Approach to Congenital Disorders: Referral Pathways for Genetic Services in the Eastern Cape

by

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NTNSES001

SUBMITTED TO THE UNIVERSITY OF CAPE TOWN

In (partial if the degree was by coursework AND dissertation) fulfilment of the requirements for the degree

MMedSc Genetic counselling

Faculty of Health Sciences

UNIVERSITY OF CAPE TOWN

Date of Submission

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DECLARATION PAGE

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Acknowledgements

I would like to extend my gratitude to the following individuals for their contributions to this research:

Firstly, I would like to acknowledge God for giving me the strength to see this through.

I would like to thank the participants of the study. I appreciate your trust and confidence in me to relay your experiences.

To my supervisor, Prof. Tina- Marié Wessels, thank you for all your support, and guidance and for being available to assist. I appreciate your feedback and recommendations.

To my co-supervisors:

Prof. Karen Fieggen, thank you for always availing yourself and your willingness to assist. All your recommendations and feedback were invaluable. Thanks for encouraging me to keep going.

The management at Oliver and Adelaide Tambo Hospital, thank you for your assistance and offering direction where needed, a special note of gratitude to Mrs Ndabisa, Mrs Bhekizulu and Ms Yolisa Ngcobo.

My fellow student colleagues, thank you for your support and encouragement.

University of Cape Town Post Graduate Funding Office (PGFO) for funding this course.

To my mother (Mangwanya,Jola,Mphankomo) and sister (Nwabisa Ntanjana), andiyonto ngaphandle kwenu. Thanks for always pushing me forward and believing in

everything I do; your unwavering support is more than I could ever ask for. To my late father (Zulu ka Ntombela) thanks for always being a guiding light.

To all my friends and the rest of my family thank you for the care, guidance, support, and encouragement. Sithokozile Bridget Moyo, thanks for being my sounding board.

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List of Abbreviations

OCA- Oculocutaneous Albinism

NTD- neural tube defects

DS-Down syndrome

NHI-National Health Insurance

TB- Tuberculosis

HIV- Human Immunodeficiency Virus

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Abstract

Congenital disorders are defined as abnormalities of either a structural or functional nature. These abnormalities are present at birth; however, they may sometimes only be observed later in life. The underlying cause of congenital disorders can be attributed to genetics or, environmental effects or both, as well as unknown factors in some instances. These disorders are known to affect approximately 1 in 15 live births, yet, in South Africa they are underreported.

The underrepresentation is partly due to the lack of skills and resources to successfully diagnose patients, therefore, contributing to the neonatal and infant mortality rate. When congenital disorders are not considered and correctly diagnosed, the implications for management and for recurrence risk in the family remains unclear. The burden of these disorders in low middle-income countries (LMIC) is underappreciated.

Referral to genetic services aids in diagnosis, management, and the interpretation of the family history to ascertain the implications and recurrence risk of a disease. The aim of this study, therefore, is to explore the experiences and perceptions of health care professionals concerning the utility of genetic services for congenital disorders in rural Eastern Cape through a descriptive phenomenology approach.

Methods: Qualitative research, drawing on principles of phenomenology was used as the study design. The sample for this study included healthcare professionals who were involved in the management of patients affected by congenital disorders. A research advertisement was disseminated with the assistance and permission from the hospital's management. Semi-structured face-to-face interviews with open-ended questions were used to collect the data and close-ended questions were used to obtain the demographic data. The interviews were recorded and transcribed verbatim. The data were analysed using a thematic data analysis approach. A total of 10 healthcare professionals were interviewed.

Results: Four themes were identified in this study. These include "Challenges in rural healthcare", "Service provision", "Genetic counselling" and "Genetic awareness and education". These themes often overlapped and could be described in three overarching ideas, namely logistic factors, human factors, and guidelines which illustrated the determinants of the approach to congenital disorders in rural areas.

Discussion and Conclusion: The study highlighted the challenges that exist for patients outside urban areas such as the lack of access to healthcare facilities and genetic information. It highlights the lack of training and guidelines on congenital disorders as a hindrance to achieving optimal patient satisfaction. Empowering healthcare providers outside of tertiary hospitals is instrumental in improving health outcomes. The research participants stressed the importance of genetic awareness among healthcare professionals, as they need to be able to empower patients by sensitising them to the causes of congenital disorders, their genetic risks, and options. Finally, the results showed that the participants had a superficial understanding of genetic counselling, thus indicating the need for raising awareness about this profession among healthcare professionals.

Chapter 1: Literature review

Background

Congenital disorders are defined as abnormalities of either a structural or functional nature. These abnormalities are present at birth; however, they may sometimes only be observed later in life. The underlying cause of congenital disorders can be attributed to developmental and genetic factors, environmental effects or both, although in some instances, the aetiology remains unknown. These disorders are known to affect approximately 1 in 15 live births, yet, in South Africa they are significantly underreported (Malherbe et al., 2017). The diagnostic pathway for congenital disorders is often lengthy and requires extensive evaluations which can be costly and require invasive testing. In early life, genetic testing is usually for the detection of aneuploidy or chromosomal structural variations and single nucleotide variants which is guided by the abnormalities noted in sick neonates (Qi et al., 2024). Access to genetic testing in low middle income countries is reported to be lower than in high income countries and is dependent on the genetic condition considered (Gatto et al., 2021)

Furthermore, the underrepresentation of congenital disorders contributing to the neonatal and infant mortality rate is partly due to the lack of skills and resources to successfully diagnose and manage patients. When congenital disorders are not considered and correctly diagnosed, the implications for management and for recurrence risk in the family remains unclear. The burden of these disorders in low middle income countries (LMIC) is underappreciated, even though in 2013 congenital disorders were reported to have been the third leading cause of death in newborns contributing to the neonatal and infant mortality rate, overtaking deaths due to infections (Malherbe et al., 2017).

In some developing countries, like Argentina, successful strategies have been implemented to reduce the mortality rate of children under the age of five. This was achieved through the seamless integration of early identification, referral, and care for children with congenital disorders. The country set up hospital-based registries in selected hospitals across several provinces, which played a crucial role in pinpointing and addressing the root causes of congenital disorders. These registries not only ensured the delivery of essential interventions but also served as a vital tool for

collecting and maintaining epidemiological data through collaboration with both private and public hospitals (Smythe et al., 2022).

Referral to genetic services aids in diagnosis, management, and the interpretation of the family history to ascertain the implications and recurrence risk of a disease or disorder (Guttmacher et al., 2007) . The aim of genetic counselling is to empower patients through education. The genetic counselling process is focused on the promotion of informed choices while facilitating the transition from diagnosis to acceptance of the risk associated with the disorder, adjusting to living with the condition and considering the familial implications (Resta et al., 2006).

However, genetic services are not always accessible, even in well-resourced countries, especially in areas that are remote (Best et al., 2022). Best et al., (2022) conducted a systematic review which included 12 studies from the United States of America, four from the United Kingdom, two from Canada, and one1 study from Brazil and Mexico respectively. They focused on assessing how existing service models, workforce capacity, logistical issues, and cultural beliefs influence access to genetic services, to provide insights into improving the delivery of genetic and genomic care for individuals with rare diseases, regardless of their geographical location.The findings highlighted systemic issues within the service delivery models which impaired the distribution of genetic services such as the lack of investment in rural services due to the underestimated demand for genetic services that is informed by the lack of awareness regarding such services and the existing referral pathways owing to geographic locations. They found that the design of service models was limited and dependent on assumptions of need and advocacy of local communities and further exacerbated by the lack of local providers and community knowledge about genetic services, however there was a desire to see geographically equitable genetic services with the first step being a needs assessment and the development of locally appropriate strategies. Workforce capacity was challenged by the inability of the healthcare providers to keep up to date on their knowledge due to the lack of contact with specialists as rural healthcare professionals usually provide generalist care. Moreover, the inadequate awareness of genetics hampered the incorporation of genetics into rural healthcare practitioners' workload due to a lack of guidelines for the management of genetic conditions. Logistical issues such as the lack of access to transportation and additional costs associated with attending genetic appointments

and transportation of genetic samples were highlighted as challenges that are faced when providing genetic services in rural areas as well as the culture of “doing without” which is often adopted in rural areas (Best et al., 2022).

Background on South Africa

South Africa has 60.6 million people spread across the country’s nine provinces, the population is reported to have grown by 19.8% since 2011 *(*Statistics South Africa | Census Dissemination*, n.d.)*.There are currently 257 municipalities in the country, of which eight are metropolitan municipalities, 44 are district municipalities, and 205 are local municipalities that have the primary role of growing local economies and providing infrastructure and services. They are allocated a budget to carry out their designated duties, and they can also raise funds through municipal rates and taxes. Moreover, they have a share in the national revenue to help them provide services to poor households and they can access additional funding through grants raised by the national government which may be conditional or non-conditional (Monkam, 2014).

The Eastern Cape consists mainly of former Homelands, which were designated for Black South Africans based on their racial classification, prior to 1994. These areas were deliberately under-developed, under-resourced, geographically segregated, and self-governed with minimal autonomy and high dependence on the national government for funding which resulted in the rise of poverty and limited opportunities for economic development (Nhlapo et al., 2011). The local government in the Eastern Cape is made up of two Metropolitan Municipalities namely, Buffalo City and Nelson Mandela Bay, while the rest of the municipalities are district municipalities.

Alfred Nzo District municipality is located on the northeastern side of the Eastern Cape province (see Figure 3) and includes Sisonke District Municipality and O. R Tambo District Municipality (EC Provincial Website, n.d.). The district has a total population of 878 635 individuals which accounts for 12.1% of the total population of the Eastern Cape province. There are 72 clinics, eight hospitals, and two community health centers in the district. The district has the most under resourced health facilities with problems such as severe staff shortages, low-security

measures, and unreliable facilities *(*Alfred Nzo District Municipality EC Profile and Analysis District Development Model*, 2020).*

The majority of the population of Alfred Nzo District Municipality are Black Africans (98.8%). Females make up majority of the population (54%) and most are young, aged 0-35 (77%). According to population statistics, 10.4% of the population has no formal schooling, 29.3% have completed secondary schooling while 17.9% have achieved higher education. 21.7% of the households do not have a formal income (Statistics South Africa, n.d.).

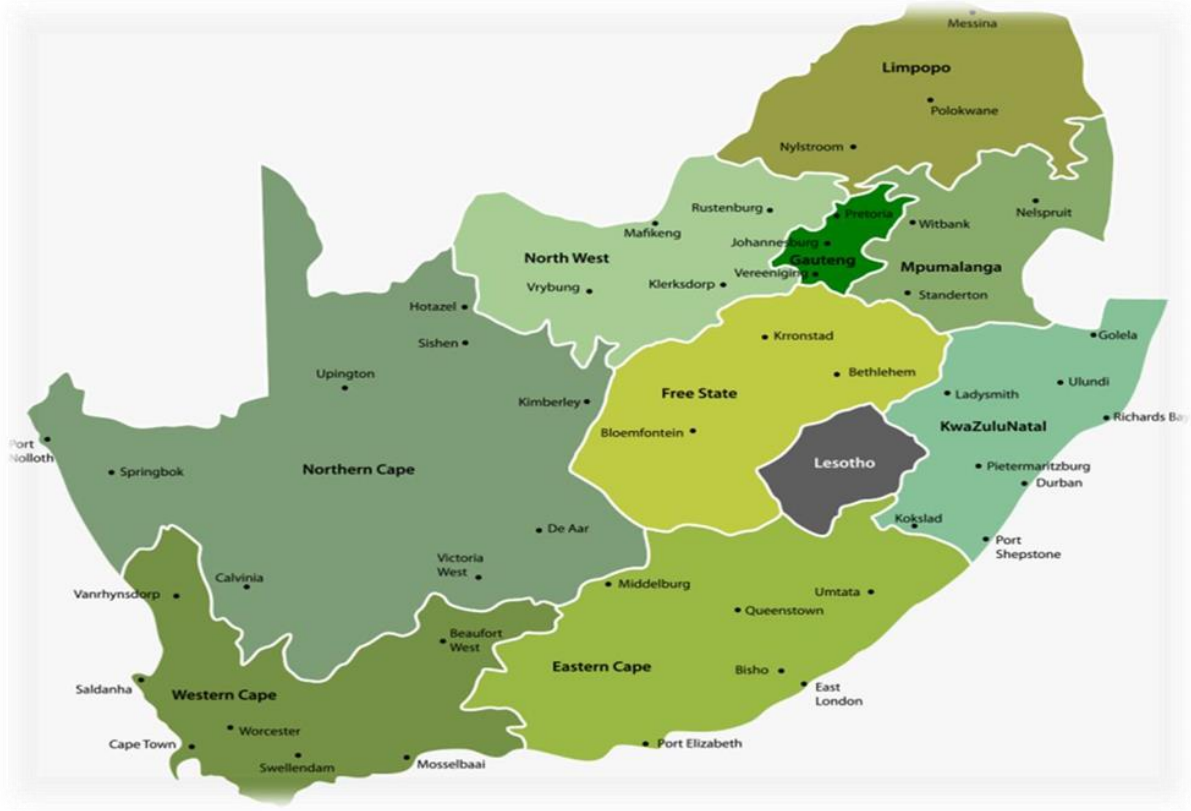

Figure 1: Map of South Africa (sourced from www.google.com)

