Research Project Mini Dissertation

Medical Practitioners' use of Genetic Counselling services in Cape Town

By

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(PRTWIL008)

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2. Abbreviations

DTC - Direct-to-Consumer

GP - General health practitioner

HBOC - Hereditary Breast and Ovarian Cancer syndrome

HREC - Human Resources Ethics Committee

MDT - Multidisciplinary team

MP - Medical practitioners

PCP - Primary care provider

POPIA - Protection of Personal Information Act (2013)

REM - Reciprocal-Engagement Model

RWOPS - The Remunerative Work Outside Public Service

SASHG - Southern African Society of Human Genetics

VUS - Variant of unknown significance

3. Summary

The genetic counselling profession has grown exponentially in the last decade. Major advances in genetic technologies contributed to the increased need for these professionals. In South Africa, medical practitioners in private practice are increasingly making use of the services of genetic counsellors in the routine clinical management of their patients. In this research study medical practitioners in private practice in Cape Town, who actively utilise genetic counselling services, were recruited to investigate why they make use of genetic counselling services. A total of twelve medical practitioners were recruited for online semi-structured interviews in Afrikaans or English. The interviews were transcribed, and data was analysed using thematic analysis. From the interviews three broad themes were identified namely: the identity of genetics and genetic counselling services, resource limitations, and the appreciable impact of genetic counselling. Some interesting points of discussion included practitioners' views on direct-to-consumer testing platforms and the ambiguity around the professional title of a genetic counsellor. This research study is, according to our knowledge, one of the first global attempts to interview medical practitioners specifically who actively make use of genetic counselling services. It provides insights that could be used to promote the service throughout South Africa and other countries across the world. Future research that includes the perspectives of medical practitioners throughout South Africa would allow for even greater perspective on utilising genetic counselling services.

4. Chapter 1: Introduction

Since the first programme in 1969 (Marks, 2004; Means, Cirino, Swenson, & Austin, 2020; Veach, Bartels, & LeRoy, 2007), genetic counselling has grown exponentially with the completion of the Human Genome Project and regular breakthroughs in the field of genetic science and technology (Means et al., 2020). In many clinical practices, genetic counsellors are appreciated as members of multidisciplinary teams (MDT) involved with patient care by providing genetic services (Carroll et al., 2021; Paneque et al., 2017; Pasquier et al., 2022). There are however many people across the world, including medical practitioners (MPs), patients, and the public, who do not understand the integration of genetic counselling in a standardised healthcare regime. In developing countries such as South Africa, communicable diseases are of great concern (J. G. Kromberg, Wessels, & Krause, 2013) and the role of genetics and genetic counselling is not yet fully integrated into healthcare.

Attempts in searching for literature evaluating the service of genetic counsellors through research platforms such as Google Scholar, PubMed, Elsevier, Nature, and the Journal of Genetic Counselling were extremely limited. Keywords used to search these platforms include genetic counsel-, understanding/knowledge, awareness, education/training, and benefit. Research articles identified using these keywords which investigated the understanding of genetic services among healthcare professionals other than medical practitioners such as nurses were not considered relevant for this literature review. The references of identified articles published in the last three decades were perused to identify additional relevant research articles.

Research endeavours appraising genetic counselling services in the last 20 years were in the form of surveys and questionnaires which usually aimed to assess the genetic knowledge of different healthcare professionals (Bensend, Veach, & Niendorf, 2014; Borle et al., 2022; Brett, Metcalfe, Amor, & Halliday, 2012; Carroll et al., 2021; Cordier, Taris, Moldovan, Sobol, & Voelckel, 2016; Düsterwald, 2015; Fountzilas et al., 2022; Thom & Haw, 2021; Walters, Aldous, & Malherbe, 2022) or assess the service from the viewpoint of patients (Davey, Rostant, Harrop, Goldblatt, & O'Leary, 2005; Macleod, Craufurd, & Booth, 2002; Pasca, Carroll, Cragun, & Cragun, 2022). This limited perspective regarding genetic counselling services inspired the conception of this research project to bridge the literature gap and explore why MPs in private practice use genetic counselling services.

What is a genetic counsellor?

The genetic counselling service can be defined, according to the National Society of Genetic Counsellors' taskforce in 2006, as a process where people are assisted in understanding the medical, psychological, and familial implications of genetic contributions to disease (Resta et al., 2006; Skirton, Cordier, Ingvoldstad, Taris, & Benjamin, 2015). In other words, genetic counsellors are professionals who are well-versed in a triad of knowledge consisting of science, medicine, and psychology (Abacan et al., 2019; Jenkins & Arribas-Ayllon, 2016; J. G. Kromberg, T.-M. Wessels, et al., 2013; Rantanen et al., 2008; Skirton et al., 2015). These individuals are trained to communicate with patients from different cultural backgrounds and can carefully

explain the genetic risks, management, and testing options that could be relevant to a patient and their family (Abacan et al., 2019; J. G. Kromberg, T.-M. Wessels, et al., 2013; Macleod et al., 2002; O'Shea et al., 2011; Rantanen et al., 2008; Skirton et al., 2015). Genetic information is shared with counselees in a non-directive manner to allow them to make autonomous decisions with regards to available genetic testing and treatment options (Biesecker & Marteau, 1999; Blacker, Racette, Sklar, & Smoller, 2005; Macleod et al., 2002).

With advancements in the genetic field the role of a genetic counsellor is ever-expanding (J. Kromberg, Parkes, & Taylor, 2006; J. G. Kromberg, T.-M. Wessels, et al., 2013; Means et al., 2020; Pasca et al., 2022). These highly trained professionals are being appointed in an assortment of settings including clinical and scientific research, policy, public health, pharmacogenetics, and more (Chou, Duncan, Hallford, Kelley, & Dean, 2021; Pasca et al., 2022). Based on the training received by these individuals (graduate or postgraduate), they are qualified to educate MPs and assist patients with cognitive processing of complicated medical and genetic concepts (Skirton et al., 2015). Compared to developing countries such as South Africa, many developed countries have implemented novel genetic technologies which provide even more opportunities to appoint trained genetic counsellors (Borle et al., 2022; Chou et al., 2021; Kim et al., 2022).

Genetics and genetic counselling in the medical profession

Medical doctors refer patients to genetic counsellors with the purpose of helping patients and their families understand the type of genetic testing options that are available to them. This allows patients to make informed decisions with regards to their own medical management (Abacan et al., 2019; Druker et al., 2017; Veach et al., 2007). During a genetics consultation, the genetic counsellor will typically obtain a detailed family history, identify at-risk family members, offer and establish additional psychological support, and explain the limitations and implications of the genetic tests with the patient and their support partners and/or families (Abacan et al., 2019; Veach et al., 2007). After this initial assessment, a follow-up consultation is arranged where the result of the genetic test is delivered and any uncertainty regarding genetic information is managed (Druker et al., 2017; Patch & Middleton, 2018). Downstream implications for the patient and their family are also discussed in this follow up consultation whereby at-risk family members will be invited for a genetic counselling consultation. During these cascade consultations, the genetic counsellor will explain the genetic risks to the family members, similar to the initial assessment, before offering genetic testing to identify the presence of the at-risk gene which has been identified in the family (Abacan et al., 2019; Veach et al., 2007).

Many medical specialists and primary care providers (PCP) practicing medicine in South Africa (Düsterwald, 2015; Thom & Haw, 2021; Walters et al., 2022) or internationally (Baars, Scherpbier, et al., 2005; Hallowell, Cooke, Crawford, Parker, & Lucassen, 2009; Harding et al., 2019; Van Langen, Birnie, Leschot, Bonsel, & Wilde, 2003) are not confident about genetic concepts, making it difficult for them to counsel patients with genetic conditions. Medical specialists are trained to focus more on disease management, but genetic factors impacting current health, continuous care

management, reproductive health, progeny, and prophylactic decisions also need to be considered and explained to patients during their clinical consultations (Makransky et al., 2016; Pasquier et al., 2022). General health practitioners as well as PCPs in South Africa, Europe and the USA were found to have limited knowledge and expertise in the field of clinical genetics (Beene-Harris, Wang, & Bach, 2007; Biesecker & Marteau, 1999; Düsterwald, 2015; Klitzman et al., 2013; Samuel, Dheensa, Farsides, Fenwick, & Lucassen, 2017; Walters et al., 2022; Wonkam, Njamnshi, & Angwafo, 2006). This is especially seen in MPs who graduated before 1990 as their curriculum did not include many genetic concepts since it was a relatively new topic (Donohue et al., 2021; Hallowell et al., 2009; Pasquier et al., 2022). In a recent study by Fountzilas and colleagues, the authors found that only 15% of 208 Greek physicians have had some training or experience in genetic counselling (Fountzilas et al., 2022).

Many MPs believe that the diagnosis and management of genetic diseases should be done solely by trained genetic medical specialists as MPs generally do not feel comfortable providing these services for patients themselves (Biesecker & Marteau, 1999; Hallowell et al., 2009; Pasquier et al., 2022). Non-genetic physicians in South Africa, the USA, and the UK believe medical specialists (i.e., cardiologists, oncologists, paediatrician) need to perform genetic counselling in their respective specialities as the medical field continues to evolve, particularly if they are more likely to encounter genetic conditions (Biesecker & Marteau, 1999; Blacker et al., 2005; Patch & Middleton, 2018; Wonkam et al., 2006). This includes MPs such as obstetricians who often attend cases related to congenital abnormalities and genetic diseases such as Cystic fibrosis, Sickle cell anaemia, and Down syndrome. This viewpoint from non-genetic MPs implies that the information provided to patients who are seen in primary care may be limited, especially if they are affected with a genetic disease that the managing clinicians are not familiar with (Brett et al., 2012; Case, Johnson, Andrews, Allard, & Kelly, 2004; Harding et al., 2019). Some MPs also have their own opinions regarding the importance of genetics and the nature of MP that is truly capable of explaining genetic concepts to patients (Biesecker & Marteau, 1999; Geller et al., 1993; Klitzman et al., 2013; Parikh, Dickerson, & Kurian, 2020; Pasquier et al., 2022; Slomp et al., 2022). Personal opinions or attitudes regarding genetic tests including single gene disorders, prenatal screening, and termination of pregnancies may be expressed by MPs to their patients, ultimately influencing their patients' decisions (Biesecker & Marteau, 1999; Simpson, Dissanayake, Wickramasinghe, & Jayasekara, 2003; Wool & Dudek, 2013). Research suggests that sharing personal views is expected by patients especially when the patients explicitly ask a doctor for advice (Geller et al., 1993).

Genetics and genetic counselling in society

Patients access medical information from the online search platforms before consulting a specialist (Case et al., 2004). Typically, patients search online for any information that could explain their symptoms, answer questions relating to their health, and provide management solutions. (Brett et al., 2012; Burke, Khoury, Stewart, & Zimmern, 2006; Case et al., 2004; Simpson et al., 2003). The online information in most of the cases is misleading and leaves patients confused which may result in patients ignoring the benefits of genetic testing. Alternatively, this information could

increase their desire to opt for genetic assessments and other predictive tests (Brett et al., 2012; Case et al., 2004; Macleod et al., 2002).

Direct-to-Consumer (DTC) genetic testing platforms have been used by people who are eager to request genetic investigations. (Koeller et al., 2017). These genetic testing platforms provide information about their genetic make-up which is helpful for patient to make decisions regarding their health, future children, and response to certain medications (Koeller et al., 2017). One commonly used DTC platform is known as 23andMe and this genetic testing platform includes screening of common cancer predisposition variants such as *BRCA1* and *BRCA2* genes in the Ashkenazi Jewish population (Majumder, Guerrini, & McGuire, 2021). The accuracy and applicability of these DTC testing platforms results is highly debated among the medical genetic community with many MPs feeling uncomfortable using these results for clinical management (Kim et al., 2022; Majumder et al., 2021). Many people who had DTC genetic testing done did not share their test results with their healthcare providers, or even a genetic counsellor, as they felt that the result was not relevant or important enough to share during their medical consultations (Kim et al., 2022; Majumder et al., 2021; van der Wouden et al., 2016). 26 million purchases were made using DTC testing platforms by 2019 and it is estimated that by 2021 DTC testing would exceed 100 million purchases (Majumder et al., 2021).

Healthcare is shifting more towards personalised medicine, which means that genetic testing, including DTC genetic testing platforms, has steadily become incorporated into standard medical care (Kim et al., 2022; Koeller et al., 2017; Pasquier et al., 2022; Pearce et al., 2019; Pritchard, Goodman, & Nadauld, 2022; Simarro, 2022). The increased demand for genetic tests warranted many health professionals, with and without formal genetics training, to request genetic testing for their patients (Donohue et al., 2021). Despite the great enthusiasm of health professionals attempting to incorporate genetics into standard medical care, the casual requests for genetic testing sets up dangerous outcomes such as test misinterpretation, unwarranted information, and other significant consequences (Donohue et al., 2021; Druker et al., 2017; Stenehjem et al., 2018).

The clinical utility of genetic testing, including DTC genetic testing platforms is being researched worldwide extensively. DTC testing allows for identification of information which patients find useful, but there remains uncertainty about the medical acceptance and clinical utility of these test results (Cornel, van El, & Borry, 2014). In the last decade research revealed that people and healthcare providers were doubtful about the clinical utility of genetic testing (Cornel et al., 2014; Parikh et al., 2020; Pearce et al., 2019). Recently however, clear utility of genetic testing has become apparent especially in cancer and hormone care management (Pasquier et al., 2022; Pritchard et al., 2022). Clinical utility refers to the ability of an investigative test to inform clinical management options that would either improve or completely prevent adverse health outcomes for an individual with a positive test result (Borle et al., 2022; Grosse & Khoury, 2006; Pritchard et al., 2022). Clinical utility for patients would include how the information of the test would benefit them and their families with regards to diagnostic implications and informed decisions on possible clinical management options in the future (Grosse & Khoury, 2006; Khoury, Jones, & Grosse, 2006; Pritchard et al., 2022).

This lacunae in the application of the genetic testing needs much more intense scientific validation. Presenting physical evidence of the impact of genetic testing would increase the uptake and use of the service in many countries especially in areas with significant financial resource constraints (Cornel et al., 2014; Stenehjem et al., 2018).

A need for genetic counselling services

With the increased demand for genetic testing, it is impossible to expect only one type of specialised MP to take on the interpretation of various genetic tests (Beene-Harris et al., 2007; Donohue et al., 2021; J. Kromberg et al., 2006; Maiese, Keehn, Lyon, Flannery, & Watson, 2019; Patch & Middleton, 2018). Access to genetic testing platforms should not be a limiting factor which determines the relevance of genetic counselling in a clinical setting. Instead, the manner in which genetic information such as the clinical diagnosis of a genetic disease, the associated symptoms, prognosis, and inheritance patterns are communicated need to be tailored to provide the best quality care management for patients (Godino & Skirton, 2012; Macleod et al., 2002; O'Shea et al., 2011; Pasquier et al., 2022; Patch & Middleton, 2018; Skirton et al., 2015). With incorporation of genetics into the medical training curriculum, more healthcare practitioners will be able to appropriately refer, utilise, and provide genetic services (Harding et al., 2019; Pasquier et al., 2022).

Incorrect risk estimation and presentation also poses a lot of problems (Biesecker & Marteau, 1999; Bonke, Stijnen, Tibben, Lindhout, & Clarke, 2005; Rodas-Pérez, Clarke, Powell, & Thorogood, 2015). A bystander effect was observed in instances where doctors simply refer patients with genetic queries to other doctors who do not have knowledge of genetics or understanding of genetic reports (Bonke et al., 2005; Maiese et al., 2019). Some primary care providers of America and the UK felt that despite being educated on the genetic causes of disease; the suitability and availability of genetic tests, benefits of testing, and management options for genetic diseases, they are still uncomfortable in explaining it to their patients (Beitsch & Whitworth, 2014; Samuel et al., 2017). This lack of comprehension ultimately leads to an escalation of medical responsibility being denied and patients not receiving appropriate medical care.

The discovery of novel disease related genes and associated guidelines for different medical conditions make it difficult for a MP to consider incorporation of medical advancements into their practices (Hallowell et al., 2009). Understandably it takes a lot of time and research for medical experts to incorporate novel genetic technologies and tests into standard health care practice. Pharmacogenomics is one such field that is not well understood yet despite its perceived importance for implementation in many healthcare practices (Godino & Skirton, 2012). Adjustment of pharmaceutical therapeutics after investigating a person's genetic profile remains infrequently implemented as the interpretation is not correctly done or understood which limits the effectiveness of personalised medicine (Burke et al., 2006; Godino & Skirton, 2012; Jenkins & Arribas-Ayllon, 2016; Zimmern & Kroese, 2007).

Multiple research groups propose implementation of genetics in clinical organisations through increasing opportunities for genetic education among healthcare providers

(Beitsch & Whitworth, 2014; Chou et al., 2021; Mikat-Stevens, Larson, & Tarini, 2015; Tutika et al., 2023), and encourage stronger collaborations with genetic counsellors (Skirton et al., 2015; Slomp et al., 2022). British doctors consult a genetic counsellor before disclosing genetic information to their patients (Patch & Middleton, 2018). Genetic counsellors are qualified to educate MPs about maladies with potential genetic components (Patch & Middleton, 2018; Rantanen et al., 2008; Skirton et al., 2015). In instances of personalising patient management, genetic counsellors can step in to discuss appropriate testing, inheritance risks and management options with different medical doctors to assist patients who are diagnosed with a genetic disease (O'Shea et al., 2011).

What is the bottom line?

Most recent population data indicates more than 60% of the current worldwide genetic counselling work force, which equates to approximately 5250 genetic counsellors, are stationed in North America (Abacan et al., 2019; Ormond et al., 2018). In comparison to North America, Africa collectively contains less than 50 active certified genetic counsellors (Abacan et al., 2019). With limited genetic counselling posts available in the current medical setting in South Africa, many certified genetic counsellors consider rather setting up a private practice of their own to interact with patients or would apply for a genetic counselling post overseas (Chou et al., 2021; Greenberg, Kromberg, Loggenberg, & Wessels, 2012). In addition to the limited number of active genetic counsellors in South Africa, access to genetic counselling is further restricted by factors including geographical distance, cultural-, economic-, and language-barriers (Delikurt, Williamson, Anastasiadou, & Skirton, 2015; J. G. R. Kromberg, E. B. Sizer, & A. L. Christianson, 2013). Furthermore, limited understanding and implementation of genetics in the medical profession affects the use of genetic counselling services. Despite these restrictions however, MPs in South Africa have actively started to utilise the service of genetic counsellors for their patients in the last decade (J. G. Kromberg, T.-M. Wessels, et al., 2013; Thom & Haw, 2021; Walters et al., 2022). The reason why MPs actively make use of genetic counselling services or find these services valuable has not yet been thoroughly investigated.

