Investigating the Opinions on Telephonic Advanced Maternal Age Genetic Counselling

By

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DECLARATION

I, *Samantha Bayley*, hereby declare that the work on which this dissertation/thesis is based is my original work (except where acknowledgements indicate otherwise) and that neither the whole work nor any part of it has been, is being, or is to be submitted for another degree in this or any other university.

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Abstract

Background: The research on telephonic genetic counselling (GC) services has increased since 2020. However, there is still limited research on the experience and opinions of this service from the patients' perspective. This is particularly true in low to middle-income settings such as South Africa. The advanced maternal age (AMA) telephonic GC service at Groote Schuur Hospital (GSH), has been implemented since COVID-19 regulations were enforced in 2020. This study aims to investigate the opinions and experiences of the patients on this telephonic service.

Methods: This qualitative study used individual semi-structured interviews, both in-person and telephonic, and followed a phenomenological approach. The data were analysed using thematic data analysis.

Results: The participants (n=9) had varying opinions about the telephonic GC service offered through GSH. The information gathering process varied for the participants; especially the differing Midwife Obstetric Units (MOUs) referrals and the information given. Some participants found online resources helpful, but not all participants felt the same. Overall, the participants felt the information given by the GC service was informative and useful. An important outcome of the research was a general trust between the GC trainee and participants but a distrust between other health care professionals and the participants. Numerous factors influenced decision-making concerning invasive testing including participants' fears and seeking control or having a sense of control based on if you would cope with having a child with DS.

Conclusion: The distrust in the healthcare system can have a significant impact on patients' understanding and decision-making in a GC session. Overall, there are benefits and barriers to be aware of, but most participants found the GC session informative and allowed them to make informed decisions. This research included a limited sample size, which prevents the generalizability of these findings.

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Dedication

I would like to dedicate this document to my parents and partner who continually support me, while providing wisdom and grace.

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Abbreviations

AMA – Advanced maternal age

DS – Down syndrome

GC – Genetic counselling

COVID-19 - Coronavirus Disease 2019

GSH – Groote Schuur Hospital

HREC - Human Research Ethics Committee

ID – Intellectual Disability

IPGC – In-person genetic counselling

MOU – Midwife Obstetric Unit

NIPT – Non-Invasive Prenatal Testing

NT – Nuchal Translucency

P1-9 – Participant 1-9

QF-PCR - Quantitative Fluorescence Polymerase Chain Reaction

RXH - Red Cross War Memorial Children’s Hospital

TBH - Tygerberg Hospital

TGC - Telephonic Genetic Counselling

TOP – Termination of pregnancy

T21 – Trisomy 21

UCT – University of Cape Town

WHO – World Health Organization

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Chapter 1: Introduction

This chapter aims to provide the necessary background information and a summary of the available literature regarding advanced maternal age genetic counselling and telecounselling.

1.1 Telehealth Platforms in Genetic Counselling

Genetic Counselling is a process of helping individuals and families understand and acclimate to the impact of the medical, psychosocial, and familial facets of genetic disease (Resta et al., 2006). Genetic Counselling (GC) involves the interpretation of medical and family histories, education on inheritance, testing, management, prevention, resources, and research (Resta et al., 2006). The goal of GC is to promote informed decision-making and adaptation to the risk or condition itself (Resta et al., 2006). Communication between client and genetic counsellor usually occurs in-person however with advancements in technology, telehealth has become more widely available and utilized.

Telehealth is used to communicate medical information between a medical professional and a patient in a virtual manner. The concept of telehealth has been a known possible venture since the early twentieth century and was introduced with the goal to provide better and wider access to health care while retaining expenses (Fritzen et al., 2019; Mitchell & Kan, 2019; Silveira et al., 2019; Van Velthoven & Cordon, 2019; Aziz et al., 2020; Breton et al., 2021; Mahtta et al., 2021; Park et al., 2021; Mechanic, Persaud & Kimball, 2022). Multiple platforms are available and frequently used in telehealth for consultations and these platforms include telephonic, video conferencing, interactive websites, applications, and videos. During the coronavirus disease 2019 (COVID-19) pandemic the adoption of telehealth rapidly increased due to lockdown regulations and minimising exposure to patients and health care workers (Mann et al., 2020). Telehealth has also become widely used in the different fields of medicine including clinical genetics and it is referred to as telegenetics. Telegenetics is a subcategory of telehealth focusing on genetic health care services, such as GC.

The increased adoption of telehealth during COVID-19 pandemic included the increase in telegenetics (Mann et al., 2020). While GC is traditionally performed in-person, telephonic GC

(TGC) was initially motivated in the cases where patients lived far from GC services to increase accessibility as well as in the case of disasters (Buchanan, Rahm & Williams, 2016; Lurie & Carr, 2018). The COVID-19 pandemic forced many GC services to adapt to telehealth service models and with this increase in telehealth services, questions regarding its quality and efficiency have been raised (Singh et al., 2021). In-person genetic counselling (IPGC) is still considered to be the "gold standard" service delivery model (Singh et al., 2021). As the COVID-19 pandemic regulations that forced the utilisation of telehealth platforms are now being withdrawn, GC services must now decide to continue with the utilisation of telehealth platforms or return to IPGC (Mahtta et al., 2021).

Telegenetics has mainly been used and researched in the pre-test cancer settings (Buchanan, Rahm & Williams, 2016). Cancer GC has used virtual communication the most compared to other areas of GC (Schwartz et al., 2014; Buchanan, Rahm & Williams, 2016) Cancer is a common condition, and cancer genetic services have grown exponentially due to genomic sequencing in both diagnostic and treatment opportunities (Buchanan, Rahm & Williams, 2016). Pre-test cancer sessions involve gathering family and medical information from the patient, as well as explaining the possible genetic cancer syndromes. These sessions do not require any physical examination but rather focus on communicating information and informed decision-making. Thus, many patients are referred to GC services. Telegenetics has allowed greater access to these services (Buchanan, Rahm & Williams, 2016; Breton et al., 2021; Mahtta et al., 2021).

Other genetic services have had less experience in telegenetics. In 2020, the Groote Schuur Hospital's advanced maternal age (AMA) service converted from an in-person service to a telegenetic service for the first time. AMA GC refers to informing women of their increased risk of their fetus having certain aneuploidies, specifically Down Syndrome, allowing women to make informed decisions regarding prenatal investigations. At Groote Schuur Hospital (GSH) the AMA GC service is currently telephonic, therefore, it is important to consider who has access to a telephone/mobile phone. In South Africa, 89.2% of people are cell phone subscribers as of 2011 (Lehohla, South Africa & Statistics South Africa, 2013). It was estimated that 25.5 million (42.1%) South Africans would be smartphone users by 2022 (S.

O’Dea, 2019). As of June 2022, 78.6% of South Africans have accessed the internet via a mobile device (Galal, 2022). This shows that although most people do have access to a cell phone, there are still many individuals that do not have access and may need to share a phone or cannot get access to a phone at all. This raises concerns about patient confidentiality when sharing phones and limiting access to the GC service (Wessels et al., 2021).

The most widely accessible and used telehealth platform is telephonic communication (Breton et al., 2021). Telephonic communication of medical information has been widely used due to most patients having their own, or access to a private phone. This is especially true in high income countries (Breton et al., 2021). Additionally, telephone calls are not a new form of communication, therefore, most individuals are comfortable with this communication platform (Gorrie et al., 2021a). Importantly, telephonic telehealth relies solely on verbal communication and no visual aids are used. More recent additions to telehealth include video conferencing/calls, interactive websites/applications, pre-recorded videos, and private messaging, which allow for the use of visual aids.

With the increased variety and availability of telegenetic communication forms, Genetic Counsellors are utilising telegenetics more over the past few years. The National Society of Genetic Counselors reported 36% of genetic counsellors use telephonic service delivery methods in 2020 (“Professional Status Survey (PSS) Executive Summary”, 2020). This increased to 74% of genetic counsellors using telephonic service delivery in 2022 (“Professional Status Survey (PSS) Executive Summary”, 2022). In 2020 the most common service delivery method was IPGC (95%) and in 2022 it is now telegenetics-audio-visual (82%) (“Professional Status Survey (PSS) Executive Summary”, 2020; “Professional Status Survey (PSS) Executive Summary”, 2022). Although, there has been this increase in telegenetics, research is mixed on whether it actually improves access to care (Hilgart et al., 2012; Buchanan, Rahm & Williams, 2016). More recent literature, states that telegenetics does improve access to care due to the virtual nature of telegenetics, the increased number of available providers and the reduction in financial burden of travel and time off work (Khan et al., 2021; Beretich, Sarasua & DeLuca, 2022). Each platform offers different advantages and disadvantages compared to each other and to in-person healthcare.

The benefits of telehealth include cost-effectiveness, improved access, timeliness of care, improved health outcomes and reduction in provider-patient supply-demand mismatch in some specialities (Mahtta et al., 2021). The delayed uptake in telehealth, until 2020, is commonly accredited to regulatory laws and suboptimal payment structures (Scott Kruse et al., 2018; Keesara, Jonas & Schulman, 2020; Mahtta et al., 2021; Park et al., 2021).

In 2020, many challenges for implementing telehealth were overcome out of necessity. Challenges included untrained staff and resistance to change (Scott Kruse et al., 2018; Emma E Thomas et al., 2022). However, some global challenges remain post COVID-19. These include concerns about patient data security, overutilisation or misuse of care, widening existing disparities in access to care, effectiveness, bandwidth, computer literacy, no personal phone, language barriers and perception of impersonal care (Scott Kruse et al., 2018; Mahtta et al., 2021). There are also concerns about explaining complexities via telehealth and the patient misunderstanding (Mahmoud, Jaramillo & Barteit, 2022). Thomas et al. highlights five requirements for overcoming such challenges for long-term telehealth utilisation. These requirements are building a skilful staff, restructuring funding, empowering patients, advancing digital networks, and incorporating telehealth into regular healthcare (Emma E Thomas et al., 2022).

As with in-person health care, telehealth presents varying difficulties in high versus low to middle income-countries. The benefits to telehealth in low to middle income-countries that have been observed include increased accessibility to healthcare, reducing financial strain and time saving, decreased need for transportation, saving resources, and continuation of care (Mahmoud, Jaramillo & Barteit, 2022). Many of the barriers experienced in low to middle income-countries are like those mentioned above, with increased emphasis on technological limitations. These technological limitations include unavailability of infrastructure or equipment, interrupted internet connection, limited technical support and technical issues during telehealth consultations (Mahmoud, Jaramillo & Barteit, 2022). In South Africa, there are additional factors to consider such as language diversity, patient literacy, and lack of familiarity with GC services (Scott, Watermeyer & Wessels, 2024). Therefore, in the South African context it is vital to remember these specific technological challenges as well as the general telehealth difficulties if telehealth is to be implemented sustainably. Although telegenetics has multiple benefits, it is not always the preferred mode of GC, as a study based in Japan found that telegenetic pre-test GC in the prenatal setting was the least preferred delivery mode for the patients (Nishiyama et al., 2021). From this study, the patients most preferred mode of delivery was IPGC, although their preference varied by experience, knowledge, attitudes and background characteristics (Nishiyama et al., 2021). The reason for telegenetics being the least preferred method was not explored in this article.

Similarly, genetic counsellors' opinions on telegenetics have been mixed (Hilgart et al., 2012; Danylchuk et al., 2021; Beretich, Sarasua & DeLuca, 2022).

Genetic counsellors have found their own challenges to telegenetics. These challenges include lack of administration, access to conferencing platforms, additional resources needed to coordinate consent to be signed and samples collected, building rapport with patients, technological issues, scheduling patients and distractions in the background (Schwartz et al., 2014; Trepanier & Allain, 2014; Buchanan, Rahm & Williams, 2016; Greenberg et al., 2020; Ahimaz et al., 2021; Gorrie et al., 2021a; Khan et al., 2021; Wessels et al., 2021).

Despite the challenges mentioned, literature from developed countries have shown that telegenetics preforms on par in many ways with IPGC. Telegenetics was shown to have comparable levels of rapport, trust, health behaviour modifications, confidence in privacy, and psychosocial outcomes (Danylchuk et al., 2021). Additionally, telegenetics had similar levels of understanding and knowledge of genetics (Gorrie et al., 2021a). Similarly, to previous research on telehealth in general, genetic counsellors have also reported advantages of telegenetics. These advantages include reduction in time waiting for appointments, reduced travel time, cost-effectiveness (for patients and the healthcare institution), decreases pressure on patients to test (more time to consider since an in-person appointment is often needed for sample collection) and decreased anxiety for some patients as they are in the comfort of their own homes (Trepanier & Allain, 2014; Buchanan, Rahm & Williams, 2016; Ahimaz et al., 2021; Gorrie et al., 2021a; Khan et al., 2021; Wessels et al., 2021). In South Africa, there are concerns about reduced health literacy becoming apparent during telegenetics and limited access to personal phones (Wessels et al., 2021).

1.2 Advanced Maternal Age Genetic Counselling

A female who is 35 years old or above at the time of conception is considered to be of AMA (Egan et al., 2000). There are various pregnancy complications associated with being of AMA (Lean et al., 2017). This research will focus on the association between AMA and the increased risk of aneuploidies (Hassold & Chiu, 1985; Eichenlaub-Ritter, 1996; Steuerwald et al., 2007).

Aneuploidy is the abnormal number of chromosomes in a cell instead of the typical 46 chromosomes. Trisomy 21 (Down Syndrome, T21), Trisomy 13 (Patau Syndrome), Trisomy 18 (Edward Syndrome) are three aneuploidy conditions that have been extensively researched and described to be associated with AMA. Aneuploidies account for 59.7% of first trimester pregnancy miscarriages (Pylyp et al., 2018). Trisomy 13, 18 and 21 have a prevalence of 1.49, 3.43 and 15.74 per 10 000 respectively in the United States (Mai et al., 2019).

Down syndrome (DS) is a pan-ethnic condition with an incidence of 2 per 1000 live births in South Africa (Delport et al., 1995; Miao et al., 2012; Loane et al., 2013; Schoonraad et al., 2020). The incidence is influenced by access to prenatal diagnostic, and pregnancy termination services and the average maternal age (Delport et al., 1995; Miao et al., 2012; Loane et al., 2013). The main features of DS include mild to moderate intellectual disability, growth retardation, and characteristic facial features (Akhtar & Bokhari, 2022). Some of the common health concerns with DS are congenital heart defects, vision issues, hearing impairment, and thyroid dysfunction (Weijerman & de Winter, 2010). Dysmorphic features of DS include flattened face, almond up-slanted eyes, single palmar crease, and others (Weijerman & de Winter, 2010). However, DS can be described as a spectrum with different individuals presenting with varied signs and severities (Akhtar & Bokhari, 2022). The cause of DS is known to be genetic with a third additional copy of chromosome 21. This can occur through nondisjunction, translocation, and mosaicism. Nondisjunction is the cause of 95% of DS cases and is associated with AMA (Coppedè, 2016). Nondisjunction describes the failure of normal chromosome segregation in meiosis resulting in an aneuploidy (Coppedè, 2016). DS caused by nondisjunction is more appropriately called T21. The remainder 5% of the cases are due to mosaicism and translocation which are not associated with AMA.

