Investigating the views and expectations of pregnant women who undergo genetic counselling for age-related risk of aneuploidy.

by

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DECLARATION

I, Nina Vorster, 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: Pregnancy at advanced maternal age (AMA) is associated with an increased risk of aneuploidy. In the Western Cape's public health sector maternal age alone is widely used to screen women for high risk of pregnancies affected by aneuploidy. The weekly pregnancy counselling clinic (PCC) at Groote Schuur Hospital (GSH) offers genetic counselling (GC) for women who are of AMA to inform them about their age-related aneuploidy risk, offer invasive diagnostic testing (IDT) and discuss the option of voluntary termination of an affected pregnancy. A recent audit at GSH showed that the uptake of IDT was low and other literature reports that South Africans tend to have a conservative view regarding termination of pregnancy (TOP). This study sought to understand what women expect from the GC service at PCC as well as what their experiences are of the service.

Methods: This qualitative phenomenological study used a pragmatic approach and participants were recruited through purposive sampling. Semi-structured, in-person interviews were conducted after women had completed their GC sessions at PCC. Thematic analysis was used to analyse the data.

Results: The results of this study suggest that participants (n=7) received very little information about their GC appointments at referring clinics, and that they generally did not have prior knowledge about age-related aneuploidy risks. Finding out about the age-related risk of aneuploidy was an emotional experience for the participants, but other factors, including normal ultrasound results, provided relief. The participants' choices regarding IDT and attitudes towards TOP reflected that of available literature as the uptake was low and most participants reported that they would not consider a TOP. The women reported that they would use the knowledge they gained during GC to educate other women in their communities about the pregnancy risks associated with increased maternal age. Generally, the participants believed that GC was useful and appreciated the opportunity.

Conclusion: The participants in this study had limited health literacy and knowledge regarding AMA risks and GC. As a result, participants had no expectations of GC. However, the participants felt that GC was useful in helping them prepare for the possibility of a child with DS, and generally viewed the service in a positive light.

Additionally, this study’s results suggests that there is a need to educate women in local communities regarding AMA pregnancy risks.

DEDICATION

I would like to dedicate this document to my parents and every person who tries to leave the world a better place than they found it.

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I don't think there are enough words in the universe to adequately express how thankful I am towards the people mentioned below. I am sure anyone who has ever submitted a dissertation can attest that there are many moments where giving up seems like an easier option and without these individuals I am sure that route would have looked so much more attractive.

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ABBREVIATIONS

AMA – Advanced maternal age

CVS – Chorionic villus sampling

DS – Down syndrome

FA – Foetal anomaly

GC – Genetic counselling

GSH – Groote Schuur Hospital

HIV – Human Immunodeficiency Virus

IDT – Invasive diagnostic testing

LR – Likelihood ratio

MOU – Midwife Obstetric Unit

NIPT – Non-Invasive Prenatal Testing

NSCG – National Society of Genetic Counselors

NT – Nuchal Translucency

PCC – Pregnancy Counselling Clinic

qfPCR - Quantitative Fluorescence Polymerase Chain Reaction

SA – South Africa

SASAS - South African social attitudes survey

TOP – Termination of pregnancy

UCT – University of Cape Town

WHO – World Health Organization

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CHAPTER ONE: INTRODUCTION

This chapter aims to provide the necessary background as well as an overview of available literature relevant to the opinions, expectations and experiences of pregnant women who undergo genetic counselling (GC) for age-related risk of aneuploidy. This includes the following subjects:

1.1 Aneuploidy and prenatal screening for aneuploidy

1.2 Approach to aneuploidy screening locally and internationally

1.3 Experiences and expectations of advanced maternal age (AMA) women undergoing GC and the factors that influence this

1.4 The study rationale

1.1 ANEUPLOIDY AND PRENATAL SCREENING

Human cells typically contain 46 chromosomes. When an individual's cells contain either more or less than 46 chromosomes, certain well-documented disease phenotypes will become apparent. Cells with an abnormal number of chromosomes are said to be aneuploid. Aneuploidy accounts for the loss of approximately half of first trimester pregnancies, with the most common form of aneuploidy being trisomy (Pylyp et al., 2018). While there is significant prenatal selection against aneuploid foetuses, several of these aneuploidy conditions can result in the birth of a live infant (Jacobs et al., 1974). The trisomic condition with the highest live birth prevalence is Down syndrome (DS) (Loane et al., 2013), where the presence of an additional chromosome 21 results in intellectual disability and other health problems. Studies of Pakistani parents (Ahmed et al., 2013) and black South African mothers (Mbazima, 2016) who have children with DS revealed that these children are often stigmatised and isolated in their communities. These studies also indicated that there is a lack of understanding regarding the mechanism of the disease and members in the participants' communities frequently thought that the affected child is cursed. DS is a pan-ethnic condition with a reported incidence ranging from 1 in 500 to 1 in 1000 depending on the average maternal age and access to prenatal diagnostic and pregnancy termination services (Delport et al., 1995; Miao et al., 2012; Loane et al., 2013). Children with DS differ from other liveborn infants with trisomies in terms of their life expectancy. For trisomy 13 and 18 median survival is only a couple of days (Meyer et al., 2016), whereas the average life-expectancy of individuals with DS who receive adequate medical care has increased with medical advancements and in some places, like Australia, is approaching the life expectancy of the general population (Glasson et al., 2002). Another published case study also documents a male who is alive in his eighth decade with complete trisomy 21 (Krinsky-McHale et al., 2008). Due to the high prevalence, the intellectual and physical complications associated with the condition and the likelihood of compatibility with prolonged survival, DS is one of the main focus areas of prenatal screening and GC programs worldwide.

Prenatal screening for DS (and other aneuploidies) aims to identify women whose pregnancies are at elevated risk of being affected with DS so that the option of further diagnostic testing can be pursued. One of the most commonly used aneuploidy screening strategies is antenatal ultrasound. Markers that are suggestive of aneuploidy during the first trimester ultrasound include increased nuchal translucency (NT), absent nasal bone, abnormal ductus venosus blood flow and tricuspid regurgitation. These markers are usually evaluated between 10 and 14 weeks gestation. Increased NT can have a sensitivity ranging between 63% and 77%% for DS while the likelihood ratio (LR) for tricuspid regurgitation in DS is 25. The implication of an absent nasal bone is however dependent on the ethnicity of the parents (O'Brien & Lichten, 2019). Ultrasound findings in the second trimester that can be suggestive of aneuploidy are short femur and humerus, echogenic bowel, an aberrant right subclavian artery, hydronephrosis, ventriculomegaly, increased nuchal fold thickness, an intracardiac echogenic focus and absent or hypoplastic nasal bone (Agathokleous et al., 2013). Studies have shown that that the likelihood of a foetus being aneuploid increases with every additional marker or anomaly that is present (Nyberg & Souter, 2001; Agathokleous et al., 2013; Kataguiri et al., 2014).

Another aneuploidy screening strategy involves the analysis of certain protein levels within the maternal blood at specific gestational periods. Biochemical markers that are typically screened for during the first trimester are pregnancy associated plasma protein A (PAPP-A) and the free beta subunit of human chorionic gonadotropin (free ß-hCG), while the quadruple screening test in second semester measures alpha-fetoprotein, unconjugated estriol, inhibin A and human chorionic gonadotropin levels (Malone et al., 2005). While these screening methods are effective in isolation, they are much more sensitive for aneuploidy when ultrasound findings and biochemical markers are used in combination. It has been shown that an increased NT with an abnormal first trimester biochemical marker has a sensitivity of 87% (Malone et al., 2005) for DS and the detection rate can increase to as high as 95% depending on the combination of screening techniques (Durković et al., 2018). This is much higher than the sensitivity range for an isolated increased NT.

Non-invasive prenatal testing (NIPT) is another option for aneuploidy screening. This is the latest addition to screening strategies and can be done throughout the pregnancy. It quantifies the chromosomes in the cell-free foetal DNA that is circulating within the maternal plasma to determine whether or not a foetus is likely to be aneuploid. A meta-analysis showed that this screening method has a sensitivity and specificity of more than 99% for DS (Taylor-Phillips et al., 2016). However, NIPT has an estimated 0,05% false positive rate for DS (Samura, 2020) and a 0,2% false negative rate for trisomy 13, 18 and 21 (van Opstal et al., 2014). Therefore invasive diagnostic testing (IDT) should still be used to confirm a diagnosis of aneuploidy. Lastly, the easiest and most affordable screening strategy is to identify pregnant women of advanced maternal age (AMA).

A correlation between maternal age and increased risk of having a pregnancy affected by DS was first noted in 1933 (Penrose, 1933) and has since been confirmed in other studies (Hassold & Chiu, 1985; Allen et al., 2009). The correlation with AMA, which is generally defined as 35 and older at the time of conception, is additionally supported by an increase in the prevalence of DS in countries such as the USA where the average maternal age has been steadily increasing (Egan et al., 2000; Shin et al., 2009). The reason for this association has not been fully elucidated. Research has shown that in oocytes obtained from women over 40, several genes involved in the cell cycle are down regulated. These included genes involved in spindle formation, commencement of anaphase after proper alignment of the chromosomes in metaphase and chromatin condensation to form chromosomes (Steuerwald et al., 2007). Additionally, older oocytes have shorter telomeres, which contribute to chromosomal instability and spindle formation and can result in nondisjunction (Steuerwald et al., 2007). Literature published in the last decade also shows that cohesion between chromosomes decreases with AMA and suggests that this is a contributing factor to the correlation between aneuploidies and AMA (Duncan et al., 2012).

The incidence of DS among AMA women differs between cohorts. Risk estimation in a Thai cohort reported that a mother who is 40 years of age has a 1 in 67 chance of having an affected foetus and these odds increased exponentially with every additional year in age to 1 in 14 at the age of 48 (Jaruthamsophon et al., 2016). In the United Kingdom (UK) a study found a risk for a liveborn infant with DS of 1 in 112 at the age of 40 and 1 in 6 when a woman is 50 years old (Morris, Mutton & Alberman, 2002). It is estimated that the detection rate for DS using only maternal age as a screening tool is 50% (Nicolaides, 2011).

1.2 APPROACH TO ANEUPLOIDY LOCALLY AND INTERNATIONALLY

In recent years, international bodies such as the Society for Obstetricians and Gynaecologists of Canada and the Board of the International Society for Prenatal Diagnosis has suggested that using maternal age as the sole indication to offer invasive testing is not justified when other screening options are available and suggested that pregnant women of all ages should be offered screening (Benn et al., 2011; Chitayat, Langlois & Wilson, 2011). However, due to resource constraints in the public sector in the Western Cape, no biochemical marker or NIPT screening is available and screening focuses on ultrasound markers and AMA only. There are several local and national policies that exist to guide the aneuploidy screening process. The most recent policy adopted by Groote Schuur Hospital (GSH) - a tertiary hospital within an urban area of the Western Cape Province – makes provision for a nuchal translucency scan in first trimester and/or a foetal anomaly (FA) scan in second trimester depending on maternal age. This policy balances screening access and resources and, as a result, women who are under 40 are referred to a tertiary hospital for possible invasive testing only if there is an abnormal ultrasound finding or soft marker. However all women aged 40 and older are required to be referred for genetic counselling (GC) in their second trimester up to 23 weeks of gestation, irrespective of whether they have had a foetal anomaly or NT scan, to be offered IDT (UCT Department of Obstetrics and Gynaecology, 2020). GSH offers a weekly multidisciplinary Pregnancy Counselling Clinic (PCC) where one of the services offered is GC for pregnant women of AMA.

The American National Society of Genetic Counselors (NSGC) defined GC as ”the process of helping people understand and adapt to the medical, psychological, and familial implications of the genetic contributions to disease” (Resta et al., 2006:79). The authors further state that the process of GC involves obtaining family histories for risk assessment, educating patients on genetic disease mechanisms, making resources available to them and, lastly, promoting adaption to a genetic diagnosis and informed decision-making (Resta et al., 2006:79).The women of AMA who attend PCC for GC of are typically referred from a lower-tiered Midwife Obstetric Unit (MOU) or secondary level hospital to GSH, between 16 and 23 weeks of gestation. When appropriate they may have an FA scan prior to GC. When the service is busy, several AMA women may initially be counselled in a group to make the process as efficient as possible. During the group sessions these women are informed of what DS is - including the intellectual and physical health problems associated with the condition, what causes DS as well as details on IDT and voluntary termination of affected pregnancies. Each woman is then seen individually, to discuss her personal risk and circumstances as well as aid her in the decision-making process.

There are three commonly used invasive diagnostic tests: chorionic villus sampling (CVS), amniocentesis and cordocentesis. GSH typically sees a second trimester referral pattern for GC and IDT, which correlates with the most appropriate time for an amniocentesis (The Canadian Early and Mid-trimester Amniocentesis Group, 1998). As a result, testing for AMA women at PCC are usually restricted to an amniocentesis. This procedure involves obtaining amniotic fluid transabdominally, allowing the chromosomes of the cells obtained from the amniotic fluid to be quantified through quantitative fluorescence polymerase chain reaction (qfPCR) or karyotyping to determine whether the foetus is aneuploid (The American College of Obstetricians and Gynecologists, 2016). It is widely believed that invasive prenatal diagnostic tests pose an increased risk of miscarriage, with the exact risk being dependent on operator experience. In contrast, a recent study argues that the miscarriage rate for invasive procedures is negligible when compared to the miscarriage rate in individuals who do not undergo invasive procedures (Beta et al., 2019). As these new findings have not yet been incorporated into any policy, a 1 in 500 miscarriage risk, based on literature and the unit's history, is communicated to the women who undergo amniocentesis at GSH. Previous research has shown that the fear of miscarriage is a significant cause of stress in individuals who undergo IDT (Scott, 2018). Another risk factor that is very relevant in a South African setting is that of vertical Human Immunodeficiency Virus (HIV) transmission from mother to child during the invasive procedure as this has been documented (Mandelbrot et al., 2009). However, it has been suggested that it is safe to perform the procedure on women who are on highly active antiretrovirals and have a suppressed viral load (Constantatos, Boutall & Stewart, 2014). A previous study has shown that HIV-transmission risk has an impact on women's decisions regarding IDT (Bee et al, 2013) and this along with the miscarriage risk has to be considered by women during their GC appointment.