Rationale

Genetic counselling is a form of highly specialised care where patients are referred to when primary and secondary level care is not adequate for their health management. MPs who practise in both the state and private healthcare sector can refer patients for genetic counselling. Unlike state-based MPs, who refer patients according to a specific management protocol, private practicing MPs intentionally refer patients for genetic counselling to receive a specific service. By investigating how and why MPs in private practice started to make use of genetic counselling services, a new perspective regarding the value of genetic counselling in the South African medical setting could become apparent. The experienced views of these referring MPs in private practice, regarding the impact of genetic counsellors and genetic counselling, may help implement the commodity of genetic counselling across the country to expand the level of quality care that can be provided to patients in the future. In short, this study attempted to investigate the perception of MPs in private practice, who utilise the

service of genetic counsellors, regarding the use of genetic counselling services in their patient management in South Africa.

Research Question

What is the perception of medical professionals in the Western Cape, who utilise the genetic counselling service, regarding the value of the service in their patient management?

Aim

The aim of this qualitative study was to explore why medical professionals make use of genetic counsellors in South Africa and acknowledge their personal experiences with regards to the genetic counselling service.

Objectives

Based on available literature exploring the impact of genetic counsellors and genetic counselling services worldwide, the following four objectives were considered for further investigation:

- Identify why, when, and how MPs in private practice started to utilise genetic counsellors/counselling services in Cape Town.
- Investigate how MPs, who actively make use of genetic counselling services in Cape Town, explain genetic concepts and the purpose/benefit of genetic counselling to their patients.
- Understand what MPs gain and/or risk losing when referring their patients for genetic counselling.
- Identify possible means by which genetic counselling can be presented to MPs who do not actively make use of genetic counsellors/counselling.

5. Chapter 2: Methodology

5.1. Study design

This research study investigated the perceived value of genetic counselling by MPs working in private practice in Cape Town. A qualitative approach based on the philosophy of phenomenology, where all meaning and value is based on personal experiences, was used (Gill, 2020; Reeves, Albert, Kuper, & Hodges, 2008). Gill describes a phenomenological research approach as an attempt of a researcher to describe and provide a faithful account of an individuals lived experience(s) and the meaning that experience conveys (Gill, 2020). To investigate and appreciate a phenomenon of interest, such as the use of genetic counselling services among MPs in Cape Town, participants with similar interest(s) are recruited to emphasise the rich accounts of the participants over the quantity of data (Gill, 2020). The aim of using a phenomenological approach is to provide understanding about an experience, i.e., to understand the experience of MPs using genetic counselling services.

Compared to a quantitative approach, which would provide insight on the measurable aspects of genetic counselling services in Cape Town, a qualitative approach was preferred for this research project as it allowed for the perspective of MPs regarding genetic counselling services to be explored, interpreted, and described (Neubauer, Witkop, & Varpio, 2019). The use of close-ended questionnaires and surveys were not considered here as these approaches were considered in other research projects which provided specific, though limited, perspectives of participants from their research samples (Düsterwald, 2015; Fountzilas et al., 2022; Pasca et al., 2022; Thom & Haw, 2021).

In-depth one-on-one interviews were considered best in obtaining rich data as it allowed for greater insight pertaining to the practices and experiences of the participants (DeJonckheere & Vaughn, 2019; Gill, 2020; Reeves et al., 2008; Saldaña, 2021). Although in-person interviews were planned, these were not possible as a result of the COVID-19 pandemic and MPs were uncomfortable participating in face-to-face interviews (Arndt et al., 2020). An online approach retained the benefits of one-to-one interviews while allowing the participants to be comfortable to consider any time suitable for them to complete the interview process (Salmons, 2014; Thunberg & Arnell, 2022).

A semi-structured question guide (Appendix A) was used to conduct the interview. The guide was constructed after reviewing many research articles and systematic reviews relating to genetic services including genetic counselling (Booth et al., 2019; Creswell, 2002; DeJonckheere & Vaughn, 2019; Saldaña, 2021). The participants' medical field of expertise was also used to guide the interview to explore genetics relating to their speciality, i.e., a breast surgeon referring patients for genetic counselling on cancer predisposition syndromes. Two systematic review articles encompassing the role of genetic services worldwide were particularly helpful in assisting with the design of this question guide (Chou et al., 2021; Skirton et al., 2015). Open-ended questions were

structured on the four objectives of this study (Appendix A and B) and converted into four broad conversation topics:

- Topic 1 – Utility of genetic counsellors
- Topic 2 – Communicating genetic concepts and purpose of referral
- Topic 3 – Perceived advantages and limitations of genetic counselling
- Topic 4 – Marketing genetic counsellors and genetic counselling services

Three mock interviews were conducted to evaluate the effectiveness of the question guide and interview schedule (Adeoye-Olatunde & Olenik, 2021; Creswell, 2002). The first mock interview was conducted in person with a family member to test the set of questions and assess the likely flow of the interview. The second mock interview was conducted with a work colleague online and was recorded to assess the online interview interface and the question prompts. The final mock interview was conducted online with a genetic counselling intern under the supervision of the research project supervisor and was recorded for review by the research team. Findings of the final mock interview were used to assess the question prompts, depth, type of questions asked, the conversation flow, and the overall length of the interview. Based on the mock interviews, the length of the online interview was approximately 30-45 minutes. After each successive interview with a MP where new prompts of conversation were identified, the question guide was adapted to add these new prompts for use in successive interviews (DeJonckheere & Vaughn, 2019). This allowed for these additional points of interest to be explored with succeeding participants. In collecting and analysing in-depth interview data, a purposive sample size of approximately ten participants is suggested to reach data saturation (Braun & Clarke, 2021; Malterud, Siersma, & Guassora, 2016; Saunders et al., 2018). Data saturation largely depended on the number of MPs interested in participating in the study and was additionally guided by the information or quality of the dialogue obtained from these interviews i.e., multiple participants expressing similar views with regards to one topic (Braun & Clarke, 2021; Lakens, 2022; Malterud et al., 2016; Saunders et al., 2018).

5.2. Research setting

Two parallel healthcare systems, private and state-based, operate in South Africa. Public or state-based healthcare is funded by the government (Mhlanga & Ndhlovu, 2021). Patients are classified according to their income and family size and are required to pay according to these classifications (H0, H1, H2, P, etc.). A patient classified as H0 for example will not be charged for healthcare services whereas an H2 patient would be required to pay a subsidised tariff. In comparison, private healthcare services are externally funded which obliges patients to pay for their medical expenses. The medical bills are usually covered in part or in full by a medical aid insurance scheme to which the patients contribute monthly.

All MPs in South Africa are trained in academic state hospitals where healthcare protocols and referral frameworks are firmly established to provide optimal patient management. In state services MPs refer patients according to standard hospital

protocols to different available medical specialists. During their training, MPs are exposed to several different disciplines, referral pathways and management guidelines. South African MPs are required to complete a medical internship and community service at a state hospital upon completion of their medical degree (Reid, Peacocke, Kornik, & Wolvaardt, 2018). MPs can practise medicine in a private capacity, in state-based only or in both a private and state-based capacity. MPs working in state and private are required to comply with the Western Cape Department of Health's remuneration policy, The Remunerative Work Outside Public Service (RWOPS) programme (Taylor & Kahn, 2014).

Genetic counselling is regarded as highly specialised care and is only available at tertiary care level in the public sector. Patients will only be referred when primary and secondary level care is not adequate for their condition (J. G. Kromberg, E. B. Sizer, & A. L. Christianson, 2013; Maphumulo & Bhengu, 2019; Young, 2016). There are limited genetic counselling services in the state and are furthermore only limited to the large tertiary teaching hospitals. In Cape Town there are three public hospitals that provide genetic counselling services to state-based patients. These hospitals employ genetic counsellors. In private a MP would refer patients directly to a genetic counsellor, who practice in the private or private and state sector, or alternatively provide their patient with the contact information of a genetic counsellor to arrange their own appointment.

The Southern African Society of Human Genetics (SASHG) is a multi-disciplinary non-discriminatory society where students, trainees, medical scientists, genetic counsellors, and other MPs working in the field of human genetics can choose to belong to. The aim of the SASHG is to advance the practice of human genetic and facilitate contact between professionals in the field. The SASHG's website (https://sashg.org/genetic_services/) was accessed to identify active certified genetic counsellors in private practice in Cape Town at the start of this project in 2019. Of the ten SASHG recognised private practising genetic counsellors, six were stationed in Cape Town. Besides these six genetic counsellors, the supervisors of this research project were able to identify two additional certified genetic counsellors stationed in Cape Town who were not listed on the SASHG website.

5.3. Participants

5.3.1. Inclusion / Exclusion criteria

The participants were private practising MPs of any discipline who actively make use of genetic counselling services in Cape Town South Africa. This includes MPs who offer medical services according to the RWOPS programme to both state- and private-based patients. For this research project the term 'medical practitioner' (MP) was used to designate a licensed healthcare practitioner who practises medicine in a private capacity such as a medical doctor, otherwise known as general practitioners (GPs), and medical specialists including oncologists, surgeons, paediatricians, etc. The term

‘medical specialist’ in this dissertation refers to a licensed healthcare worker or medical doctor who has completed advanced clinical training in a particular discipline of medicine i.e., cardiology.

The recruited MPs were required to have referred patients to a private genetic counsellor at least once. MPs who have not referred or made use of genetic counsellors before were excluded. All MPs were required to communicate in Afrikaans or English.

5.3.2. Recruitment

Criterion based sampling or purposeful sampling was used to identify the participants (Creswell, 2002). Recruitment of MPs was done by reaching out to eight genetic counsellors. Genetic counsellors who have a state funded post sometimes receive referrals from MPs in private practice. Being a small group of trained professionals; the supervisors of this research project were able to assist with the recruitment phase as they knew each genetic counsellor on a personal basis and are also members of the SASHG.

Each genetic counsellor was contacted by the researcher via email and/or telephone to ask whether they would be willing to extend invitation letters to MPs who have made use of their services, whether referring regularly or even only once. After confirming their willingness to participate in the recruitment process, they were provided with a PDF copy of the invitation and ethics approval letters. The invitation letters (in Afrikaans and English) included a brief overview of the study, the participation expectations, and informed consent form (Appendix A).

5.4. Research procedure / data collection

MPs were invited to respond to the researcher by telephone or email. Alternatively, the MP could agree for the genetic counsellor to provide the researcher with their contact details. Interested MPs were contacted by the researcher via telephone or email to schedule an interview at a time and date most suitable for them. On the day of the interview the MP was reminded of the interview time and to sign the informed consent form to confirm they were comfortable with the interview being recorded.

The interviews were directed using the semi-structured question guide (Appendix B). All interviews were conducted via the Zoom video-conference platform in either English or Afrikaans according to the preference of the participant. Open-ended questions were used to allow richer data to be collected by the researcher who was able to prompt further discussion on specific topics.

The researcher and research supervisors were all fluent in English and Afrikaans and were able to engage with transcripts and interview recordings in either language. By allowing participants to complete the interview in a language of their choosing an

additional layer of data richness was anticipated as participants used different descriptive words in the different languages to describe aspects of genetic counsellors and genetic counselling services (Baumgartner, 2012; Oxley, Günhan, Kaniamattam, & Damico, 2017; Squires, 2009). This created a sense of inclusivity where different cultural groups are recognised, allowing for different perspectives from participating healthcare professionals (Premji, Kosny, Yanar, & Begum, 2020).

Each interview was audio-recorded using the recording function of the Zoom video-conference application (Archibald, Ambagtsheer, Casey, & Lawless, 2019; Inc., 2016). The audio files were inspected at the conclusion of each interview to ensure sound files were of acceptable quality. All online interviews were, as a backup to the computer recording, audio recorded using the standard recording app available on any Apple iPhone. All recorded data were saved as individual audio files (mp3) and uploaded to a personal password protected cloud drive.

5.5. Data Analysis

The recorded interviews were transcribed verbatim in Afrikaans and English using a combined approach of manual transcription and audio-to-text conversion through the free trial version of the qualitative analysis tool: Descript (https://www.descript.com/terms) ("Descript," 2017; Saldaña, 2021). Additional notes were made on paper (Appendix C) during each interview to help direct the flow of the conversation. This allowed for impressions, non-verbal cues, and other thoughts to be written down for further analysis (Saldaña, 2021). These notes were stored in a physical project folder and later combined with the verbatim transcripts for data analysis.

After every second interview, a pre-coding process was done where the verbatim transcripts were read in conjunction with interview notes to identify new topics for discussion that could be used in subsequent interviews (Braun & Clarke, 2021; Creswell, 2002; Saldaña, 2021). Coding of the transcribed interview data started with an initial read through the transcripts to get an overall sense of the data. The interview that seemed most interesting to the researcher was selected first to help guide the coding process whereby the remaining eleven interviews were coded in no particular order. A manual coding approach was favoured because the interviews were conducted in two different languages (Saldaña, 2021).

Open coding was used to analyse the transcribed interview data (Medelyan, 2020; Saldaña, 2021). This allowed for the identification of concepts which were organised in a systematic manner and provided a platform to condense identified codes into categories and eventual themes (Figure 1) (Braun & Clarke, 2021; Saldaña, 2021; Williams & Moser, 2019). Grouping of distinct concepts in the transcripts were accomplished by using single words, short phrases, or comprehensive quotes from the participants (Table 1). Provisional codes apparent from the Afrikaans and English interviews were identified to be similar to one another. Translation, accuracy and reporting of the interview data were accomplished through the process outlined by

Oxley and colleagues (Oxley et al., 2017). Application of this process in this project is further explained in the section on rigour below.

Open coding also allows for additional notes and pre-codes to be taken into consideration. In subsequent read-throughs of the transcripts, the project research question and objectives were taken into account to identify additional codes using the open coding approach (Saldaña, 2021). This provided more comprehensive and focussed codes which could be used for downstream analysis to ultimately answer the research question.

The sets of coded data were refined, sifted, and overlapped into categories through axial coding (Saldaña, 2021; Williams & Moser, 2019) (Figure 1). This coding approach allows relationships between codes to be identified which reduces the number of codes which are ultimately condensed into categories (Table 1). After establishing categories, the final step was to condense the categories into meaningful expressions which were labelled themes (Figure 1).

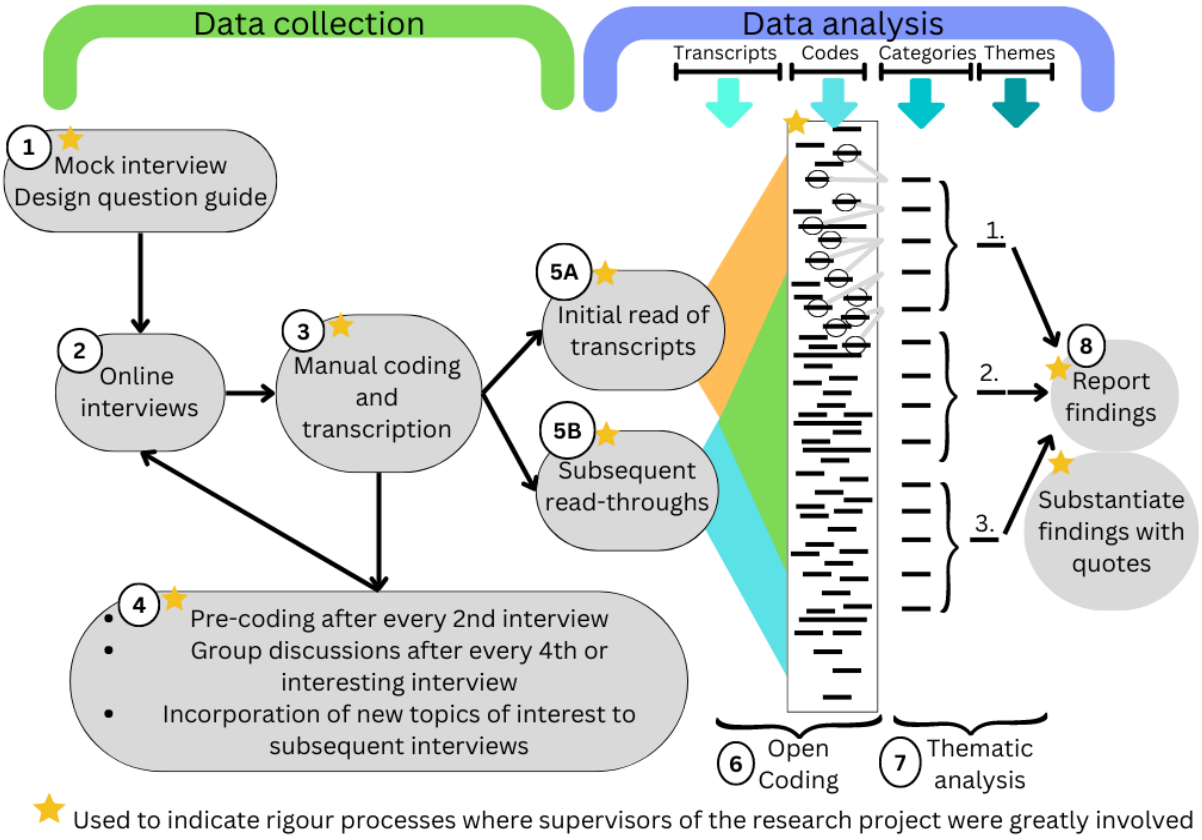

Figure 1: Coding and data analysis flow chart.

An interview guide was created after reviewing multiple articles related to the genetic counselling service. The question guide was tested through mock interviews and optimised. Online interviews were conducted in either Afrikaans of English with medical professionals after which the interviews were transcribed verbatim. The transcripts were pre-coded and discussed with the research team to identify any additional topics of interest for further discussion. After transcribing the interviews, the data was coded through open coding and eventually collapsed into overarching themes. These themes were used to the report the project findings.

Table 1 – Example of coding process with verbatim transcripts		
Interview quotes: Participant 2 - general surgeon	Code	Category
"So the counsellor should have the skill and ability to interpret the histology results and be able to counsel the patient as to what the predicted risk would be of finding a mutation that would be significant for cancer development…If it is something that can be tested for, then I think the counsellors should be able to counsel that family and advise on whether testing is indicated and at what age testing should be done."	• Expectations • Interpretation • Risk analysis • Counsel on test appropriateness and/or selection • Experienced • Age of consent for testing	Knowledge Medical expertise Counselling Skillset (Expertise)
"I mean, what are you going to do? Like, not refer? I mean when the chance of actually finding something contributory to management is there. Yeah, it is a risk, but I think one just has to accept that."	• Benefit of counselling • Benefit of testing outweighing risks • Accept the risks	Impact of service (risk)
"I would generally give them some basic guideline for referral like, a young onset, or a very strong family history (of cancer) and first-degree relatives (brief pause), a particular histological type of the cancer, a combination of different types of cancer. These are the kind of the basic things. And then what the counsellor actually does, I explain to them that you will see a counsellor. It is just a meeting. There's no testing now. You are just meeting them. They are going to ask questions about your personal life, your family life, your family members, and like for breast cancer, they are going to ask things like your age of menarche, breastfeeding, menopause onset, number of children, all these sorts of things. You prime them for that because I mean, we've actually asked them that stuff. And then I tell them if they find that there are enough significant findings in the meeting, they might recommend to you that we should actually move on and to the testing."	• Contextualise genetic risk for patients • Red flags in genetic cancer syndromes • What does a counsellor do? • It is just a meeting • Basic explanation of a genetic counsellor's role • Expectations • Indications for testing	Knowledge Medical expertise Counselling Skillset (Expertise) What is a genetic counsellor?

