie |  | 26 February 2024 |
| Last Author | Monica Araujo |  | 26 February 2024 |

# Dedication

*This research report is dedicated to my mother, Erna Duvenhage. I would never have made it this far without you. Thank you for your unwavering support and belief in me through every chapter of my life. Thank you for always pushing me to be the best version of myself and being there for the stumbles along the way.*

*You are the reason for everything.*

## Presentations arising from this research

1. World Congress on Genetic Counselling 2023, 25 – 27th October, Hinxton Hall Conference Centre, Wellcome Genome Campus, United Kingdom (virtual poster presentation).
2. Genetic Counselling South Africa Annual Research Showcase, 20 November 2023 (oral presentation).
3. Division of Human Genetics Departmental Seminar, 8th May 2024, National Health Laboratory Service (oral presentation).
4. RareX 2024, 14 – 17th February 2024, Indaba Hotel and Conference Centre, Johannesburg, South Africa (oral presentation).

# Abstract

Congenital anomalies and disorders, many being genetic, continue to have high prevalence and mortality rates globally. Prenatal healthcare providers possess the necessary skills to identify these cases before birth and refer patients for genetic counselling. This study aimed to establish the utilisation of genetic counselling services and insights into the perceptions of genetic counselling amongst prenatal healthcare providers in Gauteng, South Africa. By assessing the utilisation of genetic counselling, barriers and facilitators to referrals were highlighted, and recommendations to improve service provisions in the prenatal sector were made. An electronic survey adapted from Thom and Haw (2021) was sent to prenatal healthcare providers in both the public and private healthcare sectors. A total of 54 participants were included in this study. Results show that roughly 74% of participants are able to refer to genetic counselling services, but only 57% had made use of the service. None of the participants were able to identify all appropriate reasons for referral to genetic counselling correctly, and only 24% of participants understood the responsibilities of a genetic counsellor. Misconceptions regarding the scope of practice of genetic counsellors and uncertainties surrounding the referral process were the most significant barriers to referrals. The study revealed that although prenatal healthcare providers in Gauteng are using genetic counselling services, they are not fully utilising the service due to a lack of knowledge surrounding the profession's services. Therefore, there is a need for educational resources to bridge the knowledge gap and improve prenatal healthcare in Gauteng, South Africa.

# Acknowledgements

I would like to thank my supervisors Ms. Monica Araujo and Mrs. Katryn Fourie for their guidance and support throughout this journey. Thank you for believing in me and for always showing excitement about this project. Thank you for all your ability to help me while simultaneously showing me that I am capable. Your guidance and advice have been invaluable.

To every member of the Clinical and Counselling Section in the Division of Human Genetics, National Health Laboratory Service and University of the Witwatersrand, thank you for everything you have taught, and continue to teach me. To Dr Robyn Kerr, one chat with you changed the trajectory of my life and I could not be more thankful.

Thank you to the University of the Witwatersrand for funding my studies through the Postgraduate Merit Award.

To my fellow genetic counselling student/ intern Sebastian Barnard, thank you for your incredible support, patience, and friendship through every high and low we have faced over the past two years. I would not trade you for any other "trench buddy". A special thank you to Monica Araujo, for always having an open door and an open heart regardless of how busy you may be. You are not only a remarkable role model but a wonderful friend.

Lastly, to my family, thank you for all the love and support. Ma, there are no words to express my appreciation for every opportunity you have provided me. To my friends that became family, Gabs, Kieran, Lama, Megan, and Romi, thank you for every coffee, ice-cream, and vent session.

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# List of abbreviations

| | |
|---|---|
| CHBAH | Chris Hani Baragwanath Academic Hospital |
| CMJAH | Charlotte Maxeke Johannesburg Academic Hospital |
| ERH | Edenvale Regional Hospital |
| MO | Medical Officer |
| NHLS | National Health Laboratory Service |
| OBGYN | Obstetrician and Gynaecologist |
| RMMCH | Rahima Moosa Mother and Child Hospital |
| SASOG | South Africa Society of Obstetrics and Gynaecology |
| SASUOG | South African Society of Ultrasound in Obstetrics and Gynaecology |
| SOMSA | Society of Midwives of South Africa |
| TMH | Tambo Memorial Hospital |
| TMRH | Thelle Mogoerane Regional Hospital |
| WITS | The University of the Witwatersrand |

# Research report in the format of a “submissible” paper

This work has been written up as a paper for submission to The Journal of Genetic Counselling. The author guidelines, such as abstract, referencing and word count, have been adhered to and can be found in Appendix H. Tables and Figures have been included in the text for ease of reference.

# The Utilisation of Genetic Counselling Services Amongst Prenatal Healthcare Providers in Gauteng, South Africa

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## Abstract for paper submission

Congenital anomalies and disorders, many of which are genetic, continue to have high prevalence and mortality rates globally, particularly in developing countries. Prenatal healthcare providers possess the necessary skills to identify these cases before birth and refer patients for genetic counselling. The general lack of knowledge on the field of genetic counselling has resulted in low numbers of referrals to genetic counselling from many healthcare sectors. This study aimed to establish the utilisation of genetic counselling services and insights into the perceptions of genetic counselling amongst prenatal healthcare providers in the Gauteng Province, South Africa. By assessing the utilisation of genetic counselling, barriers and facilitators to referrals were noted, and recommendations to improve service provisions in the prenatal sector were made. An electronic survey adapted from Thom and Haw (2021) was sent to prenatal healthcare providers in both the public and private healthcare sectors. A total of 54 participants were included in this study. Results show that approximately 74% of participants are able to refer to genetic counselling services, but only 57% had made use of the service. None of the participants were able to identify all appropriate reasons for referral to genetic counselling correctly, and only 24% of participants understood the responsibilities of a genetic counsellor. Approximately 6% of participants felt comfortable with their knowledge of genetic counselling. Misconceptions surrounding the scope of practice of genetic counsellors and uncertainties surrounding the referral process were the most significant barriers to referrals. While this study showed prenatal healthcare providers in Gauteng are utilising genetic counselling services, the service is not being utilised to its full potential due to insufficient knowledge of the services offered by the profession. This demonstrates the need for educational resources to bridge the knowledge gap and improve prenatal healthcare in Gauteng, South Africa.



**Keywords:** genetic counselling, education, awareness, attitudes, prenatal diagnosis, utility

**What is known about the topic**

Studies have been done internationally and locally to assess the awareness of genetic services and their utilisation by various healthcare sectors. However, there have been no studies done focusing on the uptake of genetic counselling among prenatal healthcare providers, within South Africa or internationally.

**What this paper adds to the topic**

This paper demonstrates the utilisation and extent of knowledge of genetic counselling services amongst prenatal healthcare providers in Gauteng, South Africa. Additionally, it describes the barriers and facilitators to patient referrals and provides recommendations to enhance the service.

# 1 Introduction

Congenital anomalies remain a significant problem both globally and in South Africa, with birth incidents of approximately 3% and 7% respectively (Clarke, 2020, p. 111; Malherbe et al., 2015). Congenital disorders occur in one in every fifteen live births in South Africa (Malherbe et al., 2016). The prevalence and mortality rates of congenital anomalies and disorders remain high even though up to 70% of these can be prevented, improved or effectively managed with relatively inexpensive measures (Malherbe et al., 2015). Interventions put into place to decrease the burden of congenital disorders must look at both prevention of, and care for those affected. A 3-tiered process approaches prevention and interventions at the primary, secondary and tertiary levels. Primary prevention involves implementing basic reproductive health care such as vitamin and folic acid supplementation in pregnant patients. Secondary prevention is aimed at decreasing the number of affected children born by improving prenatal screening and diagnosis, education on avoidance of teratogenic substances and discussing termination of a pregnancy. Tertiary prevention is aimed at early detection after birth to improve medical interventions and treatments and provide counselling and psychosocial support to affected families (Malherbe et al., 2016).

Considering that 80% of congenital abnormalities may be genetic or partially genetic, medical genetic services, including genetic counselling, are in place to prevent, detect and care for patients with congenital disorders (Malherbe et al., 2016). These services allow patients to gain information and have necessary conversations regarding basic reproductive healthcare, prenatal screening and diagnosis, pregnancy options, and post-natal management and care. It is recommended that medical genetics and genetic counselling services be integrated into the 3-tiered approach to healthcare as part of a comprehensive healthcare programme to lessen the burden of inherited conditions (Kromberg & Krause, 2013).

As previously stated, prevention of congenital disorders can occur at various levels of care, all of which can be facilitated by a genetic counsellor due to their knowledge and training (Malherbe et al., 2016). Genetic counsellors are healthcare workers uniquely equipped to explain complicated information and genetic concepts to patients in a way that they understand and allows them to feel supported and empowered. Genetic counsellors provide a holistic approach to patient care through in-depth discussions with patients not only regarding the genetic condition at hand, but also the emotional toll the situation may take on the patient and their family (Catapano et al., 2022). This enables patients to take control of their healthcare and make informed choices that best suit them (Abacan et al., 2019; Kromberg, Wessels, et al., 2013; McAllister et al., 2008).

Genetic counselling is still a young profession within South Africa, with formal clinics in the state healthcare systems in Johannesburg and Cape Town starting in the late 1960s (Kromberg & Krause, 2013; Malherbe et al., 2016). South Africa has established genetic services and clinics available to the state healthcare system in four of the nine provinces, namely Gauteng, the Western Cape, the Free State and Kwa-Zulu Natal (Kromberg, Wessels, et al., 2013). These services include medical genetics, genetic counselling and genetic testing in prenatal, paediatric and adult settings (Kromberg, Sizer, et al., 2013). These services are also offered in the private sector at various independent and state-run organisations.

Although fetal medicine specialists, obstetricians, gynaecologists and sonographers perform sonography during pregnancy and identify likely abnormalities, it is often the role of genetic counsellors to propose and discuss appropriate genetic testing, obtain informed consent regarding any invasive procedures and provide counselling to the family in a comprehensible manner (Yotsumoto et al., 2016). Therefore, referrals to genetic services must be available to assist in prenatal cases (Diamonstein et al., 2018; Scelsa, 2022). While other healthcare providers may feel they are able to successfully provide genetic information to their patients,

genomic education curricula across African institutions at all levels is low. This has resulted in low genomic literacy not only in patients, but also in the majority of healthcare providers (Mboowa & Sserwadda, 2019). Therefore, patients not seen by genetic counsellors may not receive comprehensive, non-directive information which may hinder their ability to make informed decisions (Wessels et al., 2015). Genetic services may be accessed through referrals from healthcare providers at tertiary/ provincial hospitals in the state system, or through specialist referrals in the private sector. Many of the state patients are first seen at a local clinic which then refers them to a tertiary/ provincial hospital.

There are currently less than 30 registered and practicing genetic counsellors in South Africa, with only five employed in the Clinical and Counselling Unit of the Division of Human Genetics at the NHLS and the University of the Witwatersrand in Gauteng (Abacan et al., 2019; K. Fourie, personal communication, October 12, 2023). With this being the province's only state clinical genetic service offered by clinical geneticists and genetic counsellors, and one of a few in the country, the overall utilisation of genetic counselling services within the province is expected to be low due to limited access to the service. The provision of services should be improved to ensure proper patient management, however, this cannot be done without further understanding on what the service is currently being utilised for (Kromberg, Wessels, et al., 2013; Thom & Haw, 2021).

International and local studies have assessed the awareness of genetic counselling services and their utilisation by other sectors of the medical community. These studies have concluded that globally, healthcare professionals who do not work in genetics need more awareness and understanding of the work done by genetic counsellors and the value they provide (Aalfs et al., 2003; Baars et al., 2005; Suther & Goodson, 2003). The lack of knowledge of genetics and misconceptions surrounding the role of a genetic counsellor in the medical fraternity are likely the most significant barriers in referring a patient to genetic counselling (Aalfs et al., 2003;

Suther & Goodson, 2003; Thom & Haw, 2021). Not only do practitioners have insufficient genetics knowledge, but they need to be made aware of the types of patients eligible for referral to genetic services (Delikurt et al., 2015). Thom and Haw (2021) performed a similar study in South Africa to assess the awareness of genetic counselling services among allied healthcare professionals. This study concluded that allied healthcare professionals had poor awareness of genetic counselling services and the referral process and thus made little use of them (Thom & Haw, 2021). The findings from the study by Thom and Haw (2021) were consistent with similar international studies (Aalfs et al., 2003; Baars et al., 2005; Delikurt et al., 2015). Although there were similarities between local and international allied healthcare professionals regarding the awareness of genetic counselling, it is not necessarily possible to extrapolate these findings to include prenatal healthcare providers due to the differences in their training and practice fields.

# 2 Purpose of this study

This study aimed to establish the utilisation of genetic counselling services amongst prenatal healthcare providers in Gauteng, South Africa, as well as elucidate the perceptions of genetic counselling and its value among these individuals. By assessing the utilisation of genetic counselling, we were able to make recommendations to improve the service provided in the prenatal sector in Gauteng, South Africa.

# 3 Methods

## 3.1 Participants

Prenatal healthcare providers in Gauteng were recruited between April 2023 to September 2023 through six different state hospitals, three organisational mailing lists and several private practices. The state hospitals include; Chris Hani Baragwanath Academic Hospital (CHBAH), Charlotte Maxeke Johannesburg Academic Hospital (CMJAH), Edenvale Regional Hospital

(ERH), Rahima Moosa Mother and Child Hospital (RMMCH), Tambo Memorial Hospital (TMH) and Thelle Mogoerane Regional Hospital (TMRH). The organisational databases used belong to the South Africa Society of Obstetrics and Gynaecology (SASOG), the South African Society of Ultrasound in Obstetrics and Gynaecology (SASUOG), and the Society of Midwives of South Africa (SOMSA). Prenatal healthcare providers in private practices and those known to the genetics department at the NHLS were recruited in their private capacity.

The inclusion criteria for this study were professionals based in Gauteng who, at the time of the study, were practising as fetal medicine specialists, obstetricians, gynaecologists, sonographers, midwives, or fertility specialists. Specialising trainees, specifically registrars and medical officers (MOs) who had been training in these fields for more than six months, were included in the study. Six months of training should be sufficient to become familiar with indications for referral as well as the referral process. An email containing the link to the study survey, a participant information sheet (Appendix B), and informed consent (Appendix C) were distributed to the heads of the Department of Obstetrics and Gynaecology at each state hospital as well as the various organisational databases and prenatal healthcare providers in private practice. Participants were also recruited through departmental meetings personally attended by the researcher. In this study, the email containing the study link also asked potential participants to forward the email to other prenatal healthcare providers, thereby making use of snowball sampling. This sampling method relies on a recruited participant assisting the researcher in identifying other suitable potential participants (Cambridge University Press & Assessment, 2023). All participants had to give informed consent before being allowed to proceed with the study survey. Anonymity of the respondents was ensured as no identifying information was asked and no email addresses were collected. Participants were also given to option of not identifying their place of work.

Ethics clearance (Appendix E) was obtained from the Human Research Ethics Committee (Medical) of the University of the Witwatersrand (Ethics Clearance Certificate no. M230268).

## 3.2 Instrumentation

The survey design was based on the survey used by Thom and Haw (2021) in their study on the awareness of genetic counselling services among allied healthcare professionals in South Africa (Thom & Haw, 2021). Although the study by Thom and Haw (2021) looked at a different group of healthcare providers, it also aimed to determine awareness and utilisation of genetic counselling services within a South African context. Questions were tailored based on their relevance to the field of genetic counselling in prenatal healthcare rather than allied healthcare professionals. The survey was tested on a group of five individuals, none of which were genetic counsellors, within the department of human genetics at the NHLS. This helped identify any questions which needed clarification or re-wording. Minor changes were made before the survey was sent to participants. All questions in the survey were mandatory to avoid the submission of incomplete surveys.

The survey (Appendix D) included three sections; (1) the demographics and professional practice of the participant, (2) the participants' referrals to genetic counselling services, and (3) the participants' understanding and knowledge of genetic counselling services. The survey was designed to be an electronic survey in the form of a Google Form. This was to increase the ease of use by participants.

## 3.3 Data analysis

The participants were divided into subgroups based on their profession. If a participant worked in more than one field (e.g., obstetrics and fetal medicine), the field that required the most training (in this example, fetal medicine) was assigned. Obstetricians and gynaecologists were grouped together under “OBGYN”. The Excel statistical package was used to generate descriptive statistics, namely means and standard deviations. An analysis of variance

(ANOVA) test was performed to assess the normality of the demographic data and of the number of referrals. This showed a normal distribution with no statistically significant differences between the numbers of years participants had been in their current practice or the number of referrals (Due to this and the small sample size, all responses were then analysed as one group. Descriptive statistics were used to analyse the sections on knowledge and utilisation of genetic counselling services.

