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ORIGINAL RESEARCH

Mothers' Experiences of Genetic Counselling in Johannesburg, South Africa

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Abstract Genetic counselling is offered in diverse settings, and patient reactions vary due to differences in personal, family and community beliefs, local healthcare settings, as well as cultural background. Together, these factors influence how individuals experience genetic counselling. This study aimed to describe and document the experiences of thirteen mothers, with children with Down syndrome, oculocutaneous albinism or haemophilia B, who had received genetic counselling at state hospitals in Johannesburg, South Africa. A qualitative research design drawing on principles of Interpretative Phenomenological Analysis was used. Four voice-recorded focus groups were conducted and the resulting data were analysed using thematic content analysis. Five themes were identified in the data: thrown into the unknown; a worthwhile experience; a break in communication; telling the family and the community; and spreading the word. It was seen that genetic counselling cannot be viewed as a singular experience, but rather as one which is influenced by mothers' lived experiences and their interactions with other healthcare services, family and community members. The results from this study showed that genetic services and conditions were poorly understood, that the experience of genetic counselling varied amongst mothers, and on-going patient support is needed particularly when addressing family and community members. Further research is needed to assess what information is valuable to individuals during genetic counselling and how to deliver this information in a contextually appropriate manner. Greater awareness of genetic conditions is also required amongst communities and healthcare professionals. Valuable insight was gained from this study which can be used to improve local training programmes and genetic counselling services in Johannesburg, and in South Africa.

Keywords Genetic counselling · South Africa · Experiences · Information · Family · Community

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Genetic counselling is a growing profession with services being offered in many multicultural settings. According to the National Society of Genetic Counsellors (NSGC), genetic counselling can be defined as a process which assists individuals to understand and adapt to the medical, familial and emotional implications of the genetic contribution to disease (Resta et al. 2006). Few studies have assessed the value of genetic counselling and client satisfaction (Aalfs et al. 2007). Of those that have been undertaken, client experiences were generally shown to be positive; however, most of these studies were done in first world settings with Caucasian individuals (Aalfs et al. 2007).

The way in which individuals experience genetic counselling varies among countries due to differences in culture, religion, family and community values, as well as local socio-economic factors (Kinney et al. 2010). Family and community beliefs and interactions can impact whether genetic conditions are disclosed, how genetic information is interpreted, and how related healthcare decisions are made (Kinney et al. 2010; Saleh et al. 2009). Linguistic diversity, educational level, as well as a country's healthcare setting may also influence an individual's experience (Kinney et al. 2010). Language barriers can reduce understanding of the information conveyed in a consultation, limit emotional expression and result in poorer healthcare outcomes (De Maesschalk et al. 2011). Individuals from developing countries, including South Africa, may be unaware of genetic services, may become confused about the process, and may be less willing to accept the genetic information (Kinney et al. 2010; Solomon

et al. 2012). In societies where there is much cultural diversity, current genetic service provision may be incompatible with culturally determined attitudes towards genetics, testing and counselling (Barlow-Stewart et al. 2006).

Background on South Africa

South Africa provides a unique setting for genetic counselling and healthcare provision because of its cultural and linguistic diversity, socio-economic problems and apartheid history (Penn et al. 2010). 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 % Caucasian, and the remaining percentage compromised of Mixed Ancestry, Indian and Asian individuals (Statistics South Africa 2011a). Amongst individuals aged 20 years and older, an estimated 27.4 % have a Grade 12 education level, and within the population, there are an estimated 6.5 % of individuals who have no formal education (Statistics South Africa 2012a).

South Africa has eleven official spoken languages and several practiced religions including Christianity, sectarian Christian Independent churches (e.g. Zionists), Islamism, Hinduism, Buddhism, and Judaism (Statistics South Africa 2012b). There are also different cultural beliefs and practices. For example, many individuals from Black ethnic groups believe in shared decision-making amongst family and community members, have fatalistic viewpoints, believe that ancestors can affect the lives of the living, and make use of the traditional healing system in conjunction with westernised healthcare practices (Kasanga and Lwanga-Luma 2007; Kromberg and Jenkins 1997; Penn et al. 2010). Consanguineous marriages occur among some Black ethnic groups, particularly the Tswana and the Swazi (Kromberg and Jenkins 1997).

