MOTHERS' EXPERIENCES OF GENETIC COUNSELLING IN JOHANNESBURG, SOUTH AFRICA

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A research report submitted to the Faculty of Health Sciences, University of the Witwatersrand, Johannesburg, in partial fulfillment of the requirements for the degree of Master of Science in Medicine in Genetic Counselling

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DECLARATION

I, Megan Morris, declare that this research report is my own work. It is being submitted for the degree of Master of Medicine in Genetic Counselling in the University of the Witwatersrand, Johannesburg. It has not been submitted before for any degree or examination at this or any other University.

Signed:……………………
Megan Morris
Date: 9 April 2013

DEDICATION

I would like to dedicate this research to my father, Stuart Morris, and to my mother, Dalene Woolley. I love you.

ABSTRACT

Genetic counselling is an expanding profession, with many services now being offered in multicultural settings. The way in which individuals respond to genetic disorders varies greatly between countries, socio-economic groups, families, communities, religions and cultural groups. Together, these perspectives may influence how people experience genetic counselling with regard to satisfaction of the service, understanding heritability, communication, support and general healthcare provision. To address these issues standardised healthcare and genetic counselling models may need to be adjusted to prevent disparities in healthcare communication for different communities across the globe. South Africa provides a unique setting for genetic counselling because of the rich multicultural and linguistic diversity, as well as the many health and socioeconomic challenges that the country faces. Due to these diversities and challenges, further exploration into individuals' experiences of genetic counselling in South Africa is required to gain insight into the service needs for individuals.

The aim of this study was to describe and document the experience of mothers who had received genetic counselling at state hospitals in Johannesburg, South Africa, after having a child diagnosed with a genetic condition. The research design was qualitative in which thirteen women, who had received counselling between January 2010 and January 2012, agreed to participate. Four voice-recorded focus groups were conducted in a suitable African language with the use of a question-guide. All recorded data were transcribed and translated into English and Interpretative Phenomenological Analysis (IPA) was used for the analysis and development of themes.

Six main themes, together with relevant sub-themes were identified. These included a general lack of awareness of genetic counselling and genetic conditions; both positive and negative experiences of genetic counselling including a need for greater support; personal beliefs regarding genetic conditions; challenges in addressing family and community members; dissatisfaction with the healthcare system and a need for awareness campaigns. Although findings from this study cannot be generalised to all patients or all South African settings, valuable insight has been gained that may contribute towards improving genetic counselling services in Johannesburg, South Africa.

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LIST OF ABBREVIATIONS

AIDS - Autoimmune Deficiency Syndrome

BScHons - Bachelor of Science Honours

CHB - Chris Hani Baragwanath Hospital

CMJAH - Charlotte Maxeke Johannesburg Academic Hospital

CSPE - Centre for Support and Peer Education

DGMC - Donald Gordon Medical Centre

HIV - Human Immune Virus

IPA – Interpretative Phenomenological Analysis

NHLS - National Health Laboratory Services

NSGC - National Society of Genetic Counselors

OPD - Out Patient Department

RMH - Rahima Moosa Mother and Child Hospital

TCA - Thematic Content Analysis

UNISA - University of South Africa

Wits - University of the Witwatersrand

1 CHAPTER 1: INTRODUCTION

1.1 Literature Review

1.1.1 Introduction

In this literature review areas of client satisfaction, cultural attitudes and beliefs around genetic inheritance as well as cultural competency in healthcare, genetic services and counselling will be addressed. In addition, current genetic services provided in South Africa and the challenges that have arisen regarding the country's cultural, linguistic and socioeconomic diversity will be considered. This literature review will provide insight into some of the issues that have arisen from previous research on individuals' experiences of genetic counselling and healthcare services in multicultural settings. Topics discussed, will serve as a background introduction for this study, which aims to describe the experiences of state patients who have received genetic counselling in Johannesburg, South Africa after having a child diagnosed with a genetic condition.

1.1.2 Client Satisfaction Following Genetic Counselling

According to the National Society of Genetic Counselors (NSGC), genetic counselling can be defined as a process which assists individuals to understand and adapt to the medical, familial and emotional implications of genetic contribution to disease (Resta, Biesecker and Bennet. 2006). The purpose of genetic counselling is to convey relevant genetic information and reproductive options in a non-directive and supportive manner to families who are, or may be at risk to be affected by a genetic disorder (Abramovsky, Godmilow, Hirschorn *et al*., 1980.). To successfully achieve the purpose, the client must fully understand the diagnosis, testing options, the prognosis of the disorder, available resources and management options, as well as the recurrence risks, to allow informed decision making while offering emotional support and addressing any psychosocial concerns (Abramovsky *et al*., 1980; Harper. 2004; Walker. 2009).

Client satisfaction assessments in healthcare have assisted professionals to gain insight into desirable outcomes for patient care by reviewing the level of healthcare and identifying problems that need to be resolved (Aalfs, Oort, de Haes *et al*., 2007). Client satisfaction in genetic counselling is thought to cover three main areas: 1) satisfaction with the competence of the genetic counsellor in terms of how clearly the information is conveyed, how they listen to the client, address their concerns and decrease their level of anxiety; 2) the cost and availability of the service and 3) satisfaction with the personal qualities of the counsellor and their behavior towards the client (Shiloh, Avdor and Goodman. 1990). Although it has been considered important to receive feedback from users of genetic counselling services to evaluate satisfaction levels and identify areas where improvement is needed, there has been limited research in this area (Bleiker, Aaronson, Menko *et al*., 1997; Aalfs *et al*., 2007). The majority of genetic counselling studies have focused on areas such as hereditary cancers, counselling for prenatal screening, outcomes of reproductive decision making and recall of information (Collins, Halliday, Kahler *et al*., 2001; Aalfs *et al*., 2007). In addition to a lack of "client satisfaction" assessments in genetic counselling, there has also been little research on how parents who have a child with a congenital defect perceive the value of genetic counselling, or what aspects of genetic counselling clients found the most or least helpful (McCarthy Veach, Truesdell, LeRoy *et al*., 1999).

Of the few studies that have been done on client satisfaction in genetic counselling, a positive client experience was shown (Bleiker *et al*., 1997; Stadler and Mulvihill. 1998; Etchegary, Cappelli, Potter *et al*., 2010). However, most of these were done in areas of cancer or prenatal genetic counselling in European or American first world settings with higher-educated Caucasian individuals (Bleiker *et al*., 1997; Stadler and Mulvihill. 1998; Etchegary *et al*., 2010). African-American patients are not well represented in these studies and little research has been done in developing countries, particularly sub-Saharan Africa (Charles, Kessler, Stopfer, *et al.*, 2006; Meilleur, Coulibaly, Traoré *et al*., 2011). One reason for this may be due to limited genetic services in developing countries (Christianson and Zimmern. 2009). In addition to the limited number of studies done, measuring the physical and social outcomes of clinical genetic services has proven difficult (Payne, Nicholls, McAllister *et al*., 2007).

Qualitative research methods may be useful to evaluate client satisfaction, since this approach focuses on understanding a particular phenomenon rather than trying to prove any preconceived notions (McAllister, Payne, Nicholls *et al*., 2007). This is demonstrated in a qualitative study by McAllister, Payne, Macleod *et al* (2008), which found that genetic counselling services fulfill the needs of the majority of patients seeking information and support by providing relief of uncertainty, aiding individuals to adjust to a genetic condition, and addressing feelings of vulnerability. However this study was done in a first world setting and more research is needed in developing countries.

1.1.3 Cultural Attitudes and Beliefs Surrounding Genetic Inheritance

Although genetics transcends national borders, politics and cultural differences, the way in which individuals respond to genetic conditions varies greatly within and between countries, socioeconomic groups, cultures and religions (Edwards. 2010). Heritability in clinical terms is understood as something that is purely genetic; however many people perceive this phenomenon as influenced by culture, community and family beliefs (Hurst, Lieber, Lewis *et al.*, 2011).

Culture is described as a group's way of life that is passed from one generation to the next. It can largely influence human behavior in thinking, living, acting and values (Vontress. 2011). Cultural beliefs about inheritance have included: religious views; phenotypic parent-child resemblances can influence the likelihood of passing down a disease mutation; males are of a higher rank in the family thus they pass down inherited traits; that personality and maternal behaviour can influence the outcome of disease or an unborn baby; bad luck and the influence of ancestry in terms of evil ancestors and ancestral transgressions (Emslie, Hunt and Watts. 2003; Eisenbruch, Yeo, Meiser, *et al.*, 2004; Barlow-Stewart, Yeo, Meiser *et al.*, 2006; Klitzman. 2010; Hurst *et al.*, 2011). Inquiry into counsellees' understanding of the etiology of specific conditions showed that although their beliefs may be influenced by the information given to them in a genetic counselling session, this gained knowledge may either be rejected, misconstrued or may be at variance to their personal beliefs (Weil. 1991). Some people are conflicted between different beliefs of inheritance because, although they accept and understand the scientific explanation of the causation of a genetic condition, they additionally consider notions of religious conviction, cultural assumptions and thoughts around being chosen or blessed, thus impacting on how they react to the birth of a child (Weil. 1991; Eisenbruch *et al*., 2004; Black, Girotto, Chapman *et al*., 2009). Counsellors disregarding or discrediting counsellees' beliefs and values can lead to difficulties in communication (Weil. 1991). Beliefs and values should be acknowledged within acceptable limits, since support from the counsellor may assist counsellees adapt to and cope with a genetic condition (Weil. 1991).

Education levels, family systems and community attitudes have also been found to be influencing factors on an individual's beliefs regarding genetic inheritance (Browner, Preloran, Casado *et al.*, 2003; Shaw and Hurst. 2008). For example, individuals with a graduate or postgraduate level of education may accept the scientific explanation for the cause of the genetic condition more readily than other individuals who may find beliefs in environmental or spiritual causes easier to understand than unfamiliar genetic explanations and medical jargon (Weil. 1991; Shaw and Hurst. 2008).

Effective communication amongst families regarding genetic information can influence familial health, particularly around decision making, preventative measures, screening and genetic testing (Kinney, Gammon, Coxworth *et al*., 2010). The term "family" can also have different meanings within different cultures and can describe biological and social relationships which may include nuclear family members (parents, children and siblings), kin (extended relatives), household (married relatives and non-related individuals) and friendships within a community (Richards. 1996; Peterson. 2005). Studies have shown that although affected individuals may disclose their condition to their immediate family, many find it a challenge to share this information with friends and individuals in the broader community since disclosure of the prognosis, severity, treatment and clinical symptoms of a condition can influence social reactions (Plantinga, Natowicz, Kass *et al*., 2003; Sankar, Cho, Wolpe *et al.*, 2006). Communication and disclosure of genetic information within a society can be difficult because of the fear of discrimination (Plantinga *et al*., 2003). Alternatively individuals within some societies may play an important role with regard to heritability; some families within Middle- Eastern and Asian communities make use of respected community elders to assist them in decision-making (Saleh, Barlow-Stewart, Meiser *et al*., 2009). Therefore disclosure of genetic information to these community members may be valuable.

It is thus important to gain a better understanding of cultural and psychosocial issues in a particular community so as to appreciate the complexity of individual's responses to genetic conditions and to be able to assess the value of clinical services within this context (Edwards. 2010; Kinney *et al*., 2010). Healthcare professionals must not only be sensitive to cultural differences but should also be aware of individuality and not make generalisations about people from similar cultural or religious backgrounds or who speak a common language. This has been highlighted in previous healthcare studies amongst ethnic minority groups in Australia, America, East Asia, Colombia and England which have shown that although two individuals may come from the same country, believe in the same religion or speak a common language, they do not necessarily share the same set of values and beliefs (Shaw and Hurst. 2008; Black *et al*., 2009; Saleh *et al*., 2009).

1.1.4 Cultural Competence in Healthcare, Genetic Services and Counselling

As the understanding of genetics has grown due to the sequencing of the human genome, genetics in clinical care has played an increasing role in causal explanations for conditions and heritability (Hurst *et al*., 2011). Historically, users of genetic counselling services were mostly upper- to middle-class Caucasians and the genetic counselling model has mainly evolved from these Western culture values and ideals (Lum and Whipperman. 1987; Fisher. 1996). In societies where there is strong cultural diversity, current genetic service provision may be incompatible with culturally determined attitudes towards genetics, testing and counselling (Barlow-Stewart *et al*., 2006). An individual's response and attitude regarding genetic services may be influenced by: their knowledge of genetic inheritance and disease; understanding of the given information; feelings such as fear, anxiety and shame; family influences and interactions; cultural taboos as well as social circumstances (Kinney *et al*., 2010). With an ever increasing number of non-Western users of genetic counselling services and interaction of individuals from different cultural backgrounds, there is potential for conflict and ethical dilemmas over "right" and "wrong" behavior. This emphasises the need for genetic counsellors to be culturally competent (Greeson, McCarthy Veach and LeRoy. 2001). To alleviate these issues suggestions have been made that cultural competency be introduced into health education programs and curricula (Shaya and Gbarayor. 2006).

The term "culture" includes several areas such as family beliefs, religion, acceptance of consanguinity, attitudes towards non-directive counselling, gender roles as well as patient autonomy and privacy (Barlow-Stewart *et al*., 2006; Shaw and Hurst. 2008; Saleh *et al*., 2009; White. 2009; Yanni, Copeland, and Olney. 2010; Lakhani. 2012). Family beliefs are important when communicating with individuals since decisions about health in some cultures are often made by the whole family and consequently, influence decisions made by the individual who presents for counselling (Eisenbruch *et al*., 2004; Penn, Watermeyer, MacDonald *et al*., 2010; Hurst *et al*., 2011). With regard to religion, genetic testing may raise complex religious and moral issues especially when having to make difficult choices and deal with risks and probabilities. Conflict may also arise when the counsellor and counsellee have different religious beliefs (White. 2009). To address these challenges in health communication, religious attitudes should be explored during a consultation so the health professional can understand better how a patient copes with their genetic risk with regard to their religious faith (White. 2009). This will not only build stronger relationships and improve the quality of care, but can assist in establishing trust and non-judgmental attitudes between the counsellees and health care providers, where counsellees can know that their beliefs are not disregarded (Barlow-Stewart *et al*., 2006; Hurst *et al*., 2011).

Consanguinity is another cultural practice that healthcare providers should be aware of since it is common in certain populations in the Middle-East, South Asia, China, North Africa and some parts of Southern Europe (Barlow-Stewart *et al*., 2006; Shaw and Hurst. 2008). Although in the Chinese culture it is considered “improper” to marry paternal relatives as they are thought to be from the same blood line, marriages to maternal relatives are acceptable as they do not share the same surname (Barlow-Stewart *et al*., 2006). Some communities that practice consanguinity may not readily accept scientific explanations of increased risk of genetic conditions to their offspring, since it is an accepted cultural, social or religious custom

(Shaw and Hurst. 2008). Thus there is a need for culturally sensitive genetic counselling amongst these groups (Yanni *et al.*, 2010).

Non-directive counselling is the central ethos of genetic counselling. However, some researchers have debated whether this is the best form of counselling, especially in multicultural settings. It has been suggested that although non-directive counselling is suitable in Western settings, it may be inappropriate in Non-Western contexts since most individuals are familiar with direct counselling styles (Raz and Atar. 2003). Some individuals may feel that the counsellor is concealing information or may interpret a counsellor's reserved opinion as an unspoken way of approving of their decision, which may lead to confusion (Bernhardt. 1997; Michie, Bron, Bobrow *et al*., 1997; Ellington, Roter, William *et al*., 2005).

Gender roles, patient autonomy and privacy may also create challenges in healthcare communication. In some cultures, patient autonomy may be incompatible with certain cultural views and individuals may seek assistance with decision-making from respected community elders or family members (Saleh *et al*., 2009). Male authority presides in some communities and women consult with their male partner or other male family members for decision-making regarding their own health and pregnancy (Barlow-Stewart *et al*., 2006; Saleh *et al*., 2009). Alternatively, disclosure can also pose a difficulty since some populations feel it "improper" to talk about one's own medical condition, even to family and friends, making discussions about genetic risk challenging (Kinney *et al*., 2010). Due to this, problems may arise when health care professionals make use of interpreters as some cultures do not believe in disclosing personal information to strangers to protect the family reputation (Saleh *et al*., 2009).

Linguistic diversity also needs consideration when dealing with individuals. Previous studies have shown that patients with a poor understanding of a particular language obtain less information, have a lower recall of medical facts conveyed to them and so receive less adequate healthcare (De Maesschalk, Deveugele and Willems. 2011). Patients from varying cultures have reported difficulty in expressing emotions, particularly negative emotions, and healthcare professions have also found it hard to focus on non-verbal cues when speaking to individuals with different mother-tongues (De Maesschalk *et al*., 2011; Meilleur *et al*., 2011). Although translators may be helpful when a common language is not shared by the counsellor and counsellee, the translation of complex medical information, decisions around informed consent as well as changing dynamics between the counsellor, counsellee and interpreter may lead to conflict (Browner *et al*., 2003; Saleh *et al*., 2009). However, the benefit of using an interpreter may outweigh the disadvantages since greater misunderstandings between the healthcare provider and patient may occur without the presence of an interpreter (De Maesschalk *et al*., 2011). Thus recommendations have been made for the provision of interpreter and translation services in healthcare (Saleh *et al*., 2009).

Education levels may also impact on how health information is received. Individuals from some developing countries may have little knowledge about services such as genetic counselling, and as health educational material is often aimed at literate individuals, this may lead to exclusion and misunderstanding for those with lower literacy levels (Kinney *et al*., 2010; Wasserman, Maja and Wright. 2010). Studies have found that an individual’s education level may influence their decision to accept or decline genetic counselling. Those with a higher education might be more aware of the genetic basis of a health condition than those of a lower-education level, thus the latter may not see the relevance of genetics or may feel daunted by the complexity of information provided at a genetic counselling session (Culver, Burke, Yasui *et al*., 2001). To overcome these barriers, genetic counselling practices should allow for the translation of medical and scientific knowledge into an understandable and useful format for all individuals. Suggested methods to achieve this include educational community outreach and the creation of appropriate education materials (Weil. 2001). Educational tools such as audio, visual, printed and mixed media methods may be useful as well as adaptation of health information to suit the needs of individuals rather than using generalised health methods (Kreuter, Green, Capella *et al*. 2007). When designing and implementing genetic education strategies and clinical interventions, it is important to address socio-cultural factors by asking important members of the community to relay health messages and explain educational materials. This may aid in correcting myths and misunderstandings about genetic knowledge as well as promote family and societal communication. Health education should ideally be given in the vernacular and studies have indicated that for services to be successful, the information should be personally relevant for it to be cognitively processed (Kinney *et al*., 2010).

Access to healthcare, including genetic services, is also challenging in a multicultural setting (Culver *et al*., 2001). Barriers that impede access to genetic services may include transportation difficulties, poor levels of referral from other health care professionals, language diversity, the costs involved, or lack of access to healthcare facilities for individuals living in rural areas (Culver *et al*., 2001). These problems together with the other challenges faced in multicultural and linguistically diverse settings need to be addressed in order to provide effective, culturally competent healthcare services to all people.

1.1.5 Genetic Counselling and Healthcare Provision in South Africa

South Africa provides a unique setting for genetic counselling and healthcare provision because of the multicultural and linguistic diversity, socio-economic differences as well as the country's apartheid history (Penn *et al.*, 2010; Solomon, Greenberg, Futter *et al.*, 2012). There are eleven official languages spoken and several practiced religions including Christianity, sectarian Christian Independent churches (e.g. Zionists), Islamism, Hinduism and Judaism (Statistics South Africa. 2001; Ross. 2007). The population is estimated at 50.59 million people, made up of 79.5% Black (with isiZulu/isiXhosa and seSotho/seTswana making up the largest ethnic groups), 9.0% Caucasians and the remainder of the population is made up of Mixed Ancestry, Indian and Asian individuals (Todd, Haw, Kromberg *et al*., 2010; Statistics South Africa. 2011). An estimated 26.2% of the population has a Grade 12 education level and there are an estimated 7.0% of individuals who have no formal education (Statistics South Africa. 2010). There are also major issues with regard to poverty, unemployment, and access to adequate healthcare, housing, water, electricity and sanitation (Coovadia, Jewkes, Barron *et al*., 2009).