If a pregnancy is affected with aneuploidy, a woman might be able to choose to have the pregnancy terminated depending on the gestational age. The Choice of Termination of Pregnancy (TOP) Act in South Africa (SA) allows a woman to terminate a pregnancy up to 12 weeks gestation "on request". TOP is also permitted for foetal reasons between 13 and 20 weeks "if there exists a substantial risk that the fetus would suffer from a severe physical or mental abnormality" among other criteria and after 20 weeks for "severe malformation of the fetus" (Choice of Termination of Pregnancy Act, 1996:4). The interpretation of severity has not been tested legally and so there is no uniform consistency in offering TOP for DS in SA. In the Western Cape public sector, taking into account the practical realities of late referral and diagnosis and extrauterine viability of the foetus in the local setting, TOP for uncomplicated DS is usually offered up until 24 weeks of gestation.

Research that evaluated opinions on TOP in the case of foetal anomalies of a culturally representative sample in South Africa, reported that slightly more than half of the participants felt that abortion in the case of a child with malformations is wrong (Mosley et al., 2017). Additionally, a local study that retrospectively investigated the attitudes of parents with children with DS towards prenatal diagnostic testing and TOP reported that the participants thought that prenatal testing should be available to prepare for an affected child, but that they would not personally use the information to terminate the pregnancy (Scott, Futter & Wonkam, 2013). An audit of amniocentesis uptake at GSH also showed a significant decrease from the early 1980s to the 2000s (Urban et al., 2011). The reasons for this trend have not been fully explored in the local PCC setting. Considering the views expressed in Scott (2013) and Mosley (2017), this raises the question of what these women's expectations of the services at PCC are and provides the foundation for the aim of this project.

1.3 EXPERIENCES AND EXPECTATIONS OF ADVANCED MATERNAL AGE WOMEN UNDERGOING GENETIC COUNSELLING AND THE FACTORS THAT INFLUENCE IT

Very little research data is available about the opinions, experiences and expectations of women who are counselled only for AMA and the majority of literature reports on cohorts from high income countries whose demographics are vastly different from those of the general South African population. The latest census data reports that the majority of the City of Cape Town's population does not speak English as a first language and that less than half of the city's adults finished high school. The City's unemployment rate is almost 24% and a fifth of its residents live in informal housing (Statistics South Africa, 2011). Additionally, a previous review about genetic services in SA notes a lack of knowledge about the genetic services that are available to individuals (Kromberg, Sizer & Christianson, 2013). In many indigenous South African languages no specific word exists for DS (Mbazima, 2016).

A UK-based qualitative study that investigated what patients with several different genetic conditions expected from medical genetic services revealed that these individuals wanted to feel empowered. This empowerment was based on four pillars. The first pillar was decision- making - not just immediately with regards to testing and health care, but also confidence in future decisions that might be impacted on by the genetic condition. Knowledge and understanding is the next pillar, and the study found that information about how the disease is going to affect individuals' lives made them feel empowered because it created a sense of control. Another pillar was instrumentality and this referred to giving patients information about the support systems that are available to them so that they can actively engage with these. The last pillar of empowerment was to leave the medical genetic services feeling hopeful about the future (McAllister et al., 2008). Both the knowledge and instrumentality pillars mentioned in this study speak to the broader concept of health literacy, which is defined by the World Health Organization's (WHO) website 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" (World Health Organization, 1998).

Some studies done in Europe and America on cohorts that included AMA clients reported that the majority of participants found GC helpful and expected to be given information during the GC session. Some expected psychological support from their counsellor, while others also wanted the counsellor to assist in decision-making (Tóth, Nyári and Szabó, 2008; McCarthy Veach et al., 1999). Another study in Italy reported that women who undergo invasive prenatal testing are satisfied with the GC that they receive, but that it does not change their overall perception of risk. The study also reported that women who had already decided to undergo IDT prior to being counselled were unlikely to change their minds after being counselled (Godino et al., 2016).

In addition to the support and input that women who undergo GC expect from the counsellor, it has been shown that partners who accompany women to the prenatal GC consultation can also play a supportive role (Humphreys et al., 2003). In a study done to assess the experiences of black South African mothers of children with DS, the participants who received information about DS alone without any other family members present reported that they felt it a heavy burden to share with other family members and were often met with stigma and attitudes of blame (Mbazima, 2016). Additionally, they mentioned that after the initial shock, they struggled to retain all the necessary information about DS and could not relay it fully to the family (Mbazima, 2016). Partners do not always accompany the women to their appointments at the PCC and it is possible that this might contribute to the experiences of the women.

1.4 THE STUDY RATIONALE

The researcher was first introduced to the GC services for AMA women at PCC during her first year of studying to be a genetic counsellor. From the literature it seems as if uptake of IDT at GSH is low and that many women may not personally choose to terminate a pregnancy. This, as well as the general lack of literature regarding expectations of GC for age-related risk of aneuploidy in the Western Cape, suggested that further research needed to be done to understand what women’s thoughts are about the GC service at PCC. This research project therefore hopes to fill some of the gaps that exist with regards to perceptions and needs of GC for AMA women in SA and will attempt to answer the research question: What are the views and expectations of pregnant women who undergo GC for age related risk of aneuploidy at GSH?

1.4.1 AIM AND OBJECTIVES

Aim:

To investigate the views and expectations of pregnant women who undergo genetic counselling at Groote Schuur Hospital for age related risk of aneuploidy.

Objectives:

- Determine participants’ understanding and expectations of GC at PCC for AMA
- Determine whether participants’ expectations of the GC session are met
- Explore participants’ views on the referral process from their local MOU
- Determine participants’ views and opinions of the PCC’s GC services

CHAPTER TWO: METHODOLOGY

This chapter aims to familiarise the reader with the process that the researcher followed to determine the views and expectations of the participants with regards to the PCC's GC service. As such the chapter will discuss the following:

2.1 Study Design

2.2 Research Setting

2.3 Research Population

2.4 Participant Recruitment

2.5 Data Collection

2.6 Data Analysis

2.7 Ethical Considerations

2.8 Trustworthiness

2.9 Researcher Reflection

2.1 STUDY DESIGN

The researcher wanted to understand the views and expectations of pregnant women of AMA who attended GC for age related risk of aneuploidy. Qualitative research allows for the immersion of the researcher into a participant's experiences (Austin & Sutton, 2014) and was therefore an integral part of the study design. Qualitative research has previously been described as "a sequence of interpretive techniques that try to describe, decode and translate concepts and phenomena rather than to record the frequency of certain phenomena in society" (Basias & Pollalis, 2018:94). The researcher believed that a qualitative study would provide more in-depth answers as well as the chance to explore participants' attitudes towards the subject of AMA counselling and that it was better suited to addressing the aims and objectives of the project.

The underlying design of this study was based on the theory of phenomenology. This design aims to understand the experiences of individuals in a predetermined setting (Reeves et al., 2008). To help the researcher understand the views and experiences of study participants in this phenomenological study, a pragmatic approach was used.

This approach is defined as using the most reasonable methods available to investigate a phenomenon (Savin-Baden & Howell Major, 2013) and allows the researcher to combine several qualitative designs into an approach that will allow for a clear answer instead of being boxed-in through specific requirements of each individual design approach (Clarke & Visser, 2019). The pragmatic approach in this instance involved the use of individual semi-structured face-to-face interviews to obtain data and thematic analysis to understand the shared patterns of thoughts among the women with regards to the GC service (Braun & Clarke, 2006). Individual face-to-face interviews allowed the researcher the opportunity to observe non-verbal cues from participants and this helped guide the researcher in probing deeper into participants thoughts and attitudes about the research topic. The semi-structured nature of the interviews also allowed the researcher the flexibility to follow the participants' train of thought, while still ensuring that the interview answered the research question (Frances, Coughlin and Cronin, 2009).

2.2 RESEARCH SETTING

As explained in chapter one, the PCC is a weekly clinic that is held every Wednesday morning in the maternity building at GSH. Pregnant woman who are of AMA are specifically seen for GC. The GC team includes doctors, nurses and genetic counsellors, as well as supervised MMedSc Genetic Counselling students and interns who are training to be genetic counsellors. The counselling session typically takes between 30 and 45 minutes depending on each case. Each session includes a detailed history-taking to assess any additional family risks, information-giving regarding DS and AMA, personal risk information, options for further pregnancy management including IDT and options dependent on the results and, lastly, decision making. If the patient decides to accept IDT, the amniocentesis is usually performed in the afternoon of the same day and results are delivered either telephonically or in-person by the counsellor within 2 to 3 working days when the result of the qfPCR becomes available. Depending on the gestational age of the foetus, some women are also informed that they have time to consider their decision regarding IDT. In this study interviews were performed before the participants who chose IDT had undergone the procedure.

2.3 RESEARCH POPULATION

The study population consisted of pregnant women who attended the PCC at GSH for GC, solely for their age-related risk of aneuploidy. The following criteria were used to determine eligibility:

INCLUSION CRITERIA

- Participants had to be 40 or older at the time of conception
- Participants had to be scheduled for a GC consultation for increased age-related risk of having a child with chromosomal aneuploidy

EXCLUSION CRITERIA

- Women who were at an increased risk of having a child with congenital abnormalities for any reason other than advanced maternal age
- Women with foetal anomalies or markers indicative of an increased risk of aneuploidy identified on an ultrasound
- Women who were at increased risk of aneuploidy due to previous screening tests such as biochemical serum markers
- Women who declined to participate
- Women who had an intellectual impairment
- Women who were unable to be counselled in English
- Women with one or more family members affected with DS
- Women who had previously been seen at the GSH Foetal Medicine Unit
- Women who had previously received GC

2.4 PARTICIPANT RECRUITMENT

Participants who met the inclusion and exclusion criteria were identified and recruited through purposive sampling. After each participant's counselling session, the genetic counsellor who saw the eligible individual briefly explained the research project and asked permission for the researcher to come and discuss the research with the participant. If permission was granted the researcher then explained the aim of the study and each participant was assured of her unchanged, continued health care irrespective of whether or not she chose to participate. The researcher also gave a brief overview of the consent form and highlighted important points. Participants were then given time to read through the study information and consent form and ask any questions. After agreeing to partake in the study, each participant completed and signed the necessary consent form (Appendix A).

Each participant’s counselling session and interview was done in English. The PCC caters to women with a variety of home languages and translators are available when needed. However, this is a very scarce resource and translators were not available for the research study. English is the language in which most women at the PCC are comfortable conversing and was therefore likely to be inclusive of those from diverse ethnolinguistic backgrounds.

A total of 7 participants were interviewed on 5 different clinic days over a period of several weeks for this project. Interviews lasted between 33 and 55 minutes. The researcher believes that she was close to data saturation as very few new thoughts emerged from the last transcript. However, the normal PCC service for AMA women as well as face-to-face research were temporarily discontinued for several months due to the COVID-19 pandemic (See Appendix E), which limited recruitment. As a result confirmation of data saturation was not possible.

2.5 DATA COLLECTION

In the experience of the researcher it is often hard to telephonically reach patients after they attended PCC. As a result it was decided to have the interviews in a space that is convenient for both the researcher and the participant and the interviews were conducted in the same room where participants had undergone GC immediately after they have finished with the counsellor. The advantage of this was that it allowed the researcher to understand the attitudes of participants after they each had an equal amount of time to reflect on their GC experience. The ethics-approved protocol made provision for participants who wanted to participate in the study but were unavailable immediately after counselling by allowing participants who had another appointment at the hospital within two weeks to be interviewed. The two-week limit was set as previous research has proven that the accuracy of information recall decreases over time (Sherman, 2012) and it would mean that participants were still in a similar gestational period as when they were counselled. Only one counselee was approached for this option, but never contacted the researcher to confirm that they wished to participate.

Interviews started with participants completing the demographic questionnaire (Appendix B). The questionnaire included questions about the participants' age, relationship status, home language, education and job and was also used as a relationship building tool between the researcher and participant. A review article explained the importance of quickly building rapport as this helps to initially get the participant to talk and eventually builds trust so that the participant feels comfortable with sharing (DiCicco-Bloom & Crabtree, 2006). Additionally, the data from the demographic questionnaire was used to determine the diversity of the cohort and give an indication of how this cohort may differ from those in other studies in the literature.

Data was then collected in the form of a semi-structured in-depth face-to-face interview. The previously mentioned review article indicates that a question guide of approximately 10 questions should be developed to help guide the interviewer to explore the participant's answer to the research question in detail (DiCicco-Bloom & Crabtree, 2006). The semi-structured interview guide for this research was developed through four test interviews with GC students and supervisors. Both of these groups have been to the PCC and were therefore able to offer valuable insight. Test interviews are an important step in the process of developing a question guide as it allows for individuals with different viewpoints to assess and interpret the questions and also allows the researcher to identify and reconsider questions that are unclear (Bolderston, 2012). The question guide initially had several sub-questions, but after the first interviews with participants it was finalised to include 5 main questions (Appendix C) and the researcher could then listen to the responses of the participants and probe them further to understand their experience and opinions of the clinic. All interviews were recorded with the researcher's phone and were transcribed verbatim by the researcher as soon as possible after the interview.