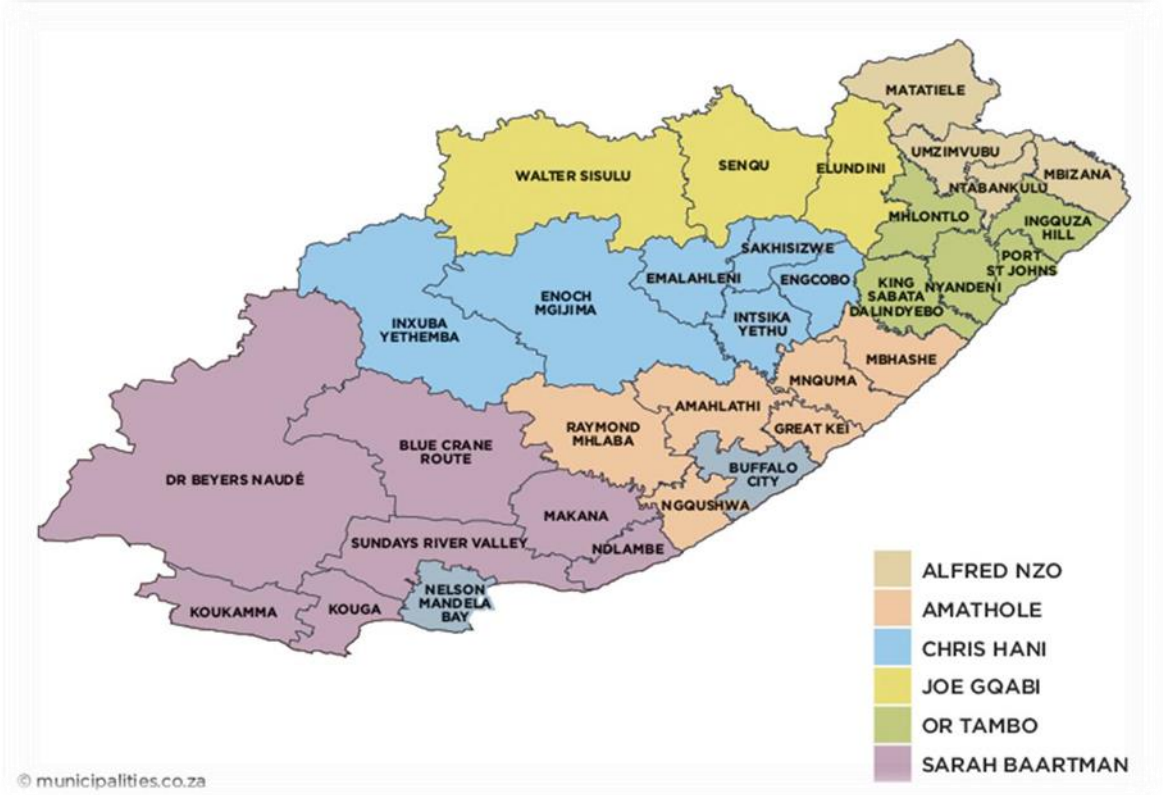

Figure 2: Map showing the Eastern Cape Municipalities (adopted from www.municipalities.co.za)

Health care system

The healthcare system in South Africa is divided into the public and private sectors. Of the population, 82.42% depend on the public sector (Africa, 2017). The private sector is funded through out-of-pocket charges as well as the aid of medical schemes. It consists of general practitioners, specialists and private hospitals. However, as the majority of South African citizens cannot afford private health care, they rely on the public health sector which is funded by the National Department of Health. The dual nature of the South African health care system has been criticised based on the unequal resource distribution (Coovadia et al., 2009). The public sector is characterised by being overburdened with providing care for the majority of the population and is under-resourced. Efforts have been made through various policies to combat these disparities in resource distribution and to expand healthcare to benefit those severely disadvantaged (Mayosi et al., 2012).

In 2011, the national white paper on National Health Insurance (NHI) in South Africa was published. The document detailed the initial vision, the goals and the

implementation strategies of the proposed National Health Insurance. The National Health insurance is aimed at bridging the gap between the private and the public sector by providing universal access to quality and affordable healthcare for all South Africans. This is proposed to be attained through the pulling of financial resources from taxes and contributions from individuals and employers, utilising the services of both public and private providers. The fund will be responsible for the purchasing of healthcare services on behalf of the citizens and the same standard of care will be expected from all the providers, the services that will be provided will be essential care such as primary care, hospitalisation, and emergency services. Foreseen challenges with the NHI include financial concerns as the implementation hinges on sufficient funds being raised to cover the growing healthcare needs of the citizens, capacity of the healthcare system to cater to the demand and the political will for long-term successful implementation.

Healthcare services

Healthcare in the public sector is provided in a tiered system which comprises different levels of care. The primary health care tier serves as the first point of service for the majority of patients. These facilities are equipped to handle basic diagnosis, prevention, and therapeutic care for common conditions. These facilities include clinics and community health centres, they also serve as the gateway for facilitating referral to higher levels of care. Challenges at this level of care include lack of adequate health care professionals to cater for the demands of the populations that they serve, therefore the waiting times at these facilities are often long and these facilities often lack specialised services (Pauw, 2022). The second tier is the district hospital level (level 1 hospital) where patients access generalised care such as slightly more extensive investigations, basic surgeries and the management of chronic illnesses. The main challenge at this level of care involves the inability to accommodate patients which results in long waiting lists to be seen by more specialised professionals and limited beds available for hospitalisation of patients in need of in-patient services (Coovadia et al., 2009). The regional and provincial tertiary hospital tier is at the apex of the public healthcare sector, it offers the most specialised care providing access to

advanced equipment and expertise for more complicated investigations and diagnoses (Tefera et al., 2020). These tertiary facilities are concentrated in urban areas with limited reach to rural residents. The challenges faced by patients include difficulty accessing information about specialised facilities and transportation; both lead to delays and ineffective access to services (Vergunst et al., 2017). This unequal distribution of resources exacerbates existing healthcare inequalities (Maphumulo & Bhengu, 2019).

Levels of decision making

In South Africa, there are three levels of government, namely national, provincial, and local government. Each level has its own responsibilities and duties, but they work together in an interconnected system (Pauw, 2022). Local governments are divided into metropolitan municipalities, which serve large cities, and district municipalities, which cover multiple local municipalities in a particular region in a province. This branched-out structure ensures that there are direct relationships and responsibilities between the national government and provinces (Monkam, 2014).

Governance and service delivery

The governance and organisation of healthcare in South Africa is regulated by the National Department of Health, which sets the national policies and provides technical support to provinces and monitors health outcomes (Pauw, 2022). The provincial departments of health are responsible for developing provincial policies in line with the national policies and delivering the healthcare services in their respective provinces. The district health authorities oversee the management of healthcare facilities and services at the district level (Pauw, 2022). Districts are responsible for the implementation of national and provincial policies by translating these policies into actions at local level. They co-ordinate service delivery by engaging with communities to understand their needs to incorporate them into their service delivery plans and are accountable to the national and provincial governments (Coovadia et al., 2009).

History of Genetic services in South Africa.

Genetic services in South Africa have been available for approximately 70 years. These services were developed and initiated in larger metropolitan areas such as Cape Town and Johannesburg. These services were later adopted by the Department of Health and Welfare in 1971(Kromberg et al., 2013). Prior to 1990, these were only available in urban areas as they were attached to academic centers and only served a small proportion of the population at the time. As a result, little epidemiological data were available on the majority of the population, specifically black South Africans. The only well-researched condition was Oculocutaneous Albinism (OCA). Down syndrome (DS) and neural tube defects (NTDs) were thought to be less prevalent in this population until the initiation of as community genetics initiative that sought to target rural populations to determine the incidence of these conditions. The program was initiated by the University of Pretoria, with the Department of Health in the Northern Transvaal as the area was known then. The surveillance program revealed that the prevalence of DS, NTDs and OCA were higher than they were thought to be (Christianson, 2001).The demand for prenatal testing increased as screening programmes were initiated. Programmes were introduced to train healthcare professionals. As part of this training program, genetic nurses were introduced to manage common genetic conditions as a response to the increased need for genetic services that stemmed from the development of outreach genetic clinics (Beighton et al., 2012).In this way, the management of common genetic disorders was done at a primary care level. Patients who needed more specialised management were seen at genetic outreach clinics or the necessary referrals were made by the trained genetic nurses. The institutions involved in the training were expanded to include Pretoria, Stellenbosch, and Bloemfontein (Christianson, 2001).

Post-democracy, enshrined in the South African constitution, section 27 is the right for everyone to have access to healthcare services. This includes individuals with congenital disorders (Malherbe et al.,2016). Since 1994 the government has attempted to ensure that all citizens receive equal access to health care, through the district health system as well as other initiatives including the nationalisation of health laboratories (Beighton et al., 2012). This aimed to improve services in both urban and rural areas. The goal was to develop sustainable rural

communities that are integrated development sites (Barron & Asia, 2001). In 2001 the human genetics policy guidelines for management and prevention of genetic disorders, birth defects, and disabilities were established to outline how genetic services should be delivered nationwide. This policy was based on the hub and spoke model (Health, 2001). This model recognises the central areas as the hubs that would service the country at large through outreach(Greenberg et al., 2012). Furthermore, in the National Health Act, genetic services are recognised as one of the required services. However, although a legislative framework exists, there are shortcomings in the implementation of these policies (Malherbe et al., 2016). These shortcomings are partly due to the prioritisation of infectious diseases, such as Human Immunodeficiency Virus (HIV) and Tuberculosis (TB) as well as the shortage of qualified personnel required to implement genetic services in the country (Greenberg et al., 2012). This has resulted in some medical genetic services declining since their inception. South Africa is a country that has failed to show a continued development and improvement of medical genetic services (H. Malherbe et al., 2016). Currently, access to these services is largely through tertiary care institutions (Kromberg et al., 2013). The services offered include prenatal genetic diagnosis, diagnostic, predictive and carrier testing as well as genetic counselling services to raise awareness regarding genetic conditions(Kromberg et al., 2013).

Health care professional's role in referral for genetic services.

Healthcare professionals at the primary and secondary care level play a pivotal role in ensuring that patients have equal access to healthcare resources as well as recognising symptoms and making the necessary referrals to ensure adequate healthcare assessments and appropriate diagnoses are made (Nguyen et al., 2022).

The referrals require good communication and understanding between the healthcare providers at all levels of care involved in the management of the patient to provide effective continuous care (O'Malley & Reschovsky, 2011).

In their review, Malherbe et al. (2017) highlight the importance of training primary healthcare staff to accurately recognise, monitor and diagnose common

congenital disorders (Malherbe et al., 2017). This is especially important in settings that lack genetic services, which is currently the case for the majority of the population (Saib et al., 2021). However, training of healthcare professionals in genetics is currently not standardised (Phaladi-Digamela et al., 2014). Primary healthcare professionals need this knowledge in order to optimise the quality of life of people affected by congenital disorders, as these disorders require lifelong management (McGrath et al., 2022). Although some of the rural population have access to genetic services through outreach clinics, these initiatives only exist in selected areas (Kromberg et al., 2013). Nurses in under-resourced areas, especially in rural areas, contribute significantly to antenatal care, labour, delivery, and care of newborns and are essential in facilitating optimal prevention and care in congenital disorders (Malherbe et al., 2017)

The importance of education and training of nurses is emphasised in the Human Genetics Policy guidelines for the management and prevention of genetic disorders, birth defects, and disabilities published in 2001, and updated in 2021. The document has established a list of priority conditions. These conditions include Down syndrome (often related to advanced maternal age); neural tube defects; fetal alcohol syndrome; and albinism. The aim of the policy is to ensure that these common conditions are identified and managed accordingly. The guidelines emphasise the level of care that should be afforded to people living with congenital disabilities and to families that are at an increased risk of inherited congenital disorders as prevention of these disorders reduces their social and economic impact. The policy further details the role of community-level care in the delivery of maternity care and genetic services for most conditions and defines referral pathways (Health, 2001). These guidelines are designed to facilitate continuous quality care for people affected by congenital disorders (van der Weijden et al., 2013).

Cultural views of congenital disorders

South Africans have diverse and complex cultural views regarding the cause of congenital disorders which reflects the country's rich diversity based on ethnicities, religious beliefs, and traditional practice. These conditions can be attributed to supernatural causes such as curses from ancestors, angered spirits,

or witchcraft. This is particularly common in rural areas and in certain cultural groups (Penn et al., 2010).

Other beliefs include linking the congenital disorder to the mother's behaviors during pregnancy such as food taboos or emotional distress due to events such as abuse or trauma during pregnancy, not mourning adequately a pregnancy loss due to miscarriage and not loving a child who was born with a congenital disorder thus leading to the punishment by a Devine power (Penn et al., 2010). Misconceptions about the causes of congenital disorders can therefore lead to stigma, blame on women who mother children with congenital disorders, and discrimination (Kromberg et al., 2013).

Cultural beliefs therefore have an impact on health-seeking behaviors when congenital disorders are believed to be caused by superstition and supernatural forces. In such cases, families may turn to alternative solutions, such as traditional healers and religious healing centers, based on their belief systems. This is especially common in areas with limited medical infrastructure, such as rural areas (Lamptey, 2019). However, many communities recognise the genetic component in congenital disorders thus acknowledging family history as a potential factor (Kromberg et al., 2013).

Rationale:

The Eastern Cape is home to around 7 million people, making it the third most populated province in the country. Of this population, approximately 4.1 million reside in non-urban areas, which include rural and peri-urban settlements. There are currently very few genetic services in most provinces of South Africa including the Eastern Cape and none in rural areas. The aim of this study, therefore, is to explore the experiences and perceptions of healthcare professionals concerning the utility of genetic services for investigating and managing congenital disorders in rural Eastern Cape, a province that does not have specialised genetic services.

Research Question:

What are the perceptions and needs concerning the utility of genetic services for congenital disorders in rural Eastern Cape?

Aims and Objectives

Aim:

To explore the perceptions and needs of health care professionals concerning the utility of genetic services when managing congenital disorders in rural Eastern Cape.

Objectives:

1. To explore the perceived role of genetic services when managing congenital disorders in the Eastern Cape.
2. To assess the current referral pathway for genetic services available to healthcare professionals outside tertiary services in the Eastern Cape.
3. To explore the perceived role of genetic counselling in the Eastern Cape.
4. To explore how existing services for congenital disorders could be improved.

Chapter 2: Methods

Study design

Rationale

The study aimed to explore the perceptions of healthcare professionals concerning the utility of genetic services for congenital disorders in a rural area in the Eastern Cape. A study of this nature has not been conducted before; thus, the researcher used an exploratory study method. This can serve as foundation for future research and can be used to uncover key issues for further investigation as well as tools for engaging with stakeholders and communities regarding genetic services in the Eastern Cape.

Researcher positionality statement

The research was conducted in the researcher's hometown. The researcher is a South African woman with post-graduate education obtained outside of the Eastern Cape province. Although the researcher was born in a local primary healthcare center referring to the district hospital where the research was conducted, she had not used the public healthcare system in the Eastern Cape. Having grown up in the research setting, the researcher was familiar with the healthcare system through secondhand experience. Her background helped in building rapport with the participants, as some of them had similar backgrounds to the researcher. Social identities have an impact on the manner in which research is conducted and analysed as researchers have lived experiences that shape their worldview which may be therefore projected in the data (Jacobson & Mustafa, 2019).