# 4 Results

A total of 74 surveys were completed. Of these, 20 surveys were excluded as they did not meet inclusion criteria. The data from the remaining 54 surveys were analysed in this study. A summary of the participants demographic data can be seen in Table 1 below.

***Table 1:*** *The demographic data of the responses included in the study.*

| Characteristics | Mean ± SD or Number (%) |
|---|---|
| **Age (years)** | 42.4 ± 10.6 |
| **Number of respondents in each profession** | |
| Fetal medicine specialist | 8 (14.8%) |
| Midwives | 11 (20.4%) |
| OBGYN | 32 (59.2%) |
| Sonographers | 3 (5.6%) |
| **Area of practice** | |
| Private only | 13 (24.1%) |
| State only | 31 (57.4%) |
| Private and State | 10 (18.5%) |
| **Years in current profession** | |
| < 5 | 18 (33.3%) |
| 5 – 10 | 14 (25.9%) |
| 10 – 15 | 7 (13.0%) |
| > 15 | 15 (27.8%) |

More than half of the participants, 59.2% (32/54), were doctors working in obstetrics and gynaecology. The mean age of participants was 42.4 years, with the youngest participant being 27 years old and the oldest being 72 years old. Of the 54 participants, more than half, 57.4%

(31/54), worked exclusively in the state healthcare sector. Years of practice in current field showed that 33.3% (18/54) of participants, had been in their current profession for less than 5 years.

## 4.1 Referrals to genetic counselling services

Of the participants, 74.1% (40/54) said they were able to refer patients to genetic counselling. The remainder of participants, 25.9% (14/54), were either unable to refer (5/14), were not sure if referrals were available to them (7/14) or did not feel referrals were within their scope of practice (2/14). Assessing referral rates, showed 57.4% (31/54) of participants had referred patients to genetic counselling services within the three months prior to their participation in the study. The number of referrals made by medical doctors (fetal medicine specialists and OBGYNs) and allied prenatal healthcare professionals (sonographers and midwives), showed no statistical difference and can be seen summarised in Figure 1**:** A summary of the number of referrals made to genetic counselling services over the three months prior to participation in the study. below. The most common number of referrals, made by 29.6% (16/54) of participants was 1 – 2 referrals and, 14.8% (8/54) had made more than 10 referrals over the three months prior to their participation in the study.

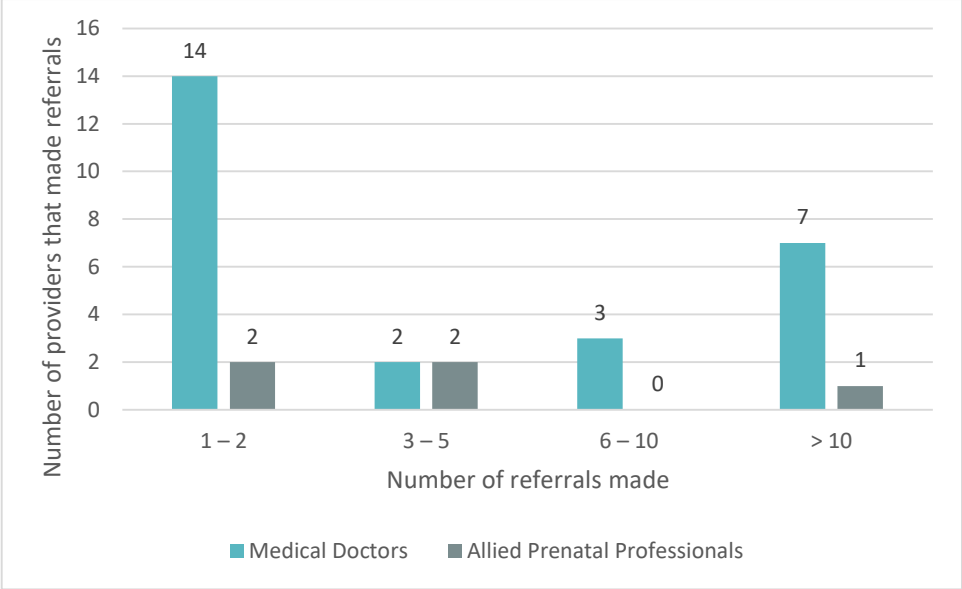


***Figure 1:*** *A summary of the number of referrals made to genetic counselling services over the three months prior to participation in the study.*

Participants were asked to identify appropriate reasons to refer a patient to genetic counselling services and were provided with a list of 18 reasons, all appropriate, to choose from. None of the respondents were able to correctly identify all eighteen appropriate reasons provided. Of the eighteen reasons provided, a minimum of half of the correct answers were correctly identified by 20.4% (11/54) of participants. When knowledge of appropriate reasons for referral was assessed per profession, 9.1% (1/11) of midwives, 25% (2/8) of fetal medicine specialists and 25% (8/32) of OBGYNs were able to correctly identify at least half of the appropriate reasons for referral. No sonographer could correctly identify more than half of the appropriate reasons for referral.

As seen in Figure 2 below, the most commonly identified reason for referral selected by 83.3% (45/54) of participants was "*The birth of a previous child with a known genetic condition*". The least commonly identified reason for referral, 14.8% (8/54), was "*In vitro fertilisation and pre-implantation counselling*". Not listed as an option but suggested verbatim by 5.6% (3/54) of participants, was "*Delivery of an abnormal baby*".

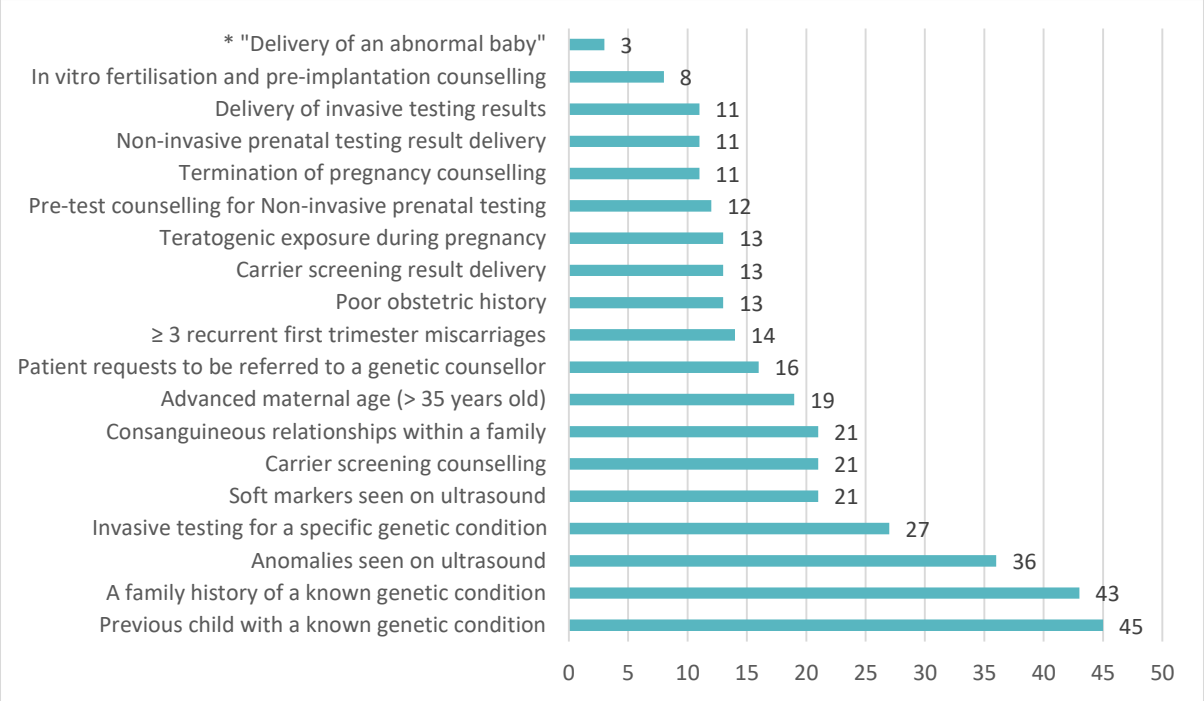


***Figure 2:*** *The number of prenatal healthcare providers that identified each reason for referral to genetic counselling services.*

Options not provided on the survey but listed as an additional answer by participants are denoted with *

## 4.2 Understanding and knowledge of genetic counselling services

### 4.2.1 Knowledge of the scope of practice of genetic counsellors

When presented with eight potential responsibilities of a genetic counsellor, 75.9% (41/54) of participants could not correctly identify the five correct responsibilities. As seen in Figure 3 below, the most commonly identified responsibility of a genetic counsellor, 94.4% (51/54), was "*To help people understand their options for genetic testing*". The three listed, but incorrect, responsibilities of a genetic counsellor were the least selected, with each being selected by between 18.5% (10/54) and 38.9% (21/54) of participants.

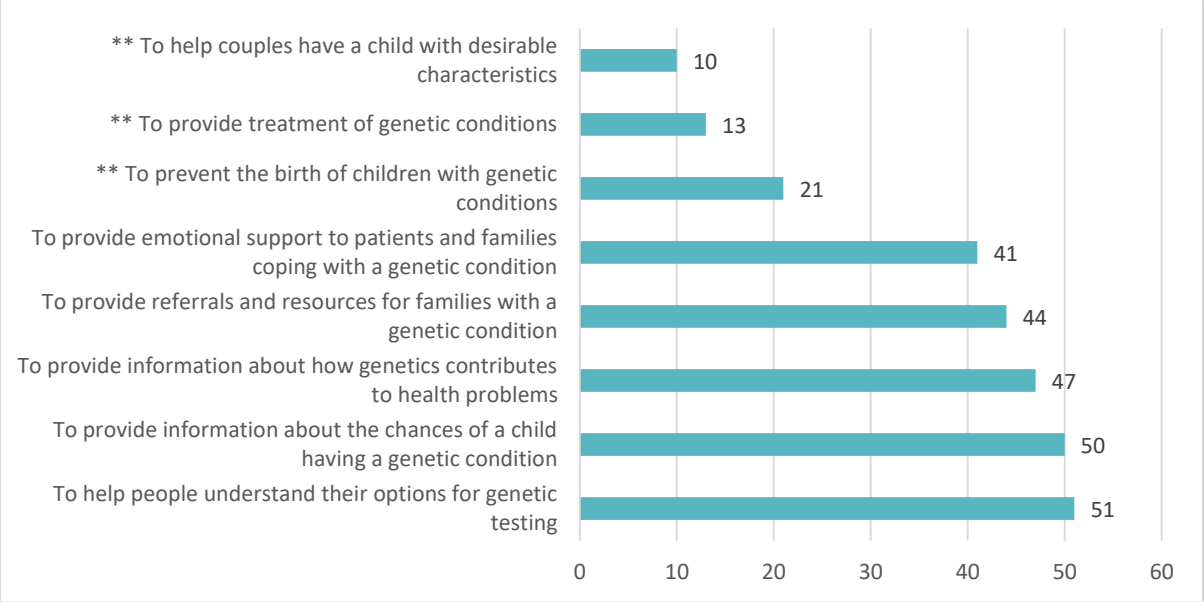


***Figure 3:*** *The number of prenatal healthcare providers that identified each responsibility as being part of a genetic counsellor's scope of practice.*
*Responsibilities outside the scope of practice of a genetic counsellor are denoted with***

### 4.2.2 Knowledge of the fields of practice of genetic counsellors

While 24.1% (13/54) of prenatal healthcare providers could correctly identify the responsibility and therefore the role of a genetic counsellor, almost double the number, 44.4% (24/54), were able to identify the various fields of practice of genetic counselling correctly. Of the fields of practice, "*Prenatal care*" was identified by 98.1% (53/54) of participants, followed by "*Paediatrics*" with 75.9% (41/54), then "*Adult*" and "*Oncology*" both with 61.31% (33/54) and "*Haematology*" the least with 57.4% (31/54).

## 4.3 Barriers and facilitators to referrals to genetic counselling services

Participants ranked the importance of various factors that may influence their decision to refer patients for genetic counselling (Table 2) as well as how frequently various factors hindered the referral process (Table 3). The majority of participants, 96.3% (52/54), stated that the severity of the disorder was an important consideration when deciding if a patient should be referred. When looking at the patient's interest in being referred, 77.8% (42/54) of participants chose this to be an important consideration. When assessing barriers to referrals, 38.9% (21/54)

of participants stated they were frequently not aware of genetic services. Participants showed confidence in their understanding of genetic counselling with 64.8% (35/54) expressing that a lack of understanding of the service is rarely a barrier to referral.

***Table 2:*** *Factors influencing prenatal healthcare providers' decision to refer patients for genetic counselling.*

| | **Very Important / Important** | **Moderately Important** | **Of Little Importance / Unimportant** |
|---|---|---|---|
| **Patients interest in genetic counselling** | 42/54 (77.8%) | 10/54 (18.5%) | 2/54 (3.7%) |
| **Severity of the disorder** | 52/54 (96.3%) | 1/54 (1.9%) | 1/54 (1.9%) |
| **Availability of testing and turnaround times** | 46/54 (85.1%) | 6/54 (11.1%) | 2/54 (3.7%) |
| **Availability of treatment for the disorder** | 40/54 (74.1%) | 10/54 (18.5%) | 4/54 (7.4%) |
| **Desire for management recommendations** | 49/54 (90.7%) | 5/54 (9.3%) | 0/54 (0.0%) |
| **Family's desire for recurrence information** | 49/54 (90.7%) | 5/54 (9.3%) | 0/54 (0.0%) |

*† for simplicity, the categories "very important" and "important" were counted together, as were the categories "of little importance" and "unimportant"*

***Table 3:*** *Factors preventing prenatal healthcare providers' decision to refer patients for genetic counselling.*

| | **Very Frequently / Frequently** | **Occasionally** | **Rarely / Never** |
|---|---|---|---|
| **Was not aware of genetic services** | 21/54 (38.9%) | 11/54 (20.4%) | 22/54 (40.7%) |
| **Do not know how to refer patients** | 17/54 (31.5%) | 14/54 (25.9%) | 23/54 (42.6%) |

| | | | |
|---|---|---|---|
| **Do not see much benefit for the patient** | 9/54 (16.7%) | 15/54 (27.8%) | 30/54 (55.6%) |
| **Do not understand what genetic counsellors do** | 13/54 (24.1%) | 6/54 (11.1%) | 35/54 (64.8%) |
| **Unable to get appropriate information from the Genetic Clinic** | 8/54 (14.8%) | 16/54 (29.6%) | 30/54 (55.6%) |
| **Do not feel it is my responsibility to refer** | 11/54 (20.4%) | 5/54 (9.3%) | 38/54 (70.4%) |

*† for simplicity, the categories "very frequently" and "frequently" were counted together, as were the categories "rarely" and "never"*

### 4.4 Knowledge improvement

Of the 54 participants, 5.6% (3/54), one OBGYN and two fetal medicine specialists, felt comfortable with the level of knowledge they have while 85.2% (46/54) were interested in increasing their knowledge of genetics and genetic counselling. The area of biggest interest was on overview of genetic disorders seen by fetal medicine providers selected by 66.7% (36/54) participants. Of the participants, 57.4% (31/54) expressed an interest in both identifying genetic resources for clients as well as new genetic technologies and advances. Referral procedures were requested by 46.3% (25/54) participants, while 38.9% (21/54) showed an interest in learning about genetic counselling and what the profession entails.

Online resources were the preferred platform for improving knowledge selected by 75.9% (41/54) of particpants, followed by academic lectures 59.3% (32/54), then written information 33.3% (18/54), and lastly discussion groups 31.5% (17/54).

## 5 Discussion

This study aimed to establish the utilisation of genetic counselling services amongst prenatal healthcare providers in Gauteng, South Africa, as well as elucidate the perceptions of genetic

counselling amongst these individuals. Participants knowledge on genetic counselling as well as their views on its utility was assessed with an online survey. Results show that while referrals to genetic counselling services are being made, there is largely an underutilisation of the service due to an insufficient understanding of the genetic counselling profession.

## 5.1 Referrals

The majority of participants (74.1%) in this study report that referrals to genetic services are available to them, yet only just over half (57.4%) had made referrals in the three months prior to participating in the study. The majority (83.9%) of referrals came from medical doctors, particularly OBGYNs. Compared to other specialities, such as family and internal medicine physicians, OBGYNs make greater use of genetic services in their day to day practice (Diamonstein et al., 2018). While it would seem that referral rates are relatively high, patient referral numbers are not. The majority of participants indicated a referral rate of 1 – 2 patients over a three-month period. These referral numbers suggest that prenatal healthcare providers are more aware of and may have better access to genetic counselling services allowing them to make more referrals than other specialties, but that their utilisation of the service to include its full potential is still low (Aalfs et al., 2003; Thom & Haw, 2021). Using 2021 census data, roughly 17,000 babies are born in Gauteng per month (Stats SA, 2022). Using the birth incidence of 1 in 15, this means there would be approximately 1,129 babies born in Gauteng per month that are affected by a congenital abnormality. In the six-month time period that referrals assessed by the study could have occurred, roughly 6,770 affected babies would have been born yet the genetics department at the NHLS only saw 173 new prenatal cases (K. Fourie, personal communication, October 12, 2023). While referrals could have been made to private facilities, given the lack of service availability it would be assumed that the difference in referral and uptake would not be explained by a lack of referrals to the NHLS alone.