2.6 DATA ANALYSIS

Thematic analysis was used to analyse the data obtained through the semi-structured interviews. This is a step-by-step process (as shown in Fig. 1) that results in the identification and analysis of topics that emerge from the data set (Braun & Clarke, 2006).

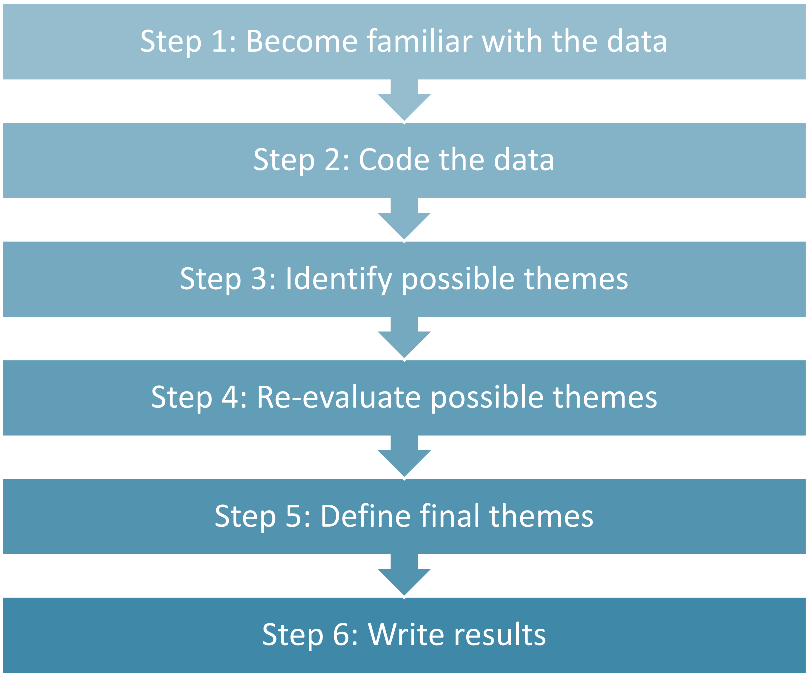

Figure 1: The 6 steps of thematic analysis (adapted from Braun & Clarke, 2006)

The researcher immersed herself in the data by transcribing the interviews and rereading the transcripts several times. The first three transcripts were used to create a coding dictionary. This was done by reading through the transcripts and highlighting codes that emerged from the data after the researcher familiarised herself with it (Stuckey, 2015). The codes from different transcripts were compared to determine overlapping codes and these were used to establish the coding dictionary. The next steps were to identify and evaluate possible themes. This was a back-and-forth process, as described in the literature (Braun & Clarke, 2006) that involved reassessing the codes and trying to see if there was any overlap that pointed to a bigger theme. Once certain codes were grouped together the researcher went back to her initial data and codes to see if the themes that were identified still supported the initial data and where necessary, made some changes before identifying the final themes as they are described in the results section.

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 636/2019 (Appendix D).

INFORMED CONSENT

Informed consent aims to present a possible research participant with all the necessary information about a study in a language that the participant understands to allow her to make a decision on her own about whether or not to participate (Nijhawan et al., 2013). The Helsinki declaration emphasises that consent should be freely given after participants are made aware of the aim, possible benefits and other factors regarding the study (World Medical Association, 2001). To adhere to this principal the researcher discussed the study as well as all the points mentioned in the consent form verbally with the participant (Appendix A) before giving the participant the opportunity to read through the document and ask questions. Each participant was assured of their continued unchanged healthcare irrespective of their participation in the study before they signed the document. All participants were also informed that they were allowed use the contact details on the consent form (Appendix A) to withdraw from the study at any time.

ANONYMITY AND CONFIDENTIALTY

Some of the most important ethical considerations where human subjects are involved are anonymity and confidentiality (Sanjari et al., 2014). To ensure that personal information was kept confidential in accordance with the Helsinki declaration, participant information was handled with the utmost care. Hard copies of transcripts were edited to omit any identifying information and all audio recordings were stored on a password-protected electronic device as well as on the cloud. The literature suggests that researchers should prevent unsanctioned access to interviews by having a secure password as well as ensuring the researcher can deactivate access to a stolen device (Barnhill & Barnhill, 2015). Both of these precautions were taken. Transcripts and recordings were given numbers to ensure that no personal information could be identified from the data and participant consent forms were kept in a locked office. The recordings and transcripts will be deleted after publication in a scientific journal. No identifying information was used in any draft or final version of the researcher’s thesis. Additionally, the large numbers of women seen at the PCC every week makes it very hard to identify an individual from the study population.

OTHER RISKS AND BENEFITS

No participant was ever in danger of any physical harm. As the information regarding aneuploidy risk can sometimes be distressing for certain individuals, the researcher reiterated that if any participant felt uncomfortable or wanted to stop, the interview could be terminated.

During the course of this research several participants had additional questions that they forgot to ask the counsellor. This did not happen often and in most situations the researcher told participants with questions that she would go and ask the original counsellor after the interview to prevent participants viewing the researcher as a genetic counsellor.

The research held no direct benefits for the participants themselves except possibly giving them the opportunity to verbalise concerns but it could benefit women who attend the clinic in future. People who participate in research have different reasons for doing so and as some participants might have been under the impression that the research will directly benefit them or their community (Sanjari et al., 2014) the researcher clearly explained that the research will hold no direct benefit for the participants (See Appendix A).

2.8 TRUSTWORTHINESS

According to the literature the trustworthiness of any qualitative research project is built on five pillars: credibility, dependability, confirmability, transferability and authenticity (Cope, 2014). This project aimed to incorporate these principles in the following manner:

- The researcher tried to ensure credibility of this data by engaging with the transcripts and interview recordings several times over a period of months. This helped the researcher to ensure that she was understanding and interpreting the participants statements in an honest fashion.
- To ensure dependability the supervisors in this project read and coded at least five transcripts and had several discussions with the researcher when themes were being developed.
- Confirmability aims to ensure that the data is not representative of research bias. The researcher spent some time at the beginning of this project to

establish her biases that could affect how she interpreted the results. Awareness of these biases ensured that the researcher engaged with results more thoroughly when they reflected her biases. The supervisors in this project were also aware of these biases and questioned the researcher's interpretation of results whenever they believed that it was influenced by her preconceived ideas.

- This project is too small to ensure transferability as it refers to whether or not research findings can be extrapolated to individuals outside of the cohort population. Due to the national COVID-19 lockdown, only a small number of the anticipated participants were recruited and the researcher was unable to reach data saturation. Therefore this research did not aim to answer the research question definitively for all AMA counselees but rather wanted to provide some insight into the experiences of this cohort specifically. This insight can be used to identify areas where more research is needed to ensure that the needs of AMA women who undergo GC are met.
- The last pillar of trustworthiness is authenticity and the researcher tried to ensure this by presenting the participants' emotions and statements as sincerely as possible.

2.9 RESEARCHER REFLECTION

Due to the nature of qualitative research, it was important for me to reflect on my biases before undertaking this project to ensure that those biases did not influence this project's results. I am a trainee genetic counsellor and have completed Medical Genetics and Principles of GC modules. As a result I interpret all patient experiences at clinics from the perspective of the literature regarding GC and my own experiences. I spent several months prior to this project at the PCC. During this time I engaged specifically with AMA women who often declined IDT and this left me wondering if AMA counselling serves a purpose as I could not understand the use of prenatal GC without IDT. As a result of this previous interaction with AMA women my greatest bias was that I believed AMA women at PCC did not want to know about the age-related aneuploidy risk and would resent the information that they received during GC and expected the project's results to reflect this. However, I also discussed this with individuals in my department who believed that the AMA counselling service was useful and important even when women do not accept IDT. For the duration of this project I constantly questioned how my biases could be reflected in the study results and discussed it with my supervisors and peers to ensure that I minimised their impact. Additionally, as the project progressed and more results became available my biases and opinions evolved and it was important that I considered how the changing biases impacted on my interpretation of future results.

CHAPTER THREE: RESULTS

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

3.1 COHORT DEMOGRAPHICS

The study's seven participants were all between the ages of 40 and 42 (see Table 1). The live birth risk of having a foetus affected with DS for women within this age range, as reported in a UK-based study, varied between 1 in 112 and 1 in 65 (Morris, Mutton & Alberman, 2002). Even though counselling and interviews were conducted in English, the participants' home languages portrayed the diversity of the cohort as it included five of the 11 official languages of South Africa, including the three most commonly spoken languages in the City of Cape Town (Statistics South Africa, 2011). The cohort had two participants with tertiary education, three who finished high school, one who attended high school but did not finish and one who only attended primary school.

Table 1: A summary of the study cohort's characteristics

Participant ID	Age	Gestation period	Relationship status	Home Language	Highest level of Education	Occupation	Decision regarding IDT
1	42	17 Weeks	Stable relationship (not married)	Ndebele	Tertiary education	Waitress	Declined
2	40	21 Weeks	Single	English/ Afrikaans	Primary School	Unemployed	Declined
3	41	21 Weeks	Married	Xhosa	High School (did not finish matric)	Cleaner	Declined
4	40	20 Weeks	Stable relationship (not married)	Afrikaans	Finished high school	Supervisor at a grocery store	Declined
5	40	21 Weeks	Stable relationship (not married)	Xhosa	Tertiary education	Health and Safety Officer	Accepted
6	40	20 Weeks	Married	Tswana	Finished high school	Unemployed	Declined
7	42	20 Weeks	Married	English/ Afrikaans	Finished high school	Receiving and Dispatch Manager	Accepted

3.2 THEMES

After analysing the participants' transcripts five major themes emerged (see Table 2). The first theme provides the background of the women's experiences when being referred to GSH and gives some insight into how this affected their appointments. The next theme then deals with participants' initial emotions at PCC when they realise that their child could be at risk for aneuploidy as well as the eventual acceptance of these risks. Theme three explores the different courses of action that participants could take after hearing about these risks and why participants made their particular choices. The fourth theme addresses the lack of wider knowledge regarding age-related aneuploidy risks as well as the women's call to action to address this particular problem. Lastly, theme five addresses how the participants found the complete GC experience. It is important to keep in mind that GC at PCC are provided by genetic counsellors, genetic nurses and medical geneticists and therefore GC in this case specifically refers to the process of receiving information on age-related aneuploidy risks and informed decision-making.

Table 2: A summary of the themes and sub-themes that resulted from data analysis

Themes	**Sub-Themes**
1. Lack of preparedness	1. Limited information from referring clinics
	2. Expectations unrelated to genetic counselling
2. An emotional experience	1. A stress-inducing situation
	2. The glass is half full
3. Decisions regarding the way forward	1. No need to test
	2. Termination of pregnancy is not an option
4. The importance of education	1. The need for education regarding aneuploidy risks
	2. Paying it forward
5. A service with a purpose	

THEME 1: LACK OF PREPAREDNESS

During the interviews it was noted that the participants all received some information regarding the reason for referral to GSH but were generally unprepared for the age-related risk information that they received from the counsellor and were caught off guard when they realised that their appointment would be in the form of a GC session. This lack of preparation also extended to participants' expectations regarding their PCC appointment as no participant had any GC-specific expectations, but several participants had other health care expectations. The sub-themes discuss the role that the referring clinic played in participants' lack of preparation as well as how it affected their expectations.

Sub-theme 1: Limited information from referring clinics

The participants were referred to PCC by their local MOUs. The data shows that the referring clinics provided varying amounts of information. In some cases, participants were only told that they were coming for a scan and had to be informed of the age-related aneuploidy risk at PCC. This was the case for participant 6 who said the following:

> Participant 6 (20 weeks gestation, declined IDT): "*So they told me that I came, I have to come here to do ultrasound, because they need to know exactly how many weeks am I, how far I am with the pregnancy. So ja*[1] *I came here, but when I got here the sister explained that uh, that the the reasons, the the actual reasons that I have to do the ultrasound is to check if the baby is OK if the baby does not have a syndrome, Down syndrome disease."*

In contrast to the participants who were completely unaware of the age-related reason for referral, other participants reported that their referring clinics explained that they were being sent to GSH as a result of their age. Despite this knowledge, these participants were still unaware of why their age warranted appointments at a tertiary hospital and were generally not informed that they would be undergoing GC. This is illustrated by participants 1 and 7 who had the following to say about their knowledge prior to arriving at PCC:

> Participant 1 (17 weeks gestation, declined IDT): *"I just came here because they referred me to Groote Schuur because of the age…, just because of the I am 42, maybe there is risk of… I don't know. They said I must come here, just because of the age… They never mentioned anything about counselling."*
>
> Participant 7 (20 weeks gestation, accepted IDT): *"[referring clinic's name] hospital sent me because I turned 42 years old last week and I'm 20 weeks pregnant so they sent me here for a genetic date as well as a marker scan uhm, … cause they say I am, as I say to them in my own words that I am above the pregnancy age, so that's why they sent me here… They said yes that uhm, what was that one nurse's exact words? We have to send you to Groote Schuur because you are older than 38 and anyone who is older than 38 has to attend either Groote Schuur or Somerset as their first appointment… Nothing further*

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[1] Afrikaans word for yes. In this case used as slang. Similar to using "yeah" in English.