The association between AMA and an increased risk for T21 was first noted by Penrose (Penrose, 1933). The reason for the association between AMA and increased risk for T21, is not fully understood, yet it is one of the most important questions in medical genetics (Allen et al., 2009). This defining age limit for AMA, 35 years, is not universally used across South Africa. Due to resource constraints at GSH, only women who are 40 years or older at the time of conception are referred for AMA counselling.

There are multiple screening methods to identify women at higher risk of having a pregnancy affected with DS. Screening is done to allow the high-risk pregnancies an opportunity for further investigation such as invasive testing. AMA is commonly used as it is the easiest and most affordable screening tool, although the detection rate for DS using only maternal age is 50% (Nicolaides, 2011). The use of a combination of screening methods along with maternal age increases this detection rate (Durković et al., 2018).

Antenatal ultrasound is one of the most used screening methods. The detection rate of DS using AMA and a first or second trimester ultrasound is up to 85% when an extended nuchal translucency ultrasound is done (Howe et al., 2000; South African Society of Obstetricians and gynecology, 2018). Additional screening methods for aneuploidy include non-invasive prenatal testing (NIPT) and serum markers. The NIPT method has a 99% detection rate for DS and is the most accurate screening test available with no associated risks to the pregnancy. However, these options are more expensive (ranging from R3900.00 to R8930.00) and not offered in South Africa’s state healthcare sector.

In South Africa, availability of resources differs compared to high income countries. The state healthcare system has limited resources and genetic services are only offered in a few tertiary hospitals across the country (Thom & Haw, 2021). The locations of these state-run genetic services are Johannesburg, Durban, and Cape Town. In Cape Town, these services are mainly delivered at GSH, Red Cross War Memorial Children's Hospital (RXH), and Tygerberg Hospital (TBH). Patients are generally referred to these services from other hospitals around the Western Cape Province. AMA GC is offered through GSH and TBH. This research will focus on the telephonic service offered through GSH as each hospital's services can differ.

Prior to the COVID-19 pandemic, AMA (40 years or older at conception) women without a marker for DS were referred from their respective Midwife Obstetric Units (MOUs), clinics or secondary hospital and had GC in-person at GSH. These women were also offered a detailed foetal anomaly ultrasound at GSH if not already performed. These were offered in their first or second trimester up to 23 weeks' gestation. The GC consisted of history-taking, to assess any additional personal or familial risks, information-giving regarding DS and AMA, risk communication, options for further pregnancy management including invasive testing and options dependent on the results and, lastly, decision-making. If the patient decided to accept invasive testing, this was usually performed on the same day as the GC session and results were delivered either telephonically or in-person by the counsellor within 2 to 3 working days when the result of the quantitative fluorescent polymerase chain reaction (QF-PCR) became available. If the pregnancy was affected with DS, the women was offered a termination of pregnancy (TOP) until 24 weeks gestation, in accordance with the South African law and the internal policy of GSH ("Termination of Pregnancy (TOP)", 2022). Each session would normally take between 30 to 45 minutes.

Previous research on in-person AMA GC at GSH has been performed which showed that overall the patients found it to be a useful service (Vorster, 2020, 2021). This research highlighted that patients have limited health literacy concerning AMA risks and GC; therefore, patients had no expectations for the GC session. One patient from this study stated that they would not have come to GSH for GC if they were only offered GC and not an ultrasound (Vorster, 2020). This patient felt it was too far to come just for GC. Overall, participants from this study still felt this GC service helped them prepare for the possibility of having a child with DS (Vorster, 2020).

Once the COVID-19 lockdown regulations were enforced, this service changed substantially to reduce the risk of exposure to both the patients and the healthcare workers. Instead of AMA women (no markers seen on ultrasound) being referred to GSH, they were informed of their higher risk (1% risk or higher depending on age) of aneuploidies due to their age and provided with a pamphlet with further information on DS and the option of invasive testing (Personal communication: Prof K Fieggen, 2024). This pamphlet has the GSH Genetics teams' call phone number on it and women are informed that if they want more information, or would like invasive testing, to contact the number listed. The women that do not contact the call phone are not seen for GC and continue their antenatal care at their MOU/clinic or secondary hospital. The women that contacted the genetics team, though WhatsApp, are asked to send photos of their maternity book to gain pertinent medical information and a genetic counsellor, trainee (GC student or intern) or genetic nurse then calls these women and telephonically counsels them. For women that do not have a smart phone, they will either provide the necessary information via a telephone call, after sending a please-call-me or a SMS, or they will be asked to attend the GSH prenatal genetic clinic. The topics discussed in this session remain the same as the above mentioned in-person sessions. The women that are interested in invasive testing will then come into the clinic at GSH. There, a genetic counselling trainee will summarise what was discussed on the phone and go through the consent for the invasive test which will be performed on the same day. As referrals are usually made in the second trimester, invasive testing is usually performed by amniocentesis. Results are then communicated approximately 3 days later, either telephonically or in-person, dependent on the result and the wishes of the woman. If the pregnancy is affected with DS, the women would again be offered a TOP until 24 weeks gestation ("Termination of Pregnancy (TOP)", 2022).

1.3 Rationale

The COVID-19 pandemic required a rapid shift in the delivery methods of GC services locally. This resulted in the transitioning of the in-person AMA pre-test GC into a telegenetics service.

During the pandemic this adjustment was necessary to maintain social distancing and limit exposure to the virus. However, now that COVID-19 restrictions have been lifted and the telegenetics service has continued, the best delivery mode of this AMA service is debatable. Investigating the specific AMA TGC service at GSH will illuminate the challenges and advantages of this service in a South African setting.

The literature shows that in many ways telegenetics preforms on par with IPGC (Danylchuk et al., 2021; Gorrie et al., 2021a). Most of the research done in telegenetics has been based in high income countries. In South Africa, a low to middle-income country, very little research in telegenetics has been conducted. Thus, this research aims to gain an understanding into telegenetics in a South African setting. Additionally, many telehealth articles are quantitative. Using qualitative study design, this research aims to gain a greater understanding of the experiences and opinions of the patients who are cared for through telegenetic services and provide guidance on the way forward for AMA GC services at GSH in the South African setting.

1.4 Research question

What are the opinions of advanced maternal age patients on the telegenetic service provided to them through Groote Schuur Hospital?

Aim

To investigate the opinions of advanced maternal age patients on the telegenetic service provided to them through Groote Schuur Hospital.

Objectives

1. To identify patients’ positive and negative experiences and opinions of the service.

2. To determine patients’ understanding of what would be offered or discussed once they contacted our GC service.
3. To identify any barriers or challenges to telegenetic services in the AMA setting in a South African population.
4. To identify patients’ preferences with regards to this telegenetic service.

Chapter 2: Methodology

This chapter aims to inform the reader with the processes that were followed by the researcher to determine the opinions of advanced maternal age patients (AMA) on the telegenetic service provided to them through Groote Schuur Hospital (GSH).

2.1 Study Design

This was a qualitative study that used a phenomenological approach. Qualitative research is a research type that investigates and analyses deeper insights of real-life situations (Tenny, Brannan & Brannan, 2023). Qualitative research can assist in further understanding quantitative research and create more investigations (Tenny, Brannan & Brannan, 2023). Phenomenology is a research design that allows for the investigation of the human experience and the meaning individuals make of these experiences (Savin-Baden & Major, 2013; Liamputtong, Anderson & Bondas, 2016). This approach aims to uncover how participants experienced an event. This leads to a description of the essence of the phenomenon (Savin-Baden & Major, 2013). The phenomenological approach was suitable to this study as it aimed to gain insight into the participants' experience and opinions of the AMA telephonic Genetic Counselling (TGC) service.

To obtain the participants experiences of the AMA telegenetic service, individual semi-structured interviews were used. Interviews are commonly used as a data collection method in phenomenology because it allows for a deeper exploration of participants' experiences. The study interviews were conducted in-person and telephonically. An in-person interview was only possible if the participant had an up-coming appointment at GSH for an invasive diagnostic test. The question guide can be found in Appendix A: Question Guide. This study was prospective as participants were recruited as they came into the Genetic Counselling (GC) service. This allowed for each participant to still have a complete memory of their TGC session.

2.2 Research setting

As described in Chapter 1, AMA cases are counselled throughout the week depending on when a woman sends her pregnancy information to the GSH Genetic Team's Call Phone via WhatsApp. The women that sent their information to the Call Phone were then contacted for a TGC session regarding their AMA risks and options available to them. The counselling was done by a GC trainee. The GC sessions were 30-45 minutes long depending on the case. Each session included family history taking, information giving about Down Syndrome (DS) and AMA, a personal risk assessment, available options going forward regarding invasive testing and TOP and decision-making. If a woman wanted invasive testing, an amniocentesis was booked for her at GSH when she was between 16-23 weeks' gestation. The amniocentesis was performed at the Fetal Medicine Unit at GSH. The QF-PCR results of the amniocentesis were delivered telephonically or in-person about 3 days later. In this study women were interviewed one to three weeks after their telephonic counselling session regardless of their decision around invasive testing and TOP. Their decision around invasive testing and TOP is not a factor in this investigation as all these women receive the same telecounselling service despite their decision on these matters.

2.3 Participants

Participants included pregnant females that were classified as AMA and who had contacted the GSH TGC service and had received telephonic AMA GC. The participants were recruited through this service. Only participants that were counselled in English, regardless of their home language were included. Thus, the interviews were held in English, and this avoided the need for a translator to be present. Since this was a minor dissertation, there were time constraints, and a translator requires a lot more time both in the interviews and in the transcription process. The researcher is also limited by only being able to speak in English. All the genetic counsellors, genetic counselling interns and genetic nurses speak English and majority of patients are counselled in English. Considering patients were proficient and comfortable to have GC in English, it was assumed that these patients would also be comfortable with an interview in English; although as English may not be a participants' first language this may impede their ability to fully express themselves. The exclusion of non-English-speakers was a limitation in the study as it reduced the participant pool.

In qualitative research the number of participants cannot be predetermined, therefore the number of participants was based on the concept of reaching data saturation. Data collection was continued until data saturation was reached, if possible, within the limited amount of time afforded for this minor dissertation (Savin-Baden & Major, 2013). It was anticipated that 8-15 participants would be suitable to address the aims and objectives of the research. A total of 9 participants were interviewed and included in this research. The sample size allowed for the aim and objectives of this research to be answered. No new themes arose in the final interview and due to time constraints data collection ceased. Data saturation is a subjective concept, which is difficult to determined completely (Braun & Clarke, 2021).

Inclusion/exclusion criteria

Inclusion:

- Females that are 40 years and older at the point of conception.
- Clients who contacted the GSH GC service after being given the service’s number at their MOU.
- Clients who were telephonically counselled on the increased risks associated with AMA by a genetic counsellor, trainee or genetic nurse and have agreed to be contacted by the researcher for possible recruitment.
- Clients who had genetic counselling performed in English irrespective of their home language.

Exclusion:

- Women below 40 years of age.
- Women who do not speak nor understand English.
- Women who had a marker for DS or foetal anomaly identified during an ultrasound as these cases are generally counselled in-person.

2.4 Recruitment

Participants were recruited via purposive sampling, which is a selection based on the rationale in terms of being the most informative (Tenny, Brannan & Brannan, 2023). To investigate the TGC service participants would have needed to experience this service. This was a prospective study which was carried out from ethical approval until completion of the research; therefore, participants were recruited from May 2023 to October 2023 after each participant had GC for AMA risk association.

Suitable possible research participants were informed about the study at the end of the TGC session by the GC trainee that conducted the session. If the individual was interested in the research and consented to being contacted by the researcher, their details were given to the researcher, Samantha Bayley, by the GC trainee. Thereafter, the researcher contacted the prospective participant and informed them in more detail about the research and determined their willingness to participate. If they were willing to participate, an information sheet and consent form on the research was provided (Appendix B: Information Sheet for Participants). Consent was taken as is detailed below (Appendix C: Consent Form).

2.5 Research Procedure/Data Collection

This research used interviews to allow exchanges between participant and the interviewer. The semi-structured interviews, which followed a few pre-set questions while including additional questions in response to the participant, allowed for flexibility in the question guide (Savin-Baden & Major, 2013). The advantage of this interview type is that it allows for diversity of responses, by allowing the participant to express themselves and focus on issues that are important to them and it is useful when the participant is only interviewed once as was the case in this study (Savin-Baden & Major, 2013). This interview type was chosen as the best way to use the limited time available, while keeping the interviews focused on the relevant topic (Savin-Baden & Major, 2013). The question guide was developed with some instruction from the previous research done on this service and the research supervisors, all with the goal of answering the research question (Vorster, 2020).

Before interviewing participants, a mock interview was conducted with a colleague from the Human Genetic clinical staff, who are familiar with the service and perform the counselling on a routine basis. This was to ascertain the pace of interviews as well as the appropriateness of the questions and if any of the questions needed to be altered.

Interviews were either held in-person at GSH or telephonically according to the participant's preference and if they had an upcoming appointment at GSH. In-person interviews are believed to be better for this research project as it investigated a telephonic service. In-person interviews allowed the researcher and the interviewee to be synchronised in time and space, which allowed for body language to be expressed (Saarijärvi & Bratt, 2021). A safer and more comfortable space can also be created with in-person interviews (Saarijärvi & Bratt, 2021). Telephonic interviews were a necessity to include as an interview option as it removed the need to travel and saved time for participants, thus, it allowed more patients to participate in the research (Saarijärvi & Bratt, 2021). The telephonic interviews would provide insight into the practical elements of telephonic communication, which would be valuable in this research. These observations were recorded as field notes, which were then incorporated into the data analysis. Originally, the researcher proposed to perform in-person interviews with women who were going to have an upcoming appointment at GSH or a MOU/regional hospital/district hospital or clinic within a 20-kilometre radius of GSH. These hospitals/clinics included:

MOU:

Retreat Midwife Obstetrics Unit

Regional:

Mowbray Maternity Hospital

New Somerset Hospital

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District:

Victoria Hospital

Clinics:

District Six Community Day Centre

Dr. Abdurahman Community Health Clinic

Goodwood Community Day Centre

Green Point Community Health Clinic

Lady Michaelis Community Health Clinic

Maitland Community Health Clinic

Parow Community Day Centre

Retreat Clinic

Retreat Community Health Centre

In order to manage time and safety, only MOUs close by and unaffected by gang violence were approached for approval so as to decrease risks to both the participants and researcher. Permission from these institutions was only granted by GSH and Greenpoint Community Health Centre, therefore, in-person interviews could have only been performed at these two hospitals. No participants were referred from Greenpoint Community Health Centre; thus, all in-person interviews were done at GSH. The women that wanted to participate in the study but couldn't be seen in-person were interviewed telephonically. An appropriate time was arranged beforehand. Participants were required to fill in or verbally agree to a consent form (Appendix C: Consent Form) and completed a short socio-demographic survey (Appendix D: Socio-demographic Survey). If participants were in-person or could sign a document electronically then the consent was given to them to sign. Telephonically interviewed participants had the consent document read to them and verbal consent was taken, and if possible, the consent was also sent to them via WhatsApp or email. Interviews were voice recorded on a password protected smartphone for both the in-person and telephonic interviews.