Research tools

This research draws on the principles of phenomenology. Phenomenology as a paradigm is based on the premise that people make sense of their world through their lived experiences (Finlay, 1999). According to Merriam & Tisdell, (2015), phenomenological studies assume that people experience the world differently, owing to the diversity in lived experiences as well as the cultures that inform the way that they approach everyday life. In this type of research, the experiences of people are grouped, analysed, and compared to identify their essence. It assumes that there is an essence to shared experiences. The essence is the core meaning that is mutually understood through a phenomenon that is commonly experienced (Merriam & Tisdell,

2015:26). In descriptive phenomenology the original data is analysed for meaning in a reflective manner, the researchers using this method are aware of their preexisting biases and therefore bracket them in order to analyse the data with minimal interference from their pre-existing judgements (Connelly,2010:127), the meaning identified is categorised according to similarities and differences that exist. The categories that are generated inform the production of themes. The results are represented by themes which are supported by quotes that represent the background of the findings (Sundler et al., 2019 :737) . This study is interested in the experiences of health care professionals at Oliver and Adelaide Tambo Hospital, this approach seemed most appropriate.

Semi-structured interviews were chosen as these are used in qualitative research to gather detailed and nuanced data about a participant's experiences and perspectives. They offer flexibility to adapt the interview to the needs of the study and the individual. The conversational nature of these interviews builds rapport between the interviewer and the participant, encouraging openness and honesty. Participants are free to share their experiences, leading to the discovery of new perspectives and insights (Melissa & Lisa, 2019).

An interview guide was developed (Appendix A), which is an essential tool for conducting successful semi-structured interviews as it provides a framework for covering key topics while allowing for flexibility to explore unexpected insights. The questions formulated should be open-ended to allow for detailed responses while the researcher remains neutral. A variety of question formats can be used, ranging from broad to more focused and probing questions to gather diverse perspectives. The sequence of the questions should ensure that both broad and narrow topics are covered to ensure specific details are captured (Melissa & Lisa, 2019). The questions included in the interview guide were formulated to include standardised questions which were posed to all the participants as well as exploratory questions. While developing the interview guide the researcher developed the questions to align with the study aim. An example of demographic questions included were regarding education and years of experience at the hospital in order to contexualise the data and seek any patterns that may exist in the sample group. A general opening question was included to establish rapport. An example of exploratory questions included questions

on department-specific activities and participants' understanding of congenital disorders. Test interviews were conducted to test the questions and amendments were made to improve the questions.

Research setting

The research was conducted at Oliver and Adelaide Tambo Hospital which is a provincially funded government hospital in the Alfred Nzo District Municipality. The healthcare professionals working at the hospital comprise 12 doctors, two clinical associates, and two dentists as well as several allied health professionals (two dieticians, four physiotherapists, one speech therapist, two social workers, four radiographers, and three occupational therapists) as well as 45 professional nursing staff and a number of auxiliary nursing staff. This hospital serves a population of approximately 200,000 people. According to the hospital's records the monthly number of infant deliveries conducted at the hospital ranges between 400-550. The hospital services 22 of the 72 clinics in the district (Alfred Nzo District Municipality EC Profile and Analysis District Development Model, 2020).

Ethical considerations

Ethical clearance

The UCT Department of Pathology Research Committee (DRC) approved the study, permission was granted by the deputy chair of the Patholgy DRC, as well as the UCT Faculty of Health Sciences Research Ethics Committee (HREC 012/2023) as indicated in appendix D. The Alfred Nzo District Manager's office also granted ethical clearance, with written permission provided by the district manager, to conduct the research as indicated in Appendix E.

Informed consent and voluntary participation

As potential research participants were required to make "informed choices" regarding their participation, the purpose of this study along with future implications were discussed with the participants and appropriate consent processes were followed. Informed consent is the cornerstone of any research, it is important to uphold the principles of autonomy and free choice. Consent is genuine and operative when it is obtained in a way that is simple and easy to comprehend. Consent should reflect the

needs of the participants and how those needs will be met. The information that informs the consent then should be transparent and detailed, not omitting any facts concerning the research and participation (Klykken, 2021). The participants provided consent after the research study was verbally explained to them. They were given an information sheet for their reference and signed a consent form, which included the research information sheet for the researcher's records for audit purposes.

Privacy and Confidentiality

The privacy and confidentiality of participants was ensured by removing all participant identifiers from the data. All documents or voice recordings containing identifying information were securely stored on an access-controlled computer. Transcription of interviews was done by the researcher. All data was de-identified using a code name or number to protect the privacy of participants. Once data analysis was completed, recordings and notes were destroyed. Due to the demographic profile of the hospital, extra precautions were needed to ensure that the participants were not identifiable from any published interview information. This meant that information such as gender, years of experience, and specialty were minimised to ensure that the privacy of the participants was not compromised.

Risks and Benefits for Participants

The researcher stressed that the details of the interview would be kept confidential, and assured the participants that they did not have to divulge any information that they did not feel comfortable sharing. The participants were assured that the contents of the interview would not be shared with other participants enrolled in the study, their colleagues or employers. Some content of the interviews could cause discomfort as the participants were talking about their work environment as well as lack of services. Participants were assured that they could opt not to answer questions that they did not feel comfortable answering. The work environment details discussed were limited to situations that were relevant to the aims of the research study. The study did not offer any direct benefits to its participants, but it aimed to identify knowledge gaps related to genetic services. The findings of this study sought to shed light on the referral pathways currently used for congenital disorders and the perceptions of healthcare professionals regarding the usefulness of genetic services. Ultimately, the results could trigger discussions on how to improve genetic services in the future.

Participants

The study included, but was not limited to, healthcare professionals that are involved in the management of patients affected by congenital disorders such as senior maternity and paediatric staff members. These participants were chosen based on their direct role in the improvement of care for, and reduction of, congenital disorders and child mortality. According to the World Health Organization antenatal care recommendations (2016) aimed at enhancing the positive pregnancy experience, close attention must be paid to women that are pregnant to identify any problems during pregnancy that would affect the pregnant woman or the child. Focused antenatal care is aimed at preventing future complications as well as identifying risk factors associated with pregnancy (World Health Organization, 2016). Health care professionals in these areas have direct exposure to congenital disorders through physical observations of the children at birth, training and experience in the management of patients through follow up, thus they are knowledgeable about the paths that are followed in diagnosing and managing congenital disorders and were therefore anticipated to be the most informative participants. Furthermore, allied healthcare professionals were also included to explore their experiences as they are involved in the long-term management of children who are living with congenital disorders (Thom & Haw, 2021).

Inclusion and Exclusion criteria

Included in this study were consenting healthcare professionals including doctors, nurses and allied health professionals at the Oliver and Adelaide Tambo District Hospital with at least one year of experience in a unit directly involved in the management of congenital disorders.

Exclusion criteria

Healthcare professionals without experience in congenital disorders and those who indicated interest and later declined participation when contacted by the researcher were excluded from the study.

Recruitment

The hospital management at Oliver and Adelaide Tambo District Hospital was contacted, and the aims and objectives of the study were explained. Permission to recruit the appropriate staff members was requested. The hospital management was asked to assist in notifying the staff members of the research. The invitation to participate in the study was open to all the health care practitioners at the hospital who met the inclusion criteria. However, due to time constraints, the researcher had set a limit of 15 interviews. This number was based on the recommendations by Guest et al (2006) for nonrandom sampling in qualitative research where they found that data saturation is usually reached after six to twelve interviews had been completed (Guest, Bunce & Johnson, 2006).

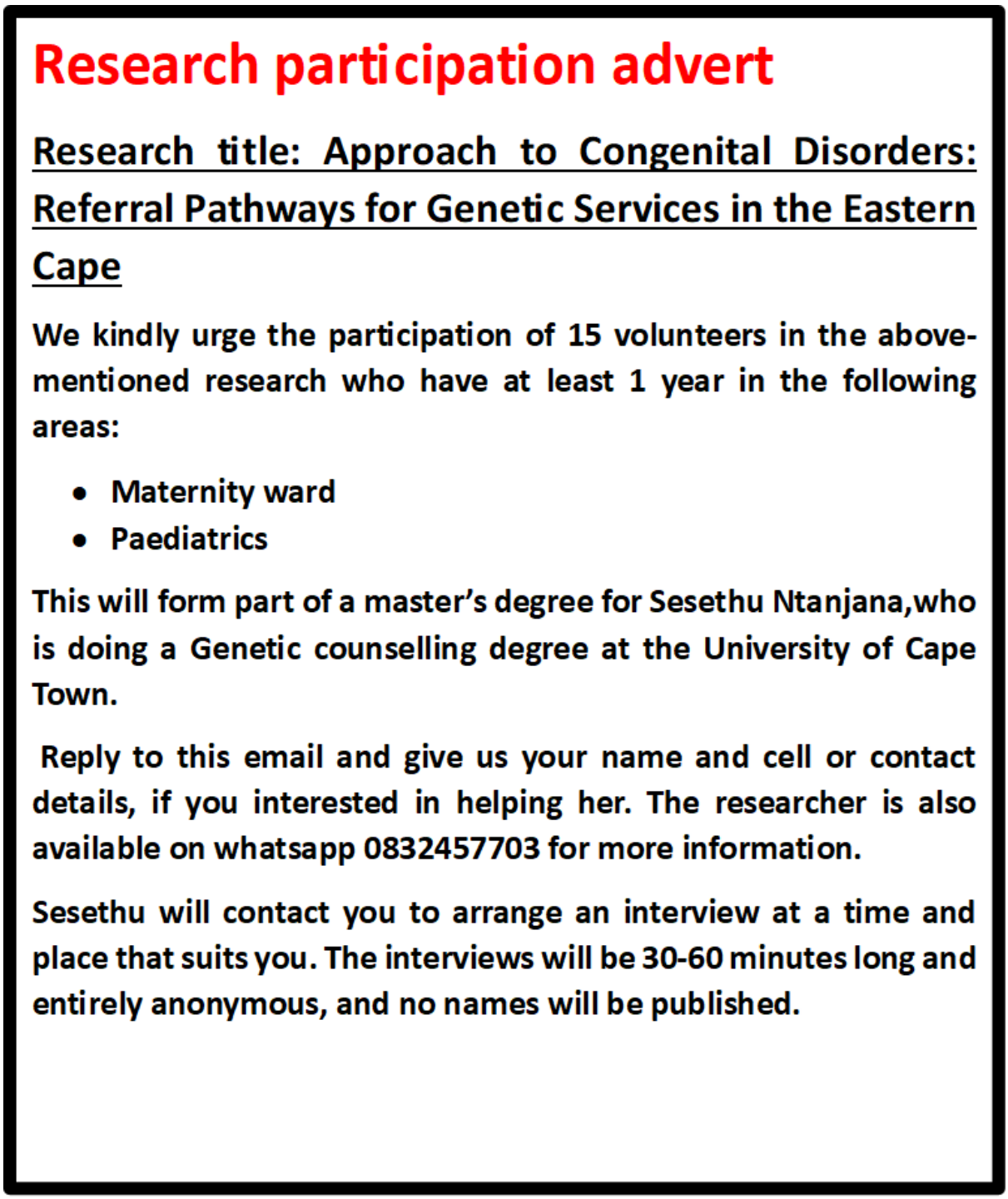

Figure 3: 1Research advert

Sampling technique

In this study, the sampling technique used was purposive sampling. This sampling technique was used in order to target individuals that have been exposed to patients who are affected by congenital disorders and would have the most to share. This technique was suitable for gaining a deeper understanding and insight into the

phenomenon under investigation to ensure the collection of useful data (Merriam & Tisdell, 2015). Interviews with the participants were then designed and conducted to address the study's aim and objectives.

Test interviews

To ensure the validity of the questions devised, three test interviews were conducted. The first was a direct face-to-face test interview with a volunteer genetic counselling student with the research supervisor present to provide feedback on the interviewing techniques and demonstrate how neutrality can be achieved in the interviewing process. Feedback from the volunteer regarding the overall experience was also incorporated to adjust the questions.

Subsequently, a volunteer genetic counselor with experience in interviewing techniques and qualitative research was interviewed by phone to provide feedback on how to improve rapport and the sequence of questions in the interview guide. This feedback was incorporated during the data collection stage of the study. Finally, before conducting interviews with more participants, a face-to-face pilot interview was conducted with a research participant. The interview transcript was shared with the supervisor for feedback.

The purpose of the test interviews was to determine the rigor of the interview guide, with the aim of improving the reliability and strength of the research findings (Majid et al., 2017). The necessary changes were made to ensure that appropriate responses were obtained. Additionally, the test interview allowed for the identification of biases. This provides an opportunity to counteract and manage these biases. (Malmqvist et al., 2019).

During the interviews the participants were asked questions according to the interview guide (refer to Appendix A). The nature of the questions used in the study included closed-ended questions for demographic and practical information and open-ended questions. These questions were followed by probing when necessary and recorded with consent. They were constantly evaluated to ensure that the data obtained was rich.

Data collection

Moderately tailored interview guides were utilised during semi-structured interviews, as outlined in Appendix A. By using these guides, the researcher was able to uncover the perceptions, experiences, and practices of healthcare professionals at the Oliver and Adelaide Tambo District Hospital regarding the investigation of congenital disorders.

The researcher conducted semi-structured interviews with 11 participants to gather individual responses but one was later discarded and excluded from analysis as the participant did not meet inclusion criteria. The data was collected in-person or over the phone. Telephonic interviews were conducted when participants were not available for in-person interviews because they allowed for flexibility. The telephonic interviews were synchronous voice-to-voice phone calls, a cost-effective way to collect data from participants. (Merriam & Tisdell, 2016). However, in-person interviews were preferred over telephonic interviews because they allowed for better rapport building and gave the interviewer an opportunity to pick up on non-verbal cues when probing participants (Merriam & Tisdell, 2016).

