Returning Individual Genetic Results to Research Participants: Experiences of Stigma in South African Families with Neurodevelopmental Disorders

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CHTNOL001

Submitted to the University of Cape Town

In partial fulfilment of the requirements for the degree

MSc (Med) Genetic Counselling

In the

Faculty of Health Science

Date of Submission: 20 January 2024

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NRF National Research Foundation

Declaration

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

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Abstract

In South Africa, there is little research about the experiences of stigma in parents or caregivers of children with a Neurodevelopmental Disorder (NDD). Likewise, the impact of genetic attribution on these experiences have not been extensively studied. The aim of this study was to explore the experiences of stigma of NeuroDev research participants and their families and how a positive genetic result influences/affects these experiences. To achieve this, the researcher looked at the experiences of parents or caregivers of NeuroDev research participants with stigma and the influence a positive genetic result may have on stigma experiences. The role of genetic counsellors in addressing instances of stigma in NDDs and genomics research was also considered.

The study drew on an interpretive phenomenological approach (IPA) as this approach views individuals as "experiential experts" where researchers engage and interpret the meaning of experiences. This research was a sub-study to the NeuroDev study. The NeuroDev study aims to account for the lack of African genetic data of NDDs by exploring the phenotypic and genetic landscape of NDDs in South Africa and Kenya. Participants of the study included fathers, mothers or caregivers of children diagnosed with an NDD, who have voluntarily participated in the NeuroDev study and who have received a positive genetic result. This is a form of purposive sampling as participants were selected based on appropriateness for the aim and objectives of this study. Data collection and exploration involved semi-structured interviews with six participants to ensure that the data obtained was deeply associated with personal experiences, views and beliefs. Interviews were conducted at Red Cross War Memorial Children's Hospital or at the University of Cape Town between March 15th to April 14th 2023 and were audio-recorded. Interviews continued for 45-90 minutes so that the researcher obtained rich, meaningful data. Transcribed interviews were then analysed using an IPA-tailored thematic analysis flow to identify recurrent themes or patterns in the data.

Findings show that participants experience public, associative and internalised stigma. It was further found that how a parent understands a diagnosis, the difficulty in raising a child with an NDD, and respective coping mechanisms affect how patients perceive a genetic result and, subsequently stigma. While a genetic result did not impact public stigma, some patients expressed that assigning meaning to a genetic cause impacts internalised stigma.

Participants understanding of genetic counselling differed. Most expressed a lack of understanding of genetic counselling. Nonetheless, the genetic counselling session enhanced participants knowledge on NDDs, is patient-centred and impacted internalised stigma. It was expected that genetic counselling would have a greater impact on other forms of stigma, however, it was not seen in this study. This study forms a basis for future research to further elucidate experiences of stigma in parents or caregivers of children with NDDs and the role genetic counsellors may play in addressing this stigma and in genomics research in South Africa.

“Only through proper education, can we then, begin to remove the social stigma that’s connected to those with disabilities”

- Robert M. Hensel

Dedication

I dedicate this dissertation to:

My Mom and Dad, Priscilla and Noel Chetty

&

My dearest cousin, Carlo Rajha.

Thank you for being my why.

Acknowledgements

To my participants, thank you for sacrificing your time to partake in my research. Without you, this would not have been possible.

To the NRF, thank you for awarding me a scholarship to complete my studies. It has been my dream to complete my MMed in Genetic Counselling. Without this funding, I may not have been able to complete my MMed and this research.

My supervisor, Dr Zandre Bruwer. Thank you for the guidance, motivation, and for reminding me of my capabilities. You have truly made this experience one to remember and I am eternally grateful for all that I have learnt from you.

To Prof. Tina-Marie Wessels. Thank you so much for dealing with my drama during this time. I truly appreciate all that you have done for me and helping me every step of the way.

To my co-supervisors, Prof Jantina de Vries and Prof Kirsten Donald. Thank you for all the support and constructive criticism. It has been a pleasure meeting you and working with you on my research.

To my colleagues, Nabeelah, Sesethu, Samantha, Lize, Monique, Aphiwe and Lesego. Thank you for listening to me when I was overwhelmed, for understanding my ramblings and inspiring me day-by-day.

To my fiancé. I love you. I love you for believing in me when I could not believe in myself. I am eternally grateful for your undying support, for dealing with my stress-related mood swings, for loving me unconditionally and for doing it all while being 1430 km away.

To my mom. I know this was not easy for you because you went through all my stress with me. Maybe a little more because you had the stresses of being a mom. Thank you for never failing me. For believing in me and reminding me that I'm only human. Thank you for grounding me while ensuring that I have the wings to fly high. I love you and I could not have done this without you.

To my dad. I love you more than you know. Thank you for supporting me and loving me unconditionally. You have always pushed me to be the best I can be and I hope you are proud

of me. Thank you for the comfort, for always believing in me and going out of your way to make me feel better.

To the Rajha's, Uncle Thegy, Aunty Beverly and Tia. Over the past few years, you have become important members of my support structure. Thank you for the pep talks, for the behind-the-scenes support, and for making sure I live my life with no regrets.

To my almost family, Yuthika, Aunty Sandhia, and Uncle Rabbichund. Thank you for reminding me that I am capable of anything.

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List of Abbreviations

ACMG-	American College of Medical Genetics and Genomics
ADHD-	Attention/Deficit Hyperactivity Disorder
AMP-	Association for Molecular Pathology
ASD-	Autism Spectrum Disorder
CD-	Communication Disorder
DSM-5-	Diagnostic and Statistical Manual of Mental Disorders, 5th edition
GDD-	Global Developmental Delay
H3Africa-	Human Heredity and Health in Africa
HREC-	Human Research Ethics Committee
ID -	Intellectual Delay
IF-	Incidental Findings
IPA-	Interpretive Phenomenological Analysis
NDD-	Neurodevelopmental Disorder
NIH-	National Institutes of Health
NHRD-	National Health Research Database
RCWMCH-	Red Cross War Memorial Children's Hospital
RHD-	Rheumatic Heart Disease
SF-	Secondary Findings
SLD-	Specific Learning Disorder
UCT-	University of Cape Town
VUS-	Variant of Uncertain Significance
WES-	Whole Exome Sequencing

Tables and Figures

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1 Chapter One: Literature Review

1.1 Chapter Introduction

In this chapter, a comprehensive overview of the available literature on subjects pertinent to this study is provided followed by the rationale, aims and objectives.

A noteworthy gap in the literature was found regarding parents and caregivers of children with neurodevelopmental disorders (NDDs) experiences of stigma in South Africa. Additionally, there was minimal literature concerning the effect genomic research and genetic attribution had on stigma in South Africa. Existing literature was paradoxical suggesting that further research is necessary to contextualise stigma, genetic attribution and NDDs. Literature on these topics is presented in this chapter.

The role of genetic counsellors in addressing stigma and in genomics research is poorly understood. Thus, literature presented expresses the need for further investigation.

Several research databases were used to acquire the literature presented in this chapter. These databases include Google Scholar, PubMed, ScienceDirect, and Scopus. Terms or phrases used to search for literature included variations of the following: neurodevelopmental disorders, autism spectrum disorder, intellectual delay, attention deficit/hyperactivity disorder, prevalence, stigma/stigmatisation, genetic essentialism, genomics research, genetic attribution, South Africa/Africa, parental perspectives/experiences, genetic counsellors.

Issues addressed in this chapter are presented under the following headings:

- Neurodevelopmental disorders in South Africa/ Globally
- Stigma
- Genetic Attribution and its Association with Stigma
- Genetic Counselling in Genomics Research

1.2 Neurodevelopmental Disorders in South Africa

1.2.1 What are Neurodevelopmental Disorders?

NDDs are a group of diverse disorders that manifest in childhood and are clinically diagnosed following paediatric, psychiatric, and specialist evaluation (Blesson & Cohen, 2020). NDDs are not clearly defined but are known to disturb developmental domains, therefore affecting gross or fine motor, cognitive, speech, and social skills (Blesson & Cohen, 2020). NDDs are not considered independent entities, but rather coexist, forming a continuum with profound variability. According to the Diagnostic and Statistical Manual of Mental Disorders, 5th edition (DSM-5), NDDs comprise of: intellectual disability (ID); global developmental delay (GDD); autism spectrum disorder (ASD); attention deficit/hyperactivity disorder (ADHD); communication disorders (CD); motor disorders (Incl. tic disorders) or specific learning disorders (SLD) (Frances et al., 2022; Savatt & Myers, 2021).

1.2.2 Hardship Associated with Neurodevelopmental Disorders

Parents note great medical, behavioural and social hardship that results from caring for children with rare NDDs (Gobrial, 2018; Currie & Szabo, 2020) with a poorer quality of life (Faden, Merdad and Faden, 2023). Parents express heightened stress, depression, and grief over the loss of a healthy child (Woodgate, Ateah & Secco, 2008; Lai et al., 2015). Other studies highlight financial complications along with difficulty finding the balance between work and caring for their children (Saunders et al., 2015; Brown & Clark, 2017). These experiences are sometimes accompanied by prolonged diagnostic periods where families traverse between doctors, diagnoses, and trying to manage their children's NDDs (Graungaard & Skov, 2007; Krabbenborg et al., 2016a) This process is also emotional (Carmichael et al., 2015; Bauskis et al., 2022), resulting in anxiety, depression, and grief.

Nonetheless, parents highlight several tools used to cope with the hardships associated with NDDs (Lai & Oei, 2014; Haakonsen et al., 2018; Gona et al., 2016). Parents are found to reframe their children's diagnosis in a more meaningful manner that aligns with their personal views, and spiritual/religious beliefs (Jegatheesan, Miller & Fowler, 2010; Gobrial, 2018). A more well-known form of coping involves seeking support from friends, family members, and support groups (Gray, 2003; Pakenham, Sofronoff & Samios, 2004; Hastings et al., 2005;

Hutton & Caron, 2005; Reddy, Fewster & Gurayah, 2019). Negative coping strategies may also arise which are maladaptive in nature. These include avoidant coping strategies such as minimising and denial (McKee et al., 2004).

1.2.3 Prevalence in South Africa

The prevalence of NDDs in South Africa has been ill-defined, however, there have been a few studies that looked at the prevalence in rural regions. Work by Kromberg et al. (1997) in Bushbuckridge South Africa has estimated a 2.2% prevalence of ID. Five years later, the estimated prevalence of ID in Bushbuckridge South Africa had shifted to 3.56% (Christianson et al., 2002). In the Manguzi sub-district of Kwa-Zulu Natal, the prevalence of childhood learning disorders, cerebral palsy, perceptual disability, and seizure disorders were estimated at 0.83% (17 participants out of the 2036 participant sample) (Couper, 2002). Other data specify a 5.5% prevalence of ADHD in the Limpopo province (Meyer et al., 2004) and, more recently, a 0.08% prevalence of ASD in the Western Cape province (Pillay, Duncan & de Vries, 2021). No other recent statistics, specific to South Africa, were identified.

1.2.4 Genetic Aetiology

Literature has indicated that there are numerous etiological factors that contribute to the development of NDDs (Au, Eaton & Dyment, 2020; Taylor & Polderman et al., 2020). That being genetic, environmental, infectious, and traumatic factors. Traumatic factors involve emotional responses to physical and psychological trauma which influence the development of NDDs (Lewis et al., 2015). With focus on genetic aetiology, NDDs may be monogenic or polygenic, and can be de novo (Levy et al., 2011) or inherited. Alternatively, they may be multifactorial with genetic and environmental factors both contributing to the development of NDDs (Taylor & Polderman, 2020). Genetic causes include copy number variants, indels, and single nucleotide variants (Au, Eaton & Dyment, 2020: Chen et al., 2022).

1.2.5 NeuroDev Study

As there is a lack of statistical data regarding NDDs in South Africa, there is minimal genomic data identifying causal variants in our populations. Karczewski et al. (2020) and Gudmundsson et al. (2022) emphasise the constraints of large genomics databases aimed at describing genomic variation. Little data on genomics databases represent African genetic ancestry. It is

also known that people of African genetic ancestry have more genetic diversity than those of other ancestral groups (Martin et al., 2018). Thus, underrepresentation of African genetic ancestry in genomics research has a great impact on accurate diagnoses and accessibility to healthcare (Popejoy et al., 2018).

To address this, the NeuroDev study aims to expand upon the phenotypic and genetic aetiologies of NDDs in South African and Kenyan populations through large scale genomics research (de Menil et al., 2019). The study has been designed to be clinically relevant and will return pertinent results to South African families and to families living within Nairobi Kenya. A description of pertinent results can be found in Chapter 2.4. Data collected from the study will also be used to aid diagnosis of syndromic developmental disorders in African populations by sharing photos, where appropriately consented, with the National Institutes of Health (NIH) Atlas of Human Malformation Syndromes in Diverse Populations and Face2Gene databases. De-identified phenotypic and genetic data will also be shared in both open and controlled access databases.

Participants for the NeuroDev study were recruited from both South African schools and hospitals. Children between the ages of 2-17 years with a clinical diagnosis of ID, GDD, ASD, ADHD, CD or SLD were considered eligible for participation in the study. These participants then underwent whole exome sequencing (WES) to identify causative variants. Single nucleotide variants and indels were confirmed by Sanger sequencing while copy number variants by array comparative genomics hybridisation. Prior to sample collection, parents or primary caregivers were given the option to indicate their interest in receiving genetic results if a causative variant (positive result) was found (de Menil et al., 2019). There are many reasons why parents may be interested in receiving a genetic result. A Masters-based research study in South Africa found that altruism, hope for a cure or treatment and for personal/family benefit, in terms of recurrence, was often cited by NeuroDev participants (Diedricks, 2020). These findings are echoed by Liang et al. (2022) and Pollard et al. (2021).

Upon conducting genomics research, such as the NeuroDev study, there has been increasing concerns of the ethical implications of genomics research. One of the few concerns noted in the NeuroDev protocol was its impact on “group-level stigmatisation” (de Menil et al., 2019). However, there has been little research on this topic in South Africa (de Vries et al., 2012; Matshabane et al., 2021).

1.3 Stigma

1.3.1 The Stigmatisation Process

Stigmatisation has been described as a societal process by which certain characteristics or traits elicit negative cognitive and behavioural responses (van der Sanden et al., 2013). In seminal work by Goffman (1963), stigma is described as an "attribute that is deeply discrediting". Hence, one's social identity is undervalued in that social setting as their traits are distinct from that considered socially normative (Kurzban & Leary, 2002). Link and Phelan (2001), highlight four components that underpin the stigma process. Firstly, a trait or characteristic is labelled. Labelled traits are then associated with negative stereotypes. Labelled individuals are then grouped leading to separation and lastly, they experience rejection, discrimination, humiliation, disgrace and status loss.