2.6 Data Analysis

The audio recordings of the interviews were transcribed verbatim using Microsoft Word and then data organising was done using NVIVO (Release 14.23.0). Microsoft word online allows for audio files to be uploaded and transcribed with a Microsoft 365 subscription ("Microsoft Word - Word Processing Software | Microsoft 365", n.d.). This transcription was manually checked by reading through the automated transcription while listening to the recordings and, where necessary, corrected by the researcher to ensure accuracy. In South Africa, there are various accents that may not be picked up by the automated transcription, which was why the manual checking was necessary. NVIVO, from QSR International, is a software program that is used in qualitative and mixed-methods research (Yeager, 2022). It was used to organise unstructured audio and text data from interviews and journal articles into categories and themes (Yeager, 2022).

The data were analysed using thematic analysis, which included sifting through the data to find repeated patterns of meaning (Braun & Clarke, 2006, 2022). Coding was an important part of thematic analysis, where the researcher performed coding at various levels to deconstruct and find links in the data (Liamputtong, Anderson & Bondas, 2016). Braun and Clark's research proposed a step-by-step process to thematic analysis (Braun & Clarke, 2006, 2022; Liamputtong, Anderson & Bondas, 2016). The general steps were as follows, although, thematic analysis is not a linear process and is a more elaborate process (adapted from (Braun & Clarke, 2006, 2022; Liamputtong, Anderson & Bondas, 2016)):

The Thematic Analysis Process

1. Familiarising yourself with the data
    a. Transcribe the data yourself.
    b. Read the transcripts multiple times and writing down initial thoughts.
2. Generating initial codes
    a. Coding interesting features of data methodically across the data set, arranging data relevant to each code.
3. Searching for themes
    a. Collating codes into tentative themes.
    b. Gather all data relevant to each possible theme.
4. Reviewing themes
    a. Ensuring that themes work in relation to the coded extracts and the entire data set.
    b. Develop a thematic 'map' of the analysis.
5. Defining and naming themes
    a. Continuing analysis to process the specifics of each theme and the general story the analysis tells, creating clear definitions and names for each theme.
6. Producing the report
    a. The final chance for analysis and selection of vivid, compelling extract examples.
    b. Final analysis of the chosen extracts and relating the analysis to the research question and literature.

Figure 1: The Thematic Analysis Process

(Adapted from (Braun & Clarke, 2006, 2022; Liamputtong, Anderson & Bondas, 2016))

As data were collected, this framework was used to guide data analysis to ensure a comprehensive analysis was done. The data transcription occurred simultaneously to allow emerging areas of interest to be explored in future interviews. This allowed for analytical assumptions to be investigated in the ongoing data collection (Thompson Burdine, Thorne & Sandhu, 2021). The researcher and the supervisors began with the thematic analysis, through coding. Four interview transcripts were blindly coded by both the student and supervisors. The differences in coding were discussed collectively. The remaining transcripts were coded by the researcher and codes were discussed with the supervisors. Once all interviews were coded the Thematic Analysis process illustrated in Figure 1 was followed by the researcher and with guidance from the research supervisors. After reviewing the codes and preliminary themes multiple times with reflection individually and with the supervisors the final codes were reached. Codes were grouped based on patterns observed by the researcher. These patterns were discussed with the supervisors and adjusted. This reflective process occurred multiple times and over numerous weeks. Finally, themes were named, and relevant quotes were chosen for the report.

2.7 Ethical Considerations

Approval by an Ethics Body

This study was approved by the Human Research Ethics Committee (HREC) of the University of Cape Town – HREC reference 022/2023 (Appendix E: HREC Approval).

Informed Consent

The aim of informed consent is to present a possible participant with the important information about the research study in a language that the participant understands (Manti & Licari, 2018). This is done so the possible participant can make an informed decision on whether they want to participate it the study. The Helsinki declaration places emphasis on consent being given freely after the study's aim, possible benefits and any other important factors have been discussed with the participant (World Medical Association, 2001). The researcher discussed the relevant facts with the possible participant and then gave the information sheet and consent form for the possible participant to read (Appendix B). These forms were either given in-person (printed out), electronically or read out to the participant verbally over the phone if the previous two options were not viable. Additionally, it was explained to them that participation was entirely voluntary and were assured that their decision to partake in the research would not influence their healthcare in any way. The participant was also informed that they could use the contact details on the consent form to withdraw from the study at any time prior to publication of the research without any ramifications. Participants were also informed that they would remain anonymous.

Anonymity and Confidentiality

To protect the participants' interests, the data were anonymised so that the participants are unidentifiable. This was completed by changing individuals' names to participant number (e.g., P1). The primary researcher, Samantha Bayley, was the only individual with access to the list that corresponds participant name to participant number. Only those directly involved in the research (including supervisors and co-supervisors) had access to the raw data collected. All data were stored securely, whereby the digital information was password protected on a personal laptop and stored on a password protected, and encrypted cloud. The hardcopies were stored securely in the postgraduate office in the division under lock and key. After publication or acceptance of this dissertation, the data will be safely discarded.

Risks and Benefits

This research was a low-risk study. The only potential risk was that the participants may become upset talking about their GC session, as these sessions can potentially be emotionally charged depending on the patient's circumstances. To reduce discomfort, participants were assured that they could abstain from any question, and they could withdraw from the study at any point. The participants were also assured that their actions would not affect their current or future medical care. Patients were also reassured that they would be referred as appropriate if it becomes evident during interviews that they were distressed or have other unmet needs that were identified.

There was no direct benefit to the participants themselves, however, a potential benefit was in that qualitative research participants have reported that talking about their experiences can be cathartic and this research may have assisted participants feel as though their opinions and experiences are being heard (Hutchinson, Wilson & Wilson, 1994).

The potential benefit of this research was to improve the GC telephonic AMA service. This helped determine areas that the patients feel were or were not constructive in the telephonic service. From this, changes can be made to the service so that other AMA women can receive an improved service.

2.8 Research Rigor

For qualitative research to be rigorous it must be credible, transferable, dependable and confirmable (Liamputtong, Anderson & Bondas, 2016). Credibility refers to believing the findings to be true. Transferability means that the findings can be applied to a similar context (Liamputtong, Anderson & Bondas, 2016). Dependability refers to if the findings correlate with the data collected or that the same conclusions would be drawn at another time (Liamputtong, Anderson & Bondas, 2016). Confirmability means that the researcher has not allowed their own values to encroach on the data to a high degree (Liamputtong, Anderson & Bondas, 2016). In order to ensure rigor, clear documentation was made throughout the planning, data collection, and analysis (Liamputtong, Anderson & Bondas, 2016). Before the interviews were performed with participants, a test interview was conducted with a colleague to ensure the questions were appropriate. The data analysis also followed a systematic process as detailed above (Liamputtong, Anderson & Bondas, 2016). Additionally, one or both supervisor/s analysed four interviews to ensure that the same themes were reached, and all interviews were discussed with the supervisors. This helped ensure dependability. Confirmability was safeguarded by the researcher establishing her own bias surrounding the telephonic AMA service at the start of this study. The awareness of this bias was shared with the supervisors so that both the researcher and the supervisors could question the researcher's interpretation was not influenced by her own prejudice.

2.9 Researcher Reflection

It was important for the researcher to reflect on her own bias in this research, due to the nature of qualitative research. As a Genetic Counselling Intern, I have spent over a year providing the telephonic AMA GC service and I have provided in-person AMA GC, although less frequently.

This experience has led to a bias of believing that although the telephonic service is convenient for many women, it doesn’t allow for the same level of psychosocial counselling as an in-person session. The previous research done on the in-person AMA GC at GSH showed that even women who declined invasive testing still found the session valuable (Vorster, 2020). Thus, my bias is that I think the in-person GC (IPGC) could be a better service for most women. For the duration of this research, I had to continually question how my bias could be reflecting in the results. This was also discussed with my supervisors and peers to minimise its impact. As the research went on my bias evolved, although I still favoured IPGC, I understood some other benefits of telegenetics. This made it important to continue this reflection and discussion to minimise the impact of new biases on my interpretation of the results.

Chapter 3: Results

This chapter will present the demographic data of the research cohort, as well as the themes that emerged from thematic analysis of the participants' transcripts.

3.1 Cohort Demographics

This research study included nine participants, all between the ages 40 and 44 (Table 1: Participants' Demographic DetailsTable 1). The Down syndrome (DS) age-related risk for these women ranged from 1/98 to 1/43. A total of 18 eligible patients were contacted to be involved in the research but only the nine agreed. Patients that declined involvement in this research, either stated that they did not have the time, or the researcher received no response after several attempts to make contact. All the research interviews were conducted in English, but the home languages of participants included English, isiXhosa, Afrikaans, Sesotho, Setswana, and Chichewa. The educational background included finishing secondary school but not Grade 12, a completed Grade 12 and tertiary education. The participants identified as cisgendered and therefore gendered pronouns will be used throughout. All participants said their religious affiliations were Christianity, except Participant 1 (P1) who described herself as Spiritual.

Table 1: Participants' Demographic Details

Participant ID	Sex	Age	Home Language	Educational Background	Religion	Residence	Employment Status
P1	F	40	Tswana	Tertiary education	Spiritual	Urban	Contract
P2	F	42	English	Tertiary education	Christian	Suburban	Contract
P3	M	40	English	Secondary but didn't finish Grade 12	Christian	Suburban	Full-time
P4	F	42	English & Afrikaans	Secondary but didn't finish Grade 12	Christian	Township	Unemployed
P5	F	41	Afrikaans	Secondary but didn't finish Grade 12	Christian	Rural	Unemployed
P6	F	41	English	Tertiary education	Christian	Suburban	Unemployed
P7	F	44	Sesotho	Grade 12	Christian	Township	Full-time
P8	F	40	Chichewa	Secondary but didn't finish Grade 12	Christian	Rural	Part-time
P9	F	42	Xhosa	Grade 12	Christian	Township	Full-time

P = Participant

Participants were referred to Groote Schuur Hospital (GSH) from six different Midwife Obstetric Units (MOUs) and hospitals and were between 10 to 21 weeks pregnant when they received telephonic Genetic Counselling (GC) (Table 2). All participants except 2 had access to their own cell phone and all participants except P3 had access to either WIFI or mobile data. Three of the participants did not accept invasive testing and the remaining accepted invasive testing in the form of an amniocentesis. Five participants were interviewed in-person, and the rest were interviewed telephonically. All these quantities are presented in Table 2. The interviews lasted between 20 to 90 minutes.

Table 2: Participant's Referral and Pregnancy History

Participant ID	Referred From	Number of Children	Number of Miscarriages/Stillbirths	Gestational Age (weeks)	Access to own Phone	Access to WIFI	Access to Mobile Data	Invasive Testing Accepted	Telephonic or In-Person Interview
P1	Wesfleur MOU	1	0	13	Yes	Yes	Yes	No	Telephonic
P2	Lentegeur	4	3	21	Yes	No	Yes	Yes	In-Person
P3	Lentegeur	4	3	21	No	No	No	Yes	In-Person
P4	Mitchel's Plain District Hospital	2	0	10	Yes	Yes	Yes	No	Telephonic
P5	Mitchells Plain MOU	4	0	17	No	Yes	No	Yes	In-Person
P6	Mowbray MOU	2	3	18	Yes	Yes	Yes	Yes	In-Person
P7	Mowbray MOU	1	1	16	Yes	Yes	Yes	Yes	Telephonic
P8	Mowbray MOU	2	0	20	Yes	Yes	Yes	No	Telephonic
P9	Gugulethu	3	0	14	Yes	No	Yes	Yes	In-Person

P = Participant

3.2 Themes

Four major themes emerged from the data (summarised in Table 3). Theme one revolves around trust, with subthemes including trusting the healthcare system and trusting the genetic counsellor. The second theme concerns decision-making, specifically about having an amniocentesis or not. The subthemes are Factors Influencing Factors Influencing Risk Perception and seeking control. Theme three encapsulated the participant's opinions about the telegenetic service by considering the practical benefits, the emotional connection, and the overall opinions of in-person versus telegenetic GC service. The final theme concerns information gathering and discusses how information was given at the MOU, the GC session and further information seeking via the internet.

Table 3: A Summary of Themes and Subthemes

Theme	Subtheme
Balancing Trust and Distrust	The Genetic Counsellor
	The Healthcare System
Exploring the Dynamics of Choice	Factors Influencing Risk Perception
	Needing More Control
A Need to Balance Practicalities with Preferences	Practical Benefits of Different Service Delivery Models
	Emotional Connection
	Overall Preferences
Unveiling Knowledge Acquisition	MOU and Pamphlet
	GC Session
	"I Googled"

3.2.1 Theme 1: Balancing Trust and Distrust

While interviewing the participants it was noted that many participants had some concerns about the trustworthiness of the genetic counsellor and/or the healthcare system. Not all participants were concerned about the trustworthiness, but a few were very concerned, which made this theme prominent. Initially, trust was not included in the interview question guide, however, after the second interview with P2 and P3, it was clear that this was an important aspect to investigate further. Although, trust was not thought to be a major factor by the researcher initially, it became an overarching theme impacting the participants' experience of the Telephonic GC (TGC) service. The subthemes describe how there were breakdowns in trust either between the participant and the genetic counsellor or between the participant and the greater health care system and staff.

Subtheme 1: The Genetic Counsellor

Trust can depend on a multitude of factors and trusting another person can come more naturally to some individuals. For some participants they viewed being open with the genetic counsellor as a necessity and as a non-issue. This was the case for P4 and P9, who described being open and honest with the genetic counsellor.

> *"I feel free to speak. I mean, I'm carrying a person inside of me, obviously I must be open. I can't hide nothing." - P4*

> *"Yeah, I think... I don't know because I trust that call. I didn't even think that, oh, this one wants to take my information and all that." - P9*

Feeling comfortable can be crucial when gaining trust. P6 mentioned how the genetic counsellor made her feel comfortable and created a safe space to facilitate a discussion. Feeling comfortable allowed for information to be shared freely without concern for judgement or false pretences. P6 shared the following on this:

> *"It was more comfortable. I think my husband cracked a few jokes there about his stress levels and it was, it was very open, it was very nice." - P6*

Other factors that influenced trust and comfort were similarities between the participant and the genetic counsellor. When similarities were noticed and valued then a more open conversation occurred. This was the case for P4, who noted that she felt more comfortable speaking to a woman since she is also a woman.