Data analysis

The data obtained from the interviews was transcribed, the IsiXhosa excerpts were translated into English by the researcher. During the translation process the excerpts were transcribed in isiXhosa and then translated to English verbatim to conserve the original meaning and content of the excerpts as much as possible (Ozolins et al., 2020). Thereafter, the excerpts were rephrased to read naturally and concisely in English. The translated excerpts were validated by a lay back translator, independent of the researcher. Upon the completion of the back translation, the back translator and the researcher discussed the excerpts and agreed on the equivalence of the translation and ambiguities were clarified to minimize the risk of nuances being lost in the translation process (Ozolins et al., 2020).

The data analysis process that was followed was as described by Braun & Clarke (2021). The researcher read through the responses and reflected on the data numerous times to explore new aspects that were not already known. Thereafter the researcher searched for meanings, and identified and noted the meanings. The meanings were described in a few words then compared to look for similarities. These

meanings were categorised according to the information that emerged. The patterns that emerged were used to develop themes. The meanings were reorganized by being written and rewritten, described throughout the text and named using descriptions derived from actual lived experiences. The researcher is a novice at qualitative research who received training in interviewing skills and qualitative research analysis, therefore the analysis was guided by a supervisor who is experienced in qualitative research. The supervisor read through the transcripts and coded independently. The codes and the themes were discussed throughout the analysis.

Research Trustworthiness

Qualitative research in its nature is subjective and can face challenges in establishing trustworthiness. Measures can be taken towards ensuring that subjectivity, biases and influences from the researcher do not impact the research findings. According to Guba and Lincoln (1986) there are four criteria that must be met to ensure research trustworthiness, namely:

- Credibility
- Dependability
- Confirmability
- Transferability

Credibility speaks to the internal validity of research, ensuring that the findings accurately represent the participants' experiences, and perspectives (Lincoln & Guba, 1986). For the study, the researcher strove for prolonged engagement with the participants to ensure the findings gathered were consistent with the details of the participants' experiences. Furthermore, the researcher transcribed the participants' interviews verbatim, including descriptions of observations made during the interview.

Dependability ensures the reliability and stability of the findings (Lincoln & Guba, 1986). This was achieved by including a detailed description of the study methodology and a comprehensive audit trail that documented the decision-making process during the research. The audit trail was developed to help identify any biases that might have developed and refine the analysis for reliable results.

The following procedure was used to show audit trail (see figure 4 and 5 below):

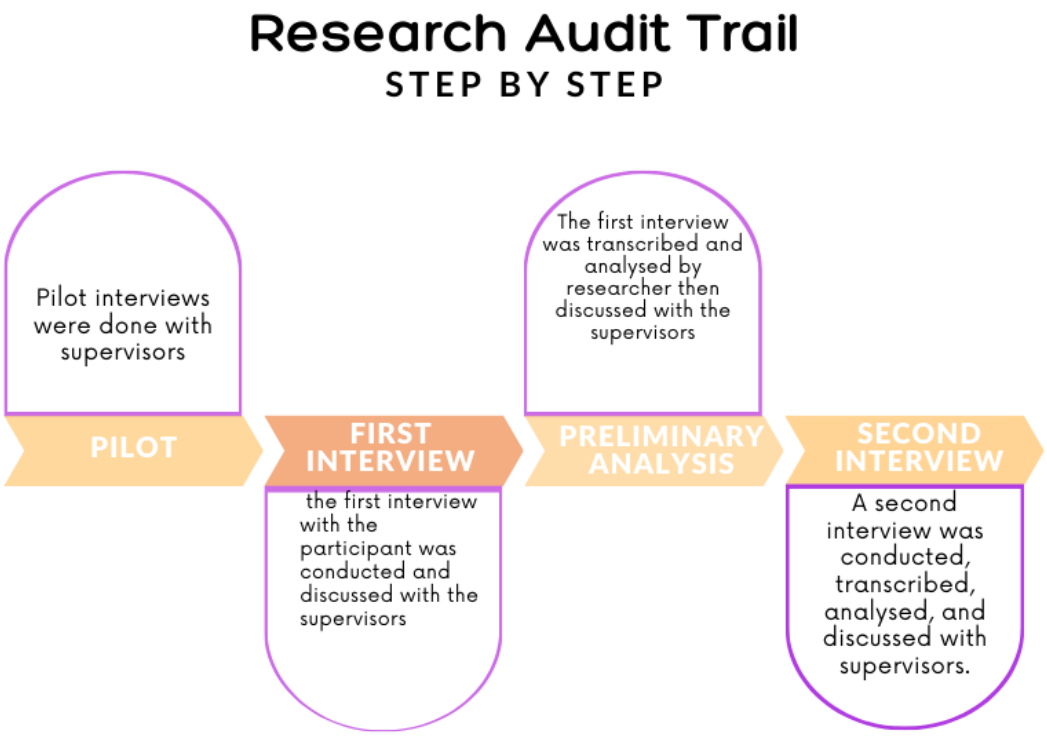

Figure 4: 2 Audit trail.

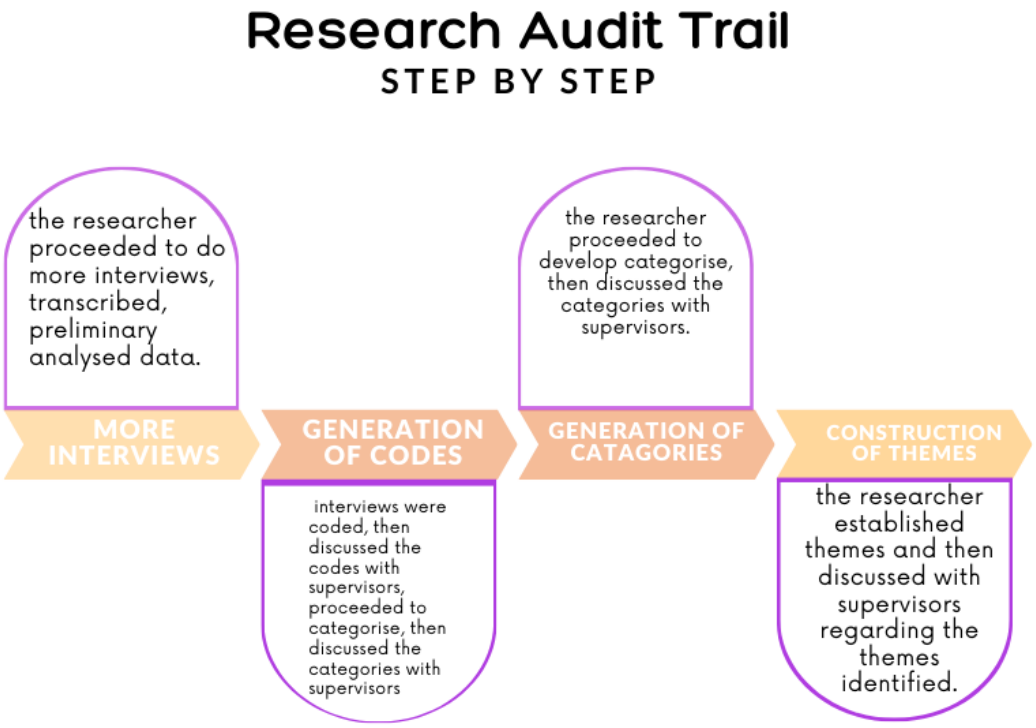

Figure 5: Audit trail

Confirmability is a crucial principle in research that ensures the research findings are not solely based on the researcher's interpretations (Lincoln & Guba, 1986). To validate research interpretation, the researcher used the analyst triangulation method, involving analysis of the data by the research supervisor for feedback throughout the research duration. This helped to validate the findings and uncover any biases that the researcher may have been blind to. Additionally, the researcher continuously reflected on their personal position during the research process and peer debriefing, and additional interviews were conducted as confirmation of data saturation. (See Appendix F for the reflective piece).

The concept of transferability in research is concerned with the extent to which the findings can be applied to other contexts (Lincoln & Guba, 1986). In this study, the researcher described the research setting in detail, and provided a comprehensive overview of the participants' characteristics in Table 1. Measures were taken to preserve the confidentiality of the participants while demonstrating the transferability of the research findings.

To demonstrate the applicability of the findings in a similar research context, a diverse sample of participants was obtained to represent different perspectives and experiences. Although not detailed for reasons of confidentiality, there was significant diversity in training background, previous work experience, duration of working in their current setting and area of work within the hospital amongst participants.

During the research process, the participants' experiences were studied, categorised, and contrasted to determine their fundamental qualities. Through this approach, valuable insights were gathered on the experiences and thought processes of healthcare professionals regarding congenital disorders and how this information informs their approach to managing these conditions in their setting (Merriam & Tisdell, 2016).

Throughout the research, the researcher reflectively examined the data to prevent any preconceived biases from influencing the findings (Connelly, 2010). The generated categories were used to inform the production of themes (Connelly, 2010). The findings were organized into themes with accompanying quotes that provide context for the results (Braun & Clarke, 2021)

Chapter 3 Research Findings

The aim of this research was to explore the management of congenital disorders in a rural setting, as well as the referral pathways for genetic services in rural Eastern Cape. The study was guided by the question: *what are the perceptions and needs surrounding the utility of genetic services for congenital disorders in rural Eastern Cape?*

14 healthcare professionals at Oliver and Adelaide Tambo District Hospital showed interest in participating in the study. One participant was excluded after an initial interview due to limited work experience at this hospital, and three were subsequently uncontactable for successful enrollment in the study. The remaining ten participants were included. Participants were interviewed at their preferred date, time, and location within the hospital.

Table 1 provides an overview of the characteristics of the participants, who included five professional nurses, four allied healthcare professionals, and one general practitioner who has an interest and experience in paediatrics. Eight interviews were in-person, and two interviews were done telephonically, the content of all the interviews was a mixture of Engish and IsiXhosa. The duration of the interviews ranged from 20 to 60 minutes. The interviews were conducted over a period of four months (May 2023-August 2023). The interviews were conducted by the researcher who is a genetic counselling student currently training at the University of Cape Town. The researcher received training in interviewing skills.

All the participants of the study were permanent employees at the hospital. They had received their training from government colleges and universities within South Africa and had one to ten years of experience working at the hospital. All participants spoke both IsiXhosa and English languages. Out of the 10 participants, three had previously worked in different hospitals.

Participant	Profession	Interview location	Language	Years of experience

1	Nurse	Hospital	English and IsiXhosa	2
2	Nurse	Hospital	English and IsiXhosa	13
3	Nurse	Hospital	English and IsiXhosa	13
4	Nurse	Hospital	English and IsiXhosa	12
5	Doctor (general practitioner)	Telephonic	English and siXhosa	1.5
6	Nurse	Hospital	English and IsiXhosa	7
7	Occupational Therapy	Hospital	English and IsiXhosa	4
8	Occupational Therapy	Hospital	English and IsiXhosa	3
9	Speech Therapy	Hospital	English and IsiXhosa	2
10	Physiotherapy	Telephonic	English and IsiXhosa	2

Table 1: characteristics of participants

The researcher conducted in-depth interviews to understand how the participants perceived genetics, genetic services, and the benefits of these services in managing congenital disorders in their hospital. The analysis of the interviews revealed four major themes accompanied by sub-themes, which often overlapped. If a participant's response fit into multiple themes, it was placed in the most appropriate category. Quotes are included to support the findings. Providing healthcare for people living with congenital disorders requires a collaborative approach between the specialists and healthcare providers who offer generalist care. However, access to these facilities poses challenges for both the healthcare professionals and the patients. Therefore, themes 1 and 2 are centred around logistic factors while themes 3 and 4 focus on human factors that inform the current approach to congenital disorders at this hospital, which provides generalist care.

Attached below (figure 6) is an overview of the themes:

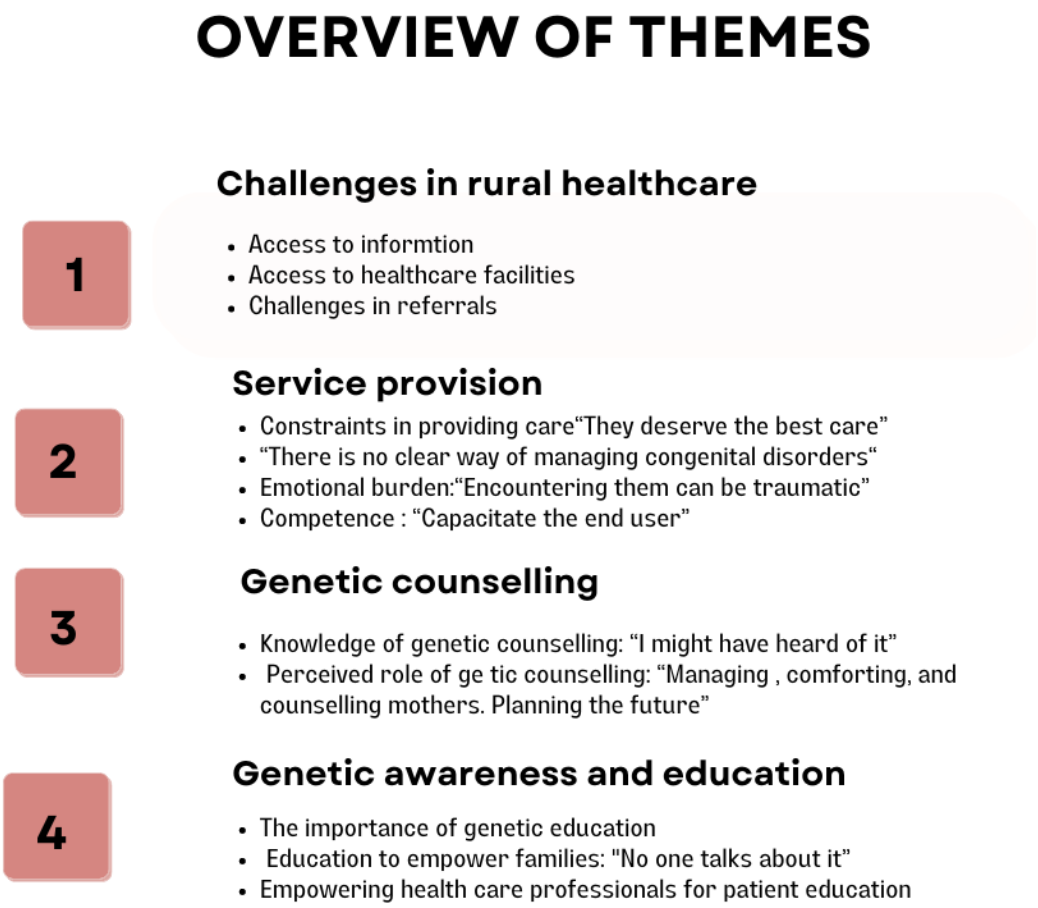

Figure 6 3: Overview of themes

3.1. Challenges in rural healthcare

The study participants highlighted that, individuals residing in rural areas face many healthcare challenges, including limited access to information. They noted that this obstacle was particularly significant for their population, compared to those living in urban areas. They felt that, owing to their geographic location, people had limited access to information about genetics and genetic services. Additionally, the participants identified that people are often unable to access health facilities, and that healthcare practitioners face difficulties when referring patients with congenital disorders to tertiary hospitals noting these as significant challenges.