> *than that… Just that. The doctor didn't even say anything to me…Doctor wrote down there a genetic date and a marker scan and I said doctor what is that? No they will explain at Groote Schuur for you. I said ok fine seeing as you can't explain I will find out by Groote Schuur how it is"*

Participant 7 makes mention of a "genetic date", but from her quote it seems apparent that she did not know that it would involve GC. She also explains that she did not receive more information when she requested it at her referring clinic and seems to have felt frustrated by the healthcare professional who would not answer her questions. She later explained that she heard from medically trained individuals in her community that there is a test that is offered to AMA women, but she still did not know that it was to test for aneuploidy or that this was the reason for her referral to GSH. It is also interesting to note that she later explained that she herself chose the words "above the pregnancy age" as a result of a fictional television program. She was not aware of what the age-related pregnancy risks are and also mentioned that she did not take any of it seriously at the time due to the fact that the TV program was fictional.

A few other participants had a different experience with regards to the information they received at their referring clinics. This was the case for participant 5 as a "friendly" nurse took the time to talk to her about DS and IDT. Despite being aware of her risk of having a child with DS, this participant did not know much more about what DS entails and only understood that she should accept IDT on the recommendation of the referring clinic. She was also unaware that she would be talking to a trained healthcare provider about DS and how an affected child might impact her life.

> Participant 5 (21 weeks gestation, accepted IDT): *"Ok and then after that even at [referring clinic's name] they did tell me about this syndrome thing… They said if you are 30 years upwards there's a chance that you get a baby with the syndrome. Hm… That's all they told me so they advise that you must do the test… they didn't mention anything about counselling... In the clinic…I only know those special need, those babies are special needs babies uh, even if, if you are working. So you must, most of the time you are gonna miss work you have to take the baby to the clinic. If it's a clinic if it's uh some of them they go to physio or depends where you have to go, but I know it's a special need baby. You must most of your time you have to dedicate to that"*

The data therefore suggests that despite the varying degrees of information regarding age-related aneuploidy risks, this cohort was not informed of the scope of their appointment prior to arriving at the clinic and the women were uniformly unprepared for their GC experience.

Sub-Theme 2: Expectations unrelated to genetic counselling

All participants in this study were booked for an ultrasound and GC appointment. There were significant expectations for an ultrasound but as none of the participants were aware of the nature of their GC appointment they did not have any prepared expectations of the GC service. From participants' quotes it seemed as if they only fleetingly considered the referral. This is illustrated in participant 7's quote, who heard about a "genetic" appointment at her referring clinic but was not informed about what it would involve.

> Participant 7 (20 weeks gestation, accepted IDT):*"I didn't think in that moment anything of it [the genetic date that the referring hospital wrote in her clinic card] No, that doctor said it's two scans, that's why I didn't think anything… So I go OK fine, a genetic date is seker a scan en 'n marker scan is maar a scan [translation: a genetic date is probably a scan and a marker scan is but a scan]. And I left it. It never crossed my mind again. What it could be or what it should be"*

Another participant also illustrated not dwelling on the referral and verbalised her absence of expectations as follows:

> Participant 1 (17 weeks gestation, declined IDT): "*No actually when they said I must come to Groote Schuur, I didn't think about, I thought ok maybe I am going to get taken care of because maybe I need more extra attention because of age so I thought maybe… at Groote Schuur, maybe there's more attention at the hospital."*

This quote highlights participant 1's expectation to receive extra attention at GSH. In a previous part of the interview this participant stated that she was happy to come to GSH as she thought that the doctors there were better qualified. This participant was not the only one in the cohort to think that. Despite understanding that they would receive extra care as a result of their advanced age, these women were still not prepared for the scope of GC.

Despite the lack of contemplation and expectation regarding the “genetic date” on their appointment cards, several participants had other healthcare and pregnancy related expectations which were often based on experiences from previous medical services at other clinics. The most pertinent expectation seemed to have been having an ultrasound. Other expectations included blood pressure tests and finding out the sex of their child.

> Participant 5 (21 weeks gestation, accepted IDT): *“[The] MOU, say I must come for ultrasound and then I did and for me the only thing I wanted to know is the gender. Because I wanted to know if it’s a boy or a girl so I found out it’s a boy again so I will have another. Three boys”*
>
> Participant 6 (20 weeks, declined IDT):*"You see? So because they only told me about the ultrasound. I knew OK I am going to do only one thing and then I have to go back home"*
>
> Participant 1 (17 weeks gestation, declined IDT): *“Uhm no I was expecting to do the blood, like blood pressure tests. I didn’t expect to come for such kind of counselling.”*

An extract from participant 4’s interview highlights the emphasis that she placed on having an ultrasound at PCC as she said that if she did not know that she would receive an ultrasound at GSH, she would not have attended her appointment.

> Participant 4 (20 weeks gestation, declined IDT): *“I wouldn’t come [only for counselling without a scan]… because they can also give me that side [at the MOU] counselling. Why must I come so far to come for counselling you see”*
>
> Interviewer: *“And if they can’t give it to you there?”*
>
> Participant 4 (20 weeks gestation, declined IDT): *“No I would still not come”*

Some participants had time at PCC between finding out that they would talk to a genetic counsellor/nurse and being counselled to consider what this would mean. Participants generally realised that their GC appointment was an independent appointment from their ultrasound when a nurse at PCC told them to go back to the waiting room where someone will call them for their GC appointment. In these cases participants sometimes drew from their other real-life experiences with counselling, which mostly seemed to have been related to HIV. Participants 4 and 7 addressed this, and participant 7 even told the researcher that she expected it to be boring based on her experience with HIV counselling.

> Participant 7 (20 weeks gestation, accepted IDT):*"You know where, we go to we also have this counselling there by us this uhm like a HIV counsellor thingy. Like so a counsellor thingy. I thought it's just gonna be that. You know a two-minute noodle thing, that's what we call our HIV… It's what we call our HIV counsellors there in [name of area where she attends her local clinic], a two minute noodle thing because it's like bla blab la blab la bla bla, OK. Thank you bye… Uh-huh, they so dead it's not even funny… I thought it was actually gonna be dull and dreary like it usually is, dull and dreary. I mean I've got absolutely nothing to compare her [the counsellor] with"*

> Participant 4 (20 weeks gestation, declined IDT): *"No I thought it was for the time when they, we heard [at Groote Schuur] we were gonna go for counselling, I thought it was gonna be for HIV. That, that's what I thought about"*

Participant 4 went further by questioning why she was not given more information at her referring clinic.

> Participant 4 (20 weeks gestation, declined IDT): *"To me it was just like why they didn't tell me in the beginning there by [name of referring clinic] that I will come for this? They didn't explain to me I am gonna come for counselling, they only said to me it's because of my age. I am gonna take another ultrasound because of my age. They are sending me here for another scan. Just to make sure that the baby is alright. They didn't mention anything about that… They were supposed to tell me that I must come here for a counselling and they will talk about DS and something like that"*

This sub-theme illustrates that the lack of preparation often went further than only a lack of information as participants did not always give their referral much thought. In the instances where participants had time to consider what to expect at their appointment, their expectations were sometimes based on limited previous experiences with other types of counselling that is unrelated to genetics and this did not result in adequate preparation for GC.

THEME 2: AN EMOTIONAL EXPERIENCE

The participants in this study often used emotive language to describe the experience of realising that their children could be affected with DS. The emotional experience did not end with learning about the age-related aneuploidy risk, as emotive descriptions were also used to describe their experiences with other parts of the counselling session, specifically after having a discussion about their specific age risk for DS. They sometimes also reported feeling reassured and optimistic after the counselling session. This theme addresses the range of emotions that participants experienced as a result of undergoing GC and other personal factors.

Sub-theme 1: A Stress-inducing situation

Most of the participants described feeling some distress during the process of GC. The main stress inducing factor was the possibility of having a child with aneuploidy as well as the desire for a "normal" child. This is illustrated by two participants who both said that they were scared when they found out about the age-related aneuploidy risk and did not want a child with DS.

> Participant 3 (21 weeks gestation, declined IDT): *"Jo[2] I find out [about the age-related risk] and then I am very scared that time… I am scared because I don't want to deliver the baby [if it] is not right… I want my babies normal like everyone, any baby, I don't want to deliver the baby [if] it's not right"*

> Participant 6 (20 weeks gestation, declined IDT): "*Yes I did panic from the first time that they told me about the disease and the uh, the risk of that it could happen to me since I am, I'm, I'm older… Jo (laughing) that I could have a, a, a baby who has the syndrome, I don't think anyone would want that for themselves so I was scared a bit"*

For some participants, the negative emotions that they experienced were largely as a result of them being unprepared to hear about age-related aneuploidy risks. This shows the impact that the lack of information and preparation discussed in theme one had for some and is highlighted by participant 4's quote below.

> Participant 4 (20 weeks gestation, declined IDT): *"I think that [not knowing that her appointment was for genetic counselling to speak about Down syndrome]*

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[2] A South African expression of surprise or disbelief

> *was actually what was making it so nerve-racking ja… Because I wasn't like in, I didn't prepare myself for it, you see"*

From the quotes in this sub-theme it seems as if the participants had a difficult time hearing about age-related aneuploidy risks and that different factors, such as being unprepared and the fear of having a child with DS contributed to the emotions that they experienced.

Sub-theme 2: The glass is half full

Despite the anxiety discussed in the previous sub-theme, it appears as if the participants often left with an overall feeling of optimism about their pregnancy. The reasons given for this optimism were diverse, including finding no concerning features on their ultrasound, faith, their perception of personal risk being low as well as previous exposure to individuals who were pregnant over the age of 40 who gave birth to children who were not affected with DS.

Some participants found comfort in the outcome of their ultrasounds and believed that their babies would be unaffected or healthy as there were no indications of DS on their scans. This was the case for participant 3 who reported initial worry that seemed to have disappeared when her scan showed no soft markers or foetal anomalies.

> Participant 3 (21 weeks gestation, declined IDT): *"Yeah I am worried about the age that time she didn't tell me, but now I am fine… Because the something change, I find my baby there in the scan, I find nothing wrong with my baby"*

The quote above is important as it not only shows the relief provided by the scan, but also speaks to an underlying concept that was coded as "misunderstanding" in several participants' interviews. In these interviews, and specifically in participant 3's transcript, it seems as if these participants believed that if there is no abnormality noted on their scans there is no risk of their child having DS.

Participant 6 reported similar optimism after her ultrasound results.

> Participant 6 (20 weeks gestation, declined IDT): "*She [the counsellor] assured me that OK I can see that the result of your scan, it shows that your baby is perfectly fine … she told me that there's no sign, my, my baby can have a syndrome so I, I me as a counsellor I am satisfied that you, the, there's 1% chance or lesser to have that. It sound good, because if there was more, more*

> *than 1 I think I would, I wouldn't be happy with that so I think it sounds good and promising"*

Participant 6's quote not only describes similar relief indicated by participant 3, but also illustrates the security that her personalised risk figure provided. In this case she understood that there was still a risk of her child being affected with DS despite the normal ultrasound, but she considered the risk to be low. She was not the only participant who found comfort after discussing her personal risk with the counsellor, as further highlighted by quotes from other participants' interviews.

> Participant 1 (17 weeks gestation, declined IDT): *"Yeah, I feel happy because I am between 99% of a chance of not having… Yes it was a good day… Because out of 85 only 1 can have and 84 cannot so"*
>
> Participant 2 (21 weeks gestation, declined IDT): *"[I was] More relaxed because she said it would be 1 out of 79 chance."*
>
> Participant 4 (20 weeks gestation, declined IDT): "*[A risk of 1 in 94 for Down syndrome] It's not really that, how can I say, not that serious to me."*

From the quotes above it seems as if these participants are perceiving their risk of having a child with DS to be low. The much higher probability of an unaffected child is contributing to their belief that their child would be unaffected and allows them to be optimistic. Thus similarly to participant 6 above, the participants understood that the risk of DS is not zero but they believe it to be low enough that they did not need to be anxious about it. This would suggest that prior to knowing the exact age-related risk these women thought the risk to be higher as they were anxious about the possibility of an affected child. In keeping with the relief provided by the "small" risk, participants also reported feeling optimistic because they knew of people who had healthy babies when they were 40 or older and that anyone could have a child with DS, even someone who is younger.

> Participant 2 (21 weeks gestation, declined IDT): *"[I felt more ease] Because they said young people could have the same problem."*
>
> Participant 4 (20 weeks gestation, declined IDT): *"But at least I can say, how can I say, in my situation because I have seen ladies who is over 40 years with normal babies. Ok the babies is just a little bit hyperactive (laughing). But that's all, but they are normal… But now I am thinking what is making me different*

> *than them, you see? Ja… Yes, and my sister she did get a baby at 45… That baby is normal"*

Another factor that gave participants hope was their faith. This is illustrated by the extracts below.

> Participant 1 (17 weeks gestation, declined IDT): *"Yeah I think if I didn't have faith I was gonna be very worried yeah I was gonna be very worried, but now because I have faith that I... to me and also the percentage, so 99 [%], only one out of 85 I think"*
>
> Participant 4 (20 weeks gestation, declined IDT): *"It's not really that, how can I say, not that serious to me (her risk to have a child affected with DS)… Because I really believe that my baby is healthy. I really believe that…(because of) the man upstairs… I think I, if I didn't believe né*[3]*, I think I would do the test if I didn't believe that my baby is healthy, but I believe my baby is healthy… Because sometimes our faith are so small. We don't believe actually, but I really believe my baby is healthy. Because God won't give me another chance, because this is now a third chance to fall pregnant. If it wasn't supposed to be then I wouldn't fall pregnant"*
>
> Participant 7 (20 weeks gestation, accepted IDT): *"I sat downstairs at the first reception as you come in… I was looking at everyone like that. I was thinking OK so I told myself God won't put something in your path that you know you can't handle"*

Participants seemed to have gone through a process of transformation with regards to their emotions during their time at PCC. This sub-theme discusses the factors that contributed to participants feeling less anxious about their age-related DS risk. It also illustrates how the information they received from the genetic counsellor allowed them to consider what exactly an age-related risk means for them personally and how it relates to their personal context and experiences. It is therefore important for genetic counsellors to understand that women process information continuously throughout the session and to allow them time for this process.