> *"No, it was fine. It was fine because it was a lady that I was speaking to. You know, female to female." - P4*

Trust was built throughout the GC session by the flow of information and the reactions of the genetic counsellor. P8 described building trust with the genetic counsellor because of the way her concerns were handled.

> *Interviewer: "Did you feel like you could trust the genetic counsellor? That she was telling you the truth?"*
>
> *P8: "I think so. She was telling me the truth."*
>
> *Interviewer: "You think so, okay. Why, why could you trust the genetic counsellor? What made you think that you could trust her?"*
>
> *P8: "Because of the way she was handling my issues."*

During the interviews, concern around trusting the genetic counsellor also depended on that these GC sessions were all telephonic. While most participants seemed to trust the GC, one participant expressed concern. On probing it was found to be mostly related to the phone call itself rather than the genetic counsellor, as according to her, it is easier to be dishonest over the phone. These comments were focused on telephonic conversations being at the heart of distrust rather than the GC trainee.

> *"And so, for me, sometimes, uh, uh, doing it over the phone, I like, you're hiding behind the phone call…Um, like, because everybody, anybody can lie over the phone." - P2*

Additionally, to the distrust in telephonic conversations, having the GC sessions over the phone limited the ways of building trust as the nonverbal expressions could not be observed by the participants. P2 and P4 both described not being able to relax and trust the genetic counsellor over the phone because they could not see their facial expressions and/or body language.

> *"Oh, maybe that person would've made me feel more relaxed, maybe to see my expression on my face, because we don't see each other. We're just talking on the phone." - P4*

> *"Yeah, that's over the phone. Anybody can lie about something. Anybody can make up a story. You can't see that person's facial expression. You can't see that person's body reaction." - P2*

Subtheme 2: The Healthcare System

Although participants generally trusted the GC trainee, there were opinions of distrusting the healthcare system. Participants mentioned feeling as though they weren't being told everything by their healthcare professionals. This idea was based around something being wrong with their pregnancy that they weren't being told about. The comments highlighted below show that P8 thought that there may be more discussions about her pregnancy between the health care professionals and not everything was disclosed to her:

> *P8: "Maybe, like, uh, maybe she (the genetic counsellor) knows something, and I don't. Because maybe the doctors share things, you know… I thought something that I don't know. Maybe my medical doctor didn't want to tell me."*
>
> *Interviewer: "Do you feel like nurses and doctors don't tell you everything?"*

> *P8: "Yes."*

There were thoughts that more details would have been shared with the participant if they had received their healthcare in the private healthcare system compared to the state healthcare system. Suggesting a greater trust in private healthcare compared to state healthcare.

> *P4: "I mean, they could have told me more about like the scan lady, or the sister could have told me this is the procedure you're gonna go through because of your age and blah, blah, blah. But I think it's because it's a local clinic. That's why they don't do it. But I think if I would've gone private, they would've been more specific with me.."*

P4 was also distrusting of the state healthcare system because of the lack of information given to her at her MOU. P4 described thinking there was another reason for her referral and was upset that she was only given more information after contacting the GC service at GSH.

> *P4: "They [health care providers at the MOU] could've told me before the time. I mean, if they would've told me what the procedure is about, then I would've been feeling more relaxed. But now I must phone Groote Schuur for this procedure, they could've told me, I must go for this procedure. But they didn't tell."*

P6 was also upset about her referral process as it was very confusing. She thought an increased nuchal translucency (NT) was detected on a prior ultrasound at her MOU. This concern around the increased NT, which the GC trainee had told her was not the case, caused a lot of confusion and distrust. When P6, was interviewed she was still unsure whether to have an amniocentesis or not because she was unsure what to believe about the risk of DS regarding her pregnancy. P6 wanted to have another ultrasound at GSH to reassure her and confirm risks. She had also been told that the ultrasounds at GSH are better and therefore more trustworthy.

> *P6: "Do they, don't they just maybe do one last check before they do the amnio because what if the amnio is unnecessary? What if there is no thickness in the neck anymore?"*

> *Interviewer: "Okay, I hear you. You're worried that they're going to find something else or, also you've heard all this different information over your course."*
>
> *P6: "Yeah, I'm worried that they're probably going to do it for nothing."*

There was also distrust in what the invasive testing process/ procedure is and the true purpose of this test. This concern shows a greater distrust in the healthcare system as a whole and that the invasive test would cause harm, either intentionally or not.

> *Interviewer: " And you said, um, you thought that you are going to be given a procedure that's going to..."*
>
> *P2: "Yes going to be, um, aggressive harm the baby, um, or harm me, or it's, I'm going to be in pain. That was my thinking of if they do this type of procedure. Um, I didn't know that it's going to be done with a needle. Uh, obviously my mind being, uh, they're going to scratch inside of me and they put something inside me to see or stuff like that."*

Summary of Theme 1

This theme shows that trust can be an issue for people depending on several factors and that some participants distrust a TGC session and others distrust the greater healthcare system. There was distrust present with in-person referrals from the MOU and thus telephonic was not the only factor to cause distrust. Trust was also influenced by multiple factors such as personality, comfortability, gender, and how concerns are addressed. These factors allowed for trusting relationships between most participants and their GC trainee.

3.2.2 Theme 2: Exploring the Dynamics of Choice

The second theme that emerged from the data was Exploring the Dynamics of Choice , which navigates whether to have an amniocentesis or not. All participants were offered an amniocentesis in view of their maternal age to determine whether their fetus has DS or not. All participants except P9 had the option of having the amniocentesis solely for informative reasons even if they would not consider a termination of pregnancy (TOP). During the later period of research recruitment, GSH implemented a new policy regarding invasive prenatal testing due to hospital budget restrictions, therefore, P9 was only offered an amniocentesis if she would consider a TOP for an affected pregnancy. The decision-making theme raised two subthemes: Factors Influencing Risk Perception and Needing More control.

Subtheme 1: Factors Influencing Risk Perception

Participants' risk perception varied and was influenced by several factors. Part of a participant's risk perception was based on if they felt they could believe the risk values they were given. P2 felt that there needed to be more women having DS children, which would mean the risk for her should be higher, as shown in the following statement:

> *"One out of 500, but [that's] not a lot. But um, for me, it's hard to believe that one out of 500 ladies' child comes out Down syndrome. There has to be more." - P2*

This distrust in how frequently a child with DS is born, influenced this participant's decision-making around having an amniocentesis or not. Other participants viewed the risks they were given as comforting and a lower risk, referring to both the miscarriage risk associated with the amniocentesis and the risk of having a child with DS, than they had anticipated. P2 and 6 share how the risk of a miscarriage associated with an amniocentesis at GSH was acceptable to them.

> *"And then I started understanding like one out of 500 people stuff, like every person that comes in that happens to, so that also like now gave me some assurance that, um, it's only one out 500. It's not like it's a higher number. So that was also fine." - P2*

> *"But once you have the conversation, like it's one in a hundred or five hundred, I can't remember what you said, but then you realize, ah, it's not that bad. You have that same (miscarriage) risk, even if you don't have an amnio, basically. I mean, you don't know what happens (if you have an amniocentesis), what's going to happen." - P6*

The amniocentesis procedure associated risks were well understood by P5. Prior to the GC session as she, had an amniocentesis in a previous pregnancy with no complications. This previous experience provided insight about the procedure and assisted with decision-making. She did, however, find the explanation from the GC trainee to be valuable as it provided her with information she did not receive previously.

> *"Because I had an amnio with my previous pregnancy. The one previous pregnancy, I actually knew what is, what is going to happen and... but they didn't tell me anything like [the genetic counsellor] did." - P5*

P6, perceived the risk of her fetus having DS as high because of this supposed increased NT and she based her decision to have an amniocentesis on this. She expressed that she would not have considered invasive testing if it was her age-related risk alone without any markers.

> *"And if nothing else pointed towards doing an amnio, I wouldn't have done it. If it was just my age, I wouldn't because that's, for me, that's textbook." - P6*

A factor that influenced risk perception and one's perceived capability to cope with invasive testing was a participant's support structure. P2 discussed how her support system gave her the confidence to contact the service and decide on the best route for her regarding invasive testing.

> *"And so, at the end of the day, I think those ladies that don't opt for this [invasive testing], I don't think they've got that support structure. Like other ladies gonna have, people that can support them, family members, a counsellor is there, family members where they're at home that supports them. So I've got that structure, so I'm glad that I've got that support at the home, and the support in church. So I've got all that love around me, so, for me, it's like I can make." - P2*

Another influence on risk perception was participants' fear of the invasive testing. P3 discussed a previous negative experience of losing his previous partner and child in a car accident. This experience rooted his current fear about the amniocentesis, which P2 (P3's current partner) wanted.

> *"I just want my baby and my wife to be fine That is it. And that's the only question I have, you know? Seriously, I mean, I wouldn't have taken off work today if it wasn't important for me. I mean, if this was like a normal visit, I would've been fine. I would've gone with, but [it] wouldn't have been so nerve wrecking." - P3*

The fear that influenced the decision-making around invasive testing, also involved fear of having a child with a disability, the emotional and financial implications as well as the stigma that can come with it. P6 discussed the guilt she felt for choosing the amniocentesis but that the fear of not being able to cope along with stigma outweighed that guilt and hence chose to have the invasive test.

> *"But I think it's a personal choice that we have the right to make. Um, but the guilt is there. You know, especially if you're a Christian, it's not kind of easy to just pick the amnio and then decide if anything, if anything is wrong. Are you going to keep it, depending on how serious it is or terminate it. It's sort of like saying, I want this baby, but I want it under conditions. But at the same time, you have to look at your lifestyle, and you have to look at it financially, and you have to look at it, are you emotionally able to cope? And with that, saying that, I am too soft to go through something like that, and always wonder, if my child has Down syndrome, how is society looking at him?" - P6*

Subtheme 2: Needing More Control

Additionally, to the risk perception described above, participants discussed their feelings around seeking some control of their situation and how the sense of control influenced their decision on invasive testing. Many participants expressed wanting to be prepared for what to expect. P4 discussed wanting to have the invasive test at an earlier stage in her pregnancy so that it would not be as much of a shock. For P4, finding out about if the fetus had DS or not earlier in her pregnancy would help her prepare and have control of her situation compared to finding out at a later gestation.

> *P4: "I can tell you now, I will handle it if my baby [has Down Syndrome], I don't say it's gonna happen that, my baby will come out Down Syndrome. I will tell you now, I will handle it, but I knew this procedure just to prevent it. What I've Google is just a prevention like for you to find out before the time if it is so or not."*
>
> *Interviewer: "So when you say it's a prevention, do you mean it would stop the Down Syndrome or do you mean it would pick it up?"*
>
> *P4: "They would pick it up that . Like my, my early stages now, I come by maybe say eight months, and now I find out my baby's Down Syndrome and then it's like a shock to me."*

Knowing what to expect allowed for a sense of control. This was a motivation to have invasive testing was also so that participants could prepare both physically and emotionally.

> *"Let's go for the procedure because then we can prepare ourselves emotionally and physically, we know what to expect at the end of the day." - P2*

Knowing someone with a child that has DS and seeing the impact it has had on their lives was a motivation to wanting to be prepared. This insight motivated P2 to have invasive testing as P2 knew they would not want the same situation as the acquaintance with a child with DS, thus, influencing TOP considerations as well. P2 was motivated to control her situation and prepare for what is to come.

> *"So basically, I could see what she was going through. So that's what also uh, push me to say, let me rather go into the procedure so that I can prepare." - P2*

Participants also made their decision on invasive testing based on how they thought they would cope or handle having a child with DS. P2 described invasive testing being necessary because they did not think they would be able to cope with a child with DS.

> *"And because at the end of the day, I don't wanna, if I say, no, baby born now I get this [Down Syndrome], my baby's not normal now, I don't know what to do, how to handle it." - P2*

Not being able to cope with a child with DS was also expressed by P9, who described not being able to continue working if she had an affected child.

> *"So, if the child is, this child is having a problem, then I can ask her to... Making abortion and all that because to have a child (with Down syndrome) is just gonna make me not to go to work and all that." - P9*

Other participants felt they would accept having a child with DS and wouldn't have invasive testing. P4 described being prepared, by having done her research though the GC session and that she and her husband decided not to have the invasive test due to the miscarriage risk.

> *"It's a rare case that can lead to a miscarriage during this procedure and I told my husband and he said to me, you're not gonna go forward with this, with this procedure. I thought, fine. But I've done my research." - P4*

Belief in the ability of caring for a child with DS was another factor influencing decision-making. P4 described believing she could handle having a child with DS because of her faith and described how her pastor told her that God won't put something in her path that she couldn't handle.

> *"Hy't gesê God sal nie iets op jou pad plaas as jy dit nie kan 'handle' nie." - P4*

> Translation: He (the pastor) said that God won't put something in your path that you cannot handle.

Summary of Theme 2

This theme showed the various factors that influence decision-making especially concerning invasive testing. Participants were influenced by their risk perception which was also influenced by their fears. Participants also wanted to be able to control their situation by preparing themselves or they had varying senses of control based on if they thought they could handle having a child with DS or not.

3.2.3 Theme 3: A Need to Balance Practicalities with Preferences

The third theme that emerged from the interviews, discusses the practical benefits compared to the deeper emotional and content preferences in telephonic (TGC) and in-person GC (IPGC). The practical benefits are the first subtheme, followed by opinions on the emotional connection between the GC and the participant as the second subtheme. The final subtheme describes the overall preferences that participants described between having an IPGC service or a TGC service.

Subtheme 1: Practical Benefits of Different Service Delivery Models

Participants described opinions over practical benefits to both IPGC and TGC sessions. A difference noted by participants between in-person and telephonic is that one requires travel and the other does not. P4 did not mind the idea of needing to travel to GSH and would have travelled for the GC session.

> *Interviewer: "If you were given the option to have a session like this in-person rather than over the phone, would that be something that you would want?"*

> *P4: "I would want that seeing that I'm at home because I don't, I don't mind coming to Groote Schuur. I'll come for the session. I don't mind."*

Conversely, P1 indicated that it would be too hard to travel in and thus preferred the TGC session.

> *P1: "But like if I had to do it in-person, I would have to like drive all the way today to the doctor's office that we had such conversation…I'd rather have it over the phone."*

The preference of a TGC session over an IPGC session was also influenced by the work schedule of the patients. P1 described how an in-person session would have been very difficult due to her working hours.

> *"The thing is in-person it will be difficult cause I work 6 to 6" - P1*

Other participants described that they would have preferred to take time off work to attend in-person. P7 described that she had her GC session while at work, which meant she was disrupted and couldn't speak freely, therefore her work situation led her to prefer the idea of an IPGC session.

> *P7: "Um, no, it was, it [The TGC session] was fine, but then, when she called me, I was still at work and I can't speak, I can't speak or really, I can't concentrate when I'm at work. There are people who are disturbing me."*

Another benefit of an in-person session that P4 mentioned was that she would have felt more comfortable to use both her home languages – English and Afrikaans. She felt the telephonic session limited her discussion to one language.