South Africa is made up of public state funded healthcare services (utilised by approximately 61.2 % of the population) and private healthcare services (utilised by an estimated 24.3 % of the population) (Statistics South Africa 2011b). Black individuals make up 81.3 % of those who use state healthcare services, and Caucasian individuals make up 88 % of the users of private healthcare facilities (Statistics South Africa 2011b). According to the South African National Health Act of 2003, all state run healthcare facilities are obliged to provide services to all pregnant and lactating women and children under the age of 6 years at no cost. In addition, women have free access to termination of pregnancy services upon request, during the first 12 weeks of gestation, and thereafter in consultation with a medical practitioner if the mother or the foetus' life is at risk (Kromberg et al. 2013a). Based on the above, it can be concluded that South Africa has high levels of ethnic diversity, particularly among non-Caucasian groups, with multiple linguistic characteristics and low levels of education. Additionally, there are major issues with regard to poverty, unemployment, and access to adequate healthcare, housing, water, electricity and sanitation (Coovadia et al. 2009). Again, this is concentrated among the Black majority of the population.

South Africa's genetic disease burden is thought to be about 53.4 per 1000 live births, with some of the more common conditions including Down Syndrome (approximately 1 in 525 births), oculocutaneous albinism (OCA) (1 in 3900 Black individuals) and cystic fibrosis (1 in 3000 Caucasian individuals) (Kromberg et al. 2013a). Support services for individuals with congenital disorders include state disability grants for severely affected individuals and some special educational and care facilities (although limited mainly to urban areas) (Kromberg et al. 2013a). There are also existing genetic support groups such as the South African Haemophilia Foundation, Down Syndrome South Africa, the Rare Disease Society of South Africa, and the South African Inherited Disorders Association (SAIDA) (Down Syndrome South Africa 2014; Kromberg et al. 2013b; Rare Disease Society of South Africa 2014; South African Haemophilia Foundation 2014).

Genetic Counselling Services in South Africa

Recognised genetic counselling services have been available locally since the 1970s, mainly at tertiary state healthcare facilities (Jenkins 1990). Prior to 1994 (during the apartheid era), most genetic counselling services were utilised by middle- to upper-class urban Caucasian individuals; however Black South Africans had improved access after a genetic clinic was established at Chris Hani Baragwanath Hospital in Soweto in 1987 (Kromberg and Jenkins 1988). A formal genetic counselling training program was first established in 1988, and is grounded on the principles of a non-directive and client-centred approach that aims to empower individuals to use genetic information in a way that will allow personal control, reduce psychological stress and permit the making of decisions in a meaningful way (Kessler 1997; Kromberg et al. 2013b).

Genetic services are currently offered in five major urban centres (Johannesburg, Cape Town, Stellenbosch, Bloemfontein and Durban) mostly at tertiary level public healthcare facilities (Kromberg et al. 2013a). Development of the profession, however, has been slow, with only 11 medical geneticists and 10 genetic counsellors practising at present (Kromberg et al. 2013b). The staff demographics are also not representative of the South African population, since the majority of personnel are female Caucasians, most of whom are only conversant in English (Wessels 2013).

Individuals presenting for genetic counselling are counselled by a clinical geneticist or a genetic counsellor, and they are mostly referred by other healthcare professionals (almost all cases seen in the state healthcare system) or are self-referred. A genetic counselling session is typically 60 min in length and involves: gathering personal information; explaining the genetics, medical concepts and the relevant risks related to the diagnosis; supporting decision making; and offering psychosocial counselling. Almost all sessions are done in English, but when communication is compromised, a lay person, a family member, or a nurse is asked to assist, as there are no formal interpreters available at the clinics (Wessels 2013). There is no cost for a consultation with a genetic counsellor, or a clinical geneticist, if an individual is seen within a public healthcare facility, and a single consultation fee in a private healthcare setting is approximately R850.00 (about US$85).

An audit was conducted between 2007 and 2008 assessing the number of cases counselled in Cape Town and Johannesburg. The findings show that in 2008, of 3365 cases seen, individuals from Black ethnic groups made up 28 % of cases seen in Cape Town and 65 % of cases seen in Johannesburg. Individuals from Mixed Ancestry populations represented 63 % of the cases seen in Cape Town and 7 % of the cases seen in Johannesburg (Kromberg et al. 2013b). Of the 3365 cases seen, 1316 cases had been counselled by genetic counsellors, and 2049 cases had been seen by clinical geneticists. Results from this study also show that 85 % of the individuals had been counselled in state tertiary level healthcare facilities in Cape Town, and 75 % of individuals in such facilities in Johannesburg (Kromberg et al. 2013b).

