OPINIONS AND PRACTICES OF MEDICAL SPECIALISTS CONCERNING MEDICAL GENETIC SERVICES AT THE PIETERSBURG AND MANKWENG ACADEMIC TEACHING HOSPITALS IN THE LIMPOPO PROVINCE

by

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DECLARATION

I, *Kathrine Elizabeth Scholtz*, hereby declare that the work on which this dissertation is based is my original work (except where acknowledgements indicate otherwise) and that neither the whole work nor any part of it has been, is being, or is to be submitted for another degree in this or any other university.

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LIST OF ABBREVIATIONS

AMA – Advanced Maternal Age

CD – Congenital Disorder

DMD – Duchenne Muscular Dystrophy

DoH – Department of Health

FMED – Family Medicine

HIV/AIDS – Human Immunodeficiency Virus/Acquired Immunodeficiency Syndrome

HNA – Health Needs Assessment

HoD – Head of Department

HPCSA – Health Professions Council of South Africa

HREC – University of Cape Town Human Research Ethics Committee

IBM SPSS – International Business Machines Corporation Statistical Package for the Social Sciences

INTMED – Internal Medicine

KAP – Knowledge, Attitude and Practices

LDoH – Limpopo Department of Health

LMIC – Low- to middle-income country

MBChB – Bachelor of Medicine and Bachelor of Surgery

n – total number of observations

NCHPEG – National Coalition for Health Professional Education in Genetics

NDoH – National Department of Health

NHLS – National Health Laboratory Service

OBGYN – Obstetrics and Gynaecology

PCH – Paediatrics and Child Health

PMHC – Pietersburg/Mankweng Hospital Complex

PMREC – Pietersburg/Mankweng Research Ethics Committee

SG – Surgery

TB – Tuberculosis

TOP – Termination of Pregnancy

UK – United Kingdom

USA – United States of America

WHO – World Health Organisation

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ABSTRACT

Globally, approximately 7.9 million children are born annually with a genetic or partially genetic condition, with an estimated 3.3 million children under the age of five dying because of a serious congenital disorder (World Health Organization & March of Dimes, 2006). Early diagnosis and intervention through screening strategies, either during antenatal care or at birth, and access to proper treatment and health care can save the life of a child with a serious and life-threatening disability.

Based on the National Department of Health Human Genetics Policy guidelines (2001), the Limpopo province would require 12 full-time medical geneticists (two per 1 million people), 48 genetic counsellors (four per medical geneticist) and 60 medical scientists (five per medical geneticist). To date, however, no medical geneticist or genetic counsellor posts have been created in the province and the provincial National Health Laboratory Service does not have a genetics diagnostic laboratory. Thus, there are currently no medical genetic services available or outreach programmes within either the public or private healthcare systems in Limpopo. Any patients requiring medical genetic services are referred to out-of-province services in Gauteng.

The aim of this study was to canvas the opinions and shed light on the practices of medical specialists at the Pietersburg/Mankweng Hospital Complex in Limpopo with regard to medical genetic services, and whether there is a perceived need for or benefit of locally available medical genetic services.

METHODS

This study employed a mixed methods approach comprising a survey questionnaire (Phase 1), followed by semi-structured follow-up interviews (Phase 2) based on questions related to the aims and objectives of this study. The survey questionnaire was an online questionnaire consisting of thirty-six questions posed to determine the demographics, education, knowledge, referral patterns and opinions about comprehensive medical genetic services of the participants. The sample population consisted of 56 medical

specialists from the departments of Family Medicine; Internal Medicine; Obstetrics and Gynaecology; Paediatrics and Child Health; and Surgery. A total of 32 responses were received. Results for this phase of the study were analysed using descriptive statistics. Semi-structured one-to-one interviews with open-ended questions were used to collect data for Phase 2. A total of 11 medical specialists were interviewed. Participant interviews were recorded and transcribed verbatim. The data were analysed using a thematic data analysis approach.

RESULTS

Results from Phase 1 showed that 44% of participants self-reported their genetics knowledge as fair; 41% felt somewhat comfortable discussing genetic information and risks with patients; and 44% felt that they were somewhat confident in their knowledge of available genetic tests and testing options. All participants were of the opinion that genetic counselling and genetic testing services are a necessity in the province, while 97% of the respondents felt that local access to medical geneticists was a necessity. While most medical specialists (88%) reported having treated patients in the past 12 months who would have benefitted from seeing a medical geneticist, only 13% reported referring these patients to a medical geneticist. Similarly, 81% of medical specialists indicated that they had seen patients or families in the past 12 months who would have benefitted from a genetic counselling service, however, only 28% had referred patients to such a service. With regard to genetic testing services, 94% of medical specialists reported seeing patients in the past 12 months who would have benefitted from genetic testing, and 50% of the respondents had sent samples for genetic testing.

Results from the semi-structured one-to-one interviews highlighted the current lack of capacity, knowledge and resources available locally in Limpopo, and the challenges faced by patients when utilising out-of-province services in terms of social and financial costs, thus creating barriers to accessing comprehensive medical genetic services. Results also showed a perceived need for and benefit of locally available medical genetic services in the province to be able to provide comprehensive care to patients.

CONCLUSIONS

While there is a clear benefit and need for comprehensive medical genetic services in Limpopo, a cost-effective and clearly thought-out strategy needs to be established that does not place strain on an already overburdened and under-resourced provincial public healthcare system.

1. INTRODUCTION

Congenital disorders (CDs) affect approximately 1 in every 15 live births and are the third leading cause of deaths in neonates, globally (World Health Organization, 2011). Approximately 7.9 million children are born annually with a genetic or partially genetic condition, with an estimated 3.3 million children under the age of five dying as a result of a serious CD (World Health Organization & March of Dimes, 2006). Although congenital disorders are a health concern globally and are found in all populations, more than 90% occur in low- to middle-income countries (LMICs), where 95% of deaths due to these disorders occur. This unequal distribution is accounted for by lack of appropriate health services, poverty and a higher prevalence of pregnant women of advanced maternal age and consanguinity in these countries (World Health Organization, 2011).

According to the World Health Organization (WHO) (World Health Organization, 2011, p.iv) community genetics is defined as “the art and science of the responsible and realistic application of health and disease-related genetics and genomics knowledge and technologies in human populations (communities) to the benefit of individual persons”. The prevention of congenital disorders and genetic disease at the community level and the care of affected individuals and families are the primary goals of community genetics. Interventions that are aimed at the prevention, detection and treatment of CDs are collectively referred to as medical genetic services. Such medical genetic services include the diagnosis and treatment of congenital disorders as well as providing counselling and psychosocial support to patients and families (World Health Organization, 2011).

The provision of appropriate medical genetic services can save the life of a child with a serious and life-threatening disability. Thirty percent (30%) of deaths in the first year of life due to a CD can be prevented if appropriate services are available (Malherbe, Christianson & Aldous, 2015).

1.1. Medical Genetic Services in South Africa

Historically, medical genetic services in the country have been restricted to urban areas in provinces where services were established and provided by human genetics departments and medical schools with isolated outreach programmes to various rural areas and underserviced provinces (Kromberg, Sizer & Christianson, 2013). Initially, in the mid-1990s, the National Department of Health (NDoH) seemed to place a focus on the care and prevention of CDs. As a result, in 2001, a WHO sponsored national task team published the Human Genetics Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities (National Department of Health, 2001). This policy provided a framework for clinical and laboratory service delivery for the treatment and prevention of CDs in the country. Priority disorders were also identified, including trisomy 21/Down syndrome, neural tube defects, albinism, cleft-lip and palate, and club feet. In addition, the financial cost of the burden of CDs and personnel requirements were estimated (National Department of Health, 2001). By 2005, the National Guidelines for the Care and Prevention of the Most Common Genetic Disorders, Birth Defects and Disabilities was published. This document described the most common CDs in South Africa and provide strategies for the care and prevention of these CDs. One such strategy is the establishment of and access to comprehensive genetic services across all provinces in the country (National Department of Health, 2005).

As a result of the National Department of Health having to prioritise other health issues, particularly the HIV/AIDS pandemic and the emerging tuberculosis (TB) epidemic, as well as the shortage of posts and qualified personnel and lack of knowledge and awareness of genetic services and the benefit to affected individuals, their families, their communities and the economy (Kromberg, Sizer & Christianson, 2013), medical genetic services were never established or implemented at a national level. Despite the lack of national implementation, four academic departments of human genetics were able to establish clinical and laboratory medical genetic services. In addition, in collaboration with the National Health Laboratory Service (NHLS), two of those four institutions established the

requisite training programs for medical genetic services. Six of the nine provinces in South Africa, however, remain without medical genetic services except for isolated outreach programmes to some areas (Kromberg, Sizer & Christianson, 2013).