> *"I feel more, I would feel more comfortable speaking both my languages also." - P4*

Some participants mentioned a disadvantage of the telephonic service for them was that their partner was not involved. P2 mentioned that an in-person session would have allowed her partner to attend the session with her and that he would have other questions that she wouldn't be able to answer.

> *"And my husband would also, would've been present, but to find out more…He's gonna have different types of questions, which I won't be able to answer." - P2*

While there were challenges with the telephonic service, which were dependent on an individual's circumstances, as described above, several participants also discussed the benefits to the TGC session. A benefit of the telephonic service was how quickly participants are contacted after going to their MOU. P2 commented on how quickly she was contacted by the GC service.

> *"Easy, convenient, no hassles. Um, didn't struggle to get in touch with her. Um, didn't take her long to get in touch with me. I didn't wait for a week or two weeks for her to call. So it was actually a good service. Very good service." - P2*

The TGC service was found to be convenient as participants could use WhatsApp to get in contact with the GC and received a prompt response. This surprised P6 who got a response from a member of the GC team at night via WhatsApp.

> *Interviewer: "But you also found it helpful to be able to WhatsApp the genetic counsellor afterwards."*
>
> *P6: "Oh, very much. I didn't expect such a quick reply last night."*

A concern for the telephonic service is that access to a private mobile phone is not a given in South Africa. However, P5 who shares her phone with her son, did not find this impacted her TGC session.

> *P5: "It's actually my phone, but he [her son] uses it a lot."*
>
> *Interviewer: "He uses it a lot. Okay. And... It didn't make it any more difficult trying to have the conversation over the phone?"*
>
> *P5: "No."*

Further benefits of the TGC session were that for the participants that accepted the amniocentesis, there were remarks about how the TGC session allowed them to know what to expect when then came for the invasive testing. Participants 5 and 6 described this benefit.

> *"Because I, because that's when I got the, when I got here like today, I know what to, what to expect." - P5*

> *"...it was more personal, I think, and that is what I liked about it. It was more comfortable. I think my husband cracked a few jokes there about his stress levels and it was, it was very open, it was very nice. Made me feel better about today [invasive testing appointment]." - P6*

Along with the mentioned practical benefits of telephonic sessions, the use of telephonic communication has increased since the coronavirus disease 2019 (COVID-19) pandemic. This normalcy of telephonic communication was mentioned by P6.

> *"We got used to doing everything over the phone these days. Right. So it doesn't really matter if there's a personal touch of face-to-face." - P6*

Subtheme 2: Emotional Connection

The participants had varying experiences of the emotional connection with their GC trainee, and this resulted in varying emotional responses and opinions on the telephonic service. A TGC session did not seem to cause more anxiety for participants which was particularly voiced by P5, who did not find it stressful receiving the information telephonically.

Moreover, P8 indicated that having the GC session telephonically reduced her anxiety. This was based on her beliefs that if she had an in-person session it would mean that something was wrong with her fetus. Therefore, for her, a telephonic session was less threatening.

> *"Maybe because I ended up thinking, if I would've gone [to GSH], maybe there is something that is wrong." - P8*

Most participants described a positive emotional connection with their GC trainee. Participants described their GC trainee being empathetic, friendly, calming, patient and passionate about their work.

> *"It was very empathetic and very, like I was speaking to a friend that was advising me on something." - P6*

> *"When I started talking to [the genetic counsellor], um, her voice calmed me down." - P2*

> *"No, she wasn't like that the Grinch kinda schoolteacher. She was friendly." - P1*

> *"Very patient. Sincerely patient. That's what I picked up. Um, and like I explained, it's not just I'm doing my job, I kind of picked up a passion for what she was doing." - P6*

Despite this, participants also described that the emotional content and connection between themselves and their GC trainee was limited. P1 and P2, although previously describing their GC trainees to be friendly and calming, both stated that the emotional content and connection were limited. P4 and P9 described that an IPGC session would have allowed for more encouragement and a better connection with the GC trainee.

> *"So for me, for me, it wasn't much of an emotional connection. It's just she gave me information, I asked her questions, she answered the questions, finish with the conversation." - P2*

> *"Like there was no emotions. I was just curious." - P1*

> *"Yes. Maybe she could have encouraged me, maybe, and then I could have told my husband. But the thing that you see, but there was no encouragement." - P4*

> *"Uh, I would choose the face to face. The face to face. Though also, you know, in any form, there's nothing, there's nothing wrong with because there's clear questions and all that, but also it makes a difference to see someone who is explaining you something that you're going to go through." - P9*

P4 suggested that these GC sessions should be in-person to improve the emotional connection and emotional understanding.

> *"It was all right, but I think in this process they must have a session actually face-to-face with, with people because you know you in your pregnancy stages, so you are very emotional." - P4*

There were also suggestions of partners being included so that their emotional wellbeing could also be cared for and considered.

> *"It should be something with them [partners] also, you know? They need some counselling and need some what they call it, emotional content." - P3*

In addition to partners' emotional wellbeing as a reason for their involvement; another reason of preference for IPGC with partners was to diminish blame, which would improve the emotional connection between the couple. When partners do not have GC and do not understand, blame can be put on the women for the decision they make.

> *"He didn't get this interview and then he don't know deeper then. Now, he's going to blame me, maybe, if he can say, if he can say, yeah, I took the child out because of this and this. He's not, he's not going to understand me. But, on my side, I do understand you." - P9*

Subtheme 3: Overall Preferences

Overall, the participants expressed different preferences regarding having a telephonic or IPGC session. P5 described enjoying the telephonic more as she could limit the amount of conversation as she does not like talking a lot:

> *"The one thing I don't like, I don't like talking too much." - P5*

P5 also described feeling more comfortable to ask all her questions over the phone and that she otherwise would not have asked if it was an IPGC session.

> *Interviewer: "What did you like about it being telephonic?"*
>
> *P5: "Then I can ask whatever I want to ask, I can, I can say what I want to say. Because I don't think I would have asked all the questions if it were face to face. I would feel*

> *fine (having an in-person session), but... I would have been evasive or something. I wouldn't have answered everything I did on the phone."*

Contrastingly, other participants felt the telephonic session limited what questions they could ask. They felt they would have asked more questions if they had an in-person session. An in-person session would have also allowed them to feel more comfortable.

> *"Phones actually does limit everybody. It does limit you. To you, uh, Um, you do, you can't ask the questions that you do, ask the questions that you want to ask, but not the real question that you really wanna ask." - P2*

> *"Maybe I would've asked more questions besides scan. I would've asked more questions. You know sometimes it's better to talk with the person face to face as over the phone." - P4*

> *"Then I could've asked more, like, then I would've feel more comfortable actually." - P4*

The use as visual aids was mentioned by some of the participants to be the benefit of an IPGC over telephonic. They expressed that it would have improved their understanding and accepting the information.

> *"Um, I think it should also have shown me statistics in the numbers, actual numbers, because it is also, it also would've been more reassuring for me 'cause I would see it instead of hearing it." - P2*

> *"I can ask her questions, yeah, understand everything, maybe she can show me some pictures, or videos, something like that." - P7*

Other participants didn't have a preference between telephonic and in-person. These participants felt the session would have been the same either way and would not have a particular preference if they had to choose.

> *Interviewer: "So if you were given the option to have the conversation with the genetic counsellor in-person, would you have wanted that or did you like that it was on the phone?"*
>
> *P8: "Anything."*

> *Interviewer: "And you don't feel it would've made any difference having the same conversation in-person?"*
>
> *P1: "No, no difference."*

P2 and P3, who are spouses, had different preferences to each other. P2 (wife) preferred in-person whereas P3 (husband) preferred telephonic.

> *P3: "Would've been more stressful in-person."*
>
> *Interviewer: "More stressful in-person?"*
>
> *P2: "For me, it was more stressful over the phone."*

Summary of Theme 3

This theme showed that there was not a clear consensus on what participants preferred between IPGC and TGC for AMA. There were practical benefits to TGC sessions such as not needing to travel but there were also practical benefits to an in-person session such as having a focused space without distractions. The emotional connection was noted to be present by some but not by all or to a sufficient extent with the majority of participants thinking there would be a better connection in-person. The importance of having an emotional connection also varied between participants.

3.2.4 Theme 4: Unveiling Knowledge Acquisition

The final theme emerged when discussing where participants obtained their information from. The main information sources were from their MOU (including the pamphlet given to some of the participants), the GC trainee, and online resources. A primary aim of GC is to explain complex information, therefore, determining the usefulness of different information sources reflects their experience of the GC session. The focus on learning and information seeking also depended on the participant's personality.

> *"Yes. I like to learn new stuff every time. I'm such a person." - P4*

> *"Now, I can learn and the next person, maybe that person can ask me, what is this? Then I can explain to them. I didn't go maybe for it [the amniocentesis], but the this is this, I learn, like to learn for the next person." - P4*

Subtheme 1: MOU and Pamphlet

At the MOUs participants were told about the GC service in several ways. These ways included a one-on-one conversation, a group announcement or by being given the pamphlet. A few participants described having a very limited discussion or not receiving enough information from their MOU.

> *"Uh, (they) told me in passing, you know, as they do their information." - P1*

> *"She didn't explain to me, she just gave the pamphlet and said, I must read through it and there's the number of Groot Schuur there and I must just WhatsApp them and they would explain further." - P2*

> *"It was a five-minute conversation." - P6*

> *"Um, just that they didn't go into detail about it. And they'd rather prefer that, the genetic counsellor talk to me about it instead of them telling me, because I think some of them, or like the nurses, I don't think they know much about the procedure or they, um, so they just said that once I talk to the genetic counsellor, she will go more into detail about it. And I don't think they would've been able to answer the questions that I asked the genetic counsellor over the phone." - P2*

P9 explained that she was told about the GC service in a group setting with other AMA women. P9 shared that she wasn't given a space to ask any questions at the MOU.

> *P9: "Yeah, we were all together."*
>
> *Interviewer: "All together. Okay. And did were you able to ask questions or anything like that from her?"*
>
> *P9: "Uh, no. They didn't give us chance. Didn't give, and they didn't explain for us."*

The core reason for the referral or the option to contact the GC service was understood by some participants while others were unsure why they were referred or had different expectations for the service. P1 knew the referral was about her age-related risk of having a child with DS.

> *"They told me that usually women at my age, at 40 plus, they tend to have a high risk in kids not developing well. They would, they had come, they become disabled or Down syndrome." - P1*

P5 also understood about the age-related risk of DS, however, she thought she had to have the amniocentesis from her referral rather than it being her choice.

> *"They only told me because, just because if I, just because I'm over 40, I have to do an amnio." - P5*

P4 was expecting to get an ultrasound from GSH and wasn't expecting the AMA GC service.

> *"I didn't expect that because I knew they're gonna send me to Groote Schuur, but I didn't know they're gonna send me for this. I thought this maybe just for a scan." - P4*

Some participants' risk perception was also altered by the information they received by their MOU. P6 stated that the miscarriage risk associated with the amniocentesis was lower than what she thought it would be before being given information at her MOU.

> *"...explaining about the risks of the amnio and what you think the risks are and what it actually is, is really not that bad. I mean, the perception that I'm having an amnio is you're going to have it and you're going to have a miscarriage." - P6*

The referral process from the MOU changed during the recruitment process in September 2023 which was after P6. Originally the MOU gave basic information, which may or may not have included a pamphlet, and then the GC service's number was given to the participant. The participant was given the option to contact GSH for more information or not. Following a very low service uptake, and incomplete information received from the patients, causing delays, the service changed to patients being referred directly by their MOU via the VULA application, which is a mobile device application that allows for online referrals to be made between different MOUs/Hospitals and different services. This new referral system aimed to streamline the service, ensure accurate information was received about the patients' gestation and other factors and assist with potentially more patients being reached. While it was only meant to replace the fact that women had to message themselves, it also often meant that women were not given the option to have their information given to the GC service and therefore they received GC whether they wanted it or not. P7 was not given the option but if she was, she would have wanted the GC service.

> *Interviewer: "Was it an option for them to give your number? Or did you just, or was that just what they said they were going to do?"*
>
> *P7: "No, they told me that they were going to do it themselves."*
>
> *Interviewer: "Okay, so it wasn't really an option?"*
>
> *P7: "No."*
>
> *Interviewer: "How did you feel about having to have this genetic counselling service?"*
>
> *P7: I think that it was fine. It was alright that I can know everything.*
>
> *Interviewer: "And if it was an option for you to not have had this genetic counselling service, would you have chosen not to have it?"*
>
> *P7: "No, I was still going to do it."*

Other patients also voiced that they would have preferred more information about the referral. P4 recommended that more information be given by the MOU, especially about the invasive testing options.

> *"They can tell the people what is this procedure. Can have sessions where they can tell them it about these procedures. Why must you go to Groote Schuur or WhatsApp? Be more open about it. Because it's not, not always at my age that get thousands of babies." - P4*

The pamphlet was designed by the GC service and four of the nine participants received it from their MOU. There were differing views on the content of the information pamphlet from those that received it. Some, like P5 and P9, thought it to be useful as it prepared them for what to expect.

> *"It was helpful. And it gave me a little bit information what is going to happen and what Down syndrome is and stuff like that." - P5*

> *"Yes. Yes, when I read the page and then I decided, yeah, I must [contact the GC service]... So that I can get more information. Maybe I understand a bit, then I need someone who's gonna explain for me." - P9*

However, P3 found the images on the pamphlet stressful. P2 found the pamphlet more useful as it answered some of her questions.

> *P3: "It made me nervous."*
>
> *Interviewer: "And for you (P2)?"*
>
> *P2: "Um, not really."*
>
> *Interviewer: "You found the pamphlet fine?"*
>
> *P2: "Yes. It answered some of my questions and when I spoke to the counsellor, she answered the rest of my questions, the rest of them.*
>
> *Interviewer: "What about the pamphlet made you (P3) nervous?"*
>
> *P3: "The needle into the stomach."*

P5 mentioned that the pamphlet was too long and that she only read what she thought was important.

> *P5: "I didn't read the whole thing."*
>
> *Interviewer: "Why didn't you read the whole thing? Was it too long? Was it too long?."*

> *P5: "It was too long. I only, I only read the important [parts]."*

Subtheme 2: GC Session

The main source of information regarding DS and invasive testing was provided to the participants via the GC trainees who tele-counselled them. Most participants were not entirely sure what to expect from this service as demonstrated in the subtheme above. However, P6 had a better understanding of the service and the reason for her referral compared to the other participants, even though she did not receive a pamphlet at her MOU.

> *"Well, I just, I took it as I had something to do with it. Mine and his background, our genetics, our DNA, any complications, or anybody with any defects in our family. So I, I kind of got there, but I thought it was just a conversation about educating us on it and then maybe taking a step further and then seeing what's going on." - P6*

Along with a more accurate expectation for the service, P6 was satisfied with how the information was explained to her by the GC trainee.