Purpose of the Study

Although genetic counselling services have grown in South Africa, very few studies have assessed client satisfaction. It is thought that evaluating genetic counselling sessions is useful since this can provide insight into the effectiveness of service delivery. Understanding the experiences of users of genetic counselling services could lead to improving the quality and effectiveness of such services, and to the development of an approach that would be suitable for all individuals, regardless of their cultural or linguistic background.

Two previous assessments of client satisfaction conducted in the 1980s in South Africa showed a positive client experience; however, the majority of the participants in both studies were Caucasian (which could be attributed to the prevailing situation in South Africa at the time) (Kopinsky 1984; Levy 1989). With regard to participants from other ethnic groups, issues such as language barriers, as well as offering a first world service in a third world setting were noted (Levy 1989). Although South African counsellors are aware of cultural diversity, studies have revealed that modes of inheritance and medical concepts are poorly grasped by individuals presenting for genetic counselling (Solomon et al. 2012). Challenges can also arise in counselling since some people may also hold a variety of cultural beliefs regarding the causes of genetic conditions, for example, that affected individuals are forms of punishment to families from their ancestors; conditions arise because of transgressions such as eating forbidden food or marrying across different cultural groups; or that a mother got a "fright" during pregnancy causing their child to be disabled (Penn et al. 2010; Solomon et al. 2012). Together, these findings suggested that further investigation into the use of western genetic counselling models, as well as the evaluation of the experiences of individuals from different ethnic groups in South Africa are needed.

This study, therefore, aimed to describe and document the experiences of Black mothers of children with genetic conditions when receiving genetic counselling in a state hospital facility in Johannesburg, South Africa. This included the types of experiences, what mothers had found the most and least helpful with regard to genetic counselling, and what outside factors influenced their perception of the service.

Methods

Design

The study design was qualitative and drew on the principles of Interpretative Phenomenological Analysis (IPA). IPA was suitable for this study as the method does not depend on a predetermined hypothesis and utilises small, purposively-selected samples in which participants are prompted with open-ended exploratory-type questions (Smith et al. 2009). A flexible question guide is used to encourage 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 et al. 2001; Smith et al. 2009). Focus groups were used for data collection as this method has been successful in a previous South African study in a similar context, as it simulated rapport amongst individuals from the same background, thus encouraging participation (Penn et al. 2010).

Ethical approval for this study was granted on 27/01/2012 by the Human Research Ethics Committee (Medical), University of the Witwatersrand (Ethics Clearance Code: M111160).

Participants

The Division of Human Genetics, National Health Laboratory Service (NHLS) and the University of the Witwatersrand have

Genetic Clinics that are run at tertiary level state academic hospitals in Johannesburg, namely Chris Hani Baragwanath Hospital (CHB), Charlotte Maxeke Johannesburg Academic Hospital (CMJAH) and Rahima Moosa Mother and Child Hospital (RMH). For this study, purposive sampling was undertaken using the file-records of patients seen at these Genetic Clinics. Patients who use these clinics are mostly from the Black population.

The study population comprised Black mothers (from various local, Southern African Black ethnic groups), with a child with a genetic condition, who had received genetic counselling (by a genetic counsellor or a clinical geneticist) at one of the state-run Genetic Clinics. The selection criteria for the study were mothers who: had a live child younger than 5 years of age diagnosed with a genetic condition, resided in Johannesburg; received counselling between 2010 and 2012, were conversant in isiZulu, isiXhosa, seTswana and/or seSotho, and who had easy access to the study site. The exclusion criteria were: mothers who had children that had died or who had lethal conditions, and mothers living outside of Johannesburg.

Mothers who met the criteria (n=50), were contacted telephonically by two research assistants and the purpose of the study was explained. Although 22 mothers telephonically agreed to participate, only 13 attended the focus groups. Those who did not attend cited financial constraints, or work commitments, or that their child was sick, as their reasons for not attending the groups, when contacted again afterwards. Of the 13 mothers who participated, nine had children with Down syndrome, three had children with oculocutaneous albinism (OCA), and one woman had a child with haemophilia B. Each woman completed a consent form and received an information sheet before each focus group commenced.

