Psychiatric genetic counselling: Current landscape and opinions of psychiatric healthcare professionals in public hospitals in Cape Town, South Africa

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Declaration

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Abstract

Psychiatric conditions are common, and psychiatric genetic counselling (PGC) aims to help people understand their, or a family member's, mental illness better, as well as help them understand the factors that can contribute to the development of a mental illness. However, PGC services are not implemented routinely despite reported patient benefits of increased patient empowerment and self-efficacy. This study serves as a pilot project to investigate current practices of mental illness counselling services, focusing on aetiology, provided by psychiatric healthcare professionals at a single unit in the public health sector in Cape Town, South Africa. A pragmatic qualitative research approach, using semi-structured interviews, was used to explore the questions patients ask in terms of recurrence and causes of their mental illness, as well as healthcare provider's (HCPs) opinions on offering a PGC service. Ten interviews were thematically analysed. Results showed that PGC is not being routinely implemented in psychiatric consultations, patients and families have limited genetic knowledge, and family members frequently ask about the mental health condition and its causes. Psychiatric HCPs seemed to believe PGC is likely to be beneficial to the patient, their families and mental health services, and would use such a service if available to them. However, there were several perceived barriers to establishing a PGC service, such as limited resources and available time, which makes a PGC service unfeasible at this point. These results will contribute to the body of knowledge on PGC (such as potential applications, benefits and obstacles, and acceptability of PGC) and may serve as a starting point to considering the implementation of a PGC service in Cape Town, South Africa.

Keywords:
Psychiatric genetic counselling, mental illness, qualitative research

Vir Oupa Willie,

Ek wens oupa kon die een sien.

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Last, but certainly not the least, Geoffrey Ellis. Thank you for making every small win feel like a giant victory. Thank you for holding my hand through it all.

Table of Contents

List of terms and abbreviations

22Q11.2DS	22Q11.2 deletion syndrome
DTC	Direct-to-consumer
GC	Genetic counsellor / genetic counselling
GCOS	Genetic Counselling Outcomes Scale
GSH	Groote Schuur Hospital
GWAS	Genome-wide association studies
HCP	Healthcare professional
HREC	Human Research Ethics Committee
IMSES	Illness Management Self Efficacy Scale
NDD	Neurodevelopmental Disorders
NHRD	National Health Research Database
NSGC	National Society of Genetic Counseling
OCD	Obsessive Compulsive Disorder
OPD	Outpatient department
PGC	Psychiatric Genetic Counselling
PRS	Polygenic Risk Score
QD	Qualitative Description
SA	South Africa
SIPD	Substance Induced Psychotic Disorder
SNP	Single nucleotide polymorphism
UCT	University of Cape Town
UK	United Kingdom
USA	United States of America

List of tables and figures

Chapter 1: Introduction

1.1. Heritability of psychiatric conditions

Mental health conditions are common, one out of eight individuals will experience a mental illness in their lives, and in 2019 an estimated 970.1 million people were living with a mental illness (GBD 2019 Mental Disorders Collaborators, 2019; World Health Organization, 2022). Data from South African estimates showed that the lifetime prevalence of any psychiatric disorder is 30.3% (Stein et al., 2008).

Psychiatric conditions are believed to be multifactorial i.e. develop as a result of an interplay between environmental and genetic factors. Many mental health conditions are highly heritable, indicating substantial genetic influences contributing to these conditions. Heritability of a trait or phenotype is defined as the proportion of total variance in a population that is attributed to variation in genetic factors (Visscher, Hill & Wray, 2008). Traditionally, heritability scores are estimated from observed and expected outcomes between relatives, such as the difference in correlation of a trait between monozygotic and dizygotic twin pairs (Visscher, Hill & Wray, 2008). Heritability scores for some common psychiatric conditions have been estimated to be 70 – 85% for schizophrenia, 60 – 85% for bipolar disorders, and 40% for major depressive disorder (reviewed by Burmeister, McInnis and Zöllner (2008)). These scores are higher than heritability scores for breast cancer (30-56%), a condition for which several genetic risk factors are well established, and affected individuals and families are frequently referred for GC (Locatelli, Lichtenstein & Yashin, 2004; Möller et al., 2016; Schildkraut, Risch & Thompson, 1989). This may suggest that there may be scope for genetics professionals in the field of psychiatry.

Absolute empiric recurrence risks of some psychiatric conditions are available (Peay, 2020). For example, individuals have an increased risk of 5-13% (general population risk 1%) and 6-15% (general population risk 0.8-1.6%) to have schizophrenia and bipolar disorder respectively, if they have an affected parent (Peay, 2020). These empiric risks are dependent on the relationship to the affected individual and the specific diagnosis. For example, the empiric risks change to 9-16% and 5-20% for schizophrenia and bipolar disorder respectively, if the individual in question has an affected sibling (Peay, 2020). Certain genetic disorders, such as 22q11.2 deletion syndrome (22q11.2DS), have significant genetic risk factors for psychiatric conditions such as schizophrenia (Bassett & Chow, 2008). A previous study found that 22.6% of adults with 22q11.2DS presenting at a cardiac clinic had schizophrenia or schizoaffective disorder (Bassett et al., 2005). Psychiatric disorders thus have a polygenic architecture, therefore there is no single gene diagnostic test.

Genetic testing for psychiatric conditions is not routinely implemented unless there are syndromic concerns, such as in the case of copy number variants like 22q11.2DS. Advancements in psychiatric genetics and research into the use of polygenic risk scores (PRS) in clinical practice is currently a hot topic and widely debated. A PRS represents an individual's cumulative risk for a particular phenotype based on the weighted sum of individual single nucleotide polymorphisms (SNPs) identified from genome-wide association studies (GWAS) (Moorthy et al., 2023). Although these methods are not used in clinical practice, psychiatric genetic HCPs have expressed their opinions that PRS may potentially be useful in clinical practice in the next decade (Moorthy et al., 2023). Even though genetic testing and use of PRS in psychiatric clinical practice is not yet being implemented, it has been indicated that GCs would be useful in psychiatry when PRS are routinely implemented (Wouters et al., 2024). It has however been reported that a common misconception is that GC should only be provided when genetic testing is available (Moldovan et al., 2019). There may be scope for GCs in the psychiatric space, despite the lack of testing availability, which will be discussed in section 1.3.

Psychoeducation, an educational process frequently used in psychiatry, involves providing families and patients with information about their illness, including its diagnosis and treatment (Motlova et al., 2017). Psychoeducation involves providing patients and families with disease-specific information such as recognition and management of symptoms, potential genetic implications of the illness, as well as techniques and skills that can be used to promote healthy lifestyles, identifying stressors and educating family members on the effects of the illness and support strategies in disease management (Motlova et al., 2017). A survey assessing the provision of psychoeducation programmes for patients with mental illnesses in the UK found that 45% of community mental health teams do not provide any psychoeducational programmes, most commonly because of financial and workload constraints and lack of expertise in providing the service (Getachew, Dimic & Priebe, 2009). Motlova et al. (2017) suggest that lack of knowledge and skills in providing psychoeducation, as a result of a lack of training thereof, might be the biggest obstacle in routine psychoeducation implementation.

Additionally, time limitations when consulting with patients and families may prevent routine psychoeducation. However, it was reported that psychoeducation may save time in the long run by preventing patient relapses and encouraging family engagement in care (Motlova et al., 2017).

1.2. What is genetic counselling?

Genetic counselling (GC) is the the process of assisting people to understand and adapt to various aspects of a genetic disease, such as the medical, psychological, and familial implications (Resta et al., 2006). The process of genetic counselling assesses the chance a disease will occur, based on the interpretation of family and medical histories (Resta et al., 2006). Genetic counsellors (GCs) provide education about the inheritance pattern and testing of diseases, as well as the management and prevention thereof (Resta et al., 2006). Ultimately, GC promotes informed choices and adaptation to the genetic condition individuals are diagnosed with or are at risk of developing (Resta et al., 2006). Such GC interactions typically involve taking family medical histories from patients, performing risk assessments, providing risk counselling and identifying at-risk relatives (Ormond et al., 2024b). GCs also play a role in educating patients about their condition and the implications to family members, informing patients of testing options, facilitating informed decision making and explaining genetic test results (Ormond et al., 2024b). GCs play an important role in providing psychosocial support throughout these tasks and assist individuals to make meaning of the genetic contributions of their condition (Ormond et al., 2024b). More recently, the role of GCs has been expanding into more non-traditional roles such as laboratory GCs or GCs specialising in certain clinical areas as “mainstreaming” of genetic services are increasing (Ormond et al., 2024a). Although this is beyond the scope of the current research, it is important to note that GCs also work in more areas than previously reported and that the role of GCs are expanding globally.

GC, as a profession, is limited in South Africa (SA). The first GC training programme was established in 1988, and there are 28 GCs practicing in SA based on 2024 estimates (Gomes et al., 2024; Ormond et al., 2018). In comparison, the United States of America (USA) have 6517 GCs registered with the American Board of Genetic Counseling, and there are about 300 GCs registered in the United Kingdom (UK) (Middleton et al., 2023; Ormond et al., 2024a). A 2023 survey conducted by the National Society of Genetic Counseling (NSGC) in the USA reported the top speciality areas that GCs are involved with which include, but are not limited to, cancer genetics, prenatal genetics, paediatric genetics, reproductive screening, and adult genetics (National Society of Genetic Counselors, 2023:8). While specialisation is common in countries like the USA and UK, due to smaller numbers of GCs in SA, the majority of GCs work as generalists, which requires them to work in all referral areas (Ormond et al., 2018).

1.3. Psychiatric genetic counselling as a subspecialty of genetic counselling

PGC for psychiatric conditions seeks to assist people in understanding the psychiatric condition that they or family members have, and assists in understanding the factors that can contribute to the development of the diagnosis (Austin, 2020; Resta et al., 2006). PGC is a growing subspeciality in the field of GC, and with mental health conditions being as common as discussed in section 1.2 (GBD 2019 Mental Disorders Collaborators, 2019; World Health Organization, 2022), the need for GC in routine psychiatric practice is increasing (Moldovan et al., 2019; Nurnberger Jr et al., 2018).

GC as a professional field is in various stages of establishment across the world (Ormond et al., 2018). In addition, PGC as a speciality field of GC is still in its infancy stages, as very few countries have established PGC services. The first specialist PGC clinic, the Adapt clinic, was established in 2012 in Vancouver, Canada (Austin, 2020; Inglis et al., 2015). Since then, two more specialised PGC clinics were established in Cardiff and Tennessee (Austin, 2023; Hopkins, 2022). The UK and Romania offer PGC as part of research projects, and PGC is only just emerging in other countries like Portugal, Sweden, and Russia (Moldovan et al., 2019). PGC is likely facing challenges with being implemented compared to other specialties like cancer genetics, as cancer genetic tests can lead to medically actionable plans (Singh et al., 2023). The field is also possibly struggling to advance due to competing priorities within healthcare systems and the additional strain of implementing genetic services in an already constrained healthcare system (Moldovan et al., 2019). However, there is evidence that such a service is desired from both the patient and healthcare professionals (HCP) (Jenkins & Arribas-Ayllon, 2016; Martorell et al., 2019). These will be discussed in section 1.3.b.

Inglis et al. (2015), described the Adapt clinic’s practice, which included eliciting a detailed psychiatric family history and the patient’s understanding of the cause of their illness. This was then followed by providing the patient with more information on the genetic and environmental risk factors that contribute to their illness, mental health protective strategies they may employ, and communicating risks for family members when requested (Inglis et al., 2015). After receiving PGC, educational material and referrals were provided to the patient and they received a routine telephonic follow-up discussion with a GC after one month, to establish whether they wish to return to discuss any other issues (Austin, 2020). Results from evaluating this clinic showed that patients' main reasons for attending the Adapt clinic were to understand the cause of mental illness (58.7%), as well as the desire to learn ways to protect mental health (21.3%), the chance of illness recurrence for their child (52.0%) or illness recurrence overall (i.e. for themselves, a child, and other relatives) (64.0%) (Inglis et al., 2015).

There are several challenges that may hinder the implementation of a PGC service. Few clinicians refer to genetic services for psychiatric indications, thus GC positions are not prioritised in those settings (Chanouha et al., 2023). Additionally, GC is not an established profession in many countries, and there are few training courses in PGC for GCs (Moldovan et al., 2019), so establishing PGC services is challenging. Additionally, the lack of clinical genetic testing for psychiatric conditions may make GCs uncomfortable in providing PGC, potentially contributing to implementation challenges (Booke et al., 2020).

A major setback for PGC was when the Adapt clinic closed in 2023, after 11 years of practice (Austin, 2023). One of the reasons the Adapt clinic closed was due to the need for GCs to work in other programs, such as general genetics, because of lengthy waitlists for such services (Austin, 2023). The closing of the first specialised PGC clinic in the world highlights the challenges that GCs practicing in this field face, despite reported patient benefits and outcomes discussed in the next section.

a. Outcomes of PGC

A large body of research suggests that PGC is beneficial to patients (Austin, & Honer, 2008; Hippman et al., 2016; Huynh et al., 2023; Inglis et al., 2015; Morris et al., 2024; Morris et al., 2021; Semaka & Austin, 2019). Reported patient benefits of PGC include significant increases in patient empowerment and self-efficacy as measured by the Genetic Counselling Outcomes scale (GCOS) and Illness Management Self Efficacy Scale (IMSES) respectively (Inglis et al., 2015). Here, a 2015 study evaluated 75 patients' GCOS and IMSES scores prior to and one month after PGC, at the Adapt clinic (Inglis et al., 2015). PGC significantly increased patients' empowerment only one month after PGC ($p < 0.0001$), with an effect size d=1, which is highly significant (Inglis et al., 2015). Self-efficacy, measured by the IMSES, also increased significantly ($p = 0.011$; effect size, d=0.43) one month after PGC (Inglis et al., 2015). Quantitative questionnaires completed after participants received PGC showed decreased confusion and concern for other family members becoming affected with a mental illness and gave participants a new perspective about the causes of a mental illness (Austin & Honer, 2008). Overall, patients found a PGC service useful and have described it to be an “empowering encounter” that provides patients with a new perspective on managing, and a deeper acceptance of their mental illness (Austin & Honer, 2008; Semaka & Austin, 2019). This “encounter” contributed to patients feeling fewer negative emotions regarding their mental illness, such as blame, guilt and shame. Patients also reported being able to better manage and protect their mental health (Semaka & Austin, 2019).

PGC has also been shown to potentially lead to changes in behaviours (such as increased physical activity and others discussed further below) which may protect people affected with a mental illness (Huynh et al., 2023). PGC encompasses more than genetic risk communication and focuses on a psychotherapeutically oriented information exchange that helps patients make meaning of the contributors to the causes of their mental illness (Austin, 2020; Huynh et al., 2023). An exploratory study interviewing eight patients with lived experiences of a mental illness who received PGC in Vancouver, Canada, revealed that PGC helped patients accept their mental illness by enabling them to have a new perspective on their diagnosis (Huynh et al., 2023). Patients held different misconceptions on the causes of their mental illness, and PGC helped patients understand the multifactorial nature of these disorders (Huynh et al., 2023). Additionally, PGC allowed patients to release some of the negative emotions related to their initial understanding of their diagnosis and enhanced their sense of empowerment and self-efficacy (Huynh et al., 2023). From this, patients reported making changes to their management behaviours like engaging in more protective actions (increased physical activity, better treatment adherence, improved sleep, and mindfulness about nutritional intake), which improved their mental health outcomes (Huynh et al., 2023).

Studies have also aimed to evaluate the impact of PGC on psychiatric hospitalisation and psychopathology (Morris et al., 2024; Morris et al., 2021). Morris et al. (2024) found that total days in hospital decreased after PGC (1085 to 669), and there were fewer hospitalisations post-PGC than pre-PGC. However, these differences in hospitalisations were not statistically significant (p = 0.011, OR = 1.69) (Morris et al., 2024). While patients who received PGC appeared to have less psychiatric hospitalisations after a PGC intervention (which may in turn influence the economic impact of psychiatric hospitalisations), the study suggested that further research characterising groups of patients who benefit more, or less from such a service is needed (Morris et al., 2024). An earlier study by Morris et al (2021), which aimed to investigate frequency of psychotic symptoms, seemed to indicate that these could be reduced by the implementation of PGC. They reported that PGC significantly decreased psychotic symptoms in participants, which could not be explained by adherence to psychiatric medication (Morris et al., 2021). However, this study had several limitations, including a small sample size and possible participant ascertainment bias, thus more research is needed to elucidate the impact of PGC in larger studies (Morris et al., 2021). Furthermore, there have been no randomised control trials evaluating the long-term impact of PGC as an intervention, and this may further assist in establishing the benefits and outcomes of PGC.

b. Attitudes towards PGC

Knowledge about PGC is limited among HCPs and there are varying perceptions regarding what PGC entails (Moldovan et al., 2019). Attitudes toward PGC among users and professionals have been investigated in the past. A survey of mental health professionals and users in Spain showed that most professionals (59%) and patients and their relatives (80%) felt that a GC unit in psychiatry would be useful, and 41% of professionals would often or almost always refer their patients to a GC unit if it was possible (Martorell et al., 2019). Even though few patients asked about the genetics of their disease (12%) or the possibility of their children inheriting the disease (19%) to their treating clinician, almost half (47%) of users believed that psychiatric disorders have a genetic basis and 43% were worried about transmitting their illness to their children (Martorell et al., 2019).

In a previous study, some psychiatrists felt that it was within their scope of practice to provide individuals and families with some genetic information as part of a typical consult, but that time was the biggest limiting factor of having such an in-depth discussion (Cardiff, Ecker & Austin, 2023). Research has shown that psychiatrists have a misconception regarding the role of a GC in psychiatry, mainly attributing the role of a GC to discussions around pharmacogenetics, family planning and genetic testing for conditions that may have psychiatric manifestations like 22q11.2DS (Cardiff, Ecker & Austin, 2023). Psychiatrists were not aware of the psychotherapeutic aspects of a GC's role (Cardiff, Ecker & Austin, 2023). Thus, referrals to PGC from psychiatry is mostly based on perceptions about the aim of PGC (Leach et al., 2016). Psychiatrists were most likely to refer patients for PGC when genetic testing is a consideration, when family planning options need to be discussed, or when patients express an interest in more information regarding the causes and recurrence risks of their illness that psychiatrists feel they cannot discuss adequately, due to time constraints or feeling like they lacked the expertise to address that need (Cardiff, Ecker & Austin, 2023; Leach et al., 2016).