> *"I was happy with it. I mean, not happy in the sense about the amnio, but happy in the sense of the information that I received and the manner it was given to me." - P6*

Other participants were also happy with the service, as they learnt new information. This learning seemed to be very valuable to a few participants.

> *"Um, it was a relief for me to know stuff. She told me stuff I didn't know about. It was a relief for me." - P5*

P7 felt that the GC session would be valuable for AMA women as she learnt new information from her session.

> *Interviewer: "Do you think other women might choose not to have that conversation with the genetic counsellor?"*
>
> *P7: "No...I think they should."*
>
> *Interviewer: "You think they should?"*
>
> *P7: "Yeah."*
>
> *Interviewer: "Why do you think they should have it?"*
>
> *P7: "Because there's a lot that you don't know."*

P4 was shocked by the new information on what causes DS, but still found value in the education she received.

> *"It was actually quite shocking 'cause I never knew about this stuff, so. I thought to myself, I thought, I never wondered where, where does Down Syndrome come from, but now I see all this." - P4*

P5 also discussed how the GC session was helpful as it explained the different options available to her.

> *"She [the genetic counsellor] told me, like the information she gave me about Down Syndrome, what it is, and what my options are. I can keep the baby or I can let the baby go." - P5*

Subtheme 3: "I Googled"

Before or after the TGC service, participants may or may not have gone to the internet to find out more information. Some participants did search the internet and found the information online helpful. These participants either wanted more information or wanted to clarify the information that was given by the genetic counsellor.

> *"No, I was still curious, wanting to find more, because after speaking with [the genetic counsellor], I did go to Google to find out that more than what she said." - P1*

> *"She [the GC trainee] was talking about it. She told me what is gonna happen, but I wasn't like, you know, not actually taking note, but I wasn't like familiar. That's why I went to go Google it." - P4*

Not all experiences of googling information were viewed as beneficial. Some participants felt that when someone is explaining information to you, it is understandable and more personalised. Hence P2 researched online minimally.

> *"When you do go on Google, it's almost like you get more confused. So then I would rather do face to face and ask you physically, ask doctor. Because I know you guys [Genetic Counselling service] can provide and you can break it down for, that's why I don't use Google that much because they don't give you the information that you really want to know or want to see." - P2*

> *"You know, it's easy to just type it in Google, but it's not the same thing like actually getting it from somebody." - P6*

A difference between online resources and the TGC session is the use of visual aids. P6 felt that the visual aids are helpful, however, whether these visual aids were shown to her by the GC trainee or through her own Googling, it wouldn't have made a difference.

> *"I think whether I watched it on my own or genetic counsellor showed me, I think it would have been the same." - P6*

P4 shared that visual aids are helpful but that she would have wanted them in her GC session so that she wouldn't have to search the internet for them afterwards.

> *Interviewer: "Do you think it would've been helpful to have photos or images in the session that helped explain different things?"*
>
> *P4: "Maybe that would've helped, yeah. I mean, now I have to go Google myself afterwards."*

P1 commented that the photos she found through her own internet search were traumatising and that just talking would have been better.

> *"Photos are traumatising, it's okay just talking. That's why I say after talking to her, I Googled and she, yeah, the possibilities are quite depressive." - P1*

Although for P1 the images online were not helpful, for others visual aids helped in their understanding of the information.

> *"And I watch the video, what they do. It was a scan to see where the baby's laying and then pull fluid from, through your navel, from the water that the baby's laying in and stuff." - P4*

Not all participants felt the need to look for more information online because the information provided in the genetic counselling session was sufficient for them.

> *Interviewer: "Didn't go Google?*
>
> *P9: "I didn't."*
>
> *Interviewer: "You felt fine just hearing from the genetic counsellor what she explained?"*

> *P9: "Yeah."*

> *"Um, but I didn't want to get too much information where it could stress me out. So, but at the same time, I think she's [the genetic counsellor] given us all the information that we needed. I'm sure if I had to Google something now to give me exactly the same answer that she gave us." - P6*

This statement by P6 also showed her trust in the GC trainee to provide all necessary information to her. This again shows how trust was pertinent throughout multiple aspects of this research.

Summary of Theme 4

This theme showed that participants were referred in various ways and not everyone was given the pamphlet. The information given by the GC trainee was helpful but the expectations for the service were not always accurate. Searching the internet was useful for many participants especially because of visual aids. However, visual aids were not considered helpful by all participants but rather harmful by some. Overall different sources of information were used by all participants and the usefulness varied depending on personal preferences.

Chapter 4: Discussion and Conclusion

This research study was conducted with the aim of answering this research question: What are the opinions of advanced maternal age (AMA) patients on the telegenetic service provided to them through Groote Schuur Hospital (GSH)?

A primary goal of GC is decision-making. To facilitate this goal of decision-making there are tools that facilitate this process. These tools include providing options and their outcomes, personalize this information, allow for an understanding that the patient can participate in the decision-making, acknowledge the scientific uncertainties, identify the potential benefits and harms, aid patients' discussion concerning their values and improve the skills for collaborative decision-making (Uhlmann, Wendy R, Jane L Schuette, & Beverly M Yashar, 2009). These tools can be used in multiple service delivery models, including telephonic. They aid a GC session in multiple ways, importantly they allow for patient satisfaction with GC and do not increase anxiety (Uhlmann, Wendy R, Jane L Schuette, & Beverly M Yashar, 2009). Therefore, to assist patients' having a positive experience of GC, appropriate decision-making, with the use of the tools mentioned, should be done. Decision-making and ultimately participants' experience of the GC service provided to them by GSH will be explored in this chapter.

Telegenetics has been available for many years and has increased significantly since COVID-19 (Gorrie et al., 2021a). In the past, especially in the midst of COVID-19, telegenetics was viewed positively by patients and genetic counsellors (Buchanan et al., 2015; Zierhut et al., 2018; Dratch et al., 2021; Gorrie et al., 2021a). This research explored the opinions of AMA participants who had a telegenetic service provided to them through GSH. This is based post COVID-19, when social distancing is not a requirement in South Africa. Both positive and negative experience were described by participants in this research. The reason for these opinions will be explored further in this chapter, discussing trust, decision-making, information gathering and psychosocial factors. Overall, opinions of the telephonic GC session alone, not including referrals, were positive and allowed for informed decision-making and satisfaction with the service.

As described in Chapter 3, multiple themes emerged from the interviews that convey a variety of opinions on the telegenetic service at GSH. The four themes described were ‘Balancing Trust and Distrust’, ‘Exploring the Dynamics of Choice’, ‘ A Need to Balance Practicalities with Preferences’ and ‘Unveiling Knowledge Acquisition’. While these themes describe distinct opinions, they also collectively describe the experience and opinions of the GSH AMA telecounselling service. In this chapter, a rounded approach to investigate how these themes answered the research question, objectives and corresponded with existing data. Finally, the limitations, strengths and implications of this study will be discussed.

Trust and Its Influence on Genetic Counselling

For information to be given effectively a level of trust needs to be achieved between the Genetic Counsellor and the patient (Green et al., 2023). Trust was an important theme that emerged from the interviews, this included trust between the GC trainee and patient as well as the trust between the nurses and the patient at their Midwife Obstetric Units (MOUs). The trust between GC trainee and participant was generally described to be good. This corresponds with the basis of a genetic counsellor-patient relationship, which is described to be based in trust, openness, and respect (Evans et al., 2004; Anderson, 2015). In previous literature there have been varying reports around telegenetics’ impact on trust. In two studies investigating Huntington Disease GC and cancer GC, it was found that there was no difference in trust between telegenetics and in-person GC (IPGC) (Sutphen et al., 2010; Hawkins et al., 2013; Danylchuk et al., 2021). From the genetic counsellor’s perspective, in a study by Badje (2021), a genetic counsellor noted that she felt patients still trusted the genetic counsellor in telegenetics to the same degree as with IPGC (Badje, 2021). The general trust between participant and GC trainee in this research, was attributed to the participant’s personality, feeling more comfortable with the GC trainee, being of the same gender, and competency. There is an agreement with past literature of professional competency being important in trust building in a nursing setting (Ozaras & Abaan, 2018). While Ozaras and Abaan did not look at patient factors, other papers have mentioned these to be known factors that affect the clinician-patient trust relationship (Dinç & Gastmans, 2013; Sheppard et al., 2013).

While all the participants seemed to trust the GC trainee, the main exception was P2 who described the importance of facial expressions in determining if someone is being truthful. In previous literature in cancer telegenetics, a similar level of distrust regarding the information given, due to a GC session being telegenetic, was described (Zilliacus et al., 2010). A lower level of trust and rapport in telegenetics compared to IPGC was also seen in a more recent study looking at patients with a high risk for a *BRCA1/2* pathogenic variant (Peshkin et al., 2016; Danylchuk et al., 2021). Therefore, additional time and effort towards rapport building in a telegenetic session may be beneficial. Other reasons for distrusting telegenetics have been due to concerns about privacy, confidentiality and doubting the efficacy of telegenetics (Green et al., 2023).

The importance of facial expressions mentioned by P2, was similarly discussed by Ozaras & Abaan's research, which described graciousness as being important (2018). Specifically under graciousness, being ready to smile was the important factor in building trust (Ozaras & Abaan, 2018). This shows the importance of non-verbal communication in building trust and how a telephonic session can lack this aspect. It can also be said that medical mistrust will influence if a patient engages with GC or not (Sheppard et al., 2013). This is an important consideration for the AMA women that did not contact the GSH GC service.

Although, the referral process has become more non-optional now, this may introduce more patients that mistrust their GC trainee due to medical mistrust. This, however, was not shown in this research but the sample size post change in policy is a limitation in this research. The main feelings of mistrust, that was identified in this research, were centred around participants' experiences at their MOUs or at the greater healthcare system. Partiticpants commented on feeling they were not given all the information from their MOU, that the Private Healthcare System would have given them more information, conflicting information made it difficult to know what to believe and that healthcare professionals can cause harm, wheather intentionally or not. The concerns about not being given enough information related to the professional incapability, and communication problems discussed by Ozaras & Abaan (2018). Participants commented that they felt the nurses wouldn't have been able to answer their questions, which related to the professional incapability resulting in mistrust, described by Ozaras & Abaan (2018).

Trust in the private healthcare system over the state healthcare system was an important contributing factor to the mistrust some of the patients in this study had. In South Africa the healthcare system comprises of the state and private services. In the state system women receive free prenatal healthcare, whereas private is paid for either by the patient or through a medical aid (Morris et al., 2015; Statistics South Africa, 2011b). Only 24.3% of the population utilises private healthcare compared to 61.2% using state healthcare (Morris et al., 2015; Statistics South Africa, 2011b). Therefore, private healthcare should be less overburdened and healthcare professions should have more time to discuss information with patients. This would explain why a greater trust in the private healthcare system was mentioned and how it links to professional incapability, since little time can result in reduced efficiency of patient communication (Ozaras & Abaan, 2018). The state system is also divided into different levels of care at the clinics and hospitals that have varying levels of resources and trained professionals. Thus, the greater trust in healthcare professionals and their resources, specifically ultrasound machines at a tertiary level hospital, such as GSH, is linked to the professional capability as well (Ozaras & Abaan, 2018). This is why P6 wanted a final ultrasound at GSH before deciding on the amniocentesis. A distrust to the point of thinking healthcare professionals would likely cause harm, even if unintentionally, is quite considerable and was only mentioned by P2. Trust building is a process that can involve trust being broken and re-established, however, various factors may hinder the trusting relationship (Dinç & Gastmans, 2013). Trust between the patient and the GC trainee is important as it allows for open communication and expressing one's emotions. It is also important in the patient being able to believe the information the GC trainee provided, which in turn influences decision-making.

Decision-Making and The Emotional Experience of Genetic Counselling

Decision-making is a complex process that is central to GC that involves risk communication and risk perception (Weil, 2000; Wessels & Koole, 2019). Overall participants in this research trusted their GC trainee, which allowed for open communication. The exception to this trusting relationship was shared by P2, who expressed questioning the GC trainee as P2 felt that anyone can lie over the phone. This doubt around the information shared with P2 influenced her risk perception, as she did not trust the incidence of DS values provided by the GC trainee. In another study investigating the influence of various knowledge sources on a woman's decision regarding amniocentesis, their view on knowledge sources was greater than or equal to the influence of their personal abortion views (Markens S, Browner CH, & Preloran HM, 2010). This study also found that the knowledge from 'expert' and 'lay' sources is in a complicated and interlinked relationship that is not straightforward or one-sided (Markens S, Browner CH, & Preloran HM, 2010). Therefore, trusting the GC trainee is important but not the only factor in risk perception. P2 in this study, knew an acquaintance with a child with DS, which was her frame of reference for risks surrounding DS. This prior frame of reference is described as an anchor, which limits the extent that new information is judged (Tversky & Kahneman, 1974; Weil, 2000). This anchor would have influenced P2's risk perception.

For P3 a more personal experience of pain and loss influenced his decision-making and his risk perception. P3 described wanting his wife and child to safe since he had already lost a previous partner and child in a car accident. This traumatic experience influenced P3's risk perception and fixed his focus on an unharmed family. This again relates to anchoring (Weil, 2000). These past experiences or frames of reference are common considerations in GC along with many more, such as a patient's view on the seriousness of the disorder and beliefs about the aetiology (Uhlmann, Wendy R, Jane L Schuette, & Beverly M Yashar, 2009). The factors described by the participants focused on seeking control. In prenatal GC, depending on where you live and the Termination of Pregnancy (TOP) legislation, prenatal testing and TOP options allow for patients to decide what they want to do. In this a sense of control can be obtained. The Genetic Counsellor must encourage the patient to make active decisions that align with their needs and values (Stoll & Jackson, 2020).

Along with anchoring, availability and representativeness are the three major cognitive processes that impact risk perception and decision-making (Tversky & Kahneman, 1974; Weil, 2000). Availability describes how easily examples of potential conclusions are brought to mind, while representativeness describes when individuals use characteristics inappropriately from a small sample size group to predict the outcomes in a larger group (Tversky & Kahneman, 1974; Weil, 2000). An example of the process of availability was shown P2 who's friend has a child with DS. This personal experience was a significant motivator for P2 to have the amniocentesis as P2 felt she would need to prepare for that outcome.