### 5.1.1 Referral process

The majority of participants have the option to make referrals to genetic counselling services, yet only just over half had recently made any referrals. This suggests a lack of understanding of the referral process. This was again emphasized by approximately a third of participants stating they were unaware of genetic services and/or frequently unaware of the referral process. This study indicates that prenatal healthcare providers have better knowledge on the service and the referral process compared to allied health workers from a similar study by Thom and Haw (2021) which showed that approximately 50% of allied healthcare professionals were unaware of genetic services and 60% were unaware of the referral process. The other consideration is that while prenatal healthcare providers may know about referrals, their work environment may have a hierarchy through which referrals have to go. One participant indicated this when they said: "*Referral is usually through fetal medicine*". There is however no way to confirm if those referrals were reported in this study. Knowledge of the referral process is however only truly beneficial when appropriate referrals are being made.

### 5.1.2 The suitability of referrals

One of the participants, a fetal medicine specialist with more than 15 years' experience stated: "*There is definitely a significant role [for] genetics and genetic counselling [in fetal medicine]. Teamwork with obstetricians and fetal medicine specialists is very important*". This is a valuable insight, but the teamwork as described requires sufficient knowledge on genetics and appropriate referrals. Considering only approximately 20% of participants could correctly identify at least half of the given appropriate reasons for referral, it is clear there is a definite knowledge gap.

Though not all patients require genetic counselling, patients who are considered to have a high-risk pregnancy may be referred to genetic services for a variety of reasons. These reason include but are not limited to; a woman being of advanced maternal age (>35 years old), anomalies or soft markers seen on ultrasound, a previous child with/ family history of a known genetic

condition, poor obstetric history, invasive and non-invasive testing, termination of pregnancy counselling and teratogenic exposure.

The majority of participants (96.3%) felt that the severity of the condition was a very important factor to warrant referral to genetic counselling services. This is concerning for a multitude of reasons. Severity is known to be subjective and there are no clear or specific legal or social parameters that allow conditions to be classified as severe or not (Boardman & Clark, 2022; Dive et al., 2023). Not only is assessing severity subjective, but it rarely takes into account the patient's point of view or the psychosocial aspects of the case (Dive et al., 2023; Stenmarck et al., 2023).. Referrals based on severity are also problematic as not all genetic conditions always have a visible effect on the fetus. For example, Trisomy 21 (commonly referred to as Down Syndrome), one of the most common aneuploidies with an incidence of roughly 1/700, results in varying degrees of both intellectual and developmental delay (Jiang et al., 2017). Some might consider this a severe condition, yet, close to 30% of cases will go undiagnosed during the prenatal period (Jiang et al., 2017). Meaning that if no anomalies are seen on scan, which is not uncommon, or picked up from maternal serum screening, the healthcare provider would not be able to refer based on severity. Severity again becomes a questionable reason for referral when it has been shown that pregnant individuals experience increased worry and uncertainty after being diagnosed as having a high-risk pregnancy (Lou et al., 2016). Cleft/lip and/or palate is a common and generally repairable congenital anomaly that can be seen in isolation, approximately 50% of cases, or as part of a syndrome and is generally not considered to be "severe" (Stock et al., 2019). Given the complicated multifactorial or syndromic influence of genetics on clefts, providers should refer patients to genetic counselling services. This is specifically to help guide the patient and their family members through the complexities of the condition in a sensitive and knowledgeable manner (Stock et al., 2019). A large majority of participants felt that referrals were necessary when a patient had a previous child (83.3%) with,

or a family history (79.6%) of a known genetic condition. While these referrals are appropriate and necessary due to potential recurrence risk, they negate the fact that most congenital anomalies are multifactorial or of unknown etiology (Glinianaia et al., 2017; Verma, 2021). They also do not take into consideration cases of *de novo* mutations which affects 1/295 births but will have a recurrence risk of less than 1/1000 for most couples (Bernkopf et al., 2023; McRae et al., 2017). It is also important to note that in most cases, carriers of recessive conditions, such as cystic fibrosis, will have no family history of the condition (Holt & Trepanier, 2013, p. 19).

While nearly 76% of participants understood that the role of a genetic counsellor is to provide emotional/ psychosocial support, only approximately 20% of providers would refer for reasons where this support is needed such as termination of pregnancy counselling or delivery of testing results (invasive or non-invasive). Yet it has been shown that genetic counselling sessions pre- and post-termination for fetal anomaly, improves the ability of individuals to cope better with long-term grief (Smith et al., 2021). This speaks to the misconceptions on the benefits of referring a patient for genetic counselling regardless of the pregnancy outcome. A study by Godino et al., (2016) showed that invasive testing is still preferred over non-invasive screening among women of advanced maternal age. This denotes the importance of ensuring patients are supplied with the correct information to allow this type of decision making (Godino et al., 2015; Smith et al., 2021). Similar points were made in a study by Salema et al., (2018), where patients who had received high-risk prenatal screening results emphasized the importance of the information and guidance provided by their genetic counsellor. However, the same study described the anxiety caused by referral to genetic counselling services without extensive explanation as to what the role of a genetic counsellor is and why the patient was being referred (Salema et al., 2018). A study done by Maio et al., (2013) revealed that 69% of the general Canadian population had never heard of genetic counselling. Due to the difference in genomic

literacy between Canada, a developed country, and a developing country such as South Africa, it is expected that the level of knowledge of genetic counselling would be even lower in South Africa. This is an important consideration since the majority of participants in this study (77.8%) considered the interest in genetic counselling shown by the patient to be an important factor when considering referral. This once again demonstrates that prenatal healthcare providers should be better informed on the genetic counselling profession as to be able to explain the necessity of it to the patients they refer (Maio et al., 2013). The majority of participants stated the desire for management recommendations to be a reason for referral yet do not seem to understand the full capacity of a genetic counsellor and the benefit referrals may have.

## 5.2 Barriers to referrals

When asked about barriers to referral, only 24.1% of participants stated that they were not clear on the role and scope of a genetic counsellor. However, the majority of participants (75.9%) were unable to correctly identify the responsibilities of a genetic counsellor when provided with a number of options. This indicates not only that participants were not well informed on the scope of genetic counselling, but also the fact that they overestimated their knowledge and understanding of that role. Some particular areas where this misunderstanding was evident was when the majority of providers noted the importance of genetic counsellors in providing patients with information on conditions and testing options, but the importance of the psychosocial and emotional support provided by genetic counsellors was not as widely known. The effective communication and informed shared decision-making needed in the prenatal sector should be provided by a genetic counsellor (Metcalfe, 2018). The importance of support from genetic counsellors was demonstrated in a study by Voorwinden et al., (2020) who found that patients who received genetic counselling, one-on-one or in a group, showed significant improvement in personal empowerment, personal control, and anxiety.

## 5.3 Facilitators of referrals

Although this study demonstrates that genetic counselling services are being utilised amongst prenatal healthcare providers in Gauteng, it indicates that the service is not being utilised to its full potential due to misconceptions on the role and scope of a genetic counsellor. still being underutilised. Increasing utilisation is not possible if there is a lack of knowledge on genetics and genetic counselling. Considering the limited number of genetic counsellors available in Gauteng, and the importance of having an integrated team, it is important that other health professionals have at least basic genetic knowledge to assess which patients may be appropriate for referral (Metcalfe, 2018; Tonkin et al., 2018).

It is therefore promising to see that more than 85% of participants were interested in increasing their knowledge of genetics and genetic counselling. Education of prenatal healthcare providers should be tailored to various ages and should be provided at various levels during the clinicians training (Diamonstein et al., 2018). Given that the majority of participants preferred online resources, distribution of information may be more efficient and may have further reaching implications if distributed through electronic media.

Information sessions focusing on the genetics in the prenatal sector can be hosted on a virtual platform to allow for prenatal healthcare providers, at various levels of training, across the country to join. Physical resources discussing genetics in the prenatal setting such as information sheets or “yes-no” flow charts outlining referral criteria should be distributed to the various hospitals. These should also be distributed as digital copies for ease of access should the healthcare provider not have a physical copy available.

## 5.4 Chapter summary

This study showed that a large majority of prenatal healthcare providers are aware of and can refer patients to genetic counselling services. However, not all of those aware of the genetic counselling services have previously referred patients, and even less have referred patients

within the three months prior to the study. While there is a general lack of understanding of appropriate reasons for referral and the scope of practice of a genetic counsellor, the majority of participants showed a keen interest to increase their knowledge of genetics and genetic counselling. By improving education in these areas, it is possible to improve the appropriate utilisation of genetic counselling amongst prenatal healthcare providers and thus improve prenatal care.

## 5.5 Study limitations

This study provides information about the utilisation of genetic counselling amongst a small sample size of prenatal healthcare providers in only one province in South Africa. The province the study took place in, Gauteng, is one of only four provinces in the country that has genetic services. Many of the hospitals and doctors that participated in the study were already referring patients to genetic services. Sample bias may have occurred in this study as prenatal healthcare providers who were already aware of genetic services may have been more likely to complete the survey. Doctors in private practice may have been less inclined to answer if they have in-house genetic counsellors, an example being doctors working in fertility centres as part of a multidisciplinary team. Extrapolation to other provinces would need to be investigated as prenatal healthcare providers in other provinces may have even less knowledge of genetic counselling and may utilise the service far less. Despite this, the study demonstrates a recurring theme that those not directly involved in genetic services, have misconceptions about what genetic counselling entails. The questionnaire used in this study has not yet been validated.

## 5.6 Practice recommendations

This study showed that while prenatal healthcare providers are utilising genetic counselling services, the full scope of the profession is not being used. This can lead to gaps in patient care. This study indicates the need for educating prenatal healthcare workers on genetics and the role of the genetic counsellor in clinical practice to allow for appropriate referrals to genetic

counselling services. Education of this nature should continue throughout the course of the prenatal healthcare professionals training. This will not only help to ensure that all healthcare providers attain training at some point during their training but will also help to keep education and training updated to current practices.

### 5.7 Research recommendations

It is recommended that a study be done to assess the utilisation of the service amongst prenatal healthcare providers in other provinces. This will help assess the overall utilisation of the service across South Africa and identify areas where education on genetic counselling services are most needed. In addition, the same study could be done after educational resources have been distributed to prenatal healthcare providers to assess the impact of education on referral rates. Qualitative studies assessing the prenatal healthcare provider's perception of genetic counselling in the preconception, prenatal and postnatal periods would give insight into when patients find genetic counselling most beneficial.

## 6 Conclusions

Genetic counselling is crucial to basic healthcare and is integral to lessening the burden of congenital anomalies. While genetic counselling services are being utilised by prenatal healthcare providers in Gauteng, the service is not being utilised to its full potential. This study found that most prenatal healthcare providers are aware of genetic counselling services and are able to make referrals. However, they have limited knowledge of the scope of practice of genetic counsellors and, methods of referral and appropriate reasons for referral to genetic counselling services. These are large barriers to the referral process as appropriate patients are not referred. The contribution of these barriers to low referral rates can be lessened through education of prenatal healthcare providers on genetics and genetic counselling. This would allow for better prenatal patient care and potentially help to decrease the prevalence of congenital anomalies.

# 7 Author contributions

All authors contributed to the conception and design of the study. MD collected and analysed the data. She drafted the work for publication and takes full responsibility for the integrity of the data. All authors contributed to the interpretation of the data and take responsibility for the accuracy of the data analysis. MA and KF critically revised the contents of the manuscript. All authors agree to be accountable for all aspects of the text and signed off on the final version before submission.

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# 9 Compliance with ethical standards

## 9.1 Conflict of interest

The authors declare they no conflict of interest.

## 9.2 Human studies and informed consent

The study was approved by the University of the Witwatersrand Ethics Committee (Medical) (Certificate No. M230268). Participants had to sign consent before they could participate in the study. The data collected was kept confidential and only accessible by the authors of the study.

## 9.3 Animal studies

No animal studies were carried out by the authors for this article.

### 9.4 Data availability statement

In compliance with the University of the Witwatersrand Ethics Committee (Medical) privacy restrictions, while no self-identifying information was asked, to protect the privacy of the research participants none of the study data is publicly available.

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# Appendices

## Appendix A: Approved research protocol

UNIVERSITY OF THE WITWATERSRAND, JOHANNESBURG

FACULTY OF HEALTH SCIENCES

| | | | |
|---|---|---|---|
| **CANDIDATE'S SURNAME:** [Please print] Duvenhage | **FIRST NAME/S:** Megan | **STUDENT NUMBER:** 1076441 | |
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| **DEGREE FOR WHICH PROTOCOL IS BEING SUBMITTED:** MSc (Med) in Genetic Counselling | | | |
| **PART-TIME OR FULL-TIME:** | Full-Time | | |
| **FIRST REGISTERED FOR THIS DEGREE:** | **TERM :** First | **YEAR:** 2022 | |
| **DEPARTMENT:** Human Genetics School of Pathology | | | |
| **TITLE OF PROPOSED RESEARCH:** The Utilisation of Genetic Counselling Services Amongst Prenatal Healthcare Providers in Gauteng, South Africa | | | |
| **CANDIDATE'S SIGNATURE:** Duvenhage | | **DATE:** 21 March 2023 | |
| **SUPERVISOR 1 (NAME & SURNAME):** | Monica Arajuo | **% Supervision:** 50 | |
| **SUPERVISOR'S QUALIFICATIONS:** | MSc (Med) Genetic Counselling | | |
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| **SUPERVISOR 2 (NAME & SURNAME):** | Katryn Fourie | **% Supervision:** 50 | |
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| **SUPERVISOR 3 (NAME & SURNAME):** | NA | **% Supervision:** NA | |
| **SUPERVISOR'S QUALIFICATIONS:** | NA | | |
| **SUPERVISOR'S ADDRESS / TEL / E-MAIL:** | NA | | |

***SYNOPSIS OF RESEARCH*:** ***(Brief summary of proposed research project; between 200-300 words only; with sub-headings: an introduction and justification for study, aim/s, proposed methodology and expected outcome/s)***
**[Use reverse side of this page if more space is required]**

Genetic conditions may be present from birth and can affect isolated systems or may be multi-systemic. It has therefore become common to see patients with genetic conditions being managed by various specialities in the medical field including medical geneticists and genetic counsellors. Genetic conditions and congenital anomalies are often detectable during pregnancy. The prevalence and mortality rates of congenital anomalies and disorders remain high, despite the fact that up to 70% can be prevented, improved, or effectively managed with relatively inexpensive measures. Prevention of congenital disorders

can occur at various levels and can all be facilitated by a genetic counsellor due to their knowledge and training. Prenatal healthcare providers are uniquely suited to detecting a number of possible abnormalities a fetus may have before birth. This allows for action to be taken in whatever form appropriate to manage the case.

Local and international studies have assessed the awareness of genetic counselling services and their utilisation by other sectors of the medical community. These studies have concluded that healthcare professionals globally, that do not work in genetics, have little awareness and understanding of the work done by genetic counsellors and the value they provide. While there has been an increase in the popularity of genetic counselling, it is evident from these studies that not all medical professionals see the need to utilise the service. The aim of the project is to establish the uptake of genetic counselling services amongst prenatal healthcare providers in Gauteng, South Africa as well as insights into the perceptions of genetic counselling and its usefulness by these individuals. The information provided by this study will give insight on how to best improve the utilisation and efficacy of the limited genetic counselling services available in Gauteng, South Africa.

| | |
|---|---|
| **WITS ETHICS NOT REQUIRED:** Yes No<br>**WITS ETHICS PENDING:** Yes No<br>**WITS ETHICS APPROVED:** Yes No<br>**(Circle appropriate symbol)***<br><br>***Please note the final human ethics clearance certificate or animal ethics certificate must be available prior to starting research** | **IF Y SUPPLY ETHICS CLEARANCE CERTIFICATE AS ATTACHMENT AND INCLUDE ETHICS NUMBER HERE:** |
| **As supervisor/s, I/we confirm that I have read the protocol which has been submitted for assessment.** | |

| | | |
|---|---|---|
| **SIGNATURE OF SUPERVISOR/S:** |  <br>…….<br>Monica Arajuo |  <br>…….<br>Katryn Fourie |
| **SIGNATURE PG OFFICE STAFF**<br><br>…….. | **REGISTERED**<br><br>**YES…..** **NO…..** | **STAMP** |

i

**PLAGIARISM DECLARATION TO BE SIGNED BY ALL HIGHER DEGREE STUDENTS**

I **Megan Duvenhage** (Student number: **1076441)** am a student registered for the degree of **MSc (Med) Genetic Counselling** in the academic year **2023**.