3.1.1 Access to information

The study participants emphasised the significance of having access to information in healthcare seeking and provision. They noted that non-urban regions may face a disadvantage in acquiring information that they consider vital, such as knowledge about their genetic status and the risk of genetic disorders. In contrast, the participants believed that individuals living in urban areas have access to more information and are therefore better equipped to seek information about their genetic status. The participants pointed out that urban residents have more options available to gain this knowledge and can choose to use the private sector if the required services are not available in the public health sector.

"*Rural areas lack access to important information, like genetics. Urban areas have better access to knowledge and private care for genetic conditions. It's important to improve access to information in rural areas*." -Participant 10

Another challenge that participants noted was the lack of information in their environment on causes of congenital disorders which they feel limits their understanding and ability to make a diagnosis Consequently, healthcare practitioners struggle to provide optimal treatment, leading to potentially unfavourable outcomes for patients who are living with conditions that could be easily treated when the diagnosis is obtained timeously.

"Early diagnosis leads to better treatment, especially in rural areas where a lot is missed or omitted for children. "– Participant 7

3.1.2 Access to healthcare facilities

The travelling distance for patients to receive healthcare services was another concern that was mentioned by the participants as a challenge.

"Unfortunately, transportation is always an issue for patients to get from their homes to these clinics. Additionally, finances are also a challenge because patients have to pay to get to these clinics."- Participant 7

The participants shared that transportation issues further hampered attendance and adherence to treatment plans for patients. To overcome this, they explained that their district hospital had implemented outreach programs led by allied healthcare

professionals to provide primary healthcare service centres with occupational therapy and physiotherapy. Through these outreach programs, they would identify patients in need of their services who were unable to reach the hospital due to the distance they would have to travel and lack of financial means to pay for the transportation. This outreach effort was aimed at reducing the healthcare costs accrued by the patients, but the hospital's budgetary constraints have hindered its sustainability.

"*I feel having outreaches would be more helpful but currently in our institution for instance this year we did not have outreach, so we miss out on a lot of cases, so I feel Primary Health care needs to be emphasized*." – Participant 7

The participants believed that the transportation issues take away the opportunity for identifying more cases at the primary healthcare level. With congenital disorders such as Down syndrome, the participants believed that adherence to such rehabilitation therapies improves the outcomes of patients as the parents also learn of ways to encourage the patients through understanding in interacting with the healthcare professionals.

"*We have two Down syndrome patients in our unit. One of them has improved with compliant treatment and understanding from the grandmother, who knows how to help the child even if someone else brings them in they know what to do, they understand the child's condition and they understand how they need to help the child*." – Participant 7

3.1.3 Challenges in referrals

Participants noted that referral timeframes are determined by case urgency, with urgent cases referred to higher levels of care relatively quicker.

"They say the baby must go to uMthatha maybe after a month or maybe after 6 months"- Participant 4

However, waiting times for specialist appointments at the nearest tertiary hospital were deemed excessively long in some cases, as participant 4 and 2 indicated.

"*Some take up to 3 or 6 months."* -Participant 2

The participants detailed having experienced difficulties while referring patients between their hospital and the designated referral tertiary hospital.

*“If it is not an emergency, they stay here until there are beds, in Nelson Mandela, they usually say that there are no beds, they are full, they say we must keep the baby here with spina bifida its almost two weeks we are still waiting for the bed.”***-**
Participant 2

This was mainly due to resource constraints at Nelson Mandela Academic Hospital, such as a limited number of beds available for patients who require early interventions.

*“If it is not an emergency, they stay here until there are beds, in Nelson Mandela, they usually say that there are no beds, they are full, they say we must keep the baby here with spina bifida its almost two weeks we are still waiting for the bed.”***-**
Participant 2

They believed that this was because the tertiary hospital served a large catchment area for which they do not have enough resources and capacity to cater to the demand of the hospitals referring to them.

“uMthatha is busy. It provides services for the whole of the Transkei. Yes, the central region and eastern region which is this side of Eastern Cape until Queen's Town”. -
Participant 7

These are the challenges that the participants highlighted in relation to healthcare services in rural areas. These challenges informed the service that is available for the management of congenital disorders in their setting.

3.2.Service Provision.

All participants believed that the current service for congenital disorders had certain limitations. One of the main reasons for this was that the hospital is classified as a level 1 hospital, according to the current healthcare tier system. To improve the service, participants suggested the implementation of clear management guidelines for congenital disorders and the up skilling of professionals as well as expanding the current service by providing access to specialists in their environment. They believed that this would help to address the discrepancies in patient experience between urban and rural areas, which was considered the key contributing factor to the quality of care currently being provided to people living with congenital disorders in their area. The participants emphasised the importance of such guidelines in effectively detecting, monitoring, and providing appropriate interventions for patients. They also stressed the need for continuous educational programs tailored to guide healthcare professionals in managing congenital disorders, thus empowering them to be more effective in their respective roles at their level of occupation.

3.2.1 Constraints in providing care : “They deserve the best care”.

The participants felt that the service that they are providing to patients living with congenital disorders is lacking because of the limitations in the skillset available at the hospital and the investigations that are possible. One participant mentioned cases of advanced maternal age as an example. The participant felt that in cases of advanced maternal age, there are limitations to the investigations that they are able to offer for the expectant women who are at an increased risk for aneuploidy. Another example given was that they can only offer a basic ultrasound, and, as a result, they hardly ever pick up foetal anomalies antenatally. This participant believed that if there was equipment and skilled personnel to conduct the necessary investigations, more congenital disorders would be picked up earlier. The participants expressed that even though they may have the knowledge of the investigations that they ought to carry out, this is sometimes restricted by the availability of resources in their setting.

“For instance, no one can do a proper ultrasound because there is no sonographer, we are just doing basic ultrasound based on our skills, it is not easy for us to identify a congenital malformation on the pregnant mom.”- Participant 5

Another participant added that the limitations posed by the lack of skilled professionals in the detection of congenital disorders prenatally meant that the options for the clients are also limited. The participant believed that, if they could detect multiple congenital anomalies reliably in a foetus, then they would be able to present the option of legal termination of pregnancy, should the client want to consider that option and, even if not, the ability to detect the conditions would help in the preparation of the mother for the child’s condition and the implications for the pregnancy.

“*As soon as the mother is pregnant you need to know, you need to do these investigations so that you know what to expect and to give options because if you see early that it is going to be multiple [congenital abnormalities] you can give an option of keeping the baby or not, or, for her to understand during pregnancy what to expect.*”- Participant 3

Furthermore, the participants felt that these limitations are disadvantageous to patients who do not have the financial resources to seek healthcare in the private health sector. Similar sentiments were also shared regarding diagnosing congenital disorders. The diagnostic route for some patients with better resources often involved seeking healthcare in neighbouring, well-resourced provinces. These patients would go to these provinces, obtain a diagnosis, and come to their hospital with a referral letter detailing the diagnosis and management plan.

“Some of the mothers they are working in other provinces, a client [comes to us] from that province and we manage them with what we have available after they have a diagnosis”-Participant 6

Overall, the participants felt that it is frustrating working in a resource constrained environment while striving to provide the best care possible and this subjects them to additional pressure.

"We strive to provide the best care even to those in rural areas without medical aid. If we cannot handle a situation or lack resources, we refer patients to uMthatha. It can be frustrating to work with limited resources, but we do our best with what is available.""-Participant 3

Moreover, the participants felt that the communication regarding the management of congenital disorders between the different levels of care could aid in improving their patient care. They believe this would allow them to understand the investigations that are done outside of their level of care. Currently they do not have enough information from the down referrals concerning the investigations that have been carried out or other referrals that may have been made to obtain a diagnosis. They are only provided a discharge summary which contains the diagnosis and management plan going forward.

"When the patient comes to us, they only have a referral letter and a discharge summary written by the doctors who were attending to the patient that talks about the diagnosis and the treatment other than that, it is not clear what they have done or who saw the patient," -Participant 5

The participants also pointed out that internal communication regarding management of congenital disorders from the recipients of training needed to improve and that information gained may be best shared through written communication.

"*Our ward doctor went for a workshop in uMthatha for congenital disorders and she gave us a guide, but we don't have something that is written*"- Participant 3

The recipients of training were of the view that the training on congenital disorders was efficient. However, they do not have access to a specialist in their vicinity skilled enough to be able to pick up cases with subtle signs and they rely on telephonic discussions with the consultants at the tertiary hospital. They are worried that some information may not be relayed effectively, and that the seriousness of the case being discussed may be missed, leading to patients not receiving the care that they may need.

"I was fortunate enough to attend training on congenital disorders, but outreaches help when you have missed something, the consultant will be able to pick that up, and if you have a case that you are unsure, you think that it could be this or maybe not. When you discuss a patient over the phone it depends on how you discuss you might not convey the message in the way that you want to, but if they are here, they will be able to see that this is serious"-Participant 5

3.2.2 There is no clear way of managing congenital disorders

The participants reported having an effective monitoring system for congenital disorders through the district health office. Their district health officials were able to identify that there was a significant number of congenital disorders reported by the hospital, therefore a training program was initiated to train more healthcare professionals in congenital disorders. The participants, however, feel that this training should be expanded because there are still not enough trained healthcare professionals to meet the demand. This results in a lack of sufficient support for the healthcare professionals who are not trained on congenital disorders who do not know how to approach these disorders.

One participant explained that when the doctors who are trained in congenital disorders are not available to discuss the cases, they are left to figure out how to proceed without clear guidelines. One participant gave as an example that, if the trained doctor is not on call, then they must find an interim solution until the doctor is available despite some congenital disorders requiring immediate attention and direction.

"What you do when you come across a situation like this [congenital disorder], to me it is not very clear what to do" - Participant 6

The participants thought that having access to specialists in their environment would aid in guiding them on how to approach congenital disorders. Having such guidance would subsequently boost their confidence. They believe that this would also afford them the opportunity to learn more, as they would be exposed to someone with more skills and more experience in managing these conditions.

"There is no clear way in terms of how we are managing congenital abnormalities as to, like if you deliver now a baby with spina bifida then the doctor comes in and then this is what is done there is no support, there's no support at all, we are improvising, like doing things you don't know." Participant 4

Another participant expressed that the lack of guidance, support and knowledge can leave them feeling incompetent because they may not do all investigations necessary for a case, due to a lack of knowledge even if the resources are available. The healthcare professionals often felt that they are out of their depth in approaching congenital disorders.

“We would learn a lot if we had our specialists here, not having knowledge and the necessary resources to carry out the required tasks can make you seem incompetent". – Participant 3

3.2.3 Emotional burden: “Encountering them can be traumatic”.

The participants reported that they frequently encounter difficulties while dealing with patients with congenital disorders. Some participants reported feeling anxious when they have patients that are at an increased risk of having children with congenital disorders because they find it challenging managing the emotional reactions in parents whose newborn children are diagnosed with a congenital disorder. The unpredictable nature of these cases and the added responsibility of handling families have resulted in traumatic situations for some of the participants. They attributed their trauma to feeling disempowered by not knowing how to guide the parents effectively as they themselves do not have sufficient information on the cause and management of different congenital disorders. One participant gave an example of severe cleft lip and palate in a baby. In these situations, the mother’s ability to bond with the infant is often disrupted as the cleft interferes with the mother’s ability to breastfeed resulting, sometimes, in adverse reactions driven by fear and frustration felt by the mother. In turn, staff who lack confidence as they are poorly informed on how to proceed, are unable to offer necessary reassurance or a clear management plan to the parents. This is further complicated by the long waiting times for appointments where the parents may be able receive a management plan. The participants felt that this takes an emotional toll on them as it is a traumatising, disempowering experience.

“Even encountering them [congenital disorders] can be traumatic.”- Participant 2

“You delivery a baby with cleft palate, so that baby cannot feed properly, you call the doctor, the doctor will say cleft lip or cleft palate, that’s not an emergency, what do you do?.. In the time that I havec been here, there has been times where mothers would actually want to murder their own babies, it is very traumatizing, then baby must go to uMthatha maybe after a month or maybe after 6 months, but who is supposed to be telling the mother how to take care of this baby, me, and I don’t even know how to” - Participant 4

The participants felt that training on congenital disorders would help in empowering them with the knowledge they need to manage patients with congenital disorders and provide information required by anxious parents.

3.2. 4 Competence : "Capacitate the end user".

The participants felt that their academic training should equip them with the skills they needed to carry out their main professional functions. Some of them shared that, as part of their academic training, the emphasis was on providing personalised treatment for patients. They believed however that their limited knowledge of genetics restricts their ability to tailor treatments for patients who are affected by congenital disorders.

"In school you are taught the theory when you get here you know the theory" – Participant 9

For instance, those who had trained in midwifery mentioned that the focus in their training was on handling the possible situations they may encounter often, such as, a sick neonate and mother. Therefore, they did not have substantial amounts of time dedicated to congenital disorders and genetics.

"Training, training, training, because if you capacitate people that are end users then babies stand a chance and mothers stand a better chance of dealing with such situations, like specific training not a package that comes with a whole lot of other stuff, specifically training to deal with congenital abnormalities."-Participant 4

Additionally, they also felt that the rarity of some congenital disorders makes it difficult for them to be able to confidently rely on the knowledge they acquired while still doing their academic training. They do not trust their ability to recall some of the conditions they covered and also noted that, since the training is based on theory, it becomes difficult when they encounter these conditions in practice. Because the congenital disorder present may not appear exactly as described in theory, this would then require them to think critically in order to tailor effective treatment.