Cultural attitudes, including the belief that a son only inherits from his father and a daughter only from her mother, that ancestors can influence the lives of the living and various religious beliefs all play a role in how inheritance is understood (Schapira and Goodwin. 1937; Kromberg and Jenkins. 1988; Kibuka-Sebitosi. 2007). The importance of family and community also play a large role in South African cultures and ideas of kinship may be contrary to Western understandings of inheritance (Meiser, Irle, Lobb *et al*., 2008, Penn *et al*., 2010). Decisions are often made on a family and community level, with community elders being greatly respected (Kasanga and Lwanga-Lumu. 2007). Additionally, the traditional healing system is widely accessible in South Africa, with many people making use of both the Western and traditional healer systems (Penn *et al*., 2010).

It has been demonstrated in some international studies that first world practices of genetic counselling and genetic services may not be suitable to middle- and low-income countries because of divergences in genetic literacy, language barriers and social customs (Christianson, Howson and Modell. 2006; Penn *et al*., 2010). These findings raise concern over the suitability of current genetic services offered to South African populations, since the genetic counselling model implemented is largely taken from British traditions and first world practices (Jenkins. 1990; Penn *et al*., 2010; Solomon *et al*., 20l2).

Genetic counselling services in South Africa were first established during the 1950s, however most services prior to 1994 were only available to the urban, “educated” Caucasian population who could afford them (Jenkins. 1990). A survey done in 1985 revealed that of 4,583 patients seen at genetic clinics in that year, only 18% were Black South Africans, despite Black individuals making up 74% of the county’s population at the time (Christianson, Venter, Modiba *et al*., 2000). In 1987, a genetic counselling clinic was established at Chris Hani Baragwanath Hospital (CHB) in Soweto, which predominantly provided (and still today provides) a service to the Black community from the surrounding areas (Levy. 1989). Genetic services have since been offered in major cities, tertiary institutions and some rural primary level outreach clinics throughout South Africa (Kromberg and Jenkins. 1988; Todd *et al*., 2010).

Despite this progression in medical genetic services, a recent article published by Todd *et al*., (2010) revealed that there were only 12 recognised medical geneticists and 11 practicing genetic counsellors in South Africa. In the greater Johannesburg area, genetic counselling services are currently offered by the Division of Human Genetics, National Health Laboratory Service (NHLS) and University of the Witwatersrand (Wits) at CHB, Charlotte Maxeke Johannesburg Academic Hospital (CMJAH), Rahima Moosa Mother and Child Hospital (RMH) as well as the Donald Gordon Medical Centre (DGMC) (Todd. 2007).

Numerous difficulties have arisen in providing healthcare in South Africa due to language barriers and a poor understanding of cultural differences (Solomon *et al*., 2012). English is the main language for communication in South Africa and few native English speakers are able to converse in an African language (Penn. 2007). A previous study by Levin (2006) showed that many individuals using healthcare services had a poor understanding of the language used by healthcare professionals leading to dissatisfaction, difficulty in understanding medical terminology and fear of not understanding a diagnosis and treatment, as well as difficulty adhering to prescribed medication (Levin. 2006). Education levels also vary greatly in South Africa and prior assessments in healthcare have shown that if an individual has a limited ability to read and understand healthcare information, this leads to poor health outcomes (Wasserman *et al.*, 2010).

South African studies have revealed that many genetic conditions do not have "traditional names" thus individuals from different cultural backgrounds often do not recognise conditions explained to them during a genetic counselling consultation (Christianson and Kromberg. 1996; Penn *et al.*, 2010). Some of the previously studied genetic conditions in children in South Africa include Down syndrome, albinism and haemophilia (Kromberg and Jenkins.1984; Christianson and Kromberg. 1996; Solomon *et al.*, 2012). These studies, together with other research, showed that many caregivers do not understand the medical causes of these conditions, that modes of inheritance and medical concepts such as "genes", "chromosomes" or "cells" were poorly grasped and that individuals held cultural beliefs and superstitions around the causes for these conditions (Kromberg and Jenkins. 1984; Christianson and Kromberg. 1996; Penn *et al.*, 2010; Solomon *et al.*, 2012). These findings lead one to question whether genetic counselling services are beneficial to communities where there is little knowledge of the conditions for which counselling and health management is offered (Christianson and Kromberg. 1996). Implementation of western counselling models may thus be unsuitable given South Africa's rich multicultural heritage and varying traditional beliefs on inheritance (Penn *et al.*, 2010).

Despite these previously mentioned barriers to effective genetic counselling implementation in South Africa, prior assessments on genetic counselling services have shown a positive outcome with regard to client satisfaction (Kopinsky. 1984; Levy. 1989). Kopinsky (1984) assessed 44 families in South Africa, who had children with Down syndrome, and had received genetic counselling between the years 1972-1979 and found that satisfaction levels amongst the families were high. Some suggestions made by participants were the expansion of services, a greater need of awareness of genetic conditions amongst the public and other healthcare professionals, and for improved co-ordination and management amongst different healthcare services (Kopinsky. 1984). Although an informative study, most participants were Caucasian, thus the study did not represent all ethnic groups in South Africa. Levy's (1989) study assessed 162 participants' experience of the genetic counselling services, the majority of whom expressed a positive experience. Similarly, a limitation was that of the 162 participants included in the study, only 25 subjects were Black South African individuals. The greatest represented group was Caucasian individuals and the remainder was individuals of Indian or Mixed Ancestry (Levy. 1989). Pre-counselling interviews revealed that most Black individuals had never heard of genetic counselling prior to their appointments nor did they understand their reason for referral. Post-counselling interviews showed that although most Black individuals could still not explain the concept of genetic counselling, 95% of the participants expressed that they were satisfied with the service and felt that their questions had been answered and expectations met (Levy. 1989). Problems reported by both participants and counsellors included the difficulty of communicating across different languages; the challenge of introducing first world services to third world communities as well as trying to overcome cross-cultural barriers. Further suggestions made were for ongoing support groups, the use of interpreters in genetic counselling sessions, producing culturally relevant educational material that is adapted to the literacy and language levels of the target population groups and greater public education on genetic counselling services (Levy. 1989).

These proposals have been echoed by other researchers who have made similar or additional suggestions. These include: modifying counselling styles so as to incorporate family members; the use of a mediator in the form of a respected member of the community or family to assist both the patient and healthcare provider overcome cultural and language barriers and using lay expressions and words to explain genetic inheritance (Penn *et al*., 2010; Solomon *et al*., 2012). Improvement and availability of health information given in the vernacular and at an understandable literacy level for all patients, as well as developing a better understanding of patients' cultural beliefs has also been recommended (Wasserman *et al*., 2010; Solomon *et al*., 2012).

1.2 Rationale for Study

Based on the literature, it can be seen that genetic counselling is an expanding healthcare profession and although genetic counselling values remain universal, healthcare settings vary greatly which affects the outcome of service delivery (Edwards. 2010). It is becoming increasingly apparent that besides standardised clinical models, genetic counselling models need to be adjusted to prevent disparities in healthcare communication due to cultural and linguistic diversity. These approaches need to be assessed globally to gain insight into the effectiveness of service delivery and how it has been received by clients. Little is known about the experiences of patients who have received genetic counselling in South Africa currently. Although both Kopinsky's and Levy's assessments in 1984 and 1989 were informative, to the researcher's knowledge no similar assessments have been repeated. Very few state patients' experiences of genetic counselling were included in these previous South African studies and since state patients form the majority of individuals seen for genetic counselling at the Division of Human Genetics, NHLS and Wits today, further research is warranted. An understanding of their experiences could provide an opportunity to improve the quality and effectiveness of service delivery and provide suggestions for a genetic counselling approach that is suitable to all individuals regardless of their cultural or linguistic background.

The purpose of this study is to describe and document the experiences of mothers of children with a genetic condition. These women received genetic counselling at a state hospital facility in Johannesburg, South Africa. The aim of the study is to gain a better understanding of these women's experience. The results of this research may provide insight for strategies that can be used to improve genetic counselling services in South Africa.

1.3 Research Aims and Objectives

The aim of this study is to explore the experiences of mothers who have received genetic counselling in a state hospital facility in Johannesburg, South Africa after having a child diagnosed with a genetic condition. Due to the methodology used, the researcher could not develop any theories on what to expect from the data, thus there are no specific objectives, except to describe and document the participants’ thoughts and feelings regarding the genetic counselling received in Johannesburg, South Africa.

2 CHAPTER 2: METHODS

2.1 Introduction

This chapter describes the methodological framework used for this study, the population and sample used, how the data was collected and analysed, the research assistants who were involved and how academic rigour was maintained. This was a prospective study in which new data was gained from selected participants. File records of patients seen at the Genetic Clinics run by the Division of Human Genetics were used to identify potential participants based on the research inclusion criteria. These records are kept by the Clinical and Counselling Unit of the Division of Human Genetics, NHLS, University of the Witwatersrand, Johannesburg, South Africa. Ethical approval was granted from the Human Research Ethics Committee (Medical), University of the Witwatersrand to conduct this study (Ethics Clearance Code: M111160) (Appendix A).

2.2 Methodological Framework for Study: Interpretive Phenomenological Analysis

The study design was qualitative, using a social constructivism research paradigm, which focuses on how an individual sees ‘them self’ (Atwater. 1996). It is influenced by how they view the world, their social experiences with other individuals and the acquisition of knowledge (Atwater. 1996). Prior research has shown that qualitative research methods may better capture the complexity and process of human interactions than quantitative research methods (Beeson. 1997). Qualitative methods have also been shown to be effective in clinical genetics and genetic counselling service evaluations as they have assisted in understanding the patients' experiences as well as determining what patients want from the service (McAllister *et al.*, 2008). As the aim of the study was to gain insight into mothers' experiences of genetic counselling, interpretive phenomenological analysis (IPA) was selected. IPA draws on aspects of phenomenology (the study of experience), hermeneutics (the study of interpretation) and idiography (the study of the 'particular') (Smith, Flowers and Larkin. 2009). It aims to understand how people perceive a particular experience in a particular context, how they interpret it, and how they make meaning of it in their personal and social worlds (Smith and Osborn. 2007; Smith *et al.*, 2009). This method does not depend on a predetermined hypothesis and utilises small, purposively-selected subjects in which participants are prompted with open-ended exploratory-type questions which allow them to speak freely about their experience (Smith *et al.*, 2009).

There are several methods to collect data in IPA; namely individual interviews, the use of diaries, computer-mediated information and focus groups (Smith *et al.*, 2009). Since a verbatim record of the data is required, all conversational interviews are video- and/or voice-recorded. It is also important that participants have a good rapport with the interviewer and feel comfortable in the interview setting (Smith *et al.*, 2009). A flexible question guide is used to prompt discussion and to ensure that areas of interest are covered, however the participants act as the main conversationalists and may thus redirect the discussion to topics that are not part of the interview schedule (Flowers, Knussen and Duncan. 2001; Smith *et al.*, 2009). All collected data are then transcribed verbatim. Since IPA focuses on the content of the participants' accounts, documentation of the length of pauses or non-verbal expressions is not required (Smith *et al.*, 2009).

During the analysis, IPA does not focus on one set “method”, but rather uses a principle that aims to describe a participant’s point-of-view of an experienced phenomenon and to interpret what it means to the participant (Flowers *et al*., 2001, Smith *et al*., 2009). IPA analysis requires reading and re-reading the original data and making initial notes and comments (Smith *et al.*, 2009). From this, themes start to emerge from individual interviews and across interviews, with the researcher making connections between recurrent themes to create super-ordinate themes (Smith *et al*., 2009). These themes are then interpreted so as to understand and capture the meaning of the phenomenon experienced by participants (Flowers *et al*., 2001; Smith *et al*., 2009).

For this particular project, focus group interviews were selected since they create a non-judgmental atmosphere that encourages a range of different opinions, therefore allowing a broader and deeper understanding of the issues discussed (Bloomberg and Volpe. 2008). Individuals may also be more willing to express their feelings in a group of people with similar issues. A focus-group participant number of four to five individuals has been recommended for IPA research and it has been suggested that an incentive may be necessary to encourage participants to take part in the focus group (Burns and Grove. 2001; Smith *et al*., 2009). Previous research by Penn *et al* (2010) showed that focus group interviews have proved effective in the South African setting, as a rapport is created amongst participants, which facilitates participation in the conversation. This encouraged the researcher to make use of this particular interviewing-method.

2.3 Study Population

The study population comprised of women who had received genetic counselling, after having a child diagnosed with a genetic condition, at one of the Genetic Clinics run by the Division of Human Genetics, NHLS and the University of the Witwatersrand, at a state-run hospital.

2.4 Study Sample

Purposive sampling using file records of patients seen at the Genetic Clinics run by the Division of Human Genetics was performed. The patients had been seen with their mothers at clinics at CHB, CMJAH and RMH. Most patients from the research sample had been seen by a genetic counsellor, although some had been seen by a clinical geneticist. The sample selection was based on mothers who have had a recent genetic counselling interaction, who could converse in the appropriate language and who has easy access to the research site.
The inclusion- and exclusion-criteria are listed below:

<u>Inclusion Criteria:</u>

- Women who had a living child younger than five years of age who had been diagnosed post delivery with a non-lethal genetic condition.
- Women residing in the greater Johannesburg area.
- Women who had received genetic counselling between January 2010 and January 2012.
- Women who could converse in isiZulu, isiXhosa, seTswana and seSotho.

<u>Exclusion Criteria:</u>

- Women whose child had died since being seen at the Genetic Clinic.
- Women who had children with a lethal genetic condition.

- Women not residing in the greater Johannesburg area.
- Women from Caucasian, Asian or Mixed Ancestry populations.

Patients who met the criteria for the study were contacted telephonically by two research assistants. The purpose of the study was explained to each client and they were invited to participate in the research. Thirteen Black South African women who were counselled in the state healthcare system agreed to participate. Of these women, nine have children with Down syndrome, three have children with albinism and one woman has a child with haemophilia B.

2.5 Research Assistants

Five assistants helped with the research project. They contacted potential participants telephonically, assisted in facilitating the focus groups as well as in the transcription and translation of collected data.

Two research assistants, employed by the Division of Human Genetics, NHLS, Braamfontein, Johannesburg, contacted the potential participants telephonically. They are both fluent in isiZulu and seTswana and the conversations were conducted in the respective languages. The purpose of the research was explained, the individuals were invited to participate and the voluntary nature of the study was emphasised.

The focus group facilitator was a Black South African woman experienced in qualitative interviewing and focus group facilitation. At the time of the interviews she was employed at the Centre for Support and Peer Education (CSPE) in Johannesburg. She has a Bachelor of

Arts degree from the University of the Witwatersrand and a qualification in Human Immune Virus/Autoimmune Deficiency Syndrome (HIV/AIDS) Care and Counselling from the University of South Africa (UNISA). Her mother tongue is isiZulu, but she is fluent in isiXhosa, seSotho and seTswana, and focus groups were conducted in these four languages.

Two senior students assisted in the transcription and translation of collected audio-recorded data from the focus groups. One has a Bachelor of Science Honours (BScHons) degree from Wits and has since worked in the corporate industry. She is currently completing a part-time degree in psychology at Wits. The other has completed a Bachelor of Science degree at Wits and is in the process of completing a BScHons degree. Both assistants are able to converse in English, isiZulu, isiXhosa, seSotho and seTswana.

2.6 Data Collection

Data were obtained through the conduction of focus groups. Permission had been granted by the NHLS Outpatient Department (OPD) in Braamfontein, Johannesburg, to perform the focus groups at their location. This location was ideal as it provided a relaxed and secure environment with minimal disruptions that facilitated open discussion. In addition it is closely situated to Park Station, a large converging site for public transport services and easily accessible for the participants. All participants signed consent forms and were provided with an information sheet (Appendix B).

The first focus group was used to assess the process, the suitability of questions and to gauge how long it would take to conduct a focus group. Adjustments were then made accordingly. A loosely-based question guide was developed by the researcher to prompt discussion within the focus groups (See Appendix C). The focus groups were formed according to the participants' preferred language. IsiZulu and isiXhosa speaking individuals were grouped together since both are Nguni languages and are similar in dialect (B Baloyi 2012, Personal Communication, January). Similarly, seTswana and seSotho speaking individuals were placed together because of the similar dialects (B Baloyi 2012, Personal Communication. January). As the researcher is only conversant in English and Afrikaans and wanted to minimise any language and cultural issues that may arise, the researcher felt that the data collection would be richer if participants were given the opportunity to express themselves in a language in which they were most comfortable. The researcher was however present in all sessions to observe, take notes, moderate the session and answer any questions that may have arisen.

Four focus groups were conducted (Table 2.1). Each focus group took 60 to 90 minutes and all sessions were voice-recorded. The women were asked not to bring their children to the sessions however three participants brought their children to focus groups one, three and four. This did however not change the dynamic of the sessions nor cause disruption. Refreshments and lunch were served at each session and the participants were compensated for their transport fees to and from the focus group venue. Before each session, the purpose of the study was explained and the voluntary nature of attendance was emphasised. All women agreed to participate and written consent was obtained. The researcher felt that the facilitator asked appropriate questions and allowed the participants to guide the conversations. Although some participants were more reserved, the majority were very communicative and readily expressed their opinions and experiences of genetic counselling.

Table 2.1 Composition of Focus Groups

Focus Group	Date: 2012	Participant Number	Interview Language	Children’s conditions
1	31.01	3	isiZulu	All Down syndrome.
2	02.02	3	seTswana	1 Down syndrome, 2 albinism.
3	01.03	4	isiZulu	2 Down syndrome, 1 albinism, 1 haemophilia B.
4	01.03	3	seTswana	All Down syndrome.

2.7 Data Analysis

The data obtained from the focus group voice-recordings were transcribed with the assistance of a software program called SoundScriber®. These transcriptions were then translated into English and the data from each focus group were analysed thereafter. Focus group one and two were transcribed and translated simultaneously and were analysed before focus group three and four were transcribed and translated in order to clarify and assess the value of the transcription and translation styles used and to see whether the data obtained from the question guide were useful.

Two senior students assisted in the transcriptions and translations and each focus group was cross-checked by each student for inconsistencies. The audio-recordings and the researcher’s notes from the focus groups were given to the two assistants to help identify the conversations and participants. The participants’ initials were used to protect their identities during the production of the transcriptions. As the research assistants were not native English speakers, spelling and grammatical errors were corrected, but the translated transcripts were largely left in their original format.

The researcher used thematic content analysis (TCA) to analyse the data. TCA was selected as IPA required the extraction of "content areas" or "coding" so as to categorize and analyse themes to create a comprehensive picture of an individual's experience of a phenomenon. Each focus group's data were analysed separately and then the data from all focus groups were cross-analysed and compared. Categories and recurrent topics were noted and sorted for the development of "content areas" or "codes". These "content areas" were further sorted for the development of emergent themes with cross-connections then made to form "super-ordinate" themes. For example in Chapter 3: "Theme 1: Thrown into the Unknown.: General Lack of Awareness" was initially coded as "lack of knowledge". The research supervisors individually analysed sections of the original translated transcriptions to verify emerging themes, and the themes were then discussed between the researcher and the supervisors until concurrence was reached. The research findings were also discussed with specialists who have valuable experience in similar qualitative research within the healthcare communication and qualitative fields. Although the researcher aimed to remain objective, she was continuously aware of the potential bias inherent in a study of this nature. To address this, the researcher had regular interaction with her supervisors and a research group.

2.8 Ensuring Academic Rigour

Rigour in qualitative studies is maintained through openness, careful adherence to the philosophical perspective, careful data collection, thorough and systematic data analysis, meticulous consideration of all data in the development of individual theories as well as presenting a coherent and transparent report of the study methods and results (Burns and Grove. 2001; Smith *et al*., 2009).

To ensure academic rigour, the researcher did not develop any prior theories on what to expect from the data and the phenomenological research method was adhered to closely. Thorough documentation was made for each step of the research process and the researcher maintained transparency throughout the project. All research assistants contributed to the rich data obtained. The focus group facilitator contributed to obtaining valuable data since she has previous experience in conducting focus groups and doing qualitative research. The focus groups were closely monitored by the researcher to ensure that they were conducted uniformly, that discussions were in-depth and that participants felt at ease. The methods used in translation and transcription of the data were closely assessed to prevent inconsistencies. Both research assistants involved in the transcriptions and translations were able to understand and converse in all the respective languages and all data transcribed and translated were cross-checked.