Data Collection and Analysis

Five Black South African women were involved as research assistants (selected as they could speak several local African languages) in the study. Two were Human Genetics staff employed at the NHLS, two were post-graduate students at the University of the Witwatersrand, and one was employed at a private non-governmental organisation. The assistants contacted potential participants and assisted in facilitating the focus groups, as well as doing the transcription and translation of data. The facilitator of the focus groups was a Black psychologist experienced in qualitative interviewing. She also had a qualification in Human Immuno-deficiency Virus/Acquired Immune Deficiency Syndrome (HIV/AIDS) care and counselling, thus had experience in discussing sensitive topics.

A question guide was used to prompt discussion in the focus groups. The question guide (Table 1) consisted of seven open-ended questions and was developed based on the experience of the authors and the focus group facilitator.

Table 1 Focus Group Question Guide

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

Four focus groups were conducted at the NHLS Outpatient Department (OPD) in Braamfontein, Johannesburg. The first focus group was used as a pilot study to assess the suitability of the process and the selected questions, and adjustments were made according to the findings. Information from this focus group, however, was included in the final results presented here. All focus groups were voice-recorded, and took between 60 and 90 min. The focus groups were formed according to the participants' preferred language and the similarity in dialects (isiZulu and isiXhosa speaking participants were grouped together, as were seTswana and seSotho speaking participants). The first author was present in all the focus groups to observe, take notes and to answer any questions. The researcher and focus group facilitator felt that data saturation had been reached after the fourth focus group. Table 2 shows the composition of the focus groups.

Responses obtained from the focus groups were transcribed by two research assistants with the use of a software program called SoundScriber® and then translated into English. All data were transcribed and translated independently, and then cross correlated to ensure consistency. In the transcripts, participants were assigned numbers to protect their identities. The focus group facilitator was not involved in the transcriptions or coding, and the researcher was not involved in the transcribing and translating of data (as she is unable to speak, or understand, a local Black African language).

Thematic content analysis was used to analysis the data within the transcripts. It is a technique used in qualitative research to analyse texts and to sort recurrent topics into "content areas" (also known as "themes" or "codes"). These themes are then further interpreted and cross-compared so as to as to explore and capture the meaning of the phenomenon described (Joffe and Yardley 2004). The first author was the main coder of the data, and the themes found were discussed with the research supervisors (the second and the third authors) to ensure inter-coder reliability. Results were also presented and discussed with other senior researchers experienced in this field so as to ensure academic rigour.

Table 2 Composition of Focus Groups

Focus Group	Date	Number of Participants	Interview Language	Children's Conditions
1	31/01/2012	3	isiZulu	3 Down syndrome
2	02/02/2012	3	seTswana	1 Down syndrome, 2 OCA
3	01/03/2012	4	isiZulu	2 Down syndrome, 1 OCA, 1 haemophilia B
4	01/03/2012	3	seTswana	3 Down syndrome

Results

The experiences of the mothers who participated in this study are represented below in the form of themes. Although several themes were identified, only those that were discussed most frequently amongst the focus groups are presented here. Those discussed in this article are shown in Table 3. Excerpts from the English transcriptions are included in this section to support the selected themes. Although the questions asked during the focus groups were aimed mainly at the genetic counselling experience, question seven asked participants to think more widely about their community. This brought in conversations about their lived experiences; their families and communities, and the South African state healthcare service. This overall finding suggests that genetic counselling cannot be viewed only as a singular personalised experience, but rather one that forms part of an individual's bigger "life-world".

Theme 1: Thrown into the Unknown (n=4 groups)

Based on participants' responses, it seemed that neither the mothers nor their communities knew about genetic inheritance and they were not aware of the genetic services available in Johannesburg. All but two mothers prior to receiving genetic counselling had never heard of the service or of the word "genetic" and did not know the cause of their child's condition. All mothers had been referred by other healthcare professionals, and very few knew what to expect.

> *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.*
> (Mother of a child with Down syndrome)

Table 3 Main Themes Identified

Theme 1: Thrown into the Unknown
Theme 2: A Worthwhile Experience
Theme 3: A Break in Communication
Theme 4: Telling the Family and the Community
Theme 5: Spreading the Word

The results showed that most of the mothers in this study prior to having had genetic counselling, had very little, or no knowledge of genetic conditions and services. Two mothers stated that unless an individual or a family member had been to genetic counselling, people in the community would not have heard of it before.