"There is no special task for genetics, as you go through your studies, yes, we were told at school about genetic disorders, but I think we also need like in-service training to be reminded of such things and how to deal with them"-Participant 2

"If you don't see those cases [congenital disorders], you forget because it is not something that you see every day." Participant 1

"*Most of the time it's not really like a cookie cutter. It's not like you can read a textbook and translates it to real life. You need to do a little bit of thinking. So that your treatment matches the patient exactly*."-Participant 9

The participants believed that hands-on training on the management of congenital disorders and genetics was an important component of providing holistic care to patients and providing effective appropriate treatment.

3.3.Genetic counselling

The participants' knowledge of genetic counselling varied with some who had never heard of genetic counselling to others who know of genetic counselling through their academic or professional careers. Some of the participants had work experience from other provinces where they were exposed to genetic counselling while some participants believed that they did not have enough knowledge of what genetic counselling entailed.

Participants generally felt that genetic counseling is a means of offering psychosocial support that would enhance the overall care given to people living with congenital disorders in a setting with limited resources. They perceived this to be important because they felt that a busy environment leaves little time for patient counseling and difficult news delivery. According to the participants, genetic counseling services could support individuals with congenital disorders in making decisions about their reproductive health. Collectively they believed that having access to genetic counseling would afford them the chance to learn more about genetics from a more knowledgeable person in the field.

3.3.1 Knowledge of genetic counselling: "I might have heard of it".

While the participants in the study had varying levels of awareness regarding genetic counselling, a common thread emerged showing a lack of comprehensive understanding of the profession. Some of the participants recalled being exposed to genetic counselling during their academic training but felt that they had a limited idea of the profession, based solely on the job title.

"Oh, we heard about it a little in like university, but it was so brief, not even, like, explaining what genetic counseling is" **-**Participant 8

For instance, one participant recalled hearing about genetic counselling in university as one of the professions that exist in the medical field in a conversation about the possible career options.

"I have heard about it but not in depth"- Participant 9

Some participants, through professional experience with congenital disorders outside of their current place of employment, recognised the value of genetic counsellors within prenatal care teams. A participant recalled having seen pregnant women referred for genetic counselling when a foetal anomaly was detected during antenatal visits while another participant had been exposed to genetic counselling when patients who had a family history of a genetic condition or congenital disorder would be referred for genetic counselling.

"I have heard about that, usually most of the time when they are doing the scan, and they see abnormal things, they see them [genetic counsellors]"- Participant 2

"What I have seen in [another province], clients would then be referred if they have a congenital abnormality in the family for genetic counselling and also looking at the current clinical picture, maybe you have an advanced maternal age or maybe someone has polyhydramnios and then those types of clients would then be referred for genetic counselling" -Participant 4

"I think I might have heard of it, but I don't really know what it is, if I was to explain."- Participant 10

Participants highlighted the need for broader dissemination of information about genetic counselling as their understanding of the profession is superficial and linked to specific situations.

3.3.2 Perceived role of genetic counselling: "Managing, comforting, and counselling mothers. Planning the future"

The participants were of the view that genetic counselling is important for fostering informed decision-making, especially prior to making big life decisions like starting a family. For example, one participant indicated that not knowing one's genetic status can be disadvantageous because, in situations where testing to find out if a person is at risk of children with a particular genetic condition is unavailable, such reproductive choices are left to chance if no genetic counselling or testing is accessible. Since

patients in other parts of the country can get some of these services, the limited availability of this service in other areas results in a concerning inequality. This lack of access calls into question equity and emphasises the necessity to ensure equitable access to healthcare. The expansion of access to services such as genetic counselling and testing could improve the lives of those who may be at risk or affected by a congenital disorder.

"Genetic counselling helps in planning for the future, especially before making life decisions. This is not widely available, which is unfortunate, it is sad, and inequality is very loud, lets expand access to such, someone who is in Cape Town can go for genetic counselling before they even decide to reproduce so you are prepared, you can plan your life, so you have better chances of making it, whereas thina (we) this side we are just blinded, it's sad."-Participant 4

From the responses it was evident that participants' views on the utility of genetic counselling often centred around the role of genetic counsellors providing psychosocial support, especially following a postnatal diagnosis of a congenital disorder which is a common occurrence in their environment.

"*Managing such cases [congenital disorders] involving mothers is particularly crucial. Some mothers may not take the news well, and they require someone to comfort and counsel them, but healthcare professionals are often busy with moving from one patient to another, and some mothers need individual attention and someone to talk to".* -Participant 1

They expressed that genetic counselling would benefit patients by providing education about the disorder and aiding healthcare professionals in bridging the gap between diagnosis and patient education. This is crucial in resource-limited environments with limited staff, as patient education promotes adherence to treatment plans. The participants felt that they are often very busy and lack the ability to dedicate sufficient time required by their patients to educate and offer the support they would like the patients and families to have.

"Rural hospitals are often understaffed, leading to a large number of patients and few workers. Patients do not receive adequate education about their condition and treatment plans, which hinders compliance". Participant 9

The participants expressed the belief that genetic counselling plays a vital role in helping individuals cope with congenital disorders, particularly given the absence of cure for those with underlying genetic causes. By offering patients a chance to discover ways to adjust to life with such conditions, genetic counselling can facilitate the adaptation process.

"A genetic counsellor helps to counsel people with conditions that cannot be changed since they are born with it. The counsellor's job is to show them how to live with it." -Participant 6

They also felt that they needed access to genetic counselling training and workshops to improve their current skillset so that they can have more knowledge when encountering congenital disorders.

"We need genetic counselling training or workshop from an expert to gain more knowledge."-Participant 3

Additionally, genetic counseling was recognised by participants as a valuable resource within the hospital system and could aid in providing an efficient referral process and facilitating communication between genetic testing laboratories and healthcare professionals at varying levels of care.

"I think genetic counsellors would be valuable in hospitals at the district and tertiary level, such as Mthatha, Livingston and Frere Hospital. Genetic counselling services could work with the lab and assist with referrals."- Participant 7

These findings revealed a fragmented understanding of genetic counselling among the study participants. While glimpses of its value emerged, a comprehensive perspective on its diverse roles and functions were not evident.

3.4. Genetic awareness and education

The participants pointed out the importance of promoting awareness of genetics and the role of healthcare professionals in this regard. They believed that through creating more genetic awareness, healthcare professionals can become familiar with genetic conditions, identify them early in those affected and assess the needs of patients from an early age, when interventions are still possible. The consensus from the participants was that early detection is crucial in improving the outcomes of patients

with congenital disorders. Furthermore, participants believed that awareness of health care practitioners could inform awareness of the communities that they serve, affording these communities the opportunity to learn more about the conditions in their families and empowering them with information that is factual and dispels misinformation and myths that may exist regarding congenital disorders.

3.4.1 The importance of genetic education

The participants stressed the importance of awareness regarding genetics based on their insecurities about what they know about genetics. They felt that they did not know enough about how genetics contributes to the conditions they see in their respective areas. One way they thought of addressing this lack of information is to have awareness campaigns aimed at healthcare professionals focusing on how genetics contributes to child development. Moreover, they felt that if health care professionals understood the utility of genetics in their respective areas, it could help foster collaboration between the different disciplines. Participants felt strongly that it is highly beneficial to have a multidisciplinary approach to congenital disorders. They expanded on this need for awareness and pointed out that they believed that all healthcare professionals at all levels of healthcare needed to be aware of genetics.

"I think it's something that everyone at all levels of healthcare should be aware of, being aware of genetics."- Participant 9

This builds on the multidisciplinary approach and the belief that this would lead to expedited referrals and timeous diagnoses. Patients will therefore be seen sooner, and appropriate management can be put in place earlier which would help the families to deal with their situation.

"We need an awareness campaign to stress the importance of genetics in child development. This will remind our medical and clinical staff, as well as our patients, of the significance of a multidisciplinary team, we can expedite referrals and provide holistic healthcare to treat children earlier."- Participant 7

The need for raising awareness on genetics was expanded to include education of people in the community as participants have seen the stigma people experience.

3.4.2 Education to empower families: “No one talks about it.”

The study participants highlighted the critical need for greater awareness among healthcare professionals regarding the balance between genetics and community experiences. They emphasised the importance of engaging with families who arrive seeking information shaped by their local narratives and personal encounters. A particular concern revolved around the spreading of misinformation about the cause of congenital disorders.

They also felt that with the growth in digital access to information, patients were becoming more empowered and had questions regarding their conditions and needed to be met with healthcare professionals who are able to engage with them in a productive manner that addresses their concerns.

Participants also raised concerns regarding the tendency within communities to shy away from open discussions surrounding congenital disorders, even those with a demonstrably high prevalence. They mentioned that a telling example is albinism, a condition present within the community yet rarely acknowledged in open conversation. The participants highlighted that when it is mentioned, it often emerged as a cautionary tale, fuelled by inaccurate beliefs and unfounded fears.

“take the case of albinism for example, you know, it is prevalent, it is there ikhona apha ezilalini (it exists here in rural areas) but no one really talks about it, the only thing that people know is that you don’t laugh at someone with a genetic or congenital abnormality because you might have it, based on myths and stereotypes but you do get mothers that are worried at times because people are accessing information online” - Participant 4

These were motivations for the necessity of empowering healthcare professionals with knowledge and tools to navigate these sensitive topics with families, dispelling misinformation while fostering productive and informative dialogues.

3.4.3 Empowering health care professionals for patient education

The participants put emphasis on the critical need of patient education and its reliance on the knowledge of medical experts. The participants emphasised that medical professionals' capacity to recognise congenital disorders empowers them to raise awareness through sharing relevant information, such as to the possibility of recurrence, with the patient. This conversation was deemed important as it gives parents an awareness of the family's risk related to the condition, which better prepares them to make decisions and navigate the healthcare system. Positive health outcomes can be promoted and maintained with the help of such knowledgeable and capable patients.

"Knowing about genetics can help create awareness among parents if a certain condition is hereditary, it would be nice to be able to tell the parents that your second child might also have the condition or what's going on with that….and for me to advocate for the child." - Participant 9

Adding to the earlier observation, participants conveyed a strong conviction that this paradigm change in education could challenge the deeply rooted belief that people with congenital disorders have limited potential. They maintained that this resulted from a lack of knowledge and comprehension.

"I think it's also more like cultural where people are like OK this child is doomed for the rest of their life [due to a congenital disorder] and they don't like seek intervention for that, but it's slowly changing that people are seeking help for the conditions that they have. But we're still quite behind, and especially with education about the conditions." - Participant 8

The participants envisioned an empowered community actively seeking interventions and adopting an informed, proactive attitude towards their health. But bringing about this change required the participation of health care professionals who have the abilities and expertise required to empower and educate their patients. They believed that genetic counselling could potentially bridge this gap as people in rural areas do not have adequate information and education regarding genetic conditions.

Chapter 4 Discussion and Conclusion

The aim of the current study was to explore the perceptions of healthcare professionals concerning the utility of genetic services for congenital disorders in a rural area in the Eastern Cape. The findings of this study are in line with those of a recent systematic review which was conducted to identify the barriers and the enablers of geographic inequity in accessing clinical genomic and genetic services for non-cancer related rare diseases (Best et al., 2022). As described in the results, the themes that emerged from the data often overlapped, therefore the discussion will be structured according to the three overarching themes (see figure 7) that illustrate the determinants of the approach to congenital disorders as illustrated by the study findings.

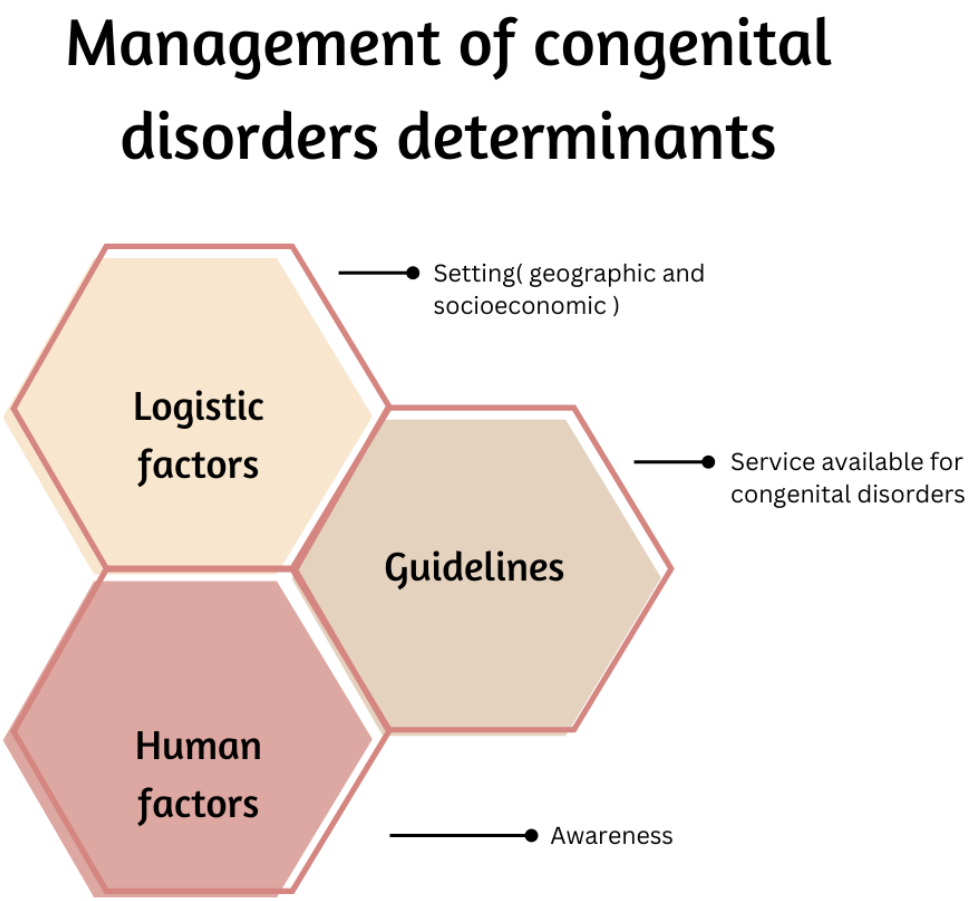

Figure 7 4: Management of congenital disorders determinants.