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 for the above degree 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.
- I have included as an appendix a report from "Turnitin" (or other approved plagiarism detection) software indicating the level of plagiarism in my research document.

Signature: Duvenhage Date: 21 March 2023

UNIVERSITY OF THE
WITWATERSRAND,
JOHANNESBURG

**Research Proposal**

# The Utilisation of Genetic Counselling Services Amongst Prenatal Healthcare Providers in Gauteng, South Africa

by

**Megan Duvenhage**

1076441

Master of Science in Medicine in Genetic Counselling

*Supervised by:*

Ms. Monica Araujo (MSc (Med) Genetic Counselling)

Ms. Katryn Fourie (MSc (Med) Genetic Counselling)

# 1 Background

Genetic information is found in every cell and genetic conditions may therefore affect many different areas or systems of the body. These conditions may be present from birth and can affect isolated systems or may be multi-systemic. It has therefore become common to see patients with genetic conditions being managed by various specialities in the medical field including medical geneticists and genetic counsellors. For genetic counsellors, this has resulted in genetic counselling having a global presence in research and nearly all specialties of medical care, especially obstetrics, rare diseases in paediatric care, oncology, cardiology and nephrology (Abacan *et al.*, 2019). Resta et al., (2006) defines genetic counselling as "the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease" (Resta *et al.*, 2006). The global presence of genetic counselling and its utilisation is ever increasing due to the increasing demand for the service shown by patients and medical professionals alike.

International studies have assessed the awareness of genetic counselling services and their utilisation by other sectors of the medical community. These studies have concluded that healthcare professionals globally, that do not work in genetics, have little awareness and understanding of the work done by genetic counsellors and the value they provide (Aalfs *et al.*, 2003; Suther & Goodson, 2003; Baars *et al.*, 2005). A similar study was performed in South Africa by Thom and Haw (2021), to assess the awareness of genetic counselling services among allied healthcare professionals. This study came to the same conclusion, namely that allied healthcare professionals had a poor awareness of genetic counselling services and thus made little use of them (Thom & Haw, 2021).

While there has been an increase in the popularity of genetic counselling, it is evident from these studies that not all medical professionals see the need to utilise the service. Roughly 5%

of children globally are born with congenital anomalies and/or significant physical or mental handicap (Harper, 2010). Congenital disorders are present in one in every fifteen live births in South Africa (Malherbe *et al.*, 2015). These anomalies could potentially be detected during a pregnancy. This, however, would require those involved in prenatal healthcare to have an understanding of genetic counselling and the benefits it could present in the management of pregnancies.

## 1.1 Genetics in prenatal medicine

Genetic conditions are often detectable during pregnancy, and due to improved testing, identifying them has become easier. Anomalies on an ultrasound, a personal family history of a genetic condition, and advanced parental age are often indicators for genetic testing. The identification of the underlying cause of abnormalities seen in a fetus is important not only to ascertain the risks posed to the fetus, but what medical management they need. By identifying the cause, recurrence risks can be given to the family and any at-risk family members may be identified. It also ensures that expectant parents are given the correct information regarding their options. These options may include potential testing, as well as giving them a choice to either continue with the pregnancy or to have a termination (Wojcik *et al.*, 2020).

## 1.2 Genetic counselling in prenatal medicine

The prevalence and mortality rates of congenital anomalies and disorders remain high, despite the fact that up to 70% can be prevented, improved or effectively managed with relatively inexpensive measures (Malherbe *et al.*, 2015). Interventions put into place to decrease the burden of congenital disorders must look at both prevention and care (Malherbe *et al.*, 2016). Prevention of congenital disorders can occur at various levels and can all be facilitated by a genetic counsellor due to their knowledge and training. It can occur through primary prevention with the implementation of basic reproductive health care such as vitamin and folic acid

supplementation in pregnant patients. Secondary prevention is aimed at decreasing the number of affected children that are born by improving prenatal screening and diagnosis, education on avoidance of teratogenic substances and discussing termination of a pregnancy. Tertiary prevention is the final way in which the prevention of congenital disorders can occur. It is aimed at early detection after birth to improve medical interventions and treatments and provide counselling and psychosocial support to affected families (Malherbe *et al.*, 2016).

### 1.2.1 The role of the genetic counsellor in prenatal medicine

Genetic counsellors have a large knowledge of human genetics and genetic testing. They are equipped to explain complicated information and genetic concepts to patients in a way that they understand and that allows them to feel supported and empowered. This allows the patients the ability to take control of their own healthcare and make informed choices that best suit them (McAllister *et al.*, 2008; Kromberg, Wessels, *et al.*, 2013; Abacan *et al.*, 2019). While fetal medicine specialists, obstetricians and sonographers perform sonography during pregnancy and identify likely abnormalities, it is often the role of genetic counsellors to propose and discuss appropriate testing, obtain informed consent regarding any invasive procedures and provide counselling to the family in a way that they understand (Yotsumoto *et al.*, 2016). It is therefore important that genetic services are available to assist in these cases (Scelsa, 2022). Prenatal genetic counselling helps ensure that the patient's autonomy regarding their pregnancy and childbirth are respected and that they are equipped with the sufficient information to make informed decisions regarding their pregnancy (Yotsumoto *et al.*, 2016).

Medical genetic services, including genetic counselling, are in place to prevent, detect and care for patients with congenital disorders (Malherbe *et al.*, 2016). It is recommended that medical genetics and genetic counselling services be offered as an integral part of basic healthcare as part of a comprehensive healthcare programme with aims to lessen the burden of inherited conditions (Kromberg & Krause, 2013). This can only successfully be achieved with the help

of other departments that refer suitable patients to these services. Prenatal healthcare providers should therefore, at least refer any patients that have a pregnancy that is affected by or at risk of a congenital disorder, patients at reproductive risk of having children with a congenital disorder and couples that are affected by a genetic disorder but still want to have unaffected children (Malherbe *et al.*, 2016).

### 1.2.2 Genetic counselling in prenatal healthcare in South Africa

South Africa has established genetic services and clinics available to the state healthcare system in three of the nine provinces. These are Gauteng, the Western Cape, and Kwa-Zulu Natal (Kromberg, Wessels, *et al.*, 2013). These services include prenatal genetic diagnosis, diagnostic, predictive and carrier testing and genetic counselling (Kromberg, Sizer, *et al.*, 2013). These services are also offered in the private sector at various independent and state run organisations. Genetic counselling is still a young profession within South Africa with formal clinics in the state healthcare systems in Johannesburg and Cape Town only having started in the late 1960's (Kromberg & Krause, 2013; Malherbe *et al.*, 2016). Genetic counsellors play a significant role in the provision of genetic services in the country and often undertake the role of referring patients to specialists and allied healthcare professionals for appropriate management. This occurs as other healthcare providers are not always aware of the types of management required by a patient with a genetic condition (Kromberg, Wessels, *et al.*, 2013; Thom & Haw, 2021).

### 1.2.3 Genetic counselling in prenatal healthcare in Gauteng

Since the late 1960's the National Health Laboratory Service (NHLS) in Braamfontein Johannesburg has offered prenatal genetic diagnosis as well as carrier screening in high risk populations (Kromberg & Krause, 2013). Genetic services, medical and counselling, are available at the tertiary care hospitals in Johannesburg on a weekly basis. Patients are usually seen in these clinics through referrals from other departments or from local clinics. Genetic counselling services for prenatal healthcare in Johannesburg are currently offered by the

Clinical and Counselling Section of the Division of Human Genetics at the NHLS and University of the Witwatersrand for both state and private patients. There are currently three state prenatal clinics scheduled each week, one at Rahima Moosa Mother and Child Hospital and two at Chris Hani Baragwanath Academic Hospital. Private patients that have been referred for genetic counselling are seen at the Wits Donald Gordan Medical Centre.

Prenatal healthcare providers are uniquely suited to detecting a number of possible abnormalities a fetus may have before birth. This allows for action to be taken in whatever form appropriate to manage the case. Since many congenital abnormalities may be genetic, it is important that genetic counsellors are involved in these cases. While prenatal referrals for conditions such as Down syndrome are regularly made, others involving neural tube defects or consanguineous relationships are less often referred and as a result do not receive genetic counselling (Kromberg, Wessels, *et al.*, 2013). These are examples of cases that could easily be referred from prenatal healthcare providers. While genetic services are regularly available in Johannesburg, the utilisation of genetic counselling services has been low and should be improved to ensure proper patient management (Kromberg, Wessels, *et al.*, 2013; Thom & Haw, 2021).

### 1.3 Barriers to referrals

There is insufficient information on the barriers to referrals to genetic services within South Africa. This has resulted in information on the topic being guided by the findings of similar studies in other countries. A study carried out in America by Aalfs et al. (2003) found that general practitioners do not often see or treat patients with genetic conditions and perceive them as rare cases. This results in a lack of awareness and referral to genetic services and genetic counselling due to lack of interest and knowledge in genetics. The lack of knowledge of genetics and misconceptions of the role of a genetic counsellor in the medical fraternity are

likely the biggest barriers in referring a patient to genetic counselling (Aalfs *et al.*, 2003; Suther & Goodson, 2003; Thom & Haw, 2021). Not only do practitioners not have knowledge regarding genetics, but they are also unaware of the types of patients that would be eligible for referral to genetic services (Delikurt *et al.*, 2015). Aalfs et al., (2003) found that many medical providers feel that genetic counselling has little benefit when it comes to therapeutic and preventative measures and that counselling should take place before, rather than during, a pregnancy (Aalfs *et al.*, 2003). Their focus on only treating the possible condition may cause a lack of awareness of how genetic counselling may help facilitate earlier testing to increase patient choice, help patients address their options to have an unaffected pregnancy or to inform the patients of the risks to them, their future pregnancies and other family members (Aalfs *et al.*, 2003).

The results of the study by Aalfs et al. (2003), while insightful, cannot be extrapolated into a South African setting due to the extensive differences between America, a first world country, and South Africa, a developing nation. Recently published data by Ormond et al. (2018) and Abacan et al,. (2019) found that there are only roughly 7 000 genetic counsellors worldwide with only 25 genetic counsellors practicing in South Africa (Ormond *et al.*, 2018; Abacan *et al.*, 2019). This has caused a gap in the knowledge of genetic counselling and the perceptions of South African healthcare workers towards genetic counselling services.

Thom and Haw (2021) found that while half of allied healthcare workers in South Africa were aware of the availability of genetic counselling services, only half of those providers made use of the service. The lack of referrals coming from allied healthcare workers not only stemmed from a lack of knowledge on genetics and genetic counselling, but also from a lack of knowledge on the referral process. The findings from the study conducted by Thom and Haw (2021) were consistent with similar studies performed internationally. While there were similarities between local and international allied healthcare professionals regarding the

awareness of genetic counselling, it is not necessarily possible to extrapolate these findings with regards to prenatal healthcare providers.

## 2 Rationale

Studies have been done internationally to assess the awareness of genetic services and their utilisation by various medical sectors. However, there have been no studies done focusing on the uptake of genetic counselling among prenatal healthcare providers, internationally or within South Africa. Due to their unique position dealing with reproduction, prenatal healthcare providers have the ability to potentially diagnose an affected fetus before birth or implantation in in vitro fertilisation. Due to this and the role of genetics in reproduction, these service providers and their patients would likely benefit from the genetic services available to them. It is therefore important to find out if prenatal healthcare providers are aware of genetic counselling services available to them and what reasons they have for utilising or not utilising the service. The information provided by this study will give insight on how to best improve the utilisation and efficacy of the limited genetic counselling services available in Gauteng, South Africa.

## 3 Aim

The aim of the project is to establish the uptake of genetic counselling services amongst prenatal healthcare providers in Gauteng, South Africa as well as insights into the perceptions of genetic counselling and its usefulness by these individuals.

## 4 Study Objectives

1. To determine if, and why, prenatal healthcare providers refer patients to genetic counselling services.
2. To determine the factors that prenatal healthcare providers find important when deciding whether to refer a patient to genetic counselling services or not.

3. To identify the barriers and facilitators that prenatal healthcare providers experience when making a referral to genetic counselling services.
4. To assess how best to improve the knowledge of genetic counselling amongst prenatal healthcare providers.
5. To assess how the referral process and patient access to genetic counselling may be improved.

# 5 Methods

## 5.1 Instrumentation

The survey intended for use is based on the survey used by Thom et al., (2021). This original survey was developed to determine the awareness of genetic counselling services among allied healthcare professionals in South Africa (Thom & Haw, 2021). While this study and the one performed by Thom et al., (2021) look at different healthcare providers, they both seek to answer similar questions surround the awareness and utilisation of genetic counselling services. The survey will be created and completed electronically using Google Forms. The survey will contain both checkboxes as well as short answer questions.

The survey will be made up of three sections: 1) demographics and professional practice of the participant, 2) the participants referrals to genetic counselling services, and 3) the participants understanding of genetic counselling services. Questions in section 1 will remain the same as Thom et al.'s survey while the rest of the questions will be tailored based on their relevance to the field of genetic counselling in prenatal medicine rather than allied healthcare professionals. All questions in the survey will be made mandatory to avoid the submission of incomplete surveys.

**The functionality of the survey will be tested by analysing the answers from first 5 respondents and determining if any changes need to be made to the survey.**

## 5.2 Participant recruitment

The inclusion criteria for this study will be professionals in Gauteng that are currently practising as; fetal medicine specialists, obstetricians, sonographers, midwives, or fertility specialists. Specialising registrars that have been training in these fields for more than six months will also been included in the study, while those that have been in their current position for less than six months will be excluded.

### 5.2.1 Participant sample pool

Potential participants will be identified through databases such as the South African Society or Ultrasound in Obstetrics and Gynaecology (SASUOG), the Fetal Medicine Foundation (FMF) of South Africa and **the Society of Midwives of South Africa**. They will also be identified by making contact with the heads of the fetal medicine units at the various hospitals in and around Johannesburg. These hospitals include Chris Hani Baragwanath, Rahima Moosa Mother and Child, Charlotte Maxeke, Tambo Memorial, Far East Rand, Edenvale, Steve Biko, Thelle Mogoerane and Pholosong.

Lastly they will be identified by requesting help from genetic counsellors to identify prenatal healthcare providers that they feel would be appropriate for the study and asking them to forward the information onto the potential participant. The potential participants will be contacted via email and invited to complete the questionnaire.

**It is expected that there are possibly around 500 people in Gauteng that would be suitable participants for this study. Using a sample size calculator available freely on SurveyMonkey (https://www.surveymonkey.com/mp/sample-size-calculator/) it is shown that a sufficient sample size would be to receive 218 completed surveys. This is based on a 95% confidence level with a 5% margin of error.**

$$sample\ size = \frac{\frac{z^2 \times p(1-p)}{e^2}}{1 + \left(\frac{z^2 \times p(1-p)}{e^2 N}\right)}$$

$$sample\ size = \frac{\frac{(1.96)^2 \times 0.5(1-0.5)}{0.05^2}}{1 + \left(\frac{(1.96)^2 \times 0.5(1-0.5)}{0.05^2(500)}\right)}$$

$$sample\ size = 217,25$$

$$\therefore 218\ completed\ surveys\ would\ be\ required$$

#### 5.2.2 Participant information sheet

The email sent out will contain a body of information on the study, its rationale and aims. This information will explain that the study will be carried out in the form of an online survey which will be completed anonymously by the participant. After the relevant information is provided, there will be a link which will redirect the participant to the survey.

#### 5.2.3 Participant consent form

Before the participant can begin the survey they will need to give consent. The first page of the survey will provide the terms of consenting to the study. Consent needs to be given, in the form of ticking a checkbox, before the participant is able to continue with the survey.

### 5.3 Data capture

Once the survey has been completed the participant will submit their answers and Google Forms will capture the results. The captured data will be available directly after submission. All captured responses will be pulled into Excel for analysis.

### 5.4 Data analysis

Statistical analysis will be performed for demographic question responses using the Excel statistical package. A Shapiro-Wilk test will be used to assess the normality of the demographic data. **The participants will be divided into subgroups based on 1) their profession and 2) their age and 3) the number of years of experience they have in their current field. Descriptive statistics (mainly percentages and means) will then be used to analyse the sections on knowledge and utilisation of genetic counselling services amongst each subgroup. This will enable us to see if there is a correlation between the participants'**

**profession and age when it comes to their knowledge of and referral to genetic counselling services.**

## 6 Ethics

Ethics clearance from the University of the Witwatersrand Human Research Ethics Committee (Medical) will be applied for on 7 February 2023 and should be approved by March 2023.

Letters have been sent to the CEO's of the hospitals mentioned in section 5.2.1.