> *It's not popular. You wouldn't get to find out about genetic unless you were in that situation.*
> (Mother of a child with OCA)

Theme 2: A Worthwhile Experience (n=4 groups)

Although there were mixed reactions towards the experiences of genetic counselling amongst the focus group participants, all but three mothers described a generally positive experience. Reasons for satisfaction included: receiving adequate information; the use of understandable language and diagrams by counsellors/geneticists to explain concepts; being given contact numbers and pamphlets to take home; referral to support groups; allowing questions; and offering emotional support. Many of the mothers seemed to value the information provided, and most were able to explain the basic clinical features, inheritance patterns and genetic cause of their children's condition during the focus groups. Most of the mothers, who had a good experience, also commented that they had been respected in the session, and that the genetic counsellor/geneticist had shown compassion and empathy towards them, unlike some of the healthcare staff from other state healthcare services.

> *Firstly, the people they know how to communicate with us and it was the first time seeing healthcare professionals who are like this…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…*
> (Mother of a child with Down syndrome)

Receiving adequate support seemed to be a major contributing factor to the mothers' positive experience, particularly those who had children with Down syndrome. Five mothers appreciated the referrals made to community based support

groups, and felt that continued long-term support from genetic counsellors would also be beneficial.

So that is why I say that the support group has also helped a lot, you meet up with other mothers, one tells 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.
(Mother of a child with Down syndrome)

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?
(Mother of a child with Down syndrome)

The mothers who had a child with either OCA (3) or haemophilia B (1), however, felt that one genetic counselling session had been enough and that they had received sufficient information.

Theme 3: A Break in Communication (n=2 groups)

Three of the mothers in this study reported having a negative genetic counselling experience. Reasons for this included poor counsellor-patient communication such as language barriers and misunderstanding the questions asked during the consultation, a lack of privacy, as well as inappropriate timing of genetic counselling (including the length of the session, as well as the period at which counselling was received). Some of the issues raised were not unique to the genetic counselling services, but also applied to other services within the South African state healthcare system.

Some of the mothers said that healthcare staff (including genetic counsellors) often could not converse with patients in their mother tongue and a second healthcare professional (often a nurse) would have to assist with the communication process by translating and explaining the information to them. One mother described her anxiety and difficulty in trying to communicate with some of the doctors at the hospital when she first learned about her child's diagnosis of Down syndrome.

I felt like I could cry so much…this then caused them to look for a nurse for me…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…then I ended up accepting that my child has Down syndrome.
(Mother of a child with Down syndrome)

One mother, who has a child with OCA, had not understood the relevance of some of the questions asked during the initial genetic counselling consultation and had found the session very long with too much information. She has also thought that some of the inquiries about her family members had been intrusive.

But you know sometimes…the doctors want information…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?
(Mother of a child with OCA)

Another mother with a child with OCA described feeling uncomfortable that several students had observed her genetic counselling consultation and that her confidentiality had been breached.

When I went for that… uhm counselling… 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…it is very uncomfortable.
(Mother of a child with OCA)

In this study, two women who had children with Down syndrome were only counselled about their children's diagnosis when their children were 8 months and 2 years old, respectively. They thought that receiving information at such a late stage was stressful and they had been worried about their children's unexplained slow development. These mothers, together with 10 others, felt that the best time to receive genetic counselling was soon after the birth of a child. However, one mother who had a child with Down syndrome felt that receiving counselling 3 days after his birth had been very overwhelming because at the time she was still in shock about the diagnosis.

Theme 4: Telling the Family and the Community (n=4 groups)

Family and community members played a significant role in the mothers' lives. The mothers' views of their genetic counselling experiences and their willingness to accept their child's condition seemed to be influenced by these individuals' understanding of the service, their level of support, and their attitudes towards the genetic condition. Ways of communicating and interacting with family and community became a dominant topic in every focus group.

Six of the 13 mothers acknowledged that their families were very supportive and openly accepted and loved their child, which assisted them to cope with their situation.

Well, wherever I go, they receive my child gladly.
(Mother of a child with Down syndrome)

The focus group participants' responses illustrated that some family members are important support figures and may assist a mother in conveying information to other family members. One participant, who has a child with OCA, had found it beneficial that her mother had accompanied her to genetic counselling and thereafter explained the condition to the rest of her family. Some mothers, especially those who had children with Down syndrome, admitted that it was difficult to discuss the genetic information with their family, as some members could not understand, or would not believe them.