For the analysis of data, all focus groups' data were read and re-read several times. Data from all focus groups were analysed uniformly and thoroughly. In-depth discussions were held with the research supervisors regarding the research results to verify themes. Additional input and discussions were held with Professor Claire Penn from the Department of Speech Pathology, as well as members of the Health Communication Project at Wits, all of whom are specialists within the fields of healthcare communication and qualitative research. The research findings were also presented at a Health Communication Project meeting on 02 August 2012. Several translated transcriptions were included in the results to support interpretations as well as to represent the “voices” of the participants. All data were considered during the development of themes and the researcher tried to maintain a non-biased and open perspective to the research.

In summary, this research aimed to describe the experiences of mothers who received genetic counselling at a state healthcare facility in Johannesburg, South Africa after having a child diagnosed with a genetic condition using a qualitative IPA method. Focus groups were used to collect data which were transcribed and translated for the analysis and the development of themes. All methods used, as well as emergent themes, were carefully documented, analysed and discussed so as to ensure academic rigour and transparency.

3 CHAPTER 3: RESULTS AND DISCUSSION

3.1 Introduction

This chapter will include a brief description of the participants (Table 3.1) and will present and discuss the major themes identified from the data together with the relevant sub-themes. Relevant issues are illustrated by including excerpts from the translated transcriptions to validate the topics discussed and to include the "voices" of the participants.

3.2 Description of the Participants

Participants were not asked directly about their demographic information during the focus groups. However some of this information was obtained from their patient files.

Table 3.1 Participant Demographic Information

Focus Group	Participant	Age	Nationality	Home Language	Occupation	Child's Condition
1	Ms. E.	37	South African	Zulu	Unemployed	Down syndrome
1	Ms. P.	38	South African	Xhosa	Waitress	Down Syndrome
1	Ms. S.	42	South African	Zulu	Cashier	Down syndrome
2	Ms. T.	22	South African	Tswana	Student	Albinism
2	Ms. K.	19	South African	Tswana	Unemployed	Albinism
2	Ms. H.	41	South African	Sotho	Unemployed	Down syndrome
3	Ms. J	37	South African	Zulu	Unemployed	Down syndrome
3	Ms. F.	21	South African	Xhosa	Nursing student	Haemophilia B
3	Ms. P.K.	43	South African	Zulu	Unemployed	Down syndrome
3	Ms. A.	25	Zimbabwean	Shona*	Hairdresser	Albinism
4	Ms. N	23	South African	Sotho	Call Centre employee	Down syndrome
4	Ms. M	33	South African	Tswana	Student	Down syndrome
4	Ms. P.J.	43	South African	Tswana	Waitress	Down syndrome

*Ms. A. could also speak Zulu.

3.3 Themes Identified

Themes were developed by the researcher after reading through the transcripts several times and linking topics that arose during the focus groups. These themes, together with the relevant sub-themes were then verified through discussions with the research supervisors and other research experts. Table 3.2 is a summary of the 6 main themes and sub-themes identified from the translated focus group data. Each theme is presented together with the sub-themes, and discussed and compared to previous research. Examples of related excerpts are included to provide proof and strengthen the arguments made. The excerpts include the English as well as the original language.

Table 3.2 Themes Identified

Theme 1: Thrown into the Unknown: General Lack of Awareness **Sub-themes:** • Lack of Awareness of Genetic Counselling • Lack of Awareness of the Genetic Condition
Theme 2: Experience of Genetic Counselling **Sub-themes:** • Was it Helpful? • Value of the Information Provided: “They Explained Everything” • The Important Role of Partners • When is the Right Time for Counselling? • Support as a “Life-Line” • Transport Difficulty and Access to Services
Theme 3: Personal Beliefs **Sub-themes:** • Religion • Traditions • Language Barriers
Theme 4: Addressing the Community and the Family • Community’s Views • Telling the Family
Theme 5: Dissatisfaction with the Healthcare System **Sub-themes:** • Feeling Unheard and Disrespected • State Healthcare Professionals: “They Are Very Much Ignorant”
Theme 6: Greater Need for Awareness Campaigns

3.4 Theme 1: Thrown into the Unknown: General Lack of Awareness

Most participants, prior to receiving genetic counselling, did not know what genetic counselling was, nor the cause of their children’s conditions, and had carried a sense of fear and guilt. Few participants were told, when referred to genetic counselling by other health professionals, what to expect from the session. Overall, these concerns caused a sense of the “unknown” amongst the participants. These issues will be explored in more detail in the following two sub-themes.

3.4.1 Lack of Awareness of Genetic Counselling

Overall, it was found that the women in the study did not know what genetic counselling was, they did not understand why they had been referred and they were unaware of the availability of services. Previous research has shown that members of the public have a poor concept about basic genetic information, often do not understand the terminology used such as “genes” and “chromosomes,” and few have a clear understanding on what to expect from a genetic counselling consultation (Davey, Rostant, Harrop *et al*., 2005; Klitzman. 2010; Solomon *et al*., 2012). This was shown in this study as eleven of the participants expressed that they had never heard the words “genetic” or “genetic counselling” and did not know what to expect from the session (see Example 3.4.1).

> ***"I wasn't aware of what they were talking about but I just went to that appointment so that I can find out more about it. I didn't know what Genetic Counselling was and they didn't say that I was going for Genetic Counselling, they just said I was going for "genetic" and did not explain that I was going for counselling."***
>
> *Bengingekho aware bakhuluma ngani but I just ngaya kuleyo appointment yahkona ngifun'ukwazi ukuthi kungani. Bengazi ukuthi yini iGenetic Counselling. Futhi abangazana bathu uya kugenetic bangazange bay'chaza ukuthi icounselling."*
>
> (Example 3.4.1: Ms. P., has a child with Down syndrome, Focus Group 1)

A South African study conducted in 1987 revealed that many individuals, particularly Black South African individuals, did not understand what genetic counselling was nor the reason for their referral (Levy. 1989). To the researcher's knowledge, no research regarding the public's awareness of genetic counselling in South Africa has since been repeated apart from this current study, which has shown similar results. Research has shown that genetic counselling uptake is generally higher amongst Caucasian individuals in predominantly English-speaking countries than amongst individuals from other cultural, ethnic or linguistic backgrounds and that most clients are referred by another healthcare professional, except for some middle- to upper-class Caucasian individuals who are self-referred (Hutson. 2003; Davey *et al*., 2005; Barlow-Stewart *et al*., 2006). Most of the participants in this study came from lower socioeconomic settings and all of the participants were from Black ethnic backgrounds.

Studies have shown that individuals' reasons for attending a genetic counselling session is influenced by education levels, wanting more information on the risks and management of a known genetic condition, interest in genetic testing and whether clients have been correctly informed about genetic counselling (Collins *et al*., 2001; Culver *et al*., 2001; Davey *et al*., 2005). In this study, most participants had not been told by their referring healthcare professional as to what to expect, and most said they only came to genetic counselling out of instruction or curiosity. Only two participants said that they had heard the word "genetic" and had an idea of what to expect, but did not know exactly what genetic counselling entailed; one of them is a nursing student and the other had read about it in her sibling's high-school biology textbook (Example 3.4.2).

> ***"When I went, because I had read about Down syndrome, like my siblings when they did biology at school, they covered it."***
>
> *"Nna ha ne kea because nna keile kabala ka Down syndrome. Like ngoana hesa etsa biology."*
>
> (Example 3.4.2: Ms. N., has a child with Down syndrome, Focus Group 4)

The findings from this study are similar to other research, both locally and internationally.

3.4.2 Lack of Awareness of the Genetic Condition

Most participants who had children with Down syndrome, although they recognised that their child had a "problem" before receiving genetic counselling, did not know what had caused it. Two women said that they had noticed that their child was "different", but did not understand what the condition was and they had an underlying feeling of fear as to what was happening with their child. In addition, some expressed that they initially felt guilty, as they thought they might have caused the problem (Example 3.4.3).

> ***"So when I got there, they told me what a genetic problem is, that we are going to talk about the child and why the child is like this. Then they explained to me that it is not my fault that the child is like this and that there is nothing wrong that I did for the child to turn out like this and then they explained that it is because of my age. They then further explained why he is like this. I was relieved because I thought there was something wrong that I had done."***
>
> *"So ngithe mang'fika bangitshela ukuthi yini iGenetic problem, sizokhuluma ukuthi yini iGenetic counselling, sizokhuluma nge ndaba yomywana ukuthi why esona; then baye bang'cazela ukuthi it's not my fault ukuthi lomtwana abe so akwenzanga, akukho nix engiy'yenzile ukuthi abe ka so, base bayachaza it's because of my age bachaza ukuthi abe kanjalo so ngaye ngakhuleka ngo I thought ukuthi something ewrong engiy'yenzile ukuze abe ka so umtwana.*
>
> (Example 3.4.3: Ms. P., has a child with Down syndrome, Focus Group 1)

As there was a lack of knowledge about the aetiology, many of the partipants felt that they had caused the condition in their child. One woman expressed that she thought that she had unknowingly taken pills or drank something that had caused her child to have Down syndrome. Other research has shown that women believe that drinking alcohol or taking medication during pregnancy can cause disability in a child and many have cultural, religious and personal beliefs on the cause of a condition (Weil. 1991; Penn *et al.*, 2010).

In this study, several participants had previously heard of, or recognised the conditions mentioned to them, but did not fully understand what they were, and almost all of the women did not know about the genetic cause. One participant who has a child with haemophilia B, said she had heard about haemophilia but did not know at the time that there were different types. Most participants who have a child with albinism recognised the condition, however did not understand how an individual gets the condition. Two women, with children with Down syndrome, said that they had read or heard about the condition in the media, but neither knew the cause, nor what to expect for their children (Example 3.4.4).

> ***"Ok when I went, I went there not knowing. I read about Down syndrome in the magazines and in the "Great Expectations" (magazine), but I, the results of trisomy 21, I was confused because I don't know what to expect from the child, and I don't see anything wrong, apart from the flat nose, I was in denial."***
>
> *"Ok nna ha kea for, keile ke sa tsebe, no Down syndrome ke badile ka yona mo di magazining le mo di "Great Expectations", mara ke, de results tsa trisomy 21, I was confused because I don't know ke expectang mo ngoaneng, and nna I don't see any wrong ya ngaphandle kwe flat nose, nna ne ke le mo denial."*
>
> (Example 3.4.4: Ms. M., has a child with
> Down syndrome, Focus Group 4)

Previous South African research, has shown that mothers who have children with Down syndrome and haemophilia have a poor understanding and recognition of these conditions, with many mothers or care-takers not recognising the words "Down syndrome" or "haemophilia", nor understanding the medical terminology used and often do not recognise the clinical features mentioned to them (Christianson and Kromberg. 1996; Solomons *et al.*, 2012). Conversely, mothers who have children with albinism recognise the condition as it is fairly common in Sub-Saharan ethnic groups and because the features are so strikingly different from pigmented individuals (Lund. 2005). One of the participants in the study understood that albinism is a condition that can occur in a family, and said that she had distant relatives who had albinism and knew there might be a chance that she or another family member could have an affected child (Example 3.4.5).

> ***"I don't want to lie, it came as a shock. After he was born, like, obviously those initial 2 minutes, it was like ok, but afterwards, I thought ok this child is beautiful, he's different, and I knew there was a possibility that I could get one. Not that we have close relative who are albino, but distant distant. So we always talked about it… that maybe it could come up in the family. So it wasn't a huge shock or surprise."***
>
> *"Haai, I don't want to lie. Ha fetsa ho thlaga, ok obviously that 2 minutes you know, but after I was like ok, ngoana o omontle yang, o sharp o different and I knew gore there was a possibility nenka mo krea. Not that we have like ba, di close relatives tsa hae but distant distant, we always talked about it hore you know mayb ba ka tlhagella in the family, ok, so it wasn't like a huge shock or surprise."*
>
> (Example 3.4.5: Ms. T., has a child with albinism, Focus Group 2)

More research is needed regarding the public's knowledge of genetic counselling and genetic conditions and genetic counsellors need to be sensitive to individual's perceptions.

3.5 Theme 2: Experience of Genetic Counselling

Participants had different experiences of genetic counselling. Several sub-themes have been identified and the specific issues that arose during the focus groups will be discussed.

3.5.1 Was it Helpful?

Clients expressed both positive and negative experiences of genetic counselling; those that had a good experience commented on the way they had been respected in the session and that the genetic counsellor/geneticist had showed compassion and empathy towards them (Example 3.5.1).

> Ms. S.:***"Firstly, the people they know how to communicate with us and it was the first time seeing doctors who are like this. Because we generally know what people who work in hospitals are like so it was the first time seeing health professionals who even have time for you and you can even ask questions thereafter. Even if you don't understand English, they try speak to you so that you can understand and tell you and explain it to you, read to you, show you pictures and they also give you contact numbers where we can reach them if we want to call them for any reason."***
>
> *"Into'qala, bayakwazi ukukhuluma nathi, uku..yazi ngithi ngiyaqala ukubona mina aboDoctor abanje. Siyazi like abantu abasebenza eyibhedlela the most banjani, ngiyaqala ukubona… ba banes'khatisakho, ungababuza anything.. uyahlala so and noma ungazwisisi mahlaumbe understand iEnglish, azame ukuhuluma slow nawe azame ukuthi understand; ak'tshele, ak'chazisele, akuphe iyncwadi akufundele like slow like makaneyitombe akubonise ukuthi abakho ba right; ene bayasinika ama number ukuthi makabe sifuna ukuphona for anything."*
>
> Ms. H.:***"Yes shame sis, they even ask about your background and they are very understanding and at that time you are going through so much pain, the Genetic Doctor will show you that he's also feeling very compassionate and they also show that they are also sharing your pain because they acknowledge that it's not easy to hear this from them and we start crying and they're even patient as they wait for you to finish crying."***
>
> *"Uya shame they did shame… and akubuze nangebackground yakho yabona, ukuthi kanje ekhaya yabona aunderstande. Even noma kunzima yabona ngalesoskhathi usebuhlungwini yabona…o..o.. that thing yabona ufila… akhbonise uGenetic Doctor ukuthi he's feeling pain also shame athi akukho mnandi masik'tshela, inyembezi bese zithi, abekezele ukuthi zimane ziqale ziphele lezinyembezi.*"
>
> (Example 3.5.1: Ms. S. and Ms. H., both have children with Down syndrome, Focus Group 1)

There has been very little research on the experiences of clients seen for genetic counselling in South Africa. Solomon *et al* (2012) examined the experience of caregivers of a child with a genetic condition, however to the researcher's knowledge, the last study to assess client experiences of genetic counselling in particular, was conducted in 1989 (Levy. 1989; Solomon *et al*., 2012). Participants in the focus groups, particularly group 1, valued qualities of respect and empathy in the genetic counsellor or geneticist. Previous research has also shown that clients value genetic counsellors' friendliness, respect and personal interest in them; and that they feel appreciated and that the personal attributes of the counsellor and how they relate to the patient on a personal level, greatly influences the experience and how they perceive the information (Skirton. 2001; Davey *et al.*, 2005). Users of primary healthcare services in South Africa are often apprehensive about attending public healthcare facilities due to the anticipated disrespect by healthcare professionals, lack of patient dignity, long queues, feeling disempowered and inadequate provision of information about the medical condition concerned (Harris, Goudge, Ataguba *et al*., 2011; Vivian, Naidu, Keikelame *et al*., 2011). Participants in this study seemed surprised when they attended genetic counselling as they had anticipated the same type of behavior towards them that they received from other staff in the South African healthcare system.

Another example of the appreciation for genetic counselling can be seen in the words of Ms. H. She said that it was through genetic counselling that she learnt about her child's condition and that if it had not been for genetic counselling, she would not have fully understood Down syndrome (Example 3.5.2).

> ***"I often say to people that I'm also grateful to the girl who told me. It was late at 8 months. Even now, I still maintain that she helped me a lot. If it wasn't for her, we would still be in the dark."***
>
> *"Lenna ke ebe ke tshelo ko bathing ke re ke leboha ngoanyana osale a mpolella. Ntshoetse bang mpolelle, so le nou ke satsho hore onthusitse, if ne ese ka ena nkebe today esetse ele boroko."*
>
> (Example 3.5.2: Ms. H., has a child with
> Down syndrome, Focus Group 2)

Overall, reasons for satisfaction with genetic counselling in this study included: receiving adequate information about the genetic condition, the use of appropriate understandable language and diagrams by counsellors to explain concepts, giving participants contact numbers and pamphlets to take home, referring participants to appropriate support groups, allowing participants to ask questions, and the offer of emotional support and respect. These findings are in keeping with some of the findings in other research by Michie *et al* (1997) and Davey *et al* (2005).

There were some participants who were dissatisfied with their genetic counselling experience. One participant, who has a child with albinism, did not find it helpful as compared to others. She found the session very long with a lot of information and she seemed annoyed that the counsellor asked her intrusive questions about her family members (Example 3.5.3).

> ***"It helped a little bit, not a lot."***
>
> ***"But you know sometimes, I don't know the doctor's name, the doctors want information that they want to gather. They ask who in the family had it. That I didn't find so important, it's not like after that you managed to go figure out ok exactly this is what happened because what does it help at the end? What are they going to do about it?"***
>
> *"Nna eng thsusitse ha nyane."*
>
> *"You know sometimes, I don't know ke doctor mang mare nkare they just want the information for the bona fela, ho botsa hore ko kae ko familing you know, ke mang ke mang so? That I didn't find so helpful because e tlo re thusa ka eng at the end? What are they going to do about it?"*
>
> (Example 3.5.3: Ms T., has a child with albinism, Focus Group 2)

Studies have shown that patients who are dissatisfied with the service, did not have their questions answered; genetic counsellors or the clinical geneticist had not made sufficient effort to contact other healthcare professionals involved in the patients's management, patients had received negative answers which they had not anticipated, or they had not received adequate information on how the condition would affect their daily lives (Bleiker *et al*., 1997; Bernhardt, Biesecker, and Mastromarino. 2000; Davey *et al*., 2005). Ms. T. (Example 3.5.3) may not have understood the relevance of the questioning about albinism or identifying other genetic conditions in her family, either because the genetic counsellor had not explained the purpose of obtaining a family pedigree, or she may not have understood the concept of heritability, particularly autosomal recessive inheritance, the manner in which albinism is inherited. The term "family" may also hold a different meaning for some individuals and may not only relate to the immediate family members, but may also represent extended family members or non-related individuals living together in the same household (Richards. 1996; Peterson. 2005). Some individuals do not feel comfortable conveying sensitive information about their families to strangers because they want to protect their family's reputation (Saleh *et al*., 2009). Ms. T. may have been protective about discussing her family members with the genetic counsellor and may also have understood the term 'family' in a different context to the counsellor.

Another participant, Ms. A. (Example 3.5.4), also did not have a positive experience of genetic counselling as she felt uncomfortable that there were several students observing the session and that her confidentiality had not been taken into consideration.

> ***"I when I went for that…. Uhm counselling at general at general hospital, at Johannesburg general, there were four ladies in the room with me. Sometimes you know there are people who don't like to express themselves in front of a crowd of people, you know that sometimes you can feel emotional because it's not easy for me to have that baby you understand. Of course now I'm ok with it but when you have the baby and you see them two ladies, the genetic counselor and the assistant and then there'll be two students (Facilitator: "Who are there to observe."), you understand it's very uncomfortable."***
>
> *"Mina when I went for that…uhm counseling at general at general hospital, Johannesburg, there were four ladies in one room with me. Sometimes uyazi there are people that don't like to express themselves in front of a crowd of people you know that sometimes you can feel emotional because it's not easy for me to have that baby you understand. Of course now I'm ok with it but when you have the baby and you see them two ladies, the genetic counselor and the assistant neh and then there'll be two…. Uhm I think they are students (Facilitator: "bazo observa.")… uya understand, it's very uncomfortable."*
>
> (Example 3.5.4: Ms A., has a child with albinism, Focus Group 3)

CHB, CMJAH (Previously called Johannesburg Hospital) and RMH (previously called Coronation Hospital) are teaching hospitals used by the University of the Witwatersrand Medical School, and genetic counselling services are offered at these hospitals as well as at Donald Gordon Medical Centre (Myers and Bibl. 1995; Todd. 2007). Genetic counselling students observe genetic counsellor/geneticist-patient interactions from their first year of study and in some universities' medical curricula, medical students have exposure to patients from their fourth year of study (Du Preez, Pickworth and van Rooyen. 2007). At times, a genetic counselling student as well as several other nursing or medical students are present in a consultation to observe the genetic counsellor/geneticist-patient interaction. Research in hospitals in South Africa have shown that patient confidentiality and privacy is often not respected and patients are dissatisfied with the way their information is openly heard by other individuals without their consent (Harris *et al*., 2011; Vivian *et al*., 2011).