## 7 Timeline

| | *Dec '22* | *Jan '23* | *Feb '23* | *Mar '23* | *Apr '23* | *May '23* | *Jun '23* | *Jul '23* | *Aug '23* |
|---|---|---|---|---|---|---|---|---|---|
| ***Literature Review*** | | | | | | | | | |
| ***Protocol Write-up*** | | | | | | | | | |
| ***Ethics*** | | | | | | | | | |
| ***Protocol Assessment*** | | | | | | | | | |
| ***Data Collection*** | | | | | | | | | |
| ***Data Analysis*** | | | | | | | | | |
| ***Report Write-up*** | | | | | | | | | |

The research project should be completed by end of August 2023. This allows for three months during which it can be examined before my degree ends in December of 2023.

## 8 Funding

This project will make use of Google Forms, a publicly available, free survey creation platform. No further funding will be required.

## 9 Limitations

1. There may be an insufficient number of participants to get an accurate reflection of the awareness of genetic counselling amongst prenatal healthcare providers in Gauteng.
2. There is a possibility of sample bias as prenatal healthcare providers who are already aware of the available genetic services in Johannesburg, may be more compelled to complete the survey.

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UNIVERSITY OF THE WITWATERSRAND, JOHANNESBURG

FACULTY OF HEALTH SCIENCES

**PROTOCOL ASSESSORS MEETING**

Candidate Full Name: . . . . . Megan Duvenhage . . . . . .

Student Number: . . . . . 1076441 . . . . . Date: . . . . . 23rd February 2023 . . . . . .

School / Department / Division: . . . . . Pathology / Human Genetics . . . . . .

**1. Type of study** *(tick all that apply)*:

X Quantitative
o Qualitative
o Mixed Methods
o Laboratory
o Clinical
o Other, please specify……

**2. Is title of the study appropriate** (*preferably fewer than 20 words*)**?** Yes

Comments: _____

**3. Are the study objectives clear and linked to the research aim and title?** Yes

Comments: _____

**4. Is the design of the study appropriate to meet the study objectives?** Yes

Comments: _____

**5. Are the proposed methods and tools appropriate to meet the research objectives?** Yes, but...

Comments (the following changes should be made to the 'Methods' section of the protocol unless otherwise stated):

- Please provide an estimate of the possible sample size for this study – online calculator: https://www.surveymonkey.com/mp/sample-size-calculator/
- Add a sample size calculation. If the calculation provides a number that is not attainable then that is OK. You have already suggested that there may be '…an insufficient number of participants…' in the 'Limitations' section of the protocol so you can refer to the sample size calculation as evidence of this
- In the 'Data analysis' section more detail is required. Will data be compared between the different medical specialists groups? If so, how? Will data be analysed sub-grouped by age or gender of participants?
- In questionnaire, would it not be easier just to ask age rather than asking for participant to pick an age sub-group? Better to use continuous data than categorical. If you do keep as categorical then the age sub-groups are in a strange format e.g. '20 ≥ 29' It should be '20-29, 30-39' etc…
- Questionnaire should be piloted in a small group of appropriate participants to ensure that it is functional
- Is there not a better way to phrase question 3? Is it not possible that someone may have referred someone for counselling over 3 months ago?

**6. Is the study feasible within the resources of:**

a) The applicant? Possibly

b) The department? Yes

c) The time frame? Yes

Concern was expressed on whether enough people would return the completed questionnaire. It was suggested that the student must use all means necessary to obtain a sufficient number e.g. send e-mail reminders, visit the appropriate departments, give a presentation on the study to the departments.

**7. If this is a PhD protocol assessment:** **Not applicable**

a) Is the content original? Yes No

b) Does the content show the scope and depth of a PhD? Yes No

Comments: _____

_____

_____

_____

**Do you recommend:**

i. Additional revision/amendment of the protocol? Please be specific on the recommendations being made:

_____

_____

_____

_____

ii. The appointment of the proposed Supervisor? Yes

Nominee/s: _____

_____

_____

iii. The appointment of the proposed Co-Supervisor/s and/or additional co-supervisors? Yes

Nominee/s: _____

_____

_____

iv. Has the Chair of the Assessor Group signed the RECOMMENDATION FOR APPOINTMENT OF SUPERVISOR(S) OF RESEARCH REPORT, DISSERTATION OR THESIS form? Please attach. Yes

v. Has the Chair informed the student and supervisor about the Wits ethics requirements, and that if required, they must have either a Wits Human Research Ethics clearance certificate or a Wits Animal Research Ethics clearance certificate? Yes

vi. Based on the protocol provided (including any proposed changes by the protocol assessor group), does the student require:

| | | |
|---|---|---|
| 1. Human Research Ethics clearance certificate | Yes | |
| 2. Animal Research Ethics clearance certificate | | No |
| 3. No human or animal ethics certificate is required | | No |
| 4. Unclear, will seek appropriate guidance from the HREC/AREC committees | | No |

vii. Has the Postgraduate student and supervisor/s signed the ethics declaration form Yes

**Overall recommendation regarding the protocol:**

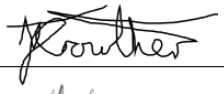
i. Revision of the protocol to the satisfaction of the Supervisor (**NB: if HoD approval is also required, please specify**): **Yes**
*(**Candidate**: one copy, list of corrections with page numbers and Supervisor approval letter – submit to PG Office).*

ii. Revision of the protocol to the satisfaction of the Assessor Group/Chair: No
*(**Candidate**: one copy, list of corrections with page numbers, Supervisor approval letter – submit to PG Office and PG Office to forward to the Assessor Group Chair).*

iii. Revision of the protocol and resubmission of the revised protocol to the next Assessor Group Meeting: No
*(**Candidate**: six copies, list of corrections with page numbers, Supervisor approval letter – submit one copy to PG Office / 5 to school assessor group administrator / for PhD, all six copies to be submitted to the PG Office).*

iv. Candidate goes ahead (*no revision required*): No

**Details of Assessors:**

| Name: | Email: | Sign: |
|---|---|---|
| Nigel Crowther | nigel.crowther@nhls.ac.za |  |
| Carolyn Padoa | carolyn.padoa@nhls.ac.za |  |
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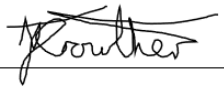
| | | |
| | | |
| | | |

**Details of Assessor Group Chair:**

| Name: | Email: | Sign: |
|---|---|---|
| Nigel Crowther | nigel.crowther@nhls.ac.za |  |

Date: 22nd February, 2023

NATIONAL HEALTH LABORATORY SERVICE
School of Pathology, University of the Witwatersrand

DIVISION OF HUMAN GENETICS

UNIVERSITY OF THE WITWATERSRAND, JOHANNESBURG

Hospital Street, Johannesburg, 2001 | PO Box 1038, Johannesburg, 2000
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19 March 2023

Postgraduate Office
Faculty of Health Science
Philip V Tobias Building

**RE: Amendments to MSc (Med) Genetic Counselling Protocol Submission**

To whom it may concern,

Amendments to the protocol entitled "The Utilisation of Genetic Counselling Services Amongst Prenatal Healthcare Providers in Gauteng, South Africa" have been made according to the suggestions made at the protocol assessors meeting on 22nd February 2023. The specific changes can be found in the attached document.

This letter serves as proof that we, the supervisors, are aware of and have approved the changes made to the protocol by the student, Megan Duvenhage (1076441).

Yours sincerely,

**Monica Araujo**
**MSc (Med) Genetic Counselling**
**Supervisor**

**Katryn Fourie**
**MSc (Med) Genetic Counselling**
**Co-supervisor**

Clinical & Counselling Section | Division of Human Genetics | School of Pathology
Faculty of Health Sciences | University of the Witwatersrand | National Health Laboratory Service
☎ 011 489 9223

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19 March 2023

Postgraduate Office
Faculty of Health Science
Philip V Tobias Building

**RE: Amendments to MSc (Med) Genetic Counselling Protocol Submission**

To whom it may concern,

Amendments to the protocol entitled "The Utilisation of Genetic Counselling Services Amongst Prenatal Healthcare Providers in Gauteng, South Africa" have been made according to the suggestions made at the protocol assessors meeting on 22nd February 2023. The specific changes were indicated with **bold font** in the adjusted protocol. The changes made can be found detailed below.

| Section | Page | Amendment Made |
|---|---|---|
| 5.1 | 8 | A sentence regarding the testing the functionality of the survey was added. |
| 5.2.1 | 9 | A further federation to contact was added. |
| 5.2.1 | 9 & 10 | A calculation and explanation were added to explain the relevant sufficient sample size that would be required for the study. |
| 5.4 | 10 | A paragraph was added to explain how the participants will be divided into subgroups to improve data analysis. |
| Appendix | ii | Heading "Section A" added |
| Appendix | ii | Question 1 was changed to a short answer question from checkbox categories |
| Appendix | iii | Heading "Section B" added |
| Appendix | iii | A new was added prior to question 3. |
| Appendix | iii | Question 3 was reworded and is now numbered question 4. |
| Appendix | vi | Heading "Section C" added |

Should these changes not be clear or should there be any questions please contact me.

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Yours sincerely,

**Megan Duvenhage**
**MSc (Med) Genetic Counselling Intern**

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## Appendix B: Participant information sheet

Return to methods.

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6 February 2023

To whom it may concern,

**RE: Invitation to participate in research study**

I, Megan Duvenhage, under the supervision of Monica Araujo and Katryn Fourie, from the Division of Human Genetics, am conducting a study in partial fulfillment of my MSc (Med) in Genetic Counselling. This study is entitled "The Utilisation of Genetic Counselling Services Amongst Prenatal Healthcare Providers in Gauteng, South Africa".

**Study aims:**
This project aims to establish the utility of genetic counselling services amongst prenatal healthcare providers as well as insights into the perceptions of genetic counselling and its usefulness by these individuals. The results obtained will provide information on the use of genetic counselling services and how they may be improved as to better patient care and management in the prenatal sector.

**Study objectives:**

1. To determine if, and why, prenatal healthcare providers refer patients to genetic counselling services.
2. To determine the factors that prenatal healthcare providers find important when deciding whether to refer a patient to genetic counselling services or not.
3. To identify the barriers and facilitators that prenatal healthcare providers experience when making a referral to genetic counselling services.
4. To assess how best to improve the knowledge of genetic counselling amongst prenatal healthcare providers.
5. To assess how the referral process and patient access to genetic counselling may be improved.

**Study methods:**
Should you wish to participate you will be sent a link to an online survey on Google Forms. There will a consent page that will need to be accepted before you may begin the survey. The survey should take 15 – 20 minutes to complete and will be made up of three sections: 1) demographics and professional practice of the participant, 2) the participants referrals to genetic counselling services, and 3) the participants understanding of genetic counselling services. It is expected that the survey is completed honestly and completely.

The information provided by you will be used anonymously and in combination with the data from other participants to curate a data set that will give insights into the above-mentioned topic.
The use the information you will provide is subject to:

1. Approval from the University of the Witwatersrand Human Research Ethics Committee (Medical)
2. Anonymity (i.e. there are no questions that require any self-identifying data)

If research arises that needs more detailed information you will be approached for further specific consent first. If at any time you choose to stop the survey and thereby withdraw consent, you will be free to do so and will not be prejudiced in any way.

**Benefits of being in the study:**
While there are no direct benefits for those wishing to partake in the study, the information provided by this study will give insight on how to best improve the utilisation and efficacy of the limited genetic counselling services available in Gauteng, South Africa.

**Thank you for reading this Study Information Sheet.**

This study has been approved by the Human Research Ethics Committee (Medical) of the University of the Witwatersrand, Johannesburg ("Committee"). A principal function of this Committee is to safeguard the rights and dignity of all human subjects who agree to participate in a research project and the integrity of the research.

If you have any concern over the way the study is being conducted, please contact the Chairperson of this Committee who is Dr Clement Penny, who may be contacted on telephone number 011 717 2301, or by e-mail on Clement.Penny@wits.ac.za. The telephone numbers for the Committee secretariat are 011 717 2700/1234 and the e-mail addresses are Zanele.Ndlovu@wits.ac.za and Rhulani.Mukansi@wits.ac.za

Yours sincerely,

**Megan Duvenhage**
**MSc (Med) Genetic Counselling Intern**

**Monica Araujo**
**MSc (Med) Genetic Counsellor**

Clinical & Counselling Section | Division of Human Genetics | School of Pathology
Faculty of Health Sciences | University of the Witwatersrand | National Health Laboratory Service
☎ 011 489 9227/ 082 887 8338 | ✉ megan.duvenhage@nhls.ac.za

## Appendix C: Participant informed consent

Return to methods.

NATIONAL HEALTH LABORATORY SERVICE
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**CONSENT FORM**
**PRENATAL HEALTHCARE PROVIDER GENETIC COUNSELLING SURVEY**

Dear Participant,

By continuing with this survey you are agreeing to participate in quantitative research being conducted into the "Awareness of Genetic Counselling Services Amongst Prenatal Healthcare Providers in Johannesburg, South Africa". This survey contains questions pertaining to your profession as well as your knowledge on genetic counselling and the availability of genetic counselling in your field of expertise.

The information provided by you will be used anonymously and in combination with the data from other participants to curate a data set that will give insights into the above mentioned topic.

The use the information you will provide is subject to:

1. Approval from the University of the Witwatersrand Human Research Ethics Committee (Medical)
2. Anonymity (i.e. there are no questions that require any self-identifying data)

If research arises that needs more detailed information you will be approached for further specific consent first. If at any time you choose to stop the survey and thereby withdraw consent, you will be free to do so and will not be prejudiced in any way.

**PARTICIPANT FIRST NAME:** .....

**PARTICIPANT SURNAME**: .....

SA ID/Passport No: .....

Contact Number: ..... Email Address: .....

I ..... agree to the above mentioned terms of the study, am aware of how my data will be used and willing choose to participate

Signature: ..... Date: .....

## Appendix D: Study survey

Return to instrumentation.

# The Utilisation of Genetic Counselling Services Amongst Prenatal Healthcare Providers in Gauteng, South Africa

If you have any questions or concerns please contact me at megan.duvenhage@nhls.ac.za or 011 489 9227 or 082 887 8338

* Indicates required question

### Consent to participate in study

Dear Participant,

By continuing with this survey you are agreeing to participate in quantitative research being conducted into the "Awareness of Genetic Counselling Services Amongst Prenatal Healthcare Providers in Gauteng, South Africa". This survey contains questions pertaining to your profession as well as your knowledge of genetic counselling and the availability of genetic counselling in your field of expertise.

The information provided by you will be used anonymously and in combination with the data from other participants to curate a data set that will give insights into the above-mentioned topic.

The use of the information you will provide is subject to:

1. Approval from the University of the Witwatersrand Human Research Ethics Committee (Medical)
2. Anonymity (i.e. there are no questions that require any self-identifying data)

If research arises that needs more detailed information you will be approached for further specific consent first. If at any time you choose to stop the survey and thereby withdraw consent, you will be free to do so and will not be prejudiced in any way.

1. **By checking the box below you are providing consent to proceed with the survey and participate in the research being conducted** *

*Tick all that apply.*

☐ I, the participant, agree to the above mentioned terms of the study, am aware of how my data will be used and willingly choose to participate

## Professional Practice

The following questions pertain to your profession and practice.

*Please answer by checking the correct box/s and by filling in information where applicable*

2. **Do you practice within Gauteng?** *

*Tick all that apply.*

- [ ] Yes
- [ ] No

3. **What is your current age?** *

_____

4. **What level of qualification do you hold?** *

_____

5. **What is your current profession?** *

*If applicable, multiple may be selected*

If it is not listed please choose "Other" and supply your job title

*Tick all that apply.*

- [ ] Fetal Medicine Specialist
- [ ] Fetal Medicine Registrar
- [ ] Fetal Medicine MO
- [ ] Obstetrician
- [ ] Obstetrics Registrar
- [ ] Obstetrics MO
- [ ] Gynaecologist
- [ ] Gynaecology registrar
- [ ] Gynaecology MO
- [ ] Sonographer
- [ ] Midwife
- [ ] Fertility Specialist
- [ ] Other: _____

6. **How long have you been working in your current profession?** *

*Tick all that apply.*

- [ ] Less than 6 Months
- [ ] Less than 5 years
- [ ] 5 – 10 years
- [ ] 10 – 15 years
- [ ] More than 15 years
- [ ] No longer practising

7. **In which of the South African healthcare systems do you work?** *

*Tick all that apply.*

- [ ] State
- [ ] Private
- [ ] Both

8. **In which hospital/s and or clinic/s do you practice?** *

*Should you not wish to disclose this information please write "NA"*

_____

9. **Which department do you practice under?** *

_____

## Referrals to Genetic Counselling

The following questions pertain to your referrals to genetic counselling services.