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[3] Afrikaans colloquialism for "right" or "isn't that so"

THEME 3: DECISIONS REGARDING THE WAY FORWARD

This theme speaks to the belief of the majority of participants that they did not feel they needed to intervene after the information that they received during GC. This attitude extended to both deciding about IDT as well as considerations regarding TOP. Participants gave a variety of reasons to explain their decisions.

Sub-theme 1: No need to test

The decision about whether or not to have an amniocentesis was a pertinent point of discussion. Indecisiveness and distress were reported by participants about the miscarriage risk and the decision-making process. The majority of participants declined IDT and reasons cited included a variety of motives such as the belief that the foetus is unaffected and the optimism following a normal ultrasound result. Participants described some distress surrounding the decision-making process and it seems as if much of this was based on their fear of having a miscarriage.

> Participant 4 (20 weeks gestation, declined IDT): *"[It was nerve-racking] Cause I was in a, a, I was in two. I didn't know if I must take the test or must I leave the test. But then I prefer to leave the test, and I believe that my baby is ok"*

> Participant 5 (21 weeks gestation, accepted IDT): *"Jo you know, I get worried thinking that uh, I am worried about the test because she mentioned something that there must be a miscarriage. There's only one in fif, 500… Yeah so that's… you fear that what if, but my God he will never allow that"*

> Participant 7 (20 weeks gestation, accepted IDT):*"Yoh I cried at first, and [counsellor's name] asked me how, what was it, miscarriage, possibility of miscarriage, I cried, oh jinne*[4]*, hm-hmmm. I've never had a miscarriage in my life"*

Similar to participant 4 above, other participants were also indecisive about testing initially. In some cases, the deciding factor was the outcome of the ultrasound which gave no indication that their babies were more likely to have DS. This is illustrated by the quotes from participants 3 and 6.

> Participant 3 (21 weeks gestation, declined IDT): *"I say me, the first time I say yes [to amniocentesis], she ask me why you want to check but you see the scan,*

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[4] An Afrikaans expression that is an exclamation of shock or surprise

> *she see the baby is fine? I say uh-hu no there's no need to check, cause I saw, the baby is ok… Everything is ok. But the first time I say I want to check, but now I say ah-ah there's no need to check because I saw my baby is fine"*
>
> Participant 6 (20 weeks gestation, declined IDT): "*And then they told me again about the risk of doing the test. That it's only 1 in 500 that you can get a miscarriage from doing the test. So the other thing is that they said my ultrasound, ehh, the scanner does not show any sign that the baby might have the disease so I am thinking that I, I don't need to do the test. Looking at the result of the ultrasound and the result of the questionnaire of family history"*

The family history that participant 6 is referring to above, is the one taken by the genetic counsellor at the beginning of her counselling session. It is interesting that she considered this when deciding whether or not to undergo IDT and this highlights how different aspects of her PCC experience affected her decision-making.

Other reasons for declining IDT included acceptance of a child with DS, and the belief that knowing that their child was affected would take the joy out of the pregnancy. The quotes below demonstrate these reasons.

> Participant 1 (17 weeks gestation, declined IDT): "*It's not important for me to get tested. I think whatever if it is a Down syndrome or it's not a Down syndrome, it's my baby."*
>
> Participant 4 (20 weeks gestation, declined IDT): *"Uh, she [the counsellor] was OK with it. But actually she wanted me to take the test. But I didn't want to take the test… Because I want to enjoy my pregnancy… Yes, I don't want to be worried about that or thinking about that now… It will mess up everything. I don't think I will feel about the pregnancy the way I am feeling about it now"*

As with all research projects there were participants with contrasting views and two of the participants accepted invasive testing. Participant 7 explained that she has four other children to think about and she had to prepare her family if they were going to have a child with DS. Her quote also illustrates the uncertainty that would have persisted if she didn't accept the amniocentesis and how it would have been present throughout her pregnancy. This seems to suggest that the participant has a need to know definitively whether her child will be affected with DS and is using the GC-service to fulfil this need. The quote also conveys some of the consideration that goes into making a decision about prenatal testing, as well as the responsibility that this places on the women at PCC.

> Participant 7 (20 weeks gestation, accepted IDT):*"So for me to say ag*[5]*, I am not going to worry with this amnio man, I know everything is gonna be fine, but then in the back of my mind, it is going to nibble here… Eventually it's gonna come to the forefront. Then the day I give birth and there is actually something wrong with it, what do you think the response of my kids is gonna be, and my husband's gonna be when I tell them that I have the choice to find out… So I can't just decide for myself, I have to decide for all of us… No then, irrespective of how I feel about, I might get a miscarriage and whatever and whatever, I want to have this [the amniocentesis] one… I want to have it done, it's a… for myself and my family, I want to have it done… You can't dump a surprise on a family, on the day the child is born, you can't. That man is gonna tell me you knew all along here the divorce comes. You knew all along and you kept quiet for how many months."*

This sub-theme provided valuable insights into the factors that influence participants' decisions regarding IDT. One of the reasons for offering IDT was that women with affected children would be offered the option to terminate their pregnancy, but most of the participants did note that they would not consider a TOP. This is the subject of the next sub-theme.

Sub-theme 2: Termination of pregnancy is not an option

As the option to terminate a pregnancy affected with DS was discussed during the participants' counselling session, it was one of the subjects that emerged during the participants' interviews. Analysis of the interviews suggested that most of the women in this study would not personally consider TOP. Several of them believed it was wrong to terminate a pregnancy while others cited their acceptance of any child and the belief that their baby would be fine.

> Participant 1 (17 weeks gestation, declined IDT): *"Actually, me I didn't mind about the Down syndrome stuff because I was thinking that's something that can happen anytime. If it happens to you that's your baby so you have to keep it…Because for me it like to take out the baby it feels like killing… it feels like*

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[5] Afrikaans dismissive expression

> *killing, so I don't think… I am fine with whatever is happening to me. If it's a Down syndrome or it's not a Down syndrome I think I am fine with it"*
>
> Participant 4 (20 weeks gestation, declined IDT): *"It is, it is, it's not nice to hear that [abortion options], hm-mm. Because I want to go on with my pregnancy… And I believe there's nothing wrong with my pregnancy."*
>
> Participant 3 (21 weeks gestation, declined IDT): *"But it's [abortion] not easy to do, it's not easy to take your baby out. Sometimes you're gonna take*[6] *[keep your baby] like that… Ah it's my baby I am gonna take [keep] it, cause it's my baby"*

The quotes by participants 1 and 3 hints at a broader acceptance of a child affected with DS. This thought is echoed by participant 4, who understands that a child affected with DS is different from other children, but still considers the child to be "normal".

> Participant 4 (20 weeks gestation, declined IDT): *"It's just that something is a little bit different about them towards the other kids. But to me they look normal*[7]*"*

Only one of the two participants who accepted IDT mentioned that she would consider a TOP. This participant, a working mother whose moral code and faith were primarily against abortion, believed that her circumstances would make it impossible to raise a child with DS. Despite her realisation about practical implications of a child with DS, this would not be an easy decision for her to make.

> Participant 5 (21 weeks gestation, accepted IDT):*"Jo, it's not easy because I am a working mother and then I have other kids, so the only thing I will think of terminate the pregnancy… Yes because jo it's going to be taking too much of my time…Hm cause I have to look after, this baby he will have a special need. So sometimes I won't have time I have to work, I still have this two, so it's going to be too much. That's why I said everything depends to the results. But I know my decision already… It's not good but you have to…You don't feel good but at the end of the day you're thinking about the future, you're thinking about the other people, but involve in your life, so at least I have two healthy boys. So to me it's not nice to kill, but you have to terminate it… It's a tough one…My faith*

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[6] In this context "take" differs from the first sentence and refers to the fact that the participant will keep her child, rather than abort it

[7] In this case "normal" refers to being physically normal

> *impact a lot but terminating it's against my religion, but at the end I am the one who is gonna suffer… My God knows everything he knows everything, but I have to take that decision…It feels bad man, because you are killing someone. Maybe that kid can change or can be normal one but there's ncinci*[8] *possibilities or to be like that but it it's bad but you have to take that decision… It's not a good one but at the end you must look at my picture and say there's one, two, three [referring to chromosomes at position 21] and the way I work I travel a lot around Cape Town so it's gonna be another thing, who's gonna look after the kid? Until I came home past 7? You see I wake up early so it's gonna be very difficult for me"*

The quote above not only illustrates the complex emotions and considerations regarding the termination of an affected pregnancy, but it also shows how this participant accepted IDT so that she would have the option to consider a TOP if the child is affected. It therefore illustrates how the topics in these two sub-themes can influence each other.

THEME 4: THE IMPORTANCE OF EDUCATION

Different aspects of education was a topic that frequently came up during the interviews. It became apparent that the participants in this study were not informed about risks of AMA pregnancies and that the participants themselves thought that their communities were not sufficiently educated regarding this topic. The participants believed that this is important to address as it would allow for informed decision-making when women choose to fall pregnant at an advanced age. The participants also gave several suggestions to address the lack of knowledge in their communities and specifically discussed their responsibility to tell other women about these age-related risks.

Sub-theme 1: The need for education regarding aneuploidy risks

Several participants commented on the fact that they did not know about the aneuploidy risk with older age prior to their appointment. Some of them explained that they knew there were risks related to pregnancy when you were older but they did not realise that this included children with DS or other aneuploidies. This is illustrated by

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[8] An isiXhosa word meaning little

the extracts from two participants' interviews below where they explain that the day of their PCC appointment was the first time they realised that they were at risk of having a child with DS.

> Participant 3 (21 weeks gestation, declined IDT): *"Cause today is my first time to see the baby, you see, it's the first time for someone to told me if you're old you get a baby like that [with Down syndrome]"*
>
> Participant 6 (20 weeks gestation, declined IDT): "*No, I knew that mostly women when they fall pregnant they are older, there's lots of risks, but I didn't know exactly what"*

It seems as if the participants' personal lack of knowledge resulted in them believing that there was a similar gap of knowledge in their communities at large. Some participants suggested that filling this educational void would empower women to make active choices about pregnancy at an older age. This is supported by participant 1's belief that if people were properly educated, she thought it would help them to actively decide whether a pregnancy at an advanced age is something they wanted to have.

> Participant 1 (17 weeks gestation, declined IDT): "*Yeah it's [the service] very good. I wish they can really teach about that maybe at the schools… I think you can also do that maybe, before it happens for people so for someone to get pregnant if you can catch them in time…It helps so you can choose if you want to be pregnant at old age or not*"

Several participants expanded on the impact that increased awareness in local communities could have by explaining how it would have affected their personal choices and behaviour. This is evident in extracts from interviews of participants 3 and 6.

> Participant 3 (21 weeks gestation, declined IDT): *"Yeah I know the age is the problem… if I know before [falling pregnant] even me I am gonna stop to be pregnant"*
>
> Participant 6 (20 weeks gestation, declined IDT): "*[If people knew about the age-related risk of aneuploidy] they will be careful I think, they will play safe. They will be careful. I am guessing they won't be pregnant at that age. Like right now I am thinking of, of, of uh what do they call it when you when they take out my womb [hysterectomy]… I am thinking of doing that cause I don't want any*

> *mistake in the future of falling pregnant and it's a mistake. I don't want that mistake… Like falling pregnant because really I…Yeah I did not plan it [the pregnancy], truly speaking I did not plan it."*

When asked what she would have done differently had she known about the age-related aneuploidy risk, participant 6 went on to explain that the information would have helped her to make a final decision about having a procedure that would prevent her from having any more children. She explained that the information would have silenced any doubts in her mind that were keeping her from taking such a definitive action.

> Participant 6 (20 weeks gestation, declined IDT): "*I think I would have made sure that I, I do the hystte [hysterectomy]… Yes I think it [the information regarding age-related aneuploidy] would have helped me to make a final, you know you make a decision and then you have this, uh doubts at the back of your mind. But when you know, when you, when you know more and you, you have facts. Like you have plenty of reasons to do what you want to do"*

As a result of this lack of education regarding age-related aneuploidy risks, there was a sense among some participants that they have missed out on the opportunity to be health literate about this subject. This is illustrated best by participant 5's explanation of the missed opportunities in her local community.

> Participant 5 (21 weeks gestation, accepted IDT): *"I feel robbed, Because we don't get chances, or opportunities to be educated about most of things. We only get some few things in our location so I think it's high time now they go out and educate people. Like now, I think after I did the, did the uh, what do you call this now? The papsmear – now they educate people about papsmear. People know that there's HIV, there's papsmear. So now they need to educate more about this so it's high time they go out and educate."*

She offered the following suggestion on how to address the problem.

> Participant 5 (21 weeks gestation, accepted IDT): *"It's not right, they're supposed to tell people or you, your organization must go out there. Don't stay here at Groote Schuur. Go out to our public clinics, so that people know about this. Because most of the women, in our culture you must get pregnant when you are older, not when you are young so now it is going to be different you*

> *see… That's why I said you must go to our clinics so that people will know about this"*

This quote speaks to the role that genetic counsellors and public health campaigns can play in educating local communities. This participant felt that counsellors should make more effort to reach women in their local communities because they would not reach these women if they only stay at GSH.

This sub-theme highlights the need for educational programs that inform women of AMA-related pregnancy risks. Additionally, it also shows the impact that the participants believe information could have on women's choices regarding pregnancy.

Sub-theme 2: Paying it forward

One of the striking results during this project was that when asked how they would use the information that they received during their GC appointment, many of the participants said that they would use it to inform and educate other people. They often discussed how they didn't want other people to have a similar experience of finding out about this age-related aneuploidy risk when they are pregnant already and explained that they would pay it forward by using their knowledge to educate other women.