3.5.2 Value of the Information Provided: “They Explained Everything”

The value of the information provided to the participants during their genetic counselling sessions was another topic that was frequently discussed. Many of the participants seemed to value the information provided to them and although some the information was not understood correctly, many participants were generally able to explain clinical features of, as well as the genetic cause of their children’s conditions (Example 3.5.5).

> Ms. H.:***“Yes so it becomes a mess because it causes the child to be like this because a person needs to have 23 so when those chromosomes are equal then they make up 46 from the mother and father to be normal. So instead of making 23, I made 24, so that is why my child was like this, so it was because of the chromosomes.”***
>
> *“Eya kwamele edibane ayenza ngakhi 23 bese adibane 46, 23 ubab 23 umama. So lezoyi chromosomes mayelingene ayeza 46 uba normal…but umakuyanza u23 kwaba 24 that is why kuba nama results anje ku my child so, ama chromosomes.”*
>
> Ms. S.:***“Yes I understood because I even got books and they told me to read them at home so that I can understand what’s going on yes.”***
>
> *“Ya mina ngi understandle ngoba ngize ngathola neyicwadi, then bang’nikeza ukuthi ngifunde ekhaya ngiyazi ukuthi kahle kwenzakalani yah.”*
>
> (Example 3.5.5: Ms. H and Ms. S., both have children with Down syndrome, Focus Group 1)

The ability to recall this information may indicate that the information provided to the patients during a genetic counselling session may have been personally relevant, thus beneficial in some way. The patients seemed to know about the inheritance patterns, what management and treatment their children required and they expressed that the genetic counselling sessions had been informative, that their questions had been satisfactorily answered and that they had received helpful resources, as one participant explained (Example 3.5.6).

> ***"Plus even the pamphlets they gave us explained almost everything, you know how it was that the child should be like this, that you and the father had the gene for the condition and the combination of your genes would create Phindile*, like everything. They explained everything. They told me that though there isn't someone like that from both families, but you and the father have the genes, that's why the child is like this."***
>
> *"Plus le di pamphlets tse ba re fileng ke tsona di explaina almost everything, you know, hore go tlile joang motho abe so ho tsho hore oena le ntate oa hae le ena na nale di genes, le oena onale tsona, so hale hlakana, obviously di tlo etsa trouble in everything. They told me that though there isn't someone like that from both families, but you and the father both have the genes, that's why the child abile teng."**Child's name has been changed.
>
> (Example 3.5.6: Ms. K., has a child with albinism, Focus Group 2)

Studies have shown that patients feel more prepared to cope with a condition after a genetic counselling session than they were beforehand, as they believe that they gained resources, information and psychological support to understand the condition (Davey *et al*., 2005).

Although some participants found that the information gained at their genetic counselling sessions was beneficial, others did not find the session as valuable. This stemmed from a lack of understanding of the genetic terminology and "medical jargon" discussed during their consultation, and they expressed that the information given to them, and the diagrams shown, had been confusing (Example 3.5.7).

> ***"But I just feel that there are some things they will rather express or elaborate to other women who are not well equipped educationally you know… because they'll just be drawing some signs. This is an albino, for you to get an albino this is A and B. So sometimes you do not understand, you won't even get the sense of what they are trying to say to you, you see. So sometimes I think maybe they must just try explain in a way that someone can understand, you understand?"***
>
> *".....But I just feel that there are some things they will rather express or elaborate to other women that are not well-equipped educationally you know.... Because they'll just be drawing some signs, this is the albino, for you to get an albino this is A and B. So sometimes I think maybe they must just try and explain in a way that someone can understand, you understand?"*
>
> (Example 3.5.7: Ms. A., has a child with albinism, Focus Group 3)

The concepts of "genes" and "chromosomes" and how they relate to inheritance are difficult to understand not only in developing countries, but also in first world nations (Solomon *et al*., 2012). Education level is an important factor to keep in mind when explaining information to an individual as research has shown that health literacy is often related to this (Kagee, Le Roux and Dick. 2007). Individuals with a higher education level may show a greater interest in learning about genetics, whereas those who do not, may be less interested and or might feel intimidated by the language and terminology used (Culver *et al*., 2001).

With regard to the South African setting, many individuals counselled at CHB, CMJAH and RMH are from previously disadvantaged areas, with many of these communities not being given adequate educational opportunities (Kagee *et al*., 2007). Due to the country's apartheid history as well as current difficulties in the education system, many individuals have poor numeracy, literacy and analytical skills (Coovadia *et al*., 2009). Although it is unknown what the participants' educational levels are, based on the previous research by Kagee *et al.,* (2007) and Coovadia *et al*., (2009) the findings in this study may explain why some of the participants felt overwhelmed and intimidated by the genetic information explained to them during their genetic counselling consultation. However, these points need to be further explored to gain a better understanding of this issue. This may assist in deciding what information conveyed during a genetic counselling consultation is beneficial to the patient, and what is not.

With regard to what information is considered to be beneficial or important to a patient, information not recalled by patients may be regarded as irrelevant, or not directly beneficial to themselves or their child (Michie *et al*., 1997). In this study, this was noted particularly when recurrence risk information was discussed, as is shown in the example of Ms. F. (Example 3.5.8), who believed that haemophilia "skipped generations".

> ***"….. cos I, I know that it, it skips a generation and then another one comes up, but sometimes things don't happen the way, it's the only thing that concerns me."***
>
> *"…… cos I, I know that it, ifitisa generation and the nhotla engwe, but sometimes things don't happen the way, ke the only thing eke na le yona."*
>
> (Example 3.5.8: Ms. F., has a child with haemophilia, Focus Group 3)

Although she is a nursing student, Ms. F. believed that the condition's pattern of inheritance was different from what had been explained to her. This issue can also be seen by another participant, Ms. A., who has a child with albinism. Ms. A. felt that she had a higher risk than other individuals who are carriers for albinism, and felt that her chance of having another affected child in a future pregnancy was 50% (rather than the 25% recurrence risk that had been explained to her).

It has been shown that unless information such as risk is individualised, patients may not see the relevance of the risk to them, and may not see the significance of this information (Lobb, Butow, Barratt *et al*., 2004; Fransen, Meertens and Schrander-Stumpel. 2006; Kinney *et al*., 2010). More research is needed on this topic, to see what information is beneficial and what is not, and how this information is personally understood and interpreted in individuals own lives.

3.5.3 The Important Role of Partners

The role of a father within society had been portrayed religiously, morally, methaphorically and culturally for centuries (Richter and Morrell. 2006). The fathers of the children in this study seemed to play an important role in the participants' and children's lives, and recommendations were made on several occasions by the participants for their partners to attend genetic counselling. Participants felt that genetic counsellors need to emphasise that the counselling is for 'couples' not only for mothers (Example 3.5.9) as they felt that it would be more beneficial for their partner to hear the information from a health professional rather than from themselves.

> ***"I think that most of the time when there are counselling sessions, for genetic counselling, they must try and call the couple, you see they were still together when they had the child."***
>
> *"I think ukuthi most of the time makunama counseling ne, njengalawa macounseling, ama genetic counseling, they must try and call the couple, yabona they were still together when they had the child."*
>
> (Example 3.5.9: Ms. A., has a child with albinism, Focus Group 3)

Some participant's partners, however were not interested in attending counselling; men often do not feel comfortable with the concept of "counselling" and discussing their emotions and often seek less emotional help than women (Collins *et al*., 2001; Pelchat, Lefebvre and Perreault. 2003). In focus group four, Ms. P.J., had difficulty trying to convince her partner to attend genetic counselling (Example 3.5.10).

> ***"He doesn't want anything to do with counselling, so for me to have to be told something there, and then come back and tell him that, it wasn't easy for me, you see. Because when I told him again, it's as though that was when the pain would hit me. I always felt that pain. Because they said the child would be like this and that, and you'd find that you couldn't. So I told him that if he couldn't come and listen for himself, I wouldn't be able to come back and relate everything to him. Lucky enough, in his family they understood, it's whereby they assisted and told him everything, then he understood and eventually then accepted, at long last. But the issue of him not wanting to go for counselling stressed me, especially after the baby fell sick, apart from the Down syndrome. They called him for counselling, but then he is sick now. He has high blood pressure etc."***
>
> *"Kwa into ehlangene ne counselling aka funi no kui bona, so for mina ukuthi ma ngi tshelwa into laphayan, ngi phinde futhi ngizomtshela yena, it wasn't easy for me, uyabona. Because, ma ngimtshela yena, manje, ingathi ipain iyangena lakimi. I always feel that pain. Cos umntwana kuthwa uzaba so uzaba so uthole ukuthi angkhoni, uya understanda. So ngamtshela, maube awukhoni ukuzozimamelela, mina ngeke ngikhone ngi zo buya ngizo tshela wena. Lucky enough okwabo bayayazi, uya understand, it's whereby bona basiza bamtshela everything everything wabe uyaiunderstanda ke, wabe uyayi accepta, at long last. But iproblem angayifuni icounselling is because into yakhe I, iyangstressa because akafuni ukuya for icounselling and especially after umtwana egulile ke, ngaphandle kuwe Down syndrome. Bayambiza for icounselling, but then, uyaqala ukugula manje hama high blood ini ini."*
>
> (Example 3.5.10: Ms. P.J., has a child with Down syndrome, Focus Group 4)

As mothers are usually emotionally expressive, they may benefit more than men from support groups as they may experience higher levels of emotional stress, unease and depression with the adaptation to a physically or mentally delayed child (Pelchat, Disson, Ricard *et al*., 1999a;

Pelchat, Ricard, Bouchard *et al*., 1999b). Fathers however, tend to be more cognitively engaged and more concerned about supporting the child financially (Pelchat *et al*., 1999a; Pelchat *et al*., 1999b; Pelchat *et al*., 2003). Ms. P.J.'s (Example 3.5.10) partner may not have seen the relevance of counselling or may not have felt comfortable with the concept of "counselling". Alternatively, the researcher feels that he may have been in denial about his child's condition, and listening to the information about his child's diagnosis may have been difficult for him.

From this study, it was also noted that some participants were no longer in contact with their child's father due to the child's condition. Research has shown that a father's acceptance to his physically or mentally delayed child is more difficult than a mother's (Damrosch and Perry. 1989). This is supported in this study, where Ms J., spoke of how her partner could no longer cope with the child's condition and his frequent illnesses, insisted that they leave the house and has since not supported or contacted Ms. J. (example 3.5.11).

> ***"He wouldn't sleep, he was a restless child, and he would cough a lot. His problem is that he coughs, he's blocked, we have to make him this thing, medication, the bottle until sunrise. Then his father I think was tired of living that life so he chased us away. So we moved out, we left, and now we live in a shack with my sister, outside in a shack. Then after that, he has never given his child any attention, up to today, he can't even look at him when we walk on the streets, which means he goes out telling his neighbours that Sipho* is a disabled child, and of which he is his blood and speaks ill of him. He doesn't even maintain him, and he doesn't even care what is happening in his life. I am raising him myself, but Sipho and I are happy."***
>
> *"bekangakhlai, bekaumtwana o resteless, uya khwehlela kakhulu. Iproblem yake ukuthi uya kwehlela, ublocked, kemele senze ama lokuzana imedication, ibodlele kuzekuse. Then ubabakhe I think bekasa khathele yilelo life leyo so wasixhosha, sahambe samoova out sahamba, futhi manje sihlala kwiskack nosister wami, singphandle kushack. Then, emvakwalokho, vele nje ngathi since from lapho azange kube umuntu ophinde wamnaka up until namhlanje, ohluleka nokumbheka nasestradidini, ene which means ngathi uhamba atshela ama neighbours ache ukuthi uSipho* umtwana wesiqhwala and of which he's his blood but umkhuluma kabi. Akamu maintaini, aka akanandaba nje ukuthi ilife yakhe kwenzakalani. Ngizikhulisela yena ngikwami, but mina no Sipho sihappy."*
> *Child's name has been changed.
>
> (Example 3.5.11: Ms. J., has a child with Down syndrome, Focus Group 3)

The roles of men and women in modern society are constantly changing due to adjustments in social and political views however; traditionally the father is able to have a greater choice with his involvement with the child than the mother (Meyer. 1986; Damrosch and Perry. 1989; Pelchat *et al*., 2003). His attitude towards his child may also have a large influence on whether a child is accepted or rejected by the family (Meyer. 1986; Damrosch and Perry. 1989).

Participants, especially those who had children with recessive genetic disorders like albinism, expressed difficulty in explaining to their partner that both he and his family were also carriers for the condition, not only the mother and her family. This is seen in the example of Ms. K. (Example 3.5.12), who has a child with albinism. She expressed that her partner and his family would not believe that he was a carrier for albinism and that because he and his family said that they did not have any family members with a disability, it was not possible that it came from "their side of the family". This made communication between them very difficult (Example 3.5.12).

> ***"It is typical of them to be surprised about something like this, asking how such an abnormality could occur in the family. They said they don't have such a person in their family. Where does it come from? They ask all sorts of questions, then at the end they were ok."***
>
> (no vernacular- participant spoke in English for this statement)
>
> (Example 3.5.12: Ms. K., has a child with albinism, Focus Group 2)

In some communities, mothers are blamed for causing a child to have a physical or intellectual disability; and if a woman is thought to bring any misfortune to the family, she will be cast out so as to prevent her passing down negative traits to any future children (Barlow-Stewart *et al*., 2006; Sheets, Baty, Vázquez *et al*., 2011). Ms. K., together with other participants, felt that if their partners attended genetic counselling, they would have a better understanding of the cause, clinical features and management of the condition and could assist and support the mothers to convey information to their respective families.

3.5.4 When is the Right Time for Counselling?

In this study, two women, who have children with Down syndrome, were only counselled about their children's condition when their children were eight months and two years old, respectively. They both acknowledged that this was very late for them to find out about the condition and that they had been worried about their child’s slow development. Previous research, performed both nationally and internationally, has shown that parents often recognise that their child is developmentally delayed, causing them to become anxious, but often do not know the cause or name of their child’s condition, and only receive counselling long after their child had been born (Christianson and Kromberg. 1996; Collins *et al*., 2001; Solomon *et al*., 2012). In this study, some mothers who have children with albinism, felt the same way as the two mothers who have children with Down syndrome, and felt that the best time to receive genetic counselling was soon after the birth of their child (Example 3.5.13), (Example 3.5.14).

> ***“But by then I’d accepted, but then the counselling member helped me to understand further, and more. But for me to understand that my child is not like others, the information was late.”***
>
> *“But nna by then ne se amohetse vele okile oa bona, but then counselling le yona eentse ke amohele more and ke tsebe more ka ene okile oa bona.”*
>
> (Example 3.5.13: Ms. K., has a child with albinism, Focus Group 2)

> ***“Yes, rather than finding out and getting scared at a later stage why a child is like this or that. You know at least, that this is the situation.”***
>
> *“Ea ja thusa. Because like ha ne ke tlo tshoga at a later stage gore ok ngoana why ale so you know immediately gore ok this is it.”*
>
> (Example 3.5.14: Ms. T., has a child with albinism, Focus Group 2)

These findings, together with other studies, show that parents of a child born with a genetic condition often have many questions about the cause, recurrence risk and prognosis soon after birth. Research has shown that parents prefer receiving genetic counselling and information about their child's diagnosis either before, or soon after birth (Collins *et al*., 2001; Sheets *et al*., 2011).

Although several participants expressed wanting genetic counselling soon after the birth of their child, it seemed for Ms. P.J. (Example 3.5.15), that having received genetic counselling three days after the birth of her child had been very overwhelming as she was still trying to come to terms with her child's diagnosis of Down syndrome.

> ***"Me, to be honest, at that time, they spoke and spoke about genetic counselling, but then I was just listening because most of the time I was just thinking about how one gets Down syndrome. But they told me how to handle everything to do with Downs. But I was having so many questions you understand, so it was difficult for me.***
>
> ***"Me, at first, like, I didn't even know the name genetic because at the time I was confused, so they had to explain to me what was happening, then after they told me what was happening, then did I understand it, what counselling is about, although I don't even remember what they said about counselling because at the time I was a little bit stressed."***
>
> *"Mina to be honest i genetic counselling nga lesasikhati, bakhulumile bakhulumile, but then I was just listening ne, because most of the time I was just thinking about this thing uya understand ukuthi how to get it. But they told me how to handle everything of Down. But I was having so many questions you understand, so uthole ukthi nakumi it's difficult, no*
>
> *"Mina I felt mina, I didn't even know the name genetics because ngalesa sikathi, I was confused neh. Jah. They had to explain to me ukuthi kuyenzakala ini la, then after they told me. Ukuthi kewnza kalani, I did understand ukuthi icounselling it's about what, although I don't remember bathini nge ndaba ye counselling because nge lesasikhathi I was a little bit stressed."*
>
> (Example: 3.5.15: Ms. P.J., has a child with Down syndrome, Focus Group 4)

Studies have shown that some parents prefer having genetic counselling at a later stage as they are still coming to terms with the child's diagnosis soon after birth and are grieving about the ‘loss’ of what was expected to be a healthy child (Collins *et al*., 2001; Muggli, Collins and Marraffa. 2009). This was shown in another example in this study, where another participant, Ms. S. (Example 3.5.16), had also not found genetic counselling helpful so soon after the birth of her child, as she was grieving and in shock from hearing about the diagnosis in her child.

> ***“Oh my I was crying, I cried as the doctor showed me pictures. I just asked myself what kind of thing is this. He showed me pictures like with children who have like teeth, eyes like this, who you’d look at. This child, these children, you see.”***
>
> *“Yoh beng’khala, beng’khala udoctor mina mayenbonisa iyithombe ngibuze ikhuthi yinto enjani yona le like, wang’khiphela iithombe ke wang’bonisa ukuhti like abantwana aba so like amazinyo, namehlo, nobuso nje mawumbhekile ukuthi yazini. Lomtwana, labantwana aba uyabona.”*
>
> (Example 3.5.16: Ms. S., has a child with Down syndrome, Focus Group 1)

Ms. P.J. and Ms. S. may have benefited from genetic counselling at a later stage when they had had time to grieve and work through their emotions. There are several stages of grief including: 1) shock (especially when an unexpected diagnosis occurs); 2) denial (a form of holding onto hope and not acknowledging their circumstance); 3) physical and physiological disturbances (for example loss of appetite, insomnia, depression, anxiety); 4) guilt; 5) anger (this can be directed at the counsellor, the medical profession, the person/thing that they lost etc); 6) idealisation (thinking about how their situation could have been different); 7) realisation (the point at which the patient recognizes their situation) and lastly 8) acceptance (when an individual is admits to their loss) (Ormerod and Huebner. 1988). The length of time and sequence of these stages, as well reaching the point of acceptance varies greatly between different individuals and it is important for counsellors to respect this (Ormerod and Huebner. 1988). The researcher feels that once acceptance has been reached, a parent/patient may be more receptive towards receiving information and may gain greater benefit from a counselling session. The timing of receiving genetic counselling thus needs to be considered. Genetic counsellors need to try and be more flexible in accommodating patients with preferences to the timing of receiving genetic counselling. Realistically however, practicing this approach in Genetic Clinics in the state healthcare system may prove to be more challenging. Genetic Clinics not only rely on referrals from other healthcare services (thus cannot anticipate the timing between a patient/parent receiving a diagnosis and receiving genetic counselling); but currently have a high patient volume with a shortage of staff, thus may not be able to reschedule patient bookings at their preference.

3.5.5 Support as a "life-line"

The need for ongoing support was emphasised by the participants. They expressed that support was needed not only from genetic counselling, but also from other families who have children with a genetic condition as well as other community services. Two participants, who have children with Down syndrome, expressed that after receiving genetic counselling, one has to go out and "face the world" on one's own (Example 3.5.17).