1.2. Medical Genetic Services in the Limpopo Province

1.2.1. A brief history

In 1995, the first data on the incidence of congenital disorders in a black Africa rural population was published from a study initiated in the Limpopo Province (at the time the Northern Province) to evaluate the potential burden of CDs with the aim to plan for future prenatal, genetic and paediatric services within the province (Venter et al., 1995). This study reported an incidence of severe congenital disorders of 14.97 per 1 000 livebirths, with an incidence of trisomy 21/Down syndrome (at the time thought to be uncommon in black populations) of 2.10 per 1 000 live births, which was attributed to 44% of the mothers being over the age of 40 years (advanced maternal age), and an incidence of neural tube defects of 3.55 per 1 000 livebirths (Christianson et al., 1995; Venter et al., 1995).

The results of this study and a collaboration between the University of the North (now University of Limpopo) and the University of Pretoria resulted in a clinical outreach programme being established in the area, which ran for a total of 7 years, from 1990 – 1996 (Christianson et al., 1995, 2000). As part of this outreach, medical geneticists visited the province three to four times per year to provide week-long clinics, while nursing sisters from six collaborating rural hospitals were trained in genetics and the diagnosis, counselling and management of common congenital disorders, and co-ordinated genetic clinics at these rural hospitals (Christianson et al., 2000; Koomen et al., 2000). However, a lack of political will, funding and active collaboration of health administrators resulted in the outreach programme no longer being feasible (Christianson et al., 2000).

In 2006, a task team was established by the Limpopo Provincial Department of Health and Social Development implement medical genetic services within the province, in

accordance with the National Guidelines for the Care and Prevention of the Most Common Genetic Disorders, Birth Defects and Disabilities (National Department of Health, 2005). From this task team, in 2007, the Greater Sekhukhune-CAPABILITY Outreach Project was initiated to pilot health care services aimed at the prevention and care of CDs in a district within the Limpopo Province (Gregersen et al., 2013). In the Limpopo Province at the time, 26.8% of medical practitioner posts and 15% of nursing posts were vacant. By 2008 this had increased significantly to 35.4% of positions vacant for medical practitioners and 43.7% for nursing staff. The total percentage of vacant positions in the province at the time, across all health professional posts, was 42.1% (Gregersen et al., 2013). As a result of these serious staff shortages, compounded by the effect of the burden of HIV/AIDS and TB on the healthcare system in the province, CAPABILITY concluded that the development of medical genetic services within the province would not be feasible under those circumstances and the outreach was concluded. It was proposed that a Health Needs Assessment be conducted in order to better understand the needs of the province and the capacity within the province and to plan for future medical genetic services (Gregersen et al., 2013). No further formal outreaches have been conducted in the province since.

1.2.2. The current landscape in Limpopo

The Limpopo Province has the fifth largest population in South Africa with approximately 6 million inhabitants, 10.2% of the national population (Limpopo Province Treasury, 2019). An estimated 80% of the population live in rural areas, with 78.9% living below the poverty line. A total of 92.8% of the provincial population relies on the public health system (Massyn, Pillay & Padarath, 2019), which comprises of 2 tertiary (academic) hospitals, 3 specialised hospitals, 5 regional hospitals (secondary care), and 30 district (primary care)hospitals. These hospitals are supported by 25 community health centres and 452 fixed clinics (Limpopo Province Department of Health, 2016).

Based on the National Department of Health Human Genetics Policy guidelines (2001), the Limpopo Province would require 12 full-time medical geneticists (2 per 1 million

people), 48 genetic counsellors (4 per medical geneticist) and 60 medical scientists (5 per medical geneticist). However, to date there have been no medical geneticist or genetic counsellor posts created in the province and the provincial NHLS does not have a genetic diagnostic laboratory. Thus, there are currently no medical genetic services available or outreach programmes in either the public or private healthcare systems in Limpopo. Any patients requiring medical genetic services are referred to Gauteng (Dr C Sutton, personal communication).

1.3. Rationale for the Study

The rationale for this study stems from a desire to establish a local dedicated medical genetic service to address the needs of the large population reliant on the public healthcare system in Limpopo. According to the WHO (World Health Organization, 2011), burden-of-disease and epidemiologic data as well as a Health Needs Assessment (HNA) analysis are essential first steps for LMICs wanting to implement appropriate care and prevention strategies for congenital disorders and genetic conditions. Christianson et al. (2013) describe an approach to applying a HNA within LMIC by considering the questions “Where are we going?” (the strategic aims), “Where are we now?” (the situational analysis), and “How do we get there?” (the gap analysis). The key component of the gap analysis in a HNA is an understanding of the health needs of the population. This is done through epidemiological investigations as well as the knowledge, attitudes and practices (KAP) of the health professionals in the area and other key stakeholders, such as policymakers and the public (Christianson et al., 2013). This study sought to address one area of the gap analysis for a HNA on the care and prevention of CDs in Limpopo by collecting data on the self-reported knowledge, opinions and practices of medical specialists at the Pietersburg/Mankweng Hospital Complex (PMHC) with regards to medical genetic services.

Several studies, both nationally and internationally, have identified a lack of genetic knowledge and understanding of medical genetic services as barriers to the provision of comprehensive care to patients with genetic conditions (Hofman et al., 1993; Hunter et

al., 1998; Baars, Henneman & ten Kate, 2005; Klitzman et al., 2013; Rinke et al., 2014; Chambers et al., 2015; Düsterwald, 2015; Harding et al., 2019; Walters, Aldous & Malherbe, 2022). However, no previous research has addressed these issues in the Limpopo province in South Africa.

1.3.1. Aim

The aim of this study was to determine the self-reported knowledge, opinions and practices of medical specialists concerning medical genetic services in Limpopo and whether there is a perceived need for or benefit of locally available medical genetics services.

1.3.2. Objectives

- To determine what medical specialists knowledge and opinions are about the different aspects of medical genetic services;
- To determine what medical genetic services, if any, medical specialists in the province use;
- To determine whether there is a perceived need for and benefit of a local dedicated medical genetic service in the province;
- To identify the needs and expectations of medical specialists regarding medical genetic services.

2. METHODOLOGY

2.1. Introduction

The aim of this study was to gather and analyse the knowledge, opinions and practices of medical specialists concerning medical genetic services in the Limpopo province using a mixed methods approach to answer the following main research question: “How do medical specialists at the tertiary hospitals in Limpopo view medical genetic services?”, by answering the following sub research questions:

- What do medical specialists know about the different aspects of medical genetic services in the province and in South Africa?
- What are medical specialists opinions of the different aspects of comprehensive medical genetic services; medical geneticists, genetic counsellors and genetic testing?
- What medical genetic services, if any, do medical specialists in the province use?
- Do medical specialists see a need for and benefit to comprehensive medical genetic services in their discipline?
- What are the needs and expectations of medical specialists regarding medical genetic services?

This chapter describes the participants and the methods used in this study, including the study design; the study population and participant recruitment; data collection and data analysis; and ethical considerations.

2.2. Study Design

This study employed a mixed methods approach which involves the collection, analyses and integration of both quantitative and qualitative research methods in a single study (Byrne & Humble, 2006). Specifically, a sequential explanatory design: participant selection model was employed for the current study, which is a two-phase mixed methods design in which the quantitative characteristics of the participants are used to guide

selection of participants for the qualitative phase (Creswell & Plano Clark, 2007). This study design was chosen to purposefully select medical specialists practicing at the PMHC in the departments of Family Medicine (FMED), Internal Medicine (INTMED), Obstetrics and Gynaecology (OBGYN), Paediatrics and Child Health (PCH) and Surgery (SG), who had seen patients that would benefit from comprehensive medical genetic services in the past 12 months and to provide both a broad and in-depth understanding of how medical specialists in Limpopo view medical genetic services in their context.

2.2.1. Phase 1: Survey questionnaire

The aim of the cross sectional survey questionnaire was two-fold. Firstly, to obtain a general understanding of the knowledge, opinions and practices of medical specialists. Secondly, the baseline survey was used as a sampling tool to select participants with knowledge of and experience with medical genetic services to be included in the subsequent qualitative phase of the study.

2.2.2. Phase 2: Qualitative, semi-structured follow-up interviews

Qualitative semi-structured follow-up interviews were conducted in order to strengthen the findings from the survey questionnaire and to gain a more in-depth understanding of how medical specialists in Limpopo view medical genetic services in their context. Semi-structured qualitative interviews provide a flexible approach that is responsive to participants' perspectives and, therefore, enables the researcher to gain rich, detailed insights into participants' knowledge, opinions, attitudes and practices (Bryman, 2016). The focus of these interviews was to gain insights into medical specialists' genetics knowledge, opinions and referral practices, and whether the medical specialists feel that there is a need for and benefit from comprehensive medical genetic services within their discipline in Limpopo.