> *…in genetic counselling where they explained things to 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?*
> (Mother of a child with Down syndrome)

It seemed that one family member's erroneous belief can also greatly influence other family members' perceptions about a child's condition. One mother seemed frustrated because her mother-in-law, who had accompanied her to the consultation, had been dismissive about the information provided.

> *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.*
> (Mother of a child with Down syndrome)

There were mixed feelings amongst the mothers regarding the community's perception of an individual who looks different from other people, or is intellectually and/or physically disabled. Six mothers said that the community was very accepting of their child and they had felt comfortable to introduce their child to strangers and people in the community. However, other participants felt differently, with some not disclosing their child's genetic condition to their neighbours because of a fear of discrimination.

> *It's just that…some people, you know, discourage people and speak foul words.*
> (Mother of a child with OCA)

Those mothers, who were more cautious about negative community perceptions, said that if they were asked about their child's condition they would avoid talking about it. They also spoke of stories that they had heard of other families hiding their disabled children, of mothers poisoning them, and fathers ending the relationship with the mother, due to the child's condition. Four of the mothers who had children with Down syndrome also said that, like themselves, some community members did not know what the condition was when it is first mentioned to them.

> *Some people don't know what Down syndrome is as I didn't know what is was.*
> (Mother of a child with Down syndrome)

Overall, these disclosures in the focus groups suggested the community may have a limited understanding of the causes of genetic conditions and that disability may be a frightening concept to some individuals.

Theme 5: Spreading the Word (n=3 groups)

The lack of awareness of genetic conditions and genetic services amongst the community and other healthcare professionals caused much discussion within most of the focus groups.

> *I think it [genetic services] 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?*
> (Mother of a child with OCA)

Five mothers said that they knew of other affected children in the community whose families did not know the cause of their conditions, and it was mentioned that healthcare workers, particularly those in smaller satellite clinics, did not always recognise or know how to manage children with genetic conditions. The participants felt that if the community, as well as healthcare professionals, were more knowledgeable about genetic conditions, parents would be more prepared, children with genetic conditions would be better managed, and the emotional stress placed on families could be alleviated.

In summary, the findings from this study showed both positive and negative genetic counselling experiences. Some of the factors which assisted in creating a positive experience included: respect, compassion and support towards the mother and the relatives, the provision of adequate information at an appropriate time, and the referrals made. Factors that contributed to a negative experience included: the lack of confidentiality in some consultations, asking patients personal questions, lengthy consultations, language barriers, mothers' challenges in conveying information to family members without the presence of a genetic counsellor, and the stress placed on worried mothers when genetic counselling was provided late. It was also noted that the mothers struggled to separate their genetic counselling from other lived experiences, and that their experiences are influenced by those from other

healthcare services, as well those occurring in interaction with family and community members.

Discussion

The results of the present study provide valuable insight into the experiences of mothers, who have children with genetic conditions, and are counselled at state run hospitals in Johannesburg, South Africa. The mothers' perception of their child's condition, and genetic counselling, seemed to be influenced by encounters with other health services, as well as those with their families and community members. This could suggest that genetic counselling is viewed within a broader social construct. Community and family values as well as particular healthcare settings have been shown to influence individuals' perceptions of genetic counselling services (Kinney et al. 2010). It has been noted in interviews with women from African-American and Hispanic communities that genetic knowledge is often understood through their own prior knowledge, as well as their familial and cultural viewpoints (Hurst et al. 2011). Genetic counsellors need to be aware of these social influences since genetic counselling involves not only the health and emotional well-being of one individual, but often of their whole family (Resta et al. 2006).

When focusing on the particular findings from this study, it was noted that the majority of the mothers did not know what genetic counselling was, or why they had been referred. Most of them had also not heard of their child's genetic condition before. This lack of knowledge has been shown in previous South African studies (Levy 1989; Solomon et al. 2012). Additional genetic counselling funding, marketing and awareness are required amongst the South African community. Genetic counselling services, together with government and healthcare officials, need to make a greater effort to create awareness within the community through the distribution of pamphlets and using local platforms such as schools and community centres to host awareness days. In addition, local clinics need to be informed through educational days. Furthermore, integrating knowledge about genetic conditions and services into the training curriculums of allied healthcare professionals may be beneficial. These marketing and awareness suggestions, however, require further study.