5.6. Rigour

Guidance and support were regularly requested from the research supervisors. The first few meetings discussed the research aim, design, ethical concerns and -aspects, coding approaches, and data collection aspects of this research project. Additional research meetings were arranged to discuss the introduction/literature review, research methodologies, results, and discussion chapters. The supervisors provided input on the mock interviews through direct and indirect supervision (Figure 1).

After every third or fourth interview, or after an outlier or deviant interview with a participant, individual- and group discussions with the supervisors were arranged. During these group meetings, new topics emerged for succeeding interviews, and these were discussed with the supervisors. At the end of the verbatim transcription process, each of the two supervisors were provided with three random interview transcripts, in both English and Afrikaans for review. Once reviewed, a team research meeting was arranged to interrogate the provisional codes.

The data and findings of this research project were reported in English and thus the Afrikaans interview quotes had to be translated into English. To preserve rigour of the data throughout the process of translation, each Afrikaans quote was translated by both the researcher and a supervisor who were both first language Afrikaans speakers. After translation of the quotes, the remaining (first language English speaker) supervisor would inspect the quotes from both parties to assess whether the content was accurately transcribed (Oxley et al., 2017; Premji et al., 2020). Team meetings were arranged to assess the proposed quotes and whether their translations supported the findings of this research project.

5.7. Ethical considerations

Ethics approval for this research study was obtained from the University of Cape Town Human Resources Ethics Committee (HREC REF: 113/2021). In the ethics application the risks and benefits for the participants were taken into consideration including the health and safety of participants during the COVID pandemic. The principles outlined in the Declaration of Helsinki for all human or animal experimental investigations (Association, 2013) as well as the Protection of Personal Information Act 2013 (POPIA) regarding data protection for health research (De Bruyn, 2014; Staunton et al., 2020) were considered in the design of this research project. Each participant recruited in this research project was required to complete an informed consent form (Appendix A) before conduction of the interview. Participants were informed that the obtained data will remain anonymous, and that no reimbursement of any sort will take place at the conclusion of the interview. Due to the small number of active certified genetic counsellors in Cape Town, the data and other information from this project was carefully inspected to conceal information that might link or reveal the identities of any participants.

6. Chapter 3: Results

6.1. Sample characteristics

The cohort consisted of 12 MPs. All MPs were recruited through five of the eight private genetic counsellors in Cape Town. For a detailed outline of the recruited medical practitioners, please refer to Table 2 below.

Table 2: Sample Characteristics

Participant recruitment	**Number/total**
Genetic counsellors contacted	8
Genetic counsellors willing or agreed to participate	5
Attempts to request genetic counsellors' assistance with recruitment of participants	2
Total number medical practitioners (MPs) invited according to genetic counsellors **(*A single MP may have been contacted by multiple genetic counsellors)**	*41
Interview demographics	
Total number consented and completed interview	12
Afrikaans interviews	7
English interviews	5
Shortest interview	~24 minutes
Longest interview	~53 minutes
Average interview time	~36 minutes
Medical practitioner field of expertise	
Surgeons	6
Breast[1]	2
General[1]	2
Oncology	1
Plastic	1
Foetal medicine specialists	2
Paediatrician	2
Radiologist	1
Physician	1
Total number of MPs practising in both a state and private capacity (RWOPS programme) **(Indicated with superscript [1])**	3
Recruited MPs gender split (M = Male; F = Female)	6M; 6F
Referrals for genetic counselling	
Total estimated number of referrals per month for all participating MPs	52-81 patients
Minimum number of referrals per month per person	1 patient
Maximum estimated number of referrals per month per person	20-25 patients

6.2. Interview findings

The MPs reflected on how they perceive and communicate the benefit or impact of genetic counselling services in South Africa. Many MPs shared their experiences as trainees in state hospitals and compared these to their private practice. Besides the question guide that was used to conduct the online interviews (Appendix A and Appendix B), interesting conversation topics came up which were explored in later interviews. These additional conversation topics include: the use of direct-to-consumer platforms, the limitations and barriers associated with incorporating genetics services in the current South African medical setting, the misunderstood title of the genetic counselling profession, the clinical benefit and cost effectiveness of genetic testing in South Africa, and finally the dependence of MPs on genetic counsellors as a human resource. From the interviews, the following themes and subthemes were recognised: the identity of genetics and genetic counselling services; resource limitations; and the appreciable impact of genetic counselling (Figure 3). Additional illustrative quotations which were not included in the text can be viewed in the supplementary material section (Appendix D).

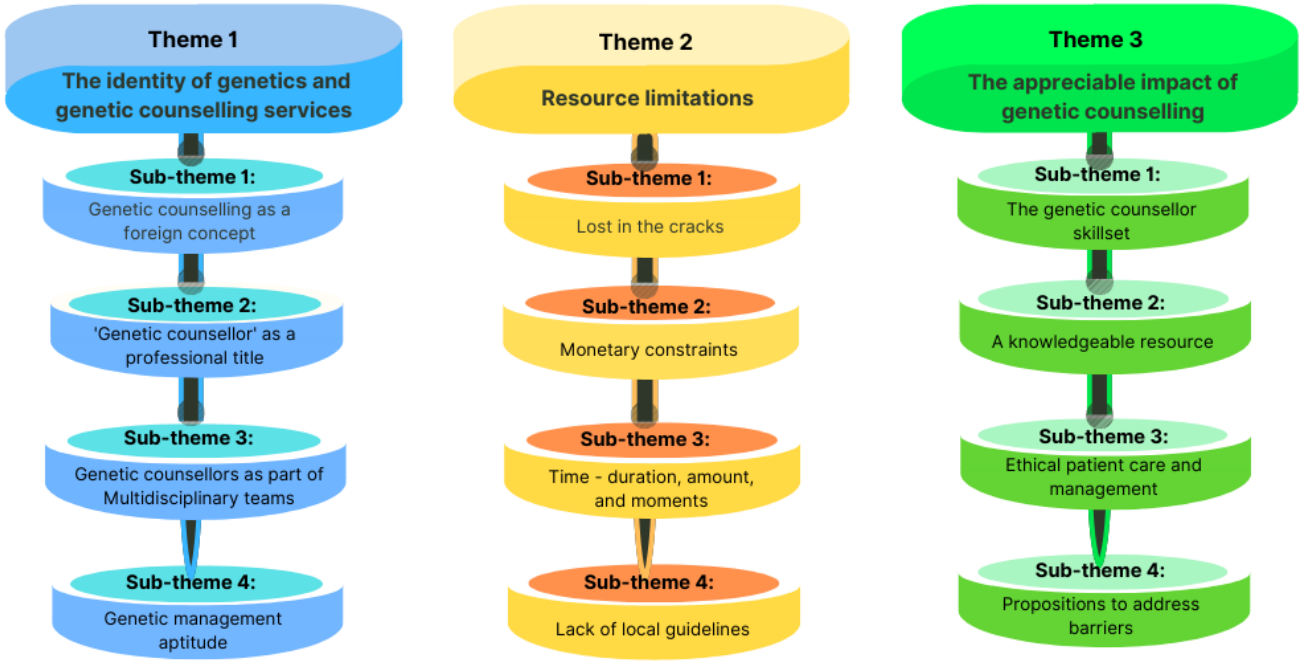

Figure 3: Themes and subthemes:

Indicated here are the three main themes namely the identity of genetics and genetic counselling services, resource limitations, and the appreciable impact of genetic counselling. The main themes are shown with their associated sub-themes which were derived from online interviews with private practicing medical practitioners regarding genetic counselling services in Cape Town.

6.3. Theme 1: The identity of genetics and genetic counselling services

Conversations with MPs largely revolved around the identity of genetic counselling services. Here, identity refers to distinctive properties and assets of the genetic counsellor and the service they provide.

6.3.1. Subtheme 1: Genetic counselling as a foreign concept

Some participants explained that their introduction to genetic counselling came from recommendations or pamphlets sent by pathology labs. Others were exposed to specific clinical environments where genetic investigations were part of the clinical workup of a patient, i.e., patients with a strong family history of cancer. Participants described themselves and others as being unaware or uninformed about genetic counselling services, how they relate to the medical management of their patient and who/what genetic counsellors are and how MPs can contact them to utilise the service. The inability of MPs to describe or explain genetic services and the access thereof to their patients was highlighted by the participants as being the result of a lack of exposure to genetic counselling in medical training.

> *"...if you do not know that genetic counselling as a speciality, or as a discipline exists, you are not going to refer someone to them." – Participant 1, Surgeon*

Some participants claimed to only have been made aware of these services incidentally when talking with their colleagues. The majority recalled first being introduced to genetic counselling services during their post graduate training.

> *"I certainly did not know about it (genetic counselling) in my undergraduate training. We were not exposed to it. So, I did not know that that existed. When, after I qualified, I did not know that there is such a thing as a geneticist." – Participant 1, Surgeon*

Genetics was originally assumed by some participants in their early training to be a separate component of science that is not directly involved with the work of MPs. Some participants described their original assumption of genetics being that of a luxury service, or a discipline that is only dabbled with by super specialist doctors and medical researchers. One participant described thinking of genetics as some foreign “airy-fairy” concept.

> *"In my undergraduate training… I did not know much about it or how it could be applied, it was this airy-fairy fancy (foreign concept) called genetics." – Participant 6, Surgeon*

Participants explained that they are academically trained to understand and apply medical principles to intersect with probable management plans to address a patients' medical concern. They do not necessarily discuss all the intricacies and implications of their diagnoses with patients. One participant stated that their focus during training was on clinical presentation and management and genetics was simply an add-on to provide some additional background to explain the clinical presentation of a disease.

> *"…our curriculum naturally focuses more on the clinical and management aspects rather than the physical genetics thereof…" – Participant 5, Surgeon*

Most participants did not know how genetics can be incorporated into general practice as academics only introduced genetic terminology and facts for examination purposes. They could also not recall whether genetic services, including genetic counselling, were available, or conveyed to them during their undergraduate medical training. The genetics of some conditions were discussed in medical handbooks during training, but these were not detailed or up to date with the recent advances in genetic/genomic medicine. Many genetic conditions that these participants have been exposed to, were also not covered during their academic programme. One participant claimed that the genetic information taught during undergraduate training was very limited with paediatric medicine being their only clear memory of genetics concepts.

> *"And I think our training, you know, our basic training as gynaecologists in terms of genetic conditions and the application thereof in obstetrics is definitely not sufficient." – Participant 12, Foetal medicine specialist*

When patients reach out for specialist care, they expect to receive the best care possible and would not necessarily doubt their doctor's judgement. Many participants expressed that nobody knows all the answers and stated that they would confidently rely on the expertise of their peers to help resolve difficult questions including genetic concepts to improve their patient management. More than one participant stated that it is never too late to refer to genetics.

> *"...we get so many patients walking through our doors who should have had genetic screening a long time ago… with the excuse, 'oh I never thought about it, and it is actually too late to do anything about it'. It is never too late to do the right thing." – Participant 5, Surgeon*

One participant, who has referred patients for genetic counselling services in a state hospital before practising medicine in a private capacity, described some of the difficulties they have faced when referring patients for genetic assessment. They explained that MPs will have no 'working understanding' of what genetic counsellors are if they do not physically see where they are and what service they provide.

> *"…you will have to ask a recent medical graduate if they have ever seen a genetic counsellor in the ward, or in the clinic, or in something at an academic hospital. So, I think that it is very important that these people (genetic counsellors) are fully integrated in academic units. When I was at [name of hospital] …you wrote the patient a letter and sent the patient into the ether of referrals, and then you hope that the patient has seen the genetic counsellor. And months later a genetic report may or may not appear." – Participant 4, Surgeon*

Patients usually consult their family doctor or general practitioner first before being referred to a specialist for further care. If doctors are exposed earlier to the impact and accessibility of genetic counselling, participants believed that this would allow for a more streamlined approach in having genetic assessment done before being referred downstream for more specialised care. Several participants felt that general practitioners, gynaecologists, and family doctors need to be more involved with care coordination as they are trained MPs compared to genetic counsellors who are trained medical scientists.

> *"…if we (MPs) only find out so late in our lives about the context of who this person (genetic counsellor) is, then the general population will know even less about who this person actually is and that such people exist." – Participant 7, Foetal medicine specialist*

According to many participants, the use of genetics in practice is based on the perspective and personality of an individual. They felt that there was no point in giving patients direct access to something they would not understand or be able to use. Some participants explained that they are aware of MPs in similar fields of expertise who would not consider genetics services because they would either not benefit financially themselves, or they would just consider alternative investigations because they do not want to engage with the foreign concept of genetics or genetic counsellor. This includes DTC platforms and random genetic test results. The same sentiment was reflected when talking about other MPs' access to genetic services: 'if they do not know how to use it, then they will not (use it)'.

> *"Well, I think… yeah, it is difficult. Yeah, somebody who does not want to use such a service most likely has a personality like 'yeah, I know enough' and then does everything themselves…" – Participant 6, Surgeon*

Many participants claimed that there are still doctors who question why they as a specialist would consider referring their patient to a genetic counsellor. One participant depicted this phenomenon of being ignorant, which was echoed by many participants, as a "blind spot" in the training of MPs.

"I could see that for many of them it was a bit of a foreign concept, why would I suggest that the patient also talk with a genetic counsellor? Why do not I just quickly do it myself? So, I think it is actually a reasonable ignorance or a blind spot."
– Participant 11, Paediatrician

6.3.2. Subtheme 2: 'Genetic counsellor' as a professional title

An interesting development from these interviews was around the title 'genetic counsellor'. Patients are assumed by some participants to know the difference between psychologist, counsellor, and genetics. Just over half of the participants (six Afrikaans and one English) stated that they would describe the individual comprehensively to their patients rather than just make use of the professional title. They explained that the title (Afrikaans or English) could be misunderstood by patients as being someone who will provide them with either psychological counselling or general advice. Participants generally felt that patients have a negative view of psychology or counselling services as the patients feel that these referrals suggest that there is 'something wrong with their head'.

"When you tell someone, you want them to see a genetic-… especially in English a 'genetic counsellor'. Immediately these people think, but a counsellor you know, I do not want to see that person, understand? Because here in South Africa there is a negative association with psychologists and counsellors and things like that."
– Participant 4, Surgeon

When asked about the reluctance in using the professional title of a genetic counsellor with their patients, the participants explained that the English title 'genetic counsellor' is not descriptive enough and "undermines" (participant 12) the value that the service provides. The Afrikaans professional title '*genetiese raadgewer'*, which directly translates to English as 'genetic advice giver', was considered more descriptive by the participants, but still not comprehensive enough and subject to misinterpretation. Participants revealed that some MPs struggle to recognise the merit of genetic counselling for their patients or themselves as the referring doctors. They explained that MPs have their own impression around the title 'genetic counsellor/counselling' and its associated service.

"Genetic counsellor (Afrikaans term) sounds a little like, how can I describe this? Like, someone who is just going to give you a little bit of their opinion and it is not a comprehensive term in my opinion." – Participant 12, Foetal medicine specialist

6.3.3. Subtheme 3: Genetic counsellors as part of multidisciplinary teams

Another frequently discussed aspect appreciated by many participants was having a genetic counsellor as part of their multidisciplinary team (MDT). When discussing patients in MDT meetings, one participant stated that they experience the work of genetic counsellors as diligent and thorough.

> *"I respect someone else's privilege to work diligently, and I think it is nice to work with someone in a team who works the same way we do." – Participant 5, Surgeon*

Participants who explained that they were not comfortable making decisions on genetic testing alone. They believe that the presence of a genetic counsellor in MDT meetings allows for greater discussion between attending MPs regarding clinical management options and genetic testing options.

> *"So obviously I would turn to them (other MPs), but it would be really nice if I'm not the one deciding that. Do you know what I mean?... not just reading it as a guideline and saying, well this is what they did in America, therefore this is what we should do." – Participant 9, Surgeon*

In addition, these MDT meetings allow for easier patient referrals to be done. Most participants explained that they felt most comfortable referring their patient to a genetic counsellor to explore genetic testing and management options of their genetic diagnoses with the patient. Some suggested that requests for genetic testing should automatically be accompanied by a referral to a genetic counsellor. Others proposed that private practices, labs, and hospitals be provided with a comprehensive list of trained genetic counsellors in an area. This was explained as assisting the requesting MP in making appropriate referrals for their patient to explain the result of the genetic test. One participant felt that genetic tests with clear clinical utility and downstream clinical management implications should be explained in more detail to managing clinicians. Their reasoning being that there could be major health implications for a patient besides providing peace of mind.

> *"If they are going to do a panel test based on multiple genes, they can decide which ones to pick out, which one is more relevant. I mean, I cannot do that. I'm not… you know. I do not have expertise to do those type of things." – Participant 1, Surgeon*

Participants who are greatly involved in MDTs expect a genetic counsellor to attend and participate in MDT meetings. Participants felt that the involvement and input of genetic counsellors in MDT meetings help to build a relationship of trust. These meetings also serve as ideal learning opportunities as patients with rare genetic diagnoses are discussed, or difficult genetic test results are interpreted, i.e.,

microarray and variant of unknown significance (VUS) results. Some participants explained that a genetic counsellor should sometimes take the leading role in suggesting appropriate screening programs based on current global medical guidelines, especially with very rare genetic diagnoses. One participant stated that they can depend on the genetic counsellor to facilitate a management plan for a patient who requires input from multiple different medical specialists. Most participants felt that a genetic counsellor should triage between a patient, their family, and the managing clinician. In other words, assist in coordinating patient care.

> *"The genetic counsellor in my opinion has the primary role of bringing continuity. To ensure the… family is looked after and to give me advice on the level of concern I should have in terms of-… especially if it is a negative (result) or VUS, then it is important for me to have that information. And that (information) is not always in the report. This is once again where the multidisciplinary meeting and those things become very important." – Participant 4, Surgeon*

Many participants shared their personal experience with patients that were diagnosed with some or other genetic condition. They described the management of private patients as very stressful and full of anxiety, especially when discussing decisions for future clinical management and genetic testing. Most participants felt that they could contact a genetic counsellor at any time to help 'bounce ideas' or to consider what management options can be offered to a patient.

> *"And I still remember the first patient that I had, I phoned [the genetic counsellor] and I was like: 'so what am I going to tell her? (Laughs) Am I supposed to tell her she needs a bilateral mastectomy or not? Like, what are the guidelines? What do your guidelines say? So, this is what I've read in the literature.'" – Participant 9, Surgeon*

Shared patient responsibility was another aspect frequently mentioned by many participants. One participant stated that in times when their personal energy reserves were depleted after a difficult day, they were relieved to know that there was someone they could refer their patient to, who would provide them with medical and emotional support. Some participants felt that they could also debrief with a genetic counsellor and contextualise on the next steps to consider especially for difficult patient cases, i.e., a *BRCA1/2* positive patient who is in denial of their test result and does not want consultants or any other medical specialists to assist with further management (participant 9).