2.3. Study Population and Study Sample

2.3.1. Study population

This study was conducted with medical specialists practicing at the PMHC, a combination of two hospitals (the Pietersburg Hospital Campus and the Mankweng Hospital Campus) that provide tertiary health care services in Limpopo. The Pietersburg Hospital Campus is situated in the city of Polokwane, the capital of the Limpopo province. The Mankweng Hospital Campus is situated in Mankweng, a semi-rural area approximately 1km from the University of Limpopo and approximately 30km from Polokwane. Both hospitals are linked to the School of Medicine at the University of Limpopo, which offers an undergraduate degree in Bachelor of Medicine and Bachelor of Surgery (MBChB) and postgraduate specialisation in various clinical disciplines, including: Anaesthesiology, Dermatology, Forensic Pathology, Family Medicine, Internal Medicine, General Surgery, Plastic Surgery, Paediatrics and Child Health, Paediatric Pulmonology, Public Health Medicine and Psychiatry. While there is a Human Genetics Unit within the Department of Pathology in the School of Medicine at the University of Limpopo, there is no Medical Genetics department nor specialist training in medical genetics.

Christianson, Howson & Modell (2006) list family planning, antenatal care, infant and child health (paediatrics) and surgery as services that can be orientated towards the care and prevention of CDs with the support of medical genetic services. For this reason, the study consisted of medical specialists from FMED, INTMED, OBGYN, PCH and SG.

2.3.2. Participant recruitment

Purposive sampling was used to recruit participants for this study. Purposive sampling is a sampling technique used to purposefully select individuals that are knowledgeable about or have experience with a specific phenomenon. Such individuals are ‘information-rich’ and are more likely to provide the best information in order to address the research objectives (Palinkas et al., 2015). For this study, medical specialists from the FMED, INTMED, OBGYN, PCH and SG were specifically selected because these disciplines are

the most relevant in terms of medical genetic services and were most likely to have had experience with patients with a genetic or suspected genetic condition and medical genetic services.

According to Fincham (2008), survey questionnaires can be administered telephonically, in person, mailed, e-mailed, internet mediated or through a combination of these methods. As participant recruitment for this study occurred during the Covid-19 pandemic, the use of emailing and internet mediated recruitment methods were deemed most appropriate. A similar recruitment strategy to that described by Cunningham et al. (2015) was employed in this study. The main recruitment strategy outlined in this article was the involvement of key individuals to facilitate the recruitment process (Cunningham et al., 2015). An email invitation was sent to all heads of departments (HoDs) detailing the purpose of the study and providing information about the survey questionnaire and follow-up in-depth interviews. A brief problem statement was also provided with a link to the online survey questionnaire (Appendix A). HoDs were requested to disseminate the project information and online link to all medical specialists within their department. Two follow-up reminder emails were sent out a week apart. This initial recruitment strategy yielded a very poor response rate and HoDs were contacted telephonically to discuss the project and participant recruitment. According to Thorpe et al. (2009), an effective strategy for increasing response rates when surveying physicians is personalised correspondence. Contact details, including email addresses and telephone numbers of individual medical specialists were obtained from the HoDs from the departments of FMED, INTMED, OBGYN, and SG. Only email addresses were provided by the HoD of PCH. Personalised email invitations were then sent directly to the medical specialists from these departments. While this did elicit a few more responses, the number of responses was still insufficient and so telephonic methods were employed to recruit additional participants. This use of a combination of recruitment methods resulted in a total of 32 out of 56 medical specialists completing the survey questionnaire.

Participants for the qualitative phase of the study were identified from the answers provided in the survey questionnaire. Medical specialists who answered ‘yes’ to the

questions asking whether they had seen any patients in the past 12 months who would benefit from consulting a medical geneticist, genetic counsellor or genetic testing were recruited for the qualitative phase of the study. Two medical specialists, one from SG and one from FMED did not meet these inclusion criteria.

The researcher contacted the medical specialists individually to request their participation in the follow-up interviews. One participant from OBGYN declined to participate and cited a lack of time as their reason. One participant from SG indicated a willingness to participate; however, the interview was rescheduled numerous times because of their busy schedule and did not take place. Other participants identified could not be reached telephonically and so recruitment was concluded. The initial aim was to recruit 10 to 15 medical specialists with a minimum of one participant from each department. This was achieved and a total of 11 participants were interviewed. Table 1 provides a breakdown of the participants from each department that completed the survey questionnaire and were interviewed.

Table 1: Participants recruited per department

Phase of Research Project	**Department of Family Medicine (n)**	**Department of Internal Medicine (n)**	**Department of Obstetrics and Gynaecology (n)**	**Department of Paediatrics and Child Health (n)**	**Department of Surgery (n)**	**TOTAL (n)**
Survey Questionnaire	4	4	12	6	6	**32**
One-on-one Follow-up Interviews	1	1	4	3	2	**11**

2.4. Data Collection

Data collection for the quantitative phase of the study was conducted using a close-ended survey questionnaire consisting of a section on medical specialist demographics and professional training; knowledge about medical genetics and medical genetic services; and opinion of medical genetic services and personal practices with regards to medical genetic services in their context (Appendix A). According to the WHO's Guide to Developing Knowledge, Attitude and Practice Surveys (2008), "a KAP survey is a representative study of a specific population to collect information on what is known, believed and done in relation to a particular topic" – in this case, medical genetic services. As this is a mini dissertation, an extensive KAP survey was not conducted in this study because of time-constraints. As such, the baseline survey only consisted of a limited number of questions aimed at obtaining a brief overview of the knowledge, opinions and practices of medical specialists with regards to medical genetic services in Limpopo, and to select participants for the follow-up semi-structured interviews.

The survey questionnaire and follow-up semi-structured interview guide were developed by the researcher and the research supervisors, a paediatric medical specialist and a qualified genetic counsellor, drawing on similar research conducted with medical students and physicians (Wonkam, Njamnshi & Angwafo, 2006), general practitioners (Düsterwald, 2015) and nursing students (Maradiegue et al., 2005) as a guide. The baseline survey questionnaire included sections on personal information, demographic information and professional education and training (16 questions); knowledge about medical genetics and medical genetic services (5 questions); opinion of medical genetic services (4 questions); and referral practices relating to medical genetic services in their context (11 questions) (Appendix A). The survey questionnaire was used to identify participants for the second qualitative phase of the study, thus the need for a section requiring personal and contact information. It was made explicit during quantitative recruitment that all personal information would be kept confidential and that participants could be contacted to participate in the follow-up interviews.

Data collection for the qualitative phase of the study was conducted by undertaking semi-structured one-on-one interviews (by the student) using an interview guide (Appendix B). A pilot study was conducted with a general practitioner from Limpopo and a medical geneticist registrar from the University of Cape Town in order to ensure the internal validity of the interview guide. The semi-structured one-on-one interviews were conducted in the same way as to be conducted in the main study and the participants were asked to provide feedback on clarity, feasibility, and suitability for the research aim and objectives. In order to ensure consistency and reliability, all interviews were conducted by the researcher. Data collection was conducted during the Covid-19 pandemic and, as such, the majority of the interviews were conducted via the electronic platform Zoom. Two interviews were conducted face-to-face with participants at the Pietersburg Hospital Campus, in the participants private offices. Participants were asked central questions followed by probing questions from the interview guide based on their responses. According to Polit and Beck (2012), probing is a research technique used to elicit a more in-depth explanation from participants in order to obtain more meaningful data. All interviews were audio-recorded and permission to record interviews was obtained prior to commencement of the interview sessions. The duration of each interview session was between 30 minutes to an hour.

2.5. Data Analysis

2.5.1. Phase 1: Quantitative analysis

The raw data generated from the online Google form survey questionnaire was exported into Microsoft Excel for basic descriptive statistical analysis, inferential statistical analysis and content analysis. Basic descriptive statistical analysis was conducted to summarise the data numerically as frequencies and percentages. Data was organised into tables and graphs for comparison and interpretation purposes. For inferential statistical analysis, the data was imported into International Business Machines Corporation Statistical Package for the Social Sciences (IBM SPSS) Version 28.0.1.0. Because of the small sample size,

Fisher's exact test was used to determine whether there was a significant association between the following categorical variances:

- demographic data and referral practices to medical genetic services;
- professional training and education and referral practices to medical genetic services;
- genetics education and referral practices to medical genetic services; and
- genetics knowledge and referral practices to medical genetic services.

Content analysis was conducted for the open-ended questions in order to gain a better understanding of the responses to the multiple choice or dichotomous questions. Some open-ended questions were excluded from the analysis for the sake of brevity. Open-ended questions analysed for this report included:

- a question asking respondents to spontaneously cite three conditions that would benefit from medical genetic services;
- a question asking respondents what would help improve their confidence in genetics knowledge, discussing genetic information and risks with patients and knowledge of available genetic tests and testing options; and
- a question at the end of the survey asking respondents whether there was anything that they would like to add.