1.3.2 Types of Stigma

Negative attitudes that provoke fear, avoidance and discrimination are referred to as public stigma. Often, those affected with conditions are recurrently stigmatised, and face alienation, rejection, and discrimination (Sankar et al., 2006; Aldersey, 2012). In ancient times, those suffering from mental conditions were perceived as either demonic or possessed. This perception has not drastically changed since then as those suffering from mental conditions are still viewed negatively. Recent work in the United States of America implies that children with ASD are frequently labelled as "dangerous", "weird" or "mentally-ill" (Serchuk et al., 2021; Kinnear et al., 2016). This is exemplified by research looking at stigma experienced by Ghanaian Mothers of children with ASD (Oti-Boadi, Dankyi & Kwakye-Nuako, 2020). It was revealed that other members of the community perceived children with ASD as "violent", "bewitched" and would rather avoid interaction. In some case, parents were deemed as inadequate as ASD is often an "invisible" NDD (Werner & Shulman, 2013). Other studies have shown that children with SLDs are stigmatised as "stupid" and "lazy" (Haft et al., 2019) while children with ID are stigmatised as "aggressive" and "childish" (Werner, 2015).

Public stigma is shifted to parents of children with NDDs due to their relationship with the affected person. This is referred to as association stigma or "courtesy" stigma by Goffman (1963). Work on stigma experienced by parents and children with developmental disorders in

Ethiopia found that parents were socially excluded by family members due to fear of status-loss and shame, faced teasing, and were often blamed for their child's condition. If not blamed, their children's conditions were attributed to a curse (Tekola et al., 2020). Similar findings were observed in research focusing on Ghanaian and Kenyan Mothers of children with ASD (Oti-Boadi, Dankyi & Kwakye-Nuako, 2020). Research looking at the lived experiences of caregivers of children with ASD in Kenya had found that the perceived cause is related to punishment from higher beings, and that one would need to get rid of their child or perform sacred rituals to ease the punishment (Cloete & Obaigwa, 2019). Other causes considered by community members included marrying into the incorrect tribe, supernatural activities, or maternal taboos. Within the nuclear family, blame, fear, and shame may be exemplified by spousal temperament. A study conducted in the Katutura Township near Windhoek found that fathers of children with ID often rejected mothers, questioned their fidelity and ceased to care for the affected child (Ntswane & Van Rhyn, 2007).

Parents or caregivers may also experience associative stigma in alternate aspects of life. They may struggle to find employment that is flexible and allows them to attend their children's regular medical appointments. Coupled with medical bills, parents and caregivers may experience cumulative financial strain (Oti-Boadi, 2017; Broady, Stoyles & Morse, 2017; Brown & Clark, 2017).

Association stigma can also lead to internalised stigma which is when parents or caregivers internalise stigma experiences and incorporate them into their psychological identity (Papadopoulos et al., 2019). This may lead to heightened anger, despair, and a negative self-concept (Cohrs & Leslie, 2017). A study looking at experiences of internalised stigma in parents of children with ASD in Nigeria had shown that parents feel inadequate, out of place and misunderstood (Oduyemi et al., 2021). Parents of children with ADHD recall feeling depressed which ultimately affected their quality of life (Chen et al., 2021; Chang et al., 2020).

These experiences often lead to social withdrawal (Ngo et al., 2012). In some instances, mothers isolate their children as they disapprove of the way other people treat them (Oti-Boadi, 2017). Internalised stigma is amplified in instances where there is lack of support from family members and friends. This creates room to adopt negative stigmas leading to major depression and complicated grief (Chen et al., 2021; Chang et al., 2020).

1.4 Genetic Attribution and its Association with Stigma

1.4.1 What is Genetic Essentialism?

The exponential growth in understanding human genetic constitution has triggered "genetic essentialist" thinking (Phelan et al., 2005). Genetic essentialists believe that genes are regarded as the foundation of one's individual identity or "essence". This includes ones' religious and political beliefs, personality, behaviour, and physicality (Dar-Nimrod & Heine, 2011). It further suggests that genetic characteristics traverse through generations. Those who inherently share genetic information are, thus, viewed as similar beings. Therefore, an association between one's genetic make-up and their behaviour or physicality underpin genetic essentialist thinking. This association results in four consequences which are highlighted by Dar-Nimrod & Heine (2011). Genetic essentialist views result in the belief that genetic characteristics are irreversible and deterministic. It further suggests that the presence or absence of a genetic change solely determines the presence or absence of a characteristic. Individuals who share a genetic constitution are then grouped and lastly, cognitive biases are generated based on natural genetic constitution (Dar-Nimrod & Heine, 2011). These cognitive biases suggest that "unnatural" characteristics, behaviours or physical attributes may be negatively viewed. Like that in stigmatisation, this creates an environment for stereotyping and prejudice.

1.4.2 Genetic Essentialism and Stigma

The rise of genomics research has fostered concerns regarding stigmatisation of conditions with a genetic cause. Literature suggests that these concerns arise as a result of the eugenics movement which largely happened, but is not limited to, the historical past (Donovan, 2022). Eugenics is a pseudoscience aimed at enhancing the genetic pool by selectively breeding for desired traits. Here, traits are categorised into those considered socially "desirable" versus "undesirable". These categorisations are said to support genetic essentialist sentiments, therefore, promoting prejudice and discrimination (Donovan, 2022).

When genetic attributions are considered within genetic essentialist frameworks, it is expected that public understanding of health and disorders may be distorted, resulting in stigmatisation (Donovan, 2022). Work by Tekola et al. (2009) revealed that proposed genetic research for podoconiosis (a chronic inflammatory disease caused by red clay soil exposure) in Ethiopia

has the potential to increase pre-existing stigmas for those affected. Sharing genetic material by means of relation with an affected individual similarly imparts associative stigma (Haslam, 2011). Tekola et al. (2009) further stress that stigma is heightened for family members of individuals affected with podoconiosis due to their familial relation. Internalisations may lead to an altered self-concept or induce shame and embarrassment (Mercer et al., 2006). Research on parental perspectives on the cause of ASD in children has shown that guilt and blame had arisen in family members that believed their genes contributed to their child's ASD (Mercer et al., 2006).

Interestingly, it has also been proposed that genetic attribution may lessen prejudice and discrimination, enhance tolerance or pity (Corrigan et al., 2003), and reduce feelings of blame, and guilt signifying that existing research is paradoxical (Bar, 1999). The main argument supporting genetic attribution and reduced stigma is that shifting responsibility to a genetic cause lessens individual responsibility and subsequent blame or guilt (Bar, 1999). In South Africa, Matshabane et al. (2021) suggests that heightened stigma as a result of genetic attribution in Xhosa people with schizophrenia is improbable and that a genetic cause may ease blame. Work by Faure et al. (2019) details that multiple causal factors play a role in rheumatic heart disease (RHD) stigmatisation in South Africa and that it is unlikely that genetic attribution would increase stigma alone. With this inconsistency, the association between genomics research and stigma is a topic of debate, yet, it has remained understudied in African populations (Faure et al., 2019; Matshabane et al., 2021).

1.5 Genetic Counsellors: Guiding Genomic Research

1.5.1 Role of Genetic Counsellors

Genetic counsellors are internationally recognised healthcare professionals trained to communicate tailored information and facilitate understanding and adaptation to all existing implications of a genetic disease (Biesecker, 2001; Resta et al., 2006). Implications include the risks of disease or recurrence, management, surveillance, the options available to manage these risks, and existing psychosocial entanglements (Biesecker, 2001).

In recent years, they have been integrated into laboratory and genomics research settings where their roles have shifted. Genetic counsellors are required to interpret complex genomics data

and return findings where applicable. This depends on the feedback policy of the respective study. Literature in the United States of America and South Africa suggests that genetic counsellors play a positive role in genetic result delivery (Waxler et al., 2013; Morris et al., 2015). They are shown to convey genetic results in an empathetic and sympathetic manner allowing parents to adjust and adapt to complex information, and a confirmed genetic diagnosis (Macleod, Craufurd & Booth, 2002; Ashtiani et al., 2014) In work by Matimba et al. (2022), it was suggested that genetic results from genomics studies should be delivered by medical professionals with specific preference to genetic counsellors. It has also been suggested that genetic counsellors are involved in pre-test and post-test counselling in genomics research (Krabbenborg et al., 2016a).

1.5.2 Cultural and Ethnic Relevance in Genomics Research

Complex genomic data exists within a realm of variables (Chapple, May & Campion, 1995). Beliefs, meaning and comprehension are largely influenced by one's ethnicity, culture, religion and psychosocial context. Hence, the term ethnocultural, as described by Weil (2000), is understood as a cultural and ethnic "sense of identity" which frames one's beliefs, practices, and worldview. This framework forms the foundations for the stigmatisation process in individuals and communities alike. Studies highlight that the relationship between stigma experiences and causal beliefs traverse rich culture and deep-seated inequalities of South African communities (Faure et al., 2019; Matshabane et al., 2021). Studies quote supernatural interpretations such as punishment for past transgressions (Etieyibo & Omiegbe, 2016; Oti-Boadi, 2017; Bunning et al., 2017; Tekola et al., 2020; Namazzi et al., 2020), medical and genetic interpretations (Elder, 2001; Zuckerman, Lindly & Sinche, 2016), and some utilise multiple explanatory models (Shyu, Tsai & Tsau, 2010; Ravindran & Myers, 2013; Faure et al., 2019; Matshabane et al., 2021; Kengne Kamga et al, 2021). Similarly, the meaning of a genetic result and reasoning of participating in a genomics study may differ between communities.

South Africa is referred to as the "rainbow nation" since it is home to numerous languages and cultures which are observed as diverse and dynamic. According to the 2022 South African National Census, which was published on the 10th of October 2023, the South African population consists of 62 million people in which those of Black African Ancestry constitute 81,4% of the population, those of Mixed Ancestry constitute 8,1% of the population and those

of White European/Afrikaner Ancestry constitute 7.2% of the population. Lastly, those of Indian or Asian Ancestry constitute 2,7% (Department of Statistics South Africa, 2023). In South Africa, those of Mixed Ancestry have a culmination of Indian, Indonesian, Khoi-San and African descent and are predominantly found in the Western Cape (Daya et al., 2013). Within these populations, stratifications exist with reference to ethnicity, language, culture, and religion. Together with South Africa's infamous political and socio-economic past, its dynamic multicultural nature spawns' kaleidoscopic worldviews, preferences, and beliefs on what is and is not socially normative. It further creates stigma experiences that may be distinct from other settings.

Commentary by de Vries et al. (2020) looking at stigma in African genomics research discussed spiritual and supernatural attribution of disease in African communities, gendered blame, polygamy, group customs, and ancestry as sources of stigma. While this work highlights that stigma is often pre-existing rather than provoked by genomics studies, it further suggests that it is necessary for researchers to be culturally and ethnically competent to recognise stigma that may arise in genomics research (de Vries et al., 2020). In the same breath, it is important for genetic counsellors to be knowledgeable on cultural and ethnic differences to ensure equitable provision of genetic counselling, comprehension of complex data, contextual psychosocial counselling and stigma identification and alleviation.

1.6 Rationale

Understanding the effect genomics research has on stigma has been a concern in recent years. However, there has been limited research on the experiences of stigma of parents and caregivers of children with NDDs and how a positive genetic result could affect these experiences in a South African context (Matshabane et al., 2021). The issue lies within the rapid expansion of genomics research and how the public would perceive genetic attribution. Genomics research has been newly introduced in Africa with the NeuroDev study being one of the first of its kind (de Menil et al., 2019). Considering the cultural and ethnic diversity coupled with varying belief systems and inequalities in South Africa, one cannot infer the effect genomics research has on our populations. Rather, one would need to determine how conducting genomics research in South Africa affects our populations. This includes personal experiences of stigma, how a positive genetic result alters these experiences for parents and caregivers of children with NDDs and the prevalent challenges families face. Understanding

these experiences is important to contextualise genomics research in South Africa, understand existing stigmas that parents or caregivers of children with NDDs face and if genomics research impacts these experiences. It may further be used to direct genomics research and result communication in South Africa.

Genetic counsellors engage in patient-centred conversations of genetics, related conditions and respective psychosocial impact with hopes to facilitate patient understanding, management and adaptation to a genetic condition and existing psychosocial complications (Patch & Middleton, 2018). To employ meaningful conversations around the psychosocial impact of stigma and the prospect of a genetic cause, genetic counsellors would require contextualisation in a South African setting. Understanding these social processes in a South African context may define points of concern when considering genomics research in South Africa. It may further assist genetic counsellors in addressing the impact of stigma leading to patient empowerment and adaptation to disease and genetic attribution.

On a more personal level, interest in this topic was sparked by reflecting on personal experiences. The researcher had a cousin with a chromosomal translocation involving chromosomes 14 and 9 which came with many challenges, including ID. The researcher had first-hand experience of public stigma, associative stigma and internalised stigma. This was one of the reasons the researcher chose to be a genetic counsellor at a young age. The researcher wanted to help families deal with these experiences. Therefore, the researcher believed this research was appropriate and necessary to, not only, shed light on stigma experiences in South African populations and how it changes with a genetic result, but to also be aware of stigmas and develop their skills as a patient-centred genetic counsellor. The post research reflection is found in ***Appendix A***.

1.7 Aims and Objectives

1.7.1 Aim

The aim of this study is to explore the experiences of stigma of NeuroDev research participants and their families and how a positive genetic result influences/affects these experiences.

1.7.2 Objectives

To determine:

1. The experiences of parents or caregivers of NeuroDev research participants with stigma.
2. The influence a positive genetic finding may have on stigma experienced by parents or caregivers of NeuroDev research participants.
3. The role of genetic counsellors in addressing instances of stigma in NDDs and genomics research.

1.8 Chapter Summary

Existing literature on subjects pertinent to this study, as well as the aims and objectives were discussed. In the following chapters, the methodological approach and findings of the study will be presented.

2 Chapter Two: Methodology

2.1 Chapter Introduction

In this chapter, the methodological approach employed by the researcher to determine experiences of stigma, stigma experiences after receiving a positive genetic result and the role of genetic counsellors in addressing instances of stigma in genomics research is communicated. The study design, research setting, research participants, recruitment, data collection, data analysis and ethical considerations are discussed below. The reasons for the selection of the approach and a discussion of its appropriateness are provided. The chapter concludes with a discussion of the measures taken to ensure trustworthiness and ethical requirements of the research.

2.2 Study Design

A qualitative cross-sectional study design was applied. Qualitative research focuses on understanding social phenomena in their existing contexts. This includes understanding people's social reality, beliefs, behaviours, or experiences (Pathak, Jena & Kalra, 2013). The researcher considered a qualitative study design as the most applicable for the aims and objectives of this study as it gives a voice to the research participants.