> *"So, it helps a lot I think for people to look after their own personal reserves, and it allows for a bit of shared responsibility of the patient too. Another contact point in case there are questions that, you know, in case they (the patients) want to talk about something. So definitely it gives us a great amount of relief on an emotional level, but also with*

regards to time, you know. So I think it is really something which is very supportive for us in our practice." – Participant 12, Foetal medicine specialist

6.3.4. Subtheme 4: Genetic management aptitude

Medical practitioners who are personally interested in the incorporation of genetics in medicine, find it easier to consider genetic testing options. Most participants felt that they would ask a genetic counsellor for help if they do not understand certain concepts or if they require assistance with requesting genetic testing. Most participants explained that they do not want to be the sole decision maker when considering a genetic test for their patient. The administration associated with requesting a genetic test is another facet with which participants were not comfortable with.

> *"So, I would never test the patient on my own. I would always send the patient to a genetic counsellor to decide whether this patient warrants testing or not. I would not send a test off myself" – Participant 1, Surgeon*

When exploring genetic concepts and testing options with the participants, some shared their experience with patients who have asked about DTC genetic testing platforms. One participant explained having done DTC genetic testing on themselves to gain perspective in discussing these options with their own patients in the future. The outcome of this experience was extremely traumatic for the participant as they did not receive any counselling prior to or after the test results and were met with confusing results and possible genetic predispositions that "sits in the back of their head" every day.

> *"Um, yeah… so I have to say after that experience, it was… it is basically, you know, you throw out your net and you wonder what comes in. And if you get some, you catch something you were not expecting. It can be quite a shock." – Participant 3, Physician*

From this experience they would never consider broad panel testing for their patients, except if they know what they are testing for. They also strongly advocate the importance of receiving genetic counselling before and after genetic testing. Another participant claimed they were horrified by associates at a congress testing positive for a *BRCA1/2* variant through free DTC genetic testing platforms and not being offered proper counselling, screening, or clinical follow-ups. Half of the participants felt that DTC genetic testing platforms cause more harm than good with participants comparing it to a minefield or Pandora's Box. Most participants felt that the information obtained from these tests are not informative or reliable at all and only cause unnecessary anxiety. None of the recruited foetal medicine specialists have encountered patients who have considered this route of genetic investigation and none of the participants

felt comfortable with this broad type of genetic testing. These aspects lacking from DTC platforms were thought by the participants to be a feature of a genetic counsellor's everyday toolkit.

> *"…if looking at the genetics of a condition is part of the diagnosis, investigation, or treatment, then your genetic counselling has to be part of that." – Participant 3, Physician*

More than half of the participants felt hesitant to communicate genetic information as they felt ill-equipped with only "half-information" (Participant 5). Some participants assume that their patients have some basic understanding of genetics and introduce genetic investigations using broad terms such as 'genes' or 'chromosomes'. Select MPs would make use of popular media icons to explain genetic inheritance, such as Angelina Jolie and *BRCA1/2* genes associated with Hereditary Breast and Ovarian Cancer (HBOC) syndrome.

> *"…I think you often use fairly basic or crude examples in the popular media… It is quite easy to explain to most people, you know, would know about BRCA[1/2] because it is had quite a lot of press in the last couple of years." – Participant 2, Surgeon*

Only a small number of participants felt comfortable discussing genetic risks associated with certain genetic conditions, i.e., risks of developing cancer and Mendelian inheritance patterns. Three participants explained that they are not trained in communicating difficult diagnoses with patients and their current perception of empathy was only learned through lived experiences at different clinics. The participants find it extremely valuable to send their patients to a genetic counsellor, not only to discuss their genetic diagnosis, but to also provide a genetics specialist opinion about their condition. Another participant stated that their first point of call is always the genetic counsellor when they can visibly see that a patient is struggling to cope with their diagnosis.

> *"I mean, we did not get taught empathy as medical students… we had to learn the pathology of all the things, but not necessarily how to convey that information to patients." – Participant 1, Surgeon*

Participants believed that patients get referred from specialist to specialist who cannot provide genetic services which ultimately delay the care and attention patients require. Additionally, a problem of setting patients on a predetermined path is created by referring patients to multiple different specialists for clinical review and generates decisions that patients might not necessarily want to make. For example, offering a patient the option of a prophylactic bilateral mastectomy or offering a termination of pregnancy for a precious pregnancy which screens high risk for Down syndrome.

> *"…you find the patients have very different needs… she wanted to be in control and not let us guide her as such… but obviously, that will still be her choice." – Participant 9, Surgeon*

If genetic test results suggest changes to current clinical management for a patient, both the family doctor and MP involved with the case should be informed, allowing for a team-based approach to management options to be considered going forward. This endorsement creates a sense of interconnectedness between the patient, the genetic counsellor, and the medical team which some participants referred to as holistic management.

> *"The benefit is just, it is holistic patient handling and I mean you actually prevent a lot of drama from happening later on… it is just correct holistic management from the get-go." – Participant 8, Radiologist*

6.4. Theme 2: Resource limitations

Besides the ambiguity regarding the identity of genetics and genetic counselling services, participants expanded on other barriers which play a major role in limiting referrals to genetic services in South African healthcare. Four subthemes about restricted resources were discussed.

6.4.1. Subtheme 1: Lost in the cracks

Many of the participants who worked in state hospital settings especially during their medical training, felt that genetic counselling services as a human resource were extremely scarce. This discrepancy further highlights the constraints of various resources available in healthcare.

> *"…it (genetic counselling) should be promoted because, you know… we have not got enough of that service available to patients at the moment." – Participant 2, Surgeon*

If there are no genetic counselling or associated services available in an area, participants believe that local MPs will not be aware of it and will not refer their patients for genetic counselling. Some participants felt that once a MP has contacted a genetic counsellor, referrals will be easy since genetic counselling consultations can be done either in person or remotely.

> *"...we were limited in what we were allowed to refer. Whereas in private obviously I can refer more broadly as I think is necessary and then rather let the patients and [the genetic counsellor] decide whether or not it is needed." – Participant 9, Surgeon*

Participants believe that patients get lost in the referral system due to the limited number of genetic counsellors currently working in South Africa. The workload is assumed to be high with a greater chance for patient referrals to 'slip through the cracks'. Some participants were convinced that patients were less likely to miss their appointments or get lost in the referral system if they are handed over in person to a genetic counsellor.

> *"I think even if you did refer patients somewhere they got lost in the cracks because… you did not have that daily contact in the hallways and say 'oh, have you seen this patient?' You know, there are sometimes weeks that go by where you do not necessarily know if they (the patient) have been seen." – Participant 5, Surgeon*

Considering that many systems in South Africa only have remote access to a genetic counsellor, participants felt that the chances of patients getting lost in the referral system are that much greater.

> *"So, I have had many of my BRCA[1/2] patients tell me that they struggled for almost six months to have someone (genetic counsellor) see them." – Participant 4, Surgeon*

With specific reference to genetic investigations, participants believe that a trained professional is required to identify at-risk patients who would benefit most from a genetic test and downstream clinical management. Some participants consider this a cost-effective strategy where at-risk patients are more accurately identified and offered appropriate genetic testing options. In addition, it highlights the lack of experts currently available who are trained and able to identify these at-risk patients. One participant stated that resource constraints including finances and human resources could mean that genetic screening will only be considered as an opportunistic tool rather than a standard clinical tool that improves standard medical management.

> *"…it is difficult in resource limited settings. I mean, you know, it is actually only a very small number of patients that have access to it (screening options) …" – Participant 2, Surgeon*

6.4.2. Subtheme 2: Monetary constraints

A resource constraint voiced by all participants was the financial impact of genetic testing on patients. Insurance companies do not cover every aspect of health care let alone genetic testing. Many patients in South Africa do not have funds available to even accommodate transport to a hospital. Participants believed that this lack of funds is one of the main reasons why patients do not attend their genetic counselling appointments.

"...I think people, uhm, you know, you need to acknowledge that the patients are suffering financially." – Participant 12, Foetal medicine specialist

Genetic investigation costs are covered sometimes by the government or public health clinics e.g., prenatal investigations and hereditary breast and ovarian cancers. However, there are other genetic testing options which are not covered by the government and are very expensive. Some participants were not entirely sure about the costs associated with genetic testing and many were hesitant to consider broad genetic testing panels as incidental findings could have great implications for patients' life insurance policies and medical aid.

"And I think that is also not something that an insurance broker will tell anyone… Ultimately it is to know how much money you are willing to give out to get an answer you want to know about." – Participant 11, Paediatrician

The financial burden on patients caused participants to refer their patients to only a limited number of specialists to help contain their expenses. Most participants felt that the benefits a patient receives in seeing a genetic counsellor outweighs the consultation fees. Participants felt that if patients are provided with appropriate information and understand the reasoning behind the offer of genetic testing, the patients themselves will be able to justify whether they would want to proceed with genetic testing or not.

"The only disadvantage is probably the additional cost as a patient will have to pay an additional consultation fee." – Participant 4, Surgeon

In cases where patient finances were really constrained, participants explained that they were willing to triage with a genetic counsellor and provide genetic counselling themselves to the benefit of the patient.

"So sometimes it (funds) is a bit of a hampering. So, in that case, I will make contact with the genetic counsellor myself and ask if there is anything else that I should say and usually we help one another in that manner so that money does not stand in the way of a patient getting a little bit more information that is essential for them." – Participant 7, Foetal medicine specialist

6.4.3. Subtheme 3: Time - duration, amount, and moments

Participants felt they cannot spend an hour per patient to explain the intricacies of a genetic diagnosis. This would dramatically limit the number of patients a doctor can attend to in one day and create tremendous pressure:

"So, I do not usually have enough time to give them (patients) a lot of information…" – Participant 9, Surgeon

The urgency for MPs to manage as many patients as possible in one day makes them more attuned to the amount of time spent per patient per day rather than spending that extra moment with the patient as they process the reality of their situation. Participants explained that sometimes when they tell the patient about their diagnosis, the patient "closes up" (participant 12) completely and does not want to engage with them anymore.

"We do not always have the opportunity visit after visit to specifically focus on that (processing of information and associated emotions of the patient) …it is not just an issue of timing of the discussion, but also time. And I think the genetic counsellor often has longer consultations and time to give attention to those aspects as well." – Participant 12, Foetal medicine specialist

Participants explained that they appreciate being in the moment with their patient, but do not always find it easy to engage with difficult emotions and psychosocial circumstances. The patients are overwhelmed with information and emotions and cannot process anything at that moment in time. Many of the participants also emphasised, they cannot do everything. It becomes a question of how their medical expertise would be utilised best, i.e., a specialist surgeon who can operate on extremely rare and malignant cancers or consult with a patient for an hour about the genetic implications of their diagnosis.

"…then we do the test. Then we need to have post-test counselling or debriefing. Now I am more than comfortable to do all those things, but it takes time." – Participant 4, Surgeon

In referring patients to a genetic counsellor, participants believe they save time to attend to other patients. The opportunity to consult a genetic counsellor not only allows questions to be asked but also allows the patient to address their concerns and fears with another healthcare professional with whom they can build rapport.

"...it actually gives us time to focus more on our job description..." – Participant 5, Surgeon

Participants felt that referring to a genetic counsellor also serves as a follow up for patients to process complicated information that was originally discussed with their managing clinician and to voice any additional concerns that were not addressed previously. They also appreciate knowing that their patient will have the opportunity to consult someone for a longer period than they are able to provide themselves.

"I think for me the most important is the time it takes to really explain and lay out the information in detail... let's say the reflective handover of information which requires a lot of time." – Participant 11, Paediatrician

6.4.4. Subtheme 4: Lack of local guidelines

In many of these interviews the need for a guideline or systematic approach was raised to identify patients who would benefit most from genetic testing. Participants explained that they currently make use of a combination of American and European clinical practice guidelines for management of their patients. They felt that they constantly need to deviate from these international guideline's suggestions due to a variety of resource constraints in South Africa.

"So, I think yes, we do need to have a separate set of guidelines. What works for Europe and America does not necessarily work for us. And some of these genetic things, it is very difficult… our resources do not necessarily give us the opportunity to do it the way it should be done" – Participant 1, Surgeon

The participants offered genetic testing to their patients to help with a diagnosis. Most believed that if they know what genetic condition(s) they can test for, in other words the type of genetic tests that can be requested in the South African setting, they would be better equipped to sift out patients who would be ideal candidates for testing. Their biggest concern was not offering genetic testing to the patient(s) who would benefit most from a genetic diagnosis and downstream clinical management. This includes early medical interventions such as regular screening for early onset cancers or possible termination of pregnancy for life limiting conditions such as trisomy 18.

"I think you as the breast cancer survivor needs genetic counselling and testing, and then having done that, then we look at your daughter based on that plus your husband, you know. So yeah, I have a lot of different uses that would not necessarily be listed, but somehow the genetic counsellor makes it work for me." – Participant 9, Surgeon

A genetic counsellor was described as guiding clinicians to consider the best clinical management options for their patients based on their research and clinical expertise on what available resources are present in the current healthcare system. Some participants felt that the integration of genetics into South African healthcare could lay the foundation of applicable clinical guidelines befitting other developing countries as well. One participant who is directly involved with the training of undergraduate medical students mentioned that they cannot understand how medical students need to be educated and trained in 1st world clinical practice standards when most of those resources are not even available in a 3rd world country.

> *"I think we need to, in everything we do in South Africa, we need to actually have our own set of guidelines… We actually need to reinvent the wheel for low- and middle-income countries. You know, for everything we do there actually needs to be a different set of guidelines for low- and middle-income countries."*
> *– Participant 1, Surgeon*

Many participants voiced the hope for the design of genetic service guidelines which MPs can use to refer patients for genetic counselling or request specific genetic tests since many felt that they need to constantly deviate from international guidelines due to resource limitations in South Africa. Participants thought that by involving more genetic and medical experts in the design of these guidelines, a resource-focussed strategy of incorporating genetic services in our country would become possible. Some participants suggested the design of something like a standard 'watch-and-worry tick list' which provides a set of relevant clinical signs or red flags that could be used by MPs to better refer patients for genetic counselling.

6.5. Theme 3: The appreciable impact of genetic counselling

Participants recognised several qualities that they value most when referring their patients for genetic counselling. Most commented that they cannot identify any obvious risk when referring their patients to a genetic counsellor. They believe that the benefits of referring patients outweigh any risk that may become apparent.

6.5.1. Subtheme 1: The genetic counsellor skillset

The participants described how they explain the role of a genetic counsellor to their patients to "plant a seed" (participant 9) for the genetics consultation. When referring patients for genetic counselling, the participants outlined several expectations of a genetic counsellor. Participants expect their patient to have a detailed discussion with a professional with regards to their medical enquiry, i.e., clinical investigation with associated symptoms or characteristics, family history suggestive of an underlying genetic condition, or concerning prenatal scan findings. They consider genetic counsellors to be experts in gathering clinically relevant information including detailed family histories, significant personal/psychosocial circumstances, and other information which may inform further clinical management or further medical investigation.

> *"I think it (genetic counselling) is like an essential adjunct, how can I say… essential additional service for any practice especially those who deal with malignant or genetic conditions. It is someone that can help with patient empowering and decision making and also help me to make better decisions in terms of further management decisions."*
> *– Participant 6, Surgeon*

Genetic counsellors are expected by participants to identify at-risk patients who will benefit most from genetic testing, i.e., patients who are classified as high risk to have inherited a genetic mutation/variant that predisposes them to develop early onset cancers such as those seen in HBOC syndrome. Genetic counsellors are also expected to discuss the concepts and implications of risk with patients, as well as explain the pros and cons associated with various genetic testing options. Participants also expect genetic counsellors to build a relationship with their patient to help them comprehend the impact of their medical diagnosis for themselves and their family. Finally, genetic counsellors are expected to provide MPs with relevant information pertaining to care and management for the patient, i.e., screening options that should be considered to monitor the well-being of their patient. One participant was able to summarise the skills of a genetic counsellor very clearly:

> *"…number one is that the counsellor is a source of information in terms of genetics: what is genetics, what is a mutation, and all these technical information for the patients. Number two is the genetic counsellor is a source of sifting. So, in other words… these people (genetic counsellors) are in the position to triage with people (medical practitioners). They are able to say, 'these are risk patterns, and these people would require testing yes or no'. Number three is coordination. Coordination between patients, the genetic counsellor's information and the oncology team, and coordination with the family of the patient. I think it is an important role and I think I will say the fourth role… is the important role of being part of the multidisciplinary team." – Participant 4, Surgeon*

In addition to the skills outlined above, many participants discussed their expectations with regards to delivering genetic test results. Once the results of a genetic test are received, a genetic counsellor can deliver and clarify the results to the patient including the possible clinical manifestations and inheritance risks for other family members. In instances where a VUS is identified; the implications of the VUS will be clarified to both the patient and the managing clinician. A subset of participants prefers a genetic counsellor to discuss genetic information and test results with their patients as some of the information is too difficult even for them to comprehend.

> *"Many times, you get these variants of unknown… you know, significance. I think a counsellor is probably more adept at giving that information or interpreting that information for a patient than we are as clinicians." – Participant 2, Surgeon*

Some of the participants who are surgical specialists explained that they would feel more confident about how a VUS could impact surgical decisions they can offer to their patient after discussion with a genetic counsellor. Genetic counsellors are also

expected to “keep a finger on the pulse” (participant 2) with regards to the resolution of VUS’s and inform affected patients about the implications thereof.

> *“So, I mean, I think I’d rather leave that to the experts and take guidance from them as to what the implications of that are. I mean, I fully understand that we do not use VUS’s to make clinical decisions, but at the same time we cannot disregard them completely.” – Participant 2, Surgeon*

Most participants felt that they cannot tailor genetic information as effectively as a genetic counsellor. One participant (participant 5) used the metaphor “let the cobbler stick to his last” to explain that experts of genetics or clinical management should provide the best service for their patient in their disciplines. Many participants stated that everyone has their role to play. All participants felt content with handing over patients to a professional who can confidently and accurately explain genetic information through a trained viewpoint.

> *“Yes, and to help interpret in [an] understandable language for the patients and their families. So, yes. I am part of the conversation, but I am more part in directing the conversation with the initial information the mutual sharing of information is a very, very big role of a genetic counsellor I think.” – Participant 6, Surgeon*

Most participants felt that a genetic counsellor should be the person to offer cascade testing to at-risk family members of an affected patient, and to assist in arranging continuous care management, i.e., screening for development of HBOC syndrome associated cancers.

> *“Yeah, I think the genetic counsellor is very good at, you know, if a certain gene has been picked up… that predisposes you also for cancer x-y-z. The genetic counsellor will propose annual colonoscopies and I mean I think we follow it (propositions)… we definitely allow the genetic counsellor to take the lead in terms of those types of suggestions.” – Participant 8, Surgeon*

There was a consensus that genetic counsellors have a scarce skill set which has a great impact on patient and family care management. As one participant explained:

> *“I think a person should go to someone who treats the same type of condition regularly, and then a person will get insight, a judgingly fair opinion.” – Participant 4, Surgeon*

6.5.2. Subtheme 2: A knowledgeable resource

Most participants resort to communicating with their colleagues, rereading their old handbooks, or exploring online resource platforms including medical forums, research articles, and international guidelines to find out more about clinical and genetic information relevant for their patient management. Some claimed that a genetic counsellor is an extended resource of information that can provide with more up-to-date information with regards to advances in genetic technologies including clinical management. One participant stated that “we (medical practitioners/specialists) have a responsibility to patients to never stop learning and provide the best medical care we can” (participant 2).