2.5.2. Phase 2: Qualitative analysis

For the second qualitative phase of the study, a thematic analysis approach was used to find patterns of meaning within the data set (Braun & Clarke, 2006). Braun and Clarke (2006) provide a step-by-step guide to doing thematic analysis comprising of six phases, namely familiarising one's self with the data; generating initial codes; searching for themes; reviewing themes; defining and naming themes; and producing the report.

The first step involves actively immersing one's self in the data set to familiarise one's self with the content. Audio recordings for each interview were uploaded to the online

transcription program, Grain (Grain Intelligence Inc., 2021), to generate a written form of each transcript. The researcher then checked each transcript against the original recording to ensure accuracy and to become familiar with the data. The transcribed interviews were then read and re-read, and interesting data extracts were highlighted. Furthermore, notes written in the margin.

The transcribed interviews were then uploaded into NVivo Qualitative Data Analysis Software Release 1.7.1 data analysis software in order to manage the data and to create the initial codes. This phase assists researchers to organise the data into meaningful groups and codes. Extracts of data were coded inclusively, meaning surrounding data was included in the codes in order to avoid loss of context (Braun & Clarke, 2006). Because a large number of codes were created, the codes were grouped into categories to make data management easier.

The next analysis phase involved the identification of relationships between codes and categories and the different codes and categories were grouped together under potential themes and subthemes. Because thematic analysis is a recursive process rather than a linear process (Braun & Clarke, 2006), at this point the write-up of the qualitative analysis commenced in an attempt to make sense of the data. During the write-up of the report, the themes and subthemes were reviewed, redefined and renamed until a final report was produced that was an accurate reflection of the stories in the data. The interpretation of the data was validated by the main research supervisor and an independent social sciences researcher in order to avoid bias.

2.6. Ethical Considerations

2.6.1. Ethical clearance

Prior to commencement of the study, ethical approval was obtained from the University of Cape Town Human Research Ethics Committee (HREC REF: 654/2020) (Appendix C). Ethical approval and permission was also sought from the Pietersburg/Mankweng Research Ethics Committee (PMREC 17 MARCH UL 2021/C) (Appendix D) because

participants in this study were from the public hospitals that fall within the authority of the Limpopo Provincial Government. Permission to conduct research in departmental facilities was also obtain from the Limpopo Department of Health (LDoH) (Appendix E).

2.6.2. Informed consent and voluntary participation

Informed consent is grounded in the value of self-determination of an individual and respect for their dignity (Glickman et al., 2009). According to the WHO and the Council for International Organisations of Medical Science, informed consent is consent provided by an individual who fully understands the purpose and nature of the research project, what is required of them during the study and any risks or benefits that may result from participation in the study (World Health Organization & Council for International Organizations of Medical Sciences, 2017). For Phase 1 of the study, the first section of the online Google form contained an information sheet detailing the purpose of the study and how data would be collected. The second section dealt with informed consent, where their rights as a participant were explained, that participation would be voluntary, that participants could refuse to answer any questions without consequence and that participants could withdraw permission at any time prior to publication of the research findings. It was also explained in this section that completion and submitting of the online Google form provided consent to participation in this phase of the study and permission for the researcher to contact them to participate in Phase 2 of the study. Because all interviews, except for two, were conducted online via Zoom during Phase 2 of the study, participants were sent an electronic copy of the information sheet and consent form (Appendix F) prior to the interview. The consent forms were signed and emailed to the researcher. At the start of each interview the researcher went through the information sheet and consent form again with each participant and afforded them an opportunity to ask questions for clarity prior to commencement of the interviews. All participants in this study were practising medical specialists and were, therefore, capable of providing consent.

2.6.3. Privacy and confidentiality

The privacy and confidentiality of participants was ensured by omitting all participant identifiers from the data. All documents and voice recordings containing identifying information were securely stored on an access controlled computer. Each participant was given a unique number that was used in the presentation of the results. Because of the small sample population within each of the five departments, extra care was taken to ensure that the participants are not identifiable from any published interview information. **PX** was the participant number used for any interview data reported that may result in the identification of an individual.

2.6.4. Potential risks and benefits to participants

There were no risks or discomfort to participants associated with this study as the questions and content were not of a sensitive nature and participation was voluntary. Participants were given the option not to answer any of the questions in the survey questionnaire or the one-on-one semi-structured interviews, and could withdraw at any point.

There was no direct benefit of the study to the individual participants; however, the proposed research has highlighted a need for and the benefit of medical genetic services in Limpopo as well as a potential strategy for the implementation of these services in the province in a cost-effective and feasible way.

3. RESULTS

The results from the online survey questionnaire are presented here, followed by the results from the one-on-one interviews.

3.1. Phase 1: Survey Questionnaire

3.1.1. Sample demographics

Thirty-two (32) responses were received from the 56 eligible participants from the five departments targeted, namely FMED, INTMED, OBGYN, PCH and SG at the PMHC. This gives an overall response rate of 57%. Individual response rates per department are reported in Table 2 below.

Table 2: Response Rates

Variable	**Department of Family Medicine (n)**	**Department of Internal Medicine (n)**	**Department of Obstetrics and Gynaecology (n)**	**Department of Paediatrics and Child Health (n)**	**Department of Surgery (n)**	**TOTAL (n)**
Respondents	4	4	12	6	6	32
Total Medical Specialists	5	9	19	16	7	54
Response Rate	**80%**	**44%**	**63%**	**38%**	**85%**	**57%**

The sample demographics are summarised in Table 3 as a complete data set.

3.1.1.1. Gender

Overall, 72% (n=23/32) of the respondents were male and 28% (n=9/32) were female. According to Tiwari et al. (2021), of the 15 304 medical specialists registered with the Health Professions Council of South Africa (HPCSA) in 2019, 68% were male and 32% were female (Tiwari et al., 2021).

3.1.1.2. Age

The majority of participants (41%; n=13/32) were in the 41-50 years of age category, with the smallest number of participants being from the over 60 years of age category (13%; n=4/32). The 30-40 and the 51-60 age categories accounted for 22% (n=7/32) and 25% (n=8/32), respectively.

3.1.1.3. Education and years in practice

Overall, the majority of participants (56%; n=18/32) had completed their undergraduate training in the last 10–20 years and their specialist training less than 10 years ago. The majority of participants (77%; n=23/30) completed their undergraduate training in South Africa, with 23% (n=7/30) having completed their undergraduate training overseas. Two participants did not provide an answer to this question. The majority of participants (88%; n=28/32) completed their specialist training in South Africa, with 13% (n=4/32) having completed their specialist training overseas.

3.1.1.4. Specific fields of interest

The vast majority of respondents (91%; n=29/32) reported having a specific field of interest within their discipline. Professional interests described were varied but included areas relevant to the field of genetics, such as breast and colorectal surgery, neurogenetics, infertility and reproductive health, neurology, haematology, gynaecological oncology, foetal maternal medicine, haemophilia, cardiology and neonatology. Specific fields of interest not relevant to genetics included trauma, general surgery and urogynaecology.

Table 3: Demographic Characteristics of Respondents

Variable	TOTAL (n;%)	Department of Family Medicine	Department of Internal Medicine	Department of Obstetrics and Gynaecology	Department of Paediatrics and Child Health	Department of Surgery
Gender						
Male	23 (72%)	2 (50%)	3 (75%)	9 (75%)	4 (67%)	5 (83%)
Female	9 (28%)	2 (50%)	1 (25%)	3 (25%)	2 (33%)	1 (17%)
Total	**32 (100%)**	**4 (100%)**	**4 (100%)**	**12 (100%)**	**6 (100%)**	**6 (100%)**
Age						
<30	0 (0%)	0 (0%)	0 (0%)	0 (0%)	0 (0%)	0 (0.%)
30-40	7 (22%)	1 (25%)	1 (25%)	1 (8%)	2 (33%)	2 (33%)
41-50	13 (41%)	2 (50%)	1 (25%)	7 (58%)	2 (33%)	1 (17%)
51-60	8 (25%)	1 (25%)	1 (25%)	3 (25%)	1 (17%)	2 (33%)
>60	4 (13%)	0 (0%)	1 (25%)	1 (8%)	1 (17%)	1 (17%)
Total	**32 (100%)**[1]	**4 (100%)**	**4 (100%)**	**12 (100%)**	**6 (100%)**	**6 (100%)**
Years since undergraduate training completed						
<10	1 (3%)	0 (0%)	0 (0%)	0 (0%)	1 (17%)	0 (0%)
10-20	18 (56%)	3 (75%)	2 (50%)	7 (58%)	3 (50%)	3 (50%)
21-30	5 (16%)	1 (25%)	0 (0%)	2 (17%)	1 (17%)	1 (17%)