GCs surveyed from Australia and New Zealand believe there is sufficient evidence to support PGC, even though most do not routinely see patients with a primary indication of a personal or family history of a psychiatric disorder (Isbister et al., 2023). Similar findings were seen in the UK (Rowlatt, McAllister & Cuthbert, 2022). GCs from Australia and New Zeeland also indicated their interest in attending education opportunities on PGC, and most would incorporate PGC into their practice in the future (Isbister et al., 2023). A workshop ("Psychiatric Genetic Counseling for Genetic Counselors" (PG4GC)) was developed by Jehannine Austin to increase confidence and competence of GCs to provide PGC in their own practices (Dillon et al., 2022; Mack et al., 2024). This two-day workshop left participants feeling empowered and comfortable to provide PGC to their patients (Dillon et al., 2022). Participants also reported that they already had the skills and knowledge to provide PGC (Dillon et al., 2022), highlighting that no extra training would be necessary for GCs once a PGC service was in place. A larger study evaluating the effectiveness of 13 training groups of the PG4GC workshop showed that this workshop increased participants' comfort and knowledge of providing PGC, and left participants feeling more equipped to provide PGC (Mack et al., 2024). Overall, PG4GC was shown to be an effective training tool, enabling more GCs to develop the skills to provide PGC, which will facilitate the implementation of PGC in more settings (Dillon et al., 2022; Mack et al., 2024).

A study in the UK investigating the demand for PGC as well as the possible benefits and barriers, found that psychiatric HCPs (psychiatrists and psychiatric nurses) believe that a specialist psychiatric genetic service would be useful and desirable, however participants indicated that low patient demand may be a barrier to such a service (Jenkins & Arribas-Ayllon, 2016). In the same study, psychiatric HCPs believed that PGC can provide key benefits to patients such as reducing stigma and guilt and provide aetiological information about a psychological condition which may reduce familial anxiety (Jenkins & Arribas-Ayllon, 2016).

However, participants indicated that the patient demand for psychiatric genetics was low, and the impact of uncertainty of psychiatric diagnoses, where many disorders have overlapping features, was a concern for the use of PGC and may be a barrier to this service (Jenkins & Arribas-Ayllon, 2016). Similarly, Rowlatt, McAllister and Cuthbert (2022) found that genetic HCPs felt that they needed more resources, such as increased workforce capacity and key information such as testing availability, risk estimates, clinical features and symptoms, impact, and diagnostic criteria of mental illnesses to feel more comfortable to provide PGC. While both positive (such as reducing anxiety, guilt and shame, feeling validated) and negative (pessimism, unhappiness, and frustration) emotional outcomes of PGC were anticipated, some HCPs had a positive attitude toward delivering PGC, and others were more hesitant (Rowlatt, McAllister & Cuthbert, 2022).

c. Implementation challenges

As reviewed by Austin & Honer (2005), there are several reasons why GC for psychiatric conditions is complex and challenging. Some of these reasons include that patients and families are profoundly impacted by stigma and guilt, the role of genetics in mental illness is a growing field of research, the environment plays an important factor in disease pathogenesis, and recurrence risks are difficult to establish (Austin & Honer, 2005).

The lack of understanding of what PGC services can provide to patients is a major barrier to implementation of these services. Previous work showed that most people believe GC is a service that assists couples to have children with desired traits, advise them on whether to have children, and prevent hereditary conditions and abnormalities (Maio et al., 2013). It is also a common misconception that GC is only indicated when there is a genetic test available for the condition in question (Moldovan et al., 2019). Such misconceptions about the profession may prevent the service being used and may prevent patients and families benefiting from PGC.

Implementation of a new PGC service will likely need to be established by working closely with psychiatrists, as most patients and families which may benefit from PGC will be in the care of a psychiatric service. Physicians were found to refer to PGC based on their perception that PGC is purely an information-providing service and will refer to PGC if they believe they could not address the patient's needs themselves (Leach et al., 2016). This shows that awareness of the scope of PGC and outcomes of GCs in this area is imperative to motivate for this service to be accessible to a broader range of patients (Leach et al., 2016).

1.4. Rationale

PGC is a growing specialty worldwide, but it is not yet offered routinely, despite research suggesting benefits to patients such as increased patient empowerment and self-efficacy and a potential to decrease psychotic symptoms experienced (Inglis et al., 2015; Morris et al., 2021). A specialist PGC services is not offered in SA. Based on previous PGC outcomes (as discussed in section 1.3) and with a lifetime prevalence of 30.3%, individuals with a psychiatric disorder, and their families, may benefit from such a service (Stein et al., 2008). Thus, investigation into the current landscape of PGC services in SA may help determine if such a service would be valuable to mental healthcare users, and to guide the potential implementation of such a service in future. In this context, the current landscape of PGC refers to investigating what, if any, kind of "informal PGC" is currently happening at the clinics and hospitals where patients receive care. Given the proposed benefits of PGC reported previously, and the large burden of mental illness in SA, this project aims to investigate the current landscape and opinions of PGC services among psychiatric HCPs, who frequently interact with patients who have psychiatric conditions, on offering a PGC service. Exploring psychiatric HCPs' views on PGC and what kind of "informal PGC" is currently happening may help guide the potential implementation of an established PGC service in the future.

1.5. Aim and Objectives

a. Aim

To investigate the current landscape of PGC services and opinions of psychiatric HCPs, who work in the public health sector within the Groote Schuur Hospital catchment area in Cape Town, on providing a PGC service.

b. Objectives

1. Explore the types of questions psychiatric HCPs experience patients and families to ask when accessing psychiatric services in terms of aetiology and recurrence risks of their illnesses.

2. Explore the opinions of psychiatric HCP’s regarding their ability to address concerns in relation to aetiology and recurrence risk of psychiatric illnesses.
3. Explore psychiatric HCP’s understanding of genetic counselling as a profession.
4. Investigate psychiatric HCP’s views on the need for additional PGC services in their setting.
5. Determine what psychiatric HCP’s believe the benefits of a PGC service will be.
6. Explore the perceived barriers of implementing a PGC service.

Chapter 2: Methods

2.1. Study Design

This study followed a pragmatic qualitative research approach, also known as qualitative description (QD) (Sandelowski, 2000). Qualitative research is useful in developing knowledge in less understood areas of healthcare (Fossey et al., 2002). Due to the nature of the research question, we do not have any prior knowledge to build on, thus a qualitative research approach will allow us to explore this research question. Due to the nature of this study, which aimed to understand the current landscape of PGC in the public sector in Cape Town SA, with the anticipation of potential implementation of PGC services in the future, a pragmatic approach was best suited. Pragmatic research aims to provide a rich descriptive summary of an event or subject, using the everyday terms and vocabulary of the event (Neergaard et al., 2009; Sandelowski, 2000). This is a practical approach to answering research questions that provides descriptive accounts that may inform professional practices (Savin-Baden & Major, 2013). This research approach "draws upon the most sensible and practical methods available in order to answer a given research question" (Savin-Baden & Major, 2013:171).

Pragmatic qualitative research offers many advantages due to the flexibility of this approach. One of the advantages is that this approach does not require philosophical or methodological "orthodoxy", so researchers are free to use the most appropriate approaches to best answer their research question (Savin-Baden & Major, 2013). However, there are disadvantages to this approach as it may be viewed as less theoretically rigorous and there are no clear standards for evaluating pragmatic qualitative research (Savin-Baden & Major, 2013). To ensure quality in this study, the researcher sought descriptive validity, as discussed in section 2.9 "Validity and Reliability", by peer examination of the themes and findings during data analysis, as suggested by Savin-Baden and Major (2013). The study adopted a post-positivist paradigm, which acknowledges that the researcher in not objectively able to view the world, but that knowledge should be generated as objectively as possible (Maksimovic & Evtimov, 2023).

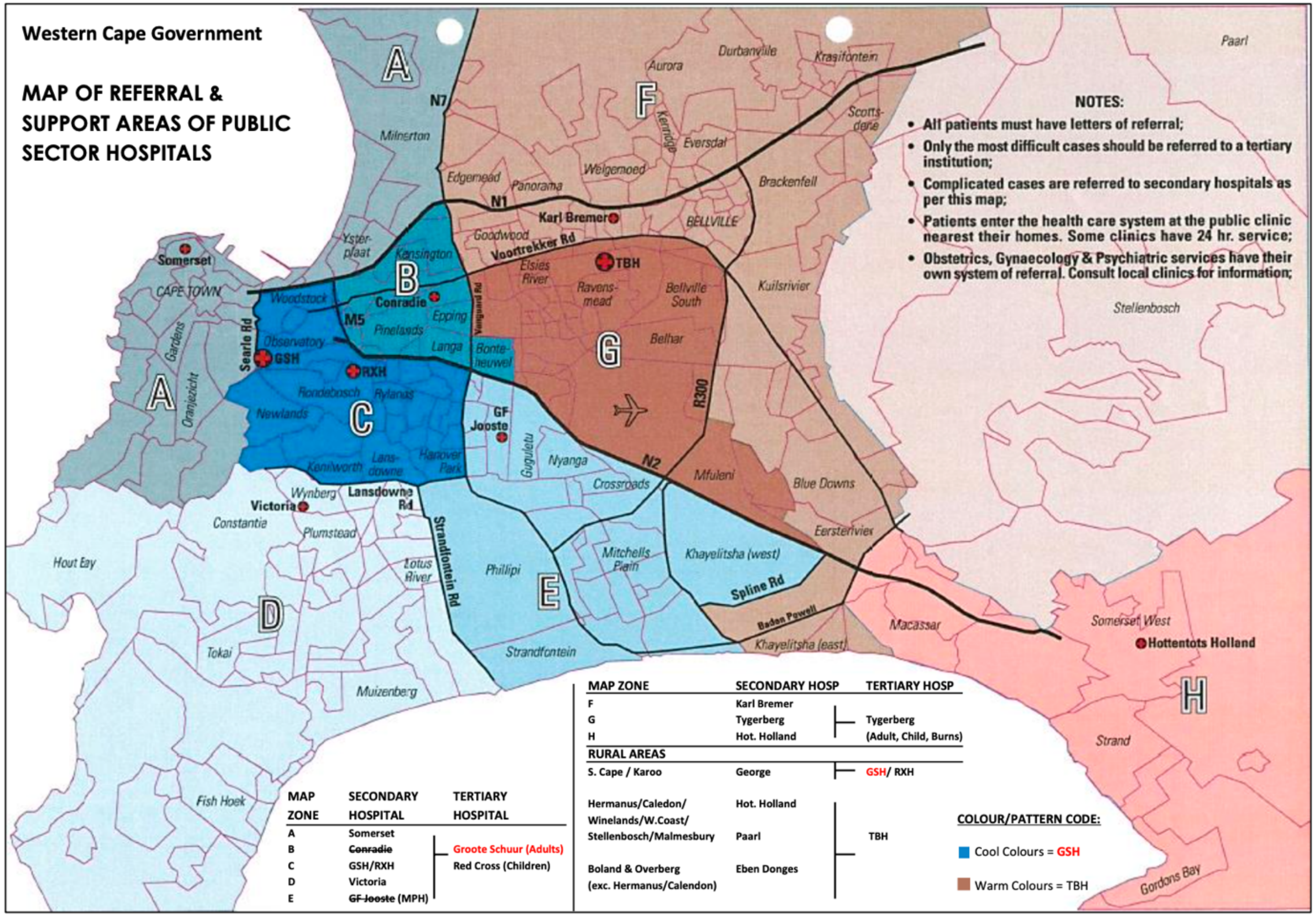
2.2. Ethical considerations

This study was approved by the University of Cape Town Human Research Ethics Committee (HREC REF 926/2023, Appendix A). Approval from the relevant institutions to conduct research was sought via the National Health Research Database (NHRD) under the reference WC_202312_028. Ethical approval was sought from the NHRD, Western Cape Department of Health, for each psychiatric unit/institution targeted. These locations included, but were not limited to: Alexandra Hospital, GSH, Lentegeur Hospital, Valkenburg Hospital, New Somerset Hospital, Mitchell's Plain District Hospital, Victoria Hospital. Institution specific ethical approvals are available on request in an effort to protect participants anonymity. Other ethical considerations such as informed consent and privacy and confidentiality are described in sections 2.10 and 2.11.

2.3. Research setting

The aim of this research study was to understand the current landscape of PGC services in the public sector in Cape Town, SA. Thus, the study took place in hospitals, clinics and psychiatric hospitals that fall within the Groote Schuur Hospital (GSH) catchment area in Cape Town. Due to the nature of this research project (the research project is a minor dissertation in partial fulfilment of the degree MMedSc Genetic Counselling), the research was limited to the GSH catchment area. The GSH catchment area (Figure 1, cool colours A-E) includes primary care facilities and district/regional hospitals in the Cape Metropole, that refer to GSH, a tertiary hospital.

Care for patients with a mental illness works in a tiered manner in the public healthcare system. This tiered approach includes primary care facilities (mental health clinics), secondary care (district hospitals) and tertiary care (psychiatric hospitals) (Lund et al., 2010). Patients with stable conditions are discharged from mental health clinics (primary care), when possible, and referred to general clinics and community health centers for their required mental health care (Lund et al., 2010). A community mental health clinic generally is staffed with a professional nurse and a psychiatric registrar (Lund et al., 2010). Patients who require admission to a psychiatric hospital will typically present to a primary mental health clinic first, where they will then be referred to secondary or tertiary care facilities for medical assessment and a 72-hour observation in accordance with the Mental Health Care Act (2002) (Lund et al., 2010). In the West Metro (Figure 2), there are approximately 30 psychiatrists, 30 psychiatric registrars, 30 psychologists and more nurses and psychiatric nurses (D Stein, personal communication 2024, November 1).



Western Cape Government

MAP OF REFERRAL & SUPPORT AREAS OF PUBLIC SECTOR HOSPITALS

NOTES:

- All patients must have letters of referral;
- Only the most difficult cases should be referred to a tertiary institution;
- Complicated cases are referred to secondary hospitals as per this map;
- Patients enter the health care system at the public clinic nearest their homes. Some clinics have 24 hr. service;
- Obstetrics, Gynaecology & Psychiatric services have their own system of referral. Consult local clinics for information;

MAP ZONE	SECONDARY HOSPITAL	TERTIARY HOSPITAL
A	Somerset	Groote Schuur (Adults) Red Cross (Children)
B	~~Conradie~~	
C	GSH/RXH	
D	Victoria	
E	~~GF Jooste~~ (MPH)	

MAP ZONE	SECONDARY HOSP	TERTIARY HOSP
F	Karl Bremer	Tygerberg (Adult, Child, Burns)
G	Tygerberg	
H	Hot. Holland	
RURAL AREAS		
S. Cape / Karoo	George	GSH/ RXH
Hermanus/Caledon/	Hot. Holland	TBH
Winelands/W.Coast/ Stellenbosch/Malmesbury	Paarl	
Boland & Overberg (exc. Hermanus/Calendon)	Eben Donges	

COLOUR/PATTERN CODE:

🟦 Cool Colours = GSH

🟫 Warm Colours = TBH

Figure 1: Map of referral and support areas of public sector hospitals (electronic communication from GSH). GSH catchment area = cool colours A-E.

Figure 2: Map of health subdistricts in Cape Town (Redfern, Westwood & Donald, 2016).

2.4. Research Participants

a. Inclusion criteria

Participants targeted to take part in this study included psychiatric nurses, psychiatrists, psychiatric registrars, medical officers working in psychiatry, psychologists, psychology interns, and other HCPs working at a psychiatric unit in the GSH catchment area. The study participants worked in psychiatric units and had regular interactions with patients with a mental health condition. The requirement for participants to have regular interactions with patients in a clinical setting was to be able to provide in depth accounts of their experience working with mental health patients and express their outlook on the implementation of a PGC service. Participants did not need to have any prior genetic training.

b. Exclusion criteria

Individuals who were not proficient in English were excluded from the study. The research study was conducted in Cape Town, SA, thus HCPs are expected to be proficient in English, as this is the language used in medical communication in this setting. This also eliminated the need for a translator to be present during the interview and the need for post-interview translation, and transcription, of the interview to English. Additionally, this is a minor dissertation with no funding available for translation. Psychiatric HCPs who did not work in a clinical setting and see patients regularly, and junior staff members with less than one year of experience working with patients were excluded from the study, this was to ensure participants had adequate experience working clinically with patients to speak on their experiences during the interview.

2.5. Recruitment

Recruitment occurred purposively and using snowball sampling. Purposive sampling allows researchers to approach appropriate participants that are most likely to yield useful and relevant responses and helps select participants in a way that uses limited resources, effectively (Kelly, Bourgeault & Dingwall, 2010; Palinkas et al., 2015). Furthermore, purposive sampling aids in matching participants to the aims and objectives of a study, which will improve research rigour and trustworthiness (Campbell et al., 2020). Purposive sampling was best suited for this study due to the limited number of potential participants and because the research question explores opinions of psychiatric HCPs, these participants needed to be purposively sought to accurately address the research aim. All psychiatric nurses, psychiatrists and other psychiatric HCPs working at the identified psychiatric units within the catchment area were identified to be of interest and contacted for participation in the study. An invitation letter and the study information sheet (Appendix B and Appendix C) were sent to members of the Department of Psychiatry and Mental Health at the University of Cape Town and GSH by the secretary of the department, informing them about the research study and providing contact details of the researcher, should they want to participate in the study. Research supervisors also contacted any individuals they knew who worked in psychiatry and asked if the research student can contact them for participation in this research study. Thereafter, the researcher contacted the consenting psychiatric HCPs to discuss possible participation in the study. In addition the researcher attended the psychiatry staff meetings to explain the purpose of the study invited potential participants to contact her if they were interested. Because potential participants were likely colleagues or acquaintances of research supervisors, this may have motivated them more to participate, however it likely did not have an impact on data collection or analysis, as identifying information were removed from interview transcripts that were shared with research supervisors. Participant anonymity was also ensure during the informed consent process. If individuals were interested in participating in the study, they were then invited for individual interviews. The researcher provided the HCPs with the study information sheet (Appendix C, via email, telephonically, or in person) and thereafter used snowball sampling to recruit more participants. Snowball sampling is a recruitment technique where participants are asked to refer more participants (Naderifar, Goli & Ghaljaei, 2017). Thus, interested participants were asked for assistance to provide contact information for more eligible participants.