> *"They can look at the specific syndromes or whatever there is, and they can decide what to do… I do not have the expertise to do those types of things." – Participant 1, Surgeon*

The participants believe that genetic counsellors are exposed to many interesting clinical cases which can be of educational value to many MPs. They view genetic counsellors as a knowledgeable resource of medical and genetic information.

> *“So, most of that information I get from the counsellors… and many of the counsellors know what the condition is, like [name of genetic counsellor] knows what test can give you a good idea of what is going on… but I like to lean on what the genetic counsellor has to say.” – Participant 6, Surgeon*

Many participants appreciated the feedback obtained from genetic counsellors. The majority felt that they always know the outcome when their patient has seen a genetic counsellor. Most participants described the summaries and feedback they receive from genetic counsellors as very comprehensive, and they would solely use these summaries to decide regarding clinical management for their patient.

> *“But I think the important thing is I think especially from [name of genetic counsellor] I get a very detailed comprehensive report. More detail than just that about the gene.” – Participant 4, Surgeon*

Participants reflected that they want rational answers when requesting genetic tests for their patients. By being provided with comprehensive feedback or written reports, (even if the genetic test result returns as negative) they feel that it clarifies unknown patient information and answers many unresolved questions. Some participants felt that the feedback from genetic counsellors assists in resolving mismanagement by clarifying ambiguity and provides patients with a sense of closure in knowing the outcome of the genetic investigation. It provided some participants with a clear guide of clinical information that they needed to share with their patient during their follow up consultations. Genetic consultations that happened prior to clinical follow-ups with the participants, as the managing clinicians, were also described as providing them a

platform to simply check in with certain medical/clinical information. This saved a lot of time for participants during their consultations as they did not have to re-explain many concepts that were already explained by the genetic counsellor.

> *"And I think if someone was already seen by a genetic counsellor, which happens every now and again, I find that they (the patients) know exactly what we are talking about. You do not have to explain concepts or tests, you can just refer to them. For example, NIPT... you will immediately see that they understand what it is." – Participant 12, Foetal medicine specialist*

6.5.3. Subtheme 3: Ethical patient care and management

Different participants commented on their appreciation of how genetic counsellors incorporate the pillars of medical ethics when consulting their patients. In respecting patient autonomy, genetic counsellors were described as empowering patients with relevant information and options that the patient and their family could consider going forward, i.e., cancer genetic testing including possible cascade testing for at-risk family members.

> *"So ultimately it is almost more of a financial decision as to how much money you are willing to give out to get that extra peace of mind." – Participant 11, Paediatrician*

Some participants recognise that they have their own ideas on what would be the 'correct' decision with regards to genetic testing and management plans. Here many participants commented on how they know of many MPs who make decisions on behalf of their patient. Many participants explained that some doctors prefer to work in their "comfort zone" (participant 12) and work like a person who follows a set "recipe" (participant 12) and likes to do what is correct according to their standards.

> *I think there is a big gap when it comes to general gynaecologists and obstetricians. I feel they need to be bolder to contact genetic counsellors directly. They just follow a general recipe. I think there is a big gap in the market where patients should be informed earlier about their options to make a decision themselves about the route that they want to take, the money they are willing to set aside for certain testing, and how important it will be for them. – Participant 12, Foetal medicine specialist*

Many participants believe that patients have the right to decide their own management and that the time for doctors as the "autocratic" (participant 6) decision maker has long passed and that this manner of convenience is the incorrect manner of empowering patients. They felt that the approach of a genetic counsellor in allowing patients to be more involved in the decision-making process is a more empowering and beneficial

manner of inspiring patient autonomy. This approach was explained as also providing great opportunities for a genetic counsellor to build rapport and form part of the patients' medical journey.

> *"So, I think the time is gone where you as a doctor say: 'this is what is wrong with you, this is the treatment that you are going to get…' that does not work. I find that the outcomes of patients in the long run are much better if they feel that they are part of their own management… it is subjective, but it feels to me that patients just get through their journey much easier if they know what it is going on." – Participant 6, Surgeon*

This manner of assisting patients to comprehend what they do not want with regards to genetic investigations was regarded as an important and uncomfortable process which some participants were not comfortable dealing with alone. Many of the participants describe how genetic counsellors tailor their sessions for patients and their families by explaining the associated limitations, applicability, and possible outcomes of genetic testing. This custom-made information session involves the patient and their family directly into their own care management and allows for family alignment and shared informed decision making to occur. One participant stated that it always feels like a failure to them if a family unit is "not on the same page" with regards to the management plan and regarding the input of genetic counsellor as key to help resolve this conflict of interest:

> *For me the expectation is that they (the genetic counsellor) actually help the patient to make a decision that fits with them… and it is definitely not always possible, but if it helps to get the patient and her husband on the same page, that they make the right decision that is not just important for them as an individual, but for them as a couple… It always feels like a failure to me if you can clearly see that these two people (the couple) are not on the same page. – Participant 11, Paediatrician*

Participants reflected on what they observe in referring their patients for genetic counselling. Most of the participants commented on a noticeable difference in their patients' demeanour after they have consulted with a genetic counsellor. Some labelled genetic consultations as a place where patients are provided with peace of mind and where their newfound reality is contextualised with information which incorporates possible options for further medical management and genetic investigations where appropriate. One participant described the role of a genetic counsellor for patients as being that of a "medical translator" (participant 6). Patients are free to ask questions about their diagnosis and process information at their own time whilst becoming more aware of how they can be directly involved with their own management plan. Most participants felt content in supporting patients with informed decisions after they have consulted a genetic counsellor.

"You (genetic counsellors) are like a preparer. A bearer of bad news, but also someone with information and a plan, and also, a handshake, or a hug, you know. It is a consoler and also a counsellor. Almost a consoling counsellor." – Participant 7, Foetal medicine specialist

6.5.4. Subtheme 4: Propositions to address barriers

During the interviews participants were prompted to propose solutions which could address some of the barriers they have described to incorporating genetic counselling services into South African healthcare. Most participants felt that engaging with the medical community through physical attendance of genetic counsellors at clinics, MDT meetings, and specialty forums, more doctors would be aware of the availability of the service. This increases the possibility of these MPs reaching out to genetic counsellors for advice and open the possibility of potential patient referrals in their practices in the future.

"…I think all of us want to practise medicine as best we can. I think when people can see from examples, there is actually an advantage to handling someone correctly from the start… I think the proof of the pudding is in the eating. To see the practical impact on a patient, that is maybe the route where it is important." – Participant 5, Surgeon

Participants felt that early exposure to genetic services will make MPs more willing to make use of it, similarly to the way MPs readily refer patients for physiotherapy or occupational therapy. The impact of genetic counselling was described as best understood when participants had the opportunity to sit in and observe a clinical case with a genetic counsellor. One participant stated that this kind of exposure to genetic counselling made a "lasting impression" (participant 7). Participants felt that exposure provided the best knowledge with many claiming that applications in medicine are best learned through lived experiences.

"And during our postgraduate training we also sat in with genetic counsellors, so that was very valuable. Not only did we see 'oh this is the person', but we could also listen how they do it. And it was, it kind of made a lasting impression on me." – Participant 7, Foetal medicine specialist

Participants claimed that by engaging with a patient alongside a genetic counsellor, they could experience first-hand what the service could offer their patients in the future. They also appreciated the impact of heritability on family management. Most participants believed that undergraduate medical students need to be exposed earlier to the service to appreciate what a genetic counsellor 'brings to the table'.

"You know the moment you are exposed to something, then you realise what you are missing out on or what is available… If you do

> *not know, then you are blissfully unaware that that component is there." – Participant 12, Foetal medicine specialist*

More than half of the participants felt that genetic counselling needs to be more incorporated into the undergraduate medical academic curriculum. Most felt that they were introduced far too late in their specialist training to appreciate the influence and application of genetic services into their medical practices. By educating medical students about the limitations and extent of genetic services in the country, they will be able to understand how and when to request genetic testing for their patients.

> *"I think to bring it into undergraduate and postgraduate teaching is important… I think the more you are exposed to the concept, the more likely you are to make use of it." – Participant 9, Surgeon*

By only providing a clinic a single telephone number to call if a need arises for genetic counselling was thought by some participants to drive disconnection. The lack of physical attendance at clinics was described by some participants as giving the impression that genetic counsellors do not want to engage with the MPs working there. Some participants felt that they do not know if they provide enough information when referring patients to a genetic counsellor. A few participants claimed that it was through the exposure and physical presence of genetic counsellors that they were introduced to the existence of the service in South Africa. Genetic counsellors were requested by participants to take control and introduce themselves to the MPs in the community.

> *"Be there, be available. Do not hide in your office… if there is a breast clinic once a week, then you sit there once a week in the breast clinic and that is how patients are going to be picked up. Nothing like 'page me if you have a patient you want to discuss with me.' That will never happen." – Participant 8, Radiologist*

Many participants felt that the best way for MPs to become more involved with genetic counselling services, was to show them the obvious benefits of the service. Examples of this type of service 'advertisement' include having genetic counsellors present case presentations, educational learning courses, journal clubs, and being part of clinical case discussions during clinical meetings. Here a genetic counsellor can also define the probable pros and cons of genetic services that could be relevant for patient management.

> *"I think also the manner in which the package is sold at the end of the day in the context of the patient, it is not a threat, it is a factual evaluation process with merit. I am taking a long time to say to something very simple which is that I think it (genetic counselling) is underutilised." – Participant 5, Surgeon*

In clearly stating the medical clinical utility of genetic testing, participants felt that MPs will be able to appreciate the impact of genetic investigations and hopefully inspire the use of these services in their own practices. Participants felt that MPs working in the same clinical environment may not always talk to one another about genetic services or take it for granted in a particular healthcare setting. Some participants proposed that they themselves who actively utilise genetic services should advertise the service more readily to the rest of the medical community.

> *"…sometimes the obvious advantages need to be highlighted clearly for someone (laughs) and hope they see it." – Participant 6, Surgeon*

6.6. Conclusions

Participants from this sample felt that they initially did not know what genetic counsellor or genetic counselling services are. Some explained how they did not know at the start of their medical career how genetics could be incorporated into their practices. Majority of the participants believed that they were only exposed to genetic counselling services during their post graduate training and shared their experiences of how they recognised the impact of genetic counsellors in MDT meetings. Many participants explained that they do not feel confident sharing their genetic understanding with patients which ultimately delays the care they require.

The integration of genetic services into South African healthcare systems according to participants from this sample are impacted by several barriers including a shortage of trained genetic professionals, which include genetic counsellors, financial resource constraints, time, and the absence of local guidelines for patient management. Despite these constraints to integrating genetic services in the country, this sample highlighted qualities of genetic counselling services which they value in their private practices and provided with propositions to address these barriers and help improve the uptake of genetics and genetic counselling services in South Africa.

Participants described what they expect to be the skillset of a genetic counsellor followed by the role that a genetic counsellor plays in their practice both for their patients and for themselves as a MP. They felt that genetic counsellors are patient-focussed medical professionals who honour the pillars of medical ethics.

7. Chapter 4: Discussion

To our knowledge, this research project is one of the first attempts to explore the views of MPs who actively make use of genetic counselling services. It is the first in South Africa and has shed some light on how MPs in private practice, who work with genetic counsellors, view their service. The general impression obtained from these participants regarding the impact of genetic counselling was positive. Participants believed that genetic counselling complements their patient care to such an extent that they felt they would provide an inferior service if they did not have regular input and support from a genetic counsellor. The role of genetic counsellors was extrapolated by participants as being educators and advisors on the clinical utility of current and available genetic investigation options. Furthermore, results indicated that genetic counselling services in Cape Town South Africa are impacted by several constraints ranging from recognising genetic counselling services to limited resources in the current healthcare setting.

Research studies have explored the perceptions of patients, general health care professionals, and trained genetic specialists regarding the integration and impact of genetic services and included groups of MPs who may or may not have been exposed to genetic counselling services. Most of these studies were conducted in developed countries such as the USA and Europe and concluded that MPs do not feel competent with their genetic knowledge to provide genetic services and would prefer the involvement of trained professionals, including genetic counsellors, to provide genetics services (Beitsch & Whitworth, 2014; Hallowell et al., 2009; Harris, Crumb, Crowe, & McKinney, 2020; Means et al., 2020; Pasca et al., 2022; Pestoff, Johansson, Nilsen, & Gunnarsson, 2019; Rosenbaum et al., 2020; Slomp et al., 2022; Wadman, Conway, Garbarini, & Baker, 2022; Wool & Dudek, 2013). In comparison to developed countries, developing countries such as South Africa do not readily have access to equipment, trained professionals, or the human- and financial resources to engage with novel genetic technologies (Abacan et al., 2019) – a viewpoint which was expressed by many participants in this sample. These shortcomings impact the awareness of genetic developments in developing countries and correlates with the limited available research on perspectives of genetic services in countries such as South Africa (Düsterwald, 2015; J. G. Kromberg, E. B. Sizer, et al., 2013; Thom & Haw, 2021; Walters et al., 2022). Supporting the findings of other research groups (Skirton et al., 2015; Thom & Haw, 2021), participants in this research project stated that genetic counselling is an “underutilised service” (participant 6) as there are still very few MPs, according to participants, who refer patients to a genetic counsellor for a consultation as they themselves do not understand what this service is or what this service provides their patients.

Findings from this research add to previous studies which alluded to a direct correlation between a lack of understanding of genetics/genomic medicine and the ultimate use of genetic services (Baars, Henneman, & ten Kate, 2005; Baars, Scherpbier, et al., 2005; Düsterwald, 2015; Harding et al., 2019; Maiese et al., 2019; Medendorp, van den Heuvel, Han, Hillen, & Smets, 2020; Mikat-Stevens et al., 2015; Pearce et al., 2019; Thom & Haw, 2021). A systematic review by Mikat-Stevens and

colleagues captured how the field of genetics develops at an exponential rate, whilst many practising MPs have training on genetic advances that were made more than a decade ago (Mikat-Stevens et al., 2015). The average time for a medical specialist, which includes all participants in this research study, to complete their training is approximately 12 years depending on the medical field of interest. This by extension means that a large portion of the current medical workforce have completed their training before the completion of the Human Genome Project and the beginning of the human genomics era (Papaioannou & Kampourakis, 2020; Pasca et al., 2022; Pasquier et al., 2022; Patch & Middleton, 2018). MPs are in other words required to provide a medical service which they are not familiar with, which risks misinterpretation and ultimately results in mismanagement of patient care. This increased pressure for non-genetically trained MPs to provide genetic services accentuates the gap where genetic counsellors can step in to provide genetic services as an “adjunct” (participant 6) for MPs. The lack of genetic knowledge has been frequently reported to be a barrier to MPs providing adequate medical management and treatment for their patients with potential inherited conditions (Carroll et al., 2021; Düsterwald, 2015; Harding et al., 2019; Papaioannou & Kampourakis, 2020; Parikh et al., 2020; Pasquier et al., 2022; Pearce et al., 2019; Walters et al., 2022). Lack of genetic knowledge being a barrier in using genetic services was echoed by participants as most of them could only recall being exposed to genetic counselling services and its application during their specialist training.

Minimal access to genetic services including genetic testing was described by many participants as being the direct result of a lack of genetics knowledge and -experience in the medical field. Many MPs, including participants from this sample, were not aware or have not yet ‘discovered’ genetic services or -resources in their country or healthcare setting (Kim et al., 2022; Mikat-Stevens et al., 2015; Papaioannou & Kampourakis, 2020; Pearce et al., 2019; Thom & Haw, 2021). Participants explained that they initially did not understand the identity of a genetic counsellor and their role. After experiencing the service first hand by observing a genetic counselling session with a patient, participants believed they could more clearly understand the relevance of services that can be offered in a particular health care system, similar to what was recently described by other research groups (Pasquier et al., 2022; Scherr et al., 2022). Similar to the study by Pasquier and colleagues (Pasquier et al., 2022), participants valued the involvement of a genetic counsellor particularly as part of a multidisciplinary or care management team. Literature suggested the incorporation of genetic counsellors and genetic specialists into MDTs (Skirton et al., 2015; Slomp et al., 2022; Tutika et al., 2023) whereas these participants were adamant that genetic counsellors perform a crucial role in MDTs. Participants expressed how MDT meetings provide an ideal platform for genetic counsellors to share their expertise, educate MPs, and propose up-to-date clinical management guidelines for complicated patient cases who are diagnosed with a genetic disease. These MDT meetings, according to participants, also provide opportunities for MPs to discuss patients with a genetic counsellor where patients could ultimately be referred to a genetic counsellor for a consultation.

With recent efforts to increase awareness and incorporation of genetics into the different spheres of healthcare, the demand for genetic testing and other genetic services are now exceeding the capacity of the available genetic workforce (Attard, Carmany, & Trepanier, 2019; Borle et al., 2022; Cornel et al., 2014; Harding et al., 2019; Stenehjem et al., 2018; Tutika et al., 2023). Genetic conditions can affect any organ or system of the human body, and along with continuous advances in medical research, it comes as no surprise that genetic services and the associated patient related administration are complex and time-consuming (Attard et al., 2019; Tutika et al., 2023). Most participants felt that they did not have the necessary time to consult with their patients about all the intricacies of genetics and to complete the associated administrative work. These interviews captured how participants clearly appreciate the input and time that a genetic counsellor spends with their patients as they can unpack and address all the medical and emotional issues with the patient in a safe and systematic manner. Participants in this sample echoed the findings by Hannig and colleagues where genetic counsellors were thought to have more time to devote to their patients to review medical records, consult, and sift out patients who would require additional clinical workup (Hannig et al., 2014). Supporting the recommendations of other research articles, participants suggested that by employing more genetically trained experts including genetic counsellors into the medical field, the increased demand for genetic testing will be addressed (Attard et al., 2019; Papaioannou & Kampourakis, 2020; Pasquier et al., 2022; Stenehjem et al., 2018).

Misconceptions about the role of genetic counselling is apparent among health professionals in South Africa (Thom & Haw, 2021) and other countries worldwide (Cordier et al., 2016; Delikurt et al., 2015; Paneque et al., 2016; Pasca et al., 2022; Slomp et al., 2022). Many discussions with the participants in this project revolved around misconceptions around the role of a genetic counsellor. Participants broadly defined a genetic counsellor as: an expert who seeks out relevant patient information; a risk analyst who identifies candidate patients for genetic testing; an information provider; and an educator. These qualities align very well with the definition of a genetic counselling ‘expert’ described by Miranda and colleagues (Miranda, Veach, Martyr, & LeRoy, 2016). Some participants explained that they depend on the input of a genetic counsellor to help clarify additional clinical management options to be considered for their patients as they themselves do not always have the time to access current literature for updates regarding clinical management for patients with specific genetic conditions. This additional aspect valued by participants aligns the role of a genetic counsellor to that of an educator and informant for patients, doctors, and other medical societies (J. G. Kromberg, E. B. Sizer, et al., 2013; J. G. Kromberg, T.-M. Wessels, et al., 2013; Means et al., 2020; Papaioannou & Kampourakis, 2020).