[1] Percentages may not sum to 100 due to rounding

31-40	7 (22%)	0 (0%)	1 (25%)	3 (25%)	1 (17%)	2 (33%)
>41	1 (3%)	0 (0%)	1 (25%)	0 (0%)	0 (0%)	0 (0%)
Total	**32 (100%)**	**4 (100%)**	**4 (100%)**	**12 (100%)**	**6 (100%)**	**6 (100%)**
Years since specialist training completed						
<10	18 (56%)	3 (75%)	1 (25%)	9 (75%)	2 (33%)	3 (50%)
10-20	7 (22%)	1 (25%)	1 (25%)	1 (8%)	3 (50%)	1 (17%)
21-30	6 (19%)	0 (0%)	1 (25%)	2 (17%)	1 (17%)	2 (33%)
31-40	1 (3%)	0 (0%)	1 (25%)	0 (0%)	0 (0%)	0 (0%)
>41	0 (0%)	0 (0%)	0 (0%)	0 (0%)	0 (0%)	0 (0%)
Total	**32 (100%)**	**4 (100%)**	**4 (100%)**	**12 (100%)**	**6 (100%)**	**6 (100%)**
Years working as a specialist						
<10	20 (63%)	3 (75%)	2 (50%)	9 (75%)	3 (50%)	3 (50%)
10-20	5 (16%)	1 (25%)	0 (0%)	1 (8%)	2 (33%)	1 (17%)
21-30	6 (19%)	0 (0%)	1 (25%)	2 (17%)	1 (17%)	2 (33%)
31-40	1 (3%)	0 (0%)	1 (25%)	0 (0%)	0 (0%)	0 (0%)
>41	0 (0%)	0 (0%)	0 (0%)	0 (0%)	0 (0%)	0 (0%)
Total	**32 (100%)**	**4 (100%)**	**4 (100%)**	**12 (100%)**	**6 (100%)**	**6 (100%)**

3.1.2. Genetics education and knowledge

In order to gain a better understanding of current genetics knowledge and knowledge of genetic conditions, medical specialists were asked about genetic education and training received during their undergraduate training and their specialist training, as well as

continuing education. Responses to the questions about genetics education are summarised in Table 4 as a complete data set.

3.1.2.1. Genetics education and continuing education

Overall, 75% (n=24/32) of respondents reported having less than 10 hours of genetic education during their undergraduate training. Two respondents (6%; n=2/32) reported not receiving any genetics education during their undergraduate training.

To the question: “How much genetics education did you receive during your specialist training?”, 41% (n=13/32) reported to not receiving any genetics education during their specialist training. The majority of these respondents were from FMED, INTMED and SG, with all participants from INTMED reporting no genetics education during specialist training. Interestingly, all respondents from FMED completed their specialist training at the same institution. One respondent who completed their specialist training 10–20 years ago reported receiving >10 lectures on genetics. The remaining respondents completed their training <10 years ago and all reported receiving no genetics education during their specialist training. Of the six respondents (19%; n=6/32) who reported receiving >10 hours of genetics education, five were from OBGYN, three of whom underwent specialist training at the University of Cape Town.

In response to the question about continuing medical education, 78% (n=25/32) of the respondents reported dedicating more than three hours per week to continuing education; however, only 28% (n=9/32) reported continuing medical education that included genetics. Such continuing medical education was reported to be in the form of self-directed learning involving journals, books and webinars. One respondent indicated participation in a Department of Health (DoH) genetics training course. Other forms of continuing medical genetics education included informal discussions, academic meetings and presentations.

Table 4: Genetics Education and Continuing Medical Education

Variable	TOTAL (n;%)	Department of Family Medicine	Department of Internal Medicine	Department of Obstetrics and Gynaecology	Department of Paediatrics and Child Health	Department of Surgery
Genetics lectures received during undergraduate training						
None	2 (6%)	1 (25%)	0 (0%)	0 (0%)	0 (0%)	1 (17%)
1-3	11 (34%)	1 (25%)	2 (50%)	4 (33%)	1 (17%)	3 (50%)
4-6	8 (25%)	2 (50%)	0 (0%)	3 (25%)	2 (33%)	1 (17%)
7-10	3 (9%)	0 (0%)	0 (0%)	2 (17%)	0 (0%)	1 (17%)
>10	4 (13%)	0 (0%)	1 (25%)	2 (17%)	1 (17%)	0 (0%)
Don't know	4 (13%)	0 (0%)	1 (25%)	1 (8%)	2 (33%)	0 (0%)
Total	**32 (100%)[2]**	**4 (100%)**	**4 (100%)**	**12 (100%)**	**6 (100%)**	**6 (100%)**
Genetics education received during specialist training						
None	13 (41%)	3 (75%)	4 (100%)	2 (17%)	0 (0%)	4 (67%)
1-3	7 (22%)	0 (0%)	0 (0%)	3 (25%)	3 (50%)	1 (17%)
4-6	3 (9%)	0 (0%)	0 (0%)	1 (8%)	2 (33%)	0 (0%)
7-10	2 (6%)	0 (0%)	0 (0%)	1 (8%)	0 (0%)	1 (17%)
>10	6 (19%)	1 (25%)	0 (0%)	5 (42%)	0 (0%)	0 (0%)
Don't know	1 (3%)	0 (0%)	0 (0%)	0 (0%)	1 (17%)	0 (0%)

[2] Percentages may not sum to 100 due to rounding

Total	**32 (100%)**	**4 (100%)**	**4 (100%)**	**12 (100%)**	**6 (100%)**	**6 (100%)**
Hours per week dedicated to continuing medical education						
0	0 (0%)	0 (0%)	0 (0%)	0 (0%)	0 (0%)	0 (0%)
1	0 (0%)	0 (0%)	0 (0%)	0 (0%)	0 (0%)	0 (0%)
2	4 (13%)	0 (0%)	1 (25%)	1 (8%)	2 (33%)	0 (0%)
3	3 (9%)	0 (0%)	0 (0%)	2 (17%)	1 (17%)	0 (0%)
>3	25 (78%)	4 (100%)	3 (75%)	9 (75%)	3 (50%)	6 (100%)
Total	**32 (100%)**	**4 (100%)**	**4 (100%)**	**12 (100%)**	**6 (100%)**	**6 (100%)**
Continuing education included genetics education						
Yes	9 (28%)	0 (0%)	2 (50%)	4 (33%)	1 (17%)	2 (33%)
No	23 (72%)	4 (100%)	2 (50)	8 (67%)	5 (83%)	4 (67%)
Total	**32 (100%)**	**4 (100%)**	**4 (100%)**	**12 (100%)**	**6 (100%)**	**6 (100%)**

3.1.2.2. Genetics knowledge and confidence in treating patients with genetic conditions

For this section, respondents were asked to rate their knowledge of genetics, comfort in discussing genetic information and risks with patients as well as confidence in their knowledge of available genetic testing options. The medical specialists were also asked to spontaneously cite three genetic conditions that would benefit from medical genetic services. This section was included in order to better understand how confident medical specialists are in identifying patients with possible genetic conditions.

The medical specialists were first asked to rate their knowledge of genetics as excellent, very good, good, fair or poor. The majority of respondents (44%; n=14/32) self-reported their genetics knowledge to be fair, with an equal number of respondents (25%; n=8/32)

indicating that they thought their genetics knowledge was either poor or good. Only two respondents (6%; n=2/32) felt that their knowledge of genetics was very good, while no respondents self-reported their genetics knowledge as excellent (Figure 1A).

The medical specialists were then asked to indicate how confident they were with discussing genetic information and risks with patients. Most of the respondents (41%; n=13/32) indicated that they felt somewhat comfortable, while only three respondents (9%; n=3/32) reported being very comfortable discussing genetic information and risks with patients. An equal number of medical specialists (25%; n=8/32) indicated that they were neither comfortable nor uncomfortable discussing genetic information and risks with patients (Figure 1B).

The next section asked respondents to rate their confidence in their knowledge of available genetic tests and testing options. The majority of the medical specialists (44%; n=14/32) self-reported being somewhat confident in their knowledge of available genetic tests and testing options, while almost a third of the respondents (31%; n=10/32) felt that they were not confident. Only two respondents (6%; n=2/32) felt very confident in their knowledge of available genetic tests and testing options, while the remaining 19% (n=6/32) indicated that they felt confident (Figure 1C).

Lastly, respondents were asked to spontaneously cite three conditions that would benefit from medical genetic services. The majority of medical specialists (84%; n=25/32) were able to spontaneously cite three conditions, with two respondents (6%; n=2/32) citing four different conditions. Two of the respondents (6%; n=2/32) were only able to spontaneously cite two conditions, while three respondents (9%; n=3/32) were only able to cite one condition (Figure 1D).

A total of 46 different conditions were mentioned in the 90 total responses recorded. Trisomy 21/Down syndrome was the most commonly cited condition, with 16 respondents (50%; n=16/32) citing this as one of the conditions that would benefit from medical genetic services. Breast cancer and trisomy 18/Edwards syndrome were the second most commonly cited conditions with 6 respondents (19%; n=6/32) citing this as a condition

that would benefit from medical genetic services. The complete list of conditions cited and the response rate can be found in Appendix G.