Logistic factors

The participants of the study brought attention to the problem of accessible healthcare in rural areas, which is a well-documented challenge in the literature. Rural healthcare is often lacking in resources such as healthcare facilities and adequate staff. These resources are commonly concentrated in urban areas, leaving rural areas underserved. These challenges are not unique to South Africa but are general trends that are seen in rural populations in other parts of the world as well (Best et al., 2022).

Transportation and communication are often lacking in rural areas making it hard for people residing in these areas to access healthcare facilities. Poor communication

networks can prevent people from accessing healthcare services (Strasser, 2003). Vergunst et al., (2017) also put forth that people living in rural areas face numerous challenges pertaining to accessing healthcare. Hindrances include poorly developed health infrastructure, high prevalence of chronic illness and disability as well as socioeconomic challenges and physical barriers to accessing healthcare facilities which include the unavailability of transportation to these facilities in the absence of reliable public transport (Vergunst et al., 2017). People in rural areas face an additional challenge of not being able to have access to genetic information as genetic services are often limited to tertiary hospitals located in urban areas of specific provinces. This means that patients living in remote areas may need to travel long distances to access these services while some may not be aware of them (Kromberg et al., 2013). Similarly, the participants in this study were of the view that patients struggled to report to primary healthcare facilities due to geographic barriers and unreliable transportation.

Additionally due to their geographic location, the participants expressed that their patients did not have access to genetic services, thus the approach to congenital disorders as detailed by the participants includes relying on physical examination by healthcare professionals who may or may not have been trained to do so, carrying out limited investigations and then referring the patients to a higher level of healthcare. This approach is similar to one described by Saib et al., 2021 in their study. They described the approach to congenital disorders in Kwa-Zulu Natal when there was no geneticist and patients with complicated congenital disorders were referred to a genetic clinic headed by a paediatrician with a genetics interest who had training through the medical genetics education program (MGEP). This is a post-graduate distance learning education program which was implemented with the Congenital Disorders Course Book, promoting independent, home-based learning (Saib et al., 2021). However, since there is no specialist-led genetic clinic available at Oliver and Adelaide Tambo Hospital, staff rely on the use of telemedicine to manage complex cases, consulting with a paediatrician in a tertiary hospital.

The practice of employing telemedicine in remote areas has been recommended as illustrated in a study by McGrath et al. (2022) which recognised finances and travelling distance as significant barriers to care for rural patients with a congenital

disorder. The authors suggested telemedicine as a viable solution to address the gap in care for people living with a congenital disorder in rural areas (McGrath et al., 2022).Telemedicine is an efficient way of delivering clinical care to patients remotely, especially in resource-constrained areas such as rural locations where access to specialists is limited and reduces overall costs, waiting times, and unnecessary travelling for patients (Gajarawala & Pelkowski, 2021). Geographic positioning of patients and healthcare providers affect access to information regarding genetics and the main challenge is the unequal distribution of specialists between urban and rural areas (Best et al., 2022). Discussions with specialists help the healthcare professionals make appropriate referrals to their designated tertiary hospital as level one hospitals are only equipped to offer generalist medical care (Pauw, 2022). Best et al., (2022) highlighted that healthcare professionals lack informal interaction with specialists in the field since the specialist are in urban areas (Best et al., 2022), similarly, the healthcare professionals interviewed for this study had limited interaction with specialists. They believed that having access to a specialist would have afforded them more learning opportunities and the specialist would be able to pick up cases that they may have otherwise missed. The participants felt that they would benefit from specialist outreach programs. However, accessing specialists outside of tertiary hospitals in South Africa is a difficult task due to the shortage of specialists in the public sector (Coovadia et al., 2009).

A review conducted by Mayosi et al., in 2012 attributed this shortage to the unequal distribution of specialists between the public and private medical sectors. The majority of specialists are located in the private sector in urban areas (Mayosi et al., 2012). The difference in the care that is available in private and public health sectors further complicate the effective management of congenital disorders as health care practitioners miss the opportunity to learn and develop due to a lack of specialists in their environment, more so in rural areas. Given that a large portion of the South African population reside in rural areas, the number of specialists that are situated in these areas, serving this population is not sufficient. Strides have been made to ensure equal access to healthcare through the proposition and effecting of policies (Coovadia et al., 2009). Although some efforts have been made through the initiation of outreach programs, patients still need to be referred to tertiary institutions for investigations.

Communication between the levels of care should facilitate a smooth transition between these levels and was also highlighted in this study.

The participants in this study reported inconsistencies in communication with tertiary institutions regarding information pertaining to the patients. According to a study conducted by O'Malley and Reschovsky (2011) which looked into the communication between primary care physicians and specialists, primary healthcare providers were unlikely to receive results from specialists after having referred patients to them. As a result, primary healthcare providers believed that their capacity to deliver high-quality care was hampered (O'Malley & Reschovsky. These findings are consistent with the feelings expressed by this study's participants about the lack of communication between them and the specialists in referral centers despite their belief that communication is a vital component in ensuring continuity and coordination of care.

Human factors

The participants felt that genetic awareness is an important step in providing personalised care for patients living with congenital disorders in line with the paper by Guttmacher et al. (2007) where the importance of having a good understanding of genetics is underscored. This should include the principles of inheritance, genetic variation and technologies that are available for investigating the underlying causes of congenital disorders (Guttmacher et al., 2007). Healthcare professionals should be able to interpret genetic results and their implications for their families (Malherbe et al., 2017). However, according to Jenkins (1990), genetic awareness in rural South Africa, where predominantly black South Africans reside, is limited. Medical genetics has historically been limited to serving a small portion of the black South African population because these services were based in urban academic centers, often serving the interests of researchers, meaning that limited epidemiological information was available on this portion of the population (Jenkins, 1990).

Historically, the driver of increased genetic awareness in underserved populations was the community genetics initiative (Beighton et al., 2012). This program saw more congenital disorders being recognised and diagnosed and the healthcare professionals at the hospitals that were recipients of the outreach program gained experience through exposure to medical geneticists. This was achieved through the training of nursing sisters in genetics who were able to identify congenital disorders

and improve the ascertainment of their prevalence (Malherbe et al., 2017). This demonstrates that genetic awareness is dependent on exposure to skilled healthcare professionals, as the participants in this study indicated. It further highlights that accurate diagnosis is crucial for the accurate representation of the prevalence of congenital disorders.

Additionally, the participants were of the view that genetic awareness can help combat the stigma surrounding congenital disorders by promoting understanding and education about the genetic factors that contribute to these conditions, such as in the case of albinism which is prevalent but seldom discussed. This stigma is often based on beliefs regarding the origin of the condition as mentioned by the participants of this study.

As stated by Kromberg et al. (2013), South Africa is a melting pot of cultures and beliefs, with people in rural areas attributing congenital disorders to various causes. These beliefs may perpetuate stigma towards those affected in communities. Dispelling misconceptions and stereotypes about these disorders can foster empathy and support for individuals with congenital disorders. Healthcare professionals therefore need to be trained in understanding the disorders and have cultural competence for effective communication and service delivery (Kromberg et al., 2013). Awareness raised through campaigns by healthcare professionals and community engagement is important as alluded to by this study's participants.

Furthermore, genetic awareness can highlight the impact of environmental factors on the development and management of congenital disorders by explaining the complex interactions between genetics and the environment, as some congenital disorders are a result of this interaction (Beighton et al., 2012). Another contributor to the restricted reach of genetic awareness is that there is a shortage of trained genetic professionals as there are few geneticists and genetic counsellors in the country (Beighton et al., 2012).

Questions regarding genetic counselling uncovered that the participants had limited knowledge of genetic counseling. They attributed this to a lack of exposure to genetic services which includes genetic counseling. This is not surprising, as historically, the awareness of genetic counseling among Black South Africans in non-urban areas has been limited.

Furthermore, the participants reported that they had no more than a brief introduction to genetics and genetic counselling in their academic training. Shifting priorities and policies have decreased the awareness of genetics among healthcare professionals. Included in this decline is the knowledge regarding genetic counselling (Kamp et al., 2021). In a research study conducted in South Africa by Phaladi-Digamela, Mulaudzi & Maja, (2014) to ascertain the perception of midwifery educators on the genetic knowledge of advanced midwifery students, the authors found that the educators thought that the lack of standardisation of the genetics education was concerning because it results in the students being inadequately equipped for genetic healthcare. They agreed that genetic education plays a role in the promotion of health, prevention of disease and the development of new roles such as genetic counselling and identification and management of genetic disorders (Phaladi-Digamela et al., 2014). The experience and lack of confidence of the participants in this study support the notion that there is limited training in genetics. Likewise, in another recent study by Thom and Haw (2022), which investigated the knowledge of genetic counselling among allied healthcare practitioners recorded a lack of knowledge in these health care professionals who play an important role in the identification and care of patients with congenital disorders that may have an underlying genetic cause (Thom & Haw, 2021).

The participants had a basic understanding of genetic counselling but were not aware of the full scope of the profession. The role of genetic counsellors is varied and constantly changing, and at times overlaps with other healthcare professionals such as doctors, nurses, and social workers. Due to limited general awareness of the profession, there are often misunderstandings about the role of genetic counsellors (Harris et al., 2023). The opinions of the participants on the utility of genetic counseling when dealing with congenital disorders was that it would be a means of providing psychosocial support to help patients cope with congenital disorders and make informed reproductive decisions. This perception is in line with how Resta et al. (2006) define the genetic counselling process (Resta et al., 2006). However, genetic counselling is not only limited to reproductive counselling and prenatal genetic testing.

Guidelines

This study revealed that the process followed when approaching congenital disorders at Oliver and Adelaide Tambo is broadly in line with the National Department of Health

Clinical Guidelines for Genetic Services updated in 2021. According to these guidelines healthcare practitioners at primary care level should identify congenital abnormalities in neonates and provide immediate care for these children at birth with a follow-up visit if necessary, for review and confirmation of the diagnosis. Upon confirming the diagnosis, the patients are then referred for specialized management (Clinical Guidelines for Genetics Services - 2021, n.d.). This process requires healthcare practitioners who are trained in the appropriate reporting and surveillance of the patients as part of their management plan. As mentioned by Nguyen et al., 2022, healthcare workers play an important role in ensuring that patients receive access to quality healthcare timeously (Nguyen et al., 2022). The participants also recognise their role as being pivotal in ensuring that their patients receive access to quality healthcare such as access to specialists for review in cases where a congenital disorder is suspected despite limited access to training and guidelines.

However, in provinces where there is limited access to specialists, who can aid in the diagnosis of congenital disorders such as paeditricians and medical geneticists, this can pose a challenge, as the participants of this study noted. The data also revealed that the participants lack clarity on the different roles of healthcare professionals outside of the district hospital level, as well as limited understanding of the role of genetic counsellors when it comes to management of congenital disorders.

Patients are referred to a higher level of care when they cannot be assisted at the district level as management frequently requires specialists who can order the correct tests for effective diagnosis and management (Saib et al., 2021). Consequently, the participants have adopted the approach of using telemedicine, although it has limitations. The data also revealed that there is a lack of clarity on the different roles of healthcare professionals outside of the district hospital level, as well as limited understanding of the role of genetic counsellors when it comes to management of congenital disorders. Furthermore, the participants of the study highlighted the need for clear management guidelines for congenital disorders tailored for their hospital. They stated that the availability of these guidelines can serve as an important foundation for the referral processes for people living with congenital disorders and improve the quality of service that they receive in their setting. Effective coordination of care is important in the provision of services for congenital disorders. The development of such guidelines allows the healthcare professionals who come into

first contact with patients the opportunity to address potential complications early on and ensure appropriate follow-up plans are in place. This is especially important in congenital disorders as these conditions typically require lifelong management (McGrath et al., 2022).

Furthermore, such guidelines are useful in ensuring standardisation of care, which includes initial assessments that are to be done as well as standardiation of referral pathways that facilitate the smooth transition between the different levels of care. Healthcare practitioners who are uncertain of the management of congenital disorders can also aggravate the anxiety of the patient's families who may already be emotionally challenged by the congenital disorder, therefore the participants feel that having these guidelines will help in managing the anxieties of their patients as well as provide structure for them in their decision making process.

The participants reported inconsistencies in care owing to the structural and systematic discrepancies that exist between rural and urban areas, these discrepancies were further compounded by the lack of clear guidelines for the management of congenital disorder tailored to their hospital, taking into account their capacity and location. Consequently, the participants noted that the health outcomes of people living with congenital disorders are compromised by this. Studies have shown that the development and adaptation of guidelines facilitates the decision-making process and further fosters shared decision-making between the healthcare provider and the patients. This leads to improved health outcomes and increased health care equity (van der Weijden et al., 2013).

Conclusion

Four main themes have emerged from the data analysis. The first two themes focus on the logistical factors of providing healthcare services in rural areas, while the last two themes relate to the human aspects of the service provided to people living with congenital disorders in these areas.

The first theme highlights the challenges in rural healthcare, including difficulties in accessing information and physical access to healthcare facilities. These challenges make it harder for patients to seek healthcare in rural areas.

The second theme focuses on the provision of healthcare services in rural areas. Healthcare practitioners working in rural areas often work under pressure with minimal support when dealing with congenital disorders. The lack of training on the approach to congenital disorders can affect patient care, as it affects symptom management, physical functioning, and understanding the condition. Empowering healthcare professionals is essential to achieving optimal patient satisfaction.

The third theme is genetic awareness. Healthcare professionals need to recognise genetic and congenital disorders and initiate intervention strategies timeously to tailor therapy and be proactive in their approach. Improved awareness and knowledge would improve the quality of communication between healthcare providers and patients, promote understanding and empathy, combat stigma, and improve treatment plan adherence. By improving awareness and sensitising people to the underlying causes of congenital disorders, communities can better understand and empathise with patients, leading to improved health outcomes and collaboration between healthcare providers and the community.