*Please answer by checking the correct box/s and by filling in information where applicable*

10. **Are you able to refer to genetic counselling services?** *

*Tick all that apply.*

- [ ] Yes
- [ ] No
- [ ] I am not sure
- [ ] It is not within my scope of practice

11. **If you answered "Yes" to the above question, how/ to whom would you refer your patients? If you did not answer yes please write "NA"** *

_____

12. Have you ever referred a patient to genetic counselling services? *

*Mark only one oval.*

- ◯ Yes
- ◯ No

13. **If you answered "Yes" to the previous question, how many referrals to genetic counselling services have you made in the last three months?** *

*Tick all that apply.*

- ☐ None
- ☐ 1 – 2
- ☐ 3 – 5
- ☐ 6 – 10
- ☐ > 10
- ☐ Not applicable - I previously answered "No"

14. **For which of the following reasons would you refer a patient for genetic counselling.** *

*Select all that apply*

*Tick all that apply.*

- [ ] A family history of a known genetic condition
- [ ] The birth of a previous child with a known genetic condition
- [ ] Poor obstetric history
- [ ] ≥ 3 recurrent first trimester miscarriages
- [ ] Soft markers seen on ultrasound
- [ ] Anomalies seen on ultrasound
- [ ] Termination of pregnancy counselling
- [ ] Pre-test counselling for Non-invasive prenatal testing counselling
- [ ] Non-invasive prenatal testing result delivery
- [ ] Carrier screening counselling
- [ ] Carrier screening result delivery
- [ ] Advanced maternal age (> 35 years old)
- [ ] Invasive testing required during pregnancy for a specific genetic condition in the fetus
- [ ] Delivery of invasive testing results
- [ ] Consanguineous relationships within a family
- [ ] Teratogenic exposure during pregnancy
- [ ] Patient requests to be referred to a genetic counsellor
- [ ] In vitro fertilisation and pre-implantation counselling
- [ ] Other: _____

15. **If anything, what seen on a scan would make you consider referring a patient for genetic counselling?** *

**(Please list examples or write "NA" if not applicable)**

_____

_____

_____

_____

16. If anything, what in a patient's medical history would make you consider referring a patient for genetic counselling * 

(Please list examples or write "NA" if not applicable)

17. **How important are each of the following factors when considering a referral to genetic counselling?** *

*Please select a level of importance for each option*

*Mark only one oval per row.*

| | Very important | Important | Moderately Important | Of Little Importance | Unimportant |
|---|---|---|---|---|---|
| **Patients interest in genetic counselling** | ◯ | ◯ | ◯ | ◯ | ◯ |
| **Severity of the disorder** | ◯ | ◯ | ◯ | ◯ | ◯ |
| **Availability of testing and turnaround times** | ◯ | ◯ | ◯ | ◯ | ◯ |
| **Availability of treatment for the disorder** | ◯ | ◯ | ◯ | ◯ | ◯ |
| **Desire for management recommendations** | ◯ | ◯ | ◯ | ◯ | ◯ |
| **Family's desire for recurrence information** | ◯ | ◯ | ◯ | ◯ | ◯ |

18. **If no referral is made to genetic counselling, how frequently are each of the following statements a reason?** *

*Please select how frequently each occurs*

*Mark only one oval per row.*

| | Very Frequently | Frequently | Occasionally | Rarely | Never |
|---|---|---|---|---|---|
| **Was not aware of genetic services** | ⬭ | ⬭ | ⬭ | ⬭ | ⬭ |
| **Do not know how to refer patients** | ⬭ | ⬭ | ⬭ | ⬭ | ⬭ |
| **Do not see much benefit for the patient** | ⬭ | ⬭ | ⬭ | ⬭ | ⬭ |
| **Do not understand what genetic counsellors do** | ⬭ | ⬭ | ⬭ | ⬭ | ⬭ |
| **Unable to get appropriate information from the genetic clinic** | ⬭ | ⬭ | ⬭ | ⬭ | ⬭ |
| **Do not feel it is my responsibility to refer** | ⬭ | ⬭ | ⬭ | ⬭ | ⬭ |

**The following questions pertain to your understanding of genetic counselling services.**

*Please answer by ticking the correct box/s and by filling in information where applicable*

19. **Of the following, which do you feel is the responsibility of a genetic counsellor?** *

*Select all that apply*

*Tick all that apply.*

- [ ] To help couples have a child with desirable characteristics
- [ ] To provide emotional support to patients and families coping with a genetic condition
- [ ] To provide information about how genetics contributes to health problems
- [ ] To provide treatment of genetic conditions
- [ ] To provide information about the chances of a child having a genetic condition
- [ ] To help people understand their options for genetic testing
- [ ] To provide referrals and resources for families with a genetic condition
- [ ] To prevent the birth of children with genetic conditions

20. **As per your knowledge, which of the following fields do genetic counsellors service.** *

*Select all that apply*

*Tick all that apply.*

- [ ] Prenatal care
- [ ] Oncology
- [ ] Paediatrics
- [ ] Haematology
- [ ] Adult

21. **How interested would you be in furthering your knowledge on genetics and genetic counselling?** *

*Tick all that apply.*

- [ ] Very interested
- [ ] It is something I would consider
- [ ] Indifferent
- [ ] Uninterested
- [ ] I feel comfortable with the level of knowledge I have

22. **If you are interested in increasing your knowledge of genetics, which of the following areas would you like to improve on?** *

*Select all that apply*

*Tick all that apply.*

- [ ] Overview of genetic disorders commonly seen by fetal medicine providers
- [ ] Identifying genetic resources for clients
- [ ] Genetic counselling and what the profession entails
- [ ] Referral procedure to genetic clinics
- [ ] New genetic technologies and advances

23. **If you would like to increase your knowledge, which of the following educational resources do you feel would be the most beneficial?** *

*Select all that apply*

*Tick all that apply.*

- [ ] Online resources
- [ ] Written information
- [ ] Academic lectures
- [ ] Discussion groups

24. **We welcome any additional thoughts and comments**

## Appendix E: Ethics clearance certificate

Return to Methods

M230268 MED23-02-036

UNIVERSITY OF THE WITWATERSRAND, JOHANNESBURG

R14/49 Miss Megan Duvenhage

**HUMAN RESEARCH ETHICS COMMITTEE (MEDICAL)**

**CLEARANCE CERTIFICATE NO. M230268 MED23-02-036**

| | |
|---|---|
| **NAME: (Principal Investigator)** | Miss Megan Duvenhage |
| **DEPARTMENT:** | School of Pathology, Division of Human Genetics<br>Edenvale Regional Hospital, Tambo Memorial Hospital,<br>Thelle Mogoerane Regional Hospital, SASOG, SASUOG<br>Chris Hani Baragwanath Academic Hospital, RMMCH,<br>Charlotte Maxeke Johannesburg Hospital |
| **PROJECT TITLE:** | The Utilisation of Genetic Counselling Services amongst Prenatal Healthcare Providers in Gauteng, South Africa |
| **DATE CONSIDERED:** | 24/02/2023 |
| **DECISION:** | Approved unconditionally |
| **CONDITIONS:** | |
| **SUPERVISOR:** | Miss M Araujo and Mrs K Fourie |
| **APPROVED BY:** | Prof P Ruff, Chairperson, HREC (Medical) |
| **DATE OF APPROVAL:** | 24/08/2023 **EXPIRY DATE:** 24/08/2028 |

**This clearance certificate is valid for 5 years from date of approval. Extension may be applied for.**

**DECLARATION OF INVESTIGATORS**

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** in January each year until study is closed. Failure to submit annual report will result in ethics clearance certificate suspension. 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). Email signed copy of this ethics clearance certificate prior to commencing with the study hrec-medical.researchoffice@wits.ac.za

Duvenhage

Principal Investigator Signature

24/08/2023

Date

**PLEASE QUOTE THE PROTOCOL NUMBER IN ALL ENQUIRIES**

## Appendix F: Plagiarism declaration

UNIVERSITY OF THE WITWATERSRAND, JOHANNESBURG

FACULTY OF HEALTH SCIENCES

**PLAGIARISM DECLARATION TO BE SIGNED BY ALL HIGHER DEGREE STUDENTS**

SENATE PLAGIARISM POLICY: APPENDIX ONE

I Megan Duvenhage (Student number: 1076441) am a student registered for the degree of MSc (Med) in Genetic Counselling in the academic year 2023.

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 for the above degree 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.
- I have included as an appendix a report from "Turnitin" (or other approved plagiarism detection) software indicating the level of plagiarism in my research document.

Signature:  Date: 4 November 2023

1

1.

## Appendix G: Turn-it-in-report

Megan_Duvenhage_MSc_Writeup_Final_for_Turnitin-1.docx

ORIGINALITY REPORT

| 10% | 5% | 9% | 4% |
|---|---|---|---|
| SIMILARITY INDEX | INTERNET SOURCES | PUBLICATIONS | STUDENT PAPERS |

PRIMARY SOURCES

| | | |
|---|---|---|
| 1 | Jamey Thom, Tabitha Haw. "Awareness of genetic counseling services among allied healthcare professionals in South Africa", Journal of Genetic Counseling, 2021<br>Publication | 5% |
| 2 | pericles.pericles-prod.literatumonline.com<br>Internet Source | <1% |
| 3 | orca.cardiff.ac.uk<br>Internet Source | <1% |
| 4 | Submitted to University of Witwatersrand<br>Student Paper | <1% |
| 5 | news.mak.ac.ug<br>Internet Source | <1% |
| 6 | Submitted to University of Technology, Sydney<br>Student Paper | <1% |
| 7 | studyres.com<br>Internet Source | <1% |
| 8 | link.springer.com<br>Internet Source | <1% |

| | | |
|---|---|---|
| 9 | Parece, Lucian. "Designing an Accessible Form for Pre-Session Education of Cancer Genetic Counseling Patients", Brandeis University, 2023<br>Publication | <1 % |
| 10 | openrepository.aut.ac.nz<br>Internet Source | <1 % |
| 11 | Jennifer G. R. Kromberg, Elaine B. Sizer, Arnold L. Christianson. "Genetic services and testing in South Africa", Journal of Community Genetics, 2012<br>Publication | <1 % |
| 12 | core.ac.uk<br>Internet Source | <1 % |
| 13 | www.medimond.com<br>Internet Source | <1 % |
| 14 | arno.uvt.nl<br>Internet Source | <1 % |
| 15 | Mashiane, Salome Edith. "Ultrasound Biosafety: Knowledge and Practices of Health Practitioners Who Perform Obstetric Scans in Sa", University of Johannesburg (South africa), 2021<br>Publication | <1 % |
| 16 | wol-prod-cdn.literatumonline.com<br>Internet Source | <1 % |

| | | |
|---|---|---|
| 17 | hdl.handle.net<br>Internet Source | <1% |
| 18 | Submitted to Padua College Limited<br>Student Paper | <1% |
| 19 | etd.uwc.ac.za<br>Internet Source | <1% |
| 20 | Karrie A. Hines. "Genetic Counselors' Perceived Responsibilities Regarding Reproductive Issues for Patients at Risk for Huntington Disease", Journal of Genetic Counseling, 10/23/2009<br>Publication | <1% |

| | | | |
|---|---|---|---|
| Exclude quotes | Off | Exclude matches | < 10 words |
| Exclude bibliography | Off | | |

## Appendix H: Journal of Genetic Counselling author guidelines

Return to submission cover sheet.

*(Retrieved from https://onlinelibrary.wiley.com/page/journal/15733599/homepage/author-guidelines)*

Journal of Genetic Counseling

# AUTHOR GUIDELINES

## SECTIONS



### 1. AIMS AND SCOPE

The Journal of Genetic Counseling (JOURNAL), published for the National Society of Genetic Counselors, is a timely, international forum addressing all aspects of the discipline and practice of genetic counseling. The JOURNAL focuses on the critical questions and challenges that arise at the interface between rapid advances in genetics and technology and the impact on individuals and communities at genetic risk. The publication provides genetic counselors, other clinicians and health educators, laboratory geneticists, bioethicists, legal scholars, social scientists, and other researchers with a premier resource on genetic counseling topics in national, international, and cross-national contexts.

As a crucial resource for genetic counselors and associated professionals, the JOURNAL'S primary purpose is to report original research in the following areas:

- **Genetic Counseling Theory, Methods, and Practice:** addresses theory development and evaluation, methods development and evaluation, current practice, and/or outcomes research relevant to the discipline and practice of genetic counseling in clinical or non-clinical settings;
- **Public Health, Public Policy, and Access and Genetics Service Delivery:** addresses public health genomics, health behaviors, policy aspects related to genetic counseling and genetic testing, precision medicine, models of genetics services delivery;
- **Education and Genetics Professional Workforce Issues:** addresses educational training, professional development, and workforce topics related to genetic counseling;
- **Ethical, Legal, Psychological, and Social Issues:** addresses ethical, legal, psychological, and/or social issues related to genetic counseling, genetic testing, genetic services, and/or genetic information regarding individuals, communities, and the public
- **Risk Assessment:** addresses algorithms, theoretical models, or empirical data for use in genetic counseling risk assessment
- **Minority and Health Disparities:** addresses diversity, equity, and inclusion topics relevant to the practice and discipline of genetic counseling

**Note:** The Journal of Genetic Counseling does not publish research involving non-human animals.

*Return to Guideline Sections*

## 2. SUBMISSION

Once the submission materials have been prepared in accordance with the Author Guidelines, manuscripts should be submitted via the JOURNAL'S Editorial Manager site: https://www.editorialmanager.com/jogc/default.aspx. More details on how to use Editorial Manager are also available at https://www.editorialmanager.com/jogc/default.aspx.

**Need assistance?** For help with submissions, please contact the Editorial Office at JOGC@Wiley.com. When necessary, the Editorial Office staff may refer questions to the Editor-in-Chief.

A manuscript is considered for review and possible publication on the condition that it is submitted solely to the JOURNAL, and that the manuscript or a substantial portion of it is not under consideration elsewhere. Preprints and/or presentation of the content at meetings prior to submission are acceptable. However, authors should kindly note that submission implies that the content has not been published or submitted for publication elsewhere except as a brief abstract in the proceedings of a scientific meeting or symposium or as a preprint. Note, the JOURNAL uses iThenticate's CrossCheck software to detect instances of overlapping and similar text in submitted manuscripts.

The submission system will prompt the author to use an ORCiD ID (a unique author identifier) to help distinguish their work from that of other researchers. Click here to find out more.

**Data Protection Statement:** By submitting or reviewing a manuscript for the JOURNAL, your name, email address, and affiliation, and other contact details the JOURNAL might require, will be used for the regular operations of the JOURNAL, including, when necessary, sharing with the publisher (Wiley) and partners for production and publication. The JOURNAL and the publisher recognize the importance of protecting the personal information collected from users and have practices in place to maintain the security, integrity, and privacy of the personal data collected and processed. You can learn more here.

*Return to Guideline Sections*

## 3. MANUSCRIPT TYPES AND GENERAL REQUIREMENTS

**MANUSCRIPT TYPES (see Table for additional details)**

**Original Article:** Manuscript reporting original quantitative, qualitative or mixed methods research using a form of systematic study or inquiry to address a question relevant to the discipline and practice of genetic counseling. Systematic reviews included in this category.

**Brief Report:** Manuscript reporting an observation that adds to the knowledge of the discipline and practice of genetic counseling.

**Case Study:** Manuscript addressing issues relevant to discipline and practice of genetic counseling by demonstrating and stimulating thought about a difficult ethical, counseling, or genetic testing situation the author has encountered.

**Professional Issue:** Manuscript reporting reflections by the author(s) on the discipline and practice of genetic counseling.

**Review:** Manuscript summarizing the literature on a focused topic. Authors should contact the Editor-in-Chief prior to submission.

**Commentary:** Manuscript addressing matters of interest or controversy to the readership. Generally commissioned by the Editor-in-Chief.

**Correspondence:** Manuscript addressing work previously published in the JOURNAL

**Practice Resource, Practice Guideline, Focused Revision:** Manuscripts addressing specific areas of genetic counseling practice. Commissioned by the National Society of Genetic Counselors' Practice Guidelines Committee.

**Book Review:** Manuscript that reviews a book of relevance to the JOURNAL scope.

**Conference Report:** Manuscript with executive summary of important conference and/or select conference abstracts. Authors should contact Editor-in-Chief prior to submission.

**Corrigenda and Errata:** Manuscript describing corrections to a work previously published in the JOURNAL.

## GENERAL REQUIREMENTS

### Format

- double-spaced
- 1 inch margins
- 12 point font (Arial or Times New Roman).
- Footnotes should be avoided in the main text. When their use is absolutely necessary, footnotes should be numbered consecutively using Arabic numerals and should be typed at the bottom of the page to which they refer. Place a line above the footnote, so it is set off from the text. Use the appropriate superscript numeral for citation in the text.