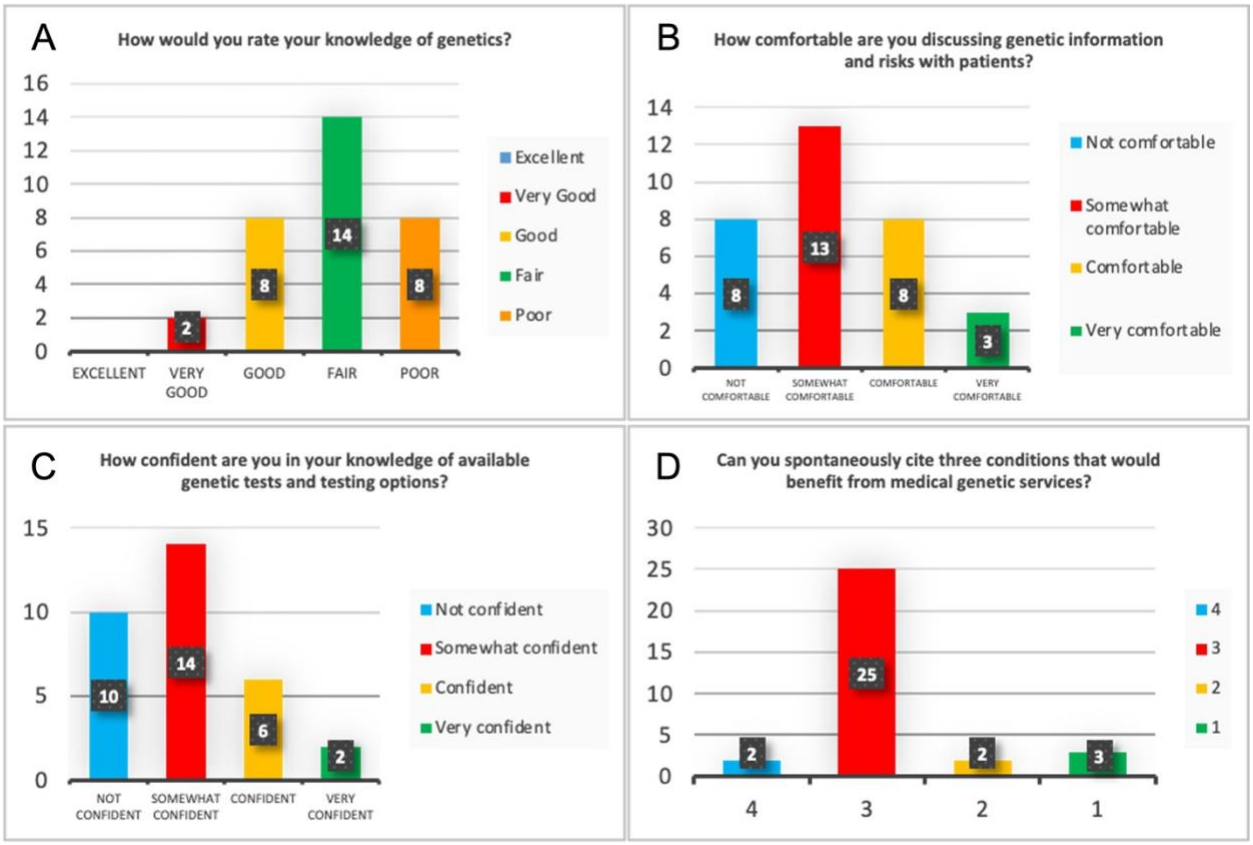

Figure 1: Medical specialists' self-reported genetics knowledge and confidence in treating patients with genetic conditions. A) Self-reported genetics knowledge. *Twenty-five percent (25%; n=8/32) of respondents reported their genetics knowledge as poor, 44% (n=14/32) as fair; 25% (n=8/32) as good; and 6% (n=2/32) as very good.* ***B) Comfort in discussing genetic information and risk with patients.*** *Eight respondents (25%; n=8/32) were not comfortable discussing genetic information and risks with patients; 13 respondents (41%; n=13/32) were somewhat comfortable; 8 respondents (25%; n=8/32) were comfortable; and 3 respondents (9%; n=3/32) were very comfortable discussing genetic information and risks with patients.* ***C) Confidence in knowledge of available genetic testing options.*** *Ten respondents (31%; n=10/32) were not confident in their knowledge of genetic tests and available testing options; 14 respondents (44%; n=14/32) were somewhat confident; 6 respondents (19%; n=6/32) were confident; and 2 respondents (6%; n=2/32) were very confident in their knowledge of genetic tests and available testing options.* ***D) Three conditions that would benefit from medical genetic services.*** *Three respondents (9%; n=3/32) were able to spontaneously cite one genetic condition that would benefit from medical genetic services; 2 respondents (6%; n=2/32) were able to cite two genetic conditions; 25 respondents (78%; 25/32) were able to cite three genetic conditions; and 2 respondents (6%; n=2/32) cited four conditions that would benefit from medical genetic services.*

The responses from the respondents in the five different departments were then analysed separately in order to get a better understanding of how confident the respondents from the different specialities are in identifying patients with possible genetic conditions, as well as treating and managing these patients.

In FMED, all respondents (100%; n=4/4) self-rated their genetics knowledge as poor (Figure 2A). This is consistent with their responses to never having included genetics education in their continuing medical education. In addition, all except one respondent recalled receiving no genetics education during specialist training. Interestingly, the respondent who had received genetics education during specialist training recalled >10 lectures, yet self-rated their genetics knowledge as poor.

Responses varied considerably when asked how comfortable they are discussing genetic information and risks with patients, with one respondent (25%; n=1/4) each reporting being not comfortable, somewhat comfortable, comfortable and very comfortable (Figure 2B). Responses were more consistent when reporting on confidence in their knowledge of available genetic testing and testing options, with the majority of respondents (75%; n=3/4) indicating that they were not confident. Only one respondent (25%; n=1/4) felt confident in their knowledge of available genetic testing options (Figure 2C). This was the same respondent who reported being very comfortable discussing genetics information and risks with patients; however, this respondent also self-reported their genetics knowledge as poor.

Despite this self-reported lack of comfort or confidence, three quarters (75%; n=3/4) of the medical specialists from FMED were able to spontaneously cite three conditions that would benefit from medical genetic services. One respondent (25%; n=1/4) could only spontaneously recall two conditions (Figure 2D). Interestingly, this was the same respondent who reported being very comfortable discussing genetic information and risks with patients and self-reported that they were very confident in their knowledge of available genetic tests and testing options. The most commonly cited genetic conditions

by medical specialists from FMED were trisomy 21/Down syndrome (100%; n=4/4) and trisomy 18/Edwards syndrome (50%; n=2/4).

Medical specialists from FMED felt that both practical and theoretical training, workshops, lectures and contact with a genetics team would improve their comfort and confidence when dealing with patients with genetic conditions.