2.6. Data Collection

Individual semi-structured interviews, typically used for pragmatic qualitative studies, were conducted as a means of data collection (Neergaard et al., 2009). Semi-structured interviews as a data collection method are versatile and allows all participants to be asked the same set of questions, but in a flexible framework, as the interviewer can improvise and follow up with questions based on the participant's responses (Dearnley, 2005; Kallio et al., 2016). The interview guide serves merely as a structure for discussion during the interview, it is not followed strictly and guides the participant on what to talk about and expand on (Kallio et al., 2016). The open and flexible nature of such interviews also allows for rich answers, so that new ideas can emerge (Dearnley, 2005). Some questions of the semi-structured interview guide (Appendix D) were based on the study by Jenkins and Arribas-Ayllon (2016), which explored accounts of psychiatric HCPs working in the UK on their opinions of the prospects of introducing GC for psychiatric indications in their setting. The interview guide went through two rounds of internal testing, where the guide was reviewed by the research supervisors (Barriball & While, 1994; Chenail, 2011). This step allowed for the interview guide to be refined by removing ambiguities and biases arising from the interviewer (student researcher), as research supervisors were external to the original development of the interview guide and could point out potential vagueness and biases the researcher may have had (Barriball & While, 1994; Chenail, 2011). The interview guide also went through a round of external assessment, where the guide was reviewed by an interdepartmental research committee, consisting of a genetic counsellor and researcher in psychiatry who were not involved in the current research project. Suggested changes were made based on their reviews such as rephrasing questions, making some interview questions more specific, and adding appropriate probes after certain questions. This helped establish the appropriateness and comprehensiveness of the guide (Kallio et al., 2016).

Once approval from the relevant ethics committees was received (HREC REF 926/2023), initial practice interviews (three) were conducted with colleagues (for example genetic nurses, medical genetic registrars and genetic counselling interns) in the Division of Human Genetics at the University of Cape Town. These colleagues were appropriate practice interview participants as they were easily accessible and could provide insight into the interview process as many of them have conducted and/or participated in qualitative studies before. Practice interviews were not performed with psychiatric HCPs due to the limited number of possible participants, thus we aimed to avoid restricting the actual number of potential participants from which interview data may be analysed. Practice interviews allowed the researcher to test the guide, become familiar with the questions and flow, confirm if the interview guide covers all relevant topics, and to identify if any changes to the interview guide were needed (Kallio et al., 2016). The practice interviews also provided an estimate of the total time needed to conduct an interview. All three practice interviews were transcribed and transcripts were reviewed by the researcher and research supervisor. Minor changes were made to the interview guide after practice interviews to better align with the aims of the study and assist with the flow of the interview.

After the practice interviews, the researcher recruited participants to the research study. The researcher conducted all the interviews between April to August 2024. Interviews occurred via video call (via Microsoft Teams) and in-person at the University of Cape Town Health Science Faculty, or the relevant psychiatric unit where the participant was employed. Participants were given the information sheet and were required to sign a consent form (Appendix E) before conducting the interview. Interviews were audio-recorded and lasted between 34-70 minutes. Field notes were made during and after interviews, noting important points raised. Interviewing continued until data saturation was achieved. Data saturation is a measurement used to determine when sampling can stop, in the case of this research, when interviewing can stop. Barney and Anselm (1973:61) defined saturation as “no additional data are being found whereby the sociologist can develop properties of the category”.

2.7. Data Analysis

Audio-recorded interviews were transcribed using Descript software. Descript is a secure platform that can automatically and accurately transcribe audio files into text (Descript, 2023). After transcription using Descript, the transcribed interviews were compared to the original recorded interviews to ensure accuracy of transcripts. Once transcripts were completed, these were uploaded to NVivo, which was used as a data organising and analysis tool. NVivo is a qualitative data analysis software tool that helps researchers analyse text and subsequently identify themes and patterns in interview data, helping the researcher draw meaningful conclusions during the data analysis stage of qualitative research (Information and Communication Technology Services University of Cape Town, 2023). Transcriptions were subsequently analysed by a modified version of thematic analysis, named coding reliability thematic analysis, which typically involves generating themes that consist of summaries of the most frequent things that relate to certain topics in question (Braun & Clarke, 2021). The method of analysis followed the same steps as a similar study by Rowlatt, McAllister & Cuthbert (2022). The researcher started with familiarising herself with the data by reading and re-reading transcripts and making notes of any interesting components of the interviews. This was followed by reading the transcripts again and assigning initial codes to each line or phrase. Initial codes were then grouped according to their similarities and differences. Initial themes were identified and further refined as codes and themes were being developed (Rowlatt, McAllister & Cuthbert, 2022). This modified approach to thematic analysis can be appropriate for a post-positivist research paradigm (Braun & Clarke, 2019). To ensure agreement on codes, the research supervisor reviewed codes derived from the first two interviews, after which frequent meetings were held to discuss codes, themes, and research findings. This is in keeping with a coding reliability thematic analysis approach (Braun & Clarke, 2021).

Data analysis occurred concurrently with data collection, as the aim was to interview participants until data saturation is reached. Thus, when an interview was completed, the interview was transcribed and coded before the next interview occurred. This allowed for new ideas or concepts that emerged to be addressed in subsequent interviews and allowed for determination of data saturation, which was the point in data collection when no additional issues are identified, data begin to repeat, and further data collection becomes redundant (Barney & Anselm, 1973; Kerr, Nixon, &Wild, 2010). A final (tenth) interview was conducted to determine whether saturation was reached after analysing all prior interviews, and no additional issues were identified.

Where results are reported in Chapter 3, direct quotations from participants are included to illustrate and emphasise themes and ideas that emerged from the interview data. Some quotations were shortened, by removing filler (such as “um”, “yeah” and others) or repeated words (Cristancho, Watling & Lingard, 2021). Square brackets were also used to clarify quotations (Cristancho, Watling & Lingard, 2021). These alterations did not change the meaning of the quotation in any way.

2.8. Reflexivity and trustworthiness

Reflexivity as reviewed by Dowling (2006) is the researcher’s attention to their role in the research, and continuous evaluation on how their experiences shaped the research project stages. Thus, throughout this research, all steps taken during the research process were documented, to ensure accountability and reproducibility. To ensure reflexivity and trustworthiness, the researcher documented reflections after each interview and employed reflective practices with the research supervisors, with regular meetings during the analysis phase, to maintain an objective view, document how their experiences shaped the research project stages, and to remove biases from the analysis phase. An additional reflection of the researcher’s own stance and potential biases is included in section 4.5.

2.9. Validity and reliability

Validity and reliability in qualitative research refers to the suitability of the processes employed in the study, and the replicability of the results, respectively (Leung, 2015). Noble and Smith (2015) suggest some strategies to ensure validity of qualitative research and many of these were employed in this study to ensure validity and reliability of the research. The researcher documented all steps of the research process, so to ensure accurate record keeping and accountability, as mentioned above. The interview recordings were transcribed verbatim to accurately reflect what was said, and the researcher listened to the recordings again to become familiar with the ideas that emerged from interviews. Finally, themes and codes derived from the interview transcripts were checked by the research supervisor to ensure accurate data analysis and interpretations of the interviews.

2.10. Informed consent

The purpose and aim of this research were discussed with participants at the start of each interview. Participants were informed that their participation in this study is entirely voluntary and they had the option to opt out of the study at any time. Participants were given a study information sheet (Appendix C) and an informed consent form (Appendix E) to sign before commencing with the interview. The responsibilities of participants, required for this study, included their honesty and time during the interview process.

2.11. Privacy and Confidentiality

Participants' privacy and confidentiality were protected throughout the study. Participants were informed that their interviews would be recorded and that the recordings would be discarded once the dissertation was examined. Any identifying information, such as names or places of work, were excluded from the transcript data, and each participant received a participant number to maintain anonymity. Participants were informed that sections of their interviews may be quoted in academic articles, posters, presentations and the research report. To ensure confidentiality when reporting results, only the participant number, occupation and level of clinic/hospital where they worked at were included in the transcripts and final dissertation. This was done due to the limited participant target sample in this study, which may possibly be identified with such information. No other identifying information was used in data analysis or included in the final dissertation. Quotations were also anonymised to protect participants' identity. Participants were advised that only the researcher and her supervisors had access to the participant's personal information. All data (audio and written) generated from this study were kept on the researcher's password protected computer, and in secure cloud storage to ensure that the research data is managed effectively.

2.12. Risks and Benefits to Participants

There were no foreseeable risks to the participants of this study. Participants were reassured that they may choose not to participate at any time, and they may choose not to answer questions during the interviews. Participants were also reassured that their responses were anonymised, to protect their identities, so as not to pose any risks to their roles as HCPs. Additionally, this study employed interviews as a means of data collection, thus participants took time out of their schedules to be interviewed. The researcher ensured that the interviews would not exceed the time participants were willing to allocate, ensuring they would not face any inconvenience or delays in resuming their work.

The potential benefits to the participants would be that this study may inform the larger scientific and GC community of the current state of GC in psychiatric units of the public health sector in Cape Town. Previous studies showed healthcare workers perceived PGC to be beneficial to patients, but due to time limitations, they do not often engage in lengthy discussions about genetics with their patients (Cardiff, Ecker & Austin, 2023; Jenkins & Arribas-Ayllon, 2016). This study may spark interest and guide further research into PGC services in SA, which could potentially support the current roles of psychiatric HCPs caring for their patients, in psychiatric units (by possibly aiding in lengthy, time-consuming discussions and empowering their patients (Inglis et al., 2015)).

Chapter 3: Results

3.1. Setting of psychiatric services

Most participants worked in both inpatient and outpatient services (Table 1). Generally, their encounters with patients in inpatient services differed from those interactions with outpatients. Participants described that typically, different psychiatric conditions are seen in the inpatient vs outpatient services. Patients experiencing an acute psychotic episode are managed as an inpatient, whereas outpatient services manage patients who have had a diagnosis for a longer period of time and focuses on long term management of patients. Patients are typically seen every few months or weeks in outpatient clinics, whereas inpatients are managed in hospital and discharged with a management plan for follow-up treatment at their local clinic. Because inpatient services mostly deal with acutely ill and psychotic patients, participants reported that patients are often too unwell to ask questions on the causes or risks to family members of their illness. It seemed that family members take on the role and responsibility of asking for more information or clarity when a loved one is admitted. Patients managed in outpatients are typically more stable and have more insight into their condition. Here it seems that HCPs receive more questions from the patient about their treatment or other aspects which are discussed later in this chapter. Most participants received questions about causes of psychiatric conditions from patients and families, although fewer reported patients and families asking about inheritance risks of a psychiatric condition. Many participants reported that more counselling happens in the outpatient clinics than inpatient services, and inpatient counselling generally happens only when patients ask. Although psychoeducation is a routine part of interactions with patients, some participants felt that not enough psychoeducation is routinely being done.

3.2. Participant Demographics

A total of 10 participants were included in the research study (Table 1). Potential participants were contacted via email, or during their ward rounds with snowball sampling being used to recruit further from there. There were approximately five potential participants whom the researcher contacted but did not receive any feedback from after several attempts to make contact. P5 attributed the novelty of the field of PGC as an explanation to why HCPs were hesitant to participate, as they may be unsure as how to respond to questions on this topic in an interview.

> *"You'll probably find it a little bit in terms of like, that with the uptake of the study. People are like, 'Oh gosh, I don't know what I'm going to say there.' And that's why there's a little bit of like almost hesitation to like to say, 'Well, I don't know what I'm going to add in the 40-minute interview. I don't know where I'm going to go. I don't know what my thoughts are because it's quite new.'" – P5*

Three eligible participants declined involvement in the research, either due to not being available for an interview or not wanting an interview to be audio-recorded. All interviews were conducted in English. Most interviews were conducted in person (n=8), and only two interviews were conducted online via Microsoft Teams. All interviews were conducted with only the researcher and participant present. Most of the participants were female (n=8), and more than half of participants were between the ages of 25-34 years or 35-44 years (n=8). A total of four Consultant Psychiatrists, four Psychiatry Registrars, one Professional Nurse and one Medical Officer were included in the study. Only one participant worked exclusively in inpatient services and two participants worked in outpatient services only.

Table 1: Participant demographics

	n
Self-reported sex	
Female	8
Male	2
Age	
25-34	4
35-44	4
45-54	1
55-64	1
Job Title	
Consultant Psychiatrist	4
Psychiatric Registrar	4
Medical Officer	1
Registered Nurse	1
Work setting	
Outpatient	2
Inpatient	1
Mixed services	7

3.3. Themes

Four main themes emerged from the research (Table 2). Theme one describes the uncertainty of genetics and psychiatry. Here, the uncertain role of genetics in psychiatry, uncertainties around the causes of psychiatric conditions, and the uncertainties that patients and families face when dealing with mental illnesses, are discussed. The second theme encapsulates knowledge, and how knowledge guides information seeking behaviour. The one subtheme discusses the impact of the stage of the diagnostic journey on the knowledge patients have and seek. The other subthemes explore patient and HCP's genetic knowledge, and HCPs understanding of PGC. Theme three is concerned with the long-term benefits that PGC is perceived to have on illness outcomes and the patient and family experience. And finally, theme four speaks about the ideal world of implementing a new PGC service, barriers that might hinder setup of a new service, and ways to possibly mitigate these barriers. The subthemes of time and accessibility are discussed further.

Table 2: Summary of main themes and subthemes

Theme	Subtheme
1. Genetics and psychiatry: Facing uncertainty	1.1. The role of genetics in psychiatry 1.2. Uncertainties of cause 1.3. Uncertainty from patients and families: “Is it normal?”
2. The impact of knowing	2.1. Stage of diagnosis 2.2. Knowledge on genetics 2.3. Knowledge of psychiatric genetic counselling
3. Long-term benefit only	3.1. Illness outcomes 3.2. Patient and family experiences
4. The ideal world	4.1. Time 4.2. Accessibility

3.3.1. Theme 1: Genetics and psychiatry: Facing uncertainty

Uncertainty was a core theme that participants spoke about. Feelings of uncertainty were not only experienced by the psychiatric HCPs (the participants), but also reported to be experienced by patients and their families. The role of genetics in psychiatry and the inherited factors that can contribute to the development of a psychiatric condition is discussed in subtheme one. The second subtheme focuses on the uncertainties around the causes of psychiatric conditions, both from a provider’s and patient’s point of view; while subtheme three explores the uncertainties that patients and families experience when they or a loved one are affected with a psychiatric condition.

Subtheme 1: The role of genetics in psychiatry

All participants discussed the role and impact of inherited factors on psychiatric conditions, and these factors are recognised to influence the development of a psychiatric condition. However, they reported that it is not a straightforward process to identify the exact underlying genetic influence.

> *“And I guess from a penetrance point of view, there's so many other life factors that end up impacting it that maybe it's not as easy to see sometimes, even though it's clear that there is this really strong genetic component.” – P5*

P7 explained that this inability to pinpoint an exact genetic cause in patients with a psychiatric condition influences the type of counselling that can occur, and what can be said to them and the family about their predisposition to developing a psychiatric illness. This uncertainty made P7 skeptical as to what an additional counselling service could communicate to patients.

"So, almost like a vague explanation around genetic predisposition, and then environment acts on that is kind of, is enough to, you know what I mean? Because you're not going to say it's a specific gene on a specific allele, and this is something you can do about it" – P7

Participants noted that the uncertainty of the role of genetics in the development of psychiatric conditions will make implementation of an additional PGC service complicated. P5 mentioned that other specialties like neurogenetics, where there are more definitive causes of certain illnesses (like Huntington's disease as P4 mentioned), makes it easier to motivate for an additional counselling service there.

"We've got a lot of stigma towards mental health as being all wishy washy and neuro, like neurogenetics is like, you know, that's the hard science. That's an easy sell." – P5

"For something like Huntington's, it's a lot easier, you know, so then that that's much more simple to explain" – P4

P7 described that when they were training as a registrar, research in the field of genetics and psychiatry was expanding rapidly, and they expected that a clear role of genetics in psychiatry would be identified that may influence diagnostic testing and management. However, this anticipation was never realised, which left many professionals in the field disappointed.

"I think again what it comes down to is I guess the disappointment around... So we, I really would, if you spoke to me as a registrar, I would have thought by this stage there will be genetic testing for schizophrenia. Because it's such an obviously remarkable illness. It seems quite similar to other people with schizophrenia. And the complexity of it, and the genetic inheritance and all that makes me think that there's got to be a 'Where's the gene? We need a test.'" – P7

P9 echoed P7 and stated that the unknowns in psychiatry, and the role of genetics in psychiatry, that is even evident in the literature, can cause anxiety. They felt that clarity about some genetic and psychiatric influences may make managing certain patients easier.

> *"There is a little bit of anxiety around all the unknowns, you know, because once in a while I spoke about breaking it down to basics, but it's not all our patients that have a low level of education. You get the ones that are also you know very, very highly learned you know. So once in a while I do get a lot of anxiety around the fact that there is still a lot of unknowns in the literature. There is still a lot of studies that we are doing and that still need to be done to really know what are the genes that are contributing to mental illnesses. So partly uncomfortable. And then there are other times as well where I wish this was something that you know could be accelerated in terms of what we know around it because it would make things so much easier." – P9*

Subtheme 2: Uncertainty of cause

As discussed above, the HCPs recognised that there is a genetic contribution to psychiatric illnesses, but the exact cause is not always known. Most participants explained their understanding of the cause of psychiatric conditions to be multifactorial, with the environment playing an important role in the development of psychiatric illnesses.