As shown in Chapter 3, some participants found the miscarriage risk associated with the amniocentesis, as reassuring and lower than expected. Other participants considered the miscarriage risk to be too high. Regardless of a woman's view on the miscarriage risk, this risk was often discussed as part of the decision-making process. In a study investigating decision-making for prenatal genetic testing, it was found that worry was the most important factor influencing their decision (Kenen et al., 2000a). This worry was based on the fear of invasive testing and worry about the health of their fetus (Kenen et al., 2000a). Worrying about the health of their fetus was described by P3 and P4, even though P3 (along with his partner, P2) had the amniocentesis and P4 chose not to. Other studies investigating the factors influencing prenatal invasive testing have identified that a woman's priori opinion has the largest impact on her decision (Vergani et al., 2002; Grinshpun-Cohen et al., 2015). The priori opinion is similar to anchoring (Tversky & Kahneman, 1974). In Grinshpun-Cohen et al.'s study, psychological factors that were considered to influence a woman's priori opinion were age-related-risk perception, pregnancy termination acceptability, and amniocentesis risk tolerance (Grinshpun-Cohen et al., 2015). Vergani et al.'s study found that woman with a lower age-related risk and multiple previous miscarriages are significantly less likely to have an original opinion in favour of invasive testing (Vergani et al., 2002). However, in this study P2, P3, P6 and P7 were the only participants to have had one or more previous miscarriages and they all accepted invasive testing. Previous miscarriages also relates to anchoring and representativeness (Tversky & Kahneman, 1974). But as in the current study, miscarriage history does not represent the only factor in the decision-making process as shown in this study.

In previous literature, having support from one's partner, family or friends allowed for more freedom in a woman's decision-making as she was aware that no matter her decision, she would be supported (LeRoy, Veach & Callanan, 2020). This support structure, allowing freedom of decision-making was described by P2 , who discussed her support structure allowing her the freedom to contact the GC service and have invasive testing. In addition, other participants in this research also discussed how other people, such as their partners, were involved in their decision-making process. It can be seen that in pregnancies, a partner's perceived support, can lower levels of anxiety and depression for a couple (Darwiche et al., 2019). Societal opinions and stigma can also influence decision-making, as described by P6 in this research. Mothers of children with DS have been reported to be aware of stigmatizations and try to protect their child from it (Deakin & Jahoda, 2020). Cultural influences can influence your view of stigma and its level of importance in your decision-making (Li, Chandrasekharan & Allyse, 2017). In South Africa, Ali et al (2015) reported that experiences of stigma have been reported by adults with intellectual disability (ID) , with the Black African ethnic group being more likely to be physically attacked and stared at compared to the mixed and white ethnic individuals with ID (Ali et al., 2015). Therefore, stigma is a reality for individuals with intellectual disability in South Africa and can play a role when deciding on prenatal invasive testing and TOP, as it did for P6. However, for South African mothers of children with DS, when asked about their opinions on invasive testing, they were for others being able to choose invasive testing in order to prepare for having a child with DS. When the same group was asked their opinion on termination of affected pregnancy, they were all uncomfortable with this due to their own experience with DS, religious, moral and ethical beliefs (Scott, Futter & Wonkam, 2013). Thus, experiences of stigmatisation was not described as a factor in decision-making regarding invasive testing or TOP for this group (Scott, Futter & Wonkam, 2013).

Complex emotions are involved in the decision-making process with regards to invasive prenatal testing. Psychosocial aspects are an integral part of GC and need to be addressed (Resta et al., 2006). From the participants' perspective in this study, some participants were happy with the emotional connection they felt with their GC trainee. Feelings of being comforted and the counsellor being empathetic, patient, and friendly were described. Participants also mentioned that having the session telephonically did not make it more stressful. Instead, it was calming for P8 in the study as it indicated that there were no issues with the fetus, as her perception was that if it was in-person there would be something wrong. Like with some of the participants of this study, patient satisfaction with telegenetics has been reported to be high regarding psychosocial outcomes in previous studies (Buchanan, Rahm &

Williams, 2016; Danylchuk et al., 2021). Both studies by Buchanan et al. and Danylchuk et al. showed that patient satisfaction was high with telegenetics (Buchanan, Rahm & Williams, 2016; Danylchuk et al., 2021).

Telegenetics has previously been found to cause a barrier with regards to emotional connection between counsellor and patient with previous studies (Zilliacus et al., 2010; Gorrie et al., 2021b). Similarly, some participants in the current study expressed that there was no emotional connection or not enough. In this study lack of emotional connection was described from two different perspectives; positively as it allowed for more focus on the information and negatively for those that would have preferred more psychosocial counselling. This lack of psychosocial counselling has been reported as a concern from the Genetic Counsellor's perspective as well, with concerns about not consistently detecting body language cues even with video calls, thus limiting psychosocial counselling and GC in general (Badje, 2021). There have been mixed reviews on the effectiveness of psychosocial support through telegenetics previously (Zilliacus et al., 2010; Hilgart et al., 2012; Otten et al., 2016; Badje, 2021). The studies by Zilliacus et al. (2010) and Hilgart et al. (2012) recommend telegenetics, both clinical and GC, when the need for psychosocial support is low (Zilliacus et al., 2010; Hilgart et al., 2012). However, in the case of an AMA referral at GSH it would be difficult to differentiate this in a standard referral as no details regarding the emotional state of the patient are given. This recommendation is easier to implement in cancer genetics when a known recent diagnosis has been made (Zilliacus et al., 2010). However, in prenatal AMA counselling, it is difficult to determine who would require more psychosocial support and who would not, as stated in this research some of the participants prefer the telecounselling, whereas others prefer IPGC because women tend to be more emotional during pregnancy. Linking to how telegenetics might be more appropriate to those who require less psychosocial support (Zilliacus et al., 2010).

Some participants suggested that the telegenetic service should include the partners in the GC. P3, husband to P2, mentioned how partners should be involved because they also need the psychosocial counselling. There is limited research on the paternal perspective in prenatal GC (Browner & Preloran, 1999; Kenen et al., 2000b; Lafans, Veach & LeRoy, 2003). In a study looking at telephonic delivery of prenatal DS result, many women were not with their partners when receiving the result call (Lou et al., 2019). The result caused grief and a need for comfort and support, which was lacking since their partner was not present (Lou et al., 2019). Another perspective from P9, was that because the father had not received GC and therefore did not understand the situation as well as her, who had been counselled, he would blame her if she chose to terminate the pregnancy. In this case, GC for the father may have reduced blame towards P9. In Lou *et al.'s* study, the women felt that being responsible to tell their partner the result was difficult as they would be delivering bad news. Partners also had different questions that the woman could not answer, or she had forgotten the information provided to her because of the stress (Lou et al., 2019). Delivering results to partners can be difficult especially when understandings differ and decisions regarding prenatal testing and/or TOP vary. Therefore, when scheduling a GC call, as seen by Lou et al (2019) and this current study, it can be beneficial to arrange or recommend that the partner be present, if the woman would want that.

The Transfer of Information in The Telephonic and In-Person Approaches

The reason for why the GC session is needed is vital when communicating and referring patients. Participants shared their referral process, and it showed many differences. At some MOUs the participants were informed about AMA GC individually while others were informed in a group setting, however, regardless of how they received the information there was still some misinformation and inconsistencies on the depth of information provided by the nursing staff. These differences in the information provided by the MOUs were also noted in previous research done on GSH's (in-person) AMA GC service (Vorster, 2020). Vorster (2020) found that patients lacked an awareness of what to expect from GC as this wasn't clearly explained at their MOU. Even in other settings, referrals from General Practitioners to GC services vary with the extent of information shared with the patient on what to expect from the service (Aalfs et al., 2003). Meaning that depending on the level of information shared, this will influence the patient's expectations of the GC service. This is vitally important to consider when a GC service is optional for the patient to contact as they may not know what they are declining. This was the concern with the previously designed AMA referral process.

The lack of detailed information provided at the MOUs instilled doubt surrounding the healthcare workers' capabilities in P2. This in turn can influence the patient's trust towards these workers and distrust was a major motif for P2. This was described in Trust and Its Influence on Genetic Counselling above where Ozaras & Abaan (2018) noted a similar phenomenon regarding trusting nurse practitioners (Ozaras & Abaan, 2018). Considering only certain tertiary level hospitals in the state system have Genetic Counsellors/ GC trainees; GC on AMA can only be done at these institutions. The referral system still needs to be in place; however, this process results in limited information being shared at the MOU level. A difficult balance needs to be reached where enough information is given so that the patient is aware of why the referral is made but not too much information as it isn't the duty of nurses to provide GC. In the current study, participants described wanting the GC session at their MOUs rather than being referred to GSH and having a delayed process. This want was also described in Vorster's (2020) research and that this unmet want translated into frustration (Vorster, 2020). Considering this was described by this research and Vorster (2020), it is a future recommendation to implement GC services at the MOUs. Further reason for this recommendation, Vorster's study (2020) also found that the reason for referral from MOUs to GC services were not always accurately explained. Many participants in Vorster's research described thinking that the reason for referral was for an ultrasound (Vorster, 2020). Additionally, in this study, P6 mentioned that their risk perception on the miscarriage risk for an invasive test was altered by the information given at their MOU, which caused a lot of unnecessary anxiety. This shows that accurate information regarding the referral is vital to mitigate this effect.

In addition to anxiety caused by incomplete or inaccurate referral information, anxiety was also described by P3 in relation to the images included on the pamphlet given at the MOUs. This pamphlet was designed by the Clinical Genetics Team during Coronavirus Disease 2019 (COVID-19) pandemic to be given at the MOUs to assist with information giving. Opinions on the pamphlet varied among participants that received it. P2 described that her husband, P3, was very stressed when looking at the diagram of the amniocentesis. P5's complaint about the pamphlet was that it was too long and that she only read the important parts, but she did not clarify which sections these were. In a previous study looking at what was the most important information to share about DS, in the prenatal setting, the variable range of intellectual disability, hypotonia, providing printed information sheets and others were all noted to be highly important (Sheets et al., 2011). Therefore, written information sheets are important. In keeping with Sheets et al, (2011) this study also found that some participants felt the pamphlet was helpful and gave them new information on DS. Although this information was useful, P9 still wanted a person to explain everything fully to her, thus, reinforcing the value of GC.

The practical benefits of the telegenetic service were also discussed in this research, in 3.2.3 Theme 3: A Need to Balance Practicalities with Preferences. These practicalities of decreased financial strain, timeous service, continuation of care, and decreased need for transportation described in Chapter 3, were also described by Mahtta *et al.* (2021) and Mahmoud, Jaramillo & Barteit (2022). These specific practicalities along with increased accessibility to healthcare, and saving resources were emphasised as important in low to middle income countries (Mahmoud, Jaramillo & Barteit, 2022). Additional advantages of telehealth services, which were not described by this research, included: cost-effectiveness, improved health outcomes and reduction in provider-patient supply-demand mismatch in some specialities (Mahtta et al., 2021). P6 commented on the commonness of telephone communication, especially since the COVID-19 pandemic and how that resulted in a positive view of the telephonic GC (TGC) session. This result ties well with a previous study wherein, the use of the telephone for telehealth communication is the most common because individuals are more comfortable and familiar with it than other forms, thus, being familiar with the mode of communication improves a patient's experience of it in telehealth situations (Gorrie et al., 2021b).

It is undeniable that there are some practical advantages to telegenetics and it has been described as comparable and non-inferior to IPGC (Danylchuk et al., 2021; Green et al., 2023). However, there are practical disadvantages to consider in telegenetics as well. Such disadvantages described by participants in this study included a distracting environment, not having a partner present and not being able to use multiple languages easily. Patients being in a distracting environment has been described before from the Genetic Counsellor's perspective (Badje, 2021). These distractions can lead to patients walking away from the telephone/computer and leaving the Genetic Counsellor alone with no way to follow after the patient (Badje, 2021). This was not described by the participants of this research but rather it was experienced by the researcher in a telephonic interview done for this research; where the participant moved away from her phone during the interview resulting in a pause in conversation with the researcher not being able to communicate with the participant. These distractions both from the patient's perspective in this research and from the Genetic Counsellor's perspective in literature lead to interruptions and incomplete information transfer (Badje, 2021). Genetic counsellors can advise their patients prior to the telegenetic appointment to be in an environment with limited distractions to aid information sharing.

A partner's involvement in decisions regarding prenatal invasive testing vary depending on the couple/relationship and how decisions are made for these families/individuals (Stoll & Jackson, 2020). P2 raised the concern that if she was given an IPGC appointment then her partner would have attended with her. P2 and P3 (P2's husband) both attended the TGC session but that was coincidental rather than planned. Contrastingly, in a previous study, women that didn't require agreement on prenatal testing from the partner preferred IPGC, whereas women that did require a partner's agreement preferred classroom or group GC (Nishiyama et al., 2021). The classroom or group GC was in-person and allowed for a private one-on-one session with a Genetic Counsellor after the group session (Nishiyama et al., 2021). Although, this literature showed that of 114 participants, IPGC was preferred by 80.7%, thus it was the preferred mode generally (Nishiyama et al., 2021). Therefore, this study demonstrated that when a partner was involved an in-person session was preferred, but this was also preferred when the partner was not needed for decision-making.

Another interference in telemedicine is language barriers, which is important to consider in South Africa as there are 11 official languages. (Bigna et al., 2014; Scott Kruse et al., 2018). Although translators can be used over the phone this is very challenging and information can be lost in translation. As noted in previous literature, when language barriers are not overcome, disparities in health care are created (Saeed & Masters, 2021). Therefore, this was also important to consider when reflecting on objective 3 and the challenges to AMA telegenetics in South Africa.

Bearing in mind that language barriers can be present, the overall opinion of the participants on the information shared by the Genetic Counsellor was positive. Many participants felt the information shared was helpful and informative. A similar view of GC session in South Africa being informative was described by Morris *et al. (2015)*. In the current study, P7 described that AMA women who didn't contact the GC service should do so, because she learnt a lot and thought others would do so as well. Health literacy is defined as "the achievement of a level of knowledge, personal skills and confidence to take action to improve personal and community health by changing personal lifestyles and living conditions" by the World Health Organization's (WHO) website (World Health Organization, 2023). Health literacy in South Africa regarding genetics is limited. In a previous South African study, mothers of children with genetic conditions felt their communities were unaware of genetic conditions (Morris et al., 2015). Participants in this study commented on how the GC appointment allowed them to learn new information regarding AMA risks and DS. This was also similarly described in a previous research study investigating the formerly in-person AMA GC at GSH (Vorster, 2020).

Greater knowledge leads to greater health literacy, which allows patients to have more control and be able to make informed decisions. In this study, P4 described having done her research, through having the GC session, and thus being able to choose not to have invasive testing. Other participants chose to have invasive testing so that they could have a greater sense of control through preparation. This shows how the GC session improved health literacy regardless of decision around invasive testing. Genetic Counsellors are trained in skills that allow for complicated information to be described in lay language and thus made more understandable. Therefore, GC skills can be applied to improving health literacy (Mann et al., 2015). This was shown to be a positive of this service, with informed decision-making a primary goal. This positive of GC services has also been previously described (Stoll & Jackson, 2020).