This study was firmly established in phenomenology. According to Husserl's call "to the things themselves", phenomenology aims to describe how one's worldview is formed and concurrently experienced (Eatough & Smith, 2017). Thus, phenomenology focuses on understanding the meaning or essence of participants' contextual and personal experiences (Wagner, Kawulich & Garner, 2012). This study further drew on an interpretive phenomenological analysis (IPA) which views individuals as "experiential experts". This allowed the researcher to grasp the "sense" individuals make of their experience (Eatough & Smith, 2017). IPA utilises three approaches. These are Phenomenology, Hermeneutics, and Idiography. Phenomenology focuses on exploring individuals' subjective experiences while Hermeneutics focuses on making meaning of subjective experiences (Eatough & Smith, 2017). Idiography emphasises the uniqueness of each individuals' experiences such that they may not be generalised across different contexts (Eatough & Smith, 2017). Hence, these approaches

work synergistically to create a holistic, in-depth understanding of the lived experiences of individuals in their respective sociocultural context (Eatough & Smith, 2017).

The study involved the use of in-depth semi-structured, one-on-one, in-person interviews. The interviews consisted of short, open-ended, follow-up and probing questions to obtain experience-rich data, beliefs, and thoughts while maintaining conversation between the researcher and participants (DeJonckheere & Vaughn, 2019). The researcher customised and adjusted the flow of questions to suit individual participants responses, as well as clarified concepts briefly discussed or unclear to the researcher. This created an explorative environment in which the researcher and participants examined significant thoughts, feelings and opinions without compromising the interview structure (Alamri, 2019).

2.3 Research Setting

This research is a sub-study to the NeuroDev Study. In cases where there was a positive genetic result, these were fed back to participants who consented to receive a returnable result. Thus, participants of this study had already received a genetic result that could explain their children's NDD. These results were delivered in the context of a genetic counselling session by a genetic counsellor associated with the NeuroDev study.

2.4 Research Participants

The researcher was interested in the stigma experienced by mothers, fathers and caregivers of children diagnosed with an NDD and how a genetic diagnosis altered this experience. The specific inclusion and exclusion criteria used to determine participant eligibility are listed below.

Inclusion

- Mothers, fathers or caregivers of participants enrolled in the NeuroDev Study.
- Participants who requested the return of their results.
- Participants who had received a positive result.
- Participants with the cognitive ability to consent their participation in this study.
- Participants who have previously consented for participation in supplementary research to the NeuroDev study.

Exclusion

- Participants who have not requested for the return of pertinent results.
- Participants who have not previously consented for participation in supplementary research.
- If participants have not received a positive result.

Within the inclusion criteria, it was highlighted that only participants who had received a positive result were included in this study. A positive result describes pertinent genetic results associated with the NDD, where variants have been classified as pathogenic or likely pathogenic according to the American College of Medical Genetics and Genomics (ACMG) and the Association for Molecular Pathology (AMP) criteria and are considered returnable within the context of the NeuroDev study (Richards et al., 2015). The classification has considered inheritance, allelic data, population data, computational and predictive data, functional and phenotype data. These variants were further confirmed in a diagnostic laboratory on a second sample before the individual research results were returned as per the Human Heredity and Health in Africa (H3Africa) guidelines. The H3Africa guidelines were developed to guide the return of individual genomics research results in Africa (Matimba et al., 2016). Variants of uncertain significance (VUSs), incidental findings (IFs) and secondary findings (SFs) were not returned to patients. VUSs are variants detected in which it is unclear as to whether it is disease-causing or not. IFs are variants detected unexpectedly and are unrelated to the primary indication for testing and SFs are those that are purposefully analysed but are unrelated to the primary indication (Richards et al., 2015).

2.5 Recruitment

Purposive sampling was used to recruit participants based on the outlined criteria. Purposive sampling is a routine technique used in IPA to select participants viewed as experiential experts in the phenomenon of interest, and are best suited for the aims and objectives of the study (Palinkas et al., 2015). During the period of data collection, positive results had been returned to participants from the first year of the study. Potential participants were selected from the NeuroDev contacts database by a NeuroDev team member. Individuals from this list were then contacted telephonically by the researcher at the University of Cape Town (UCT). The

researcher was suitable in this regard since the potential participants had already consented to being contacted for additional research when recruited into the NeuroDev study. A clear description of the study was given highlighting the research plan, eligibility and purpose of the study. Fathers, mothers or caregivers willing to participate in the study were then given a date and time best suited for the participants. This was communicated telephonically, via WhatsApp, text message, or email depending on the participants preference. An information sheet was sent directly to the participants by the researcher via email or WhatsApp, and a physical copy of the information sheet was also given to the participants on the day of the interview (***Appendix B***). This process is illustrated in ***Figure 1***. Twelve families, who met the criteria, were initially contacted and six families consented to participate in this research.

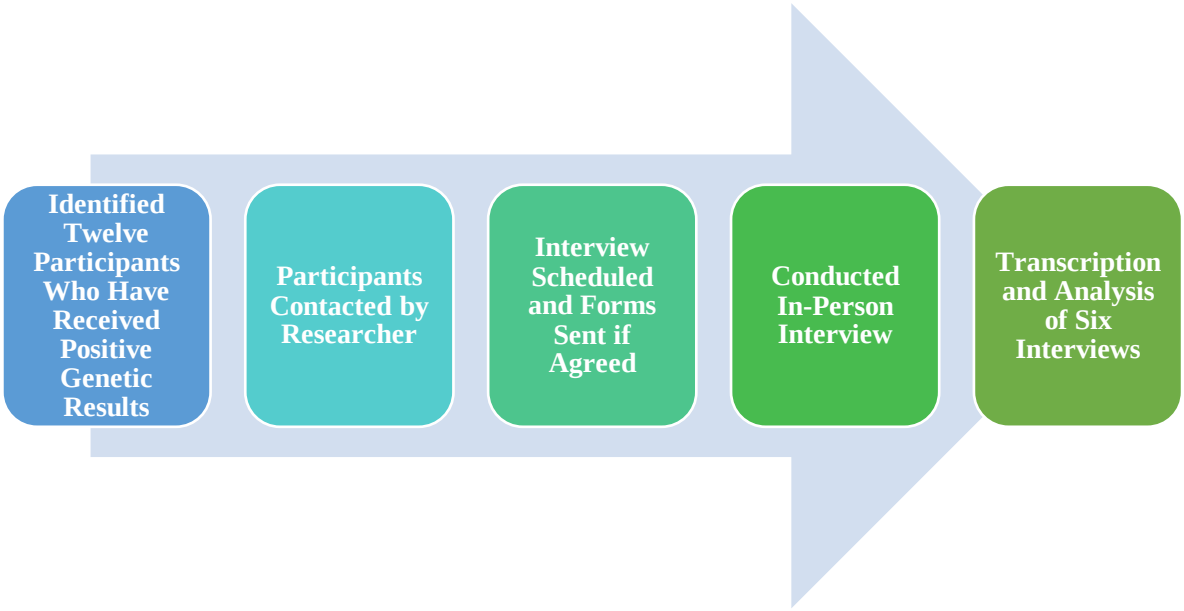

Figure 1: A diagrammatic representation documenting the process of participant recruitment to analysis.

2.6 Data Collection

Data collection and exploration involved in-depth, semi-structured interviews to ensure that the data obtained was deeply associated with personal experiences, views and beliefs pertaining to the aims and objectives of this study (DeJonckheere & Vaughn, 2019). An interview guide was created and piloted with members from the Human Genetics Department and a member of the cohort to ascertain the efficacy of the interview guide, and to make necessary alterations where applicable. After completion of the pilot interviews, points that were confusing, obscure

and redundant were identified and the question guide was refined to better suit the aims and objectives of the study. The question guide was then rechecked by the researcher's supervisors. In addition to serving as a pre-test of the interview guide, it also provided an estimate of the amount of time required to complete each interview. Interviews were conducted in a private room at either Red Cross War Memorial Children's Hospital (RCWMCH) or UCT and were audio-recorded. All the interviews were conducted by the researcher and took place during the months of March and April of 2023. A total of six semi-structured interviews were conducted. These interviews proceeded for 45 to 90 minutes allowing the researcher to obtain rich, meaningful data. The researcher further established rapport and trust with the participant, enabling them to feel comfortable and open about the sensitive topics questioned (Pietkiewicz & Smith, 2014).

Interviews were recorded as this allowed the researcher to focus on the interview, for accurate data capture and served as a means to clarify misunderstandings. It further preserved contextual cues, allowed for thorough analysis and enabled objective transcription (Rutakumwa et al., 2020). Interviews were not conducted until data saturation was reached (where no new data/themes emerge and similar themes recur) since IPA does not depend on data saturation (Saunders et al., 2018). IPA aims to thoroughly and richly examine a phenomenon, therefore studies using IPA often have small sample sizes with recommendations of six to eight participants being recognised as appropriate for an IPA study (Pietkiewicz & Smith, 2014). This is because the researcher does not want to generalise findings to a broader population but rather deeply understand individual experiences. Thus, prioritising depth over breadth.

2.7 Data Analysis

Audio-recorded interviews were employed for data collection. The interviews were then transcribed verbatim and analysed according to the process depicted in ***Figure 2***. There is no definitive method for IPA, however the researcher utilised an adapted version of that depicted by Charlick et al. (2016). The three overarching steps in this study involve Data Familiarisation, Case-by-Case Analysis and In-Depth Analysis. The individual facets of each overarching step are depicted on the following page.

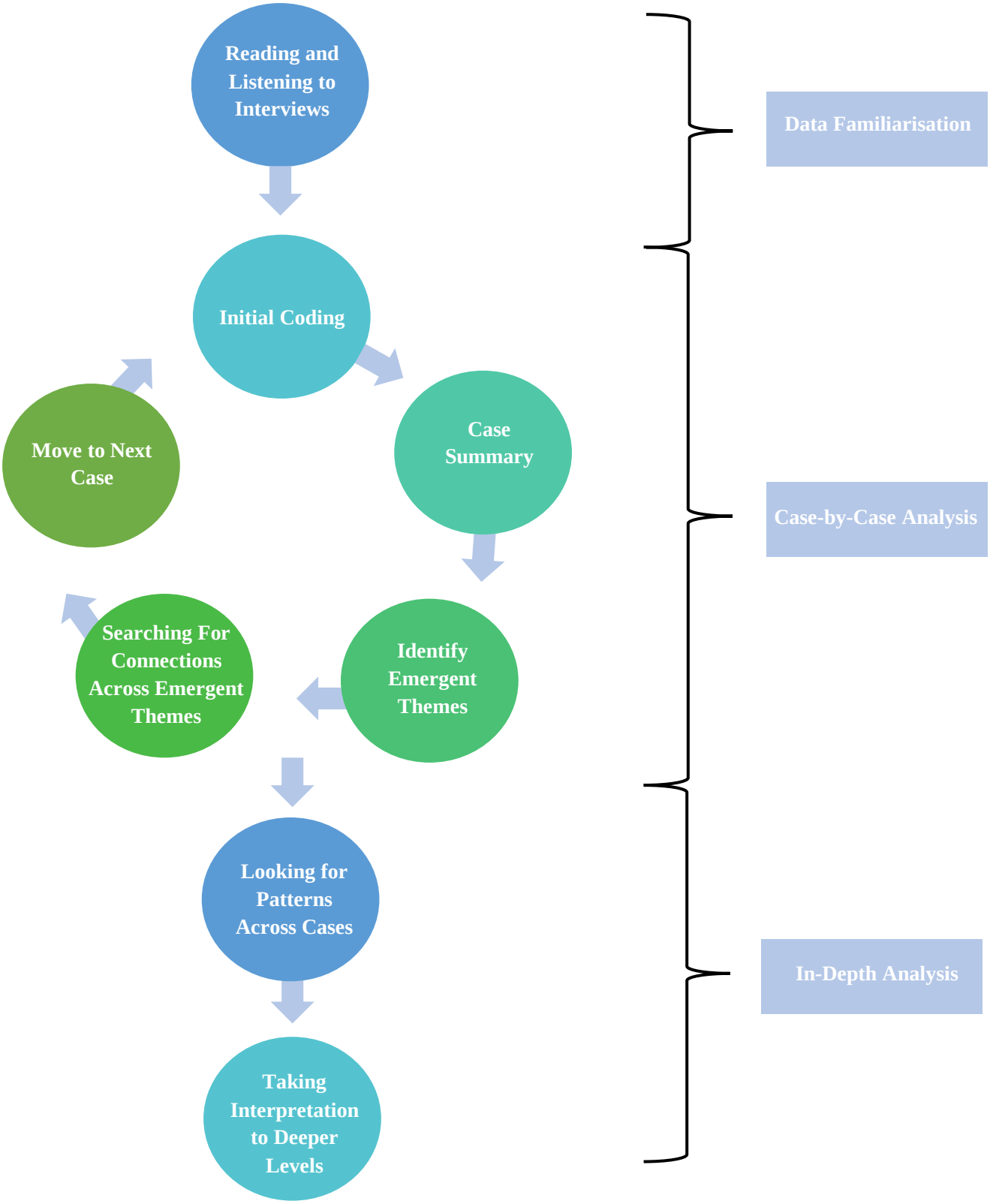

Figure 2: A diagrammatic representation of the IPA analysis approach used in this study.

The researcher familiarised herself with the data by transcribing the interviews, reviewing the transcripts and listening to the audio-recordings on multiple occasions. Each interview was examined individually before collective examination. The first transcript was reviewed by the researcher and specific units of interest; a word, phrase or sentence, with a specific subject, was flagged and identified with a code. A code labelled the topic discussed and defined the meaning of this topic. Reflective and observational notes pertaining to the context, language used, and emotional responses were also made to contextualise responses and detect hidden meanings. The final coded transcript was then reviewed by the researchers' supervisors to ensure accuracy of coding before further analysis took place. A descriptive account of this interview was created to identify the essence and core values. These descriptions were interpreted alongside the codes to examine how a participant made meaning of the phenomena against a societal, cultural and theoretical backdrop. This process was repeated for each of the six interviews. Once completed, patterns across the data were explored and interpreted to make sense of the data. As depicted in the diagram, the analytic approach is dynamic in nature rather than linear and emphasises the layered approach required to attain a profound understanding of the research topic (Charlick et al., 2016). The data obtained was then compared with available literature.

The data was managed and analysed manually. It was believed that this further aided data familiarisation. Quotes that exemplified the meaning of a theme or reflected typical or unusual statements were quoted in the report. These quotes were pseudonymised to protect the identity of research participants.