> Ms. H.:***"They shouldn't let us go"***
>
> *"Bengasiyeki."*
>
> Ms. P.:***"Like with me, I would see them before I go home to the township where you'd meet up with people because they start talking and it hurts, so this can only be removed by counselling. Where are you going to get it again?"***
>
> *"Like mina ngibabone mina before ngiye khaya eloctioni, la uhlangana khona nabantu, bayakhuluma ufeela kanjani? Leyonto leyo izosuswa yini? Icounselling? Ozodibana kuphi nayo futhi?"*
>
> (Example 3.5.17: Ms. H. and Ms. P., both have children with Down syndrome, Focus Group 1)

Both mothers said that new questions arise about the child's development and people in the community sometimes make negative remarks about the child. They believed that these particular medical questions and emotional concerns could only be answered and alleviated at a genetic counselling session. The diagnosis of a genetic condition in a newborn or infant is a very traumatic experience for a parent and the amount of support that they receive plays an important role on their ability to cope with their child's genetic condition as well as for the welfare of the child and family (Muggli *et al*., 2009). Parents and families of affected children often do not obtain the support needed, and parents have expressed that ongoing support is essential for continued advice, guidance and care (Muggli *et al*., 2009; McGrath, Stransky, Cooley *et al*., 2011).

Several of the participants who have children with Down syndrome appreciated the support they got from other health professionals, particularly the physiotherapists and occupational therapists and were grateful for the referral to support groups as they enjoyed meeting with other mothers and families (Example 3.5.18).

> ***"…so that is why I say that the support group has also helped a lot, you meet up with other mothers, one tell you that: 'Mine has a problem like this,' so they teach me like this. So you do these things to your children so that they can be ok."***
>
> *"... so that is why ngithi isupport group nayo isize kakhulu, angithi niya hlangana lapho nigabomama, omunye akutshele ukuthi: "owami benekinga eso ngamyenza so." bang' fundisa kanje' nawe so nyayiyenza lento kumtwana kho then umtwana umbona aza right."*
>
> (Example 3.5.18: Ms. H., has a child with Down syndrome, Focus Group 1)

The need for support from each other was also evident after the focus group sessions. Most participants stayed at the venue after the session had ended. They exchanged phone numbers, spoke about their children's management and showed each other photographs of their children.

The need for greater support was also noted when participants spoke about their feelings of disappointment and the sometimes "false hope" created by genetic counsellors when they invited patients to contact them after the initial consultation. Several participants felt that although genetic counsellors gave them their office contact numbers, they could not be reached (Example 3.5.19).

> ***"Just as I bumped into Amy* now, I told her that I have been looking for her and can't reach her over the phone. Like everyone that we get here is always busy, it not even during the morning (that I phone)."***
>
> *"Njengoba ngihlanga noAmy manje ngiyamtshela ukuthi kunini ngingak'tholi bantu ngik'fonela. Like bonke abantu esibatholayo la uhlala a busy akekho nasek'seni."* *Genetic Counsellor's name has been changed.
>
> (Example 3.5.19: Ms. S., has a child with Down syndrome, Focus Group 1)

Participants felt that genetic counsellors should provide their cell phone numbers so that they contact them directly and wanted to be able to come to the genetic counselling offices if they needed to. This was expressed particularly by a participant, Ms. E., who had been frustrated by not being able to contact the genetic counsellor that she had seen, despite attempting to contact her on several occasions (Example 3.5.20).

> ***"You see if they give us their cell numbers straight, it's easier for me because you would sometimes hear them say: "Hold on", or they are not available or something you see."***
>
> *"Yabona, mebes'nika straight icellphone number, it's easier for me because uzozwa sebethi: "Hold on" or akhekho or kwathini yabona."*
>
> (Example 3.5.20: Ms. E., has a child with Down syndrome, Focus Group 1)

Ms. E. also said that she did not know the physical address of the genetic counselling offices, as she would have wanted to go see the counsellor directly. These comments showed their need for ongoing support and gave a sense of the participants feeling 'let down' by the assurance of support made to them by the genetic counsellor/geneticist. For individuals seen for genetic counselling, some may have high expectations since the word 'counselling' is associated with psychological support and they often expect to receive further emotional support as well as information on how to cope with the condition (Collins *et al*., 2001; Davey *et al*., 2005). In this study, many viewed genetic counselling as a "life-line" that needed to be readily available and some felt that as the child was growing up, they would have different needs and concerns that may need to be addressed, and follow-up genetic counselling sessions and support seemed important to them (Example 3.5.21).

> ***"The first session is important but there needs to be a second one. Like I said before, I got the first session in the hospital, then I went out and faced some challenges out there and some are very disappointing. Now how do I deal with them? So a second session is important after you've faced the world."***
>
> *"Le session yok'qala iilo, ngicabang 'ukuthi iright but fanele kube neyes'bili because like as I said, bekukok'qala ngidibana nayo ngeses'bheldela, ngayithola la es'bhedlela, amachallenges eng'wathole ngaphandla futhi, ene mhlaumbe ayangidisappointa and then ngiwawipa kuphi? And then ngifisa nje kube khona eyes'bili ke after usuphumele ngaphandla wadipan nezinto eyithile."*
>
> (Example 3.5.21: Ms. P., has a child with Down syndrome, Focus Group 1)

Participants in this study, who have children with Down syndrome seemed to want ongoing genetic counselling support. This finding is similar to other research, which noted that parents who have children with Down syndrome often show higher levels of unmet support needs than parents of children with other special healthcare needs (McGrath *et al*., 2011). Mothers, who had children with albinism and haemophilia, had felt that one genetic counselling session was adequate (Example 3.5.22).

> Ms. T.:***"Once, only one time. I think once was enough for me, because the information I got was enough, it was a long session of almost an hour. I think I made notes, I would have made my own way in order to know. I think that one session was helpful."***
>
> *"Kei le hai once, only one time. I think once was enough for me because information eke kereileng it was long, almost an hour. I think if I needed to anything more lenna I'd make my own way hore keitse so I think one session ene ele enough hore ketsebe."*
>
> Ms. K.:***"Too much of something is not good."***
>
> (Example 3.5.22: Ms. T. and Ms. K., both have children with albinism, Focus Group 2)

This observation differs to previous research which found that families and caregivers of individuals affected by albinism and haemophila had expressed the benefit of support groups (Lund and Gaigher. 2002; Solomon *et al*., 2012).

The findings from this research show that greater efforts for patient and family support needs to be considered in genetic counselling services, especially for mothers of children with Down syndrome. Several proposals have been made for improving health professionals' attitudes towards parents of a child who has been recently diagnosed with a genetic condition. These include attitudes and skills such as greater empathy, encouragement, offering ongoing support (both personally and referring to other support groups) and reassurance (Wasserman, Inui, Barriatua *et al*., 1984; Muggli *et al*., 2009). These approaches may lessen the traumatic experience for parents and may increase their levels of satisfaction and adaptation to the condition (Muggli *et al*., 2009). The feasibility of providing ongoing support in genetic counselling services needs to be considered, as financial constraints, genetic counselling staff shortages and an overload of patient numbers limits the ability of providing ongoing support for all individuals. Approaches that do not require radical interventions however must be considered in order to improve genetic counselling services and to provide or facilitate support to parents and families in need.

3.5.6 Transport Difficulty and Access to Services

Some participants in this study mentioned having transport and financial difficulties in reaching healthcare services, particularly at the larger state hospitals. Not all healthcare facilities such as clinics and smaller hospitals have all necessary equipment, services and specialist healthcare professionals (Coovadia *et al*., 2009). Patients may not have a healthcare facility near their home and if indicated, patients may be referred to a larger healthcare facility which may be further away. This makes access a barrier for many South African patients, due to geographic location or expensive travelling costs (Harris *et al*., 2011). In addition, very few patients within the state healthcare system use private transport, with most either using public transport or walking to their destinations (Harris *et al*., 2011). This difficulty of access to services was expressed by two participants in the study, Ms. T., and Ms. K., (Example 3.5.23).

> Ms. T.: ***"Some people are scared to be referred to far off places."***
>
> *"Akere batho babang natsoafa gore ba lo go referela gole koa."*
>
> Ms. K.: ***"Like myself, I go to Bara, Coronation and Helen Joseph, but someone from the township will tell you that they have to use transport twice to get there and to come back. They'll tell you things like that."***
>
> *"Like nna, nna ha ke tsamae ko Bara. Ke tsamaya ko Coronation le ko Helen Joseph, and so motho otsoang lokshen otloo joetsa hore transport ke tlo e palama gai two go guthla. Ba go joetsa dintho tse joalo."*
>
> (Example 3.5.23: Ms. T. and Ms. K., have children with albinism, Focus Group 2)

Frequent hospital visits are often necessary for individuals who have genetic conditions as they require ongoing management. This places stress on parents and care-takers of affected children, especially if these patients and their families have to travel long distances to healthcare facilities. In this study, this issue was emphasised particularly by mothers who had children with Down syndrome as they said it was difficult for them to attend all the appointments on a monthly basis (Example 3.5.24).

> ***"Well you know for me, another thing is that Johannesburg (Hospital) is too far for me. Especially because I have many appointments for the child. On one day it's speech therapy, on another day I go to OT, on another day it's for something else. If only they were nearer."***
>
> *"Mina uyabona enye into, eJohannesburg (Hospital) it's too far for me. Especially, ngina ma appointment amaningi for umtwana, nge linye ilanga ngiya for ispeech therapy, nge linye ilanga for OT, ngelinye ilanga ngiya for in-in. Ingathi if bezi la eduzana."*
>
> (Example 3.5.24: Ms. P.J., has a child with Down syndrome, Focus Group 4)

To alleviate some of these travelling and financial stresses, suggestions were made by the participants to make genetic counselling services available at local clinics (Example 3.5.25).

> ***"That's why this should be available at clinics nearby. I think they must do things like this nearby, in clinics, locally because people are lazy, and will think that Helen Joseph is far, but if it's a local clinic, the person catches local transport and that's it."***
>
> *"That's why kere dintho tse so tshontso dibe available ko di kliniking, mo rekgonang because di closer to everything, because still otlo joetsa gore tsamaisa ngoana. Le wena oye for counselling eng eng otlo bona Helen Joseph e ko hole. So at least ha le mo klinking oa tseba hore, okhomba local, go fella mo."*
> (Example 3.5.25: Ms. T., has a child with albinism, Focus Group 2)

3.6 Theme 3: Personal Beliefs

3.6.1 Religion

Religion is very important in many people's lives and it can influence their personal beliefs on the cause of a genetic condition, and how they make meaning of it. Religion can be described as a person's belief in something more powerful than themselves and it often bestows a positive outlook on life (Musgrave, Allen and Allen. 2002). In South Africa there are several practiced religions including Christianity, sectarian Christian Independent churches (e.g. Zionists), Islamism, Hinduism and Judaism (Statistics South Africa. 2001; Ross. 2007). In the South African Black communities, most individuals belong to different independent Christian churches, however there remains a strong belief in the influence of ancestors and other "spirits" (Du Toit. 1980).

Religious faith has different meanings for people; either by providing answers for their purpose in life, giving an explanation as to why people suffer, by facilitating in coping mechanisms, by providing guidance on what is morally right and wrong, and by forming a basis for personal peace, hope and love (White. 2009). Religion was a frequent topic of conversation during the focus groups. Some participants believed that they must accept their child regardless of the condition as he/she is a "gift from God". Christianity and Buddism practice the notion that one must be accepting, understanding and realise that one cannot change a situation as it is the will of God (Eisenbruch *et al.*, 2004; Black *et al.*, 2009). In Example 3.6.1 one participant thought that she was being "tested" on how strong her faith was and it was "wrong" of her to question God's reason for "giving" her a child with Down syndrome.

> ***"I was asking myself, why is Themba* like this, when Tsepo* (her older child) is alright? But then I accepted that God doesn't give us all things the same. He did things his way, and I should accept it. Maybe he even wants to see my faith, how much I accept this."***
>
> *"Lenna ne ke e potsa amd then (her child) yena why ale so mos (her older child) yean o right? Mara ka gcina ke amohela. Ok Modimo harefe ka hotshoana, mohlomong mo oentse kamogo atsang, so tlameile ke amohele. Maybe oleka le teko yaka obatla hobona hore ke amohela hakakang."**Children's names have been changed.
>
> (Example 3.6.1: Ms. H., has a child with Down syndrome, Focus Group 2)

Parents of an affected child, may either believe that the condition is a punishment from God for their past sins, or that they are being tested as they are considered 'weak' before God, and may consequently feel that that their experience with the child will promote individual growth and change (Weil. 1991; White. 2009).

In this study, prayer was also considered important, as was attending church and keeping a positive outlook (Example 3.6.2).

> ***"You know, I pray for my child even when I'm just sitting. I just clothe him with the blood of Jesus. I always pray and I love my child."***
>
> *"Umkulekele umtwana wakho ngoba mina owami ngiyamkhulekela, mang; hleli nje ngiyamqcobisa nge gazi lika Jesu… ngiyamkhulekela… yazi ngiyakhuleka ngiyasho kuNkulunkulu and nguyamuamukela and ngiyam'thanda."*
>
> (Example 3.6.2: Ms. P., has a child with
> Down syndrome, Focus Group 1)

Other researchers have shown similar findings; that prayer to God to provide a miracle, spiritual guidance and emotional support will strongly influence the outcome of a situation (Weil. 1991; Ahmed, Atkin, Hewison *et al*., 2006; White. 2009). Another participant, Ms. P. (Example 3.6.3), said that she constantly prayed for her child and his condition and believed that prayer would help her and her child through their difficult situation.

> ***"The thing is I'm always busy and if I'm not busy, I go to church and I thought that if I go to church and pray, that will help me because that is where I meet God, because that is where I'll get my help you see."***
>
> *"Into ukuthi I'm always busy and if I'm not busy, I go to church and angithi uyabona nayo ichurch nakhona idibana name ngithi yabona if I pray yabona ngizosizakalala."*
>
> (Example 3.6.3: Ms. P., has a child with
> Down syndrome, Focus Group 1)

Ms. P. (Example 3.6.3) trusted wholly in her faith in God and was convinced that He would cure her child from Down syndrome and that her son would one day attend a mainstream school and lead a normal, independent life. These beliefs have shown to reduce stress levels and allow some mothers to cope better with their child's diagnosis, as has been demonstrated in previous research by Black *et al*., (2009).

In this study, prayer through "spiritual healers" was also thought to be effective as one participant described an encounter with another woman in a taxi who asked if she could hold the child and pray over him as she could see that he was sick (Example 3.6.4).

> ***"One day (in the taxi) I was sitting with another man and some lady who seems to be in touch with her spiritual self heard my child breathing heavily, showing that she couldn't restrain herself. I kept looking at her concentrating on my child, then she asked the man to swap places with her and she said that my baby has a problem, why is he breathing heavily and said that she'd like to hold my baby and she prayed for him. She said that I should dedicate him to the Lord and he will be fine and she sees the way he is breathing and it is affecting her. So I said: 'No Lady, I always ask God to take care of my baby.'"***
>
> *"Nge linye elanga benghleli omunye ubuthi wathi, losisi ngathi unomoya wezwa umtwana ephefumlela phezulu abonise ukuthi akasa khone, ngibathe ngyambheka seconcentrate kolomtwana waze wa cela komunye ubuthi ukuthi ba swap wagcina ahlale pa wathi umtwanakho une problem why a phefumula kanje yabo, wathi ngicela ukumthatha wathandaza wathi sisi always ubomnika kuNkulunkulu, uzoba right washo wathi ubona nga the way ephefumula ngakhona ne lento yakhe iaffecta yena. So ngathi: "Hayi sisi kuNkulunkulu ngiyamcelela everyday.""*
>
> (Example 3.6.4: Ms. E., has a child with
> Down syndrome, Focus Group 1)

Research has shown that some members in communities are thought to be spiritual healers and it is believed that they have the ability to cure an individual, change one's risks and to bring about good luck (Musgrave *et al*., 2002; Eisenbruch *et al*., 2004; White. 2009). Other South African studies have also shown that many individuals believe that having a disabled child is part of God's will and that prayer and laying one's hands on an affected individual is a powerful form of healing (Du Toit. 1980; Penn *et al*., 2010).

Termination of pregnancy is another sensitive topic with regard to religion. One participant, Ms. P.J. (Example 3.6.5), stated that it was better not to know about a child's genetic condition whilst still pregnant, as having a termination of pregnancy was "the wrong thing to do". This comment was not probed further by the facilitator, thus one cannot know whether Ms. P.J. felt that way because of her religious beliefs, or for other reasons.

> Facilitator:
> ***"But they can check then. Just like you have a baby with albinism, so you checked for albinism, so now you can check for Down."***
>
> Ms. P.J.:
> ***"But you are already pregnant! That means that they are going to tell you that you are going to have a Down, if you don't want the Down again, you have to make an abortion."***
>
> Facilitator:
> **"*Ya, it's your choice."***
>
> Ms. P.J.:
> ***"And it's not the right one."***
>
> (Example 3.6.5: Ms. P.J., has a child with Down syndrome, Focus Group 4)

Several religions, including Christianity prohibit termination of pregnancy and individuals believe that God will punish them if they abort a fetus, and they should rather trust that God will provide for them and their child (Ahmed *et al*., 2006; White. 2009). A South African study looking at pregnant patients' decisions after receiving genetic counselling for fetal abnormalities, showed that the majority of patients chose not to have a termination of pregnancy and that many of them opted to continue with the pregnancy, citing religious reasons and the belief in accepting one's fate (Todd. 2007). It is important for genetic counsellors to be sensitive to these religious issues when dealing with patients.

Overall, the participants in this study had and felt strong religious convictions. They felt that one must accept one's fate and be grateful for whatever type of child one has. This study, together with other research, shows that many parents and individuals have strong beliefs in fate and religion, and that it assists them in coping and understanding a genetic condition (Eisenbruch *et al*., 2004; Black *et al*., 2009; White. 2009). It is important for genetic counsellors/geneticists to be aware and sensitive to these beliefs.

3.6.2 Traditions

This topic was least mentioned during the focus group discussions, yet the researcher feels it is still important to address. In South Africa, there are several different cultural and ethnic groups including Zulu, Xhosa, Sotho, Tswana, Caucasian, Mixed Ancestry, Indian and Asian (Todd *et al*., 2010; Statistics South Africa. 2011). From the literature review, and given South Africa's rich multicultural heritage, the researcher anticipated that culture, traditions and myths would be key topics, yet they were only briefly mentioned, and when they were, the facilitator did not probe further on these topics. Despite this, a number of important factors can be gained from the focus groups in this study regarding tradition and culture.

Community elders play an important part in South African Black culture (Kasanga and Lwanga-Luma. 2007). The respect for elders' knowledge, particularly those of grandmothers, was communicated by Ms. E. (Example 3.6.6), who believed that the grandmothers in her community could assist children like those with Down syndrome who have an inability to speak. She implied that grandmothers are the "advice givers" in the community and are involved in decision making concerning a child's health. Ms. E. spoke about how parents who have children with speech impairments, approach grandmothers in their community to assist the child's healing. One grandmother had reportedly used a razor blade to make a cut underneath a child's tongue in order to "loosen" it and to promote speech. Ms. E. felt that doctors used the same approach to speech impairments as the grandmothers

> ***"Yes, even at home with our grandmothers they'd make a cut under the tongue. So that's why I sometimes see that kids with Downs are helped by doctors as they find it hard to protrude the tongue and they help them by making a cut under the tongue. Even back at home, there was a child who was unable to speak and a gogo took a razor blade and cut it underneath and now the child can speak. I see that with Downs kids have it and it is inhibiting them from speaking."***
>
> *"Mmm, because imambile uyabona mas'khula ekhaya ogogo ekhaya vele ekhaya kuye kwaba nomtwana onjalo, so ugogo wak'yenza ke loko kwak'ngakhulunya yabo. So this thing le ibambile so that's why mang'bona amaDowns eba, that's why abo doctor, they cut this thing embambile, that's why ngibona kubenzima ukuthi bethi out tongue. Mina ngabona ekhaya kukhona ek'seni uyam'khamula ayithi kha! From there wakhuluma wayengazi from there wakhuluma. So ngiyabona eyinye yezinto zama Downs ebanjwayo as long as uyithi lento uyisika ubambile."*
>
> (Example 3.6.6: Ms. E., has a child with
> Down syndrome, Focus Group 1)

Respect for elders has previously been documented and researchers have suggested the importance of involving them in the dissemination of healthcare information as they have the ability to connect healthcare professionals with individuals from the community (Saleh *et al*., 2009; Kinney *et al*., 2010). This is particularly viable in the local context since grandmothers are frequently involved in the primary care of their grandchildren.