Participants believe that genetic counsellors have a scarce skillset in the medical profession. Tailoring and communicating custom-made genetic information (Harding et al., 2019; Medendorp et al., 2020; Skirton et al., 2015) was a skill appreciated by all participants as they felt they do not feel confident or qualified to perform the role of communicating genetic information themselves. Some explained that their training was focussed on clinical management and not the communication of information as it takes a lot of time and expertise to especially convey medical genetic information in

an informative and simplified manner to patients. This adds to findings of other research which illuminate how limited genetic knowledge and other potential barriers complicate the communication process of explaining genetic information (O'Shea et al., 2011; Paneque et al., 2016). Genetic principles of cancer, including the associated personal and familial inheritance risks, management options, and the recommended medical multidisciplinary teams, are examples of concepts with underlying genetic principles that were reported to be lacking by MPs (Cornel et al., 2014; Pasquier et al., 2022; Tutika et al., 2023).

Many participants were hesitant in ordering genetic tests, and they felt that they were not experienced or knowledgeable enough to interpret and explain the results to their patients. Similar to the available literature, participants preferred a genetic counsellor to request and follow through with patients regarding genetic testing requests as ordering and interpretation of genetic tests are a standard tool in the skillset of a genetic counsellor (B. Bernhardt, 2014; B. A. Bernhardt, Biesecker, & Mastromarino, 2000; Werner-Lin, McCoyd, & Bernhardt, 2016). In previous research studies, MPs were found to order genetic tests at random without considering guidelines to identify high-risk patients (Stenehjem et al., 2018) and without being able to interpret or apply the information gained from a genetic test (Parikh et al., 2020; Pasquier et al., 2022). The lack of a genetic background or associated skills were acknowledged by participants in this and other research groups as a possible reason why MPs are generally hesitant in requesting genetic tests (Baars, Henneman, et al., 2005; Baars, Scherpbier, et al., 2005; Harding et al., 2019; Parikh et al., 2020; Slomp et al., 2022). The inability of MPs to understand and convey genetic test results to their patients is a common reported concern (Parikh et al., 2020; Pasquier et al., 2022; Pearce et al., 2019; Tutika et al., 2023). Baars and colleagues investigated the genetic proficiency of newly graduated MPs and found that they were not confident in their skills to offer genetic services (Baars, Scherpbier, et al., 2005).

A few participants felt confident explaining more complex genetic results including VUS and microarray reports with their patients as they felt they had more years of experience to understand and communicate this information. Professional development in the genetics profession is not only the result of years of experience, like in the case of these participants, but it is also the result of the quality of experiences a genetic counsellor or MP encounters in their career (Miranda et al., 2016). Furthermore, the proficiency of some participants in this sample being able to explain complex genetic results supports the findings of another systematic review in 2019 by Pearce and colleagues who found that certain medical specialty fields including oncology and neurology would require trainees to be more proficient in genetic risk assessment compared to MPs such as cardiologists and primary care practitioners (Pearce et al., 2019). Research indicated that some healthcare providers, with limited genetics training, have attempted offering genetic counselling for their patients themselves. Some of their efforts were met with adverse outcomes such as providing misinformation to their patient(s) with limited psychosocial support and requesting inappropriate genetic testing (Bensend et al., 2014; Pasquier et al., 2022; Tutika et al., 2023). Incorrect interpretation of negative and/or VUS results was of particular concern for participants as they felt this could lead to detrimental

consequences for the patients and their families ranging from improper care and management to causing unnecessary anxiety.

In addition to the genetic counselling skillset explored above, participants explained how they value the role of a genetic counsellor being that of an emotional support pillar which has also been highlighted by other research groups (Benoit, Veach, & LeRoy, 2007; B. A. Bernhardt et al., 2000; Paneque et al., 2017). For participants the genetic counsellor was explained to offer support through listening, contextualising and assist with problem solving for difficult patient cases. For patients the genetic counsellor was appreciated as a "consoling counsellor" (participant 7) especially for patients who are faced with harsh psychosocial circumstances and complicated medical diagnoses that are difficult to deal with. One participant stated how they are relieved to have trained professionals available where patients can receive empathetic assistance when they themself feel emotionally overwhelmed. Similar to the phenomena of burnout, compassion fatigue refers to how professional caregivers are overwhelmed by repeated empathetic engagement with clients (Benoit et al., 2007). The handover of patients also exhibits a relationship of trust between genetic counsellor and MP. Many participants in this sample, as well as other research studies (Cordier et al., 2016; Paneque et al., 2017), emphasised trust as one of the most important aspects in ensuring holistic care as they feel comfortable knowing that their patients will receive the same quality care that they would be able to provide themselves. Furthermore, participants respected how genetic counsellors are rooted in medical ethics and empower patients with information to help them make an informed decision with regards to the available options for their care and management (Paneque et al., 2017; Skirton et al., 2015). Genetic counsellors are specifically trained to bridge the information gap of patients with an understanding of the emotional needs of a patient (B. A. Bernhardt et al., 2000). These different qualities voiced by participants could be argued to align very well with the Reciprocal-Engagement Model (REM) of Genetic Counselling Practice coined by McCarthy Veach and colleagues (Veach et al., 2007).

An interesting aspect in this research study was regarding the professional title: 'genetic counsellor'. More than half of the recruited MPs explained that the title "undermines" (participant 12) the genetic counselling service. Most of the participants stated that patients become doubtful around the word 'counselling' or 'counsellor' due to a possible underlying psychotherapy implication, which has a negative connotation for many people, and supports the findings of other research groups (Ebsworth & Foster, 2017; Means et al., 2020). Research on the continued relevance of the title 'genetic counsellor' was undertaken in 2020 by Means and colleagues who found perceptions, similar to that raised by participants in this sample, when interviewing leaders within the genetic counselling profession (Means et al., 2020). They concluded that genetic counselling providers should take charge in helping change the narrative associated with the title rather than changing the title itself. It has taken many years for the profession to incorporate into healthcare and the relevance of the genetics field will only become bigger with the rapid development of genetic technology. This narrative change is something which is already apparent with participants in this sample. Participants prefer to describe and explain the role of a genetic counsellor to their patients rather than just providing a patient with the title or label 'genetic

counsellor'. Taking into account the lack of knowledge, lack of human resources, limited exposure, and the ambiguity of the job title, an invariable link is shown to exist between the accessibility of genetic services and its usage (Delikurt et al., 2015).

Some participants claimed that they had patients respond to receiving a genetic counselling referral as not being something they require at the time, as there is nothing 'wrong with their head'. The aversion for patients attending 'counselling' or mental health services was shown to be influenced by cultural and religious beliefs (Means et al., 2020; Pasca et al., 2022). In South Africa there exists a large variety of cultures and religious practices each with its own respective ethical norms and values, including gender roles, that could contribute to feelings of doubt around certain health care services. Considering how lack of trust and cultural and religious insensitivities can create barriers between patients and healthcare providers (Harris et al., 2020; Means et al., 2020; Pasca et al., 2022; Thompson, Bazile, & Akbar, 2004), many participants felt that clinicians' personality or attitude as well as their perspective regarding genetic services play a significant role in what management options they would offer their patients. This supports findings of many other research groups that acknowledged a significant role of a clinician's attitude in the receipt of genetic counselling services in their patients (Paneque et al., 2016; Parikh et al., 2020; Rosenbaum et al., 2020). Regardless of the limitations and barriers explored in this research project, one can only feel inspired when MPs, with specific reference to the participants in this sample, consider genetic counselling services as an "essential adjunct" (participant 6) which should be involved in more healthcare settings beside their own private practices.

8. Chapter 5: Conclusion

Medical practitioners in this research study greatly appreciate the influence of genetic counsellors in their private practice and stated that they cannot see a future in medical management without their continued input and support. Most participants were only introduced to genetic counselling services during their specialist training, and they felt that physical exposure allowed them to better understand the role of the genetic counselling service. Participants appreciate the input of genetic counsellors in MDT meetings and consider them knowledgeable resources of information who can guide the MDT to provide holistic patient care management.

By facilitating online interviews in Afrikaans or English allowed for data-rich perspectives to be collected from participants regarding genetic services in the current healthcare system in South Africa. Some topics of interest brought to light in this study include the use of DTC testing platforms in standard healthcare and ambiguity regarding the professional title of a genetic counsellor. The title 'genetic counsellor' was regarded by many participants as undermining as they felt it does not fully capture or explain the role of the trained professional. Most participants explained that they will describe 'what a genetic counsellor is' to their patients rather than providing them with only the professional title to limit ambiguous interpretation.

Besides the misleading professional title, participants felt that barriers such as the shortage of trained genetic professionals, including genetic counsellors, financial constraints, and time are resources which limit the incorporation of genetic services in South Africa. Additionally, the lack of national healthcare guidelines makes it very difficult for MPs to know what patient care management to consider when their patients are diagnosed with a rare genetic condition. Despite the constraints affecting the integration of genetic services in South Africa, the participants felt that genetic counselling services are valuable in their private practices that they "cannot even image (imagine) going without it".

The growing demand for genetic services will only increase with the advancements of genetic technologies. This by extension means that the relevance of genetic counselling will only increase as genetic investigations become more incorporated into standard medical practice. I believe this research highlights the significance of genetic counselling services and how it will flourish in the future with escalations in genomic medicine. More genetically trained experts will be required to help incorporate novel therapies and management approaches into healthcare regimes worldwide. For additional comments please refer to the reflective piece in Appendix E.

Participants proposed different approaches that they would consider helpful to promote genetic counselling services, the main proposition being that genetic counsellors need to increase their presence in clinics and MDT meetings. It is time to address this issue in South Africa and invite the medical community and the population into the genomics era with the assistance of genetic experts including genetic counsellors.

9. Chapter 6: Limitations, recommendations, and practice implications

To our knowledge this is one of the first research endeavours investigating the perceptions of MPs who actively utilise the service of genetic counsellors. To limit data heterogeneity, MPs in private practice were recruited for this study. Of the recruited MPs, all were clarified to be medical specialists. This could be interpreted as a misrepresentation or ascertainment bias. Alternatively, the recruitment of only medical specialists could also be indicative of the limited number of MPs, including PCPs, who refer patients for genetic counselling.

In this sample many surgeons were recruited compared to other medical specialties. This overrepresentation of surgeons could impact the perceived viewpoints with regards to genetic counselling services in this study. MPs such as paediatricians and foetal medicine specialists would have different medical management implications of a genetic test compared to a surgeon who would need to consider specific screening and prophylactic surgery options. Having a predominance of surgeons in the study, other aspects which might be highlighted by these other specialists would have been missed. Participants were recruited by only approaching genetic counsellors who are currently stationed in Cape Town. Future research could investigate the impact of genetic counselling services throughout South Africa by exploring the experiences of MPs practising in both private and state-based systems throughout the whole country.

The lens of completing interviews in both Afrikaans and English allowed for interesting points of discussion to come to light such as that of the professional title of a genetic counsellor and DTC testing platforms. In future research the prospect of investigating the impact of genetic counselling in more native languages could highlight even more interesting and relevant data for the future of genetic counselling and medical management in South Africa and across the world.

The integration of genomic literacy into the medical academic curricula highlighted the involvement of general practitioners and other MPs in providing and referring patients to genetic services. Participants in this and other research studies felt that PCPs including family doctors, gynaecologists, and GPs should be more involved in offering genetic services (Carroll et al., 2021; David et al., 2015; J. Kromberg et al., 2006; Mikat-Stevens et al., 2015). The involvement of PCPs was explained as helping to reduce over- and under-referring patients to specialists and assisting in minimising associated consultation and testing costs. Additionally, the hope is for PCPs to become integrated in the management process with the other specialists to provide holistic and continuous care management (Chou et al., 2021; Paneque et al., 2017). Creating stronger relationships between MPs, including medical specialists, and PCPs to ultimately improve the level of patient care management (Carroll et al., 2021)

Participants explained that they use a variety of international guidelines to make decisions on clinical management for their patients. These participants and other research groups felt, however, that the necessity to deviate from international

guidelines due to resource limitations in the country make it very difficult to incorporate genetic services into standard medical management (Mikat-Stevens et al., 2015; Slomp et al., 2022). Many research articles have proposed/described different models and frameworks that can be utilised to incorporate genetic services into modern health care systems (Borle et al., 2022; Harding et al., 2019; Macleod et al., 2002; Patch & Middleton, 2018; Pearce et al., 2019; Slomp et al., 2022). Some of these innovative models were compared by Carroll and colleagues who investigated how genetic services can be incorporated specifically into services provided by PCPs (Carroll et al., 2021). They concluded that new service delivery models should of necessity include available genetics experts, including genetic counsellors, to improve the efficacy of the genetic service that can be provided to patients. The focus group investigation done by Carroll and colleagues found that PCPs want to engage more with genetic services, but are not trained or informed enough to do so (Carroll et al., 2021). They found that PCPs do not have the genetic knowledge, access to trained personnel, or necessary guidelines to incorporate genetic services into their current health care regime. This not only supports the findings of this research, but also highlights the opportunity for possible intervention.

Most MPs, including the participants recruited for this research study, are open to educational interventions such as seminars and meeting forums to assist in resolving their genetic knowledge gap (Baars, Henneman, et al., 2005; Baars, Scherpbier, et al., 2005; Chou et al., 2021; Fountzilas et al., 2022; Harding et al., 2019; Scherr et al., 2022; Wonkam et al., 2006). Participants proposed that genetic counselling and the associated services should be incorporated earlier into the medical student undergraduate training programme.

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11. References

Abacan, M., Alsubaie, L., Barlow-Stewart, K., Caanen, B., Cordier, C., Courtney, E., . . . Gardiner, K. (2019). The global state of the genetic counseling profession. *European Journal of Human Genetics, 27*(2), 183-197.

Adeoye-Olatunde, O. A., & Olenik, N. L. (2021). Research and scholarly methods: Semi-structured interviews. *Journal of the american college of clinical pharmacy, 4*(10), 1358-1367.

Archibald, M. M., Ambagtsheer, R. C., Casey, M. G., & Lawless, M. (2019). Using zoom videoconferencing for qualitative data collection: perceptions and experiences of researchers and participants. *International journal of qualitative methods, 18*, 1609406919874596.

Arndt, C., Davies, R., Gabriel, S., Harris, L., Makrelov, K., Modise, B., . . . Anderson, L. (2020). Impact of Covid-19 on the South African economy. *Southern Africa-Towards Inclusive Economic Development Working Paper, 111*.

Attard, C. A., Carmany, E. P., & Trepanier, A. M. (2019). Genetic counselor workflow study: The times are they a-changin'? *Journal of Genetic Counseling, 28*(1), 130-140.

Baars, M. J., Henneman, L., & ten Kate, L. P. (2005). Deficiency of knowledge of genetics and genetic tests among general practitioners, gynecologists, and pediatricians: a global problem. *Genetics in Medicine, 7*(9), 605-610.

Baars, M. J., Scherpbier, A. J., Schuwirth, L. W., Henneman, L., Beemer, F. A., Cobben, J. M., . . . ten Kate, L. P. (2005). Deficient knowledge of genetics relevant for daily practice among medical students nearing graduation. *Genetics in Medicine, 7*(5), 295-301.

Baumgartner, I. (2012). Handling interpretation and representation in multilingual research: A meta-study of pragmatic issues resulting from the use of multiple languages in a qualitative information systems research work. *Qualitative Report, 17*(42), 1.

Beene-Harris, R. Y., Wang, C., & Bach, J. V. (2007). Barriers to access: results from focus groups to identify genetic service needs in the community. *Public Health Genomics, 10*(1), 10-18.

Beitsch, P. D., & Whitworth, P. W. (2014). Can breast surgeons provide breast cancer genetic testing? An American Society of Breast Surgeons survey. *Annals of surgical oncology, 21*(13), 4104-4108.

Benoit, L. G., Veach, P. M., & LeRoy, B. S. (2007). When you care enough to do your very best: Genetic counselor experiences of compassion fatigue. *Journal of Genetic Counseling, 16*(3), 299-312.

Bensend, T. A., Veach, P. M., & Niendorf, K. B. (2014). What's the harm? Genetic counselor perceptions of adverse effects of genetics service provision by non-genetics professionals. *Journal of Genetic Counseling, 23*(1), 48-63.

Bernhardt, B. (2014). Genetic counselors and the future of clinical genomics. *Genome Medicine, 6*(7), 49. doi:10.1186/gm565

Bernhardt, B. A., Biesecker, B. B., & Mastromarino, C. L. (2000). Goals, benefits, and outcomes of genetic counseling: client and genetic counselor assessment. *American journal of medical genetics, 94*(3), 189-197.

Biesecker, B. B., & Marteau, T. M. (1999). The future of genetic counselling: an international perspective. *Nature genetics, 22*(2), 133-137.

Blacker, D., Racette, S. R., Sklar, P., & Smoller, J. W. (2005). Psychiatric genetics: a survey of psychiatrists' knowledge, opinions, and practice patterns. *J Clin Psychiatry, 66*(7), 821-830.

Bonke, B., Stijnen, T., Tibben, A., Lindhout, D., & Clarke, A. J. (2005). Genetic risk estimation by healthcare professionals. *Medical journal of Australia, 182*(3), 116-118.

Booth, A., Noyes, J., Flemming, K., Moore, G., Tunçalp, Ö., & Shakibazadeh, E. (2019). Formulating questions to explore complex interventions within qualitative evidence synthesis. *BMJ global health, 4*(Suppl 1), e001107.

Borle, K., Kopac, N., Dragojlovic, N., Rodriguez Llorian, E., Friedman, J. M., Elliott, A. M., . . . Gen, C. S. (2022). Where is genetic medicine headed? Exploring the perspectives of Canadian genetic professionals on future trends using the Delphi method. *European Journal of Human Genetics*. doi:10.1038/s41431-021-01017-2

Braun, V., & Clarke, V. (2021). To saturate or not to saturate? Questioning data saturation as a useful concept for thematic analysis and sample-size rationales. *Qualitative research in sport, exercise and health, 13*(2), 201-216.

Brett, G. R., Metcalfe, S. A., Amor, D. J., & Halliday, J. L. (2012). An exploration of genetic health professionals' experience with Direct-to-Consumer genetic testing in their clinical practice. *European Journal of Human Genetics, 20*(8), 825-830.

Burke, W., Khoury, M. J., Stewart, A., & Zimmern, R. L. (2006). The path from genome-based research to population health: development of an international public health genomics network. *Genetics in Medicine, 8*(7), 451-458.

Carroll, J. C., Morrison, S., Miller, F. A., Wilson, B. J., Permaul, J. A., & Allanson, J. (2021). Anticipating the primary care role in genomic medicine: expectations of genetics health professionals. *Journal of Community Genetics, 12*(4), 559-568. doi:10.1007/s12687-021-00544-1

Case, D. O., Johnson, J. D., Andrews, J. E., Allard, S. L., & Kelly, K. M. (2004). From two-step flow to the Internet: The changing array of sources for genetics information seeking. *Journal of the American Society for Information Science and Technology, 55*(8), 660-669.