The mothers had different experiences of genetic counselling. Those who described positive experiences (10/13) had felt respected, that the counsellors showed empathy, and that their questions had been answered. Satisfaction with genetic counselling services has been described in other studies (Aalfs et al. 2007; Levy 1989). Showing respect, providing empathy, giving attention, and listening are all essential primary skills needed to provide effective communication during a patient-health professional consultation (Epner and Baile 2011). Also, mothers, particularly those with children with Down syndrome, appreciated referrals to support groups and the offer of further assistance from genetic counsellors. However, the few mothers in this study who had children with haemophilia B and OCA did not feel the need for further support. These results reflect findings from other research which show that parents of children with intellectual disabilities experience greater levels of stress and difficulty in coping, than those who have non-disabled children (Hassall et al. 2005). On-going support may be beneficial for some parents who need continued advice, guidance and care (Collins et al. 2001; Muggli et al. 2009). Another explanation could be that since OCA and haemophilia B are conditions that are more easily identified and understood within the South African community, the mothers of these children may have found one counselling session sufficient. This study, together with a previous local study, found that Down syndrome is often not recognised within the Black South African community, and that there seems to be no local name for the condition (Christianson and Kromberg 1996). This may explain why the mothers who had children with Down syndrome wanted on-going assistance and guidance from counsellors and support groups, as they may have felt overwhelmed and anxious by this "unknown" condition and did not know what to expect for the future. Although an effort is made in the Genetic Clinics to see individuals for follow-up sessions, it has been challenging to accommodate all patients due to staff shortages. Referrals to existing support groups, where more experienced parents are present, should however be continued and encouraged.

Mothers who had a negative experience mentioned language barriers, long consultations, disapproval of personal questioning, and disrespect for confidentiality as reasons. Language diversity has been studied previously, and recommendations for professionally trained interpreters have been made to overcome potential communication challenges (De Maesschalk et al. 2011; Solomon et al. 2012). Genetic counsellors also need to be sensitive to the amount of information provided to patients as previous research has suggested that "information overload" occurs in some consultations (Collins et al. 2001). Confusion may have arisen in some sessions because the counsellor did not explain the purpose of obtaining a family pedigree, or the patient may have had a poor understanding of heritability. Some individuals may also understand the term "family" in different ways, and may consider extended family members or non-related individuals living together in the same household as family (Peterson 2005; Richards 1996). It is also important to note that some individuals dislike conveying sensitive information about their families to strangers (Saleh et al. 2009). South African counsellors need to be aware of these issues, and further study is needed to assess the value of information, as well as the manner in which it is presented in consultations for

individuals from various cultural, familial and linguistic backgrounds. In doing so, an approach can be developed to provide genetic counselling information that is more appropriate in the South African context.

In South Africa, genetic services are offered at tertiary level teaching hospitals, and genetic counselling students, as well as medical students, often sit in and observe consultations. Researchers have shown that patient confidentiality may at times not be respected in these settings, which is disconcerting for some patients (Vivian et al. 2011). Although students need to be given the opportunity to learn from observed patient consultations, both students and their mentors need to be considerate of the patients' privacy. This approach may assist in improving the overall genetic counselling experience for sensitive individuals.

The timing of genetic counselling appeared to be an influential factor on how individuals experience the information and service. Early genetic counselling may be beneficial, since some mothers described their anxiety about only receiving information about their child's condition long after birth. Mothers may notice their child's developmental delay but do not know the cause, which can result in great distress (Collins et al. 2001). Receiving adequate information soon after an affected child's birth may facilitate emotional support, decrease concerns about or regarding an unknown condition, and assist parents to gain a sense of control.

This present study showed that while some mothers said that their families gladly accepted their children, others had a challenging time telling their relatives about the child's diagnosis. Generally, parents are responsible for conveying genetic information to relatives; however, the present study together with others, have shown that this can be a stressful and challenging process. Some researchers believe genetic counsellors need to have a better understanding of family communication and dynamics (Peterson 2005; Saleh et al. 2009). If counsellors can explore these issues during genetic counselling, equip parents with appropriate wording, and assist them in conveying information to their family members, then it may help alleviate their stress. Research has shown that if the family members can accept a child's condition and offer a support network, then parents generally cope better (Peterson 2005).