Another topic that arose in this study was the belief in traditional medicine. Traditional healing is a parallel health care system widely used in South Africa, and although some of the approaches vary amongst ethnic groups, the principal remains widely the same (Kale. 1995;

Penn *et al*., 2010). Individuals in African communities will consult with a traditional healer for various reasons including finding answers for the cause of a disease, for guidance on how to treat a condition and to perform needed rituals (Penn *et al*., 2010). This can be seen in this study, where one woman's partner sought guidance through a traditional healer to find out what had caused their child to have albinism (Example 3.6.7).

> ***"Yes, they should explain to him, that's what I mean that sometimes someone may believe in traditional healing, but when someone who seems like they have more information, better than they can listen."***
>
> *"Eya, bem explainele, that's what I mean ukuthi sometimes omunye umuntu abe believa emasikweni, but when they get someone ongathi une more information, better than yena they can listen."*
>
> (Example 3.6.7: Ms. A., has a child with albinism, Focus Group 3)

In some Black communities, beliefs are held that disease may be caused by the influence of ancestors and spirits, and faith is often put into the wisdom and guidance of community elders and the traditional healing system, as they are able to provide explanations to the influence of the ancestors and causes for disease (Kale. 1995; Kasanga and Lwanga-Luma. 2007; Penn *et al*., 2010). Beliefs around the cause of a condition may conflict with Western medical and genetic explanations received at medical facilities and can lead to misunderstanding due to belief in alternative myths and community explanations (Eisenbruch *et al*., 2004; Klitzman. 2010). Several Black South African ethnic groups make use of this health care system and will either consult with an inyanga (a herbalist who will make use of natural herbs and animal products to cure disease); a sangoma (a diviner who has the ability to consult with the ancestors and spirits) or a umthandazi (a type of faith healer who is part of an independent African Christian church and has the ability to heal by prayer and touch) (Kale. 1995).

Genetic counsellors need to be aware of potential misunderstandings that may arise and need to find ways in which western medicine and traditional healing approaches can work in tandem so as to benefit the care of a shared patient.

In some communities, it is believed that women are to blame if any misfortune (such as illness) is brought upon a family (Barlow-Stewart *et al.*, 2006; Penn *et al.*, 2010). It seemed that it was especially difficult for participants in this study to convey genetic information to their child's father and his family. One participant in this study was relieved to find out, that on her husband's side of the family, there is an intellectually and physically delayed child. She implied that if this had not happened, she would be blamed by her husband's family for causing her child's condition (Example 3.6.8).

> ***"Do you know how it became easy for me? When I found out that there is such a child from the father's side of the family. Do you understand? Then at least I was relieved."***
>
> *"Yazi kwa ba easy kanjani kimi? When I find out ukuthi kulo baba we ye khona umtwana oso. Uya understand? Then at least nga thola irelief."*
>
> (Example: 3.6.8: Ms. P.J., has a child with Down syndrome, Focus Group 4)

Mothers' anxiety has been previously described in both international and South African studies where they have said that their partners' families blamed them for the cause of the condition, saying that such a condition had never previously occurred in their family and as a result, they are mistreated by their husband's family (Collins *et al.*, 2001; Baker, Lund, Nyathi *et al.*, 2010). Previous South African studies have reported that Black women are often blamed by the father if the child is born with a problem, and may deny or abandon the woman and child to avoid the responsibility of having to look after the child (Chalmers. 1987; Richter and Morrell, 2006). Women are thought to cause disability since they carry “pollution in their womb” and that they contribute “bad” things to a child, whilst a man only contributes positive qualities (Penn *et al*., 2010 pp 16). However, other studies have found that in other communities, men are often blamed for causing misfortune to their children (Barratt and Penn., 2009). It is important to be aware of personal and traditional beliefs regarding gender within communities when addressing issues of disability in a genetic counselling consultation.

3.6.3 Language Barriers

Language issues were also mentioned in the focus groups and participants said that healthcare staff often could not converse with participants in their home language, and a second healthcare professional (often a nurse) or hospital staff member would have to translate and explain the information to them. Language barriers in healthcare and genetic counselling communication have been reported globally (Saleh *et al*., 2009; De Maesschalk *et al*., 2011; Meilleur *et al*., 2011). In some countries, it has been shown that language barriers lead to poorer health outcomes and patients who are not fluent in European languages are unable to express their emotions, leading to greater dissatisfaction and a decreased understanding of the patient’s needs by healthcare professionals (Saleh *et al*., 2009; De Maesschalk *et al*., 2011). This can be seen in this study, where one participant, Ms. P.K. (Example 3.6.9), described her anxiety and difficulty in trying to communicate with the doctors at the hospital when she first learnt about her child’s diagnosis of Down syndrome.

> ***"I felt like I could cry so much and exclaim: "If the doctors tried to explain to me but they didn't understand how it begins. This then caused them to look for a nurse for me, a nurse who will be able to explain things to me in my home language, who had to sit me down and explain to me how it happens that he gets Down syndrome. How it happens: one, two, three, one, two, three. Then I ended up accepting that my child has Down syndrome."***
>
> *"Bekuthi ngikhale ngithi wema, bazama ukuthi bangichazele odokotela kodwa angzange ngi understand ukuthi iza knajani, iqala kanjani. Kwase ku causa ukuthi bang'funele ama sister, osister abakwazi ukungi explainela ngesikithi ukuthi ngihlale phanse nabo bang'chazele ukuthi kwenzeka ka so ukuze athole iDown syndrome kuhambe kanje: one, two, three, one, two three. Ngagcina seng'kwamukela ke ukuthi umtwanami uDown syndrome"*
>
> (Example 3.6.9: Ms. P.K., has a child with Down syndrome, Focus Group

Language barriers are a common issue in South Africa since there are many different languages spoken; the eleven official languages, as well as languages of immigrants from other African countries (Statistics South Africa. 2001; Harris. 2002). This linguistic diversity makes communication barriers inevitable, particularly in healthcare communication, shown in several South African studies (Levin. 2006; Schlemmer and Mash. 2006; Penn. 2007). Recommendations for the training and utilisation of professional interpreters in South African state hospitals, as well as basic medical training for interpreters have been made (Schlemmer and Mash. 2006). It is thought that this service will not only improve communication, but will also enhance understanding of other cultures and improve satisfaction levels for both the healthcare professional and the patient (Schlemmer and Mash. 2006; Penn. 2007). Despite this, little has been implemented to ensure the availability of interpreters.

Although it has been recognised that interpretation services improve the quality of healthcare provision and further recommendations for funding of these services have been made, some patients have expressed their concern of having a third party member present in their medical consultations because of a breach of patient privacy (Saleh *et al*., 2009; Maesschalk *et al*., 2011). It is important for genetic counsellors to be respectful and aware of individuals' preferences when suggesting the use of an interpreter.

3.7 Theme 4: Addressing the Community and the Family

3.7.1 Community's Views

Most participants expressed that the community had very little, or no knowledge of genetic conditions or genetic counselling (Example 3.7.1).

> ***"….. I took him to church and told the Pastor and he asked me what this condition is called, and I said no they say the child has Down syndrome and he asked: "What is that?" He didn't know."***
>
> *"….Ngamisa esontweni ngabatshela ke umfundisi wang'buza ukuthi ibizwani le, ngathi hayi lomtwana bathi une Down syndrome, wathi: "Yini leyo?" akayazi."*
>
> (Example 3.7.1: Ms. P., has a child with Down syndrome, Focus Group 1)

Another participant, Ms. T. (Example 3.7.2), said that unless an individual, or one of their family members had been to genetic counselling, people in the community would not have heard of it before and genetic conditions were generally not well understood or recognised.

> ***"It's not popular. You wouldn't get to find out about genetic unless you were in that situation."***
>
> *"Ha e popular waitse. You don't get to find out about genetic if ole ko spetlele and when the situation ya yona."*
>
> (Example 3.7.2: Ms. T., has a child with albinism, Focus Group 2)

These participants, as well as several other participants in this study, raised the need for greater efforts towards community awareness of genetic conditions and genetic counselling services.

There were also mixed feelings amongst the participants regarding the community's perception of an individual who looks different from other people, or is intellectually and physically disabled. This has been shown in other studies where community perceptions of children who have a genetic condition are different, and although family members tend to be more welcoming and accepting of an affected child, community reactions are varied (Greeson *et al*., 2001; Baker *et al*., 2010).

Some participants felt that the community was very accepting of their children (Example 3.7.3).

> ***"Well, wherever I go, they receive my child gladly."***
>
> *"Na haai, mo ke tsamayang ba mo thabela."*
>
> (Example 3.7.3: Ms H., has a child with
> Down syndrome, Focus Group 2)

Parents in other studies have expressed that their communities readily accept, respect and support their children and that they do not focus on their condition or disability, but rather on their abilities and try to treat them as normal citizens of society (Greeson *et al*., 2001; Sheets *et al*., 2011). This view was held by some of the participants in this study who reported that they felt comfortable introducing their child to others, and some of the participants who had children with Down syndrome said that they were not concerned about their child's ability to function in society as they have seen well adjusted disabled individuals in the community (Example 3.7.4).

> ***"But I just wasn't able to find schools for him that will help him develop as there are many people out there who are disabled and work well, they write, they know how to talk, they are ok. There are doctors who are disabled. It is just important that you do not hide your child."***
>
> *"Uyabona like labanu aba bayasebenza, bayabhala, bayakwazi, ukukhuluma is"lungu ene bayakhuluma ba right ba right. Kukhona abo doctor abakhubazekile ungaya yonk'indwawo le. Kuya ngokuthi umtwanakho azang'umfihleumgamfihli ungamfihli."*
>
> (Example 3.7.4: Ms. S., has a child with Down syndrome, Focus Group 1)

Although this seemed to create reassurance for some participants, the disabilities seen in the community were short stature and limb defects, not Down syndrome. The researcher feels that Ms. S., together with some of the other participants may have been in denial, or did not fully understand the implications of the intellectual and physical delay in their own children or how it would affect them in the future.

Some of the mothers in the focus groups felt differently compared to the mothers who willingly introduced their children to members of the community. These mothers had not disclosed that their child had a genetic condition to their neighbours or to other individuals. Many individuals in communities do not know about genetic conditions and their cause; there are misperceptions that a condition may be contagious, that Black individuals with albinism are Caucasians, or that a condition is caused because of cultural or ritual transgressions (Sankar *et al*., 2006; Ross. 2007; Baker *et al*., 2010; Solomon *et al*., 2012). Participants said that some members of the community spoke negatively about individuals who had problems or looked different (Example 3.7.5).

> ***"It's just that I understand sis *Bongi's situation because some people, you know sis, discourage people and speak foul words."***
>
> *"Ukuthi uyabona ngiyayi understand isituation ka sis *Bongi, abanye yazi sis, they discourage you bakhuluma kabi"* *Participant's name has been changed
>
> (Example 3.7.5: Ms. T., has a child with albinism, Focus Group 2)

The participants who were more cautious about negative community perceptions, said that if they were asked about their child's condition, they would either briefly mention the child's diagnosis or they would avoid talking about the condition (Example 3.7.6).

> ***"I don't have any problem because my child goes into the streets and he does his hand gestures, then they ask me why he is like that, then I just say: "He's just like that.""***
>
> *"Mina anginankina….mina umtwanami udlala nge walking wiel njengoba ngisho aloba ayenza lezo zandla zakhe, babuze ukuthi why enza kanje ngithi unjalo."*
>
> (Example 3.7.6: Ms. E., has a child with Down syndrome, Focus Group 1)

One participant said that although her neighbours knew that her child was "different", they had never asked her what his diagnosis was, and she thought that they either felt uncomfortable addressing the topic with her, or that they didn't know what the condition was (Example 3.7.7).

> ***“Many people don’t know Down syndrome and the families, the family seem as though they are scared, when they look at the fact that…”***
>
> *“Many people hey don’t know Down syndrome and di family, family le yona okare ya tshoga, heke elebelletse gore.”*
>
> (Example 3.7.7: Ms. M., has a child with Down syndrome, Focus Group 4)

Research around Black South African women living in Cape Town, showed that the women did not like to discuss their child’s genetic condition with their neighbours, and felt that people living in the city should focus on their own problems and that they should not inquire about other individuals’ personal issues (Solomon *et al*., 2012). Similar views and experiences were expressed by some participants of the focus groups in this study. One participant, Ms. P.K. (Example 3.7.8), said that many individuals in the community are afraid or ashamed of individuals with disabilities. She spoke of stories that she had heard of families hiding their children in their homes, of mothers poisoning them and fathers ending the relationship with the mother, because the child had a disability.

> ***“I told myself that: “No I have to be strong,” so that I am able to tell other people, even those who wish to end up as some of our other children who hide themselves in the house. At some other point you end up giving the child poison to drink, killing him because they are in the situation they are in. Or you are afraid that other people or the father of the baby will dump you, or the child’s father doesn’t really understand.”***
>
> *“Ukuthi ngitshele nabanye abantu, nofisayo ngoba kuendapha vele ezinye izingane zethu sizifihla sizivalela ezindlini. Ngesinye isikathi ugcina ngane uyiphusiza uschefu, uthi uyayibulala ngoba ekulesimo ekuyiso. Or osaba ukuthi abantu noma uyise wengane uzoku dumpa, noma ubona ubab waomtwana omunye akazwisisi akuunderstandi.”*
>
> (Example 3.7.8: Ms. P.K., has a child with Down syndrome, Focus Group 3)

Research has also shown that communities may react negatively towards individuals who appear physically or intellectually different from others. Parents may be ashamed of their child, as has been shown in studies on parents who have children with Down syndrome, haemophilia and albinism (Baker *et al*., 2010; Sheets *et al*., 2011; Solomon *et al*., 2012). There are reported stories of parents hiding their children in their homes and keeping them out of view because of feared perceptions from individuals in the community (Baker *et al*., 2010; Sheets *et al*., 2011).

Overall, these disclosures in the focus groups indicate a poor understanding in the community on the causes of genetic conditions and that disability is a frightening concept to many individuals. Although the participants seemed willing to discuss genetic counselling and their child's condition with their families, most participants were more cautious about telling members of the community about their child's condition; fearful of the type of reaction they would receive. This perception may lead to decreased social support for families with affected children and parents often feel isolated in their situation (Solomon *et al*., 2012).

3.7.2 Telling the Family

Family communication is an important aspect to consider in genetic counselling (Peterson. 2005). Communication between family members not only assists in identifying at risk individuals, but may also assist an individual to adapt to their/their child's genetic condition (Weil. 2000). Most participants in this study acknowledged that their families were very supportive and openly accepted and loved their child (Example 3.7.9).

> ***"For me, at home they absolutely adore my child! Like it's like they've taken my child from me."***
>
> *"Ah mina ekhaya bayamthanda! Like bang'thathela umtwanami yoh! Yazi bangthathele umtwanami kithi… like ngilalae umtwanamo angekho."*
>
> (Example 3.7.9: Ms. E., has a child with Down syndrome, Focus Group 1)

This assisted parents in coping with the situation and acceptance of the condition, and studies have shown that disclosing information about a genetic condition to family members is easier than telling community members due to anticipated negative social reactions (Plantinga *et al*., 2003; Sankar *et al.,* 2006). Research has also shown that although genetic counsellors usually rely on the counselled individual to convey genetic information and risk to their families, many patients would prefer a healthcare professional to give this information, and want genetic counsellors to assist them by identifying and counselling family leaders who can help in conveying information (Peterson. 2005; Kinney *et al*., 2010). The value of this is shown in this study where Ms. T. appreciated that her mother conveyed information to her family on her behalf (Example 3.7.10).

> ***"I was fortunate because that same night my mother told everyone that we were ok, the child has albinism. So we returned home, the family was there, cousins welcomed him. They could see he was an albino and I didn't have to explain anything."***
>
> *"Nna ne ke le fortunate because the same night I could just tell everybody, mama ka aba bollela hore ngoana oso so, ba right and everything. But lenna ha ke kgutla, he was fine, family ene le teng, my cousins, ba mo thabela. They could see vele hore he was albino there was no need hore kenne ke explaine."*
>
> (Example 3.7.10: Ms. T., has a child with albinism, Focus Group 2)

Ms. T.'s mother had accompanied her to the genetic counselling session, and after hearing the genetic information, she assisted her daughter by telling their family members about the child’s condition when they arrived home. Ms. T. felt that her mother took control of the situation. She supported Ms. T emotionally, helped ease the burden of having to share this information and decreased the anxiety caused by the anticipated confrontation by her family members.

Some participants however, especially the mothers who had children with Down syndrome, admitted that it was difficult to convey information about their child’s diagnosis as some family members did not understand the condition. Conveying genetic information to distant, as opposed to closer relatives, may be more emotionally challenging and the refusal to accept a child’s genetic condition by other family members may lead to poor psychosocial adjustment for parents (McCauley, Feuillan, Kushner *et al.*, 2001; Peterson. 2005). One participant said that it was difficult to explain the cause of her child’s condition to her family members who live in a rural area because they had never received genetic counselling themselves, and she thought that they might think that she caused the condition (Example 3.7.11).

> ***“From there that is when I ended up in genetic counselling where they explained things to me. I was sad at the time, I felt emotional because I didn’t take it seriously when they told me. When I got there at the time I felt sad, I cried and then asked myself I don’t have any relatives who are disabled, so what will they say? Because I’m the one who received counselling and not them so they don’t have the information and now I am the one who is supposed to go back and tell them about the child… where do I start? Are they going to think that I am the one who caused this upon the child? So I felt otherwise because I didn’t know what to tell them in Mpumalanga.”***
>
> *“Beku…I was..ngaba sad that time, ngafeela emotional cause beng’ngayi thathanga serious mabang’tshela but masengifika that time, bangenza ukuthi ngibe aware of it and then ngafeela sad, ngakhala and then…mara ngazibuza umbuzo ngathi, ekhaya mangabe akekho umtwana odisabled so mina bazothi ngenzeni? Abana information. Yimi engfanele ngibatshele ngomtwana, ngizoqala ngibathini. Are they going to think I’m the one o cause lokhu kulomtwana? Yabo…so ngafeela ngenyeindlela ngoba beng’ngazi ukuthi ngito fika ngibatshele kuthini ekhaya base Mpumalanga so bengazi ukuthi batshele uk’thini, ngibatshele kanjani.”*
>
> (Example 3.7.11: Ms. S., has a child with Down syndrome, Focus Group 1)

Communicating genetic information to family members who live in distant rural areas is challenging as many of them do not understand or believe the information conveyed due to lack of awareness of genetic conditions (Solomon *et al*., 2012). Genetic counselling is important in this situation, as research has shown that it facilitates family communication and family members’ acceptance of an affected child, as well as enhancing support for the child’s parents (Fortier and Wanlass. 1984; Bernhardt *et al*., 2000).

Communication amongst family members is influenced by both verbal and non-verbal behavior as well as family beliefs and values (Peterson. 2005; Hurst *et al*., 2011). This can be seen in this study where one participant, Ms. N. (Example 3.7.12) seemed frustrated because her mother-in-law (who had accompanied her to genetic counselling) had been dismissive about the information when told about the child’s condition at the hospital, saying that the condition did not exist.

> ***“Because that day when they told me that the child has Down’s syndrome, I was with his grandmother, his father’s mother. So that lady is just a different type of person. She said that there is no such thing. So you know, to them, yes the child has Down’s syndrome, but it’s not something that they feel that they need to worry about.”***
>
> *“Because that day ha batlo njoetsa hore ngoana onale Down’s syndrome, ne kenale le ngkono wa hae, mme wa ntate wa hae. So nje that lady is just ke mme omong nje. Are ha hona tho eo hayiko lento. So you know, to them, yes ngoanan onale Down syndrome, mara it’s not something nje joanong tlameile ba wore.”*
>
> (Example 3.7.12: Ms. N., has a child with
> Down syndrome, Focus Group 4)

It is important for genetic counsellors to be aware of family dynamics and perceptions when counselling parents, as communication with the family can either facilitate or hinder a parent’s acceptance and coping with their child’s genetic condition.