> Participant 1 (17 weeks gestation, declined IDT): *"I think the service is good because some of the people they don't know about it. It's so good because maybe I can also let other people know about it the risk of having a baby at old age"*

> Participant 5 (21 weeks gestation, accepted IDT): *"I think the best thing is to pass it to other people…tell them what's happening especially the young girls, and then I will also advise them to have a, to have kids at an early stage, 30s so that they are safe from the, it doesn't say that the baby can't have the syndrome if it's, but when you are older, the risk is more… And people they just make babies they don't even think about this syndromes. This there's a syndrome sickness or syndrome what... so to me now I will preach the gospel"*

> Participant 6 (20 weeks gestation, declined IDT): "*[I am going to use this information] By telling other people the whole thing about the, the disease, the syndrome uh the whole information that I got today. I am going to share it with other people…Like I said that I think there is lack of uh this kind of information*

> *getting to people, to other people like me I didn't know about it before. So I came here I got the information so I think it is wise or it is I'll be doing other people out there justice to tell them about the info… because I think I, I it is important for me that I know this info so why not share it to other people."*

> Participant 7 (20 weeks gestation, accepted IDT):*"If someone should fall pregnant that I know of that is of my age, I want to inform them… Because I, no one gave me the chance to know. And I don't want someone else to go through that."*

Some participants, like participant 6, explicitly expressed concern about other people who, like her, are not aware of these risks and said that she believed something needed to be done. This statement echoes participant 5's statement in the previous sub-theme regarding the lack of opportunities to learn about these healthcare risks in her community.

> Participant 6 (20 weeks gestation, declined IDT): "*Ja, I think it's a, it's a problem really in our society because we need to know about these things I don't know if it's ignorance or we just don't get information enough, but I feel bad. I am thinking it's most women at my age they don't know… So I think it's bad, something has to be done to get information out there"*

From the data in this sub-theme it seems as if the participants gathered valuable information that they want other people in their communities to be aware of. The participants explained that they did not want anyone else to have a similar emotional experience in only finding out about age-related aneuploidy risks while being pregnant and were therefore willing to go and discuss their experiences with women in their communities.

THEME 5: A SERVICE WITH A PURPOSE

The final pattern that became clear after analysis, was that the participants believed that the GC service at PCC had a purpose and that they appreciated the care they had received. This appreciation is illustrated by participant 4 who listed several things she appreciated about the GC service, including the information she received and the warm tone that the counsellor used with her.

> Participant 4 (20 weeks gestation, declined IDT): *"No it [GC] helped, and I am alright with that. I appreciate it actually, yes… it was worth it [coming to GSH],*

> *At least now I know more about it [the age-related risk]… she [the counsellor] didn't make me feel like I am an outsider like that. No she was actually really friendly towards me… I was like speaking to a friend"*

Participants 5 and 6 had similar thoughts as illustrated below.

> Participant 5 (21 weeks gestation, accepted IDT): *"[If asked to explain what genetic counselling is] I'll say to them the genetic counselling is, firstly they educate you about the pregnancy, and then later you have to take decision if you want to do the test or not. But before everything they give you counselling, they talk to you, calm you down, tell you there's a risk, there's no risk. They tell you everything, they educate you about this…You know when they call you, you have that thing… maybe my baby is… and that when she is uh, you have this syndrome and then she showed me those numbers and the cells, everything. How [it] must be, so at least I have the knowledge about this syndrome… Yes to me it helps because now I have an idea what is happening"*

> Participant 6 (20 weeks gestation, declined IDT):*"Well I am happy, because I wouldn't have got this information if I didn't come here. Yes I knew about this syndrome, DS, but not, not into details. So at least now I know more about the disease. So I am happy...About the info that I got and the service that I got yes...But I feel that they did their, their job well by explaining, which is something that I didn't know before… They [genetic counsellors] are very polite people, they take time to explain things into details, they have patience. They don't just do their work just for the payroll purposes. It seems like they are doing their job for they chose, they want to do this job and that's what they wanted to do. They are not here for the money"*

In some cases participants believed that the information they received during GC is important to emotionally prepare individuals in case they do have a child with DS. This does not refer to practical preparation, but rather to preventing emotional distress due to an unexpected diagnosis. This thought is illustrated by extracts from participant 1 and 2.

> Participant 1 (17 weeks gestation, declined IDT): *"I think it's good, if you know of what to do and what not to do. I think it's important for someone to have a choice of what you want. Like when they told me about getting tested… I think*

> *it was going to be painful if I didn't know about all this information, but now, because I know about all this, there is a chance of that. But 99% of chance I have got hope that it is not going to happen to me… Yes, I think if you didn't have this information then if it just happens, I think it can hurt you."*
>
> Participant 2 (21 weeks gestation, declined IDT): *"At least now I know that this could happen, if there is gonna be problems I can handle it more"*

Other participants addressed the importance of practical preparation for the possibility of having a child with DS. Participant 7 articulated her thoughts about how a child with DS would have a profound impact on other family members and would have implications on their lifestyle. She believed that it is important to prepare for such a change and that the service that she received was therefore useful.

> Participant 7 (20 weeks gestation, accepted IDT):*"[I'll use this information to] Prepare myself. And the family as well, prepare them and knowing because if we have a child, like I said a child with Down syndrome, it's not a child whose gonna leave home at the age of 18, go and study further or go and work whatever… It's a child for life…And that needs preparing, because you can't just say for someone who doesn't know or doesn't want to do the test and now it comes out that you actually have this child… so I want to be informed… With information comes knowledge and so you learn so I can teach my kids, ok this is what is going to happen… It's a possibility of this happening, there's a risk of that happening, I can tell them so we can all as a family prepare ourselves… it's going to change our lives for good… It's not a temporary change, it's for life, so who wouldn't want to be prepared for that?"*

The participants seem to suggest that GC is a valuable service as it allows patients to mentally and practically prepare for the possibility of a child with DS. It also seems as if the participants were grateful that they have had the opportunity to learn about age-related aneuploidy risks. The participants also reported appreciation for the way in which the counsellor addressed and treated them which might suggest that the participants had different experiences with some other healthcare providers.

The results indicate that the participants' GC experience is influenced by more than just GC itself. Factors that influenced the experience included information from the referring clinic, lack of prior knowledge in local communities and the respectful manner in which counsellors treated participants. The results also indicate that it was an emotionally taxing experience to find out about age-related aneuploidy risk, but that the GC process and other factors left participants with a sense of optimism regarding their pregnancies. Generally, the results suggest a positive experience of GC that resulted in participants feeling they had been educated about the subject.

CHAPTER FOUR: DISCUSSION AND CONCLUSION

This study was conducted with the aim of answering the research question: What are the views and expectations of pregnant women who undergo GC for age related risk of aneuploidy at GSH? As shown in chapter three, this research project revealed multiple themes that touched on a diverse range of subjects relating to the participants' experiences at PCC. While each theme is a distinct entity in its own right, they collectively tell the story of women who are referred for GC at GSH where they have to come to terms with the realisation that their advanced age puts them at risk of having a child affected with aneuploidy. This chapter will take a holistic approach to see how these themes answer the research question as well as how they relate to existing data. The study's strengths and limitations as well as the implications that this project can have for the GC service at PCC and referring MOUs will also be discussed.

All participants in this study were referred to the GC service from a secondary level hospital or local clinic where they usually receive their antenatal care. From the results, it would seem as if the information received during the referral process is a major factor in setting the stage for the participants' experiences at PCC. This is in keeping with results from another South African study that aimed to understand the GC experiences of black mothers in Johannesburg who had children with genetic conditions. That study reported that participants' perceptions of GC was influenced by, among other things, interactions with other healthcare workers (Morris et al., 2015). For the most part, participants in this study appeared to have received very few details about why they were being referred to GSH and were unprepared for GC. The previously mentioned South African study revealed a similar absence of awareness regarding GC and participants' reason for referral to genetic services (Morris et al., 2015). The majority of the current study cohort understood that the reason for referral was related to the fact that they were older, but very few were aware of the implications that their age could have for their pregnancies or that DS would be a prominent point of discussion. This lack of information from the referring clinics was a clear source of frustration for some participants. This is evident in the hint of defiance with which one participant declared that if the doctor at her referring clinic did not want to give her the necessary information, she would go and find it herself at GSH and another participant explaining that more information from the referring clinic would have been helpful to her understanding of what she was letting herself in for.

From the data it seems as if participants believed that the uncertainty regarding their referral to PCC was as a result of insufficient communication at their referring clinics. However, it is also possible that the problem partially resulted from participants' inadequate prior exposure to information regarding age-related aneuploidy. The previously mentioned South African study that included a cohort of 13 black mothers who could converse in isiZulu, seTswana, isiXhosa or seSotho and had young children with genetic diseases revealed that their communities were often oblivious to genetic conditions (Morris et al., 2015). The few participants in this cohort who were aware that there were risks associated with AMA were not able to verbalise exactly what those risks are and most participants did not know that DS was a specific risk. This is in contrast to a Canadian study of first-time mothers who knew that the risk for chromosomal abnormalities increased with age, despite not being as knowledgeable about other AMA risks. This study also reported a positive correlation between health literacy and knowledge (Sheinis et al., 2017).

A study done with a cohort of isiXhosa speakers recruited in the Eastern Cape focused on developing a tool to measure health literacy in SA and reported a significant link between health literacy and education level as well as the ability to speak and read English (Marimwe & Dowse, 2019). Their conclusion was not evident in the current study where although the average level of education was higher than the average for the City of Cape Town (Statistics South Africa, 2011) and all participants were able to converse in English, health literacy about AMA did not correlate with these factors and this left some participants feeling disadvantaged. This is particularly highlighted by one participant's explanation of how she feels “robbed” that she did not have an earlier opportunity to learn about age-related aneuploidy risks. This participant made specific reference to the lack of resources in her community and this could possibly be a reflection of the inequalities in education and access to healthcare that are part of the legacy of Apartheid (Coovadia et al., 2009). In general, many women who attend PCC are likely to have lower education levels that are more representative of the City of Cape Town's average education level (Statistics SA, 2011) and the need for proper education and health literacy regarding age-related aneuploidy risks might be even more pronounced in the general population. In response to this lack of knowledge in the wider community, most of the participants mentioned that they think there needs to be an initiative to make this information available to others. This will be further discussed later.

While it was rarely directly stated, the participants' interviews all suggest that they gained some knowledge during their counselling session which they believed to be valuable. This finding highlights the educational role that genetic counsellors and/or nurses fulfil at the clinic and speaks to the first pillar of health literacy. It also fits with the American NSGC's definition of GC and its explanation that the process involves "education about inheritance, testing, management, prevention, resources and research" (Resta et al., 2006). This knowledge also plays an essential role in the process of informed decision-making (Metcalfe, 2018). In GC, the client is expected to be solely responsible for decision-making as GC is guided by the principle of non-directiveness (Weil, 2000). In prenatal GC this requires a fine balance. Previous South African research which used conversation analysis to understand how AMA women perceive the offer of IDT, revealed that it is often perceived as an invitation to accept (Wessels, Koole & Penn, 2015). This is similar to the case of one participant in the current study who believed that the counsellor wanted her to accept IDT despite the efforts of the genetic counsellor to facilitate autonomous decision making. Other participants in this study specifically addressed the non-directiveness and explained that the counsellor made it clear that the decision regarding IDT and consideration of TOP was theirs alone. Therefore the participants' statements about knowledge gained and the literature's connection between knowledge and informed decision-making would suggest that the educational aspect of GC is an essential service to aid patients who attend PCC in making decisions – in this case regarding accepting or declining IDT.

The majority of this cohort declined IDT. This is in keeping with a previously published audit that showed a reduction of more than 50% in the uptake of amniocentesis for AMA at GSH between the first five years of the 1980s and first five years of the 2000s (Urban et al., 2011). The audit also reports that nationally only a very small fraction of AMA women had an amniocentesis in 2008 (estimated to be less than 1%). This number takes into account the whole population that should have been eligible to undergo IDT for age-related risk of aneuploidy, but the audit notes that only a fraction of these individuals usually receive counselling. More than one third of women who were counselled for AMA in 2008 at GSH actually did choose to accept an amniocentesis (Urban et al., 2011). A similar decline in uptake of IDT has been noted since the early 2000s in some Korean (Kim et al., 2018) and American (Nakata, Wang & Bhatt, 2010) studies. These studies reported IDT uptake rates of 7,3% (Kim et al., 2018) and 31% (Nakata, Wang & Bhatt, 2010) respectively. The low uptake rate recorded in the South African audit as well as in the current study is therefore not unique but it is important to know that the international studies mentioned that one of the reasons for declining uptake rates were increased availability of screening tests such as maternal serum screening and NIPT. As discussed in chapter 1 these tests have a much higher sensitivity and specificity than AMA. It is unlikely that these factors are the reason for decreased uptake of amniocentesis in the public health sector in SA as they are not routinely offered in this system. As a result, the majority of women in this study declined IDT without the additional risk information that participants in the international studies may have had.