2.8 Validity and Reliability

Validity was important to demonstrate as it illustrates the accuracy and truthfulness of the results. Several steps were taken to ensure validity and reliability of the research. The interview guide was piloted with members from the Human Genetics Department and study cohort to enhance the significance of the questions asked. Furthermore, the supervisors reviewed the researchers' coding strategies and interpretations to ensure accurate and credible data analysis (Stahl & King, 2020). Differences in coding strategies or perspectives were useful to identify as it allowed the researcher to think abstractly about certain perspectives that may have been missed, thus creating credible and coherent interpretations of the data. Furthermore, the

interviews were recorded and the data was captured verbatim during transcription to enhance the validity by providing an accurate and complete record.

The researcher observed the recruitment and feedback of findings for the NeuroDev study as this allowed the researcher to immerse herself in the participants' context, engage, and reflect on potential feelings or experiences that participants have. This further promoted credibility and trustworthiness of this research as it allowed the researcher to contextualise her research (Stahl & King, 2020). Journaling was used to highlight researcher bias and therefore reduce subjectivity (Cope, 2014). This was done as a form of bracketing as the researcher felt that experiences with families in the genetic clinic may influence their perception and analysis of the results. Lastly, confirmability was shown by reporting rich-quotes that exemplified each theme to further negate researcher bias.

2.9 Ethical Consideration

Approval to complete this study was received from the Human Research Ethics Committee (HREC) of UCT under the reference 734/2022 (***Appendix E***). This approval was extended to December 2024. Further approvals were obtained from the National Health Research Database (NHRD) under the reference WC_202301_006 and RCWMCH Research Committee under the reference RCC 362 / WC_202301_006 (***Appendix F***).

2.10 Informed Consent

The purpose and outcomes of the study were verbally discussed with the participants. Participants were then given an information sheet and consent form detailing the information discussed which were signed by the participant confirming their voluntary participation in this research study and understanding of the research. It was further emphasised that participants have the ability to withdraw from the study at any time due to personal preferences. The information sheet and consent form may be found in ***Appendix C*** and ***Appendix D*** respectively.

2.11 Privacy and Confidentiality

Maintaining privacy and confidentiality of the research participants was an essential aspect of this study. Participants were advised that personal information obtained by the researcher

would remain confidential and that only the researcher would have access to the participants personal details. Interviews were audio-recorded, transcribed by the researcher and pseudonymised. Any quotes or extracts used in the study were pseudonymised as well.

Audio-recordings and transcriptions were stored on the researcher's password-protected laptop and on an external hard drive to ensure good data management. The external hard drive was safely stored by the researcher in a locked cupboard accessible to the researcher only. Participants were further informed that the audio-recordings and transcripts would be reviewed by the supervisors of the study to ensure that the data analysis was accurate. Once the research has been completed and published, the remaining audio-recordings and transcripts stored on the password protected laptop will be discarded appropriately.

2.12 Risks and Benefits to Participants

Qualitative research involves probing to gather comprehensive experiences of participants. It may also elicit overwhelming emotions such as anxiety, distress, guilt, shame or regret leading to psychological harm (Richards & Schwartz, 2002). Therefore, participants were informed that they were not required to answer questions that trigger overwhelming emotions and could withdraw from the research if needed. If participants displayed signs of deeper emotional distress, they would've been referred to appropriate healthcare professionals such as a psychologist or genetic counsellor, however, this did not occur.

Since, interviews were conducted at RCWMCH and UCT, participants were compensated for travel costs to the value of R50 (2.70 USD) per person or the transport was arranged for them. Food and refreshments were supplied for the participants, to the value of R50 per person.

There was no direct benefit to the participants and results from this study did not alter current healthcare regimes. However, the process of sharing deep-rooted experiences was viewed as cathartic as it allowed participants to share challenging concepts, thoughts and feelings in an unbiased environment. Furthermore, this research hopes to raise awareness for the stigmatisation of NDDs and how a genetic cause may alter experiences of stigma. Understanding experiences of stigma of parents or caregivers is important in recognising stigma that may arise in genomic research and genetic result communication in South Africa.

2.13 Role as a Genetic Counselling Student while Conducting Research

In a few instances, participants asked questions related to their children's NDD's or mentioned incorrect medical information. As a researcher, I could not answer the questions participants asked or correct misunderstandings even though I would do so in a clinical setting. In these instances, I would mention that I would not know and they would need to consult with a qualified healthcare professional such as a genetic counsellor.

2.14 Chapter Summary

The methodological approach used to conduct this research was detailed in the above chapter. The following chapter will focus on the findings of the study.

3 Chapter Three: Results and Discussion

3.1 Chapter Introduction

The findings of this study are presented and discussed in this chapter. This format is customary in qualitative research as it prevents unnecessary repetition (McMillan & Schumacher, 2001). The first section considers the experiences of stigma and is discussed in Chapter 3.2. The second section considers the influence that a positive genetic result has on stigma and is discussed in Chapter 3.3. Here, four primary themes were identified, namely "Understanding of the NDD", "Navigating the Challenges of NDDs", "Coping with the NDD" and "Perception of Positive Genetic Result and Their Impact on Stigma". The third section considers the role of genetic counsellors in addressing stigma as part of the genetic counselling process and is discussed in Chapter 3.5. Four primary themes emerged, namely "Resource of Information", "Addressing Stigma", "Relational Aspects of Care" and "Continuity of Care". The text below, will briefly summarise the main findings under each of these themes, and illustrate pertinent observations with direct quotes.

To contextualise participant responses, a table briefly describing each participant and their child is provided below (***Table 1***). Their children's NDD was discussed briefly, to protect the identity of each participant and ensure that the quotes remain confidential.

Table 1: Brief description of research participants and their children.

Participant Number (P)	**Brief Description of Participant**	**Child with NDD**				
		Sex	**Age (Years)**	**ASD**	**Clinical Features**	**Genetic Aetiology**
1	Participant is a mother and is of Xhosa descent. Participant and her husband care for their child.	Male	12	No	GDD ID Short stature Dysmorphism	Congenital Anomaly Syndrome

2	Participant is a mother and is of Mixed Ancestry descent. She is a single mother.	Female	9	Yes	ID Seizures	Microduplication Syndrome
3	Participant is a mother. She is a professional with a tertiary education. Participant and her husband care for their child.	Female	12	No	ID Dysmorphism Seizures	Microdeletion Syndrome
4	Participant is a mother and is of Mixed Ancestry descent. Participant and her husband care for their child.	Female	9	Yes	GDD Delayed Speech and Language	Microduplication Syndrome
5	Participant is a father and is of Mixed Ancestry descent. Participant and his wife care for their child.	Male	12	Yes	GDD ID Seizures	Single Gene Disorder
6	Participant is a mother and is of Mixed Ancestry descent. She is a single mother of two children with intellectual disabilities.	Male	9	Yes	GDD	Microdeletion Syndrome

3.2 Stigma Experiences in South African Parents and Caregivers

The researcher sought to understand whether and to what extent the research participants had experienced stigma as a consequence of their children's NDD. Stigma is described as a societal process by which certain characteristics or traits are labelled (van der Sanden et al., 2013; Donovan, 2022). In the quote below, a participant echoed this description in her own words.

"Stigma is when you have a label that prevents you from living your full life. So, I am stigmatised, but I'm not stigmatised by having a label on me, a red tongue as it was. That was stigma in the Middle Ages. There are other ways to stigmatise people and to make them feel… super inferior. And not inferior in the sense of, you know, just feel otherwise that you don't belong, that you're different, that you… feeling, being discriminated against." – P3

Participants recalled numerous experiences of public, associative and internalised stigma which led to social exclusion. Participants described being mocked, for instance where people say their children are *"mad"* (P2), *"not normal"* (P4) or use the Xhosa term *"sisdalwa"* (P1) which translates to "disabled child". A participant whose child needs to be restrained with a child safety harness in public spaces, described receiving comments like "*yeah, you walking your dog"* (P5). Similarly, participants noted that people often look at you in public spaces or waiters would get annoyed when their children will not sit still.

International literature discusses similar phenomena of children experiencing stigma (Serchuk et al., 2021; Oti-Boadi, Dankyi & Kwakye-Nuako, 2020; Haft et al., 2019; Kinnear et al., 2016; Werner, 2015). In Tanzania, parents note that the community believe children with a disability are a "bad omen" to their families and use the derogatory word "talia" to describe children with ID (Aldersey, 2012). They further note that people who practice witchcraft target and kill children with ID.

Participants recalled instances where their children were excluded from social gatherings such as birthday parties or family trips which intensified feelings of isolation and shame. One participant (P2) described that her other children would get invited for birthday parties while her daughter with an NDD would not. Instead, for her daughter, "*they'll always just send the stuff over*". Similarly, another participant (P5) describes that his child with an NDD is even

excluded from family events, like "*Sunday drives*", where "*everybody's welcome except (my son)*".

Participants felt that they were being stigmatised for their children's condition. Such feelings occurred mostly from encounters in which the complexity of their children's NDDs and behavioural manifestations became apparent. One participant (P2) recalled experiences of associative stigma at the hospital where she was reprimanded by nursing staff for not controlling her daughter.

"...the nurse came up to me and like (asked), 'whose child is that?'. And I said, 'It is my child'. And she told me: 'Can't you control your child?'. And for me it felt actually very sad because you as a nurse, this is what you do. You work with such children, like you know, you're supposed to understand and know why." – P2

Another participant spoke about how the blame comes from unexpected corners. Consider for instance the quote below.

"This was when (my daughter) was still in intensive care. And I had to go home to fetch something which was very important. And at the time there was no Uber, so I had to rely on the taxi service which was often used by the ICU parents. So, it was a regular old chap… and we got chatting. So, he asked me: 'what's (the matter with your child?)'. I explained she's very sick, she has a heart condition… and because how old I was, and I said it. And then I needed to return to the hospital. So, I called him again a few hours later and I got into the taxi and outright, he said, 'Well, I spoke to my wife and she says, you deserve this because you put your career before your family. Because you decided to have a child so late. This is clearly an act of God to punish you for.'" – P3

This participant (P3) also described that her mother-in-law, who lives overseas, blamed her for her daughter's condition. When her mother-in-law realised the profound challenges her granddaughter faces on an overseas visit, she resorted to blaming her challenges on her daughter-in-law's irresponsibility of traveling with her daughter who is "*sick*". While her mother-in-law did not directly blame her for her grandchild's NDD, she displaced her anger by redirecting her unpleasant feelings towards something more socially acceptable such as travelling with her "*sick*" granddaughter.

"*And she's a sick child….and you are the most irresponsible mother in the world for traveling across the globe with a sick child."* – P3

The view that parents may be unable to fulfil conventional parental standards or provide adequate care is seen to endorse associative stigma (Serchuk et al., 2021). Mothers are also found to be more vulnerable to this form of stigma in comparison to fathers (Aldersey, 2012). This study further found that parents of children with ASD were frequently blamed or deemed as inadequate. In some cases, fathers may abandon their families due to having a child with ID (Aldersey, 2012). Existing research supports this finding as children with ASD may not exhibit the same physical manifestations as children with ID or syndromic involvement (Werner & Shulman, 2013; Gobrial et al., 2018:). As a result, children with ASD may appear to be neurotypical, leading other people to assume that their behaviour is a product of bad parenting. Behavioural problems include aggression, destructive behaviour, communication difficulties and meltdowns which are often viewed in relation to a dysfunctional family (Reddy, Fewster & Gurayah, 2019).

These accusations and many forms of blame and exclusion compound the physical and emotional burden of caring for a child with NDDs. Participants expressed that situations such as the ones recounted, lead to them withdrawing from social settings. One participant (P1) would *"rather let (her son) play in the yard with his siblings"* to avoid children mocking him. Some participants indicated that they withdrew because they were embarrassed or felt as if their friends were frustrated with their children. Another participant (P2) felt that her friends may be "*annoyed or something*" when her daughter accompanied her at social gatherings.

However, participants were keen to point out that social withdrawal was not because they were ashamed of their children, but rather, the complexity of their children's NDD and their respective needs lead to social withdrawal.

"Not because I wanted to hide it, but because it was challenging…Toileting was an issue. There are lots of things… that we can't do as others do." – P3

The above results highlight various experiences of blame, exclusion and stigma. Strikingly, some participants described not experiencing stigma. It was implied that members of the community were quite supportive of their children rather than judgmental or exclusionary. One participant (P6) commented that she does not note stigmatising attitudes anymore as she is

"past that stage now". Another participant (P4) echoed these views. In Tanzania, parents recounted that some members of the community were supportive and accepting of children with ID (Aldersey, 2012). However, as seen in this study, not all participants felt supported.

Participants expressed experiences of internalised stigma that resulted in depression, shame and loneliness. Participants described neglecting themselves, feeling as though "*there's nobody that understands what I'm going through, how I'm feeling"* (P6), being worried about what people may say about them as parents and feeling hurt when their children were excluded.

"I just didn't wanna go out, just wanted to stay in the house. Didn't want to be around people…In a way I also neglected myself." – P2

In one case, a participant relayed how she felt alone, bitter and betrayed when a kindergarten did not accept her daughter due to her special needs.

"Yes. I did feel alone in that sense, very alone when they kicked us out from (the kindergarten) because she was, you know, in the age group of four and she still needed nappy changes. I felt very alone and very bitter, and I felt very betrayed… They are trained about inclusivity, but there is no intention…These are my lonely moments. These are my bitter moments. This is when society, you know, they don't come after me to say I have a disabled child. But there are so many ways to you know, to face what is stigma."
– P3

Feelings of blame and guilt may contribute to emotions of isolation, embarrassment and feeling outside of the norm. This may affect parents' self-concept, mental health and may impact their relationship with their children (Papadopoulos et al., 2019). In Vietnam, families of children with ID described feeling depressed, alone, neglecting themselves and feeling as if there is nobody that understands what they are going through (Ngo et al., 2012). Likewise, in Nigeria, parents of children with ASD were shown to feel inadequate, out of place and misunderstood (Oduyemi et al., 2021). Therefore, there appears to be an important similarity between the feelings described by parents in this study, and those of parents in other countries of the world.

3.3 Influence of a Genetic Result on Stigma Experiences

Secondly, this project sought to understand the impact that receiving a positive genetic result for their children had changed the feelings of stigma or exclusion that participants described. The relationship between a genetic result and stigma experiences was not straightforward. Rather, it was found that the emergent themes: "Understanding of the NDD", "Navigating the Challenges of NDDs" and "Coping with the NDD" reflect factors which uniquely influence the "Perception of a Positive Genetic Result and Stigmatising Experiences", the final theme (***Figure 3***).