> *"I don't think there is a definitive answer. So we do definitely believe that it is an element of environmental, but then also genetics and epigenetics and predisposition, and then it could be substances or medications used, there's a, you know, there's a lot of causes. So I don't think you can actually point it down to one. I don't think one thing causes mental illness." – P1*

Conversely, all participants spoke about patients ascribing environmental causes, such as stressful life events, trauma, or other environmental factors to the development of their psychiatric condition.

> *"I think most people blame their life stresses childhood trauma, or you know, their adult life stresses for whatever they are going through." – P10*

“But for the most part they would say stress. It'd be like, if they're having like marital conflict, or if they you know, if they've had childhood problems a lot of the time, even if it's a primary psychotic or mood disorder they often think it is because of just stress induced as opposed to something genetic or something biological.” – P1

In addition to stressful life events, substance abuse is common in the population according to participants interviewed. The increased rate of substance use leads to high rates of substance related disorders seen by psychiatric HCPs.

“Another very common one in Cape Town and for the profile of patients that we see is substance related disorders. So substance induced psychosis. Sometimes even just you know patients that are withdrawing from substances because they've been now admitted to hospital and they can't access their substance or preferred substance while they are admitted.” – P9

For this reason, participants described that patients and families commonly believe that psychiatric conditions are caused by substance abuse, because they are often exposed to the effects thereof. As P4 described below, families of a patient admitted for psychiatric reasons frequently ask if the patient is on drugs, and if the reason they are admitted is due to substance abuse.

“But that's what they ask always, ‘Is it because of the drugs?’ And they assume because they think that's what people see in their communities.” – P4

Because substance abuse is a prevalent problem, according to participants interviewed, HCPs felt that some patients have insight to understanding that substance use can contribute to the development of a psychiatric condition or psychotic disorder.

“Some have insight into knowing that the substance that they've been taking could cause this.” – P6

In terms of genetic factors, participants described that patients do not routinely ascribe heritable causes to the development of their mental illness.

“No one has ever offered, ‘Oh because my mom had OCD, I have OCD.’” – P4

"Even if they do have a family history, the association may not always be met." – P1

As discussed earlier, participants report that patients and families have some uncertainties and questions on the causes of psychiatric conditions. However, most are reported to believe that psychiatric conditions are caused by environmental factors. Additionally, participants reported that beliefs about spiritual or ancestral causes to mental illness are widespread.

"I think, there is a chunk that may associate their symptoms to spiritual or religious beliefs. You know so within the Xhosa speaking profile of patients, there is a lot of blurring and grey areas where some of their cultural beliefs may manifest like psychotic features or manic features. So we do get predominantly from our Xhosa speaking patients, questions around "Is this purely medical or is it ancestral? Can we arrange a visit with our family back home or with our traditional healer to get a better understanding?'" – P9

"And it also does depend [on], for instance, culturally, religious, backgrounds. So some would say maybe it's because of, you know, a calling, et cetera. There's that aspect as well. Especially the black Xhosa community. They would say, 'Oh no, we need to take them to a traditional healer. We think this is sometimes demons. We think...' Religiously they would say it's demons. Or it's an ancestral calling. That is why they're presenting like this. So, yeah, some people do have like a cultural, religious explanation for the symptoms." – P3

From the interviews it was apparent that HCPs often encounter questions regarding diagnostic tests for psychiatric illnesses and methods to identify a cause for the patient's symptoms. It seemed that families want to know how the treating team diagnosed the family member with a psychiatric condition and want some kind of proof that the diagnosis is correct. P6 explained that a medical workup is often done to exclude other medical causes of the patient's psychiatric presentation, but that can cause confusion around how a diagnosis is made when blood tests come back normal, as P9 described.

"We get a lot of questions around why we can't see something on their bloods or on their brain scan, you know, 'So you're telling me that I have schizophrenia or bipolar, but the other doctors say that my scan is normal.' And they say 'My bloods are normal.' So there's also a

lot of misunderstanding and grey areas around the diagnosis. How do we diagnose and why is it that their tests are clear, but we are saying they have a mental disorder." – P9

Subtheme 3: Uncertainty from patients and families: "Is it normal?"

Uncertainty was not only experienced by psychiatric HCPs, but participants reported that they often see their patients and families grappling with uncertainties around the diagnosis.

"Yeah, mostly it's questions like that. Like, what are these things? Is it normal? Where does it come from?" – P4

Family members who are involved in the care of their loved one often struggle with taking care of someone with a mental illness or misunderstand aspects of the illness.

"So, I think, like I said, a lot of families have lived with the stress of, you know, having a member who suffers from a mental health condition, they do not really understand it, or like, how they can help their family member better, or they are so anxious, always, and all that, that they themselves end up with conditions like depression or anxiety." – P10

Some statements from participants showed that patients and families have questions about the diagnosis but might not know which services to utilise (if available), for answering their questions, or are too scared to ask their treating doctor.

"And sometimes it comes back or maybe the person would also come from the doctor and come to you and then 'Can you just explain to me again?' because sometimes people are scared to ask doctors also some questions." – P8

Participants reported that some patients ask whether their diagnosis is "normal". P9 also discussed that patients occasionally ask why the illness is happening to them. Although, it is not clear if the "why me" questions are existential in nature, or a search for the cause of illness.

"And then we also get a lot of questions around. 'Why? Why is this happening to me when no one else in my family has had it?'" – P9

Participants indicated that the types of questions patients and families ask is dependent on many factors. Some of these include the patient's personality, demographics and level of education.

"So probably that's why I have never had people really ask me deeper questions around, you know, around genetics and all those things. But I know there are clinics, like my friend who will be coming, she is at [a clinic in an affluent area]. And, the demographics there is different. So you have people coming and saying things like, 'Oh, I recently read this article on, on this. What do you think of this medication?' Or like, 'What do you think of the risk of this, of this?' You know? So it depends on [the] profile of patients." – P10

The curiosity and uncertainty around the diagnosis or causes of the psychiatric condition are not universal. HCPs reported that while some patients or family members are curious and ask questions, many, especially in the inpatient setting, do not engage with questions around the diagnosis.

"The patients themselves, they, I don't know, they tend not to ask those kind of questions. Like, 'Why do I have this illness?' They [are] not. I don't know the curiosity level. But some of them are quite ill." – P3

Additionally, some situations do not necessarily call for uncertainty or questioning, so patients do not seek explanations. As P4 explained, a patient who had a suicide attempt will not ask questions around the reasons they feel like that.

"So because if you're coming with a suicide attempt or depression, you kind of already know why you tried to kill yourself or why you're depressed." – P4

3.3.2. Theme 2: The impact of knowing

This theme discusses the knowledge of patients and HCPs, and how knowledge and understanding influences further actions. Subtheme one explores how the stage of diagnosis and life influences what knowledge patients seek. Genetic knowledge of HCPs and patients are discussed in subtheme two and the understanding of PGC is discussed in subtheme three.

Subtheme 1: Stage of diagnosis

Participants indicated that the stage where patients are in their diagnostic journey, for example, whether they just received their diagnosis, or have been living with this diagnosis for a long time, and even their age, influenced the knowledge they seek, but also influences the services they utilise.

Participants mentioned that patients who are acutely ill or who are admitted to an inpatient ward are not likely to wonder about the causes of their psychotic episode, or the implications of their diagnosis to other family members. Most participants indicated that the most common question they receive from admitted psychiatric patients are about time of discharge. This made some participants, like P4, doubtful of the utility of a PGC service for acute cases.

"But when they're psychotic, manic phases, they don't really want to know. They just want to be out of the hospital." – P3

"I do think it would be used more in things like families with intellectual disabilities rather than the things we see here, which is like SIPD or substance induced psychosis." – P4

However, participants discussed that as the acute episode subsides and patients are engaging with their diagnosis, then questions arise and patients would start thinking about implications and causes of their episode.

"They have a lot of questions. But it's usually when they're more settled. When they start, you know, getting better." – P3

Patients who have been living and dealing with their psychiatric diagnosis for a long period of time were also thought to be less suitable candidates for PGC, or less likely to be concerned about aspects that PGC deals with, such as the causes of their mental illness. P7 explained that this might be because these patients have already obtained an understanding of their condition that has developed with living with their condition for a long time.

"If I actually look at it on a day to day, it's not like you're going to get a flood of referrals from psychiatry to try and understand. You know, a lot of people have been living with a

condition for a long period of time, and they have kind of an understanding that's developed over time." – P7

This is in contrast with the time that patients are initially diagnosed, where P3 explained that a lot of psychoeducation about the diagnosis and implications of the condition need to happen, because families and patients do not know about the illness and what it means for them going forward.

"[You get] all of those kind of questions, especially when it's first diagnosis. It's much more difficult and you have to do a lot of psychoeducation." – P3

In addition to the stage of the diagnostic journey, the patient's stage of life is likely to influence their knowledge on their condition and the impact that more knowledge will have on them. Participants identified that the best times to intervene with knowledge (such as PGC) and educate patients and families about the causes of mental illnesses and the risks to their family and children were patients who have had their first psychotic episode, and prenatal and preconception scenarios.

"I think maybe for the first timers especially, then maybe you can discharge, [and have them] follow up at genetic counselling clinic. Especially for the first timers I think." – P3

"Yeah, I mean, almost like a planning for pregnancy. If you have a patient who is like, 'I'm looking at this, at wanting to have a kid. I don't know what to do about [this]. I've been stable from a mental health point of view for two years. I don't know about my medication. I don't know about things.' You have this kind of combined service to help them think through, almost like a preconception type thing." – P5

Some participants mentioned that they hardly ever get questions about risks of mental illness to future children, but that the handful of times it has happened, the patient was pregnant.

"I think one person has asked 'What is the chance if I have it, that my child will have it?' And there was one pregnant lady, that was one pregnant patient." - P1

However, P4 indicated that they had never mentioned to any of their pregnant patients that their psychiatric condition could be heritable. They indicated that they actually did not want to discuss that possibility due to the perceived limited usefulness of that information when patients are already pregnant.

"I don't think I've ever said to anyone, 'You know, this is actually heritable.' I don't want to bring that up, I guess. But you know, it's a bit late then. And even then, I'm not sure if it would help." – P4

P6 explained that a patient's insight and wellness will also influence the content discussed with the HCP as well as the approach used to explain the causes and inheritance risks of a psychiatric condition by the HCP.

"When you're talking about mentally unwell patients. Their baseline might never be fully apsychotic and fully in touch with reality, so they might never fully understand the nature of the counselling and the information that they're gathering. But the families might. So I guess the actual condition of the patient then to understanding what you're showing to them might be an obstacle." – P6

P2 described that patients with 'softer psychiatric illnesses' are more information seeking and might be appropriate candidates for PGC.

"If a patient has, from my experience, have more sort of softer psychiatric illnesses like generalized anxiety disorders, then they will be more interested in knowing how it works and what medication should be prescribed and how the medication works and they'll ask around, and then on how the illness works. And then we'll say, 'Okay, well, this is maybe your predisposing factors and things affecting it and how it will work.' Sort of basic neuroscience around it and how medication then impacts our decision, the condition and ja." – P2

Conversely, P5 saw more scope for additional PGC services for severe mental illnesses than common mental illnesses.

"I guess I see the scope for severe mental illness a little easier than I do in common mental disorders." – P5

However, certain areas of psychiatry such as child psychiatry, intellectual disabilities and geriatric psychiatry were highlighted by others as ideal areas to implement additional counselling services, due to the family involvement found in caring for children and older adults, as well as the larger genetic influence in intellectual disabilities.

"So I think the biggest areas that would be helpful would be children, intellectual disabilities, and probably old age psychiatry." – P4

"Intellectual disability is quite sort of interesting because obviously there's a big overlap with intellectual disability and genetics. And some sort of syndromal stuff is you know, that's where we would use it." – P7

Subtheme 2: Knowledge on genetics

Genetic knowledge of the psychiatric HCPs as well as patients and families was a common theme that emerged from the interviews.

All participants explained that they have never had any specialised genetic training, and that most of their genetic knowledge was acquired through formal undergraduate training in medical school. This genetic training was very broad. Most participants who had completed or who were in a psychiatry registrar training programme noted that they received some psychiatry specific theoretical genetic training early in the programme.

"So very briefly, in our second year, we go through the theoretics of genetics and that's like you're starting from just the basics, what is DNA and the DNA sequence and things like that." – P9

"So there is that component. It's not like a heavy component, but it is there.
Interviewer: I was just going to ask, what kind of things do they teach you there?
Participant: Yes, it's the basic, basics of genetics and what would really more apply to psychiatry. And some pharmacogenetics." – P10

P4 commented further that through their own interest in a particular genetic condition, their knowledge on genetics of that condition increased by self-study.

"I am very interested in tuberous sclerosis. So I've had patients with tuberous sclerosis and I've looked into the genetics of that, but that hasn't so much been taught, that is of interest." – P4

Participants noted that they do not recall much from their training, and do not use genetics often in practice. It was discussed that typical exposure to genetics as a psychiatry trainee included theoretical teachings, and sometimes exposure to other clinics such as neurology or neurogenetics, where there is established genetic service involvement.

"In my psychiatrist training, we do a six weeks rotation with neurology. And they have a neurogenetics clinic where one of their consultants works there with their cohort of patients. So I also got a little bit of time there and I think for me that was practically [where] it made more sense practically. Because now I'm a bit more aware of clinical signs and symptoms. And I also know how it relates to the field of genetics. So I think that was probably what solidified my background knowledge for me." – P9

P5 noted that genetic information is often perceived as complex information and difficult to understand. They further explained that this complexity may be a reason why genetics has not received enough attention in their training.

"I think it's because it's quite hard and complex. Or well it's perceived as quite hard and complex. I think sometimes students don't do it as much because it's quite a lot of work to know it, to understand it well." – P5

Genetic risk explanations are also often complicated by complex risk figures for different psychiatric conditions. Those risk figures are difficult to remember, so participants reported not using figures during risk conversations, but rather qualitative risk explanations based on their foundational knowledge.

"But even if I don't know the numbers by heart, I mean, for other, you know, conditions I would then go by, you know, like, high or low, low risk. But I do, I don't think it's difficult to,

to really answer when you know, like, which, or conditions have more of a genetic component to others, then, you know, you can actually say. Yeah. Or if the demographics, like, you know, is, widely prevalent in females, then [if] you would be likely a female, you know, is already a big risk factor for you. You know?" – P10

Participants' perceived preparedness to answer genetic related questions of psychiatric conditions differed, and seemed to largely depend on the level of experience of the participant. P6 found it difficult to convey genetic concepts to patients in a manner that would be comprehendible to them. They felt that due to their limited experience in that area that it was a challenging matter to address with patients.

"So I would say it's a, it's difficult, but I mean, at least at my level. Where I haven't done it all that much. Where I haven't needed to, or have been asked to do it all that often." – P6

Trainees and junior doctors felt uncomfortable describing the concept of genetic risk, and either did not feel that they were well-equipped to answer these kinds of questions, or felt just equipped enough with the basic theoretical genetic knowledge. Alternatively, participants who had more years of work experience felt equipped to address such topics. Interestingly, P5 felt that the uncertainty of the role of genetics and genetic risk in psychiatry equipped them to relay that information to patients.

"I don't think I'm well equipped to like counsel somebody on it just in, you know, explaining to somebody in like layman's terms." – P1

"In terms of feeling equipped. Certain disorders where it's a bit more wishy washy, I can give a wishy washy answer. That should be okay." – P5

However, P5 went on and explained how conveying risk information in a counselling environment is part of what they do as psychiatrists, and that they should be able to address these aspects.

"So, unless there's someone who has like, specific counselling and understanding genetics and, and how to counsel. Psychiatrists probably have more skill than general doctors in doing that. In terms of general practitioners and that. So, I mean, I guess it is part of what we

> *do. That's something we learn is the genetic risks and how it runs. Even if we don't know all the specifics, we can help someone think it through and use our therapeutic skill there." – P5*

To increase confidence and knowledge in addressing genetic risk and aetiological aspects to patients, participants reported that more theoretical training on this topic as well as practical experience would be useful. Their suggestions were to incorporate discussions about genetics into, for example, ward rounds or observe skilled and trained professionals counsel on these topics.

> *"You know, so by the time you do get a case that's relevant, you're thinking, 'Oh my gosh, what is it that was said? What did we speak about this? It's been so long since I had a case that had a neurogenetic application.' You know? So I also feel like perhaps if it was regularly spoken about in our meetings and practically in our ward rounds as we're seeing patients, then we would remember it, when we do have to consult a patient." – P9*

> *"Probably watching it done by like maybe genetic counsellors or like an understanding of how that, is usually done, you know, by someone who has done it a million times, that's probably helpful. I mean, I think that's the best way to learn how to communicate to people is watching someone who already knows how, do it, because then you pick up on what they are putting down, you know." – P4*

P9 explained that conversations about hereditary causes of mental illnesses are tricky because they feel that patients do not have a lot of genetic knowledge.

> *"So not typically that is something that, yeah, it's a far fetch when we're going to possible genetic causes. It almost feels like you are teaching them something they've never heard of before, you know." – P9*

It seemed that participants felt that individuals would not ask about something they do not know about. P5 explained that the lack of knowledge around psychiatric conditions and basic genetics such as inheritance of conditions, may prohibit patients from asking such questions.

> *"Um, but maybe they don't know enough about, how it works in families to, be able, to [ask about] it." – P5*

However, the educational attainment of patients who utilise state mental health services vary greatly. HCPs reported that the level of education of the patient influences their level of knowledge on their illness and, in turn, influences their information-seeking behaviour and questions.

"So typically people who are, have a higher education will obviously ask more kind of technical questions to want to understand better." – P2

HCPs in this study reported that they have felt that patients needed a better understanding of their condition and would appreciate an additional counselling service, as a way to ensure their patients have enough support. P7 thought that patients do not expect an explanation of their psychiatric condition, although other participants, such as P8, reported that families do not understand mental illness and that they struggle because of this.