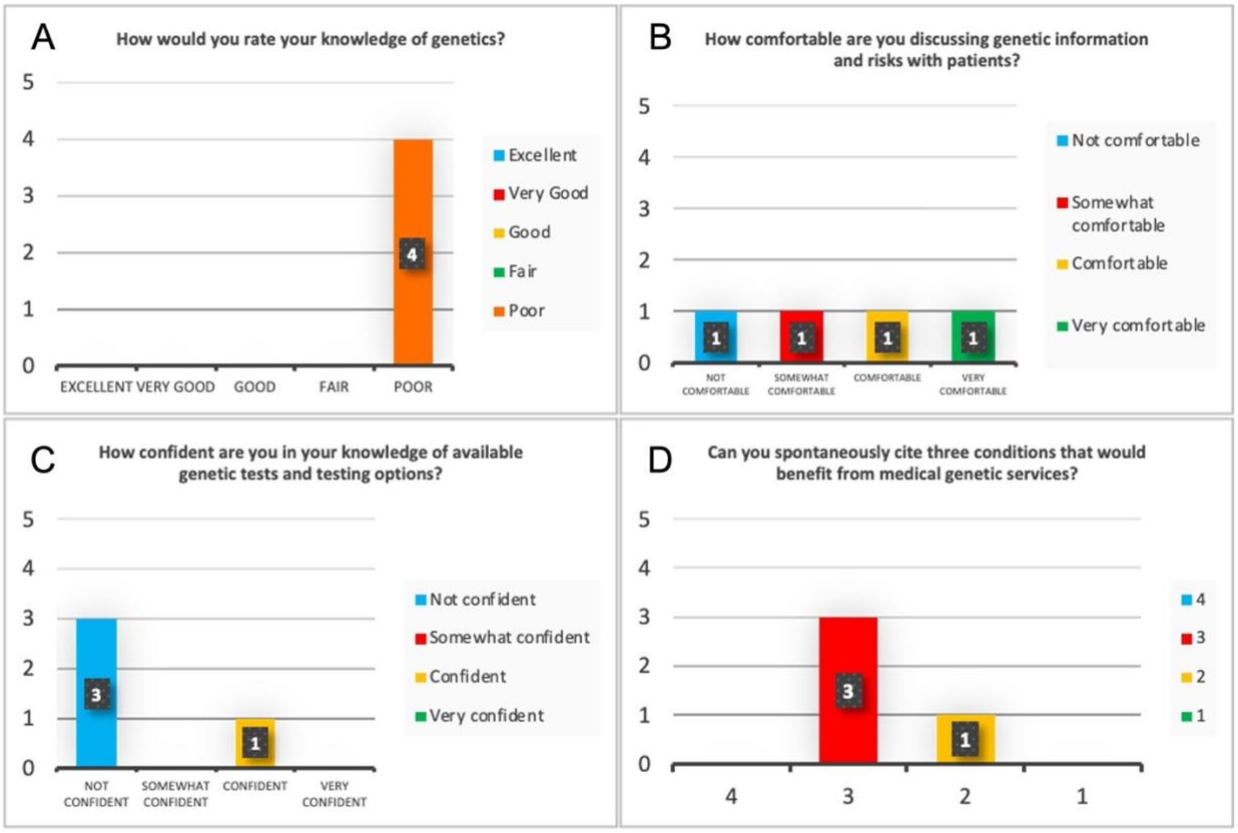

Figure 2: Medical specialists in the Department of Family Medicine's self-reported genetics knowledge and confidence in treating patients with genetic conditions. A) Self-reported genetics knowledge. *All four respondents (100%; n=4/4) from the Department of Family Medicine self-reported their genetics knowledge as poor.* ***B) Comfort in discussing genetic information and risk with patients.*** *One respondent (25%; n=1/4) was not comfortable discussing genetic information and risks with patients; while 1 respondent (25%; n=1/4) was somewhat comfortable; 1 respondent (25%; n=1/4) was comfortable; and 1 respondent (25%; n=1/4) was very comfortable..* ***C) Confidence in knowledge of available genetic testing options.*** *Three respondents (75%; n=3/4) were not confident in their knowledge of genetic tests and available testing options; while 1 respondent (25%; n=1/4) was confident.* ***D) Three conditions that would benefit from medical genetic services.*** *Three respondents (75%; n=3/4) were able to spontaneously cite three genetic condition that would benefit from medical genetic services; while 1 respondent (25%; n=1/4) was able to cite two genetic conditions.*

In INTMED, half of the respondents (50%; n=2/4) self-rated their genetics knowledge as fair, with the remaining respondents reporting their knowledge as either good (25%; n=1/4) or poor (25%; n=1/4). No respondents felt that their genetics knowledge was very good or excellent (Figure 3A). This is consistent with the fact that none of the medical specialists from this department recalled receiving any genetics education during their specialist training. The respondent who self-rated their genetics knowledge as good had completed a DoH genetics training course. Interestingly, the respondent who self-rated their genetics knowledge as poor, reported having done self-directed genetics education by reading journal articles.

When asked about their comfort when discussing genetic information and risks with patients, the majority of respondents (50%; n=2/4) reported that they were not comfortable, while an equal number (25%; n=1/4) reported being either somewhat comfortable or comfortable (Figure 3B). Interestingly, when asked about their confidence in their knowledge of available genetic tests and testing options, the majority of respondents (50%; n=2/4) reported feeling confident, while an equal number (25%; n=1/4) reported to being either not confident or somewhat confident in their knowledge (Figure 3C). The respondent that self-rated their genetics knowledge as poor, also indicated that they were not comfortable discussing genetic information and risks with patients and was not confident in their knowledge of available genetic tests and testing options. In addition, the respondent who self-rated their genetics knowledge as good, reported being comfortable discussing genetic information and risks with patients and being confident in their knowledge of available genetic tests and testing options. This was the same respondent who reported completing a DoH genetics training course.

Despite their self-reported lack of knowledge, comfort and confidence, all respondents from INTMED were able to spontaneously cite three conditions that would benefit from medical genetic services (Figure 3D). The most commonly cited genetic conditions by medical specialists from this department were neurodegenerative disorders such as Huntington’s disease, Spinocerebellar Ataxia and Duchenne Muscular Dystrophy (DMD).

Medical specialists from this department felt that training, continuing medical education and support and guidance from geneticists would help improve their confidence in treating and managing patients with genetic conditions.

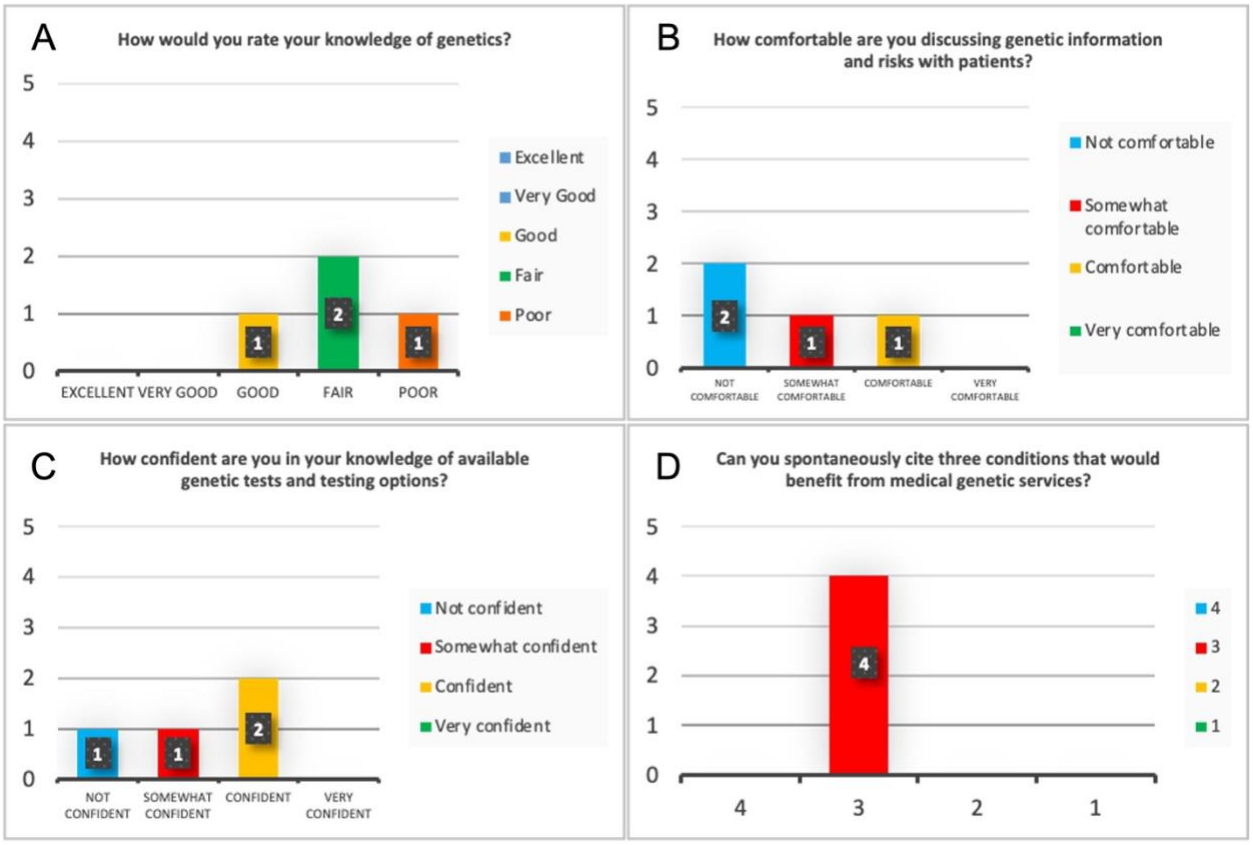

Figure 3: Medical specialists in the Department of Internal Medicine's self-reported genetics knowledge and confidence in treating patients with genetic conditions. A)Self-reported genetics knowledge. *Twenty-five percent (25%; n=1/4) of medical specialists self-reported their genetics knowledge as poor, 50% (n=2/4) as fair, and 25% (n=1/4) as good.* ***B) Comfort in discussing genetic information and risk with patients.*** *Two respondents (50%; n=2/4) were not comfortable discussing genetic information and risks with patients; while 1 respondent (25%; n=1/4) was somewhat comfortable; and 1 respondent (25%; n=1/4) was comfortable.* ***C) Confidence in knowledge of available genetic testing options.*** *One respondent (25%; n=1/4) was not confident in their knowledge of genetic tests and available testing options; one respondent (25%; n=1/4) was somewhat confident; and 2 respondents (50%; n=2/4) were confident.* ***D) Three conditions that would benefit from medical genetic services.*** *All respondents (100%; n=4/4) were able to spontaneously cite three genetic condition that would benefit from medical genetic services.*

The majority of respondents from OBGYN self-rated their genetics knowledge as good (50%; n=6/12) or very good (17%; n=2/12). A quarter of the respondents (25%; n=3/12) felt that their genetics knowledge was fair, while only one respondent (8%; n=1/12) self-rated their genetics knowledge as poor. No respondents felt that their genetics knowledge was excellent (Figure 4A). Respondents who self-rated their knowledge as fair or poor had not included any genetics education in their continuing medical education. The respondents that self-rated their knowledge as very good recalled >10 lectures on genetics during their specialist training and reported having included genetics education in their continuing medical education, by reading journal articles, attending webinars, undertaking online searches and participating in informal discussions and academic meetings.