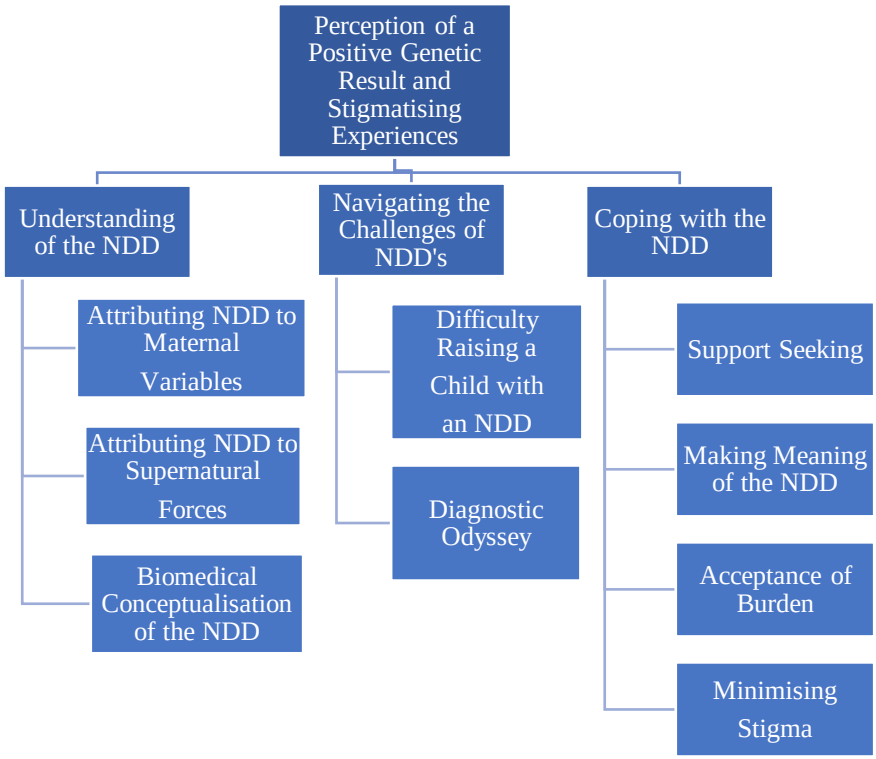

Figure 3: Hierarchical depiction of the main themes and sub-themes.

3.3.1 Understanding of the NDD

Participants of this study understood their children's conditions in different ways. Some attributed the condition to supernatural forces, others to more medical or genetic causes, and, yet again, others to things the mothers had done during pregnancy. Some participants expressed multiple explanations.

3.3.1.1 Attributing NDD to Maternal Variables

Many participants conveyed that they thought their child's condition was a result of things they did during their pregnancy. One participant (P2) thought that maybe she was too *"wild"* during her pregnancy or that her child's condition could be explained by the fact that she smoked before she found out she was pregnant. This made her feel guilty. *"I feel like it's because of me that she's this way"* and resulted in her going into a *"deep depression"*. As one participant expressed below, she thought her constant stress during the pregnancy may have been the cause of her son's condition.

"I used to stress a lot. So, I thought maybe it is the stress that caused him to be the way he is." – P1

Another participant attributed her daughter's NDD to her active lifestyle, age at time of pregnancy and the fact that she considered a termination of pregnancy. This participant further discussed the stress she experienced and fear of disappointing her mother as possible reasons for her daughter's NDD.

"I've been asking questions like could it be because I was doing sports at the time? Could it be because I've been training a lot? What could it affect? Could it affect her or, or the part where I was gonna go for an abortion with her?" – P4

A father of a child with an NDD (P5) also initially attributed the cause of his son's condition due to maternal distress during birth:

"His mother was in distress at hospital. The machines were showing that she was in distress." – P5

Mothers of children with NDDs frequently struggle with feelings of guilt and self-blame. Together with personal pressures and societal views on gendered blame, mothers are more likely to question their contribution to their children's NDDs (Gobrial, 2018).

3.3.1.2 Attributing NDD to Supernatural Forces

While a participant (P1) previously discussed her guilt about her son's condition, she also expressed great religiosity and that her son's condition may be a result of supernatural forces. She described a conversation with a friend that detailed supernatural involvement in the development of her son's NDD and that spirits may have come from the family she married into.

"I think it's something wrong, maybe Satan is doing something with (your pregnancy). Satan did something in your womb that caused (your son) to be like this…. They say like evil spirit maybe when you (were) pregnant, you were attacked by an evil spirit. Something may be coming from the family where you (were) married. It's the evil spirit from family that cause this…." – P1

She further described an experience with her "*prophet who prophesised": "I saw you are carrying a disabled child in your womb"* and that her son would be a *"little bit different from other kids".* She knew that her son would experience intellectual and developmental delays as her prophet further mentioned that her "*child might not be able to walk or talk*".

Different studies on disability in Africa have described an association with supernatural interpretations such as punishment for past transgressions, God's displeasure or an act of evil spirits (Etieyibo & Omiegbe, 2016; Oti-Boadi, 2017; Bunning et al., 2017). Furthermore, Reddy, Fewster & Gurayah, (2019) found that South African parents of children with ASD spoke about demonic possession and witchcraft. Interestingly, these views were also found in Hinduism and Christianity (Reddy, Fewster & Gurayah, 2019). Similar interpretations were described by caretakers and healthcare workers for children with NDDs in Uganda (Namazzi et al., 2020) and in Kenya (Gona et al., 2016).

3.3.1.3 Biomedical Conceptualisation of the NDD

In contrast, biomedical conceptualisations of the cause of their children's NDDs also arose. Biomedical conceptualisations include the belief that there may be a genetic or medical explanation for their children's NDD.

A participant (P3) described that she knew her daughters' NDD had a genetic cause since there were many indications suggestive of genetic involvement.

"We knew it's something genetic because we have kind of ruled out everything else. We knew it wasn't autism, we knew it wasn't ADHD or attention deficit disorder. Well, she might have some of that, but it wasn't, you know, just that it was much more. She had a heart condition at birth. So, there were a lot of proxies to say that there is something genetically different with her. But what the condition is? That remained a mystery." – P3

Yet at the same time, this participant also read peer-reviewed articles that suggested severe prenatal infection may lead to foetal abnormalities. This also made her question whether a prenatal infection could've been the cause of her daughter's NDD.

"I did think that it could have been something I did in the pregnancy, but I thought that after reading peer-reviewed articles about the fact that an infection during pregnancy in the mom can actually, especially in the very early stages of the pregnancy, can cause problems. But because we didn't know what the condition was, I didn't know. Couldn't figure out, you know, from the readings whether a severe infection or maybe, you know, not so severe infection or maybe severe, but undetected infection could have caused this." – P3

Two participants (P6 and P5) felt that pregnancy complications were a likely cause for their children's NDDs. These participants commented:

"Probably 'cause he was born premature… and also I went into early labour, but at that time they gave me morphine at (the hospital) to stop the pain." – P6

"Uh, the cord around his neck. That was my first (thought), because when he was born… his umbilical cord was wrapped around his neck twice." – P5

More than three quarters of parents that participated in a study in the United Kingdom described complications with pregnancy and delivery, and illness after birth as their causal explanations for their children's ASD (Elder, 2001). A study by Zuckerman, Lindly & Sinche (2016) in the United States of America found that majority of parents believed that their children's ASD was

a result of hereditary or genetic causes with beliefs about antenatal and environmental exposures to a lesser extent.

It is worth noting that some participants, in the present study, described and utilised multiple explanations for their children's NDD. Kengne Kamga et al. (2021) had shown that those from a Cameroonian rural community utilise multiple explanatory models to make meaning of Fragile X Syndrome. Genetic, spiritual and socioeconomic explanations were noted similarly as in this study. These findings are further substantiated by work in South Africa which discovered that Xhosa participants held multiple explanatory models for RHD (Matshabane et al., 2021) and for schizophrenia (Faure et al., 2019). Out of Africa, studies in Taiwan and India echo these findings (Shyu, Tsai & Tsau, 2010; Ravindran & Myers, 2013). Kengne Kamga et al. (2021) further noted that seeking multiple explanatory models exemplifies participants' search for personal meaning. This search for meaning is usually motivated by the desire to understand (Weil, 2000), thus, implying that participants in this study are finding a means to understand their children's NDDs.

3.3.2 Navigating the Challenges of NDDs

Participants described challenges associated with their children's NDDs. These challenges are discussed under subthemes, namely "Difficulty Raising a Child with an NDD" and "Diagnostic Odyssey".

3.3.2.1 Difficulty Raising a Child with an NDD

Participants recounted their initial frustrations with noticing differences in their children's behaviours and milestones. One participant (P4) described that *"one is more advanced than the other one"* while another (P6) described delayed milestones in the quote below.

"It was very frustrating because at the age of four he couldn't speak yet. He was still in a nappy, and it was just too much. He also started walking late, so I was like… something was wrong with my child, but I don't know what it is." – P6

Caring for a child with an NDD was described as "*being hands full*" since, as one participant put it: *"(my son is) equivalent to 10 children to look after"* (P5). Participants further expressed that caring for a child with an NDD placed a burden on their relationships.

"He places a big burden on our relationship… He demands your attention. So, if, whether it be my attention or his mother's attention, if he demands it, you can't leave him anywhere." – P5

One participant (P3) passionately discussed the physical, cognitive and emotional burden of caring for a child with an NDD. She further sympathised with other parents who struggle with daily life and resort to drastic measures to manage the load.

"I don't know how they do it. You just do it because what can you do? You know, we are not saints. You just have to do it. And some people break and kill themselves and kill their children, and you know they do horrible things and dads run away. But we shouldn't be so quick to judge…Okay, yes, it's not right. But hell, it's difficult. It's very difficult. I'm sorry…It's not just emotional, it's also physical, you know, because then you end up in all sorts of traps… We could never look after ourselves in the past 11 years because there was always demand on our time, either to keep up with our professional lives… But most of all keep up with (my daughters) demand. So, you don't, you're not exercising, you're not eating properly, you're not sitting down as a family to eat dinner. You don't go out anymore. You drink more than you're supposed to. You eat more than you're supposed to. Because you know, there was a certain time in our life… the only good thing was the food in your mouth. It was horrible. And then you keep doing it because that's the only good sensation you have. Everything else is horrible and your child is clung to a billion machines and cords and whatnot, and you don't know if she's gonna make it, but probably she will. But then what is it gonna be if she makes it? You know… all those questions." – P3

Caring for a child with an NDD is shown to affect many aspects of a parent's life (Gobrial, 2018; Currie & Szabo, 2020). Parents express great difficulty in caring for a child with an NDD, heightened stress, depression and grief over the loss of a healthy child. These findings coincide with research by Faden, Merdad and Faden (2023) who found a direct correlation between having a child with an NDD and a parent's diminished quality of life and wellbeing. These parents describe caring for a child with an NDD as a heavy burden with financial, familial and personal challenges. Similar findings are echoed by Gobrial (2018) who explored mothers experiences in Egypt.

Mothers of children with ASD experienced similar challenges. Their children frequently had "*meltdowns*" in public spaces that were difficult to contain. These difficulties lead to participants mourning the loss of a healthy child, resulting in poor mental health and a low self-esteem.

"I was feeling hurt also, but not hurt for myself because more (for my daughter), because I always used to think like she's not going to have the life that I always wanted for her, like a normal child would have, like grow up, play outside." – P2

Parents of children with ID hope that their children may one day have a family or job and live a "normal" life (Aldersey, 2012). These findings echo that seen in parents of children with ASD in China and the United States (Lai et al., 2015; Cohrs & Leslie, 2017).

Families with children with NDDs often experience financial and employment constraints in comparison to those who have children without NDDs (Saunders et al., 2015). In this study, participants voiced that finding a stable and suitable job is difficult. One participant (P1) described having to do part time jobs since she "*won't be able to work, like, for example, maybe a seven to five job*". Alternatively, participants described working night shifts or finding new jobs that allow for flexibility so that they may spend more time with their children.

"No, when I have been working at (a grocery store), I just gave it up because I said there is no time…And then eventually, I found another job. I just said I wanted to find another job that would (allow) more time seeing her." – P4

In some cases, it was found that managers may not understand when their employees repeatedly need time off for their children's appointments or admissions. As a participant (P6) put it: "*With his appointments and things like that, they don't understand*". This often results in an internal conflict as both their child's health needs and job security are important. As one participant (P6) commented:

"I'm a single parent and yes, I need to work, but also my child also needs me. So, it's like, what do I do? Where do I go first? Who do I put first? Because without the job I can't support (my children)." – P6

Literature suggests that there is great difficulty for parents to maintain a work and family life balance when their child has a developmental disability (Brown & Clark, 2017). It is thought to affect the number of hours one may work, type of job and money earned. In a qualitative study by Gobrial (2018), mothers of children with ASD in Egypt described instances of leaving their jobs to care for their children or having their husbands work multiple jobs to make ends meet. Understanding the complexities of their children's NDD and their daily needs resulted in parents feeling uneasy about their children's well-being when they may not be there to look after them. It also made them more aware of their own mortality. This resulted in anxiety, fear and worry that their children may not receive the care they need.

"I can't die before him. I need to be there for him because who else is going to be there for him?" – P5

Similar findings were discussed by Reddy, Fewster and Gurayah, (2019) when looking at lived experiences of parents of children with ASD in South Africa. Parents in Tanzania also wondered what their children's lives would be like in their absence or felt that other family members may take advantage of them (Aldersey, 2012). A parent further worried for their son's safety and feared that he may be killed.

3.3.2.2 Diagnostic Odyssey

Many participants described a diagnostic odyssey which was emotionally complex as it involves frequent doctors' visits, tests and uncertainties. One participant (P3) described their diagnostic journey as driving through a dark tunnel looking for light at the end. During that journey, they found it difficult to stay on track, however, their unwavering commitment is what keeps them striving for answers.

"So, what I'm trying to say is we always felt from that very early moment on that with (my daughter), we have been driving fast in the dark without any street lights on or our headlights on. And you're just hoping that, you know, you stay on the road and you're hoping that you're going in the right direction, and sometimes you realise, oh gosh no we are off the beaten track. We need to make our way back in the dark. But we are still together and we are still driving, and we are still going, and we don't know if we are heading in the right direction, but we have a gut feel we are." – P3

These findings are consistent with literature which describes the diagnostic odyssey as emotional for parents with rare genetic conditions (Graungaard & Skov, 2007; Krabbenborg et al., 2016a). It also exemplifies the extensive and unwavering search for answers for parents (Carmichael et al., 2015). Parents begin this journey to learn about their children's condition, what the future holds for their children and how to cope.

Another participant detailed his son's 10-year diagnostic journey where they received multiple inappropriate diagnoses. Each diagnosis resulted in a notable psychological disturbance likened to traversing the stages of grief.