"Patients don't, I suppose they don't expect an explanation as to why they should have the psychiatric illness." – P7

"Family struggles, because they don't always understand why it is so difficult for the person just to stop using the substance that they are using." – P8

Finally, participants have experienced that the knowledge of a genetic contribution to mental illness is not always readily identified by patients and families. Some patients (if there is a family history of psychiatric conditions) will recognise the importance of genetics and discuss that information. However, HCPs always enquire about family members with mental illnesses, and it is usually only after probing that patients identify other family members with similar symptoms or associate the condition with possible heritable factors. This speaks to the genetic knowledge of patients and families – only after these aspects are alluded to, are they recognised. From this result, it seemed that knowledge of the impact of the illness in other family members is not usually readily identified or questioned.

"And the ones that understand it well, you see where the light bulb starts going on, where it's like. 'Oh, OK. So my grandfather or my grandmother did have this. I see how that's possible.' It's never the first thing they're thinking of that it could be genetic." – P9

“I've had people say, oh, his grandmother had it, but I have always asked. No one's ever offered that to me. I ask about a family history because that sort of like, sort of dead bolts the diagnosis for me. So if you come in hearing voices and they're like, ‘Oh granny had schizophrenia’, I'm like ‘Okay, you're schizophrenic.’” – P4

“Once in a while, like that light bulb that I was talking about, once in a while, as you are explaining they may say, ‘Now that you mention, I remember a cousin so and so, who possibly had the same kind of symptoms. You know, what can we do about that?’ ” – P9

Subtheme 3: Knowledge of psychiatric genetic counselling

While all participants’ explanation of their understanding of genetic counselling as a profession was accurate, this question was met with a lot of doubt and discomfort. Many of them have not referred to genetic counselling in the past, and have had very little exposure to genetic counselling, only learning about it during their undergraduate studies. Participants generally explained the role of genetic counsellors as understanding and conveying information about clinical aspects of disease and the genetic causes of conditions, as well as dealing with the heritable implications of the condition and emotions that come with those discussions. Many participants also acknowledged the role of genetic testing and the involvement of other family members.

“So someone who has a knowledge of genetics and the clinical aspects of genetic disorders and the testing thereof and those who understand the… I don't know if inheritance is the right word… But the basically that the genetic implications for this patient and for their family and all their offspring.” – P6

“For me it is like, people come in for psychology sessions, that is how I'm seeing [it]. I'm not sure if it is like that, but that is kind of [it]. First of all, kind of speaking to family, this is what it is, dealing with the emotions that's coming from that, assisting them throughout the whole process, supporting them emotionally, and preparing them for what the possible outcome might be.” – P8

However, participants were unclear about the role of psychiatric genetic counselling and have not heard about such counselling being done in the mental health space. P2 provided an explanation of PGC to be about communicating genetic risk and encouraging the use of protective strategies to minimise the risk.

"I think in our setting, it, for me, it's about explaining the risk, the genetic risk, predisposition. And then the risk, and how to potentially maybe mitigate it as much as you can." – P2

3.3.3. Theme 3: Long-term benefit only

The anticipated effects of PGC were viewed by participants to only benefit the long-term care of patients i.e. PGC would have little effect on patients' management in the acute setting, but there would be improved illness outcomes in the long run. These aspects are discussed in subtheme 1. Participants also noted that a PGC service could benefit patients and families by improving the effects that a psychiatric condition has on the family and community, as discussed in subtheme 2.

Subtheme 1: Illness outcomes

Generally, it was stated that HCPs make use of services if it will impact their patients' management and care. Participants in this study believed that the underlying genetic contribution or any other causes of a patient's psychiatric illness would not necessarily change how they managed their patient. Thus, this aspect of psychiatric conditions is not given much attention when managing and treating patients in the acute inpatient phase of their illness, as highlighted by P7.

"It is a little bit strange, I suppose, because if you think about it, a lot of our, again, so we spend a lot of time learning about the genetics of it, but actually, the terrible thing is it doesn't make much difference because it doesn't affect the treatment, particularly at this stage." – P7

Participants in the study mentioned that discussions regarding inheritance and genetic risks are usually initiated by them if it is therapeutically indicated. An additional obstacle is the limited treatment options available in the state healthcare sector. Even if more information on causes could impact how a psychiatrist manages a patient experiencing a psychotic episode, they do not have a range of options available to treat their patients with, reflected by P4.

> *"Because the reason why it doesn't matter that I really care about why my patients are psychotic is because the patient is psychotic, and the treatment for psychosis is Risperidone. And that's what we have, and so that's what they get. And so the reason, in their particular case, is a little bit irrelevant to the management of it. It's the social things that we see more often that could change." – P4*

Incorporating PGC into current care of their patients was seen as mostly benefiting the patient and their families, and not the acute care of patients. However, participants believed that should PGC be implemented, it would slot in as part of the multidisciplinary team approach to care for their patients.

> *"I think if it's sort of a combined, discussion. Maybe say for example, the genetic [counsellors], because you guys can maybe give insight into things that we don't necessarily give insight to. And then along with that, the doctor that can give more information on if we're talking about causes other external causes and medical kind of causes, or medication related." – P2*

Care that is the most beneficial to the patient was important to participants. Here, P4 recognised that they might not always be the most appropriate person addressing certain aspects of a condition, which makes multidisciplinary team care ideal. This leads to the idea that it is a matter of involving the most appropriate person to have PGC discussions.

> *"We are all experts in our own thing, you know, and you can really harm someone by explaining something that you don't really understand to them. I've seen that a lot. I mean, I wouldn't want to explain to someone how their shoulder was going to be like reattached and I wouldn't want the person reattaching the shoulder to explain to them why they're mentally ill." – P4*

By educating and counselling patients on aspects that PGC addresses (such as protective factors, inheritance risks and causes of a mental illness), participants noted that the benefits of this might translate to better illness outcomes. HCPs in this study reported that patients have difficulty adjusting to taking chronic medication, understanding the reasons for the need for medication and often question the length of treatment or side effects. P5 felt that a PGC service could improve health literacy, which in turn, may improve outcomes such as function and relapse risk.

"I do think that health literacy helps improve outcomes. And if they understand the disorder, the management, and help them to empower them and give them ownership of it, I think you really can change function and relapse risk and all of that if they really understand it then." – P5

Participants also explained that patients often ask how long they will need to be on treatment for and thus default on their treatment for various reasons, as P8 explained that treatment adherence may keep patients out of work.

"So that question, I do get a lot, until when? Because many times people default… Because staying, coming here for a monthly injection means staying out of work."– P8

HCPs in this study felt that a service such as PGC might lead to better adherence and thus improve long term health outcomes like decreasing relapses, because patients will understand the need for treatment and adhere to their treatment.

"I think it would eventually lead to better adherence to medication because, you know, you kind of eliminate all the misconceptions as well with regards to that and like more motivation overall and better health seeking behaviours in the long run." – P3

"The benefits of doing that kind of counselling, you're preventing, hopefully, relapse, and then you can have lifestyle modifications. And, in the long term that then obviously has beneficial impact on the environment or the resources they usually use by, I mean, you're not filling up the hospitals." – P2

P2 indicated that PGC may be beneficial to hospital resources, and P9 shared the same view that improved knowledge may decrease relapses and perhaps help unburden the healthcare system.

"So I think overall I, I like to talk about just the healthcare system and how burdened it is and practical things that we can do to unburden. You know, and I think that would be one way to avoid this vicious cycle where patients keep coming back because there's gaps in their knowledge and in their awareness. I think it would just be part of closing up that gap and making long term care, you know, a possibility so that we don't keep having acute cases." – P9

As P2 explained, they felt the long-term benefit of PGC may be preventing relapses and patient admissions. They also pondered about evaluating the effects of such a service on actual patient outcomes.

"It would be interesting to see that once one implements it, what happens to admission rates, what happens to readmission rates, to actually objectively say whether an intervention like that is helpful." – P2

Subtheme 2: Patient and family experience

The perceived benefits by participants of an additional PGC service was focused on patient and family experiences. PGC was perceived to be a service where patients can receive more information and participants felt that having a more informed patient could alleviate pressures on the healthcare system (as discussed in subtheme 1 above). HCPs pointed out that more information via a service like PGC may have other long-term benefits for the patient and family experience such as increased understanding of familial implications, correcting misconceptions of the illness and alleviating stigma, increased knowledge on early warning signs for other at-risk relatives and when and where to seek help if other family members require. HCPs also felt that more information to plan the future, relieving anxiety about the unknowns of the illness and an overall improved understanding of the mental illness would be beneficial to the patients' and families' experience in the long term.

"And the families are also aware, maybe aware of if there is a special need that the person does need, how to actually optimize their living space and how to actually provide an appropriate comforting space." – P1

Participants reported that with this improved overall understanding, patients may have a better quality of life, feel more empowered and understood, and be of increased benefit to society.

"I think their life will have a better outcome." – P8

"Like, obviously, like, for the prognosis of the patient, I think in general, it's super important for them." – P10

"Then in society you have a more functional society because the more people relapse, um, the more they get cognitive decline, the less functional and more dependent they become." – P2

P9 discussed that there may be long-term benefits to society by raising awareness on mental health conditions, resources, and how to seek care in the community. They commented that by employing a PGC service, awareness around mental illnesses and the services available could make a difference in how communities seek help when needed.

"I think it might just in the long run it would raise awareness which is key for any patient diagnosed with a mental disorder in terms of knowing how to manage, you know, long term follow up. So I think in that sense it would really help patients just to have that awareness of what to do when we are with them at home. How can we help to keep this disease under control. But I also know that people in communities talk to other people in communities. So the next time someone is also presenting with behavioural features, it's easier for this mommy to say to the next mommy, 'You know what my son also had this and there is people who can help and there's a possibility that this, you know, we might need to explore if it's part of your family. This is what I did that could help.' You know?" – P9

Even though educating the public about mental health issues through a PGC service will benefit the community, as P9 mentioned above, it will be crucial for HCPs to be aware of the service's availability (should it be implemented) and what it can provide so that the right patients and families can be referred to use it.

"I think if the clinicians were informed of the service and knew the appropriate referrals to make, I do think it would be used." – P6

Families are often involved in a patient's care, especially when the patient is a child or elderly. In acute cases, families often have a lot of questions because the patient may not be well enough to attend to the questions they have. Participants felt that PGC could be a service catered for families who raise questions, to help them make meaning of a diagnosis, and assist with equipping families with the correct information to assist the patient's illness outcome.

"Kind of to equip the family as well in terms of dealing with what's, what's expected and kind of for them to also make meaning of what just happened because I do, I mean it's a very sensitive thing. And I do see how frustrated families become and they actually mourn, like it's... they grieve. It's like a loss when you actually tell them, 'Okay, so your child...' Because they're usually young as well and they have their lives ahead, they're in school, they're in varsity, now having first episode psychosis, they don't know what's happening. Um, so they, yeah, you, I think, especially that, on that crucial moment where, because then they, otherwise they open to like misconceptions. So then they all go read up on Google, 'Oh, the doctor said schizophrenia.' Then next thing they go read unreliable sources. They ask other people who have, you know, gone through this and they get misinformed about stuff. And then you find that sometimes they come back and they tell you, 'My child is not going to take this medication.' You're like, 'Okay, why?' They're like, 'Oh so and so took this medication. We gave them this. You know, so and we are not doing this.' You know, so now it becomes difficult because now you have to explain, 'Okay, just because the other person, your neighbour got this doesn't mean that you…', you know? So this kind of a service like would really then put things into perspective for them as well." – P3

PGC was also seen as a potential de-stigmatisation method, and by increasing patients and families' understanding of mental illnesses, it can impact their experience in a positive way. P3 mentioned that more understanding will help lift the blame that families experience.

"But I think having that understanding and having a little bit of the stigma of mental illness, um, removed is, is always a good thing." – P4

"Families also now get to know better and stop blaming themselves for what is happening" – P3

Participants reported that patients and families who are in mental health services face a lot of stigma. This often leads to patients defaulting on treatment because going to the community health center for treatment means that others would realise their reasons for attending.

> *"Where, if you're sitting in a community health setting, in the day hospital, everybody see[s] you're going to go to the mental health sister's office. You understand? That is, that is the reality of it. So people are still stigmatizing. They don't want to be seen there." – P8*

This shows that stigma surrounding mental health and even different cultural beliefs may hinder treatment of mental illnesses and how information like that discussed in PGC can be conveyed to a patient.

> *"I think some of the patients do come, and there may be a cultural component, but they may not necessarily agree with what you say or they already have their preconceived ideas of, of why they may have a mental illness on this or why their child has it." – P1*

Participants described expecting the long-term benefits of additional counselling services on causes and genetic risk to not only be improved illness outcomes for the current patient, but also for the family and future generations. Such families with histories of mental illnesses seemed to be ideal targets for a PGC service according to participants in this study. HCPs felt that conversations about the possible inheritance risks of psychiatric conditions may also benefit patients and families in the long term for early intervention. P2 felt that talking about early signs and when to seek help could be included in discussions with patients.

> *"So say for example, 'You therefore have to be on the lookout for your child's mental well-being' and that can be after the psychoeducation, there's a summary to say, 'So based on what we've discussed now, be on the outlook for your children's sake.'" – P2*

> *"Even if they have had past, relatives with mental illness, it would just be helpful to say that if you see your son or your child doing this, this and this, or something there's an increased chance, like be aware or, you know, just for early detection. I think that would help." – P1*

P4 mentioned that discussing heritability of psychiatric conditions may be an ethical issue, questioning the benefit of such conversations. This participant also assumed patients' stance on this topic and that patients and families may not want to talk about inheritance risks, due to their own perceived usefulness of this information. This lack of insight may be another ethical issue entirely.

> *"And I've never ever considered the ethics, of explaining to people that if you have schizophrenia, there's a chance that your child will. And I mean, we see tons of pregnant women because part of our thing is changing meds for pregnant women. So if I think of how many pregnant women with bipolar I see, I don't think I've ever said to anyone, "You know, this is actually heritable". I don't want to bring that up, I guess, but you know, it's a bit late then. And even then, I'm not sure if it would help." – P4*

Although questions on recurrence risks for others were not commonly reported by HCPs in this study, some participants indicated that it does happen occasionally. For example, patients would sometimes want to know how to get help for their children early on.

> *"I find majority of the time, it's more your elderly, you know? Yeah, mid midlife 40s going onwards because at that age they've probably had children or they are planning on having children and there will now be concerns around whether I need to worry for my children getting something similar. You know, so you will get questions like is there tests for example that I can do to make sure my son doesn't end up having this condition? Or in instances where perhaps they've already started picking up symptoms that are suggestive, then they may ask. Well, can I bring my children in as well for an assessment? At what point is it OK for me to bring my son in or my daughter in for an assessment? So I find it's more related to concerns about their children having similar conditions." – P9*

3.3.4. Theme 4: The ideal world

It was apparent that should a PGC service be available, participants in this study would utilise this service. However, there were many aspects participants felt would hinder the implementation and execution of an additional counselling service and the concept of an 'ideal world' scenario, where access to resources and possibilities are endless, needs to be adjusted to fit with the needs and landscape of state healthcare services. Time seemed to be a major obstacle and is discussed in subtheme one. Additionally, accessibility to services and resources are discussed in subtheme two.

Subtheme 1: Time

A major barrier to implementing conversations about genetic risks and other PGC aspects is the limited time participants reported having. Even though HCPs in psychiatry, compared to other fields of medicine typically have more time to spend with patients during consults, there is limited time to address patient questions during a consultation, especially at the primary community clinic level.

"Just for example in our community clinics sometimes we have about 10 to 15 minutes, if that, for some patients. So it's not necessarily, there's things you want to say and you want to do for the patients but may not necessarily have time, whereas [a counselling service] would really help." – P1

"We often have to make the time even. Even if it's not there (laughing) we have to make that time available. Otherwise you haven't done justice to the patient." – P9

Participants discussed that they would like to set time aside to speak to families, to answer their questions and help them understand the diagnosis, but do not have the time to do so. Thus, conversations about genetic risk do not necessarily happen, even though counselling should ideally happen during a consultation, as P2 emphasized.

"And I mean, in an ideal world, counselling should happen also in the session. That doesn't necessarily always happen" – P2

A PGC service was seen as a solution to this limited time obstacle by many participants. Such longer conversations, like those in a PGC session, could then be outsourced to save HCPs time, and would be appreciated by HCPs because that can assist with their caseload, as P9 explained.

"So I think it would be used not only because it's necessary and what should be done, but also because it would actually help in terms of our caseload. We could also be able to know that we can focus on you know, treating and diagnosing and managing symptoms. But there is a place where you know a proper in depth psychoeducation can be done. Because what normally happens is we're very rushed on our phones and we're just wanting the information

that will tell us whether this person needs to stay or not, you know and you even find unfortunately sometimes families want to speak more. And then on your side you have to cut them off. You know, so I think it would definitely be used within psychiatric services." – P9

P10 reported that when working with patients at outpatient clinics, some conversations get moved to later sessions due to the extensive work that needs to be done in one session.

"So sometimes, you know, we continue until the second session to because in one session, the history taking itself is so long and so extensive, especially, you know, in psychiatry, and when you come to, to actually your management plan you don't have that much time left to talk about, you know, the condition itself. And you are writing, yes, your precipitating, perpetuating factors and those things in your clerk. But, like, do you actually, you know, have time to explain everything? And sometimes the patients, you prefer the patients to come with the family. Also, if you need further information, or to explain to [the] family also what they're dealing with and how we are going to solve it." – P10

Subtheme 2: Accessibility

Although there were many perceived barriers to setting up a PGC service that are discussed below, participants reported that they can see the benefit to patients for an additional counselling service like this, even when they are already working in an under resourced section of healthcare.

"I think anything that supports people with mental illnesses is a positive thing. Anything that supports people with intellectual disabilities is a positive thing because there's no money spent on these things. And so we have an extremely under resourced section of health and a very high burden of disease. And so anything extra that can help people understand, I think is great." – P4

Patient and institutional finances were often discussed as a barrier to set up an additional PGC service. Institutional costs to set up a service such as administration fees and costs to employ professionals to be involved in the service, when so many services are already struggling financially, was the biggest obstacle according to P10.