With regard to community views, some mothers stated that most of their community members openly supported their affected child and were determined that their child be treated "normally." This type of support has been reiterated in other research where parents stated that their communities accept their children and try to treat them as ordinary citizens (Greeson et al. 2001). Other mothers however, felt that the members of their communities were less accepting of children with disabilities. In South Africa, the cause of genetic conditions may not be understood, and people may believe that children who are developmentally delayed and/or physically different represent forms of punishment for their parents because of cultural or ancestral transgressions (Penn et al. 2010).

Greater awareness of genetic conditions and genetic services was emphasised as important by the mothers in this study, not only amongst the public, but also among healthcare professionals, particularly those in smaller satellite clinics. This need for awareness has been described in local research (Solomon et al. 2012). A better understanding of genetic conditions and an improvement in relevant awareness and education, together with governmental input, is vital to ensure appropriate acceptance and care of affected individuals.

Study Limitations

One of the limitations of the study was that it only described and documented the experiences of a small group of mothers of children with a few genetic conditions. Also, most of the participants had a child with Down syndrome. The research only focused on one population and only interviewed a few (n =13) individuals within that group, and participant demographics were not assessed. Thus, the findings cannot be generalised to either the recruitment population or to the wider South African community. Further, the researcher was unable to converse in an African language, and could not understand the conversations in the focus groups. This limited the transparency of the data collection and analysis, as the researcher had to rely on the skills and translations of the research assistants. Self-selection by mothers into the study could also have biased the findings as it is unknown how participants differ from non-participants. Lastly, the use of a semi-structured interview protocol prevented the focus group facilitator from probing further into some comments, and some participants may have restricted their disclosure given the group setting, thus some of the deeper meanings might have remained unexplored.

Research Recommendations

Recommendations for future research include exploration of the experiences of individuals seen for genetic counselling for other genetic conditions in other South African settings, using larger sample sizes and participants from different ethnic backgrounds. The nature of the information given during genetic counselling, and counselling methods, also needs to be investigated. Further research to assess which approaches are most suitable in the local context may contribute to improving the genetic counselling experiences for individuals from diverse cultural and linguistic backgrounds in the future.

Practice Implications

Based on the present study findings, the implications for the practice of genetic counselling locally are that genetic

counsellors need to be aware of how genetic counselling experiences are influenced by individual's life experiences, so that they can modify their counselling techniques. Also, they need to provide adequate information in a respectful and understandable manner, and they need to improve co-ordination amongst genetic services and other healthcare services to ensure early diagnosis and appropriate counselling for genetic conditions. Ways of improving information provision not only during the consultations, but also to family members and communities, need to be addressed. Discussions should be held with parents during genetic counselling consultations on how they plan to tell their relatives about their child's condition, and counsellors could encourage parents to invite their relatives to counselling sessions. Training and employing professional interpreters at healthcare facilities is highly recommended, as is the development of genetic awareness campaigns. Together, these actions may not only improve the communities' and health professionals' awareness of genetic conditions, but may also decrease stigma and assist in alleviating parents' anxiety when they have to integrate their affected children into society.

Conclusion

New insight has been gained regarding South African mothers' experiences of genetic counselling in this study. Genetic counsellors need to be aware of the factors that result in both positive and negative genetic counselling experiences. Each individual is unique in how they perceive genetic counselling, what emotional and educational needs they bring to a consultation, as well as when they require such sessions. The findings show that a greater effort is needed to create public awareness of genetic services and genetic conditions in South Africa. The information provided to individuals during genetic consultations should be locally and personally relevant, and on-going patient support may be required, particularly when explaining the condition to family and community members. As South Africa is a multicultural and linguistically diverse country, cultural sensitivity in genetic counselling, as well as the provision of trained interpreters, is important to ensure that individuals derive benefit from the service. Although the findings of this study cannot be generalised to all individuals seen for genetic counselling, and therefore further research is warranted, valuable knowledge has been obtained on how mothers experience genetic counselling. It is anticipated that providing these findings to local genetic counselling training programmes and services, together with the recommendations, will assist in improving genetic counselling services in Johannesburg, and South Africa in general.

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Conflict of Interest Megan Morris, Merlyn Glass, Tina-Marié Wessels, and Jennifer G.R. Kromberg declare that they have no conflict of interest.

Informed Consent Informed consent was obtained from all participants included in the study.

Research Involving Human Participants and/or Animals All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000.

The study was approved on 27/01/2012 by the Human Research Ethics Committee (Medical), University of the Witwatersrand, Johannesburg, South Africa (Ethics Clearance Code: M111160). No animal studies were carried out by the authors for this article.

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