"But you know, what we've been told from, let's say he's been diagnosed with, uh, probably four or five different things. And only now DNA testing was done and it came out that this is the results… and all the others was wrong. All the other diagnoses (were) wrong… The worst thing that was here was that (a doctor) said: 'My educated assumption is Fragile X' before doing Fragile X testing and 'your child will never live a normal life.' And I mean, my wife left (the hospital) crying." – P5

In some cases, medical professionals may diagnose a patient based on certain clinical features in the absence of a confirmed diagnosis (Bauskis et al., 2022). Mothers in Egypt expressed that medical professionals often misdiagnosed their children and that there was a poor awareness regarding their children's ASD (Gobrial, 2018). These diagnoses, such as ID or ASD, are more general and are nonspecific. Parents that discussed their diagnostic journey felt that there was no clear pathway (Bauskis et al., 2022). Parents further described concerns about the lack of appropriate treatment and prognosis without a confirmed diagnosis.

3.3.3 Coping with the NDD

Participants highlighted several tools used to cope with these difficult challenges. They use both positive and negative coping mechanisms to navigate the challenges they experience daily. Similar findings were noted by Lai & Oei (2014) who explored coping mechanisms in parents and caregivers of children with ASD in China. In this study, a multidimensional framework of coping was discovered which included four domains, namely "Problem Solving", "Spiritual Coping", "Cognitive Reframing" and "Active Avoidance". These domains may be likened to some of the strategies found in this study. These are discussed under sub-

themes namely, “Support Seeking”, “Meaning-Making of the NDD”, “Acceptance of Burden” and “Minimising Stigma”.

3.3.3.1 Support Seeking

Familial and spousal support were highlighted as important to help individuals cope with their children’s NDD. Having immediate support was shown to reduce feelings of loneliness and foster resilience. One participant (P6) named her mother as a significant supportive figure who “*is the rock, the pillar in the house*” and guides her through this journey. This view was echoed by another participant (P2) who felt that family members were supportive and would always “*check-up*” on her or fetch her daughter when she needs personal time. In the quote below, a participant (P3) illustrates that the support she receives from her spouse helps her stay afloat. Understanding how difficult it is to care for a child with special needs, she further commended single parents with special needs children.

“Like, I'm still alive. That's how it feels. Like a lifeline. I would never be able to do it alone. Well, probably I would because what choice do you have? But I don't even want to think about how hard that is. And when I see single parents… No, they're not humans… They are angels or I don't know what they are. It's insane what a single parent would have to do.” – P3

Coping strategies are intrinsic adaptive responses that aid in stress management, develop resilience and assure family well-being. Literature suggests that parents of children with ASD may adopt cognitive strategies to manage stress (Lai & Oei, 2014), delve in spirituality and religiosity, partake in research, and seek support as a means to cope (Reddy, Fewster & Gurayah, 2019). Many studies describe support seeking as an important positive coping strategy that parents and caregivers display and is often associated with positive outcomes (Gray, 2003; Pakenham, Sofronoff & Samios, 2004; Hastings et al., 2005).

Other participants recalled their children’s school, a school for learners with special education needs, and their hospitals as sources of support. As one participant (P6) commented:

“I actually loved last year. (The school) had a parent support group and I enjoyed that… It shows you that you as a parent, whether you're single or married, you are not the only one going through this.” – P6

In New England and South Africa respectively, studies suggest that support groups create a holding environment for parents to meet like-minded families and creates a space for learning and fostering positive behaviours (Hutton & Caron, 2005; Reddy, Fewster & Gurayah, 2019).

3.3.3.2 Meaning-making of the NDD

Two participants expressed how meaning-making may be used to cope with a diagnosis, both positively or negatively. One participant (P5), for instance, expressed how his religion allowed him and his family to view his son as a gift.

"I'm Muslim. And we believe that children with a disability, as in whatever the disability, let's call it mentally, physically, however. Whatever child is born with a challenge, that child is a gift. Yeah. So now what a term the old people would use with us is: 'Hy jou Jannah. He is your paradise'. So, I would always say to them that paradise isn't cheap because it's hard work." – P5

This finding is consistent with that of Jegatheesan, Miller and Fowler (2010) who discovered that Muslim parents felt blessed to be picked to care for a "special" child and that they would be rewarded in heaven. They further found that parents believed that this was a test of their spiritual and moral conduct for which they would be judged.

This participant (P5) further created meaning by ascribing his personal development, life trajectory and family role to his sons' presence. He commented:

"He grounds me. I promise you… If it wasn't for him, I would be in a different space. I would be more than likely a different person if it wasn't for him. When I say that, I'm more grounded with him, you don't get me (out on) weekends… I will go out (with) the kids. Yes. I will spend my whole day on the weekends, from the morning to the night with (my children) doing what they want to do. So, if it wasn't for him, I'm sure I would be doing what I would have wanted to do… I've got zero friends. I've got friends, but they know they're my friends during work time… After work, I'm daddy." – P5

One participant (P1), who believed that supernatural forces were responsible for her son's NDD, ascribed meaning to the diagnosis by encouraging her to strengthen her faith so that the diagnosis doesn't drive her apart from her husband.

"That is Satan… The evil spirit wants you to divorce your husband. That's why your baby is like this. So, you have to pray to God and don't come out of your marriage. Like don't allow Satan…to ruin your life." – P1

These meanings are shown to build a profound appreciation for their children, their circumstances and perspective on life which may guide them through the complexities of caring for a child with an NDD. Regardless of how different religions and cultures make meaning, these findings suggest that religion provides a basis for understanding life's obstacles and finding purpose in adversity. It enables parents to see their children's NDD as a part of a divine plan and as a means for their children to teach them valuable lessons (Gobrial, 2018).

3.3.3.3 Acceptance of Burden

One of the most striking results to emerge was that all participants implied that comparing their children to other children with worse conditions helped them cope with and accept their children's diagnoses. Consequently, there is this acceptance of burden which is minimised when compared to vast difficulties other families face. Two quotes are presented below to illustrate this finding.

"When I…first come to (the hospital) and meet other kids with the disability, it's a different type of disability, then it gives me courage like… my son is better than others. I don't have to carry him. He's learning. Although he's learning very slow. But he's learning as he is becoming big." – P1

"There are days when I… feel really down. And then I will scroll through… how some parents are struggling. Then I will like tell myself, but here's a parent going through way more worse things than what I'm doing, than what I'm going through. And then I'll feel much better after that." – P2

The researcher felt that engaging in comparative exercises may be a way for parents to find peace, perspective, and reassurance that their children's condition is manageable in their context. It further enables parents to maintain a positive attitude in the face of adversity. Unfortunately, no specific research could be identified to substantiate this finding. While this may be labelled as a helpful coping mechanism, the researcher thought that it may invalidate their emotions and children's health challenges.

3.3.3.4 Minimising Stigma

When questioned about the words people use to describe their children, it was found that people do make stigmatising comments. A participant (P1) mentioned that people would say *"ai she's mal man"* which translates to "oh she's mad man" or *"taatie"* which is slang for "mad". She later added that these people would say it "*in a joke way not really like in a negative way*". Despite it being a joke, it is stigmatising nonetheless. Here, it was thought that humour was used to deal with microaggressions and subsequent internalised stigma. Similarly, in the quote below, one participant (P5) described older children who "*joke around*" with his son.

"The younger ones, they also play with him, so he can now play with the younger, from five years old and he can play with a 14-year-old. Although the 14-year-olds, they joke around with him. But he's sporty. He goes for it, so." – P5

The researcher felt that this coping strategy was maladaptive (McKee et al., 2004) as parents protect their psychological well-being by framing mockery in a light-hearted manner. While this may relieve emotional suffering, it may also mask their true emotions and realities they face. Therefore, it may be considered a harmful coping mechanism.

Thus far, the results have detailed the first three emergent themes, namely "Understanding of the NDD", "Navigating the Challenges of NDD's" and "Coping with the NDD". When participants were questioned about the positive genetic result received, an array of responses were received. These findings will be illustrated in the following section.

3.4 Perception of Positive Genetic Result and The Impact on Stigma

While not outright, deeper interpretation of the results had shown that participants understanding of their children's' NDD, how they navigate challenges and cope with these

challenges play key roles in how participants perceive a genetic result and stigma experiences. Interestingly, personal motivations for participation in genomics research also influenced one's view of the result. These findings are discussed under sub-themes, namely "Meaning of the Result", "Prior Understanding shapes Genetic Result Perception", "Diagnostic Odyssey shapes Genetic Result Perception", and "Positive Genetic Result's Impact on Stigma".

3.4.1 Meaning of the Result

Personal motivations for partaking in genomics research emerged when asked about the meaning of the genetic result. These motivations include diagnostic closure and hope for a cure or treatment. For most participants, there was a hope for a treatment or cure. As a participant (P5) commented: "*I was hopeful that they would find something that can be fixed."* When the results were given and there was no cure or treatment available, it resulted in disappointment.

"I thought maybe if the results will come and say maybe there is a cure to the condition, the way my son is… I feel bad, like there's nothing that the hospital can do to change the way (my son) was." – P1

It may be implied that parents were motivated by the potential of enhancing the lives of their children. A Masters-based research study in South Africa questioned parents' motivations for participating in the NeuroDev study and discovered three key reasons for parents to participate in genomics research. This included altruism, for personal and family benefit in terms of care and recurrence, and a hope for a cure or treatment (Diedricks, 2020). Surprisingly, more parents cited altruistic motives than hope for a cure or treatment, which is different to the findings of this study where most participants participated in hopes that there would be a cure or treatment. While this may be a result of a small cohort, it may also imply a hidden agenda or subliminal wishes. Liang et al. (2022) discovered similar results, indicating that altruism was a significant influence in deciding to participate in genomics research. Parents were disappointed that the results were not actionable, yielding no change in medical regimes or daily living. Despite these findings, some parents state that a positive genetic result provided diagnostic closure and created a sense of hope that a therapy will be available in the future. In a study which looked at the psychological impact of genetic results on parents, it was found that some parents valued the genetic diagnosis, others experienced a loss of hope and disappointment (Krabbenborg et al., 2016a). This was due to hoping for a better treatment or for a miracle.

The majority of participants stressed that the genetic results did not change their day-to-day living or medical regime. One participant (P3) commented: "*No, it hasn't changed in any ways. Because we knew it has to be that we just didn't know the label*". However, for some participants, the result was able to provide a sense of diagnostic closure and foster hope for future treatments. This finding is exemplified in the quote below.

"I'm just praying and hoping that they will find some kind of medication or treatment to help (my son), or not just (my son), like any other child." – P6

A significant observation was made when a participant (P3) argued that a genetic result for a rare condition is not useful. It was implied that since there is little research detailing the prognosis of a specific diagnosis, her daughter's NDD remains unpredictable. She further alludes to how this may create uncertainty for her daughter's future and health needs. While the result wasn't meaningless, the lack of actionability meant that the result did not offer any tangible resolutions.

"Because the rare genetic condition means we are, okay, closer to the truth, but what is the truth? We don't know. We just know we have scientific evidence for showing that we are closer to the truth. We have a diagnosis, but without a prognosis that diagnosis is not helpful." – P3

Participants may undergo genomic testing for diagnostic closure and to improve their well-being by means of available treatment and long-term management (Pollard et al., 2021). In a study that looked at parents' views on the diagnosis of rare genetic diseases, some parents appreciated a confirmed genetic diagnosis without clinical utility while others argued that for a genetic result to be beneficial, one would need access to treatments (Pollard et al., 2021). Similarly, parents who received a genetic diagnosis for a rare genetic disease expressed relief and disappointment. While they had received a diagnosis, often medical professionals knew little about their children's condition meaning that there was no way forward (Krabbenborg et al., 2016a).

3.4.2 Prior Understanding shapes Genetic Result Perception

Of interest here, it was found that some participants reverted back to their initial understanding of the cause or viewed the genetic result as part of the holistic cause.

"I still feel so yes. I feel maybe if they didn't give me all that morphine it maybe would've been different." – P6

"It's a contributor and to me it's, uh, you know what… I repair machinery. So, I look, I don't look at what is broken. I look what caused that thing to break in order to fix it. I can't just fix what is broken. I must look why did that thing break?... When (my son) was starved for oxygen, he was a colour blue… when he was born. So, I believe that it was a contributor to his slow development." – P5

Parents from this study could be seen to conceptualise genetic results through the lens of their initial causal beliefs of their child's NDD. Despite receiving the positive genetic result, some parents continued to believe that they could've done something different to prevent their children's NDD or that maternal stress caused the NDD. This finding suggests that while genetic information is valuable, it may hold limited importance to some parents. Parents may prioritise their beliefs as it provides them with a sense of understanding and control. Often a genetic result may be too complex to understand resulting in parents reverting to prior understandings. Another participant viewed the positive genetic result as a factor in addition to biomedical beliefs. Weil (2000) briefly introduces the concept of a hierarchy of causal explanations. Here he discussed that people may psychologically compartmentalise beliefs or rank them in order of personal importance. This suggests that while a positive genetic result may be understood or received, a parent's prior beliefs may take precedence since it provides them with support, comfort and understanding. These findings are consistent with those of Matshabane et al. (2021), who discovered that Xhosa participants with RHD held multiple causation models which featured genetics and that medical, cultural and behavioural explanations were deemed more important.

On the other hand, one parent who already suspected that her daughter's NDD was genetic was given confirmation of her suspicions. These findings reiterate that the search for meaning is largely individualistic.

3.4.3 Diagnostic Odyssey shapes Genetic Result Perception

As previously discussed, a participant (P5), recounted numerous experiences where they received incorrect diagnoses which were indescribably traumatic. It was further implied that

they had undergone multiple grief processes after each diagnosis, so that when the confirmed genetic result was received, there was no upset or disturbance.

"I still don't have that. 'Oh, wow'... It's a bit of relief to think that okay at least there's now something. But you see all the time it was just speculation." – P5

This was a significant finding and impacted the participants' view of the genetic result. However, no supporting research was found to corroborate this finding.

3.4.4 Positive Genetic Result's Impact on Stigma

There was some evidence suggesting that a positive genetic result impacted experiences of internalised stigma. Participants reported how a genetic result made them feel better as it removed their feelings of responsibility. As one participant (P2) commented: "*The weight is like off my shoulders*". She further described that she was "*short tempered*" and would "*scream or just yell*". It alluded to the idea that the genetic result removed the guilt she felt when she commented: "*I can see also a difference and feel a difference in myself*". Another participant was particularly elated that the genetic variant was paternally inherited. As mentioned in the quote below, both of her children, from separate relationships, have special needs. Despite their diagnoses being different, she still felt responsible for her children's special needs.

"The one thing for me was, at least it's not coming from me as the mother. It's the father. So yeah. That was actually the main thing for me because both my children have special needs… I thought that I was the one carrying it." – P6

This was the outcome of parents identifying an agent of responsibility other than themselves. In other words, a genetic result provides a clear scientific basis that alleviates self-blame, guilt and shame (Krabbenborg et al., 2016a).

Despite harbouring these feelings, this participant (P6) still believes that there is something she could've done to prevent her sons from having an NDD. This speaks to her struggle with internalised stigma which remains prominent despite receiving a positive genetic result.