"Obstacles, obviously admin. You know how many posts have been cut because of budget cuts and those things. So it does seem like mental health is not really featuring high on priority list. So definitely that's something that, you know, it's ambitious to think about how to support your patients further when doctors are less now. Like I'm having to see 15 patients a week, just in an afternoon of my clinic, psych patients, which is ridiculous. But we are doing it for the sake of our patients. So, now do you think that they will fund a post, you know, a proper set up, designated, you know, area, and people?" – P10

Furthermore, participants indicated that patient costs would also impact whether patients and families attend additional services, as many are already struggling to attend their current appointments.

"The reality is people don't have money. A lot of our patients [are] on grants. And it's already difficult for them to come here if they have to get a monthly injection for travel. So they will not want to give out more money to come for a counselling or for information." – P8

Resources are limited in the state healthcare system. Participants commented on current struggles in psychiatry such as understaffing and working in a severely resource constrained environment. They felt that finding people to work in a PGC service, who have the time and skill, when there are already few psychiatrists and GCs employed would be a barrier. The thought of adding more responsibilities or opening up more services was quite daunting for some participants.

"I feel like I want to tell you that there'd be a great need for it, but the reality is it's almost like we kind of, we sort of, we manage the patients under relative pressure. So you don't necessarily want to expand it and to take on a lot more" – P7

P5 spoke about the challenge of establishing how big the scope of a PGC service will be and how to raise awareness of the service. They reported that setting clear criteria for referrals and scope would be important due to the time it would take to see each candidate for PGC.

"Because the scope is we see a lot of patients. So, if you opened up to psychiatry in general and wanted to see everyone, this is impossible. Because the work that a genetic counsellor would do would take ages with each person, right? To make it so specific and nuanced." – P5

Participants also discussed how referrals should work and be set up, and it was mostly suggested to use existing formal referral platforms that contain detailed descriptions and specific information that can be screened by individuals involved in the PGC service. An *ad hoc* PGC service, like a liaison service, was ideal for some participants, as it would allow patients to receive important information to help their understanding of the cause of their diagnosis and assist with treatment adherence and understanding of treatment. Some participants felt like it would be ideal to refer patients just after they are discharged from the acute setting, and others felt that it would be an ideal time to intervene just before discharge, when the patient is doing better and getting ready to go home, as some patients do not attend follow up clinic appointments.

> *"Just at that point where you start thinking about sending them home, I think that would be the point where the genetics counselling team, if available would be of use. So even if it meant that, OK, we've treated this person for five days and now we feel that they're getting better, but they're not yet at the point of discharge, then we call genetics counselling and for you guys as well at that point in time, the patient has improved enough to be able to speak to you and to understand. You know, so you could then have time with them while they're in a good state of mind where they can understand and the family are able to know that before the patient is discharged, what do they need to know going forward. So I think it would just make for better discharge planning, you know, because unfortunately sometimes we're sending people home and they don't have an understanding of what it was that got them admitted in the first place. Then the whole cycle repeats. The families still don't know that this could be linked to my genetic line. You know. So I think if there was room for us to just do the clinical treatment and get them stable enough and before discharge, perhaps refer to you guys, yeah."*
>
> *– P9*

Practical difficulties like identifying target patients to refer to PGC, communication and language barriers, availability of patients and family, and patient motivation to attend additional services were discussed. The ability to communicate with a patient in their home language or in English is frequently difficult, especially the ability to convey complex information like genetic topics in a way that patients will understand, were raised by participants.

"I try… This is my personal experiences. I try use very, yeah, common language. Language that is easy for them to understand, you know?" – P9

Evidently, HCPs spoke about the need for a balance between what an "ideal world" scenario would be and working realistically with the resources available in our setting.

"Yeah, I mean, and it's, again, in the ideal world, it's brilliant. Because we'd see somebody and if they had a question, we could send them down the corridor and they could see somebody who's, you know, really tuned up and give them, give them sort of factual information. I think it would be a help for them." – P7

However, the reality is that we do not work in an ideal world where new services could easily be implemented. There were some suggestions given by participants to try to overcome the current resource constraints that might hinder the establishment of a new PGC service. Some of these suggestions included offering PGC in a group setting, thus reaching many patients and families experiencing similar uncertainties at the same time. P2 also indicated that it also might contribute to patients' sense of belonging.

"I think that for me, you said ideal world, but I think in a resource constrained environment, you then have multiple people that you can counsel at the same time. And because it's so broad, a lot of people could relate to many of the causes and risk factors. And then there's a sort of a sense of belonging that I'm here with other people who have the same difficulties than me." – P2

Other suggestions by participants included having the service available as a pathway to receive advice from an expert in the field. Additionally, to help make an additional counselling service more accessible, participants suggested offering PGC in the community, to alleviate traveling and cost difficulties from the patients and families or having pamphlets and online resources to easily reach more patients and families. Another suggestion was to integrate new PGC services into current services.

"I wonder, if like, if you had a mobile team going to places, hospitals, and they'd have, say for example, different days at different facilities where they have their genetic counselling day, that is maybe taking place at the same time as a psychiatric OPD day and maybe have a

group counselling session beforehand before they see their doctor or afterwards or then that there are slots where patients can be seen, that they see this person first and then go for genetic counselling, revised medication and then go see. I think in terms of resources, that's probably something that would work better if it's at the same place, same time." – P2

"It would be nice to have kind of backup with someone who has more specific skill with it. And maybe not to everyone because there's like one of you [genetic counsellor]. But information, so information we could direct patients to, I think would be really useful, like specifics around, understanding these types of concepts, something that's easily digestible. In lieu of a whole service that's there. So that might help support it, but I often use things like UpToDate patient information and stuff which sometimes has some of those information and then makes it a bit more easy. If there is something that's like that, that would be useful." – P5

3.4. Chapter Conclusion

This chapter described the results from this study as themes that were identified in the interviews. The main themes identified were *Genetics and psychiatry: Facing uncertainty* (Theme one), which addressed the uncertainty experienced by HCPs and patients and families involved in mental health services. Here the uncertain role of genetics in psychiatry and the uncertain causes of mental illnesses were discussed, with an emphasis on what an additional counselling service can offer. The uncertainties and questions that patients and families deal with when they have, or a loved one has, a psychiatric condition, were addressed. The second theme, *The impact of knowing*, focused on the knowledge that HCPs and patients and families have, how more knowledge will influence their actions and when they seek more knowledge. The stage of a patient's diagnostic journey as well as stage of life influences what knowledge of the mental illness is applicable to them but also influenced if and what kind of understanding and information they seek. Genetic knowledge of HCPs and patients were discussed, and it was clear that while most HCPs have basic genetic knowledge, patients and families lack genetic information, and this influences the counselling HCPs do, as well as the kind of questions families ask. HCPs' understanding of the GC profession was then discussed, but participants were unsure of what GC would look like in the psychiatric space. How participants viewed the effects of PGC was encapsulated in the third theme: *Long-term benefit only*. HCPs felt that PGC will improve patient's illness outcomes, but not in the acute setting. Additionally, the positive influence of PGC on long-term management and patient and family experience was explored. The final theme labelled *The ideal world* was considered where HCPs discussed some barriers and solutions they believe would be faced. Participants frequently reported having limited time in their day-to-day and that an additional counselling service could help save them time and contribute to the overall care of their patients. The service needs to be accessible to patients though, and within a resource restricted setting, solutions need to be implemented to maximise the impact a PGC service can make.

Chapter 4: Discussion and Conclusion

4.1. Discussion

This research study sought to understand the current landscape of psychiatric genetic counselling services and opinions of psychiatric HCPs, who work in the public health sector within the Groote Schuur Hospital catchment area in Cape Town, South Africa. To our knowledge, this was the first study in SA that explored HCPs' opinions of a PGC service and the perceived barriers and benefits of a new PGC service. As described in Chapter 3, from the 10 interviews conducted, four themes emerged from the data: i) Genetics and psychiatry: Facing uncertainty; ii) The impact of knowing; iii) Long-term benefit only and, iv) The ideal world. This chapter will discuss these results in the context of the setting of genetics and PGC and link with existing literature.

Perceived causes and uncertainties of psychiatric conditions

The role of genetics in psychiatry was covered in the first theme, along with the many unknowns of the field. Almost all study participants reported to believe psychiatric conditions are caused by a combination of heritable (genetic) and environmental factors. This is consistent with previous studies examining psychiatrists' and genetic professionals' perceptions on the influence of genetics on mental health, where participants acknowledged the genetic basis of psychiatric disorders (Martorell et al., 2019; Zhou et al., 2014). Conversely, participants in this study reported that the patients and families themselves largely believe the causes of psychiatric conditions to be environmental, and that some patients and families ask about the causes of psychiatric conditions. Consistent with previous studies, patients largely attribute environmental factors to cause their mental illness such as "problems at work", "problems in social environment", "negative life events" and others (Magaard, Schulz & Brütt, 2017). The same study found that patients very rarely (2.1%) attribute "predisposition" as the cause of their mental illness and many believe "self/internal states" was the causal factor (Magaard, Schulz & Brütt, 2017). However, another study found that 47% of mental health users believed there was a genetic basis to psychiatric illnesses (Martorell et al., 2019). However, the two studies mentioned above had different settings. Magaard, Schulz and Brütt (2017) surveyed inpatients (n=712) receiving psychosomatic rehabilitation in Germany, and Martorell et al. (2019) surveyed patients and their relatives (n=959) attending outpatient mental health service centres in Spain. The differences in setting and target participants across the two studies may have contributed to the large disparity in belief about genetic contribution to mental illness. More research should be conducted in the future to understand the beliefs of patients and families in a South African public health setting, as it is possible that experiences in SA will differ from other high income countries. Furthermore, a systematic review and meta-synthesis by Choudhry et al. (2016) found that perceived causes of mental health by patients could be divided into three main categories: most frequently psychosocial and environmental factors (stress), followed by spiritual or supernatural causes, and biomedical or genetic causes. These results are consistent with what was reported by participants in the current research from their experience of patients' causal beliefs of their mental illness. Patients and families largely attribute environmental factors to cause their mental illness, with some recognising a genetic contribution or spiritual causes.

The second theme included the impact of increased knowledge and understanding of mental illness on patients and families. This study found that according to HCPs, patients and families have questions when a family member or themselves are diagnosed with a psychiatric disorder, as detailed in theme two. Few participants recalled patients asking about inheritance risks in relation to their psychiatric condition. This is consistent with a previous study by Martorell et al. (2019) that found 12% and 10% of HCPs reported patients asking them about the genetics of their psychiatric condition and the inheritance risks, respectively. However, in the same study, 43% of mental health users were worried about transmitting their psychiatric condition to their offspring (Martorell et al., 2019). Furthermore, P1 reported experiencing only one previous patient ever asking them if her diagnosis of a mental illness is heritable, and that particular patient was pregnant at the time. The aspect of inheritance was likely very relevant to the patient at that point, as she was currently pregnant and this may be why such concerns were raised. It is possible that patients and families question the inheritance of the psychiatric condition that they or a family member are diagnosed with more often than reported by participants in this study, but that HCPs may not receive such questions due to time constraints during consultations, as alluded to in quotations by P1 and P9. Here participants explained that sometimes they only have 10-15 minutes to see a patient in the clinic, or that they have to make time to talk to the patient even if they do not really have time available. It is also possible that because participants in this study work in an acute psychiatric setting at a tertiary hospital, questions about inheritance may not be the main concern of patients and family members when they are acutely ill, as P3 explains that questions arise from patients when they are "better". Another aspect could be that patients may have many uncertainties regarding the inheritance risks and causes of mental illnesses, but may not be addressing these worries with their managing doctors, as exemplified by P8 who explained having patients who were too scared to ask the doctors questions.

Perceptions of PGC

Knowledge of PGC was also encompassed in theme two and participants from this study showed a good understanding of the profession of GC. While many participants were uncomfortable explaining what they believed a GC does, most recognised the involvement of other family members with a GC's role, and many associated GC with genetic testing. Participants in this study were unclear what GC would look like in the psychiatry space, and what PGC would entail. So while PGC is formally understood as the process of assisting patients and families to understand their mental illness as well as the multifactorial causes that contribute to the development, as discussed in more detail in chapter one, P2 mainly saw PGC to be about explaining risks and predisposition to mental illness. Although positive outcomes of PGC do not rely on genetic testing (Inglis et al., 2015), advances in pharmacogenetic testing and direct-to-consumer (DTC) testing may open the door for GC involvement in psychiatric care, where patients have access to such testing. Thompson, Hamilton and Hippman (2015) found that most psychiatrists surveyed from a psychiatric hospital and other community practices in the USA thought genetic data (through pharmacogenetic testing or DTC testing) would be beneficial to them and their patients and that it would be beneficial to have a GC discuss these test results with them and the patient. Although such advanced genetic testing is not frequently encountered in the public healthcare system currently, access to these genetic tests may improve in future and could see GCs working more closely with psychiatric HCPs.

Perceived preparedness to answer inheritance or genetic risk questions differed amongst participants in this study, as exemplified in subtheme 2.2. All participants had basic theoretical genetic training and most had psychiatric specific theoretical training. This supports other studies such as Hoop et al. (2008) and Jenkins and Arribas-Ayllon (2016) that showed that most psychiatrists received some level of genetic training during medical training. Additionally, it seemed that the senior HCPs who participated in this study, had more experience working as consultant psychiatrists, and felt more equipped and comfortable talking about genetic risks to patients than junior doctors with less work experience. Most participants indicated that they would feel more comfortable addressing these aspects with their patients if they had more knowledge on this subject or learned from observing GCs or other experienced individuals counsel patients on genetic risk. This is in contrast with the study by Zhou et al. (2014) which found that most psychiatrists felt competent to discuss genetic information with patients and families, and this rating of perceived competency was not significantly associated with time since most recent genetic training. HCPs in this study indicated that they do not regularly address aspects such as inheritance and genetic risk of mental illnesses with their patients. Again, the lack of experience in conversations around genetic risk in our study population could be due to the context of working in the acute psychiatric setting. In addition, high caseloads may also hinder HCPs addressing these aspects with their patients due to time constraints.

Notably, P4 mentioned that in addition to not discussing genetic risks for psychiatric conditions with their patients, they had never considered the ethics of disclosing recurrence risks of psychiatric conditions. Indeed, certain theoretical ethical issues exist that would need to be taken into consideration when discussing inheritance and genetic risk to patients (Bennett, Thirlaway & Murray, 2008; Gershon & Alliey-Rodriguez, 2013). Some of these include, preimplantation selection of embryos based on a risk for mental illness (if PRS become clinically valid), termination of pregnancy based on chance of developing a psychiatric condition, and increased harm resulting from stigma from families and the community due to the increased genetic attribution to illness (Bennett, Thirlaway & Murray, 2008; Gershon & Alliey-Rodriguez, 2013).

An interesting observation from the study was that participants differed on their outlook on which psychiatric conditions would benefit most from PGC, these were found in theme two. Some (such as P4) felt that intellectual disabilities and geriatric psychiatry would benefit most. Indeed, GCs are frequently involved in counselling families of patients diagnosed with neurodevelopmental disorders (NDDs) (Blesson & Cohen, 2020). Conversely, other participants, like P2, felt that “softer” mental illnesses (such as anxiety or depressive disorders) would benefit most. Interestingly, a study by Kalb et al. (2017) found that GC was perceived to be useful for adults with a personal or family history of alcohol addiction. There was a significant correlation between the likelihood that GC would be perceived as beneficial and participants who believed that genetic factors played a significant role in alcoholism ($\rho=.21$, df=111, $p=0.024$), as well as between unaffected individuals who were worried about alcohol addiction in themselves and perceived benefits of GC ($t=2.37$, $df=66$, $p=0.021$) (Kalb et al., 2017). Most participants attributed genetic or inherited factors to play an important role in the causes of addictions, and recurrence risks to children and siblings were perceived as 46% and 27%, respectively (Kalb et al., 2017). Although participants from the Kalb et al. (2017) study did not undergo PGC, and were only provided with a definition of PGC to estimate whether it would be useful in their scenario of a personal or family history of alcohol addiction, this was an interesting study showcasing the potential impact of PGC on addiction rather than on other mental illnesses discussed in this dissertation such as schizophrenia, anxiety disorders or bipolar disorder, that PGC is often associated with.

Impact of PGC

The current study found that many participants have a positive outlook on the potential benefits that PGC could have in assisting them, and many felt an additional counselling service like PGC would be useful to their patients, as was discussed in more detail in theme three. For example, HCPs mentioned that it may aid with saving time by being able to outsource conversations that will take longer to the most appropriate person with the necessary skills and knowledge. This is seen in P4's quotation, stating that HCPs are all experts in their own field, reflecting the need for the most appropriate people to be involved in PGC conversations. Previous studies also found that psychiatric HCPs have a positive outlook on incorporating GC into their practice, reporting that these individuals believed such a service would be useful and desirable (Aschauer, Yazdi & Aschauer, 2024; Jenkins & Arribas-Ayllon, 2016). PGC is already being offered to patients and families with psychiatric disorders or NDDs across Europe (Koido et al., 2023). However, this article reported that there is a general lack of guidelines and education on genetic testing (such as pharmacogenetic testing, Sanger sequencing and exome/genome sequencing) and counselling for psychiatric conditions (including NDDs, neurological and major psychiatric disorders) in European countries (Koido et al., 2023). The authors recommended that the development of guidelines for GC, genetic testing, and training may improve access to and the quality of these services (Koido et al., 2023). Development of evidence-based guidelines, may further help with implementation of new PGC services in countries where they are not available yet, such as SA.