### English Language

Manuscripts must be submitted in grammatically correct American English. Manuscripts that do not meet this standard cannot be reviewed. Authors for whom English is a second language should consult an English-speaking colleague or consider having their manuscript professionally edited before submission to improve the English. A list of independent suppliers of editing services can be found at https://wileyeditingservices.com/en/. All services are paid for and arranged by the author, and use of one of these services does not guarantee acceptance or preference for publication.

### Inclusive Language

An important element of the Journal of Genetic Counseling's commitment to diversity, equity, and inclusion is fostering authors' use of inclusive language in their manuscripts. To facilitate use of inclusive

language we have compiled two excellent and comprehensive resources for authors' use:

CDC guidelines - **https://ehe.jhu.edu/DEI/Health_Equity_Style_Guide_CDC_Reducing_Stigma.pdf**

APA guidelines - **https://apastyle.apa.org/style-grammar-guidelines/bias-free-language**

Specific guidance:

1. Participant-reported demographic variables should be ascertained/used when reporting demographic information on participants.
2. Authors should explain the use of person-first or non-person-first language in the manuscript. For guidance, see https://apastyle.apa.org/style-grammar-guidelines/bias-free-language
3. For guidance on reducing stigma through use of terminology describing groups, see https://ehe.jhu.edu/DEI/Health_Equity_Style_Guide_CDC_Reducing_Stigma.pdf, and https://apastyle.apa.org/style-grammar-guidelines/bias-free-language
4. For guidance on use of terminology for sex and gender, see https://apastyle.apa.org/style-grammar-guidelines/bias-free-language

**General Style Points**

- **Abbreviations:** In general, terms should not be abbreviated unless they are used repeatedly and the abbreviation is helpful to the reader. Initially, use the word in full, followed by the abbreviation in parentheses. Thereafter use the abbreviation only.
- **Units of measurement:** Measurements should be given in SI or SI-derived units. Visit the Bureau International des Poids et Mesures (BIPM) website for more information about SI units.
- **Numbers and p-values:** numbers under 10 should be spelled out, except for: measurements with a unit (8 mmol/L); age (6 weeks old), or lists with other numbers (11 cousins, 9 aunts, 4uncles). Numerical figures (excluding p-values) should not exceed 2 decimal places. For p-values less than 0.001, report as $p < 0.001$. For p values greater than 0.001, use 2-3 decimal places. Do not use a leading zero for statistics that cannot exceed 1.0.
- **Genomic Terminology and Nomenclature:** Please use the following terms:
    - *genome sequencing* instead of *whole genome sequencing*
    - *exome sequencing* instead of *whole exome sequencing*
    - *pathogenic variant* instead of *mutation*
    - *secondary finding* instead of *incidental finding*

Use of italics

- italicize gene names (e.g. *FBN1*)
- do not italicize protein names (e.g. fibrillin)

Sequence variants

- Sequence variants should be described in the text and tables using both DNA and protein designations whenever appropriate.
- Sequence variant nomenclature must follow the current HGVS guidelines; see hgvs.org, where examples of acceptable nomenclature are provided.
- Human gene nomenclature should follow the standards of the HUGO Gene Nomenclature Committee (HGNC), see https://www.genenames.org/.

- **Pedigrees:** Pedigrees should follow the recommendations for standardized nomenclature accepted by the National Society of Genetic Counselors. Authors should consult the following references for these recommendations:
    - Bennett, R. L. , Steinhaus, K. A., Uhrich, S. B., O' Sullivan, C. K., Resta, R. G. , Lochner-Doyle, D., Markel, D. S., Vincent, V., & Hamanishi, J. (1995). Recommendations for Standardized Human Pedigree Nomenclature. Journal of Genetic Counseling, 4, 267-279.
    - Bennett, R. L., Steinhaus French, K., Resta, R. G., & Lochner Doyle, D. (2008). Standardized Human Pedigree Nomenclature: Update and Assessment of the Recommendations of the National Society of Genetic Counselors. Journal of Genetic Counseling, 17, 424-433.
- **Trade Names:** Chemical substances should be referred to by the generic name only. Trade names should not be used. Drugs should be referred to by their generic names. If proprietary drugs have been used in the study, refer to these by their generic name, mentioning the proprietary name and the name and location of the manufacturer in parentheses.

*Return to Guideline Sections*

## 4. PREPARING THE SUBMISSION

**Specific Requirements** for each manuscript type are summarized in this **Table.**

### Parts of the Manuscript

The manuscript parts should be uploaded as separate files:

- cover letter
- main text file
- tables
- figures
- supplementary information files

### Cover Letter

- include a statement that the work presented in the manuscript has not been published elsewhere and is not currently under review elsewhere. Include a statement that the work is available as a preprint or was presented at a conference, if relevant.
- If the study includes original data, at least one author must confirm in the cover letter that he or she had full access to all the data in the study and takes responsibility for the integrity of the data and the accuracy of the data analysis.

### Main Text File

The **main text file** should be presented in the following order in one document (as appropriate for manuscript type):

1. Title Page
2. Abstract and keywords
3. 1-2 sentence responses to:
    1. What is known about this topic:
    2. What this paper adds to the topic:
4. Main body of paper
5. Author Contributions
6. Acknowledgements
7. Compliance with Ethical Standards
    1. Conflict of Interest
    2. Human Studies and Informed Consent
    3. Animal Studies
    4. Data Availability Statement
8. References
9. Tables
10. Figure legends
11. Figures

***Title Page***

The title page should include (in this order):

- title of the article. Authors may benefit from referring to Wiley's best practice tips on Writing for Search Engine Optimization.
- authors' names (no degrees) in the order to be published. Please denote cases of equal authorship with a superscript and footnote, e.g., for joint first or joint senior authorship, the footnote should say 'X and Y should be considered joint first author' or 'X and Y should be considered joint senior author', respectively.
- authors' institutional affiliation(s) where the work was conducted, and the author's present affiliation if different from where the work was conducted. The affiliation should comprise the department, institution (usually university or company), city, and state (or nation) and should be noted with numbered superscript to the author's name and footnote.
- Telephone number and e-mail address of the one author designated to review proofs (the corresponding author).
- Suggested running head. The suggested running head should be less than 80 characters (including spaces) and should comprise the article title or an abbreviated version thereof

***Abstract***

- unstructured, i.e., no main headings or subheadings
- maximum 300 words
- contains major keywords summarizing the work
- If reporting on a clinical trial, include the name of the trial register and the clinical trial registration number at the end of the Abstract.
- See Table for manuscript category-specific information

***Keywords***

- 3 to 6 keywords
- **Please include 2-3 keywords from** this list.

***What is known about this topic***

- 1-2 sentences

***What this paper adds to the topic***

- 1-2 sentences

***Main Body of Paper***

- **Please refer to this Table for specific manuscript type requirements**.
- For all research involving human participants, please include a statement in the **Methods** section confirming that the study was reviewed by an institutional review board/human investigations committee/ethics committee (include name of committee and IRB protocol number) and approved or waived as human subjects research.

***Author Contributions***

Prior to submitting the manuscript all authors fulfilling the International Committee of Medical Journal Editors (ICMJE) criteria for authorship should be identified and should agree on the order in which their names will be listed in the manuscript.

ICMJE criteria are:

- Substantial contributions to the conception or design of the work; or the acquisition, analysis, or interpretation of data for the work; AND
- Drafting the work or revising it critically for important intellectual content; AND
- Final approval of the version to be published; AND
- Agreement to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

**If the study includes original data,** at least one author must confirm that he or she had full access to all the data in the study and takes responsibility for the integrity of the data and the accuracy of the data analysis.

**Please insert the following statements in the Author Contributions section**, specifically identifying the relevant author(s):

*Authors X and Y confirm that they had full access to all the data in the study and take responsibility for the integrity of the data and the accuracy of the data analysis. All of the authors gave final approval of this version to be published and agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.*

**During the submission process** you will be asked to select the contribution(s) made by each author from a pre-specified drop down menu.

***Acknowledgements***

Contributions from anyone who does not meet the ICMJE criteria for authorship should be listed in an Acknowledgements section. Financial and material support should also be mentioned in this section. Authors should list all funding sources and are responsible for the accuracy of their funder designation. If in doubt, please check the Open Funder Registry for the correct nomenclature: www.crossref.org/services/funder-registry.

If this paper is to be considered for the **Journal of Genetic Counseling Best Trainee Paper award**, please include a statement indicating that the research presented in the paper was conducted while the first author was in training or to fulfill a degree requirement of the first author. See the Best Trainee Paper Award tab on the JOURNAL website for more information about this award. Thanks to anonymous reviewers is not considered appropriate to include in Acknowledgements.

***Compliance with Ethical Standards***

- ***Conflict of Interest Statement***

The JOURNAL requires that all authors disclose any potential sources of conflict of interest. Any interest or relationship, financial or otherwise, which might be perceived as influencing an author's objectivity is considered a potential source of conflict of interest. These must be disclosed when directly relevant or directly related to the work that the authors describe in their manuscript. Potential sources of conflict of interest include, but are not limited to, patent or stock ownership, membership of a company board of directors, membership of an advisory board or committee for a company, and consultancy for or receipt of speaker's fees from a company. The existence of a conflict of interest does not necessarily preclude publication in the JOURNAL.

If the authors have no conflict of interest to declare, they must also state this in the manuscript. It is the responsibility of the corresponding author to review this policy with all authors and collectively to list in the manuscript under the subheading “Conflict of Interest” *all* pertinent commercial and other relationships.

The above policies are in accordance with the Uniform Requirements for Manuscripts Submitted to Biomedical Journals produced by the International Committee of Medical Journal Editors (http://www.icmje.org/).

The Conflict of Interest Statement should **mention each author separately by name.**

Recommended wording

*Author W declares that she has no conflict of interest.*

*Author X has received research grants from Drug Company A.*

*Author Y has received a speaker honorarium from Genetic Testing Company B and owns stock in Genetic Testing Company C.*

*Author Z is founder and CEO of Genetic Education Company D.*

If multiple authors declare no conflict, this can be done in one sentence:

*Author X, Author Y and Author Z declare that they have no conflict of interest.*

Submitting authors should ensure they liaise with all co-authors to confirm agreement with the final statement.

- ***Human Studies and Informed Consent***

The JOURNAL requires that all appropriate steps were taken to obtain informed consent of all human subjects participating in the research reported in the manuscript submitted for review and possible publication, and statements to this effect must be included under this subheading. Participant anonymity should be preserved and all identifying information should be excluded in the manuscript unless the information is essential for scientific purposes and the study participants or patients (or parents or guardians) have provided written informed consent for publication. Images and information from individual participants will only be published where the authors have obtained the individual's free prior informed consent. Photographs need to be cropped sufficiently to prevent human subjects being recognized (an eye bar must not be used because of insufficient de-identification).

The editors reserve the right to reject manuscripts that do not comply with these requirements. The author will be held responsible for false statements or failure to fulfill these requirements.

For manuscripts reporting studies that involve human participants, a statement identifying the ethics committee that approved the study and confirmation that the study conforms to recognized standards is required, for example: **Declaration of Helsinki**; **US Federal Policy for the Protection of Human Subjects**; or **European Medicines Agency Guidelines for Good Clinical Practice**. It should also state clearly in the text that all persons gave their informed consent prior to their inclusion in the study.

Examples (not exhaustive)

*Approval to conduct this human subjects research was obtained by the [name of institutional review board or ethics committee]. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Informed consent was obtained from all patients for being included in the study.*

*The study was approved by the [name of institutional review board or ethics committee]. No informed consent was required from subjects as data were anonymously extracted from the [name of system]. All procedures*

*followed were in accordance with US Federal Policy for the Protection of Human Subjects.*

*This study was approved by and conducted according to the ethical standards of the [name of institutional review board or ethics committee]. All applicable international, national, and/or institutional guidelines were followed. This study was approved by the IRB after expedited review and was granted an informed consent waiver.*

*This study was conducted in accordance with all guidelines set forth by the [name of institutional review board or ethics committee]. Informed consent for genetic testing was obtained from all individuals undergoing testing, and [name of institutional review board or ethics committee] waived authorization for use of de-identified aggregate data. Individuals or institutions who opted out of this type of data use were excluded.*

*This study was reviewed and granted an exemption by the [name of institutional review board or ethics committee]. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Implied informed consent was obtained for individuals who voluntarily completed the online survey and submitted their responses.*

*Approval to conduct this human subjects research was obtained by the [name of institutional review board or ethics committee]. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Informed consent was obtained from all patients for being included in the study.*

*The study was approved by the [name of institutional review board or ethics committee]. No informed consent was required from subjects as data were anonymously extracted from the [name of system]. All procedures followed were in accordance with US Federal Policy for the Protection of Human Subjects.*

*This study was approved by and conducted according to the ethical standards of the [name of institutional review board or ethics committee]. All applicable international, national, and/or institutional guidelines were followed. This study was approved by the IRB after expedited review and was granted an informed consent waiver.*

*This study was conducted in accordance with all guidelines set forth by the [name of institutional review board or ethics committee]. Informed consent for genetic testing was obtained from all individuals undergoing testing, and [name of institutional review board or ethics committee] waived authorization for use of de-identified aggregate data. Individuals or institutions who opted out of this type of data use were excluded.*

*This study was reviewed and granted an exemption by the [name of institutional review board or ethics committee]. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Implied informed consent was obtained for individuals who voluntarily completed the online survey and submitted their responses.*

If any identifying information about participants is included in the article, the following sentence should also be included:

*Informed consent was obtained from all participants for which identifying information is included in this article.*

Authors do not need to provide a copy of the consent form to the publisher; however, in signing the author license to publish, authors are required to confirm that consent has been obtained. Wiley has a standard patient consent form available for use.

- ***Animal Studies***

The JOURNAL does not publish non-human animal studies. To affirm that this is the case for your submission, please include the following sentence under this subheading in the manuscript:

*No non-human animal studies were carried out by the authors for this article*

- ***Data Availability Statement***

Authors are required to provide a data availability statement to describe the availability or the absence of shared data. When data have been shared/are available in a repository, authors are required to include in their data availability statement a link to the repository they have used/created, and to cite the data they have shared. Whenever possible the scripts and other artefacts used to generate the analyses presented in the paper should also be publicly archived. If sharing data compromises ethical standards or legal requirements then authors are not expected to share it.

Sample statements are **available here from Wiley Author Services**.

*Example with data repository*

Data Availability Statement

*Data collected for this study are available through the publicly available repository MINDS@Uw. The data file is available at* ***http://digital.library.wisc.edu/1793/78637***

Citation in Reference List

*Donahue, A., Hall, A., & Petty, E. (2018). Genetic counselor information needs & current library services for genetic counselor. [Data file] Retrieved from* ***http://digital.library.wisc.edu/1793/78637***

Although it would be rare for a paper submitted to the JOURNAL to report novel nucleotide sequence data, should that be the case, the novel nucleotide sequence data including genetic mutations must be

submitted to a public database prior to publication and a sentence naming the database should be included in the manuscript.

***References***

The accuracy of references is the responsibility of the authors. The JOURNAL **strongly prefers references that have undergone peer review** and are not conference abstracts, unpublished masters' theses, unpublished dissertations, unpublished data in manuscripts, etc. However, if essential to the manuscript, they should be cited and referenced appropriately.

References should be prepared according to the *Publication Manual of the American Psychological Association* (6th edition). The APA website includes a range of **resources for authors** learning to write in APA style, including **an overview** of the manual, **free tutorials** on APA Style basics, and an **APA Style Blog**. For more information about APA referencing style, please also refer to the **APA FAQ**.

EndNote users can download the style **here**.

***In-text citation***

- follow the author-date method whereby the author's last name and the year of publication for the source should appear in the text, for example, (Jones, 1998)
    - publications with no authors, use brief phrase to describe publication and year of the publication, for example, ("Report of 1979 Business Meeting", 1979, p. 1) or ("NSGC Professional Status Survey: Executive Summary", 2020)
    - publications with no dates, use n.d., for example, ("American board of Genetic Counseling, Mission and History", n.d.)
- Multiple citations should be listed alphabetically by author's last name

**Personal communications**

- cite within the text as (Name of person providing the communication, personal communication, Date of communication), for example (Jane Doe, ABCD Executive Director, personal communication, September 2019)
- do not include in the reference list
- permission in writing from the communicator is required. Submit written permission with manuscript.

***Reference List***

- alphabetical by last name of first author
- the reference list is not numbered

***General Comments***

- Digital Object Identifier (DOI) should be provided for all references where available
- For journal articles, issue numbers are not included unless each issue in the volume begins with page one.
- For references with more than seven author names list first six with three dots and then last author name.