The fourth and final theme of the study on genetic counselling revealed that there is a lack of awareness about genetic counselling among healthcare professionals, which is consistent with the findings of previous studies. However, despite their limited knowledge about the profession, the participants in this study believed that access to genetic counselling and genetic services would be valued in their setting. They believe it would help improve the level of care and guide the approach to congenital disorders.

Establishing genetic services that cover the South African population requires political will and commitment even though a roadmap to achieve this is evident in National health policy guidelines. This support could improve access to genetic services, particularly for rural South Africans. With the availability of genetic services and genetic counselling in rural areas, patients can receive personalised treatment and health care professionals working in these areas can be better supported.

Strength and limitations

The qualitative nature of this study lead to obtaining beneficial in-depth information regarding the approach to congenital disorders and the perceived role of genetic services in the management of these conditions in a rural setting. The healthcare

professionals were able to express their views in a confidential manner and it was valuable to the participants to consider genetic services and their role in their setting. Participants were able to reflect on the current approach and come up with suggestions on how to improve the current practice. The limitation of the study is that it was conducted in one hospital and did not include all healthcare professionals in the hospital and only one medical practitioner was included. Views of other healthcare professionals in other rural areas may differ and were not explored. Furthermore, the study aimed to explore the perceived role of genetic counseling in the Eastern Cape, which was broader than initially intended. This would require additional research, including more healthcare facilities, to be adequately covered. Furthermore, the congenital disorders that were mentioned by participants included cleft lip/ palate, spina bifida, Down syndrome associated features, congenital heart defects, omphaloceles, talipes, and anencephaly however confirming or addressing incidence was beyond the scope of this study.

Recommendations

From the research study, it is evident that the research participants have a willingness to learn and improve their knowledge regarding genetics and congenital disorders. Initiatives to conduct training in genetic literacy and congenital disorders would be beneficial and serve to empower healthcare professionals in managing congenital disorders. Through training initiatives, the healthcare professionals in rural areas can benefit from improved job satisfaction and confidence in approaching congenital disorders and they can understand the pathway of care better. Additionally, training would improve early detection and implementation of intervention strategies to curb congenital disorders and thus child mortality thereby contributing positively to public health. This could be done through incorporating tailored genetics content in the training curriculum across health care professionals as well as initiating more continuous education programs focused on genetics and congenital disorders. Implementing clear written guidelines for the management of congenital disorders would also assist healthcare professionals in decision-making particularly in the absence of trained healthcare professionals. Genetic counselling remains unattainable for majority of South Africans as the service is confined to training centers and tertiary hospitals. Attaching genetic counsellors to district hospitals could aid in managing congenital disorders as they are trained in genetics and risk assessment, in

the absence of a medical geneticist, genetic counsellors can help in professional and patient education in line with the Department of health's clinical Guidelines for Genetic Services document of 2021. Regarding the trauma reported by participant when dealing with congenital disorders, the hospital could initiate an employee wellness program where healthcare professionals can debrief and seek counselling for improved mental health and to boost morale.

Future research

Expansion of the current study to include more health care professionals and to include other hospitals within the same district and/or similar districts situated in rural areas elsewhere in South Africa. This would provide a more comprehensive picture of how the utility of genetic services in the management of congenital disorders is viewed and shed more light on the referral pathways that exist when managing these conditions at district level. Other research opportunities of value have come to light including community engagement research focused on patients and their families', perceived utility of genetic services in the management of congenital disorders, as well as their beliefs regarding the causes of congenital disorders.

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Appendix A: Interview guide

Demographics

Age

Specialty

Qualification

Experience

Training

Interview Questions

- Could you give a brief history of your time here at Oliver and Adelaide Tambo District Hospital?
- Could you describe what happens in your department?
- What are congenital abnormalities/disorders?
- How do you approach a case of a congenital abnormality/ disorder?
- What do you feel could help you when dealing with such cases?
- How do you feel about your duties?
- What do you do for women who are expecting aged 40?
- What do you do with children with a birth defect?
- What do you understand about Genetic Services?
- What do you know about genetic counseling?
- How do you access genetic services?
- What are your needs when it comes to genetic services?

Appendix B: Consent form

Approach to Congenital Disorders: Referral Pathways for Genetic Services in the Eastern Cape

Information and Purpose: The interview for which you are being asked to participate in, is for a research study in partial fulfillment for the degree M(Med)Sc in Genetic Counselling at the University of Cape Town. The research study is focused on examining the knowledge, attitudes, and practices concerning medical genetic services for investigating congenital disorders, as well as the referral pathway followed to obtain a diagnosis at Oliver and Adelaide Tambo District Hospital. The purpose of this study is to explore your understanding and experiences of medical genetic services, more specifically genetic counseling, medical geneticist, and genetic testing services. Healthcare professionals with at least one year of experience in a unit directly involved in the management of congenital disorders will be recruited for this study.

Your Participation: Your participation in this study will consist of an interview that will be approximately 30 min to an hour long. During the interview, you will be asked a series of questions about your duties focusing on the investigation of congenital disorders in your department/ unit. You may pass on any question that makes you feel uncomfortable. At any time, you may notify the researcher that you would like to stop the interview and your participation in the study. Participation in this study is entirely voluntary. You may decide not to participate, or you may decide to leave the study at any time. Your decision will not be shared with hospital management or have any effect on your employment contract at the hospital.

Benefits and Risks: While there is no direct benefit to you for participating in this study, it has the potential to identify knowledge gaps concerning genetic services. This study aims to explore the options for improving services and could assist in determining how to improve health care for children born with congenital disorders in rural areas. The outcomes may elucidate the existing referral pathway in the Eastern

Cape that are utilized in this area for congenital disorders as well as the perceptions surrounding the utility of genetic services by Health Care Professionals and generate discussion on improving services. There are no risks associated with participating in the study.

Confidentiality: The interview will be voice recorded; however, your name will not be recorded. Your name and identifying information will not be associated with any part of the written report of the research or any publications arising from this study. All your information and interview responses will be kept confidential. The researcher will not share your responses with anyone other than the research supervisor and all attempts will be made to not include any identifying information.

Consent to take part in research:

- I,_____volunt arily agree to participate in this research study.
- I understand that even if I agree to participate now, I can withdraw at any time or refuse to answer any questions without any consequences of any kind.
- I understand that I can withdraw permission to use data from my interview before the publication of the research report or any publications, in which case the material will be deleted.
- I have had the purpose and nature of the study explained to me and I have had the opportunity to ask questions about the study.
- I understand that participation involves, an interview lasting approximately one hour.
- I understand that I will not benefit directly from participating in this research.

- I agree to my interview being audio-recorded.

- I understand that all information I provide for this study will be confidential.

- I understand that in any report on the results of this research, my identity will remain anonymous. This will be done by changing my name and disguising any details of my interview which may reveal my identity or the identity of the people I speak about.

- I understand that disguised extracts from my interview may be quoted in a research report, conference presentation, or published papers.

- I understand that signed consent forms will be retained in a locked office at the University of Cape Town and original audio recordings will be retained in a password-protected device until the research report has been submitted and reviewed by the relevant examination board at the University of Cape Town.

- I understand that a transcript of my interview in which all identifying information has been removed will be retained for two years from the date of the final examination.

- I understand that under freedom of information legalization, I am entitled to access the information I have provided at any time while it is in storage as specified above.

- I understand that I am free to contact any of the people involved in the research to seek further clarification and information.

Signature of research participant

_____

Date_____

I believe the participant is giving informed consent to participate in this study:

Signature of researcher

_____

Date_____

Researcher M(Med)Sc Genetic Counselling degree candidate:

Sesethu Ntanjana:

083 245 7703 / Email: ntnses001@myuct.ac.za **Supervisors:**

Dr Tina-Marié Wessels: 021 406 6373 / Email: tina.wessels@uct.ac.za

Dr Karen Fieggen: 0214066298/ Email: karen.fieggen@uct.ac.za

Human Research Ethics Committee

E 53, Room 46, Old Main Building, Groote Schuur Hospital, Observatory Office

Contacts:021 406 6492; 021 404 7682; 021 406 7260

Appendix C

Information Sheet for participants

Approach to Congenital Disorders: Referral Pathways for Genetic Services in the Eastern Cape

Dear participant,

A research study is being done at the University of Cape Town (UCT) through the Division of Human Genetics for a minor dissertation for MMedSc Genetic Counselling. This research project aims to explore the experiences and perceptions of healthcare professionals concerning the utility of genetic services for congenital disorders in the rural Eastern Cape.

We hope this study will help to raise awareness and gather more information concerning the utility of genetic services for congenital disorders in rural Eastern Cape, as well as investigate the referral pathways for such disorders, as there are no genetic services that are available in the rural Eastern Cape. You have been invited to participate in the study as a healthcare professional at Oliver and Adelaide Tambo District Hospital involved in the investigation and management of congenital disorders.

To gather information for this research project, an individual interview will be conducted which will take between 30-60 minutes. It will be recorded, but all your information will be kept confidential. You will be required to sign the form below that stated that you have consented to take part in the study before we begin the interview.

Participation is voluntary. You may decide not to participate, or you may decide to leave the study at any time. Your decision, whether you participate or not, will not be shared with hospital management or have any effect on your employment contract at the hospital.

If you have any questions, please contact:

Researcher

Ms Sesethu Ntanjana at 0832457703 or NTNSES001@myuct.ac.za

or

Supervisors

Dr Tina-Marié Wessels: 021 406 6373 / Email: tina.wessels@uct.ac.za

Dr Karen Fieggen: 0214066298/ Email: karen.fieggen@uct.ac.za

or

UCT ethics committee at 021 650 1236 or

hrec-enquiries@uct.ac.za

Kind regards,

Sesethu Ntanjana

Appendix D

Human Research Ethics Committee Approval

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

Room 45, E-52 Old Main Building
Groote Schuur Hospital
Observatory 7925
Tel:0214066492
Email: hrec-submissions@uct.ac.za
Website: www.health.uct.ac.za/home/human-research-ethics

14 March 2023

HREC REF: 012/2023

Prof T Wessels
Division of Human Genetics
Email: tina.wessels@uct.ac.za
Student: NTNSES001@myuct.ac.za

Dear Prof Wessels

PROJECT TITLE: APPROACH TO CONGENITAL DISORDERS: REFERRAL PATHWAYS FOR GENETIC SERVICES IN THE EASTERN CAPE
(MMED DEGREE - MISS SESETHU NTANJANA)

Thank you for submitting your study to the Faculty of Health Sciences Human Research Ethics Committee (HREC) for review. Thank you for your response dated 17 February 2023 to the comments raised by the 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 March 2024.

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)

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.

The HREC acknowledges that MMed Candidate Miss Sesethu Ntanjana is also involved in this study.

Please quote the HREC REF 012/2023 in all your correspondence.

Yours sincerely

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

Appendix E

Permission to conduct research at Oliver and Adelaide Tambo District Hospital

Province of the
EASTERN CAPE
HEALTH

81 Murray Street • Kokstad • 4700
Private Bag X3515 • Kokstad • 4700 • REPUBLIC OF SOUTH AFRICA
Tel.: +27 (0)39 797 6000• Fax: +27 (0)39 727 1044 • Website: www.ecdoh.gov.za
Email: yoliswa.ngcobo@echealth.gov.za Cell 060 544 6399
Alt Email : yoli.ngcobo7@gmail.com

To : Sesethu Ntanjana

From : The District Manager
: Alfred Nzo Health District

Date : 30 March 2023

Re : Granting of permission to conduct a research study in Alfred Nzo

Your request to conduct a research study in Alfred Nzo is hereby granted.

The terms and conditions to observe are

1. Adhering to the scope of the research as indicated in your proposal.
2. Observing and adhering to all ethical conduct and practice.
3. Observing communication protocol of the Eastern Cape Department of Health.
4. Observing all patients' rights and dignity practices.
5. The costs of the research will be fully carried by yourself and not on the department.
6. Observing and keeping to the stipulated period of the research and where necessary Indicate any factors that may result in undue delays.
7. Share the research finding with the District and the Eastern Cape Department of Health and contribute to the department's growth and development.
8. The granting of the research is purely for academic and development purposes and does not in any manner constitute or imply an employment contract or conditions of any Payment in any manner.

Throughout the duration of the research, you are requested to keep constant contact with the District Manager.

Yours in service

30/03/2023

Mrs Y.Y Ngcobo
District Manager Alfred Nzo

Appendix F

Reflection

The findings of the research study revealed that there is a need for clear management guidelines for congenital disorders, a need to raise awareness about genetic counseling as a profession among other healthcare professionals and the need for accessible equitable healthcare.

While doing my research, I was struck by the importance of history that shaped it. I also learned how crucial it is to advocate for access to healthcare, especially in impoverished areas. This has been an ongoing process, as evidenced by literature dating back to 1990. One issue I encountered during my research was the lack of monitoring of congenital disorders in rural areas. Often, individuals with disabilities are not properly diagnosed or treated, and their families may resort to superstitious explanations for their condition. As someone from a rural area, this issue was particularly personal to me.

Throughout the data collection phase I was constantly striving to improve my interviewing skills. I was conscious that participants bring their own pre-existing biases, shaped by personal experiences, social norms, and the desire to present themselves favourably. Social desirability bias leads individuals to respond in ways they believe are socially acceptable, potentially distorting their true opinions and they may feel the need to omit or embellish information based on their recollection and perception. I was also conscious of my own biases as a person pursuing a career in genetic counselling, my enquiry had to be as neutral as possible, I was conscious of not leading the participant, having one of my supervisors listen to my recorded interviews and provide feedback was helpful, I was provided tips on how to remain neutral. This was very reassuring in the beginning of my data collection as I was still very conscious and afraid as a novice in this method of research. I would clarify any statements that could be open for interpretation to ensure that I am grasping the essence of what the participants were communicating. The analysis phase was the most challenging, this was especially difficult because of my familiarity with the environment, as I conducted the study in my hometown. I had to ensure that my experiences did not interfere with the data. I did this by reflecting on my interpretations,

sharing my codes with my supervisors and constructing themes and rearranging pre-existing ones, the guidance of my supervisors is invaluable.

Overall, the research process was both challenging and rewarding. I have a deeper appreciation for those who paved the way for us through their work, healthcare professionals who work in rural healthcare, and I believe that studies involving rural populations can provide valuable insights into genetics and genomics.