Perceived benefits of a PGC service from this study, as reflected in theme 3, included improved health outcomes due to increased understanding and knowledge, treatment adherence and decreases in relapses. However, P4 explained that they felt strongly that the cause and inheritance risks of a particular mental illness does not matter to them as the treating doctor in the acute setting, due to this information having no impact on acute care of a psychotic (for example) patient. When the acute phase has passed, many participants felt that this would be an ideal time to intervene with PGC. Some suggested having a mobile PGC clinic, implementing PGC as an outpatient service or a consultation liaison type service to counsel patients and families while they are in hospital, before being discharged, to help with understanding the reasons that lead to the patient to be admitted or becoming mentally ill. Having the service brought to patients may also assist with the travel and financial burden some patients face when attending hospital appointments. It seems that early intervention with PGC is associated with larger gains in empowerment scores (measured by the GCOS) (Saxton et al., 2022). Furthermore, other studied beneficial outcomes of PGC have been reported such as an increased empowerment and self-efficacy as well as increased knowledge and accuracy of risk perceptions (Hippman et al., 2016; Inglis et al., 2015). Relapse risk prevention as a result of having better knowledge and understanding of the psychiatric condition was anticipated as a benefit of PGC in this study. Participants felt that improved understanding will improve health outcomes, such as increased adherence to treatment and decreased relapses. Few studies have explored whether PGC translates into clinical benefits. Morris et al. (2024) explored the relationship between PGC and psychiatric hospitalisations and found that PGC may lead to fewer psychiatric hospitalisations and shorter hospital stays (1085 to 669 days) pre-GC vs post-GC. However, these findings were not statistically significant and served as a preliminary study using population based data that may contain many uncontrollable confounders (Morris et al., 2024). Another study investigated whether PGC would reduce psychiatric symptoms due to increased medication adherence (Morris et al., 2021). This study found among patients who had the most psychiatric symptoms prior to PGC, there was a significant decrease in psychotic symptoms post PGC, which could not be explained by increased adherence to treatment (Morris et al., 2021). Although this is the first study to investigate this correlation, there were several limitations such as a small sample size, possible ascertainment bias, restriction to in an outpatient setting and to psychotic symptoms (Morris et al., 2021). More research is needed to quantify the effect of PGC, particularly in low- or middle-income countries. The current research study also found that some participants felt that an increased knowledge and understanding of psychiatric conditions can be shared in the community and would lead to better health-seeking behaviours, better knowledge of at home management, and awareness of where and when to seek help.

Perceived barriers to implementation

Participants felt that aspects discussed in PGC, such as inheritance risk, causes of psychiatric conditions and risks to other family members are important and should be addressed with patients. Also, participants felt that a PGC service would be useful and that they would make use of such a service if available. However, it was clear from the interviews conducted that there are several perceived barriers to implementing a new PGC service, and the "ideal world' vs what is practically available and possible in a South African setting is reflected in theme four. Several practical barriers such as institutional finances, staff availability, patient finances and travel difficulties to attend clinics were frequently discussed. Consistent with other research, the complexities and uncertainties of genetics in psychiatry (that was also clear in theme one), concerns for stigmatisation, limited capacity for autonomy of patients, staff availability, financial barriers, and lack of genetic knowledge of patients and clinicians (consistent with theme two) have also been reported as barriers to implementation of a PGC service (Jenkins & Arribas-Ayllon, 2016; Koido et al., 2023).

Future outlook

More research is needed in future to assess how a new PGC service would be implemented in order to be most beneficial to patients and staff, and to overcome the perceived barriers to implementation mentioned above. Institutional prioritisation of funding is a major driver of the creation of new GC positions (Chanouha et al., 2023). The prioritisation of institutional funds is mainly driven by HCP referral patterns which, in turn, is influenced by the availability of genetic testing (Chanouha et al., 2023). This is problematic and will make the motivation for establishing new GC positions and a PGC clinic challenging, as there is no available genetic testing for common mental illnesses that can drive HCP referrals. Future research should focus on how PGC will generate revenue and improve HCP efficiency, to motivate for new GC positions, as only evidence for patient benefit from a service does not seem to play a large role in creation of new GC positions (Chanouha et al., 2023). Additionally, P7 discussed how they already manage their patients under relative pressure, alluding to staff availability, not only limited psychiatric HCPs but also limited numbers of GCs, as a barrier to providing PGC. With only 10 GCs currently working in the state healthcare system in SA (Gomes et al., 2024), motivating for a new clinic may prove to be challenging. It is likely that the scarcity of human resources from the GC profession in SA will play a larger role in the motivation of the implementation of a new PGC service than the availability of genetic testing. Additionally, there is already an increasing unmet need for genetic specialists in SA (Gomes et al., 2024), thus the idea of establishing new services with no one to provide these services may also be an ethical issue in itself. In this case, some suggestions like the development of information pamphlets, or website resources were made to aid in addressing time and resource barriers to implementation of PGC. Written or electronic resources that could be distributed in clinics and hospitals could thus be useful in bridging this gap. Indeed, previous work has shown that educational booklets increased knowledge in patients with serious mental illnesses, compared to people who were put on a waitlist (did not receive an educational booklet) (Hippman et al., 2016).

4.2. Conclusion

In conclusion, to our knowledge, this study is the first in SA to explore opinions of psychiatric HCPs on providing a PGC service, investigating the current landscape of 'mental illness counselling' that is currently being implemented in psychiatric services at a tertiary institution in Cape Town.

This research study found that HCPs believed that patients and families frequently ask about the diagnosis of psychiatric conditions, and although risks to other family members may not often come up, patients wonder about the causes of their mental illness. Psychiatric HCPs do not frequently use their genetic training in practice, and do not routinely address causes or genetic risks with patients. Some HCPs felt more comfortable than others in addressing aspects such as genetic risk, which seemed to largely depend on the level of experience of the HCP. HCPs in this study felt that more knowledge and information or observing someone experienced in addressing genetic risks would increase their confidence in discussing it with patients. Participants from this study had a good understanding of the profession of GC, but were unsure how GC would be applied in the psychiatric space. However, upon explaining aspects that PGC would address such as causal explanations and inheritance risks, most participants felt that such a service would be useful and beneficial to patients and families. They mentioned some perceived benefits such as increased knowledge and understanding for patients and families and decreased stigmatisation of the psychiatric disorder, which may have an impact on improved treatment adherence and decreased relapses. However, there were many perceived barriers to the implementation of a new PGC service, such as limited time, resources, and financial burdens of the institution and patients. These barriers seem to make PGC unfeasible at this time and would require creative solutions to overcome should a service be implemented in future after more research.

This research is an important first step to understanding the current workings of the psychiatry services in the public health sector, which will help guide future research on PGC in SA and potential implementation of a PGC service.

4.3. Strengths

This was the first study to explore the opinions of HCPs on PGC in SA. The qualitative nature and QD design of the study allowed the researcher to answer the research question and address the aims and objectives of the research. This was the first study of the current landscape of 'informal PGC' happening in public hospitals in Cape Town, SA, which may hopefully guide future work in this area. This research study used open-ended questions in semi-structured interviews as a data collection method, which allowed for new ideas and concepts to emerge during the interview process. This also allowed for new ideas to be incorporated in future interviews. The researcher built rapport with participants by having a short introductory conversation before the interview began. The researcher also spent a fair amount of time asking routine questions to understand their work and schedule, to assist in rapport building. The interviews were conducted in-person or telephonically, at a time and place that was most convenient to participants and did not take longer than 75 minutes. This helped with recruiting participants to the study, as to limit the disruption to their normal routine as much as possible.

4.4. Limitations

This research has several limitations. There was a very limited study population of psychiatric HCPs included in this study, limited to a specific institution, due to only receiving ethical approval from a select few institutions, and responses from participants from only one institution. This resulted in a small sample size with participants only from one institution, although participation from more institutions were sought out. This sample is thus not representative of the entire public health services in Cape Town, as was originally sought out, and opinions and experiences of HCPs may differ if there was inclusion of staff at district or community level healthcare facilities. Due to time pressure of this research study being conducted as a minor dissertation, it is possible that more in-depth themes and discussions could have emerged with more time and expertise. Additionally, this study was a minor dissertation in partial fulfilment of the degree MMedSc Genetic Counselling, with limited time and resources. The researcher had not conducted qualitative research before, and thus is a novice qualitative researcher. However, attempts to circumvent this were to consult and include the research supervisors throughout the research process. Additionally, transcripts were also not returned to participants for accuracy checking and feedback, although transcripts were transcribed verbatim to ensure the interviews were accurately reported.

This study explored the views of psychiatric HCPs and their experiences working with patients and families who have a psychiatric condition. Although participants reported that from their experience most patients and families believe psychiatric conditions are caused by environmental or spiritual causes, it would be important in future to study patient causal beliefs by exploring their own views, as this is not truly representative of actual patients' causal beliefs. Furthermore, it will be important in future to explore mental health care users' questions and opinions about their own psychiatric condition, as the experience of their treating HCP (such as in this study) may not be an accurate representation of their questions and uncertainties.

4.5. Researcher reflection

In qualitative research, the positionality of the researcher is key, as this can affect all research aspects such as study design, and data collection and analysis (Wilson, Janes & Williams, 2022). Positionality, as defined by Savin-Baden and Major (2013:71) “reflects the position that the researcher has chosen to adopt within a given research study”. It was important for the researcher to identify the impact of their positionality on the research and potential bias it may have brought. As a genetic counselling trainee, I have spent over two years working in genetic clinics, interacting with and counselling patients on a regular basis. I have seen the positive impact GC can have on patients and through my training, believe that I am helping patients, empowering them and assisting in their understanding of genetic conditions. I am thus biased towards perceiving GC as beneficial, as I have seen it first hand, and am also training to practice independently as a GC. This made me biased towards believing GC for psychiatric conditions may have the same beneficial effects. For the duration of my research, I had to critically evaluate whether my bias is being reflected in the results of this study. I frequently met with my research supervisor to discuss this, and kept record of all analysis to minimise bias. Interestingly, my thoughts on PGC changed during the duration of conducting this research.

Initially I anticipated PGC would be beneficial to patients, and was unsure why we could not just start seeing patients for psychiatric indications. Towards the end of this study, it became clear to me that there are many others factors at play and even if it is expected to be beneficial, many practical barriers and logistical nuances need to be ironed out to implement new counselling services. I also learnt that keeping an open mind for the future is important after realising that there are "less traditional GC" methods we could help patients with mental illness, such as creating information sources.

4.6. Research implications/recommendations

Results from this study may serve as a starting point for future research in PGC in SA. Some potential research opportunities may include the exploration of patient and family experiences, and their opinions of an additional PGC service. Participants' responses to their perceived competency in answering genetic risk questions varied, and it may be useful in future to explore further the perceived competency and role of genetic training on their confidence when communicating genetic risks to families. Additionally, outcomes of PGC in the South African context are needed to elucidate the impact of an additional PGC service. Finally, development of PGC resources (such as pamphlets, information sheets etc.) to distribute to the public or patients in clinics may be useful in bridging the gap in service provision to individuals who may benefit from this information.

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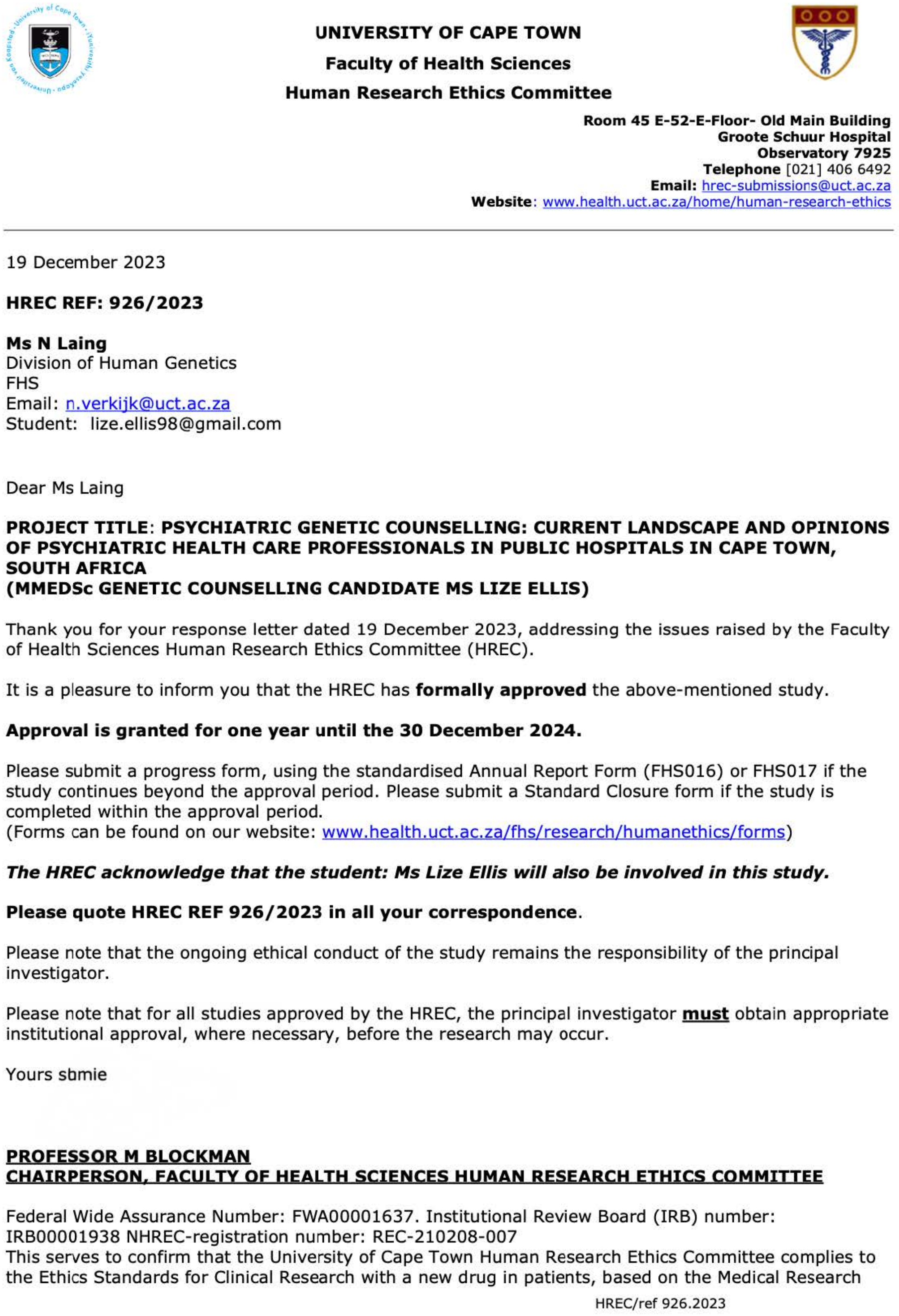
Appendix A



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

Room 45 E-52-E-Floor- Old Main Building
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19 December 2023

HREC REF: 926/2023

Ms N Laing
Division of Human Genetics
FHS
Email: n.verkijk@uct.ac.za
Student: lize.ellis98@gmail.com

Dear Ms Laing

PROJECT TITLE: **PSYCHIATRIC GENETIC COUNSELLING: CURRENT LANDSCAPE AND OPINIONS OF PSYCHIATRIC HEALTH CARE PROFESSIONALS IN PUBLIC HOSPITALS IN CAPE TOWN, SOUTH AFRICA**
(MMEDSc GENETIC COUNSELLING CANDIDATE MS LIZE ELLIS)

Thank you for your response letter dated 19 December 2023, addressing the issues raised by the Faculty of Health Sciences Human Research Ethics Committee (HREC).

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 December 2024.

Please submit a progress form, using the standardised Annual Report Form (FHS016) or FHS017 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 Lize Ellis will also be involved in this study.

Please quote HREC REF 926/2023 in all your correspondence.

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

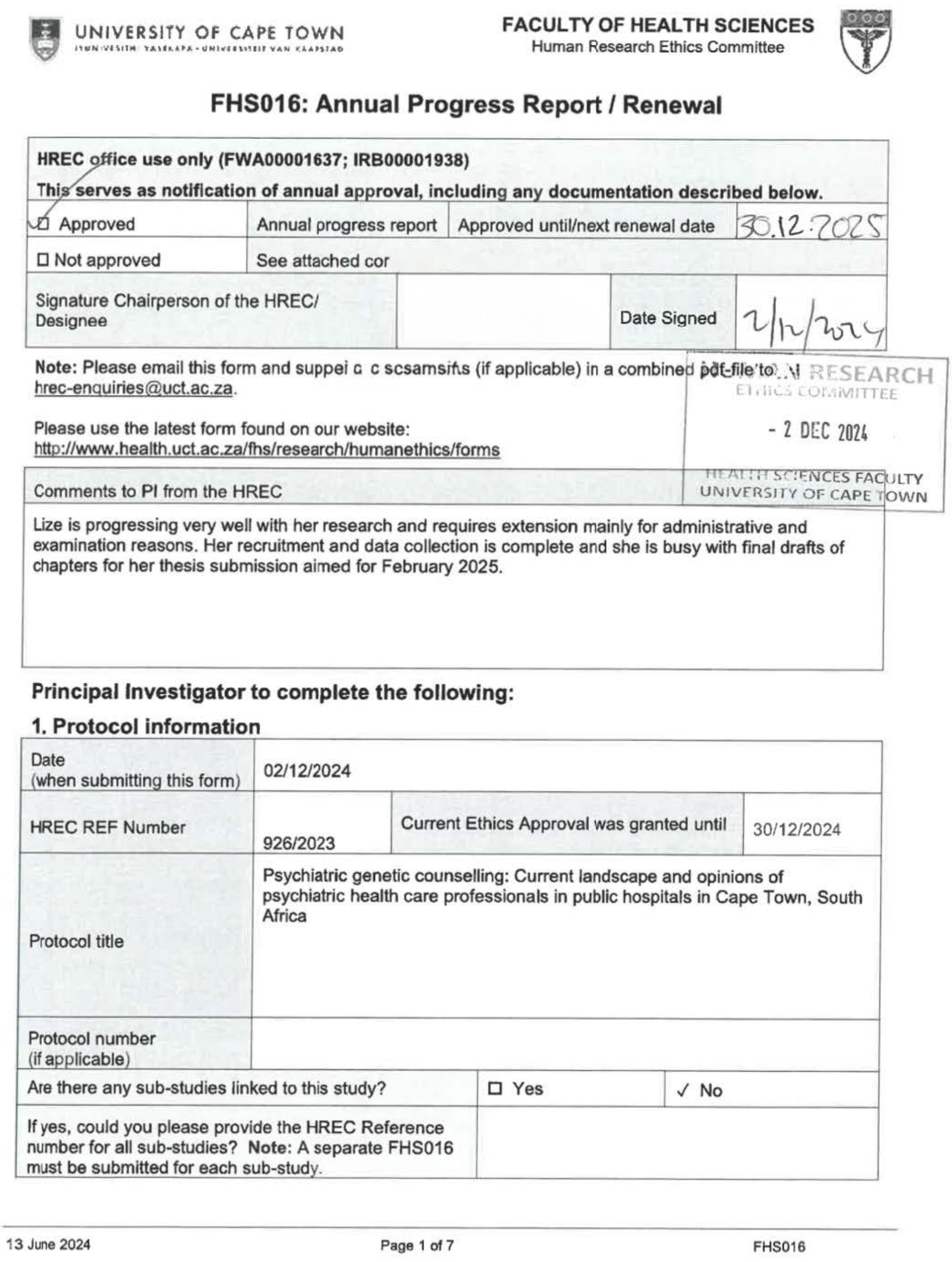
Please note that for all studies approved by the HREC, the principal investigator **must** obtain appropriate institutional approval, where necessary, before the research may occur.