"I feel there's something I could have done better. I don't know what it is or what it could be. Maybe if my diet was like it should be, then it would've been different." – P6

Seeing this divergence of beliefs signifies that she may still be navigating the complexities of coming to grips with her son's NDD. Matshabane et al. (2021), on the other hand, discovered that while Xhosa participants with RHD reported internalised stigma, a genetic attribution had no effect on internalised stigma in this community. The authors attribute these findings to pre-existing causal beliefs, structural stigma and contextual realities which influence how they conceptualise their disease.

It is worth noting that there was little evidence suggesting that a positive genetic result affected other forms of stigma. There were no changes in stigmatising experiences and this was attributed to the lack of change in society's view of difference and disability. While a positive genetic result impacts internalised stigma, it does not impact societies opinions of difference and disability. Thus, there was no change in public stigma. As one participant explained:

"I had those experiences. Not because I wasn't accepting my child. It is because I saw people don't accept difference to the whatever the norm is. I mean, you define the norm. I won't 'cause I don't know what the norm is. But I know what the not norm is." – P3

As literature proposes, a genetic result may either exacerbate pre-existing stigma (Tekola et al., 2009) or lessen public discrimination. However, in this study, a genetic result was not shown to impact public and associative stigma. Nonetheless, receiving a genetic result encourages participants to strive for harmony in acknowledging medical realities while nurturing positive coping mechanisms that build resilience for future endeavours. In one case, a participant (P2) described that she is "*doing more research, like things to help (her daughter)".* It further created a sense of belonging as the positive genetic result encouraged participants to foster new relationships and family involvement. In the quote below, one participant (P6) mentioned a family member who wanted to do more research to know how to best help the participant and her child.

"Like just pop in, see where I am. And that's the day when he sat down and he wanted to know… what is the name of that thing and he wants to research more. And then I said, you can research, it's fine, it's not a problem. He said he wants to know more and then it would just encourage me." – P6

Participants received consistent support from a variety of sources such as family and community members, spouses, schools and hospitals. This support fostered a sense of belonging and acceptance, and enhancing confidence and resilience to navigate their children's

NDD while confronting stigmatising experiences. Interestingly, mothers in Ghana noted forgiveness as a coping mechanism to deal with stigmatisation (Oti-Boadi, Dankyi & Kwakye-Nuako, 2020). Forgiveness helps convert negative feelings with positive feelings. This was, however, not seen in this study. According to one parent, regardless of a genetic result, societal judgments and biases persist, implying that it may not change deep-rooted societal beliefs. According to Matshabane et al. (2021) participants with RHD did not consider causal explanations as an essential contributor to disease-related stigma. This supports this finding in this study as it speaks to societal biases stemming from sources other than the causes of an NDD.

3.5 Role of Genetic Counsellors in Genetic Research and in Addressing Stigma.

When the role of genetic counsellors in participants' result delivery was discussed, it was found that participants' knowledge of genetic counselling was minimal. A participant (P6) mentioned that she was *"still puzzled around the genetic counselling"*. Consequently, the responses received were not as descriptive as expected. However, emergent themes suggest that genetic counsellors play an important role in genomics research and addressing instances of internalised stigma. Four main themes emerged, namely "Resource of Information", "Genetic Counsellors Impact on Stigma", "Relational Aspects of Care" and "Continuity of Care".

There has been an increasingly strong recommendation that genomics research teams have a partial ethical obligation to return positive genetic findings that may impact a participant's healthcare. The NeuroDev project is one pioneer project at UCT that sought to return positive individual genomic research results. At the time of this study, that practice was still new and little was known about whether and how people would understand such research results, and whether it would change the way they understand disease. Matimba et al. (2022) emphasises that positive genetic findings should be returned to participants by medical genetic professionals with preference to genetic counsellors. The role of a genetic counsellor, as discussed by Biesecker (2001), involves facilitating a meaningful and intimate adjustment to genetic information while reducing psychological distress in the process. It follows a psychoeducational model which marries teaching and psychosocial complexities within an established therapeutic relationship (Patch & Middleton, 2018). This model coincides with the findings in this study since it was determined that genetic counsellors served as a resource of

information on their children's newfound diagnoses, provided emotional support and addressed experiences of internalised stigma.

3.5.1 Resource of Information

Participants expressed that speaking with a genetic counsellor served as a resource of information on their children's newfound diagnoses. For one participant (P5), the session offered him *"clarity"* on his son's NDD. Likewise, another participant (P2) emphasised that the session helped her learn things she was unaware of.

"For me, it was actually interesting because… she explained to me certain things that I never knew of and I learned actually from it and it helped me also now in my…daily lifestyle."– P2

Patients find the provision of information as one of the most helpful aspects of genetic counselling and appreciate speaking to someone deemed an "expert" in the field (Macleod, Craufurd & Booth, 2002). A study looking at mothers' experiences of genetic counselling in Johannesburg South Africa had labelled the genetic counselling experience as "a worthwhile experience" (Morris et al., 2015). Mothers valued the information provided along with suitable diagrammatic explanations, emotional support and referral to external support groups. This study also highlighted negative experiences of genetic counselling. However, this was not found in the current study. Parents of children with Williams Syndrome also reported that receiving accurate information on the diagnosis and management for their children was shown to produce a more favourable genetic counselling session (Waxler et al., 2013).

3.5.2 Relational Aspects of Care

Along with learning about the diagnosis, participants expressed that they received emotional support during the result delivery session. A participant (P3) described that the counsellor was "s*ympathetic and empathetic… very professional, very kind, very accommodating."* Another participant (P2), in the quote below, commented that the counsellor established a human element into research allowing her to build a relationship and open up to the counsellor. She further went on to say that the genetic counsellor provided support and comfort.

"She wasn't just one where, you know… 'I'm just there for the blood test, I'm just there for the study'. She actually gave that… like I could see like I could bond with her, speak with

her… I felt like she wasn't just there for research alone, but she was (there) for me as a whole… It was a good thing that I went for it because sometimes you need that weight off your shoulder… I felt good about it after and during the counselling session." – P2

Another participant (P3) commented that *"shared sorrow is half sorrow"* which illustrates that sharing one's grief in an understanding environment makes it more manageable. This participant further echoed that the genetic counsellor provided an understanding ear which is "*a rare opportunity to have*".

What stands out is that despite having the counselling session, a participant (P3) felt that the rarity of her daughter's diagnosis meant that there was no further guidance that could be obtained from a genetic counselling session. Since there is no prognosis, one is still left with uncertainty. This participant further expressed that she would much rather receive support from a medical professional who is able to give concrete answers.

Parents often find it challenging to receive a genetic diagnosis because it may have a lifelong psychological impact. Parents may experience shock, fear, uncertainty, sadness and others, relief (Ashtiani et al., 2014). When a diagnosis is finally made, it may bring a sense of comfort and clarity. However, it may also trigger feelings of grief when their children's challenges are realised. It is recommended that genetic diagnoses are conveyed delicately, sensitively and empathetically (Ashtiani et al., 2014). The positive experience noted by parents by Waxler et al. (2013) was justified by the genetic counsellors' skills in conveying difficult news and providing support where necessary. In a genomics research setting, genetic counsellors may contribute to the integration of research findings into clinical practice, as well as facilitate adaptation and acceptance. They may encourage patients to gain a sense of mastery and identify effective coping strategies to navigate the psychological and physical challenges that may lie ahead (Haakonsen et al., 2018). This study also highlighted that genetic counsellors provide a listening ear which creates a safe space where parents may voice their concerns and anxieties, and facilitate acceptance of their children's NDDs.

3.5.3 Genetic Counsellors Impact on Stigma

A few participants expressed that receiving a genetic result impacted internalised stigma. A participant (P4) who understood the cause of her daughter's condition as something she did during the pregnancy expressed that the genetic result rid her of her feelings of guilt. Another

participant (P6) who struggled with internalised stigma and felt at fault for her son's condition expressed that parental testing confirmed that she was not to blame for her son's condition. Consequently, the result delivery with a genetic counsellor reduced her internalised stigma.

"When she told me that (the genetic variant) comes from the father's side, then it was like I could jump on the roof and say, it's not my fault." – P6

Mothers of children with ASD describe experiencing associative and internalised stigma and are often deemed responsible for their children's diagnoses (Oti-Boadi, Dankyi & Kwakye-Nuako, 2020). Likewise, parents of children with ASD in Nigeria felt inadequate, misunderstood and alienated (Oduyemi et al., 2021) while parents of children with ADHD reported feelings of sadness (Chang et al., 2020). The findings of the present study had shown that genetic counsellors play a role in reducing strong feelings of blame and guilt in some parents who felt at fault for their children's conditions. This was due to the genetic counsellor conveying the genetic result which may have been de novo or inherited by the other parent (Bar, 1999). Unfortunately, there is no research that looks at the efficacy of genetic counselling for NDD's to corroborate this finding. The absence of this research signifies that further research is required to broaden our understanding. Other research that looked at the understanding of genetic disease in British families found that parents may feel responsible for their children's learning disabilities (Chapple, May & Campion, 1995). Bar et al. (1999) suggests that in these cases, a genetic attribution may reduce internalised stigma by targeting self-blame.

It was further found that the genetic counsellor facilitated adjustment and gave participants tools to reframe their diagnosis. As a participant (P2) commented: "*I started thinking more positively"*. Another participant (P4), who struggled with her daughter's diagnosis and the associated uncertainty, discussed that the session with a genetic counsellor encouraged her to come to terms with her daughter's diagnosis and "(*accepted) what the future is gonna be like for (her daughter)"*.

NeuroDev participants have expressed that the information gained from the study impacted their feelings of blame as they reframed their thoughts from personal responsibility to religiosity (Diedricks, 2020). While this finding is secondary, its significance may not be understated. It implies that there is a link between the role of genetic counsellors and reduced feelings of blame and guilt in parents of children with NDDs. While it is true that receiving the

positive genetic result, alone, contributed to reducing feelings of guilt and blame, speaking to a genetic counsellor compounded these results by creating a holding environment where parents could reframe their causal beliefs and process complex genetic information. Without this, the significance of a positive genetic result and associated inheritance may not be understood and interpreted correctly. Thus, this finding highlights the positive impact a genetic counsellor plays in result delivery and the importance of including a genetic counsellor in genomics research.

While this correlation was identified, the results show no evidence that genetic counsellors directly influence other forms of stigma. Participants indicated that there was no change in relationships or societal attitudes in public spaces. This study highlighted two possible reasons for that. Firstly, participants who noted strong support structures found that families and communities remained supportive. This support also reduced the impact of public and associative stigma. As a participant (P4) highlighted, she is *"past that stage now".* Additionally, it was found that no matter the cause, society may always be judgmental and discriminatory towards differences and that genetic counselling or having a genetic explanation for an NDD would not impact public and associative stigma. A lack of public awareness and knowledge on NDD's such as ASD fosters negative stereotypes leading to public and associated stigma (Broady, Stoyles & Morse, 2017). Parents of children with ASD feel that educating the public on the difficulties children experience may combat discrimination (Woodgate, Ateah & Secco, 2008). These findings suggest that genetic counsellors operate on an individual and family level, however, a multi-stakeholder approach is necessary to raise awareness and foster inclusion in order to see changes in public and associative stigma.

3.5.4 Continuity of Care

There were some indications that participants appreciated that the genetic counsellor was involved in all steps of the NeuroDev research study. A participant (P5) felt as though *"it was somebody doing something"* and kept them updated with the progress of the study. Another participant (P6) echoed these findings by describing the involvement of the genetic counsellor in the various steps of the study.

> *"I think she did it the right way, preparing us before the test, preparing us for the waiting period and obviously after getting the results."* – P6

Krabbenborg et al. (2016b) discusses the importance of genetic counsellors in the process of WES. They suggest that genetic counsellors should be present for pre-test and post-test counselling to support families through this process. This work substantiates this statement by describing the uncertainties and losses which families experience during this process and that genetic counsellors may be able to provide adequate support to manage these uncertainties (Krabbenborg et al., 2016b). These findings further emphasise the importance of genetic counsellors in genomics research.

3.6 Chapter Summary

The findings of this study were comprehensively discussed in this chapter. A brief description of each participant and their child was presented followed by experiences of stigma, how a genetic result influences these experiences and the role of a genetic counsellor. In the final chapter, the researcher will conclude the findings and discuss the strengths, limitations and implications of this study.

4 Chapter Four: Conclusion

4.1 Chapter Introduction

The findings of this study were extensively compared to existing literature in the previous chapter. In this chapter, the findings are concluded and the strengths and limitations are considered. This chapter completes with recommendations for genomics research in South Africa and future research avenues.

4.2 Conclusion

This study showed that how a parent perceives a genetic result and how this influences stigma is largely impacted by how a parent understands a diagnosis, the difficulty in raising a child with an NDD, and the coping mechanisms parents employ. While a positive genetic result holds strong for some, others may struggle to let go of their causal beliefs or assimilate the genetic result into their causal belief framework. Parents recounted multiple forms of stigma which was compounded by the difficulty in raising a child with NDDs. These parents often embarked on a diagnostic odyssey which was described as lengthy and filled with uncertainty. It was also found that coping mechanisms often arose. These coping mechanisms, while positive and negative, were shown to help parents face adversity and deal with stigmatising experiences. The genetic result was shown to only impact internalised stigma experiences as it allowed some parents to redefine their causal belief and remove feelings of responsibility. The genetic result does not impact other forms of stigma. It was hoped that this research may provide insight on how a genetic result may influence public stigma in a South African context. However, this was not found. The researcher thought that the lack of time between the delivery of the result and the interview to observe changes in stigma experiences, or a lack of community understanding of genetics due to disparities in education may have been the reason for this finding. Furthermore, due to the sensitive nature of the topic, parents may not have communicated the positive genetic result with family or community members.

These findings show that stigma is multifaceted and has multiple external influences. It also emphasises that when conducting genomics research in South Africa, one needs to consider socio-economic, political and cultural factors which may influence stigma and shape genomics research in South Africa.

This study further looked at the role of genetic counsellors in addressing instances of stigma in NDDs and genomics research. It recognised the crucial role genetic counsellors play in addressing stigma and in genomics research. It was found that genetic counsellors play a role in reducing internalised stigma but had no effect on public and associative stigma. This is because a genetic counsellor operates on an individual level rather than community level meaning that a genetic counsellor cannot directly change societal prejudices. Genetic counsellors were found to be a resource of information, provide a human element to genomics research and provide continuity of care. Consequently, genetic counsellors are shown to play a crucial role in genomics research.

4.3 Strengths of the Study

Despite the small cohort, this study was able to explore the perspectives of South African parents of children with NDDs. It further lays the groundwork for future research on the types of stigma families experience and how a genetic result may influence stigma.