Two studies which were conducted to investigate factors that influence amniocentesis uptake among AMA cohorts reported that the most significant factor was women's theoretical attitudes towards the procedure (Vergani et al., 2002; Grinshpun-Cohen et al., 2015). Considering the lack of preparedness discussed previously, it is highly unlikely that the participants of this cohort had theoretical opinions regarding IDT before coming to PCC. Other factors that were reported in the previous studies included sonographic results, risk of miscarriage and attitudes towards TOP (Vergani et al., 2002; Grinshpun-Cohen et al., 2015). The 2002 study reported that a small percentage of women changed their attitudes towards amniocentesis after seeing the sonographic results (Vergani et al., 2002). This is in keeping with some participants who seemed inclined towards testing until they considered the outcome of the ultrasound. These participants explained that they decided not to accept IDT when they realised that there was nothing found on the scan to suggest that there was anything wrong with their foetus. It is important to keep in mind that the normal ultrasound seemed to have been misinterpreted by some participants to mean that their child would definitely not have DS. It is worth considering the implications of this possible misunderstanding as participants seemed to have used this information to make decisions regarding IDT. The risk of miscarriage might have been another consideration in declining testing, but if so it was not specifically mentioned as a reason by the participants in this cohort, but it did seem to be a factor that caused significant distress during the decision-making process. It is interesting to note that some of the women that brought up the miscarriage risk and mentioned that it caused emotional distress eventually decided to undergo IDT.

A South African study that interviewed mothers who had children with DS and were never offered IDT, reported that these individuals believed that IDT is important as it could have helped them prepare for a child who needs extra care (Scott, Futter & Wonkam, 2013). This echoes the statement of one participant who accepted IDT who said that she will have to prepare for a child who will most likely not be able to live independently as an adult and also discussed the importance of preparing the rest of her family for such a life-changing experience. Even some of the women that declined IDT mentioned that they thought the information they received from the counsellor served as a form of preparation in the event that their child is born with DS. A previous study reported that mothers who received limited counselling about DS after their child was diagnosed were often shocked and not well informed about the condition (Mbazima, 2016). It is therefore possible that the information gathered during their GC appointments could prevent the participants from having a similar experience should one of their children be affected with DS. The participants found the GC service to have a purpose, irrespective of whether or not they chose to accept IDT.

One of the reasons for offering women IDT is to have relevance to further management of the pregnancy. Typical interpretation of the South African TOP act gives women who are expecting a child with DS the legal right to choose to terminate the pregnancy up to a certain gestation and this is one of the factors participants were made aware of during the counselling session. A limited study of black female university students in KwaZulu-Natal reported an overall conservative attitude towards abortion (Patel & Myeni, 2008). Another study with a larger and more diverse cohort was published using data obtained from the annual South African Social Attitudes Survey (SASAS) in 2013. The survey had more than 2800 respondents from all nine provinces in SA and the cohort was representative of South Arica's diversity in home language and ethnicity. It reported that more than half of respondents believe that abortion is wrong even in the case of a foetal malformation (Mosley et al., 2017). These conservative views seem to be in keeping with this study cohort's views, as the majority of participants considered TOP to be an unthinkable action for themselves. Some participants also mentioned that they considered it to be wrong and equated it to killing. It is important to note that participants were only talking about TOP for their pregnancy and not about other women's consideration of TOP. The study that evaluated black female students' opinions also noted that some participants acknowledged that they would consider a TOP despite their religious objections in the case of an unwanted pregnancy (Patel & Myeni, 2008).This suggests that consideration of a TOP is a complex process that is dependent on more than just the individual's previous attitude towards the procedure. This is in keeping with the information shared by the only participant who said she would consider a TOP when she explained that although her religious beliefs oppose abortion, she would have to consider it if her child is affected with DS as she cannot practically take care of a child with special needs.

While TOP of a child affected with DS would generally not be considered by this cohort, several participants mentioned that their pregnancies were not planned and that they believed if the information regarding age-related aneuploidy risks were more commonly known in their local communities, some women might choose to not fall pregnant at such an advanced age. This is an interesting finding when one considers that participants generally felt that the age-related aneuploidy risk in their current pregnancy was low. The participants' statements that informed women might choose to not fall pregnant due to the aneuploidy risk suggests a contradiction in risk interpretation. One participant who specifically mentioned that her risk of less than 1% is low also said that she considered having a hysterectomy prior to falling pregnant and had she known about the aneuploidy risk she would have been more proactive about getting the surgery. While this relates to risk interpretation it also relates to the second pillar of health literacy – the ability to act on information received. An Iranian study that aimed to understand how women are empowered when they gather health information with a diverse cohort in terms of education, occupation and age reported on modifiable and preventative behaviours that women were empowered to take when becoming more health literate (Nasrabadi, Sabzevari & Bonabi, 2015). This correlation between health literacy and the intent to take action is further supported by a meta-analysis of 34 studies in high income countries which showed that individuals with lower health literacy are more likely to avoid preventative behaviours such as screening for sexually transmitted infections or following up on abnormal results of a Papsmear (Kilfoyle et al., 2016).

From the participants' statements regarding a woman's choice to be pregnant over the age of 40 as well as the lack of prior education, it seems as if there is not sufficient knowledge regarding age-related pregnancy risks in the participants' communities. The participants felt that genetic counsellors should take some responsibility for improving community awareness. This is not a new finding as the suggestion that genetic counsellors in SA need to improve community awareness regarding genetic diseases has previously been documented in the literature (Morris et al., 2015). The participants in this study offered some solutions about how they thought the counsellors could reach a wider audience. One participant specifically spoke about previous public health campaigns regarding HIV and Papsmears and how they, in her opinion, led to the people in her community being well informed about these topics. This notion is well accepted. A study done in several census enumeration areas of SA with a large cohort between the ages of 15 and 55 years reported that, when individuals were exposed to public health campaigns regarding HIV, they not only became more knowledgeable about the disease, but also instituted some behavioural changes to reduce the risk of HIV transmission (Peltzer et al., 2012).

In addition to the belief that genetic counsellors need to reach wider communities with this information, the women also reported that they would take the onus on themselves to inform other people in their social sphere about the risk of age-related aneuploidy. As mentioned previously, some participants reported feeling deprived of the opportunity to learn about the risks prior to falling pregnant and mentioned that they would inform other people of the risks to prevent them from having a similar experience. This willingness to "preach the gospel" highlights the importance of proper health literacy and also reflects certain qualities of Ubuntu – a practice that is widespread in some African cultures and reflects the interdependence between individuals, or as it is more commonly explained: "I am because you are" (Kamwangamalu, 1999). The desire to educate other individuals has also previously been reported in a cohort of African mothers whose children have DS (Mbazima, 2016) and is encouraging as genetic counsellors are a relatively scarce resource in SA (Kromberg, Sizer & Christianson, 2013).

The widespread lack of knowledge regarding the subject of pregnancy risks at AMA, as well as the apparent inadequate amount of information received during the referral process, resulted in the participants of this study having no expectations of their appointments that were specific to GC. This cohort-wide lack of expectations regarding GC and lack of knowledge regarding the reason for referral seem to be more pronounced than in other studies found in the literature but is not unique to this cohort. An American study that interviewed both counsellors and counselees reported that patients undergoing GC for a variety of reasons often had a general understanding of why they were going to speak to someone with a genetics background but were unsure of exactly what the appointment would entail or what form it would take (Bernhardt, Biesecker & Mastromarino, 2000). This differs slightly from the participants in this study who were not even aware that they would be speaking to a genetic counsellor but the overall limited understanding about GC seems similar and is echoed in the results of the study that investigated black mothers' experiences with GC in SA (Morris et al., 2015).

Despite there being no GC-related expectations among the participants, there were several other health care related expectations. The expectations that were reported varied and included having an ultrasound, finding out the gender of their child or further HIV counselling. An ultrasound was an expectation in most cases, even when individuals knew that they were being referred due to the risk of DS. This was such an important expectation that one participant revealed that, without the ultrasound, she did not think that coming to GSH for GC would have been worth it. This expectation was met in all the cases of this study and had a significant impact on other aspects of the counselling session, including decision-making and how the participants processed their emotions regarding the aneuploidy risk. Another observation was that many participants' only prior experience with counselling was that of HIV counselling, as this is a service that is available at most local clinics in the country (Department of Health, 2015). The period between learning that they were at PCC to see a genetic counsellor and actually seeing the counsellor afforded some participants a limited amount of time to consider their expectations. One participant explained that she could only compare it to her previous HIV counselling experience, which she did not find insightful at all and therefore expected GC to be "dull and dreary".

Participants learning unexpectedly about age-related aneuploidy risks, whether at the referring clinic or at PCC, combined with the desire for a "normal baby" resulted in some participants experiencing emotional distress. A study in Sweden on women's experiences when they received unexpected news regarding risks to their child at a second trimester ultrasound, reported that women often viewed the ultrasound as a social rather than medical event to look forward to and were often distraught when they found out about risks to the foetus as a result of soft markers. (Åhman, Runestam & Sarkadi, 2010). The women in the Swedish study did not realise that the second trimester ultrasound was part of a screening process that can inform about certain risks to the foetus. This is similar to the participants in this study who believed that they were only coming for an ultrasound and were unaware that they would also find out about the age-related aneuploidy risk. It is therefore not surprising that the participants reported initial emotional upheaval when they were informed about their risk, analogous to the increased anxiety reported among the Swedish cohort. Similar worry was reported in a study where South Korean women of AMA learned about increased risk for aneuploidy through additional screening strategies such as the quadruple serum screen (Jun, Thongpriwan & Choi, 2016). It is of interest to note that several participants in the Swedish study would have preferred not to know about the ultrasound findings and possible risks (Åhman, Runestam & Sarkadi, 2010). This was not a finding that emerged from the current study's cohort as they generally reported being very appreciative of the information. However this cohort was limited due to its size and more research is needed to determine whether a more representative sample of attendees at PCC might reflect the views of the Swedish study.

Despite the initial emotional reaction reported by the participants, it seems as if the participants left their counselling session with a sense of hope. This is an important observation as a sense of hope was one of the four pillars of empowerment that the UK-based study discussed in chapter 1 reported after investigating what patients expect from genetic services (McAllister et al., 2008). Factors that provided relief from the reported anxiety and left the participants in this cohort feeling optimistic included normal ultrasound results, their religious faith, hearing their personal aneuploidy risk which participants often thought to be low, talking to the genetic counsellor as well as previous experience with women who have had healthy children after the age of 40. This last factor is known as availability and refers to how easily someone can think of an example of a certain outcome (Weil, 2000). This phenomenon has previously been noted in a Canadian cohort where nulliparous AMA women were more likely to view the risk to their pregnancy as low when they could recall a favourable outcome in someone else's AMA pregnancy (Bayrampour et al., 2012). It is interesting to note that several of the participants have seen an individual with disabilities who they now believed had DS in their communities and were able to bring them to mind during the interview, but it seems as if this did not influence participants' view of their risk in the same way that an example of a positive outcome did. Several participants, whether directly or indirectly, also reflected on the role that the counsellor played in the process of getting to an optimistic attitude. Multiple participants commented on the warmth of the counsellors and other staff at GSH and voiced their appreciation for the information that they received. This fits with the previously discussed study of black mothers with children who had genetic diseases that reported reasons for a positive experience included information given as well as the respect that they often received from the counsellors (Morris et al., 2015). Participants in the current study also mentioned religious faith as a reason for their optimism. This phenomenon has previously been reported as a factor that helped mothers of children with DS come to terms with their child's condition (Mbazima, 2016).

CONCLUSION

In conclusion, it was found that although the women had some knowledge about age-related aneuploidy risks, the details about these risks and the options dealing with them seem to be lacking in local communities as well as at lower tier MOUs and hospitals. This coupled with the fact that the women had no knowledge about a "genetic counselling consultation" or what it entails, resulted in them not knowing what to expect from their referral to GSH. The findings from the study were interesting in that, despite this lack of preparedness and not seeking out the service, the participants appreciated the information and support they received from the counsellors. They were able to process the information discussed during the session and generally adjusted well to their risk and left feeling empowered to be able to deal with what the future holds. From the results, it seems that feeling empowered does not mean undergoing an amniocentesis as evidenced by the fact that, despite initial anxiety regarding the age-related aneuploidy risk, the majority of the women opted to not undergo IDT. They had strong feelings about the lack of knowledge in their communities and all planned to educate other women in their communities and recommended that the professionals embark on education campaigns to improve health literacy. These findings can have implications for the GC profession in the context of the Western Cape's public health system as it highlighted the needs and expectations of this specific population. There is clearly a role for GC at lower tier MOUs and the implications and recommendations for this is discussed below. This insight will offer genetic counsellors the opportunity to align the AMA counselling service more closely with the needs of the women who use it.

STRENGTHS AND LIMITATIONS

Strengths:

- The cohort is diverse in home language, level of education and employment and this makes the results of this study more likely to be applicable to the wider population that attend PCC.
- The pragmatic approach and semi-structured interviews allowed the researcher to delve deeper into certain aspects of participants' comments which allowed for richer data and a more holistic picture.
- The researcher was not involved in counselling the women that she interviewed and could therefore be viewed as an independent party by participants.

Limitations:

- The interviews for this project were conducted in the months leading up to a nationwide lockdown as a result of the worldwide COVID19 pandemic. Data collection was interrupted by this lockdown as well as the decision by the HREC of UCT to temporarily suspend all face-to-face interviews (See Appendix E) and the suspension of student attendance in clinics as a response to the pandemic. These unforeseen circumstances resulted in a smaller cohort than the researcher anticipated and as a result the researcher cannot be certain that data saturation was reached or that the findings can be extrapolated to other patients who attend PCC.
- Only women who were counselled in English were interviewed for this study and it is possible that patients who were counselled in a language other than English, possibly with the assistance of a translator, might have had a very different experience at the clinic. It is also possible that participants who were not interviewed in their home language struggled to express themselves as well as they would have in their home language.

- The interviews were conducted in the same room where the participants received GC. This might have prevented participants from separating the researcher from the service, which in turn could have resulted in participants filtering their true thoughts of the service.