3.8 Theme 5: Dissatisfaction with the Healthcare System

Although it was intended that the focus group discussions centre around the experiences of genetic counselling, participants often spoke of the other services that they had received within the state healthcare system. Most participants expressed disappointment in the services that they had received, both in the hospitals and the smaller satellite clinics in their communities. Some of the main issues were; the feeling of being unheard and disrespected, frustration with healthcare professionals’ lack of knowledge regarding genetic conditions and poor attention to medical issues.

3.8.1 Feeling Unheard and Disrespected

Many of the participants felt let down by the healthcare system and often disapproved of the way in which they had been spoken to and treated. The stories told by the participants in the focus groups are ones of disempowerment, frustration and disappointment. One woman who has a child with albinism was upset with the manner in which the healthcare staff at the hospital spoke to her whilst giving birth (Example 3.8.1). It was traumatic for her to find out that her child had albinism and she felt that the staff had not been sensitive about the issue, or supportive towards her and her partner. She also seemed angry when asked by the staff if the child's father was Caucasian.

> ***"Yes, so they just saw the head and I was already in labour and was supposed to push the baby, so they said to me: "Is the child's father white?" and the child's father was there with me at the time, they saw him, it wasn't something that they didn't notice that this is my husband or this is the father of the baby. Then they said: "Is this the father of the baby?" and I said no. So when I pushed the baby I knew automatically, what can you imagine when someone says that to you? So I just pushed the baby."***
>
> *"Eh, so they just saw the head and I was already in labour and was supposed to push the baby, bathi kimi:"ubaba womtwana ngumlungu?" and ubab womtwana bekhona with me at that time bebambonile, it wasn't something that they didn't notice ukuthi this is the husband or this is the father of the baby. Then babethi: "Ubabomtwana?" Ngathi ah ah. So when I pushed the baby I knew automatically what you can imagine when someone says something like that to you? So I just pushed the baby."*
>
> (Example 3.8.1: Ms. A., has a child with albinism, Focus Group 3)

Another issue was that the participants' requests were often ignored and although mothers enquired about their children's health, they often did not receive an adequate response or were dismissed by healthcare staff (Example 3.8.2).

> ***“Then they told me that he may be a ‘slow’ child, then I saw that he was becoming worse, so I took him to Bara. Even there they didn’t give me the attention I needed as they kept chasing me away and I’d always return the next day until they finally sent me to the genetic people. When I got there they took some blood tests and they told me.”***
>
> *“…Then bangtshela ukuthi unagthola ukuthi uzoba slow, kunabantwana abanjalo baut ngathi seyenza worse then ngamthatha ngam’sa eBara…nakhona bebangafuni ngimisa ngafosta ngilobele ngimisa ngaksasa bang’xosha bangbuyise ngibuyele until baze bang’thumela lapha eGenetic ke yilang’fike bamthatha amagazi ke base bayang’tshela’ke.”*
>
> (Example 3.8.2: Ms. S., has a child with Down syndrome, Focus Group 1)

These types of statements have been described in previous South African research which showed that many staff in state healthcare facilities hold negative attitudes towards patients; that unprofessional behavior often occurs with minimal accountability, staff absenteeism is common and incidences of neglect, emotional and physical abuse by healthcare staff, particularly by nurses, towards patients is common (Gilson and McIntyre. 2007; Coovadia *et al*., 2009; Vivian *et al*., 2011). One participant, Ms. J. (Example 3.8.3), recalled the story of how her requests for assistance were ignored by healthcare staff when she went into early labour at six months gestation.

> ***"I have complications in my pregnancy since from five months. Everytime you'd find that I wake up from bed and stand on my feet, water would rush down my legs. So everytime I went to the clinic, I was working at the time, I'd get there and explain to them at the clinic that everytime when I get up from bed, water rushes down my legs. Ok fine they'd check me, and check me and give me pills, I think the ones to strengthen the baby's bones, then after all I gave birth when I was six months pregnant. One day I woke up and there was too much water, like a dam of water. Then I decided that yoh, I won't go to work, I'll go straight to the hospital. Then I went to the hospital and the doctor examined me and took me and put me into the sonar and everything, then he told me that there is still a lot of time, that I still have three months left, your time for delivery is still far. The very same day, after I finished collecting the medication that side, I felt that no, there is something wrong with me. So I went to the toilet to check, then I found that it was. Then I went back and they told me that the doctor is off to an operation and that I should wait. When he came back he checked me, and said to me: "No, you probably have a problem because you work too much and you don't rest, so I am asking you to book you in for bed rest, then I will discharge you tomorrow". The very same day I gave birth."***
>
> *"Mina I have complications in my pregnancy since from five months. Everytime uthole ukuthi mangivuka embhedeni ngime ngeyinyawo then kwehla amanzi, so everytime mangiya eclinic, bengisebenza that time, ngifike ngibaexplainele eclinic ukuthi njalo mangehla embhedheni kwehal amanzi. Ok fine bang'checke bang'checke bese bang'nikeze amaphilisi, I think lawa awoku qinisa amathambo abe abantwana, then after all, ngitete nge six months. One day ngathi mang'vuka lamazi wabamaningi kwaba nje idam yamazi phansi then nga decida ukuthi yoh angiyi emsebenzini namhlanje ngathi igiye straight es'bhedlela, then I go to the hospital then udoctoe wang'checka wangthatha wangifaka ku sonar and everything. Wang'tshela ukuthi isikhathi sisesiningi usasele ngenye ithree months isikhathi sakho sise kude. The very same day after ngiqeda 'kuyo thatha imedication bang'yenzela iprescription ngaya makumele ngiyo thatha imedication ngale ngiyezwa ukuthi no there is something wrong ngami, ngathi athi ngiye etoilet ngiyo checka, ngitole ukuthi khonamanje ngathi soyuki. Ngibuyele back to, ngithole bathi udoctor uyonyenza ioperation then ngimlinde. Makabuye angichecka, athi kimi: "noh wena maybe une problem yokuthi usebenza kakhulu awureste, no ngicela ukuzoku booka ibed rest then ngizokudischarger tomorrow". The very same day ngateta."*
>
> (Example 3.8.3: Ms. J., has a child with Down syndrome, Focus Group 3)

Ms. J. (Example 3.8.3) felt disempowered by the way she had been disregarded, and despite the healthcare staff telling her that she was not in labour and ignoring her plea for help, she delivered her baby on the same day. Patient disempowerment has also been repeatedly described in South African literature, with many patients receiving insufficient information about their diagnosis, treatment and management (Vivian *et al*., 2011). In another example, a participant from this study, Ms. P. (Example 3.8.4) was upset because a doctor had taken blood from her child to do a thyroid functioning test, but had not explained to her who he was, nor what the test procedure was about. He also never contacted her again about the test results, nor did he give her a follow-up appointment. Ms. P. felt that the doctor had not respected her, and she had not been adequately informed about the procedure or her child's health and management.

> ***"Then they performed the thryo what what on him. One of the things I didn't like about the service, I wasn't aware about the thyro test of my child, they didn't give me further details about when I should return for a follow-up. I don't know the doctor and the doctor hasn't, up to this day, given me a date to come and see the results of the test."***
>
> *"Basebamyenzai thryo what what. Ene oku engathi lapho kuyeloservice angiyithandanga, I wasn't aware ukuthi leyo thryo test leyo fanele, bay'thatha but azange bang'tshele ukuthi ngibuye nini ng'sokwenza ifollow*-up. *Angmazu udoctor, udoctor azange ang'nike idate lokuthi ngizochecka amaresults, up until namhlanje angw'wazi."*
>
> (Example 3.8.4: Ms. P., has a child with
> Down syndrome, Focus Group 1)

Another topic that arose during the focus groups was that of xenophobia. Xenophobia can be described as a "dislike of foreigners" (Harris. 2002 pp 2). One participant, Ms. A. (Example 3.8.5), who is from Zimbabwe, mentioned that staff in local state healthcare facilities sometimes acted in a prejudiced manner towards foreigners and that she had had a negative experience when interacting with some of the medical staff.

> ***"……especially when you are a foreigner, they have this…this way of treating foreigners that you don't understand why do they have to be like that you know…."***
>
> (Example 3.8.5: Ms. A., has a child with albinism, Focus Group 3)

Ms. A. felt that the healthcare staff did not show her respect and were often dismissive towards foreigners. Since 1994, South Africa has had increasing immigration of many foreign African nationals seeking refuge, and although the government has set policies in place to allow integration into local communities, foreign nationals still face many discriminatory challenges in the country (Landau. 2006). One of the major challenges remains access to healthcare, and although policies state that foreigners have equal access to basic health care, many are expected to pay a fee for these services because they do not have the correct documentation (Landau. 2006). Foreigners have also expressed their concerns around discrimination by local healthcare staff and have said that local medical staff are at times hostile towards them (Harris. 2002; Landau. 2006).

Although not discussed in depth, and not directly said, participants also seemed to re-iterate that HIV stigma is still present amongst the public, and that people do not openly discuss HIV, especially about their own HIV status. One participant, Ms. A. (Example 3.8.6), seemed uncomfortable disclosing her HIV status to the genetic counsellor and the observing students in the genetic counselling session, even though she was concerned about how HIV could potentially affect her child's genetic condition.

> ***"Because sometimes there are other things that you'd like to expose or to ask you know. Maybe for instance you want to know okay I'm HIV positive neh, I'm not saying that I am, I am not exposing my status; and you want to know maybe that my child is an albino, how can it affect that child and there are four people in the room. How do you say that? It's not nice."***
>
> *"Because sometimes there are other things that you'd like to expose or ask you know. Maybe for instance uzofuna ukwazi ukuthi okay I'm HIV positive neh, but I'm not saying that I am, I'm not exposing my status; and you want to know maybe ukuthi untwana oyi albino, how can it affect that child and there are four people in the room. How do you say that? It's not nice."*
>
> (Example 3.8.6: Ms. A., has a child with albinism, Focus Group 3)

HIV and tuberculosis epidemics are rife in South Africa, with an estimated 5.38 million people infected with HIV, and despite large global and national efforts, there still remain extensive challenges to control these diseases (Statistics South Africa. 2011; Kromberg, Sizer and Christianson. 2012). Despite ongoing public efforts in South Africa, stigma remains one of the largest obstacles to HIV/AIDS prevention and care (Campbell, Foulis, Maimane *et al*., 2005). Although antidiscrimination laws have been created within the country as well as education programmes to reduce the levels of stigma, negative community and family perceptions still occur with a poor understanding of HIV (Campbell *et al*., 2005). Many patients, their parents as well as their families who are seen at South African genetic counselling services are affected by HIV/AIDS. It is important for genetic counsellors to be aware of this, and be sensitive towards any medical- or psychological issues that may arise. In addition, patients' rights to confidentiality needs to be respected in genetic counselling sessions.

These previous research findings, together with the experiences described by the focus group participants in this study, are distressing to the researcher from a healthcare provision, professionalism and communication perspective. Research has shown that physicians, nurses and midwives sometimes abuse their power over patients and that the lack of consequence of abuse, supervision and control, perpetuates the situation (Vivian *et al*., 2011). Suggested reasons for this type of behavior have included low staff morale, lack of supervision, poor and stressful working conditions, a decline in staff numbers (which has lead to an uneven patient: staff ratio), lack of professional role models, as well as poor training and support (Walker and Gilson. 2004; Du Preez *et al*., 2007; Gilson and McIntyre. 2007; Coovadia *et al*., 2009).

There seems to be a lack of appropriate training (both medically and professionally) in communication skills for healthcare professionals, with many healthcare staff being ill-equipped with regard to communication and psychosocial skills, as well as professional behaviour. Support systems for healthcare staff are often limited or poorly organised, leading to decreased morale, increase in stress levels and poor communication, which is often transferred onto patients (Gilson, Palmer and Schneider. 2005; Sun, Saloojee, Jansen van Rensburg *et al*., 2008; Coovadia *et al*., 2009). Greater efforts are needed to improve these attitudes and services towards patients within the South African state healthcare system and healthcare professionals need to be more aware of sensitive patient issues.

3.8.2 State Healthcare Professionals: “They Are Very Much Ignorant”

Participants in this study also expressed frustration with healthcare staff’s lack of knowledge with reference to their child’s genetic condition. This was especially emphasised for smaller clinics. Many participants felt that this lack of knowledge and availability of good services led to poor health outcomes and management. This has been mentioned in previous research, and although South Africa is considered to be a middle-income country based on its economy, it has very poor health outcomes, with some instances considered to be worse than low-income countries (Coovadia *et al*., 2009).

Some participants thought that the healthcare staff at the clinics were unaware of clinical features of certain conditions, did not do proper investigations and only did very basic routine checks, as mentioned by Ms. F. who has a child with haemophilia B (Example 3.8.7).

> ***"and some of the clinics, if like in that history of that clinic they never had such a problem. They are very much ignorant so they just do a routine check. That's why most of the Down syndromes or whatever, they don't notice because they are not doing any further investigations (people agree in background)… that's why."***
>
> (Example 3.8.7: Ms. F., has a child with haemophilia B, Focus Group 3)

In addition, participants voiced that at the clinics, there was a lack of staff training, available equipment and resources as well as poor staff-patient communication, which led to poor management of the child's condition or antenatal care. One participant in the study, who has a child with albinism who frequently got sunburnt, felt that she was not adequately informed at the clinic on how to manage her child's condition and how to take care of her child's sensitive skin (Example 3.8.8).

> ***"So sometimes with other you'd want to know how to prevent it. They don't tell you at the clinic that ok buy SPF what for that ointment, buy this lotion for her, buy these things you see."***
>
> *"So sometimes ezinyo izinto you'd want to know how to prevent it. They don't tell you at the clinic ukuthi ok buy SPF what for that ointment, buy this lotion for her, buy these things you see."*
>
> (Example 3.8.8: Ms. A., has a child with albinism, Focus Group 3)

Ms. F. (Example 3.8.9), also felt concerned that healthcare staff in the clinics and in some hospitals were not adequately trained on how to manage and treat genetic conditions.

> ***"They are ignorant, and the only thing to make things better I think health education is very much important because of some people don't have information. Even if you say one thing everyday, four times a day but somebody grasping something from them. So most of them are ignorant, they don't do those health educations, during observation they just give you pills then ask: "How are you? Are you fine?""***
>
> *"ba ignorant, and the only thing to make things better I think education is very much important because of some people don't have information; even if you say one thing everyday, four times a day but somebody grasping something from them. So most of them ba ignorant, they don't do those health educations, during analysis just bahufa dipilisi, "how are you? Are you fine?""*
>
> (Example 3.8.9: Ms. F., has a child with haemophilia B, Focus Group 3)

Ms. F. is a nursing student, so her genetic knowledge was better than the other participants and she felt that healthcare staff should be more aware of common genetic conditions. State hospitals and clinics have had difficulty in managing medical conditions appropriately due to a lack of human resources, standardisation of training across different healthcare facilities, staff supervision and support, as well as infrastructure (Coovadia *et al.*, 2009; Vivian *et al.*, 2011). The Department of Health is nationally involved in developing policies and co-coordinating the country's state healthcare (Gilson and McIntyre. 2007). Provincial departments are responsible for providing health services to state hospitals and primary healthcare clinics at a local level (Gilson and McIntyre. 2007). Although good policies are created, implementation of these are poor as a result of unsatisfactory managerial practices, failures in leadership and reduced numbers of staff (Coovadia *et al.*, 2009). Greater efforts are needed both on a national and provincial level to resolve these issues and genetic counselling practice needs to learn how to operate effectively within the boundaries of a poorly functioning healthcare system.

3.9 Theme 6: Greater Need for Awareness Campaigns

Another topic discussed in all the focus groups was a greater need for awareness in the community and healthcare facilities about genetic conditions (Example 3.9.1).

> ***"I think it should be advertised. People should be aware of it and have knowledge because it's relevant for children with albinism as well as Down syndrome. They also need to have knowledge, about why their child is a certain way. What should they do?"***
>
> *"I think tshontse bai advertise. People should be aware of it, babe le knowledge, because not only bana ba albinism, but banaleng di Down syndrome. There also needs to be like go be le, babe le knowledge gore goetsagalang ka ngoana oa bona, why ngoana oaka aleso? Tshontso ke etse eng?"*
>
> (Example 3.9.1: Ms. K., has a child with albinism, Focus Group 2)

The need for greater awareness about genetic conditions as well as genetic services in South Africa has been described in previous South African research (Christianson *et al*., 2000; Kromberg *et al*., 2012; Solomon *et al*., 2012). Genetic services are scarce in developing countries such as Africa, and in South Africa, it is estimated that there is only one genetic counsellor per 5 million people, meeting approximately only 10% of the country's genetic needs (Christianson *et al*., 2000; Edwards, Greenberg and Sahhar. 2008; Kromberg *et al*., 2012). Financial constraints, lack of political interest and coordination, a decline in staff numbers and facilities as well as deficiencies in awareness of genetic conditions all contribute to the lack of availability and awareness of genetic services (Christianson *et al*., 2000; Kromberg *et al*., 2012).

Several participants said that they knew of other affected children in the community whose families did not know the cause of their conditions. This has been mentioned in previous research, where South Africans have a poor understanding of the cause and prognosis of genetic conditions, and healthcare professionals are often not aware of the availability of genetic services or what they entail (Christianson and Kromberg. 1996; Penn *et al*., 2010; Solomon *et al*., 2012). The participants felt that if the community as well as healthcare professionals were more knowledgeable about these conditions, then parents would be better prepared, that children with genetic conditions would be better managed, and it would help alleviate the emotional stress placed on families (Example 3.9.2).

> ***"Honestly, I think it's better to accept it, and once you've accepted it, it to be taught even to our young children that get babies at a young age. Even at the clinics before they give birth, when they do all those things they do to pregnant mothers, when they do clinicals on pregnant women, they must teach them from the start that things like this do happen."***
>
> *"Mina kwamampela ngibona ngathi kungcono ukukwamukela nokuthi sikwamukelile, and then sesik'wamukelile kufundiswe nase bantwaneni njengoba abantwana bethu bathola abantwana basase bangcane. Nase maclinic before bazo teta noma mabayenziwa vele lezinto zabo mama, kwenziwa iclinical yabo mama abakhulelwe, ba ba teache from the start izinto ezinje ngalezi ziyenzeka."*
>
> (Example 3.9.2: Ms. P.K., has a child with Down syndrome, Focus Group 3)

Participants in Focus Group 2 were particularly vocal about this topic and felt that genetic conditions should get the same amount of attention and campaigning as HIV, and that genetic counsellors/geneticists, as well as other healthcare professionals and government officials should make a greater effort in creating awareness within the community and in local clinics (Example 3.9.3).

> Ms. T.:***"Maybe a workshop, like one for HIV, bringing information to the township on these kind of things. Even a 5 year old child now would know what it is."***
>
> *"Ne re re di campaign tsa HIV like just like everybody o bua ka tsona, di gona these kind of things. Le ngoana oa 5 I'm sure oa itse hore ke eng by now so."*
>
> Ms. K.:***"So as much as they put in a lot of money and advertise HIV campaigns, it should also be in the township whereby the HIV campaigns go to the extent of finding schools on weekends. Once in a while so that we can get more knowledge from local clinics."***
>
> *"So as much as ba Kenya di tsheletse tse dingata, bo advertisa di campaign tsa HIV, it should also be ko lokshin whereby, di campaign tsa HIV, they go to extent tsa go educate dikolo on weekends, gore di etsue. Something like that ka di weekend, re gone ho thola more knowledge."*
>
> (Example 3.9.3: Ms. T. and Ms. K., both have children with albinism, Focus Group 2)

"Annual Health Awareness Days" are held nationally by the Department of Health (National Department of Health. 2012), but greater effort needs to be made to reach broader communities as well as smaller healthcare facilities. The researcher feels that healthcare professionals, including genetic counsellors and geneticists need to get more directly involved in these efforts for awareness of genetic conditions.