An important aspect of health literacy comes from the patient taking initiative to investigate their situation further and the internet is an incredible tool for that. Not all participants, in this research, sought after more information after their GC session as they felt that the GC trainee had explained everything sufficiently to them and that they could trust what the counsellor had told them. Thus, trusting the GC trainee can directly impact if and/or why a patient would search for more information online. Searching for more information online would have also caused more anxiety for P6. There is a copious amount of information available online and previously it has been described that it was difficult for patients to differentiate between truth and lies (Diehl-Jones, 2020). However, despite this challenge some participants, from the current study, found online resources after their GC session useful. These participants either wanted to get more information on the topic or they found it difficult to absorb all the information in the GC session and needed to search online to clarify the facts. A study in prenatal counselling resources, described that less than 10% of Genetic Counsellors distributed website links, books or other resources, but 40.1% of patients independently accessed website links (Delgado, Schulkin & Macri, 2022). Women receiving prenatal care have turned to online recourses in order to fill their information gaps and this was noted to be a generational approach (Kraschnewski et al., 2014). Although this technology was not found to be without limitations such as giving inaccurate information and patients having to search multiple sites to find their sought after information (Kraschnewski et al., 2014). In line with Kraschnewski *et al.* (2014), participants, from this research, mentioned that searching the internet for information was confusing and that speaking to a GC trainee allowed them to hear the information that they were interested in. P6 also described how receiving information from a person is different and better than looking for it by yourself online.

Online technology provides a beneficial supplementation to a GC session, especially providing visual aids in the context of a TGC. P4 described watching a video of an amniocentesis, which helped her understand this invasive test. Although an amniocentesis could have been described over the phone, this level of detail and visual information could not have been shared. With high frequency of pregnant women searching for online resources independently, it could be valuable to have healthcare professions providing accurate and safe websites (Delgado, Schulkin & Macri, 2022). Genetic counsellors can also provide website links and contact details to relevant support groups, such as Down Syndrome South Africa. This would be important as the information online, including visual aids, can be harmful. P1 described pictures she saw online after her GC session as traumatising. Although, healthcare providers often recommend not searching online, providing safe websites is a better approach since online resources are so commonly used by patients (Delgado, Schulkin & Macri, 2022).

Conclusion

In conclusion, these results were able to answer the aims and objectives of the study proposed. This research allowed for the understanding of AMA patients' opinion on the telegenetics service provided to them, as well as the greater referral and information gathering that form part of this experience as a whole. As can be seen in chapter 3, there were both positives and negatives described by participants about the telephonic AMA GC service. A significant determinant in the participants' experience of the service was based on if/who they trusted. Overall, participants described trusting their GC trainee, which allowed for participants to trust that they were given accurate and complete information and could make informed decisions from this information provided. The only doubt surrounding the trust toward the GC trainees was from P2, who based this opinion on the fact that anyone can lie over the phone. On the other hand, there was a greater sense of distrust towards their MOUs, which created uncertainty about the information shared at the MOUs. Participants had varying experiences of their referral from the different MOUs. Some participants were not given enough information at the MOU and did not have clear or correct expectations for the GC session. The pamphlet that was occasionally given out at the MOUs was helpful to some, but it could be adjusted to not include the image of the amniocentesis and to be more succinct. The distrust in the healthcare system can have a significant impact on patients' understanding and decision-making, especially when the GC service is optional after the often-incomplete information provided at the MOU. The negatives of the service were the practical disadvantages (a distracting environment, not having a partner present and not being able to use multiple languages easily) and that the emotional connection was not always sufficient for the participant. The main concerns for TGC are the limited emotional connection and the distrust that can occur because facial expressions are not visible. The lack of visual aids is also a disadvantage described which is also why participants went to online resources post-GC. The positive opinions of the service included the practical benefits (decreased financial strain, timeous service, continuation of care, and decreased need for transportation), that it was informative, allowed a sense of control in their decision-making process, and there was some emotional connection. A benefit of the telephonic service was that for the participants that wanted an invasive test, they had clear expectations for when they came to GSH for the appointment. At the start of this research there was a concern about shared mobile phones, but this was only the case for one participant, and it did not interfere with her

TGC session. Overall preferences of a telephonic or IPGC service varied. Comfortability for patients depended on their personal preferences between telephonic and in-person. Some participants didn't have a major preference for in-person or telephonic and felt that the mode of communication would not make a difference. Overall, there are benefits and barriers to be aware of, but most participants found the GC session informative and allowed them to make informed decisions, which aligns with the goals of Genetic Counselling.

Strengths and Limitations

Strengths

- The researcher did not provide GC to any of these participants which allowed her to be viewed as an independent individual.
- The interviews were semi-structured which allowed for important or new topics that arose in the interviews to be explored further, providing richer data.
- The participants of the study have diverse backgrounds including their home language, education level, employment, and prenatal histories. This allows for a more representative study group that makes the research more applicable to the greater population that uses the GSH AMA GC service.
- Both telephonic and in-person interviews were performed. The telephonic interviews also allowed insight for the researcher into some of the advantages and disadvantages of telephonic conversations.

Limitations

- As this was a prospective study, the researcher relied on frequent referrals to the service that met the study requirements. The number of appropriate possible participants was quite limited which made recruitment slow. Since this is a minor dissertation with limited time, a smaller cohort was used than what the researcher anticipated.
- The participants of this study were counselled and interviewed in English. Women that were counselled in another language may have had a different experience of this

service. Additionally, these participants may not have been able to fully express themselves in a language which is not their home language.

- This study was conducted at for a master’s level minor dissertation, and it is therefore possible that additional themes and understandings may have been identified with additional time and expertise. Due to the limited experience of the researcher, potential bias could have arisen, which is why supervisors were consulted throughout the research process.

Implications and Recommendations

As shown, there were several factors outside of the GC appointment that influenced participant’s experiences. Therefore, the implications and recommendations will also touch on these areas.

- Genetic counsellors should ask if the AMA woman would like to have her partner included in the GC session or not.
- Genetic counsellors should have appropriate website sources that can be given to patients to allow them to find accurate information.
- The pamphlet should be given out at all MOUs but can be re-evaluated regarding its length and images included.
- The referral process also needs to be made more consistent and clearer regarding what to expect at the GC service.
- Have GC services available at the MOUs.
- Future research should aim to assess the impact and importance of the emotional connection and trust with the GC trainee in a larger cohort.
- Future research can also focus on how Genetic Counsellors build trust with their patients.

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Appendix

Appendix A: Question Guide

These questions will focus on the opinions of advanced maternal age patients on the telephonic genetic counselling service that was delivered to them. Questions could also include thoughts on possible improvement options.

Topic Guide and Prompts

Question Guide

These questions will focus on the opinions of advanced maternal age patients on the telephonic genetic counselling service that was delivered to them. Questions could also include thoughts on possible improvement options.

Topic Guide and Prompts

Opening question

- What was your impression of the telephonic advanced maternal age genetic counselling session?
    - What do you think of the telephonic genetic counselling session that you had?
    - What were you expecting when you contacted the service?

Referral system

- How were you told about our genetic counselling service?
    - What information were you provided with about advanced maternal age and the genetic counselling service?
    - What was your understanding of this information?
    - How was this information provided to you?
- What was your reason for contacting us?
    - Why did you contact the genetic counselling service?

  - Were you aware that contacting the service was optional?
- Why do you think other advanced maternal age women chose not to contact our service?
  - How did what you were told about the service at your clinic match with what was discussed in your telephonic session?
    - What did match?
    - What didn't match?

Telephone service

- How was having the session over the phone?
  - What did you like about having this discussion over the phone?
  - What did you not like about having this discussion over the phone?
  - What difference would it have made to have the session in-person?
- Have you had other healthcare sessions on the phone before?
  - Were you expecting to discuss medical information?
  - How do you feel discussing medical information over the phone?
- How did you feel in the session?
  - Were you feeling ready to talk about all the risks to your pregnancy?
  - How was it receiving the info over the phone?
  - How did it feel discussing such sensitive information on the phone?
  - How did you feel about discussing risks over the phone?
  - What did you understand about the risks discussed in your session?
- How was the emotional connection with the genetic counsellor?
  - How would this change if you could see the genetic counsellor on video or in-person?
- How did the genetic counsellor respond to your emotions/feelings?
- What information did you find difficult to understand?
  - Do you think photos and images have helped you understand the information?
  - How was the counsellor's explanation confusing?
  - Did/What did you google after your telephonic session with the counsellor?
- How did you feel about the service as a whole?

    - What would you change about the service?
    - What would you keep the same in the service?
- How did you feel about having the session over the phone and not in-person?
    - What did you like about it being telephonic?
    - What did you dislike about it being telephonic?
    - How would feel having a face-to-face session?
    - What difference would a face-to-face session have made?
    - What made receiving the information on the phone stressful?
    - If you were given the option to have the session in-person, how would that compare?
    - What information was useful or beneficial from your session?
- What were the technical issues or practical issues with your session?
    - How was scheduling the phone call?
    - How was hearing each other on the phone? Any issues hearing each other?
    - Were there any difficulties understanding each other due to language?
    - Were there any issues getting access to a phone to use for the call?
    - Were there any concerns about data or minutes usage throughout this process?
- Is there anything else you would like to add?

Appendix B: Information Sheet for Participants

Samantha Bayley Research Project 2023

Dear Participant,

A research study is being done at the University of Cape Town (UCT) through the Division of Human Genetics for a minor dissertation for MMedSc Genetic Counselling.

11 February 2024

The aim of this research is to investigate your opinions as a woman who is of advanced maternal age on the telephonic service provided to them through Groote Schuur Hospital (GSH).

We hope this study will help us identify what the patients think about the telephonic genetic counselling session so that changes can be made to improve the service.

The information for this project will be gathered through individual, private interviews, which will take between 30 to 90 minutes. The interview will be voice recorded, but all your information will be kept confidential. You will be required to sign a consent form before we begin the interview. Whether you participate in the research or not will not influence your health care in any way.

This process is voluntary, and you can withdraw from this study at any time.

If you have any questions, please contact Ms Samantha Bayley at 0743694012 or bylsam004@myuct.ac.za or Mrs Nakita Laing (Supervisor) at n.verkijk@uct.ac.za or Ms Malebo Malope (co-supervisor) at mmalope@sun.ac.za or the UCT ethics committee at 0216501236 or hrec-enquiries@uct.ac.za.

Kind regards,

Samantha Bayley

Appendix C: Consent Form

Consent Form for S Bayley Research Interviews 2023

11 February 2024

Ethical clearance given by HREC Reference Number: 022/2023

I, ____, agree that Samantha Bayley from the division of Human Genetics at the University of Cape Town may interview me for the purpose of providing information about the telephonic advanced maternal age genetic counselling service. These interviews form part of an institutional research project that includes enquiry of patient's experience and opinions on the telephonic advanced maternal age genetic counselling service.

Statement of participation:

1. I am aware that taking part in this research project is voluntary and that I can withdraw from the study (without giving reason) at any time I feel a need to.
2. I know that I may answer questions that I am comfortable with and am free to not answer any question I am not comfortable with during the interview.
3. I am aware that personal questions will be asked in the study that may bring about strong emotions.
4. I have been provided with information about the study; the aims and objectives have been explained to me and I was given enough time to decide about taking part in the study.
5. I was given the opportunity to ask questions about the research project, I understand the aims and objectives of the study and I understand that I may ask questions at any time during the research.
6. I am taking part in this study at free will and was not pressurised by anyone who is a part of this study.
7. I am aware that all my personal details will be kept private and confidential as long as there is no conflict with the legal or ethical obligations of the researcher.
8. Provided that my personal information is kept private and confidential, I agree that the results of this project be used for scientific purposes and may be published.
9. I am aware that I will not be given any money or any other form of incentive for participating in the project.

10. I am aware that if there is concern about my mental health, the researcher will refer me for counselling, and it is my choice to attend the counselling or not.
11. I am aware that any health care I am receiving will not be affected by my participation in the study.
12. I understand that the interview will be voice recorded.

I give my consent to participate in this project and agree that I am participating at my own free will.

________________________ ________________________ _______________

Name and Surname of Participant Signature Date

____

S Bayley's Signature

Human Research Ethics Committee contact information: 0216501236 or

hrec-enquiries@uct.ac.za

HREC project number: 022/2023

Appendix D: Socio-demographic Survey

11 February 2024

Socio-demographic Survey for S Bayley Research Project 2023

Participant Code: ________________________

1) Age: ________________

2) Home language: _________________________

3) Educational Background

No Formal Education	
Primary Education	
Secondary Education but no Grade 12 (Matric)	
Grade 12 (Matric)	
Higher certificate/ Diploma University Degree	

4) Religion

Christian	
Islam	
Judaism	
Traditional/ Cultural Other (Please specify):	

5) Residence

Urban (Metropolitan/city)	
Suburban (developed area outlying city)	
Township/ Location	
Rural	

6) Employment Status

Unemployed	
Part-time	
Full-time	

7) Number of Children excluding the current pregnancy: ____

8) Number of miscarriages/stillbirths excluding the current pregnancy:_____

9) Gestational age genetic counselling was received: ______ weeks.

10) Do you have access to your own cell phone?

Yes	
No	

11) Do you have access to WIFI or data?

WIFI	

Data	
Neither	

12) Did you accept invasive testing?

Yes	
No	

11 February 2024

Appendix E: HREC Approval

UNIVERSITY OF CAPE TOWN
Faculty of Health Sciences
Human Research Ethics Committee

Room 45 E-52-E-Floor- Old Main Building
Groote Schuur Hospital
Observatory 7925
Telephone [021] 406 6492
Email: hrec-submissions@uct.ac.za
Website: www.health.uct.ac.za/home/human-research-ethics

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06 February 2023

HREC REF: 022/2023

Mrs N Laing
Division of Human Genetics
Falmouth Building- FHS
Email: n.verkijk@uct.ac.za
Student: sbayley123@gmail.com

Dear Mrs Laing

PROJECT TITLE: **INVESTIGATING THE OPINIONS ON TELEPHONIC ADVANCED MATERNAL AGE GENETIC COUSELLING**
(MMEDSc GENETIC COUNSELLING - MISS SAMANTHA BAYLEY

Thank you for submitting your study to the Faculty of Health Sciences Human Research Ethics Committee (HREC) for review.

It is a pleasure to inform you that the HREC has **formally approved** the above-mentioned study.

Approval is granted for one year until the 28 February 2024.

Please submit a progress form, using the standardised Annual Report Form (FHS016) if the study continues beyond the approval period. Please submit a Standard Closure form if the study is completed within the approval period.
(Forms can be found on our website: www.health.uct.ac.za/fhs/research/humanethics/forms)

The HREC acknowledge that the student: Miss Samantha Bayley will also be involved in this study.

Please quote the HREC REF 022/2023 in all your correspondence.

Please note that the ongoing ethical conduct of the study remains the responsibility of the principal investigator.

Please note that for all studies approved by the HREC, the principal investigator **must** obtain appropriate institutional approval, where necessary, before the research may occur.

Yours sincerely

PROFESSOR M BLOCKMAN
CHAIRPERSON, FACULTY OF HEALTH SCIENCES HUMAN RESEARCH ETHICS COMMITTEE

HREC/ref 022.2023