IMPLICATIONS AND RECOMMENDATIONS

As discussed previously, several factors outside of the PCC influenced participants' experience of the clinic. As such, the implications and recommendations of this study are not only applicable to the PCC clinic, but also to other areas of healthcare:

- The referral process needs to be adjusted to address the needs of the patients. This could possibly take the shape of an explanation for the reason for referral and a pamphlet that the patient can read in her own time explaining the age-related risk of aneuploidy so that she will have a better understanding when she arrives at the clinic. Counsellors should also advocate for a better understanding of AMA GC at referral clinics so that patients can choose whether or not they want to hear about their age-related aneuploidy risk.
- Genetic counsellors should try to develop a public health campaign that targets women of reproductive age to inform them about AMA related aneuploidy risk and to empower them to make informed reproductive decisions.
- Genetic counsellors should ensure that they educate women about the limitations of antenatal ultrasounds to aid patients with informed decision-making.
- Future research should investigate how AMA women interpret risks to understand why participants viewed their age-related aneuploidy risk as low but some still mentioned that they would have taken more precautions to not fall pregnant had they known.
- Future research should compare how women who had prenatal GC experienced the birth of a child with DS with those who did not to assess whether GC without IDT really prepare parents for an affected child.
- Future research should aim to recruit a bigger cohort to assess whether the appreciation of information is representative of most PCC attendees.

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APPENDIX A

Title: Investigating the views and expectations of pregnant women who undergo genetic counselling for age-related risk to aneuploidy

I am a genetic counselling student at the University of Cape Town and am doing this research to obtain my master's degree. This project aims to investigate what women expect from and how they experience the genetic counselling that is offered at the pregnancy counselling clinic in Groote Schuur hospital. This will be investigated by post-counselling individual in-person interviews with the researcher on the same day that you undergo genetic counselling. The interviews will be done in a private room at the clinic in Groote Schuur hospital. The interview will be between 45 and 60 minutes and will be recorded for further analysis.

The Human Research Ethics Committee at the University of Cape Town has approved this research (reference: 636/2019) and it will be used to submit a mini-dissertation to the University of Cape Town as well as a possible publication in a peer reviewed academic journal.

This research might address some sensitive topics and you are allowed to refuse to answer questions that make you uncomfortable. If you have any further questions about your individual risk the researcher will refer you back to a genetic counsellor for further assistance.

Your privacy is of the utmost importance and your name will not be used in the storage of any documents or recordings. Your personal details that are used in this form will also be stored in a different location than all the other data collected during this process. Any data collected will be stored on a password-protected computer. Only the researcher and her supervisors will have access to the consent forms with your name on them – in all other documentation a numerical number will be assigned to differentiate between participants.

Participation in this study is optional. If you want your data to be removed at any point before the submission of the researcher's mini-dissertation you can contact the researcher and it will be done without any impact on the health care that you receive from Groote Schuur hospital. You do not need a reason to withdraw from the research.

There will be no financial reimbursement for your time and participation in this study and the researcher has no obligation to contact you with the outcome of this study. The results of this study will hold no benefits directly for you as the participant.

If you have any queries or worries about the research at any point after your interview, please feel free to contact the researcher's supervisors:

Dr Karen Fieggen: 021 406 629/ karen.fieggen@uct.ac.za
Nakita Laing: 021 406 6304/ n.verkijk@uct.ac.za

If you have any ethical concerns about this research, the chairperson of the ethics committee, Professor Marc Blockman, can be telephonically contacted on **021 406 6492**.

Statement of participation:

1) I have been informed of the purpose of this study and am choosing to participate **willingly and without coercion**
2) I am aware that this study holds **no direct benefit** to me or any of my immediate family
3) I am aware that participation in this study has no effect on the care that I will receive from Groote Schuur hospital and that I can **choose to withdraw** from this study **at any time**.
4) I understand that my interviews will be recorded
5) I understand that I can refuse to answer any of the researcher's questions at any time
6) I agree that the data obtained in this interview might be used for **publication** as long as all **personal information is kept private and confidential**
7) The researcher explained the study to me and answered all the questions that I had pertaining to the study. I was given enough time to consider whether I **want to participate**

I hereby give consent to participate in this research and agree that I am signing this freely and without coercion.

Participant's name	Participant's signature	Date

APPENDIX B

Demographic Information

Participant identifier: ______

1. How old are you?

---

2. What is your relationship status?

☐ Single

☐ Married

☐ Divorced

☐ Stable non-married relationship

☐ Widowed

3. What is your home language?

---

4. What is your highest education qualification?

☐ Primary School

☐ High School (did not finish matric)

☐ High School (finished matric)

☐ Tertiary education (University diploma or degree)

☐ Postgraduate degree

5. What type of residence do you live in?

☐ Formal housing in an urban area

☐ Informal housing in an urban area

☐ Formal housing in a rural area

☐ Informal housing in a rural area

6. Are you employed? If yes, what kind of work do you do?

____

7. How far along is your pregnancy?

____

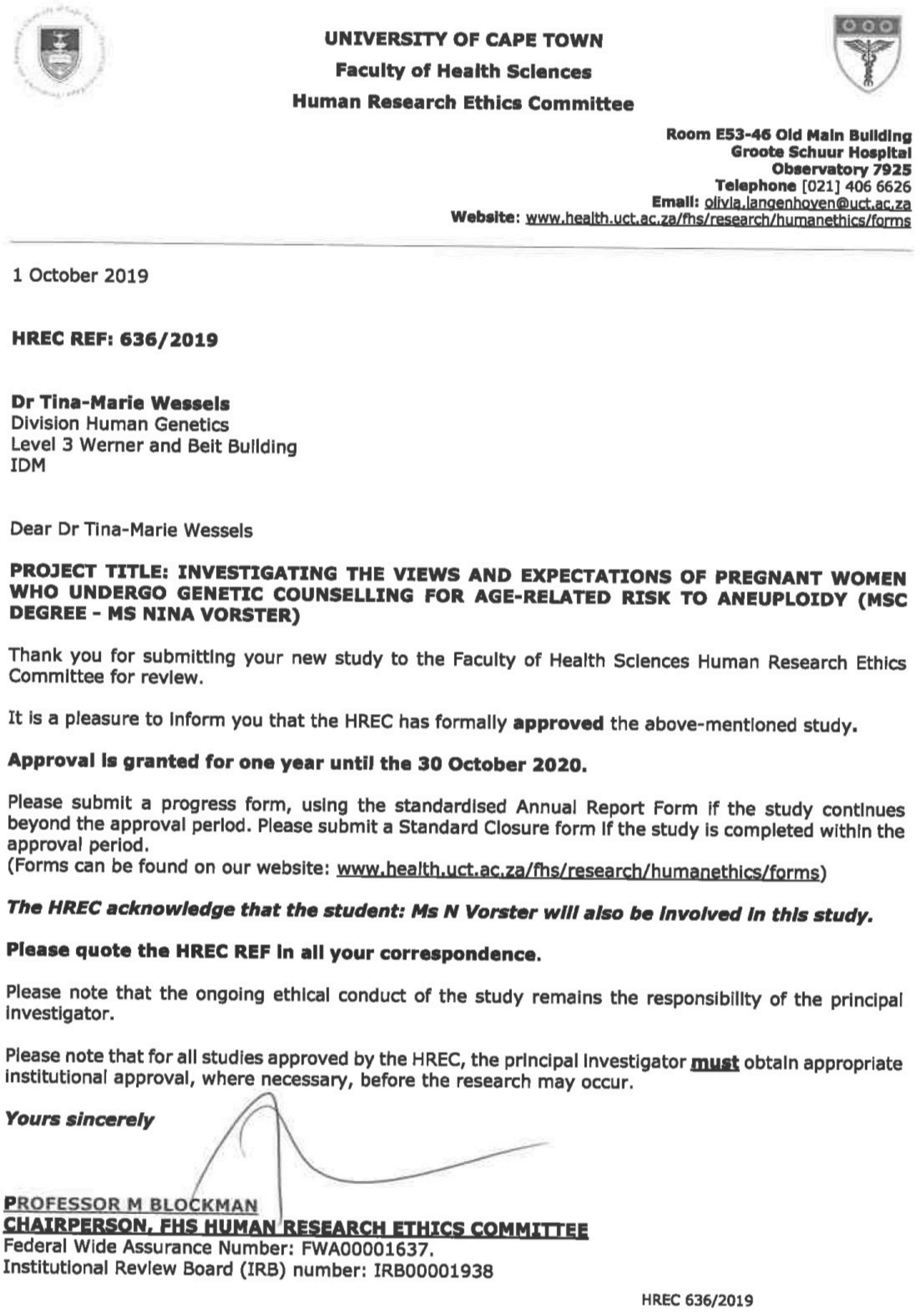
APPENDIX C

Adjusted Question Guide

Question guide

1. Can you tell me about your appointment today?
2. What did you think was going to happen in the genetic counselling session today?
3. Compared to what you thought before the session, what do you think about everything that the counsellor told you today?
4. Can you tell me your thoughts on our services?
5. Can you share your thoughts on how it was to first have a group interview followed by an individual interview?

APPENDIX D



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

Room E53-46 Old Main Building
Groote Schuur Hospital
Observatory 7925
Telephone [021] 406 6626
Email: olivia.langenhoven@uct.ac.za
Website: www.health.uct.ac.za/fhs/research/humanethics/forms

1 October 2019

HREC REF: 636/2019

Dr Tina-Marie Wessels
Division Human Genetics
Level 3 Werner and Beit Building
IDM

Dear Dr Tina-Marie Wessels

PROJECT TITLE: INVESTIGATING THE VIEWS AND EXPECTATIONS OF PREGNANT WOMEN WHO UNDERGO GENETIC COUNSELLING FOR AGE-RELATED RISK TO ANEUPLOIDY (MSC DEGREE - MS NINA VORSTER)

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

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

Approval is granted for one year until the 30 October 2020.

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

The HREC acknowledge that the student: Ms N Vorster will also be involved in this study.

Please quote the HREC REF 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, FHS HUMAN RESEARCH ETHICS COMMITTEE
Federal Wide Assurance Number: FWA00001637.
Institutional Review Board (IRB) number: IRB00001938

HREC 636/2019

NHREC-registration number: REC-210208-007

This serves to confirm that the University of Cape Town Human Research Ethics Committee complies to the Ethics Standards for Clinical Research with a new drug in patients, based on the Medical Research Council (MRC-SA), Food and Drug Administration (FDA-USA), International Council for Harmonisation of Technical Requirements for Pharmaceuticals for Human Use: Good Clinical Practice (ICH GCP), South African Good Clinical Practice Guidelines (DoH 2006), based on the Association of the British Pharmaceutical Industry Guidelines (ABPI), and Declaration of Helsinki (2013) guidelines. The Human Research Ethics Committee granting this approval is in compliance with the ICH Harmonised Tripartite Guidelines E6: Note for Guidance on Good Clinical Practice (CPMP/ICH/135/95) and FDA Code Federal Regulation Part 50, 56 and 312.

APPENDIX E

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

Room G50 Old Main Building
Groote Schuur Hospital
Observatory 7925
Telephone [021] 406 6626
Email: hrec-enquiries@uct.ac.za
Website: www.health.uct.ac.za/fhs/research/humanethics/forms

17th March 2020

Dear Heads of Departments, Department Research Committees, Researchers and Members of the Research Community

COVID-19: FACULTY OF HEALTH SCIENCES HUMAN RESEARCH ETHICS COMMITTEE RECOMMENDATIONS FOR RESEARCH STUDIES AND RESEARCH SITES INVOLVING HUMANS

We are anticipating community spread in South Africa. Therefore, all research activities where people are brought together for research purposes, where research participants and research staff and students are placed at risk by virtue of the nature of data collection, or where specimen collection poses transmission risk should be urgently reviewed to determine the risk for COVID-19 transmission. Our duty is to ensure the protection of UCT staff and students; as well as reducing the risk of transmission.

In line with recommendations to practice social distancing, we want to minimise the risk of transmission at research sites and in the context of research studies involving human participants.

Over the next week, we recommend each research study or study site develop a plan to de-escalate or suspend research activities. Limiting infections and protecting research participants and research staff and students should be the priority.

Therefore, the HREC recommendations are as follows:

1. Limiting infections and protecting research participants and research staff and students should be the priority.
2. All non-therapeutic research involving human participants should be stopped. Exceptional reasons for research to continue can be discussed with the HREC.
3. For therapeutic research and clinical research trials:
    a. All therapeutic research and clinical research trials should be placed on hold and research activities should be suspended where possible.
    b. Whenever possible, telephonic visits or follow-up should occur.
    c. Principal investigators and study sites should implement measures to ensure that there is no interruption of required medication or monitoring of adverse events.
    d. Only critical study visits where benefits significantly outweigh the potential harm of COVID-19 infection take place going forward. Study visits for critical medication or monitoring purposes should continue in consultation with the HREC.
    e. New enrolments to studies should be suspended. Potential exceptions to this recommendation can be discussed with HREC.
4. Recommendations regarding hand hygiene, cough etiquette and social distancing should be implemented immediately at study sites.

5. Researchers and study sites should develop a 'in the time of COVID-19' template register in case retrospective contact tracing becomes necessary.
6. Should any research staff, students or participants develop signs or symptoms suggestive of COVID-19, please call the National Coronavirus Hotline (0800 029 999), the Provincial hotline (021 928 4102), or WhatsApp "Hi" to 060 012 3456.

All precautions to prevent transmission and reduce risk need to be urgently implemented.

The HREC will continue to function administratively, as well as the reviewing of new submissions.

Yours sincerely

Signature removed

PROFESSOR M BLOCKMAN
CHAIRPERSON, FHS HUMAN RESEARCH ETHICS COMMITTEE