4 CHAPTER 4: CONCLUSION, STUDY LIMITATIONS AND RECOMMENDATIONS

This chapter includes a summary the overall research findings of this study, the study limitations, future recommendations for genetic counselling services as well as recommendations for future research.

Amongst the participants, prior to their own experience, there was a general lack of awareness of genetic counselling and genetic conditions. Previous South African studies have shown that many individuals either have no knowledge or a poor understanding of the genetic conditions explained to them during a genetic counselling session (Christianson and Kromberg. 1996; Christianson. 1997; Solomon *et al.*, 2012). This finding, is however not unique to a South African setting, with international research showing that members of the public often have a poor concept about basic genetic information and usually do not understand terminology used such as “genes” and “chromosomes” (Morren, Rijken, Baanders *et al.*, 2007; Klitzman. 2010).

The study participants had mixed responses about their experiences of genetic counselling. Some had a positive experience, and felt respected and felt that their questions had been answered. Some seemed to have had a sense of “empowerment”, were able to ask questions and had been able to have a conversation with the health professional on an equal level. Patient empowerment has been expressed by individuals after receiving genetic counselling in previous studies and individuals have appreciated the information received and felt that they had gained personal control over their situation (Payne *et al.*, 2007; McAllister *et al.*, 2008).

Conversely, other participants in this study expressed having a negative experience, with the main issue being confusion caused by the questions, as well as the lack of personal and family privacy. Research has shown that it is important that counsellees understand the information provided to them as it leads to better client satisfaction (Hall. 2003). It is important for genetic counsellors/geneticists to ensure that patients/parents understand the concept of genetic counselling, the relevance to their/their child's condition and to understand what the condition means for them. The purpose of gathering information such as the family pedigree, needs to be clarified and it should be done in a respectful and non-judgmental manner, as some families prefer keeping family matters private and do not like discussing their family members (Saleh *et al*., 2009). Genetic counsellors/geneticists should be able to assess a patient's readiness and interest to receive information and be sensitive to non-verbal cues. An understanding of the information and risks given should be frequently assessed by the counsellor to ensure that there is no confusion, and information should be conveyed at an appropriate level of understanding allowing for easy interpretation, and should be provided both verbally and in a written format (Culver *et al*., 2001; Wasserman *et al*., 2010).

With regard to patient confidentiality and academic teaching for medical and genetic counselling students, patients should be asked whether they feel comfortable having students present in a consultation. Genetic counsellors and staff in teaching hospitals need to be more sensitive to these issues and although students need to be given the opportunity to learn from observed patient consultations, both students and their mentors need to be respectful towards the patient's confidentiality. Allowing patients/parents to feel at ease and respecting their confidentiality, could assist in improving the overall genetic counselling experience for individuals.

The value of the information provided to a counsellee should also be considered. Counsellees from previous research have spoken about “information overload” in some genetic counselling sessions (Collins *et al*., 2001). Although it is important to provide adequate and accurate information, genetic counsellors/geneticists need to gauge whether the counsellee is ready to receive this information and information should be delivered in “bits” rather than in “bulk” so as to not overwhelm the counsellee. It is also important to acknowledge and enquire about patients’ beliefs to facilitate the communication process, convey information in an understandable manner and to make the information personally relevant to the individual (Weil. 1991; Kinney *et al*., 2010). The role of partners should also be considered when arranging genetic counselling appointments. From the focus groups, it seemed evident that many play an important role in their children’s lives, and mothers felt that it would be beneficial for them to understand the management and care for their children’s genetic conditions.

Early genetic counselling may be beneficial to patients. Receiving adequate information soon after a child has been diagnosed facilitates emotional support, decreases concerns over an unknown condition and assists parents to gain a sense of control over their current situation. However, to be able to accommodate individuals’ preferences on the time that genetic counselling takes place is challenging, as genetic counselling services rely on referrals from other healthcare professionals. Research has shown that although some patients self-refer, most individuals seen for genetic counselling are referred from general practitioners, medical specialists or other healthcare professionals (Bleiker *et al*., 1997; Davey *et al*., 2005). In the South African state healthcare system, almost all patients are referred by other healthcare professionals in the system. Increased awareness of genetic counselling by staff in antenatal and paediatric units in state hospitals and community clinics may increase the amount of referrals made, as well as sensitivity to timing. It is important for genetic counsellors/geneticists to be sensitive and aware that parents are often at different stages of grief when they are seen for genetic counselling, thus a second consultation may be more beneficial for some parents.

Participants in the study emphasised the need for greater support, with many viewing genetic counselling as a “life-line”. Studies have indicated that parents of affected children often request ongoing support, they feel that it is beneficial and it improves their overall coping and acceptance of the condition (Skirton. 2001; Muggli *et al*., 2009). Although efforts are made to see individuals for follow-up sessions, it is sometimes difficult to arrange since the compliment of geneticists and genetic counsellors at Genetic Clinics in Johannesburg are currently low. With regard to patients’ difficulties of reaching counsellors telephonically, patients are usually given the department’s secretary’s telephone number, as genetic counsellors/geneticists are often not in the office, but at hospital visits or clinic attendance and many healthcare professionals do not want to give out their personal cell phone numbers to maintain professional boundaries. A follow-up appointment or telephone call may provide further emotional support, and allow the counsellor to assess whether or not the patients are coping. Appropriate referrals can then be made if indicated.

The value of support groups must also be emphasised as they offer ongoing education, support and opportunities for parents to meet other parents who also have an affected child. Previous research has shown this (Weil. 1991; Muggli *et al*., 2009) and it was reiterated by the participants in this study.

Genetic counsellors/geneticists practicing in culturally, linguistically and ethnically diverse settings need to be aware of the influence of religion, traditions and myths in healthcare consultations. This study together with other South African research has shown the existing respect for elders in the community and that grandmothers play an important role in the upbringing of children (Penn *et al*., 2010). More emphasis needs to be placed on these South African traditions as it may benefit communication in genetic counselling consultations. Many Black Africans also make use of both Western and traditional healing systems (Solomon *et al*., 2012). Previous research has shown that instead of Western healthcare professionals dismissing the opinions of community elders and beliefs in traditional medicine, they should rather communicate with these members of the community to understand their approaches to healthcare and mutually benefit from each other in the health and treatment of a mutual patient (Kale. 1995; Penn *et al*., 2010; Solomon *et al*., 2012).

The training and hiring of professional interpreters also needs to be emphasised as mentioned in previous research (Schlemmer and Mash. 2006; Penn. 2007). Interpreters should be trained in psychosocial skills as well as basic healthcare to allow them understanding and interpretation of the information conveyed between counsellees and healthcare professionals.

Patient confidentiality and patients' level of comfort with the presence of an interpreter needs to be respected.

With regard to a patient's family, the researcher feels that genetic counsellors/geneticists place a large responsibility on counsellees to convey genetic information and risks not only to their own family, but also to their partners and to their partner's family, which may be a stressful and challenging process. The term "family" has different meaning in different communities, and decisions around an individual's health are often made at a family-level (Richards. 1996; Penn *et al*., 2010). A family's acceptance of a condition can largely influence parents and an affected individual's coping and support structures (Peterson. 2005). Genetic counsellors/geneticists should enquire about family dynamics and communication to gauge the patient's family environment. Genetic counsellors can offer their support by welcoming participants to invite their families to genetic counselling and by providing cascade letters to family members informing them about the condition and their own risks. In the same way, genetic counsellors can assist in communication and understanding of genetic conditions amongst the community. Stigma and community beliefs about genetic conditions are present in many communities (Penn *et al*., 2010; Meilleur *et al*., 2011). Genetic counsellors/geneticists, together with main role players within the community, need to find ways to address these issues.

Although the focus of this research was on mothers' experiences of genetic counselling, participants seemed to view genetic counselling as part of the greater healthcare system, and did not view the service as a separate entity. Communication, staff training and care, not only in genetic counselling services, but in the greater South African healthcare system needs to be addressed. Lack of adequate healthcare training, disrespect for patients and lack of professionalism amongst healthcare staff, as well a lack of resources seem to be evident in the participants' stories in this study, as well as in previous literature (Coovadia *et al*., 2009; Vivian *et al*., 2011). These concerns, together with limited access to genetic services (since it is only offered at tertiary level urban healthcare facilities), exacerbate the lack of knowledge amongst healthcare staff about genetic conditions, services and where to refer patients (Penchaszadeh, Christianson, Giugliani *et al*., 1999. Kromberg *et al*., 2012; Solomon *et al*., 2012). It is important for genetic counsellors/geneticists to keep these issues in mind when meeting patients for the first time, as well as when making patient referrals to other state services since these issues affect the overall experience of patients within the state healthcare system. Some suggestions to enhance the current difficulties include improved staff salaries, increases in staff numbers, ongoing training in education, medical conditions and professionalism, improved healthcare working environments, funding, support and equipment (Walker and Gilson. 2004; Du Preez *et al*., 2007; Coovadia *et al*., 2009; Vivian *et al*., 2011). Greater staff support and interaction is also needed to improve staff relationships, morale and encouragement (Sun *et al*., 2008).

Transport and geographic difficulties also need to be kept in mind when referring patients to other healthcare services. Healthcare professionals should refer patients to the nearest possible appropriate healthcare facility. Genetic counsellors/geneticists also need be sensitive to xenophobic issues when counselling foreign individuals and the researcher feels that the South African government must strive to ensure that xenophobic attitudes are reduced amongst local citizens and that foreigners have equal access to medical treatment in state health care facilities.

Genetic counsellors also need to be aware of HIV-stigma, as it still remains a large issue within South African and global societies. Although HIV-counselling is not a primary role of genetic counselling, if patients' disclose their HIV status during a session, counsellors should be sensitive to this, respect patient confidentiality and ask how they are coping with regard to their management, family communication and support. In doing so, genetic counsellors can assist patients with their ongoing management and coping skills by referring them to relevant health professionals such as doctors, social workers or psychologists as well as relevant support groups who are equipped in dealing with HIV-related issues.

Greater efforts for awareness campaigns regarding genetic conditions and genetic counselling services was raised by participants in this study. The Department of Health has annual "Health Awareness Days" for specific health-related issues, including genetic conditions such as Down syndrome, haemophilia as well as other inherited disorders (National Department of Health. 2012). Genetic counsellors and geneticists should get more directly involved in these national campaigns and awareness days at local hospitals, clinics and community forums to improve staff knowledge and awareness of genetic conditions and referral resources. Efforts of awareness amongst other health care workers may increase referrals to genetic services and ensure that affected individuals, as well as at-risk family members, receive appropriate genetic counselling and medical care. None of this will be possible without governmental support, thus it is important to lobby government to assist in creating awareness and care for individuals and families affected by genetic conditions.

The limitations of this study were:

- The study only described and documented experiences of mothers who have children affected with a genetic condition from Genetic Clinics in Johannesburg. Due to the qualitative nature of this research, the study aimed for depth and contextual authenticity. The findings can thus not be generalised to a wider population such as all patients in Johannesburg, state healthcare facilities, or genetic counselling services offered in other parts of South Africa. The findings from this research study could however be transferred to a similar context with similar type participants (using related research methods).
- The researcher is unable to speak an African Black language, and could not understand the conversations in the focus groups, nor interpret or read the transcriptions. This limited the transparency of the data collection and analysis, as the researcher could not have insight into the conversations nor the transcriptions and translations, and had to rely on the skills and interpretation of the research assistants.
- The focus group facilitator did not probe further into some of the comments made, thus some of the meanings or deeper understandings remain unexplored.

Recommendations for genetic counselling services in Johannesburg, South Africa:

- The researcher recommends that greater efforts for genetic counselling and genetic service awareness campaigns need to be made. Informative lectures and the distribution of written pamphlets, leaflets and posters to health professionals and members of the community may be helpful, and community platforms such as hospitals, clinics, support groups, community leaders and schools should be utilised to raise awareness about genetic conditions. These actions may not only improve communities' and health professionals' awareness of genetic conditions, but may also contribute to alleviating parents' anxiety in introducing their affected children into society, decreasing stigma and correcting preconceived ideas about genetic conditions.

- Genetic counsellors/geneticists should carefully assess patients' understanding of the information provided to them during genetic counselling consultations so as to ensure that the patients are not overwhelmed, that the information is personally relevant to their situations and that patients adequately benefit from the provided service.
- The researcher feels that it may be beneficial to improve communication and co-ordination with antenatal and pediatric units in hospitals to ensure early referrals to the Genetic Clinics for parents with a newly diagnosed child. In addition, a follow-up appointment for parents who are not yet emotionally ready to hear the genetic information would be beneficial. This will ensure that all parents/patients adequately benefit from genetic counselling.
- It is further recommended that fathers of the children should either be invited together with the mother if they are still in a relationship, or they should be invited separately. Genetic counsellors should also assist parents by equipping them with psychosocial skills and information in the form of pamphlets, cascade letters and copies of diagnostic results, to convey sensitive information to partners and family members.

- To enhance support for parents who have children affected with genetic conditions, the researcher feels that genetic counsellors/geneticists can facilitate support for parents by identifying other families who have an affected child who have accepted the diagnosis, are coping well and who would be willing to speak to new parents. This will assist in offering alternative support to parents outside of genetic counselling. Recommendations are made that a second genetic counselling office contact number be given to patients, or a designated area or "booking sheet" should be appointed in the hospitals. Patients can then be assisted by a healthcare professional to make their own booking, request a follow-up telephone call, or be put onto a waiting-list for another genetic counselling appointment.
- Genetic counsellors/geneticists should attempt to make greater efforts to communicate with other healthcare professionals involved in the management of a patient, to try and co-ordinate appointments on the same day if possible. This will prevent the patient and their parents/care-takers having to travel to the hospital on several occasions and incurring expensive travelling costs.
- Lastly, with regard to communication amongst different cultural and ethnic groups in South Africa, the researcher recommends ongoing training to healthcare professionals about traditional beliefs. Cultural competency needs to be practiced so effective communication can take place regardless of an individual's background and traditional beliefs. The training and hiring of professional interpreters at healthcare facilities is recommended to facilitate breakdown in language barriers and allow effective communication. Respected community members, such as elders and traditional healers should be approached to assist in communication and understanding of genetic

conditions. Together, this will not only strengthen community relationships, but may also lead to a greater acceptance of genetic conditions.

Recommendations for future research:

- The researcher feels that further research is needed into the experiences of individuals seen for genetic counselling for other genetic conditions. Larger sample sizes and additional focus groups are warranted in future research.
- Further research into individuals' experiences who received genetic counselling in other South African settings would be valuable.
- Greater understanding of different beliefs, cultures and traditions needs to be explored so as to improve understanding and respect amongst different ethnic and language groups in South Africa. The use of language interpreters in the South African healthcare setting also needs to be further assessed to gauge its value.
- Further research is needed by the Department of Health, healthcare training co-ordinators, hospital and clinic managers as well as other healthcare leaders and administrators to better understand and address the challenges that healthcare facilities and healthcare staff face. This may facilitate the development of strategies that will promote professionalism, ongoing education and training, optimal standards of care, as well as effective communication amongst, and between healthcare professionals and patients.

In conclusion, this study aimed to describe and document the experiences of mothers who had received genetic counselling at state hospitals in Johannesburg, South Africa after having a child diagnosed with a genetic condition. Thirteen Black women agreed to participate and four focus groups were conducted in an African language and recorded data was obtained. All data were transcribed and translated for the analysis and the development of themes. Valuable insight has been gained from the research findings, not only about genetic counselling services, but also about the greater South African healthcare system. Findings of this study cannot be generalised to all individuals seen for genetic counselling in South Africa however, and more research is needed on this topic. It is hoped that the observations noted in this study, together with the recommendations made, can assist in providing better genetic counselling services in Johannesburg, South Africa. The researcher feels that this study will contribute towards improving understanding and care for individuals affected by genetic conditions, not only within the healthcare system, but also with due consideration of the South African cultural context.

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7 APPENDICES

APPENDIX A: RESEARCH ETHICS CLEARANCE CODE

UNIVERSITY OF THE WITWATERSRAND, JOHANNESBURG
Division of the Deputy Registrar (Research)

HUMAN RESEARCH ETHICS COMMITTEE (MEDICAL)
R14/49 Miss Megan Morris

CLEARANCE CERTIFICATE	M111160
PROJECT	South African Women's Experience of Genetic Counselling
INVESTIGATORS	Miss Megan Morris.
DEPARTMENT	School of Pathology/Human Genetics
DATE CONSIDERED	25/11/2011
M111160DECISION OF THE COMMITTEE*	Approved unconditionally

Unless otherwise specified this ethical clearance is valid for 5 years and may be renewed upon application.

DATE 27/01/2012 **CHAIRPERSON** ....
(Professor PE Cleaton-Jones)

*Guidelines for written "informed consent" attached where applicable
cc: Supervisor : Sr Merlyn Glass

DECLARATION OF INVESTIGATOR(S)

To be completed in duplicate and ONE COPY returned to the Secretary at Room 10004, 10th Floor, Senate House, University.

I/We fully understand the conditions under which I am/we are authorized to carry out the abovementioned research and I/we guarantee to ensure compliance with these conditions. Should any departure to be contemplated from the research procedure as approved I/we undertake to resubmit the protocol to the Committee. **I agree to a completion of a yearly progress report.**

PLEASE QUOTE THE PROTOCOL NUMBER IN ALL ENQUIRIES...

APPENDIX B: PARTICIPANT INFORMATION SHEET

Mother's Experiences of Genetic Counselling in Johannesburg, South Africa

Investigator: Megan Morris, MSc (Med) Genetic Counselling Student

Good Day,

My name is Megan Morris and I am a student at the Division of Human Genetics, Faculty of Health Sciences, University of the Witwatersrand. I am doing research to find out how Black South African women, who have a child with a genetic condition, have experienced Genetic Counselling. It is important for us to know how South African clients have received our Genetic Counselling Services so that we may improve them in the future. I would like to invite you to volunteer in this study.

Group discussions with five to ten African women will be held in a language suitable for you. The discussions will be lead by an experienced Black South African woman. Your participation in this study is completely voluntary. The group discussions (with your permission) will be voice-recorded and you and the other women will hear each other's stories. Confidentiality is not possible in focus groups and the discussions may include personal thoughts and feelings. If you are uncomfortable, you do not have to share all your personal information. We kindly ask that you do not discuss what you hear from others in the group with anyone else outside of the group.

If you feel anxious due to some of the questions asked, the researcher can refer you for Genetic Counselling to a Genetic Counsellor. I would really appreciate your participation in this study, as you will be helping us learn more and help improve Genetic Counselling in South Africa. We will refund your travelling costs to the location at which the focus groups will be held and back home. Refreshments will also be served.

Your personal information will be kept strictly confidential and information obtained from the study will be anonymous. Only the researcher and authorised individuals will hear and have access to the audio-recordings, which will be kept in a secure location. Should you want to withdraw from the study, you have the right to do so at any time and your refusal or withdrawal will not affect any present or future treatments.

If you have any questions, please contact me or my colleagues on the numbers listed.

Kind regards,
Ms. Megan Morris- BScMedSci Hons Human Genetics, Genetic Counselling Student- 011-489-9223/4

Ms. Merlyn Glass- RN, RM, Dip Paeds MSc Nursing; Genetic Nurse Counsellor - 011-489-9603

Ms. Tina-Marie Wessels- MSc (Med) Genetic Counselling, Genetic Counsellor - 011-489-9243

Professor Peter Cleaton-Jones- Chairman of Human Research Ethics Committee (Medical) - 011-717-2301

APPENDIX C: FOCUS GROUP QUESTION GUIDE

1. Can you tell me about the time you attended the genetic counselling (GC) clinic?
2. What was it like for you?
3. What was helpful during GC?
4. What was the most helpful during GC?
5. What was not helpful during GC?
6. What was the most unhelpful during GC?
7. What does your community think about GC?

Please contact the following people if there are any queries:

Ms. Megan Morris- BScMedSci Hons Human Genetics, Genetic Counselling Student- 011-489-9223/4

Ms. Merlyn Glass- RN, RM, Dip Paeds MSc Nursing; Genetic Nurse Counsellor - 011-489-9603

Ms. Tina-Marie Wessels- MSc (Med) Genetic Counselling, Genetic Counsellor - 011-489-9243

Professor Peter Cleaton-Jones- Chairman of Human Research Ethics Committee (Medical) - 011-717-2301