Yours sbmie

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

Federal Wide Assurance Number: FWA00001637. Institutional Review Board (IRB) number: IRB00001938 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 2020), 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.



UNIVERSITY OF CAPE TOWN
IYUNIVESITHI YASEKAPA • UNIVERSITEIT VAN KAAPSTAD

FACULTY OF HEALTH SCIENCES
Human Research Ethics Committee

FHS016: Annual Progress Report / Renewal

HREC office use only (FWA00001637; IRB00001938) **This serves as notification of annual approval, including any documentation described below.**			
☑ Approved	Annual progress report	Approved until/next renewal date	30.12.2025
☐ Not approved	See attached cor		
Signature Chairperson of the HREC/ Designee		Date Signed	2/12/2024

Note: Please email this form and suppei c c scsamsifts (if applicable) in a combined pdf-file to: hrec-enquiries@uct.ac.za.

Please use the latest form found on our website: http://www.health.uct.ac.za/fhs/research/humanethics/forms

RESEARCH
ETHICS COMMITTEE
- 2 DEC 2024
HEALTH SCIENCES FACULTY
UNIVERSITY OF CAPE TOWN

Comments to PI from the HREC
Lize is progressing very well with her research and requires extension mainly for administrative and examination reasons. Her recruitment and data collection is complete and she is busy with final drafts of chapters for her thesis submission aimed for February 2025.

Principal Investigator to complete the following:

1. Protocol information

Date (when submitting this form)	02/12/2024		
HREC REF Number	926/2023	Current Ethics Approval was granted until	30/12/2024
Protocol title	Psychiatric genetic counselling: Current landscape and opinions of psychiatric health care professionals in public hospitals in Cape Town, South Africa		
Protocol number (if applicable)			
Are there any sub-studies linked to this study?	☐ Yes	✓ No	
If yes, could you please provide the HREC Reference number for all sub-studies? **Note:** A separate FHS016 must be submitted for each sub-study.			

13 June 2024 Page 1 of 7 FHS016

Appendix B

UNIVERSITY OF CAPE TOWN

GROOTE SCHUUR HOSPITAL

DIVISION OF HUMAN GENETICS
Faculty of Health Sciences
University of Cape Town
Observatory, 7925
South Africa
Telephone: +27 21 404 6235
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medicalgenetics.uct@gmail.com

Good day,

Thank you for taking the time to consider this urgent request.
I would like to introduce, Lize Ellis, a second year MMedSc Genetic Counselling student, who is conducting a research study on mental illness counselling and the current landscape thereof in the public sector in Cape Town.

This study aims to investigate the experiences of psychiatric health care professionals with their patients and their patients' views on the cause as well as recurrence risks of mental illness. The research also aims to identify barriers and potential benefits of providing a psychiatric genetic counselling service. **No** prior genetic experience, knowledge or training is needed to take part in the study.

If you are a psychiatric health care professionals, working at Groote Schuur Hospital, such as a psychiatric nurse, psychiatrist, registrar and medical officer, psychologist or psychology intern, we ask that you please consider participating in an interview about your experiences.

After pilot interviews, it seems like the interview will only take 30-40 minutes. The interviews can take place in person, or via Microsoft Teams, whatever is most convenient for participants in terms of place and time.

I have also attached the information sheet and consent form to participants for your convenience.

Participants can contact the researcher, Lize, via email if they are interested in participating (schliz010@myuct.ac.za), or WhatsApp (0718511708) for more information or to organise a time for an interview.

Kindest regards

Nakita Laing
n.verkijk@uct.ac.za
0214046235
0822146587

Appendix C

Information sheet

Dear participant,

You are invited to participate in a research study investigating the current landscape and opinions of genetic counselling services among psychiatric health care professionals in the public health sector in Cape Town, South Africa. This research study forms part of a minor dissertation in partial fulfilment of the degree MMedSc Genetic Counselling, at the University of Cape Town.

The study will investigate your experiences with psychiatric patients and their views on the causes and recurrence risks of their mental illness. The research aims to identify barriers and potential benefits of providing a psychiatric genetic counselling service, according to your experiences. Additionally, the study would hopefully contribute to informing the implementation and further research of such a service.

To gather information for this study, you are invited to an interview that will last approximately 30-60 minutes. The interview will be voice recorded, and recordings will be discarded after the dissertation is examined. Your confidentiality and privacy will be protected and no identifying information will be used in the study. You will be required to sign a consent form before the interview.

Participation is entirely voluntary. If you decide to participate, you may choose not to answer certain questions during the interview or choose to withdraw from the study at any time, with no penalty to you.

If you have any questions, please contact the researcher, Lize Ellis on 0718511708 via SMS or WhatsApp, or via email at schliz010@myuct.ac.za. Alternatively, you can contact the research project supervisor, Nakita Laing, at n.verkijk@uct.ac.za or the UCT ethics committee on 021 650 1236 or hrec-enquiries@uct.ac.za.

Kind regards

Lize Ellis

Appendix D

Interview Guide

Section A: Demographic questions

1. Sex
2. Age
3. Occupation
4. How many years have you been working in this role?
   a. What were you doing before/where did you work before?
5. Have you ever received any genetic training? If so, what kind of training?
   a. How long ago was this?
   b. Probe: What do you remember?
6. How many patients do you see per week (on average)? Ranges offered: 0-10; 10-20; 20-30; 30 or more.
7. Do you have or have you previously had any mental health condition?

Section B: Semi-structured interview guide

Daily/weekly overview

1. Can you describe an average day in your week?
2. What does a typical interaction with a patient look like?
3. What are some of the most common questions patients ask you?
   a. Probes: What questions do they ask regarding the causes
   b. & recurrence risks of their mental health condition?
4. What kind of mental health conditions do the patients have that you typically see?

Causes and recurrence of mental health conditions

5. Can you tell me what causes patients ascribe their mental health condition to?
   a. Probes: Are they mostly environmental, or genetic causes (inherited/family history)?
   b. What do you understand causes mental health conditions?
6. Can you tell me what patients ask with regards to their condition occurring again in the family?
7. Can you talk me through how you might describe and explain genetic risk to families?

    a. Sub-question: How do you feel about answering these questions?
    b. Do you feel equipped to answer these questions?

Genetic counselling

8. What do you understand from the term "genetic counselling"?
    a. Short explanation: Genetic counselling is where patients come and get explained about the causes of their condition and the chances of other people in their families having the same condition
9. Have you referred patients to genetic counselling before, where and why?
    a. (Have you ever referred a patient back to a doctor to talk about the causes of their condition?)
10. In your opinion, if there was a service available where patients and families could go to have counselling regarding the causes and recurrence risks of their mental health condition, do you see this service being used?
11. Why do you think so?
    a. How or where do you see such a service fitting in?
    b. Who should do the counselling/provide the service?
        i. Why do you say that?
        ii. How will this work?

Scenario

Imagine this scenario with me, we have a genetic counsellor operating a dedicated psychiatric genetic counselling clinic from Groote Schuur Hospital, where patients could be referred for counselling. At this clinic, patients are counselled to help them understand their condition better, help them understand the causes and potential risks to other family members.

12. How do you see this clinic working alongside what you do here?
13. How should referrals work to and from this psychiatric genetic counselling clinic?

Obstacles & benefits

14. Can you comment on some of the obstacles or problems that might come with having such a service in place?
    a. Practical obstacles vs emotional/psychosocial obstacles
15. What do you think will be the benefits of implementing a psychiatric genetic counselling service in the hospital?
    a. Patient benefit
    b. Service benefit

Appendix E

Informed Consent form

I, ____

____

agree to participate in this study.

I confirm that the researcher (Lize Ellis) explained the study to me in a manner that I understand what is required of me and what the study entails.

I understand the purpose of this research, as provided in the information sheet.

I understand that the objective of this study is to explore the current landscape of psychiatric genetic counselling services and opinions of psychiatric health care professionals who work in the public health sector within the Groote Schuur Hospital catchment area in Cape Town on providing a psychiatric genetic counselling service.

I understand that participation in this study will involve an interview asking my opinions about psychiatric genetic counselling services and my experiences with psychiatric patients and their views on the causes and recurrence risks of their mental illness.

I understand that the interview will take about 30-60 minutes of my time.

I understand that the interview will be conducted in English.

I confirm that all my questions about this research study has been answered satisfactorily.

I understand the purpose and aim of this research study as explained to me by the researcher and provided in the information sheet.

I understand that my participation in this study is entirely voluntary, with no financial benefit to me.

I understand that there is no direct benefit to me by participating in the research study.

I understand that I may withdraw from this study at any time, with no penalty to me.

I understand that during the interview, I may choose to refrain from answering specific questions with no consequence.

I understand that my interview will be audio-recorded, transcribed, and analysed by the researcher.

I understand that my identity and any other identifying information will be protected and that my identity will remain anonymous. I understand that my identifying information will be removed from the study data prior to analysis, and that I will be assigned a participant number to protect my identity.I understand that my audio-recordings and transcripts will be stored on the researcher’s password protected computer, to which only the researcher and the research supervisors will have access to the data.

I understand that sections of my interview may be quoted, without any identifying information, in the mini-dissertation, academic posters, presentations, or academic articles.

I understand that the study has been approved by the Human Research Ethics Committee at the Faculty of Health Sciences at the University of Cape Town. I have been given contact details of the committee should I have any questions or complaints about the research study.

The researcher has explained the information of this study to me, and I understand everything.

Signature of participant

____
(Signature)

____
(Date)

Signature of researcher

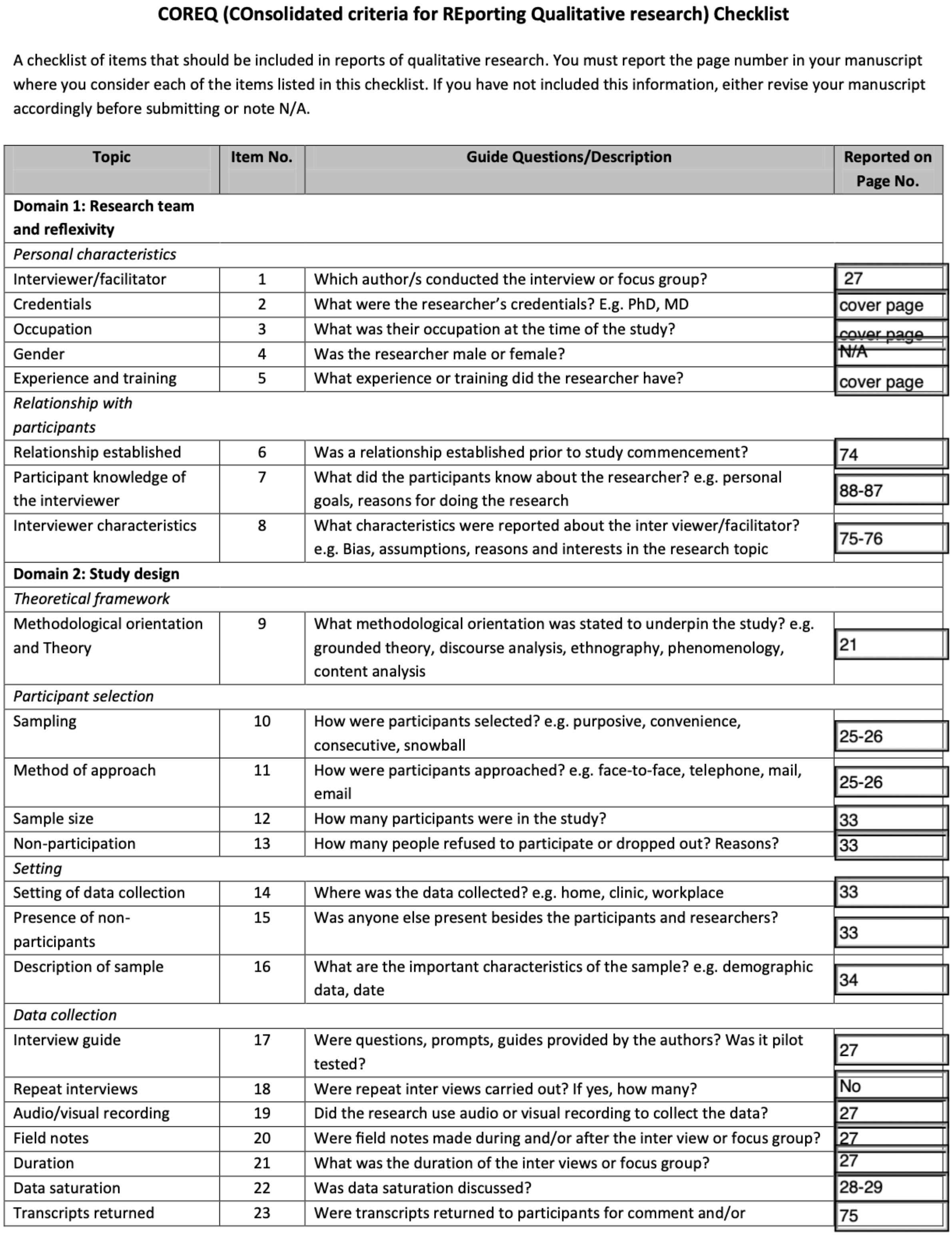
____
(Signature)

____
(Date)

The UCT's Faculty of Health Sciences Human Research Ethics Committee can be contacted on 021 406 6338 in case you have any ethical concerns or questions about your rights or welfare as a participant on this research study.

For any questions about the research project, please contact the researcher, Lize Ellis on 0718511708 via SMS or WhatsApp, or via email at schliz010@myuct.ac.za. Alternatively, you can contact the research project supervisor, Nakita Laing, at n.verkijk@uct.ac.za or the UCT ethics committee on 021 650 1236 or hrec-enquiries@uct.ac.za

Appendix F



COREQ (COnsolidated criteria for REporting Qualitative research) Checklist

A checklist of items that should be included in reports of qualitative research. You must report the page number in your manuscript where you consider each of the items listed in this checklist. If you have not included this information, either revise your manuscript accordingly before submitting or note N/A.

Topic	Item No.	Guide Questions/Description	Reported on Page No.
Domain 1: Research team and reflexivity			
Personal characteristics			
Interviewer/facilitator	1	Which author/s conducted the interview or focus group?	27
Credentials	2	What were the researcher's credentials? E.g. PhD, MD	cover page
Occupation	3	What was their occupation at the time of the study?	cover page
Gender	4	Was the researcher male or female?	N/A
Experience and training	5	What experience or training did the researcher have?	cover page
Relationship with participants			
Relationship established	6	Was a relationship established prior to study commencement?	74
Participant knowledge of the interviewer	7	What did the participants know about the researcher? e.g. personal goals, reasons for doing the research	88-87
Interviewer characteristics	8	What characteristics were reported about the inter viewer/facilitator? e.g. Bias, assumptions, reasons and interests in the research topic	75-76
Domain 2: Study design			
Theoretical framework			
Methodological orientation and Theory	9	What methodological orientation was stated to underpin the study? e.g. grounded theory, discourse analysis, ethnography, phenomenology, content analysis	21
Participant selection			
Sampling	10	How were participants selected? e.g. purposive, convenience, consecutive, snowball	25-26
Method of approach	11	How were participants approached? e.g. face-to-face, telephone, mail, email	25-26
Sample size	12	How many participants were in the study?	33
Non-participation	13	How many people refused to participate or dropped out? Reasons?	33
Setting			
Setting of data collection	14	Where was the data collected? e.g. home, clinic, workplace	33
Presence of non-participants	15	Was anyone else present besides the participants and researchers?	33
Description of sample	16	What are the important characteristics of the sample? e.g. demographic data, date	34
Data collection			
Interview guide	17	Were questions, prompts, guides provided by the authors? Was it pilot tested?	27
Repeat interviews	18	Were repeat inter views carried out? If yes, how many?	No
Audio/visual recording	19	Did the research use audio or visual recording to collect the data?	27
Field notes	20	Were field notes made during and/or after the inter view or focus group?	27
Duration	21	What was the duration of the inter views or focus group?	27
Data saturation	22	Was data saturation discussed?	28-29
Transcripts returned	23	Were transcripts returned to participants for comment and/or	75

Topic	Item No.	Guide Questions/Description	Reported on Page No.
		correction?	
Domain 3: analysis and findings			
Data analysis			
Number of data coders	24	How many data coders coded the data?	29
Description of the coding tree	25	Did authors provide a description of the coding tree?	28
Derivation of themes	26	Were themes identified in advance or derived from the data?	28
Software	27	What software, if applicable, was used to manage the data?	28
Participant checking	28	Did participants provide feedback on the findings?	75
Reporting			
Quotations presented	29	Were participant quotations presented to illustrate the themes/findings? Was each quotation identified? e.g. participant number	33-64
Data and findings consistent	30	Was there consistency between the data presented and the findings?	33-64
Clarity of major themes	31	Were major themes clearly presented in the findings?	35
Clarity of minor themes	32	Is there a description of diverse cases or discussion of minor themes?	35-64

Developed from: Tong A, Sainsbury P, Craig J. Consolidated criteria for reporting qualitative research (COREQ): a 32-item checklist for interviews and focus groups. *International Journal for Quality in Health Care*. 2007. Volume 19, Number 6: pp. 349 – 357

Once you have completed this checklist, please save a copy and upload it as part of your submission. DO NOT include this checklist as part of the main manuscript document. It must be uploaded as a separate file.