This study provided contextual understanding of the lived experiences of families with children with NDDs. It further acknowledged the diversity of experiences within the South African population and explored each unique narrative individually whereas quantitative studies may not.

The researcher was able to witness NeuroDev result delivery sessions with a genetic counsellor. This was valuable to the research process as it allowed the researcher to immerse herself in the procedure and brought awareness to some of the concerns that parents discussed with the genetic counsellor at that time. This allowed the researcher to effectively bracket her opinions, biases and preconceived notions so that the data remains authentic and reflective of the participants' experiences. This ensured that the research was credible. Each interview was conducted in the language of the participants choice and in an appropriate venue. The use of recorded, semi-structured interviews allowed the researcher to preserve the exact words used by the participants aiding analysis and interpretation.

The analysis was an ongoing process; therefore, the findings of the study drove the sampling process as the study progressed. These findings are crucial in informing practice and policy for genomics research in South Africa and the utilisation of genetic counsellors in genomics

research. It may also inform support programs and that may meet the psychological needs of parents and families with children with NDDs.

4.4 Limitations of the Study

Qualitative research methods are known to be complex for first-time qualitative researchers (Lester, Cho & Lochmiller, 2020). This suggests that data collection, analysis, and interpretation may not be of standard. However, the researcher's background as a genetic counselling student meant that she had already acquired communication and interpersonal skills as a part of the degree and/or clinical experience suggesting that the researcher was able to establish rapport and obtain meaningful information.

This research is classified as a minor dissertation and there was a time constraint of twelve months to complete this research. Furthermore, only a few hours were officially dedicated to 'research' alongside formal tuition, assignments and clinical hours.

There were only a small number of participants in this study. At the time that this project took place, only twelve NeuroDev participants had received positive results of which only six were available for an interview. This poses a selection bias as the participants were interviewed based on availability. Reasons for non-participation included travel, work and medical constraints.

There was also a limited amount of time between result delivery and the research interview for some participants. This suggests that the participant's understanding, adjustment, family/community communication of the result and stigma experiences may be skewed as there was not enough time to see or experience the noteworthy impact of the positive genetic result on their understanding of illness.

During the data collection process, participants inquired about telephonic interviews in cases where participants were unable to come to RCWMCH or UCT. Initially, it was thought that in-person interviews would be preferred due to the nature of the questions asked. However, including telephonic interviews may have allowed more participants to take part in the study.

4.5 Implications/ Recommendations

This study provided insight on parent's and caregiver's experiences of stigma and how a newfound genetic diagnosis influences these experiences. Understanding these experiences may help genetic counsellors and other medical professionals comprehend the intricacies of caring for a child with an NDD, the types of stigmatisations in South African populations and it may direct genomics research in a South African context. For example, awareness of the forms of stigma may orchestrate how genetic counsellors respond to the relational needs of parents and caregivers creating a framework of empathy and support. This research further demonstrated that genetic counsellors play a critical role in genomics research. It was shown that genetic counsellors may aid in the empathetic and humane delivery of a genomic result and associated information, along with addressing experiences of internalised stigma. From personal experience, other genomics research studies fail to incorporate genetic counsellors for feedback of genomics findings unlike that of the NeuroDev study. Consequently, these results show that genetic counsellors play a critical role in genomics research and further qualitative research into the inclusion of genetic counsellors in genomics research is proposed.

Based on these findings, the following recommendations are given when considering genomics research in South Africa:

- Consider ethical guidelines that address stigma-related concerns in genomics research.
- Inclusion of a genetic counsellor in genomics research.
- Genetic counsellors to be culturally competent and aware of the stigma associated with NDDs and how this may affect family dynamics, finances and careers.
- Ensure that genetic counsellors and researchers are aware that genetic information may have psychological implications and may influence stigmatisation.
- Ensure that there are support services for families to address their emotional and psychological needs.

4.6 Future Research

This study provides brief insight into the forms of stigma experienced by parents and caregivers of children with NDDs in a South African context. It is expected that parents not interviewed in this study may have additional experiences. South Africa is a diverse and multicultural nation; thus, it is expected that there is an array of experiences that may not have been considered in this study. Therefore, it is proposed that additional research on these experiences be conducted. It was also shown that the knowledge of genetic counsellors and their role is scarce among the lay public. Additional research looking at the understanding of genetics and the service genetic counsellors provide among the public is proposed.

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Appendix A: Research Reflection

Reflecting on where my interest in this topic began, I believe it began many years ago when I was bullied at school for having a cousin with a rare NDD. Little did I know, 15 years later, I would be conducting research on stigma experiences. Having that experience from a young age and seeing the difficulties around caring for a child with an NDD not only directed my research interests but also encouraged me to be a genetic counsellor to support families just like mine. I believed that gaining an understanding of the types of stigma other families experience would assist in molding my genetic counselling style and allow me to be an empathetic counsellor.

This study was my first venture into the qualitative research space. Coming from a quantitative background, I found it very difficult to turn off my numerical mind. It somewhat felt like a maze where some days or weeks I'd reach a dead end and other days I'd find ways closer to the exit. Conducting these interviews also allowed me to strengthen my listening and questioning skills which has helped me in my day-to-day counselling. I learnt a tremendous amount about qualitative research, IPA and about my own personal triggers. Hearing stories from patients was insightful, however, somedays I would go through depressive spells where I wouldn't feel strong enough to hold my participants emotions and construct meaningful research. But I now see the importance of my research and how a listening ear may help those out there. It was definitely a daunting experience but fulfilling to say the least. I believe having this experience ignited a flame to further my research and I hope to continue in this research space.

This research also allowed me to truly understand the impact of a result delivery. Particularly, how a sensitive, sympathetic and empathetic result delivery is beneficial and that genetic counsellors should be routinely used in genomics research. It allowed me to advocate for our role during my oral presentation at the 5th International Developmental and Paediatric Association Congress.

Appendix B: Semi-Structured Interview Guide

Introductory questions

1. What are your experiences with the NeuroDev study?
    a. What do you remember about the day you were recruited?
    b. How was the process of receiving the result?
        i. Expectations?
    c. How do you feel about having this interview today? (PROMPT: Nervous, confused, not sure)

Experiences of stigma.

2. What are your experiences with living with a child with a condition where the cause is unknown?
    a. What did you believe the cause of the condition was? (PROMPT: Act of God, curse/witchcraft, supernatural, stress, poverty, nature)
    b. What are your views on this condition happing again for a future child?
    c. Have you felt guilty or at fault for your child's condition?
        i. Could you tell me about why you feel this way?/ What does that feel like?
    d. How has this impacted your self-concept? (PROMPT: not good enough, best parent I can be)
    e. How has this impacted your social relationships? (PROMPT: self-conscious, embarrassed)
    f. Have you stopped working/close relationships/concealed these problems?
        i. What does that feel like?
    g. What is the most difficult/hurts the most?
3. How have family or people in your community responded to your child's condition.
    a. Have you been treated unfairly by your spouse?
        i. How have you been treated unfairly?
        ii. How does that feel?
    b. Have you ever been blamed for causing your child's condition?
    c. Have you had any negative experiences/attitudes because of your child's condition? (PROMPT: <u>At work</u>: missing work, exclusion, not being able to do

certain types of work, Schools: not able to attend certain schools, Public places: mall, restaurants, accessibility, transport)

d. How have people shown that they feel differently about you child? (PROMPT: avoided, insulted, treated poorly, threatened, attacked)
e. What experiences of isolation or discrimination have you experienced in your community?
f. What type of external support have you received? (Prompt: Church, family/ friends support, WhatsApp groups)

How a positive result alters experiences stigma

4. Could you tell me how a positive result makes you feel?
    a. What kind of answers does it reveal for you as a family? (PROMPT: blame, guilt, gendered blame, stigma) or how would your life change knowing that the condition has a genetic cause?
        i. Relief?
    b. How would your extended family or people in the community perceive this result?
    c. How would knowing the genetic cause of your child's condition impact relationships with family or community members? (PROMPT: Be more accepting)
    d. How would knowing the genetic cause of your child's condition change negative experiences for you? (PROMPT: At work, school, public places)

The role of genetic counsellors in alleviating stigma in genomics research

5. What could the NeuroDev study do to help you with feelings of guilt, blame, shame or discrimination?

6. What is your understanding of Genetic Counselling?
    a. What experiences have you had with a genetic counsellor?
    b. How would talking to a professional that provides support, genetic knowledge and understanding of your personal/cultural/family experiences be beneficial in this process for you? (PROMPT: explain results, what this means for the family, management)

c. How often would you like to have a session with a genetic counsellor in this process? Or when would you prefer to meet with a genetic counsellor while participating int the NeuroDev study? (PROMT: Before testing, after testing, after results have been communicated)

Ending question

7. Is there anything that we haven't discussed that you would like to talk about?
8. Is there another close family member/carer that will be willing to participate in this research?

Appendix C: Information Sheet for Research Participants

Dear participant.

You have been invited to participate in this research study due to your involvement in the clinical confirmation phase of the NeuroDev Research Study which will allow for the return of a positive genetic finding (returnable genetic result).

The study of interest is being conducted in the Human Genetics division at the University of Cape Town as a part of a minor dissertation for partial fulfilment of the MMedSc Genetic Counselling degree.

The study aims to understand the experience of parents and caregivers with stigma and how a positive result alters this experience. It further aims to ascertain the role of genetic counsellors in addressing instances of stigma in genomics research.

In order to acquire this information, interviews will be conducted at Red Cross War Memorial Children's Hospital or at the University of Cape Town where transport will be arranged for you if required. It is anticipated that these interviews will take approximately 2-3 hours in length. The interview will be audio-recorded and transcribed for the use of the researcher and her supervisors only, and will remain confidential by password protection. Certain quotes from the audio-recorded interview may be used in the final report of this study, however, identifying information will be pseudonymised.

Participation in this research study is voluntary and you may withdraw from the study at any point. Acquired information will then be discarded appropriately. There is no direct benefit to the research participants. Due to the sensitive nature of the questions, referrals to suitable health care professionals will be organised upon request.

Please contact Ms Nolene Chetty at 0796962613 or CHTNOL001@myUCT.ac.za or Zandre Bruwer at Zandre.bruwer@uct.ac.za or Associate Professor Jantina De Vries at Jantina.devries@uct.ac.za or Professor Kirsten Donald at Kirsty.donald@uct.ac.za for any further enquiries or Professor Blockman Marc.blockman@uct.ac.za for any queries related to ethical concerns/rights as a participant.

Please see attached consent form.

Regards, Nolene Chetty

Appendix D: Consent Form

Returning Individual Genetic Results to Research Participants: Experiences of Stigma from South African Families with Neurodevelopmental Disorders

I, _____,
agree to participate in the study.

I understand the subject of research and purpose as explained by the researcher and provided in the information sheet.

I confirm that the researcher (Nolene Chetty) has explained the information of the study to me in a language (English) that I am proficient in and my questions have been answered satisfactorily.

I understand that my participation in this study is voluntary and that I may withdraw at any point during the study without consequence to the medical care of my child or the process of receiving the genetic test results.

I am aware that this is a once off procedure that will be implemented in 2022/2023 at a time convenient to me.

I understand that I may refrain from answering questions that may be uncomfortable with no consequence.

I understand that I or my child/children may not directly benefit from the research.

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

I understand that my identity or other identifying information will remain anonymous. Prior to analysis, transcripts will be pseudonymised to prevent identification.

I understand that audio-recordings and transcripts will be safely stored on the researcher's password-protected device and will only be accessed by the researcher and/or her supervisors.

I understand that sections of my interview may be anonymously quoted in the minor dissertation, presentations or academic posters.

Signature of Participant

_____(Signature) _____(Date)

Signature of Researcher

_____(Signature) _____(Date)

Appendix E: HREC Approval

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

Room 45 E-52-E-Floor- Old Main Building
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12 December 2022

HREC REF: 734/2022

Dr Z Bruwer
Department of Paediatrics and Child Health
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Dear Dr Bruwer

PROJECT TITLE: RETURNING INDIVIDUAL GENETIC RESULTS TO RESEARCH PARTICIPANTS: EXPERIENCES OF STIGMA IN SOUTH AFRICAN FAMILIES WITH NEURODEVELOPMENTAL DISORDERS-SUB-STUDY LINKED TO 810/2016-MMED SCI CANDIDATE-MISS NOLENE CHETTY

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

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

Approval is granted for one year until the 30 December 2023.

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

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

Please quote the HREC REF 734 /2022 in all your correspondence.

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

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

Yours sincerely

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

HREC/ref 734.2022

Federal Wide Assurance Number: FWA00001637. Institutional Review Board (IRB) number: IRB00001938 NHREC-registration number: REC-210208-007
This serves to confirm that the University of Cape Town Human Research Ethics Committee complies to the Ethics Standards for Clinical Research with a new drug in patients, based on the Medical Research Council (MRC-SA), Food and Drug Administration (FDA-USA), International Council for Harmonisation of Technical Requirements for Pharmaceuticals for Human Use: Good Clinical Practice (ICH GCP), South African Good Clinical Practice Guidelines (DoH 2020), based on the Association of the British Pharmaceutical Industry Guidelines (ABPI), and Declaration of Helsinki (2013) guidelines. The Human Research Ethics Committee granting this approval is in compliance with the ICH Harmonised Tripartite Guidelines E6: Note for Guidance on Good Clinical Practice (CPMP/ICH/135/95) and FDA Code Federal Regulation Part 50, 56 and 312.

Appendix F: RCWMCH Approval

UNIVERSITY OF CAPE TOWN
SCHOOL OF CHILD & ADOLESCENT HEALTH
DIVISION: PAEDIATRIC MEDICINE
RED CROSS WAR MEMORIAL CHILDREN'S HOSPITAL
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Professor Kirsty Donald
Division of Developmental Paediatrics
23.01.2023
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Red Cross War Memorial Children's Hospital
Research Committee

Dear Sir/Madam

RE: Head of Division Approval to Conduct Research at Red Cross War Memorial Children's Hospital

This letter is to approve the proposed sub-study titled: Returning Individual Genetic Results to Research Participants: Experiences of Stigma in South African Families with Neurodevelopmental Disorders.

This study falls under the umbrella of the NeuroDev study which is being conducted at Red Cross War Memorial Children's Hospital. Approval for the NeuroDev study has been received from both the Western Cape Government Department of Health and Red Cross War Memorial Children's Hospital under the NeuroDev Human Research Ethics Committee (HREC) study reference 810-2016.

For the proposed sub-study, approval from the Department of Paediatrics and Child Health Research Committee as well as the HREC has been obtained. Under these conditions, approval from Red Cross War Memorial Children's Hospital is motivated. No personnel or resources will be required for this student project

Yours sincerely,

Prof. Kirsty Donald
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Prof. Rudzani Muloiwa
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