Chou, A. F., Duncan, A. R., Hallford, G., Kelley, D. M., & Dean, L. W. (2021). Barriers and strategies to integrate medical genetics and primary care in underserved populations: a scoping review. *Journal of Community Genetics, 12*(3), 291-309. doi:10.1007/s12687-021-00508-5

Cordier, C., Taris, N., Moldovan, R., Sobol, H., & Voelckel, M.-A. (2016). Genetic professionals' views on genetic counsellors: a French survey. *Journal of Community Genetics, 7*(1), 51-55.

Cornel, M. C., van El, C. G., & Borry, P. (2014). The challenge of implementing genetic tests with clinical utility while avoiding unsound applications. *Journal of Community Genetics, 5*(1), 7-12. doi:10.1007/s12687-012-0121-1

Creswell, J. W. (2002). *Educational research: Planning, conducting, and evaluating quantitative* (Vol. 7): Prentice Hall Upper Saddle River, NJ.

Davey, A., Rostant, K., Harrop, K., Goldblatt, J., & O'Leary, P. (2005). Evaluating genetic counseling: client expectations, psychological adjustment and satisfaction with service. *Journal of Genetic Counseling, 14*(3), 197-206.

David, S. P., Johnson, S. G., Berger, A. C., Feero, W. G., Terry, S. F., Green, L. A., . . . Ginsburg, G. S. (2015). Making personalized health care even more

personalized: Insights from activities of the IOM Genomics roundtable. *The Annals of Family Medicine, 13*(4), 373-380. doi:10.1370/afm.1772

DeJonckheere, M., & Vaughn, L. M. (2019). Semistructured interviewing in primary care research: a balance of relationship and rigour. *Family Medicine and Community Health, 7*(2), e000057. doi:10.1136/fmch-2018-000057

Delikurt, T., Williamson, G. R., Anastasiadou, V., & Skirton, H. (2015). A systematic review of factors that act as barriers to patient referral to genetic services. *European Journal of Human Genetics, 23*(6), 739-745.

Descript. (2017). [Voice-to-text transcription tool.] Retrieved from https://www.descript.com/terms

Donohue, K. E., Gooch, C., Katz, A., Wakelee, J., Slavotinek, A., & Korf, B. R. (2021). Pitfalls and challenges in genetic test interpretation: An exploration of genetic professionals experience with interpretation of results. *Clin Genet, 99*(5), 638-649.

Druker, H., Zelley, K., McGee, R. B., Scollon, S. R., Kohlmann, W. K., Schneider, K. A., & Wolfe Schneider, K. (2017). Genetic counselor recommendations for cancer predisposition evaluation and surveillance in the pediatric oncology patient. *Clinical Cancer Research, 23*(13), e91-e97. doi:10.1158/1078-0432.Ccr-17-0834

Düsterwald, G. (2015). *Understanding doctors' knowledge and attitudes concerning genetics and genetic services in South Africa.* University of Cape Town,

Ebsworth, S. J., & Foster, J. L. (2017). Public perceptions of mental health professionals: stigma by association? *Journal of Mental Health, 26*(5), 431-441.

Fountzilas, E., Apostolou, P., Vasiliadis, A. V., Aivazi, D., Saloustros, E., & Fostira, F. (2022). Physicians' experience, practice and education, on genetic testing and genetic counseling: a nationwide survey study in Greece. *Fam Cancer*. doi:10.1007/s10689-022-00290-4

Geller, G., Tambor, E. S., Chase, G. A., Hofman, K. J., Faden, R. R., & Holtzman, N. A. (1993). Incorporation of genetics in primary care practice: Will physicians do the counseling and will they be directive? *Archives of Family Medicine, 2*(11), 1119.

Gill, M. J. (2020). Phenomenology as qualitative methodology. *Qualitative Analysis: Eight Approaches*, 73-94.

Godino, L., & Skirton, H. (2012). A systematic review of nurses' knowledge of genetics. *Journal of Nursing Education and Practice, 2*(3), 173-184.

Greenberg, J., Kromberg, J., Loggenberg, K., & Wessels, T.-M. (2012). Genetic counseling in South Africa. *Genomics and Health in the Developing World. Oxford Mongraphs of Medical Genetics. In: Kumar D, Ed.. P.. Oxford: Oxford University Press*, 531-546.

Grosse, S. D., & Khoury, M. J. (2006). What is the clinical utility of genetic testing? *Genetics in Medicine, 8*(7), 448-450. doi:10.1097/01.gim.0000227935.26763.c6

Hallowell, N., Cooke, S., Crawford, G., Parker, M., & Lucassen, A. (2009). Healthcare professionals' and researchers' understanding of cancer genetics activities: a qualitative interview study. *Journal of Medical Ethics, 35*(2), 113-119. doi:10.1136/jme.2008.024224

Hannig, V., Cohen, M., Pfotenhauer, J., Williams, M., Morgan, T., & Phillips Iii, J. (2014). Expansion of genetic services utilizing a general genetic counseling clinic. *Journal of Genetic Counseling, 23*(1), 64-71.

Harding, B., Webber, C., Rühland, L., Dalgarno, N., Armour, C., Birtwhistle, R., . . . Phillips, S. P. (2019). Bridging the gap in genetics: a progressive model for primary to specialist care. *BMC medical education, 19*(1), 1-10.

Harris, J. R. A., Crumb, L., Crowe, A., & McKinney, J. L. G. (2020). African Americans' perceptions of mental illness and preferences for treatment. *Journal of Counselor Practice*.

Inc., Z. V. C. (2016). Security guide. Zoom Video Communications Inc. Retrieved from https://d24cgw3uvb9a9h.cloudfront.net/static/81625/doc/Zoom-Security-White-Paper.pdf

Jenkins, S., & Arribas-Ayllon, M. (2016). Genetic counselling for psychiatric disorders: accounts of psychiatric health professionals in the United Kingdom. *Journal of Genetic Counseling, 25*(6), 1243-1255.

Khoury, M. J., Jones, K., & Grosse, S. D. (2006). Quantifying the health benefits of genetic tests: the importance of a population perspective. *Genetics in Medicine, 8*(3), 191-195.

Kim, N., Kong, S.-Y., Yoo, J., Kim, D.-H., Seo, S. H., & Kim, J. (2022). Current issues, challenges, and future perspectives of genetic counseling in Korea. *alm, 42*(3), 314-320. doi:10.3343/alm.2022.42.3.314

Klitzman, R., Chung, W., Marder, K., Shanmugham, A., Chin, L. J., Stark, M., . . . Appelbaum, P. S. (2013). Attitudes and practices among internists concerning genetic testing. *Journal of Genetic Counseling, 22*(1), 90-100.

Koeller, D. R., Uhlmann, W. R., Carere, D. A., Green, R. C., Roberts, J. S., & Group, P. G. S. (2017). Utilization of genetic counseling after Direct-to-Consumer genetic testing: Findings from the impact of Personal Genomics (PGen) study. *Journal of Genetic Counseling, 26*(6), 1270-1279. doi:10.1007/s10897-017-0106-7

Kromberg, J., Parkes, J., & Taylor, S. (2006). Genetic counselling as a developing healthcare profession: a case study in the queensland context. *Australian Journal of Primary Health, 12*(1), 33-39.

Kromberg, J. G., Sizer, E. B., & Christianson, A. L. (2013). Genetic services and testing in South Africa. *Journal of Community Genetics, 4*(3), 413-423.

Kromberg, J. G., Wessels, T.-M., & Krause, A. (2013). Roles of genetic counselors in South Africa. *Journal of Genetic Counseling, 22*(6), 753-761.

Kromberg, J. G. R., Sizer, E. B., & Christianson, A. L. (2013). Genetic services and testing in South Africa. *Journal of Community Genetics, 4*(3), 413-423. doi:10.1007/s12687-012-0101-5

Lakens, D. (2022). Sample size justification. *Collabra: Psychology, 8*(1), 33267.

Macleod, R., Craufurd, D., & Booth, K. (2002). Patients' perceptions of what makes genetic counselling effective: An interpretative phenomenological analysis. *Journal of Health Psychology, 7*(2), 145-156.

Maiese, D. R., Keehn, A., Lyon, M., Flannery, D., & Watson, M. (2019). Current conditions in medical genetics practice. *Genetics in Medicine, 21*(8), 1874-1877.

Majumder, M. A., Guerrini, C. J., & McGuire, A. L. (2021). Direct-to-Consumer genetic testing: value and risk. *Annual review of medicine, 72*, 151-166.

Makransky, G., Bonde, M. T., Wulff, J. S., Wandall, J., Hood, M., Creed, P. A., . . . Nørremølle, A. (2016). Simulation based virtual learning environment in medical genetics counseling: an example of bridging the gap between theory and practice in medical education. *BMC medical education, 16*(1), 1-9.

Malterud, K., Siersma, V. D., & Guassora, A. D. (2016). Sample size in qualitative interview studies: guided by information power. *Qualitative Health Research, 26*(13), 1753-1760.

Maphumulo, W. T., & Bhengu, B. R. (2019). Challenges of quality improvement in the healthcare of South Africa post-apartheid: A critical review. *Curationis, 42*(1), e1-e9. doi:10.4102/curationis.v42i1.1901

Marks, J. H. (2004). 2003 ASHG Award for Excellence in Human Genetics Education. The importance of genetic counseling. *American journal of human genetics, 74*(3), 395-396.

Means, C., Cirino, A., Swenson, K. B., & Austin, J. (2020). I am a Genetic Counselor": A qualitative exploration of field leaders' perceptions of the title "genetic counselor. *Journal of Genetic Counseling, 29*(1), 97-104.

Medelyan, A. (2020). Coding qualitative data: How to code qualitative research. In.

Medendorp, N. M., van den Heuvel, L. M., Han, P. K., Hillen, M. A., & Smets, E. M. (2020). Communication skills training for healthcare professionals in providing genetic counseling: A scoping literature review. *Patient Educ Couns*.

Mhlanga, D., & Ndhlovu, E. (2021). Explaining the demand for private healthcare in South Africa. *Interdisciplinary Journal of Economics and Business Law, 10*(2), 98-118.

Mikat-Stevens, N. A., Larson, I. A., & Tarini, B. A. (2015). Primary-care providers' perceived barriers to integration of genetics services: a systematic review of the literature. *Genetics in Medicine, 17*(3), 169-176.

Miranda, C., Veach, P. M., Martyr, M. A., & LeRoy, B. S. (2016). Portrait of the master genetic counselor clinician: a qualitative investigation of expertise in genetic counseling. *Journal of Genetic Counseling, 25*(4), 767-785.

Neubauer, B. E., Witkop, C. T., & Varpio, L. (2019). How phenomenology can help us learn from the experiences of others. *Perspect Med Educ, 8*(2), 90-97. doi:10.1007/s40037-019-0509-2

O'Shea, R., Murphy, A. M., Treacy, E., Lynch, S. A., Thirlaway, K., & Lambert, D. (2011). Communication of genetic information by other health professionals: the role of the genetic counsellor in specialist clinics. *Journal of Genetic Counseling, 20*(2), 192-203.

Ormond, K. E., Laurino, M. Y., Barlow-Stewart, K., Wessels, T. M., Macaulay, S., Austin, J., & Middleton, A. (2018). *Genetic counseling globally: Where are we now?* Paper presented at the American Journal of Medical Genetics Part C: Seminars in Medical Genetics.

Oxley, J., Günhan, E., Kaniamattam, M., & Damico, J. (2017). Multilingual issues in qualitative research. *Clinical linguistics & phonetics, 31*(7-9), 612-630.

Paneque, M., Serra-Juhé, C., Pestoff, R., Cordier, C., Silva, J., Moldovan, R., & Ingvoldstad, C. (2017). Complementarity between medical geneticists and genetic counsellors: its added value in genetic services in Europe. *European Journal of Human Genetics, 25*(8), 918-923.

Paneque, M., Turchetti, D., Jackson, L., Lunt, P., Houwink, E., & Skirton, H. (2016). A systematic review of interventions to provide genetics education for primary care. *BMC family practice, 17*(1), 89.

Papaioannou, K., & Kampourakis, K. (2020). Health-care professionals' awareness and understanding of genomics. In *Applied Genomics and Public Health* (pp. 225-242): Elsevier.

Parikh, D. A., Dickerson, J. C., & Kurian, A. W. (2020). Health disparities in germline genetic testing for cancer susceptibility. *Current Breast Cancer Reports, 12*(2), 51-58. doi:10.1007/s12609-020-00354-3

Pasca, C., Carroll, R., Cragun, R. T., & Cragun, D. L. (2022). Knowledge and perceptions of the genetic counseling profession among a national cross-sectional sample of U.S. adults. *Journal of Genetic Counseling, 31*(1), 206-217. doi:https://doi.org/10.1002/jgc4.1473

Pasquier, L., Minguet, G., Moisdon-Chataigner, S., Jarno, P., Denizeau, P., Volf, G., . . . Moutel, G. (2022). How do non-geneticist physicians deal with genetic tests? A qualitative analysis. *European Journal of Human Genetics, 30*(3), 320-331.

Patch, C., & Middleton, A. (2018). Genetic counselling in the era of genomic medicine. *British Medical Bulletin, 126*(1), 27-36.

Pearce, C., Goettke, E., Hallowell, N., McCormack, P., Flinter, F., & McKevitt, C. (2019). Delivering genomic medicine in the United Kingdom National Health Service: a systematic review and narrative synthesis. *Genetics in Medicine, 21*(12), 2667-2675. doi:10.1038/s41436-019-0579-x

Pestoff, R., Johansson, P., Nilsen, P., & Gunnarsson, C. (2019). Factors influencing use of telegenetic counseling: perceptions of health care professionals in Sweden. *Journal of Community Genetics, 10*(3), 407-415.

Premji, S., Kosny, A., Yanar, B., & Begum, M. (2020). Tool for the meaningful consideration of language barriers in qualitative health research. *Qualitative Health Research, 30*(2), 167-181.

Pritchard, D., Goodman, C., & Nadauld, L. D. (2022). Clinical utility of genomic testing in cancer care. *JCO Precision Oncology, 6*, e2100349.

Rantanen, E., Hietala, M., Kristoffersson, U., Nippert, I., Schmidtke, J., Sequeiros, J., & Kääriäinen, H. (2008). What is ideal genetic counselling? A survey of current international guidelines. *European Journal of Human Genetics, 16*(4), 445-452.

Reeves, S., Albert, M., Kuper, A., & Hodges, B. D. (2008). Why use theories in qualitative research? *BMJ, 337*.

Reid, S. J., Peacocke, J., Kornik, S., & Wolvaardt, G. (2018). Compulsory community service for doctors in South Africa: A 15-year review. *South African Medical Journal, 108*(9).

Resta, R., Biesecker, B. B., Bennett, R. L., Blum, S., Estabrooks Hahn, S., Strecker, M. N., & Williams, J. L. (2006). A new definition of genetic counseling: National Society of Genetic Counselors' task force report. *Journal of Genetic Counseling, 15*, 77-83.

Rodas-Pérez, C., Clarke, A., Powell, J., & Thorogood, M. (2015). Challenges for providing genetic counselling in Colombian genetic clinics: the viewpoint of the physicians providing genetic consultations. *Journal of Community Genetics, 6*(3), 301-311.

Rosenbaum, M., Dineen, R., Schmitz, K., Stoll, J., Hsu, M., & Hodges, P. D. (2020). Interpreters' perceptions of culture bumps in genetic counseling. *Journal of Genetic Counseling, 29*(3), 352-364.

Saldaña, J. (2021). The coding manual for qualitative researchers. *The coding manual for qualitative researchers*, 1-440.

Salmons, J. (2014). *Qualitative online interviews: Strategies, design, and skills*: Sage Publications.

Samuel, G. N., Dheensa, S., Farsides, B., Fenwick, A., & Lucassen, A. (2017). Healthcare professionals' and patients' perspectives on consent to clinical

genetic testing: moving towards a more relational approach. *BMC Medical Ethics, 18*(1), 47. doi:10.1186/s12910-017-0207-8

Saunders, B., Sim, J., Kingstone, T., Baker, S., Waterfield, J., Bartlam, B., . . . Jinks, C. (2018). Saturation in qualitative research: exploring its conceptualization and operationalization. *Qual Quant, 52*(4), 1893-1907. doi:10.1007/s11135-017-0574-8

Scherr, C. L., Kalke, K., Ramesh, S., Fakhari, H., Dellefave-Castillo, L. M., Smith, M. E., . . . Rasmussen-Torvik, L. J. (2022). Integrating clinical genetics in cardiology: Current practices and recommendations for education. *Genetics in Medicine*. doi:https://doi.org/10.1016/j.gim.2022.02.003

Simarro, F. S. (2022). Advances in clinical genetics and its current challenges. *Anales de Pediatría (English Edition)*.

Simpson, B., Dissanayake, V., Wickramasinghe, D., & Jayasekara, R. (2003). Prenatal testing and pregnancy termination in Sri Lanka: views of medical students and doctors. *Ceylon Medical Journal, 48*, 129-132.

Skirton, H., Cordier, C., Ingvoldstad, C., Taris, N., & Benjamin, C. (2015). The role of the genetic counsellor: a systematic review of research evidence. *European Journal of Human Genetics, 23*(4), 452-458.

Slomp, C., Morris, E., Knoppers, B. M., Lynd, L. D., Dey, A., Adam, S., . . . Gen, C. S. (2022). The stepwise process of integrating a genetic counsellor into primary care. *European Journal of Human Genetics*. doi:10.1038/s41431-022-01040-x

Squires, A. (2009). Methodological challenges in cross-language qualitative research: A research review. *International journal of nursing studies, 46*(2), 277-287.

Stenehjem, D. D., Au, T., Sainski, A. M., Bauer, H., Brown, K., Lancaster, J., . . . Brixner, D. I. (2018). Impact of a genetic counseling requirement prior to genetic testing. *BMC Health Services Research, 18*(1), 1-11.

Taylor, A., & Kahn, D. (2014). The RWOPS debate - yes we can! *SAMJ: South African Medical Journal, 104*, 475-477. Retrieved from http://www.scielo.org.za/scielo.php?script=sci_arttext&pid=S0256-95742014000700014&nrm=iso

Thom, J., & Haw, T. (2021). Awareness of genetic counseling services among allied healthcare professionals in South Africa. *Journal of Genetic Counseling, 30*(6), 1649-1657.

Thompson, V. L. S., Bazile, A., & Akbar, M. (2004). African Americans' perceptions of psychotherapy and psychotherapists. *Professional psychology: Research and practice, 35*(1), 19.

Thunberg, S., & Arnell, L. (2022). Pioneering the use of technologies in qualitative research–A research review of the use of digital interviews. *International Journal of Social Research Methodology, 25*(6), 757-768.