***Reference examples***

- *Journal article with 7 or fewer authors*
  - Beers, S. R., & De Bellis, M. D. (2002). Neuropsychological function in children with maltreatment-related posttraumatic stress disorder. *The American Journal of Psychiatry, 159,* 483–486. *doi:*1176/appi.ajp.159.3.483

- *Journal article with more than 7 authors*
  - Reuter, C. M., Kohler, J. N., Bonner, D., Zastrow, D., Fernandez, L., Dries, A., ... Wheeler, M. T. (2019). Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing. *Journal of Genetic Counseling, 28,* 1107-1118. doi: 10.1002/jgc4.1161

- *Book with 7 or fewer authors*
  - Bradley-Johnson, S. (1994). *Psychoeducational assessment of students who are visually impaired or blind: Infancy through high school* (2nd ed.). Austin, TX: Pro-ed.

- *Book with 8 or more authors*
  - Gilbert, J. R., Smith, J. D., Johnson, R. S., Anderson, A., Plath, S., Martin, G., . . . White, N. (2014*). Choosing a title* (2nd ed.). New York, NY: Unnamed Publishing.

- *Internet Document*
  - Norton, R. (2006, November 4). How to train a cat to operate a light switch [Video file]. Retrieved 4/20/2020 from http://www.youtube.com/watch?v=Vja83KLQXZs
  - Note: only include the retrieval date if the content is likely to change

- *Publicly available data repository*
  - Donahue, A., Hall, A., & Petty, E. (2018). Genetic counselor information needs & current library services for genetic counselor. [Data file] Retrieved from http://digital.library.wisc.edu/1793/78637

- *Newsletter/newspaper, no author*
  - Report of 1979 Business Meeting of the NSGC. (1979). *Perspectives in Genetic Counseling*, 1(4), 1. Retrieved from nsgc.org [members only access]

- *Newsletter/newspaper, author*
  - Smith, A.C.M. (1982). "The sermon on the amount:" The status of NSGC finances. *Perspectives in Genetic Counseling*, 4(1), 2. Retrieved from nsgc.org [members only access]

- NSGC Professional Status Survey
  - NSGC Professional Status Survey: Executive Summary (2020). p. 2. Retrieved from https://www.nsgc.org/page/whoaregeneticcounselors

***Figure Legends***

Every figure must have a legend that includes the figure number and figure title. Legends should be concise but comprehensive – the figure and its legend must be understandable without reference to the text. The figure legend should Include definitions of any symbols used and define/explain all abbreviations and units of measurement.

**Additional Files**

***Tables***

Tables should be self-contained and complement, not duplicate, information contained in the text. The table must be understandable without reference to the text.

Tables should:

- be numbered and referred to by number in the text.
- have a brief explanatory title,
- have a concise but comprehensive legend in the case where additional explanation is provided using footnotes. Footnotes should be indicated by superscript lowercase letters
- define all abbreviations in the legend.
- be supplied as editable files, not pasted as images
- be uploaded as separate file(s)

***Figures***

Authors are encouraged to send the highest quality figures possible. Line art should be exported at 600 dpi or higher, and halftone images should be exported at 300 dpi or higher.

Figures should:

- be numbered and referred to by number in the text
- be clearly labeled
- be uploaded as separate file(s)

*Color figures.* Color figures may be published online free of charge.

***Supporting Information***

Supporting information is information that is not essential to the article, but provides greater depth and background. It is hosted online and appears without editing or typesetting. It may include copies of surveys or interview questions, consent forms, tables, figures, videos, datasets, etc.

Click here for Wiley's FAQs on Supporting Information.

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## 5. EDITORIAL POLICIES AND ETHICAL CONSIDERATIONS

### Peer Review and Acceptance

The acceptance criteria for all papers are the quality and originality of the research and its significance to JOURNAL readership and the practice and discipline of genetic counseling. Papers will only be sent to review if the Editors determine that the paper meets the appropriate quality and relevance requirements.

Except where otherwise stated, manuscripts are single-blind peer reviewed. Wiley's policy on the confidentiality of the review process is **available here.**

### Pre-Print Policy

The JOURNAL will consider for review articles previously available as preprints. Authors may also post the **submitted version** of a manuscript to a preprint server at any time. Authors are requested to update any pre-publication versions with a link to the final published article.

### Revisions

When submitting a revised manuscript please also submit:

- Response to reviewer comments in the form of a table with reviewers' suggestions on the left-side and edits made/how addressed on the right-side
- A marked version (tracked, highlighted, etc.) and unmarked version of revised manuscript

### Changes in Authorship

Prior to submitting the manuscript all authors should agree on the order in which their names will be listed in the manuscript. Any changes to authorship, including adding, removing or rearranging the authorship list, must be made before the manuscript has been accepted and only with approval from the Editor. The corresponding author is expected to write to the Editor with the reason for the change and will complete an Authorship Change Request form with written agreement from ALL authors, including those affected by any change. Only in exceptional circumstances will the Editor consider changes in authorship after a paper has been accepted. If a request for change in authorship is submitted after acceptance but before publication, the article will be suspended from proceeding to publication until the authorship change request has been resolved. Once an article has been published in an online issue, a completed Authorship Change Request form and a corrigendum will need to apply to reflect any allowed changes in authorship.

### Decision Appeals

Appeals should be filed within 28 days of notification of the decision. The appeal should be in the form of a letter addressed and submitted to the Journal of Genetic Counseling Editorial Office at JOGC@wiley.com. The letter should include clear and concise grounds for the appeal, including specific points of concern. The appeal will then be assessed by the Journal of Genetic Counseling management team, led by the

Editor-in-Chief, and informed by the subsequent editorial communications. You will be informed of the outcome of the appeal in writing, normally within 28 days. The decision will be final.

### Clinical Trial Registration

The JOURNAL requires that clinical trials are prospectively registered in a publicly accessible database and clinical trial registration numbers are included in all papers that report their results. Authors are asked to include the name of the trial register and the clinical trial registration number at the end of the Abstract. If the trial is not registered, or was registered retrospectively, the reasons for this should be explained.

### Research Reporting Guidelines

Accurate and complete reporting enables readers to fully appraise research, replicate it, and use it. Authors are encouraged to adhere to recognized research reporting standards. The EQUATOR Network collects more than 370 reporting guidelines for many study types, including for:

- Randomized trials: CONSORT
- Observational studies: STROBE
- Systematic reviews: PRISMA
- Qualitative research: COREQ
- Quality improvement studies: SQUIRE
- Study protocols: SPIRIT
- **Studies reporting on genetic counseling as an intervention should follow the reporting standards found here:** Standards for the Reporting of Genetic Counseling Interventions in Research and Other Studies (GCIRS)

### Publication Ethics

The JOURNAL is a member of the **Committee on Publication Ethics (COPE)**. Read Wiley's Top 10 Publishing Ethics Tips for Authors **here**. Wiley's Publication Ethics Guidelines can be found **here**.

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## 6. AUTHOR LICENSING

If a paper is accepted for publication, the author identified as the formal corresponding author will receive an email prompting them to log in to **Author Services**, where via the Wiley Author Licensing Service (WALS) they will be required to complete a copyright license agreement on behalf of all authors of the paper. This email will be issued within a few days of paper acceptance.

- For authors signing the copyright transfer agreement

If the Hybrid Open Access option [requires payment] is not selected the corresponding author will be presented with the copyright transfer agreement (CTA) to sign. The terms and conditions of the CTA can be previewed in the samples associated with the **Copyright FAQs**.

- For authors choosing Hybrid Open Access

If the Hybrid Open Access option is selected the corresponding author will have a choice of the following Creative Commons License Open Access Agreements (OAA):

Creative Commons Attribution License (CC-BY) OAA

Creative Commons Attribution Non-Commercial License (CC-BY-NC) OAA

Creative Commons Attribution Non-Commercial -NoDerivs License (CC-BY-NC-ND) OAA

General information regarding licensing and copyright is available on the **Wiley Author Services** and the **Wiley Open Access websites**.

Note to NIH, The Wellcome Trust and the Research Councils UK Grantees

Pursuant to NIH mandate, Wiley will post the accepted version of contributions authored by NIH grant-holders to PubMed Central upon acceptance. This accepted version will be made publicly available 12 months after publication. Please click **here** for further information. If you select the Hybrid Open Access option and your research is funded by The Wellcome Trust or the Research Councils UK (RCUK) you will be given the opportunity to publish your article under a CC-BY license supporting you in compliance with The Wellcome Trust and Research Councils UK requirements.

- *Self-Archiving Definitions and Policies*

Note that the JOURNAL'S standard copyright agreement allows for self-archiving of different versions of the article under specific conditions. Please click **here** for more detailed information about self-archiving definitions and policies.

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## 7. PUBLICATION PROCESS AFTER ACCEPTANCE

**Accepted Articles**

All accepted manuscripts are subject to editing. Authors have final approval of changes prior to publication.

*Proofs*

Once the paper is typeset, the author will receive an email notification with full instructions on how to provide proof corrections.

Please note that the author is responsible for all statements made in their work, including changes made during the editorial process – authors should check proofs carefully. Note that proofs should be returned within 48 hours from receipt of first proof.

*Publication Charges*. There are no publication charges for the Journal of Genetic Counseling.

*Color figures.* Color figures may be published online free of charge.

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## 8. POST PUBLICATION

### Access and Sharing

### Early View

The JOURNAL offers rapid publication via Wiley's Early View service. Early View (Online Version of Record) articles are published on Wiley Online Library before inclusion in an issue. Note that there may be a delay after corrections are received before your article appears online, as Editors also need to review proofs. Once your article is published on Early View no further changes to your article are possible. Your Early View article is fully citable and carries an online publication date and DOI for citations.

When the article is published online:

- The author receives an email alert (if requested).
- The link to the published article can be shared through social media.
- The author will have free access to the paper (after accepting the Terms & Conditions of use, they can view the article).
- The corresponding author and co-authors can nominate up to ten colleagues to receive a publication alert and free online access to the article.

For additional important information on Wiley's Reuse policy, click here.

### Promoting the Article

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### Measuring the Impact of an Article

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## 9. WILEY AUTHOR RESOURCES

### Wiley Author Resources

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## 10. EDITORIAL OFFICE CONTACT DETAILS

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*JOGC Author Guidelines updated 6/18/2020*



National Society of Genetic Counselors

*Page length excludes supplemental information. Abstract, tables, figures, and references are included in page count.

| Article Type | Description | Abstract | Key Words | What is known/what is new | Display Items | Max Page Length* | Reporting Guidelines |
|---|---|---|---|---|---|---|---|
| Original Article | Manuscripts reporting original quantitative, qualitative or mixed methods research using a form of systematic study or inquiry to address a question relevant to the discipline and practice of genetic counseling.<br><br>Systematic study or inquiry can be approached using a variety of methods, such as empirical methods, systematic literature review methods, normative or conceptual research methods.<br><br>Manuscript divided into main and subsections. Main section headings, e.g., Introduction, Methods, Results, Discussion, Conclusions, etc..<br><br>Methods subheadings should include, as appropriate: Participants, Instrumentation, Procedures, and Data Analysis.<br><br>Discussion subheadings should include, as appropriate: Study Limitations, Practice Implications<br><br>Discussion should begin with a very succinct summary of the major conclusions of the paper and then go | Yes, Unstructured 300 words max<br><br>relevance of topic/background information, study's objective, methods or methodological approach, sample, measures or main outcome variables, main results, conclusion | 3-6, including subject classifications from this list | 1-2 sentences each about what is known about the topic and what the paper adds to the topic | Max 5 tables+figures<br><br>Submit additional display items as online Supplemental Information | Quantitative research: 25 pages double-spaced<br><br>Qualitative research: 35 pages double-spaced | • Randomized trials: CONSORT<br>• Observational studies: STROBE<br>• Systematic reviews: PRISMA<br>• Qualitative research: COREQ<br>• Quality improvement studies: SQUIRE<br>• Genetic Counseling Intervention Studies: GCIRS |

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| | on to focus on the interpretation and significance of the findings with concise objective comments that describe their relation to other work in the area. It should not repeat information in the results. | | | | | | |
| Brief Report | Very brief reports offered in a letter format reporting an observation that adds to the knowledge of the discipline and practice of genetic counseling.<br><br>Manuscript is not subdivided into sections | Yes, unstructured, 300 words max<br><br>relevance of the topic/background information, study's objective, methods or methodological approach, sample, measures or main outcome variables, main results, and conclusion | 3-6, including subject classifications from this list | 1-2 sentences each about what is known about the topic and what the paper adds to the topic | Max 2 tables+figures<br><br>Submit additional display items as online Supplemental Information. | 9 pages double spaced | n/a |
| Case Study | Case studies inform/address issues relevant to genetic counseling by demonstrating and stimulating thought about a difficult ethical, counseling or genetic testing situation the author has encountered.<br><br>Manuscript should address observations of patient encounters | Yes, unstructured, 300 words max<br><br>relevance of topic/background information, | 3-6, including subject classifications from this list | 1-2 sentences each about what is known about the topic and what the paper adds to the topic | Max 2 tables+figures<br><br>Submit additional display items as online Supplemental Information. | 15 pages double-spaced | n/a |

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| | (usually 1-3) or a single small family that add substantially to the practice and discipline of genetic counseling.<br><br>Manuscript should have headings: Introduction, Case Description, Discussion<br><br>Note: the Journal of Genetic Counseling does not publish case studies whose sole or primary purpose is to report clinical and molecular information. | report, main issue, summary/conclusion | | | | | |
| Professional Issue | This article type features reflections by the author(s) on the discipline and practice of genetic counseling.<br><br>Manuscript may have headings, as appropriate | Yes, unstructured, 300 words max<br><br>Relevance of topic/background, main issue, summary/conclusion | 3-6, including subject classifications from this list | no | Max 2 tables+figures<br><br>Submit additional display items as online Supplemental Information. | 25 pages double-spaced | n/a |
| Review | Authors should contact the Editor-in-Chief prior to submission.<br><br>Note: submissions that describe a systematic process for reviewing the literature to address a research question (e.g., systematic reviews, scoping reviews) are included in the Original Article category | Yes, unstructured, 300 words max<br><br>Relevance of topic/background, review objective, methodological approach, main results, summary/conclusion | 3-6, including subject classifications from this list | no | Max 5 tables+figures<br><br>Submit additional display items online Supplemental Information. | 25 pages double-spaced | n/a |

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| Commentary | Commentaries/Editorials often address matters of interest or controversy to the readership. Generally commissioned by the Editor. | none | 3-6, including subject classifications from this list | no | None expected. Discuss with Editor | Max 15 pages double spaced | n/a |
| Correspondence | Brief, to-the-point letters to the editor that generally comment on work previously published in the Journal of Genetic Counseling. Correspondence is subject to editorial or peer review. The corresponding author of the original manuscript which is the subject of the submitted letter will be offered the opportunity to respond. If a response is provided, every effort will be made to publish these letters together. Only one round of comment is allowed. | none | none | no | none | Max 2 pages double spaced | n/a |
| Practice Resource | This article type addresses specific areas of genetic counseling practice and are commissioned by the National Society of Genetic Counselors' Practice Guidelines Committee. | Yes, unstructured, 300 words max<br><br>Per PGC template | 3-6, including subject classifications from this list | no | Discuss with editor | Discuss with editor | n/a |
| Practice Guideline | These article types address specific areas of genetic counseling practice and are commissioned by the National Society of Genetic Counselors' Practice Guidelines Committee. | Yes, unstructured, 300 words max<br><br>Per PGC template | 3-6, including subject classifications from this list | no | Discuss with editor | Discuss with editor | n/a |
| Focused Revision | These article types address specific areas of genetic counseling practice and are commissioned by the National Society of Genetic Counselors' Practice Guidelines Committee. | Yes, unstructured, use "Purpose" heading | Not required | no | Discuss with editor | Discuss with editor | n/a |

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| | | instead of "Abstract"<br><br>Per PGC template | | | | | |
| Book Review | Contact the Editor-in-Chief with a request to submit a book review. The topic of the reviewed book should be closely aligned with the mission of the Journal of Genetic Counseling. | none | none | no | none | 3 pages max, double spaced | n/a |
| Conference Report | The Journal of Genetic Counseling occasionally publishes an executive summary of an important conference or scientific meeting that involves topics related to the scope of the Journal. The Journal also on occasion publishes the abstracts of an important meeting on a selected basis. Authors should contact the Editor-in-Chief prior to submission. | none | 3-6, including subject classifications from this list | no | Discuss with editor | Discuss with editor | n/a |
| Corrigenda and Errata | Brief communications that correct errors in work previously published in the Journal of Genetic Counseling. Corrigenda correct errors that were responsibility of the author(s), and errata correct errors that are responsibility of the Journal, including editorial staff and production. These may be written by the corresponding author of the relevant manuscript or they may be composed by an editor. | no | no | no | Contact editor-in-chief | Contact editor-in-chief | n/a |

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*JOGC Author Guidelines update 6/18/2020*