Tutika, R. K., Bennett, J. A., Abraham, J., Snape, K., Tatton-Brown, K., Kemp, Z., . . . Openshaw, M. R. (2023). Mainstreaming of genomics in oncology: a nationwide survey of the genomics training needs of UK oncologists. *Clinical Medicine, 23*(1), 9-15. doi:10.7861/clinmed.2022-0372

van der Wouden, C. H., Carere, D. A., Maitland-van der Zee, A. H., Ruffin, M. T. t., Roberts, J. S., & Green, R. C. (2016). Consumer perceptions of interactions with primary care providers after Direct-to-Consumer personal genomic testing. *Ann Intern Med, 164*(8), 513-522. doi:10.7326/m15-0995

Van Langen, I., Birnie, E., Leschot, N., Bonsel, G., & Wilde, A. (2003). Genetic knowledge and counselling skills of Dutch cardiologists: sufficient for the genomics era? *European heart journal, 24*(6), 560-566.

Veach, P. M., Bartels, D. M., & LeRoy, B. S. (2007). Coming full circle: a reciprocal-engagement model of genetic counseling practice. *Journal of Genetic Counseling, 16*(6), 713-728.

Wadman, E., Conway, L., Garbarini, J., & Baker, M. (2022). Moral distress in genetic counseling: A study of North American genetic counselors. *Journal of Genetic Counseling*.

Walters, S., Aldous, C., & Malherbe, H. (2022). Addressing healthcare practitioner genetic knowledge, attitude and practices in Gauteng, South Africa–preparation for an educational intervention.

Werner-Lin, A., McCoyd, J. L., & Bernhardt, B. A. (2016). Balancing genetics (science) and counseling (art) in prenatal chromosomal microarray testing. *Journal of Genetic Counseling, 25*(5), 855-867.

Williams, M., & Moser, T. (2019). The art of coding and thematic exploration in qualitative research. *International Management Review, 15*(1), 45-55.

Wonkam, A., Njamnshi, A. K., & Angwafo, F. F. (2006). Knowledge and attitudes concerning medical genetics amongst physicians and medical students in Cameroon (sub-Saharan Africa). *Genetics in Medicine, 8*(6), 331-338.

Wool, C., & Dudek, M. (2013). Exploring the perceptions and the role of genetic counselors in the emerging field of perinatal palliative care. *Journal of Genetic Counseling, 22*(4), 533-543.

Young, M. (2016). Private vs. public healthcare in South Africa.

Zimmern, R. L., & Kroese, M. (2007). The evaluation of genetic tests. *Journal of Public Health, 29*(3), 246-250.

12. Appendix A – Invitation letter and informed consent form

Invitation letter

For attention medical practitioner.

Research study in genetic counselling in South Africa.

MMedSci student at University of Cape Town: Willem Pretorius

Research topic: Value of genetic counselling in medical practices in South Africa

Dear Doctor.

I herewith request your permission to conduct an online interview to determine the value of genetic counselling and genetic counsellors in a private medical practice. The online interviews will be structured on 4 main topics of interest and will be approximately 30-45 minutes in duration. Platforms considered to conduct these interviews include Zoom and Microsoft Teams. Due to the expected duration of the interview, and the fact that it could compromise your consulting hours, we could arrange for an interview at a date and time that would suit you even after office hours. Please note the following:

- Note that the online interview will only take place upon receipt of signed informed consent form (attached at the end of the project outline).
- For the sake of doing an in-depth qualitative analysis, each respective interview will be audio recorded.
- Privacy of information and participant confidentiality is of utmost importance and will not under any circumstance be made available for any reason. I will therefore also refer only to “genetic counsellor” and not the name of the genetic counsellor during the interview.
- In reporting the findings of this study, all participants and their responses will remain anonymous and confidential.
- All recorded data and documentation will be destroyed at the conclusion of the study and publication.
- For further information regarding the content of this study, please refer to the attached project outline.

If you are interested in participating in this study and require further information, please feel free to contact me via telephone: 076 104 2748; or email: prtwil008@myuct.ac.za.

Project title:

Exploring the value of genetic counselling by interviewing medical practitioners (MPs) who actively utilise the service in Cape Town.

Introduction and summary:

The advantage of genetic counsellors in the medical profession can be appreciated in all facets of health care ranging from prenatal to adulthood stages of normal human development. Genetic counsellors/counselling is still largely underrepresented in South Africa. With MPs of all speciality backgrounds being confronted with apparent genetic influencing factors contributing to their patients' well-being, it will become imperative for the medical work force to adapt and interpret genetic information into their standard regime of optimal patient health care. By investigating MPs who are actively utilizing the service in their private practices, it will become possible to appreciate why genetic counselling is valued by MPs in Cape Town. This investigation may also provide with a new approach in promoting the beneficial facets of genetic counselling to MPs who are unaware or unsure of this profession's role in standard health care systems.

The aim of this qualitative study is to understand why MPs make use of genetic counsellors in South Africa and acknowledge their personal experiences with regards to genetic counselling.

Objectives:

- Identify why, when, and how MPs started to utilise genetic counsellors/counselling services in Cape Town.
- Investigate how MPs, who actively make use of genetic counselling services in Cape Town, explain genetic concepts and the purpose/benefit of genetic counselling to their patients.
- Understand what MPs gain and/or risk losing when referring their patients for genetic counselling.
- Identify possible means in which genetic counselling can be presented MPs who do not actively make use of genetic counsellors/counselling.

Participants:

The participants for investigation in this study are MPs, more specifically private practicing MPs who actively make use of the service of genetic counsellors in South Africa (Cape Town).

Methods:

By responding to the researcher of this study via email or telephone call, an online face-to-face interview will be scheduled with each participating MPs whereby a set questions will be asked to investigate the core topics stipulated by the objectives. A Zoom or Microsoft Teams video call interface will be used to conduct the online interview. Estimated length of each online interview will be approximately 30-45 minutes long. The participant can decide a time and date which suits them best to do the interview regardless of office hours. Each interview will be audio recorded to allow the researcher to revisit the content and recover information that is best applicable in investigating the aim of this study.

Privacy:

The importance of privacy and confidentiality will remain of utmost importance in this study. All identifying documentation will be password protected and kept separately from the obtained interview data. In ensuring consistency of recorded thoughts, a gender- and race-neutral pseudonym will be used to establish an individual. Non-identifying descriptors may be used to identify a person when reporting a quote, i.e., Interviewee/Participant 3. No personal information of any sort will be made available at any time for any reason.

The principles outlined in the Declaration of Helsinki for all human or animal experimental investigations (Association, 2013) as well as the Protection of Personal Information Act 2013 (POPIA) regarding data protection for health research (De Bruyn, 2014; Staunton et al., 2020) have been considered when designing the protocol for this study and will be considered when reporting its findings.

Withdrawal:

Participation in this study is entirely voluntary. Should the participant feel uncomfortable with answering any of the postulated questions or prompts, they are under no obligation to do so. Should the participant no longer wish to participate in this study, they may withdraw at any point without providing any reason whatsoever. All audio recordings, transcripts and associated documentation will be discarded at the conclusion of the study and after publication.

Contact information:

For any questions or concerns please contact:

Willem Pretorius (student): tel: 076 104 2748
email: prtwil008@myuct.ac.za

Dr Tina-Marié Wessels (supervisor): tel: 021 406 6698
email: tina.wessels@uct.ac.za

Mrs Gill Düsterwald (supervisor): tel: 082 687 5857
email: gill.dusterwald@uct.ac.za

Ethics Chairperson Professor M. Blockman, Division of Pharmacology, Department of Medicine, K Floor, Old Main Building, Groote Schuur Hospital, tel: 021 406 6492

Statement of participation:

1. I am aware that my participation in this study is voluntary and that I may withdraw at any time.
2. I am aware that I am under no obligation to answer any question that I am not comfortable in answering.
3. I have received information about the study, including the aims and objectives, and have been allowed sufficient time to decide whether I would participate or not participate in the study.
4. I was provided with an opportunity to ask questions I have pertaining to the study, and I confirm that I understand the scope of the study.
5. I am aware that I may ask any additional questions I have for the duration of the study.
6. I declare that I have not been pressured in any way to participate in this study and that my participation in this study as at my own free will.
7. I agree that the results obtained from this study may be used for publication provided the anonymity and confidentiality of my participation in the study.
8. I am aware that I will not be reimbursed or receive any financial incentive from participating in this study.
9. I agree to the interview being voice-recorded.
10. I agree to the use of quotations from the interview, provided they are relevant to add value to the research.
11. I agree to offer feedback regarding the final write-up of this research to ensure accurate and relevant information is reported by the researcher for this study.

I hereby give my consent to participate in this study and that I am doing so at my own free will. I give consent for my interview to be recorded.

Name and Surname of Participant: ……

Date: ……

Signature: ……

13. Appendix B – Question guide

Medical practitioner interview 2020-2021

[Turn on the audio recording device and test it.]

Start of the session (Introduction):

- Researcher introduces themselves and the proposed research study to the participant.
- Provide an overview of how the research topic came to fruition – interest in genetic counselling, history of discrepancies regarding genetic counsellors worldwide, limited practicing genetic counsellors in South Africa.
- Have the participant read and verbally consent and/or sign the informed consent form.

Topic 1 (The origin of utilising genetic counsellors)

Purpose:

To identify why, when, and how medical practitioners started to utilise genetic counsellors/counselling services in Cape Town.

Question:

How did you first become aware of the genetic counselling service?

Prompts:

When did you start referring patients for genetic counselling?

When would you typically refer a patient for genetic counselling?

How did you start to refer patients for genetic counselling?

Why did you start to refer your patients for genetic counselling?

Topic 2 (Communicating the tenacity of genetics with patients)

Purpose:

To investigate how medical practitioners, who actively make use of genetic counselling services in Cape Town, explain genetic concepts, and the purpose/benefit of genetic counselling, to their patients.

Question:

Can you tell me how you introduce genetic concepts and genetic counselling to your patient(s)?

Prompts:

When/How do you explain a genetic diagnosis to you patient?

When/How do you explain to your patients to contact a genetic counsellor?

How do you elaborate on genetic health concerns with your patients?

Topic 3 (Perceived advantages and limitations of genetic counselling)

Purpose:

To appreciate what medical practitioners gain or risk losing in referring their patients to genetic counsellors/for genetic counselling.

Question:

What are some potential risks and rewards in referring your patients for genetic counselling?

Prompts

What do you expect from a genetic counsellor with regards to your referred patient?

What are some of the limitations in sending your patient for genetic counselling?

Tell me more about the advantages/gains in sending your patient to a genetic counsellor.

Tell me more about the disadvantages/harms in sending your patient to a genetic counsellor.

Topic 4 (Promoting genetic counselling for use by medical practitioners)

Purpose:

To identify a possible means of presenting genetic counselling to similar medical practitioners who do not actively make use of genetic counsellors/counselling.

Question:

What makes it difficult for medical practitioners like yourself to refer patients for genetic counselling?

Prompts:

Tell me more about patient referrals for genetic counselling.

How has your patient referrals changed over time*?*

What will make patient referrals to genetic counselling easier for you?

How would you encourage other medical practitioners to start making use of genetic counselling?

14. Appendix C – Case record form

Medical practitioner interview 2020-2021 (NOTES)

Start of the session (Introduction):

Participant name:

Date:

Atmosphere, initial thoughts:

Topic 1 (The origin of utilising genetic counsellors)

Question:

How did you first become aware of the genetic counselling service?

Topic 2 (Communicating the tenacity of genetics with patients)

Question:

Tell me how you would explain genetic concepts and genetic counselling to your patient(s)?

Topic 3 (Perceived advantages and limitations of genetic counselling)

Question:

What do you gain/risk losing when referring patients for genetic counselling?

Topic 4 (Promoting genetic counselling for use by medical practitioner)

Question:

What makes it difficult for medical practitioners like yourself to refer patients for genetic counselling?

Additional thoughts proceeding the session:

Reflection on session with participant:

15. Appendix D – Additional supplementary material

Table 3: Additional illustrative quotes supporting identified themes and subthemes	
Theme 1: The identity of genetics and genetic counselling services	
Subtheme 1: Genetic counselling as a foreign concept	• *"Absolutely. And how wide the field is, that is the other thing. You do not realise it. Yeah, from foetuses to cancers and everything in between." – Participant 7, Foetal medicine specialist* • *"Definitely not during undergraduate studies… we did genetics at the time, and I liked it, but I never realised that there was something like a genetic counsellor." – Participant 7, Foetal medicine specialist* • *"...the earlier you come into contact with the concept (genetic counselling), the more it will become part of your normal referral framework." – Participant 11, Paediatrician*
Subtheme 2: Genetic counsellor as a professional title	• *"...I think we are careful in how we explain it (genetic counsellor/counselling) so as to not scare off a patient…" – Participant 8, Radiologist* • *"I still get many people (medical professionals) who ask me 'who, what is a genetic counsellor?'. They do not know what it is." – Participant 4, Surgeon*
Subtheme 3: Genetic counsellor as part of multidisciplinary teams	• *"I can walk out with that patient and immediately say 'here is the [genetic counsellor].'…my experience is that if you do not have a genetic counsellor in your physical unit, then your patient uptake to go… to physically see them is very bad." – Participant 4, Surgeon*
Subtheme 4: Genetic management aptitude	• *"I explain as far as my head can go, as far as I am trained, because luckily it forms part of my training, but definitely not the level of depth you go into." – Participant 7, Foetal medicine specialist*
Theme 2: Resource limitations	
Subtheme 1: Lost in the cracks	• *"…our resources do not necessarily give us the opportunity to do it (practice medicine) the way it should be done." – Participant 1, Surgeon* • *"In my experience in the state sector we as foetal medicine specialists and the genetics specialists… even though we worked side-by-side, the integration was not so good." – Participant 12, Foetal medicine specialist*

Subtheme 2: Monetary constraints	• *"I'm also conscious that there's a financial implication for patients. So, I do as much of my own counselling as I can actually." – Participant 3, Physician* • *"(Sigh) The cost of genetic testing… very few people can afford to do it, but I understand why it is so expensive. I do not think it is an unwarranted price. It is just, you know, like what is the point sometimes." – Participant 10, Paediatrician*
Subtheme 3: Time – duration, amount, and moments	• *"You have to take their social, the whole social network and their family into consideration to understand what the impact of this problem will be on the patient. And I think a person in our field cannot get that (information) in that moment in one session. Nobody can. That is why it helps if we have someone who you can work with." – Participant 7, Foetal medicine specialist* • *"I suppose I'd like them to give the family a deeper level of understanding of the condition, which honestly, I do not have time for… I expect them to talk about whether they want to test other family members, if it is necessary or if it is appropriate." – Participant 10, Paediatrician*
Subtheme 4: Lack of local guidelines	• *"And I think someone needs to coordinate from a medical point of view that care. And we've got a problem here with that, because you know, we diagnose in passion, they (patients) actually need to be screened from multiple places and someone needs to coordinate that and make sure that everything's happening." – Participant 1, Surgeon*
Theme 3: The appreciable impact of genetic counselling	
Subtheme 1: The genetic counsellor skillset	• *"I do not think we are that advanced that we've got like a sort of mechanism in place to pick the VUS's up… I would hope the counsellor themselves would keep their finger on the pulse about that." – Participant 2, Surgeon* • *"I just want to point out that it does not need to be written. Like, I'm happy for… if someone phones me and says: 'this is, you know, this is the plan.'" – Participant 3, Physician* • *"… [the genetic counsellor] does the logistics of following up with the (genetic test) results. [The genetic counsellor] also offer (services) like risk estimations and other things besides just looking at genetic tests." – Participant 4, Surgeon* • *"It has become part of the management that I… you cannot even image going without it. So, it has become quite institutionalised." – Participant 8, Radiologist* • *There are frequently these variants of unknown significance, and it helps a lot that someone with more experience can put it into perspective. – Participant 11, Paediatrician*

Subtheme 2: A knowledgeable resource	• *"We are a complimentary system so you (the patient) will definitely benefit by speaking with someone (the genetic counsellor) … I think it (the genetic counsellor) is a good resource."– Participant 7, Foetal medicine specialist* • *"…if I have trouble with a report or the interpretation thereof, then I shall ask [name of genetic counsellor]" – Participant 8, Radiologist*
Subtheme 3: Ethical patient care and management	• *"I took a nurse up on it one day and I said: 'So, sorry, how can you do the test without counselling?' and she went: 'No but you are a doctor.' I said: 'No, but you should still be counselled-...' 'Oh no, that's not part of my job.' I got really cross with [them] because it was like, you can test someone, for me it is fine because I do understand. But because I asked [them]: 'are you not counselling me [be]cause I'm a doctor?' [They] said: 'no, I just, I do not counsel anyone.'" – Participant 3, Physician* • *"it is subjective, but I feel that my patients get through their journey easier if they… know what is going on." – Participant 6 Surgeon* • *"As long as it helps the clinician and the patient make the correct decision. And it does not make them make an incorrect decision or uninformed-…, but it will not be an uninformed decision. It will be an informed decision!" – Participant 9, Surgeon*
Subtheme 4: Propositions to address barriers	• *"I think there are few enough of these people (genetic counsellors) and I think every one of them should take responsibility to advertise the speciality or service as an entity." – Participant 4, Surgeon* • *"…if there was a genetic team at [name of hospital] they did not do a good job, I want to say, advertising their service… and not just a 'oh you can call me if there is a problem.' That is not going to work." – Participant 8, Radiologist*

16. Appendix E – Reflective piece

Reading through the latest research- and review articles relating to genetic counselling services, I had the impression that MPs, due to limited genetics knowledge and training, do not know what these services are or what these services provide their patients. These previous research endeavours also gave me the impression that many people working in the field of genetics think that other healthcare professionals do not see or appreciate what they as a genetics service provide patients.

Majority of the research assessing genetic counselling services are in the form of surveys and/or questionnaires. Despite being helpful in appraising a service or testing knowledge, I feel these assessments give a restricted and skewed look of MPs who actively use genetic counselling services. The available literature made me hypothesise that participants for this research study will not exactly know what a genetic counsellor and their services are or what these trained professionals provide their patients.

I was pleasantly surprised.

Talking with these participants made it very clear that genetic counsellors are an invaluable resource for their private medical practices. All the participants were eager to help spread awareness of genetic counselling services. They value the involvement of genetic counsellors and believe that their support, physical and/or emotional, and their genetics expertise allow them as MPs to provide better healthcare for their patients. I believe the discussions with these participants only highlight how relevant the role of a genetic counsellor will become in the years to come. Genetic counselling services will flourish along with the advances in genomic medicine continue and will require experts in the field to assist in incorporating novel therapies and management approaches into healthcare regimes worldwide.

This research project changed my approach when talking with MPs, students, and other healthcare professionals. I now actively try to initiate conversations about the use of genetics services and help these healthcare professionals become more aware of what genetics services are currently available in the South African healthcare setting. The ambiguity regarding the professional title ‘genetic counsellor’ apparent from the interviews also made me reconsider how I introduce myself to doctors and patients alike. By explaining my role as a genetic counsellor more broadly and simplified, I feel that people are more willing to engage with me and would freely ask me questions about genetics and genetics services.

I feel inspired about the future of genomic medicine in how some MPs perceive and use genetic counselling services. This research project helped me solidify my personal goal of spreading awareness of genetic counselling services in the next decade not just by adding to the number of available genetic counsellor workforce, but also to increase awareness among all healthcare professionals across South Africa.