In contrast, when asked how comfortable they are discussing genetic information and risks with patients, the majority of respondents self-reported being not comfortable (25%; n=3/12) or somewhat comfortable (33%; n=4/12). A quarter of the respondents (25%; n=3/12) indicated that they felt comfortable discussing genetic information and risks with patients, while only two respondents (17%; n=2/12) reported feeling very comfortable (Figure 4B).

The same trend was observed when asked about their confidence in knowledge of available genetic tests and testing options, with the majority of respondents self-reporting being not confident (25%; n=3/12) or somewhat confident (33%; n=4/12). A quarter of the respondents (25%; n=3/12) indicated that they felt confident and only two respondents (17%; n=2/12) reported feeling very confident (Figure 4C).

On an individual level there was no clear trend between self-rated genetics knowledge, comfort discussing genetic information and risks with patients, confidence in knowledge of available genetic tests or with testing options. However, it was interesting to note that one of the respondents who self-rated their genetics as very good also indicated that they were very comfortable discussing genetic information and risks with patients and very confident in their knowledge of available genetic tests and testing options. This same

respondent recalled receiving >10 lectures on genetics during their specialist training and had included genetics education in their continuing medical education. Similarly, the respondent who self-rated their genetics knowledge as poor, also indicated that they were not comfortable discussing genetic information and risks with patients, and were not confident in their knowledge of available genetic tests and testing options. This respondent recalled receiving no genetics lectures during their specialist training and had never included any genetics in their continuing medical education.

Consistent with the majority of medical specialists in OBGYN self-rating their genetics knowledge as good or very good, more than half of the respondents were able to spontaneously cite three genetic conditions that would benefit from medical genetic services (58%; n=7/12), while two respondents (17%; n=2/12) spontaneously cited four conditions. Three respondents were only able to spontaneously recall one such condition (25%; n=3/12; Figure 4D). The respondent who self-rated their knowledge as very good, and reported being very comfortable and very confident, was one of the respondents who was only able to recall one genetic condition that would benefit from medical genetic services. The most common group of genetic conditions cited by medical specialists from this department were chromosomal aneuploidies, such as trisomy 21/Down syndrome, trisomy 18/Edwards syndrome, trisomy 13/Patau syndrome and monosomy X/Turner syndrome (58%; n=7/12).

When asked what would help improve their confidence in genetics, and in treating and managing patients with genetic conditions, half of the respondents mentioned support, guidance and collaboration with genetic specialists and genetic teams. Practical exposure, training, workshops and short courses on genetics were also mentioned.

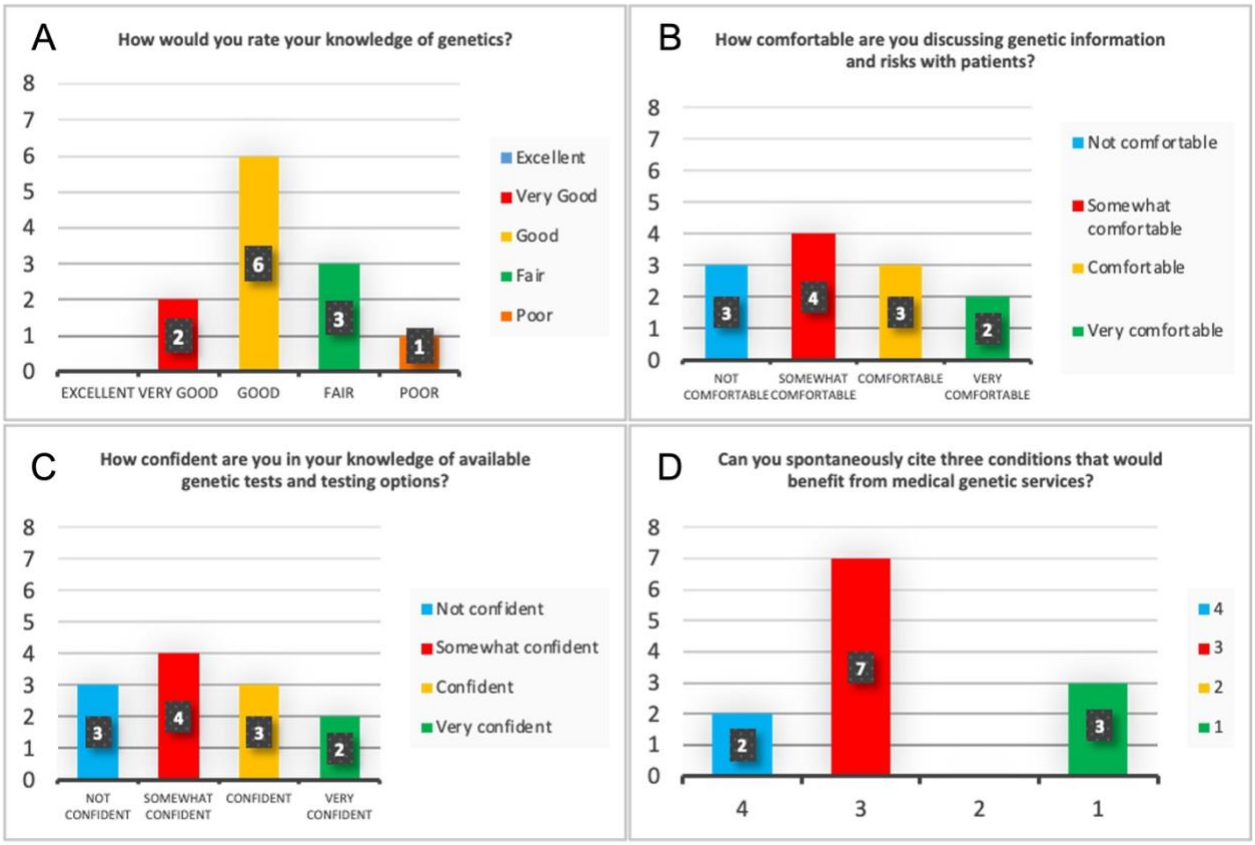

Figure 4: Medical specialists in the Department of Obstetrics and Gynaecology's self-reported genetics knowledge and confidence in treating patients with genetic conditions. A) Self-reported genetics knowledge. *One respondent (8%; n=1/12) self-reported their genetics knowledge as poor, 3 respondents (25%; n=3/12) as fair, 6 respondents (50%; n=6/12) as good, and 2 respondents (17%; n=2/12) as very good.* ***B) Comfort in discussing genetic information and risk with patients.*** *Three respondents (25%; n=3/12) were not comfortable discussing genetic information and risks with patients; while 4 respondents (33%; n=4/12) were somewhat comfortable; 3 respondents (25%; n=3/12) were comfortable and two respondents (17%; n=2/12) were very comfortable discussing genetic information and risks with patients.* ***C) Confidence in knowledge of available genetic testing options.*** *Three respondents (25%; n=3/12) were not confident in their knowledge of genetic tests and available testing options; 4 respondents (33%; n=4/12) were somewhat confident; 3 respondents (25%; n=3/12) were confident and 2 respondents (17%; n=2/12) were very confident in their knowledge of genetic tests and available testing options.* ***D) Three conditions that would benefit from medical genetic services.*** *Seven respondents (58%; n=7/12) were able to spontaneously cite three genetic condition that would benefit from medical genetic services; while 3 respondents (25%; n=3/12) were only able to spontaneously cite one genetic condition and 2 respondents (17%; n=2/12) were able to spontaneously cite four genetic conditions.*

An analysis of the responses from PCH revealed that the majority of respondents self-rated their genetics knowledge as fair (83%; n=5/6), while only one respondent felt that their knowledge was good (17%; 1/6). No respondents felt that their knowledge was excellent, very good or poor (Figure 5A). The respondent who rated their genetics knowledge as good reported to having included genetics education in their continuing medical education and by participating in self-directed learning from journals and textbooks.

When describing their comfort with discussing genetic information and risks with patients, most of the respondents (83%; n=5/6) felt somewhat comfortable, while one respondent (17%; n=1/6) reported not being comfortable. No respondents indicated feeling comfortable or very comfortable. On an individual level, the respondent who self-rated their knowledge as good only reported being somewhat comfortable discussing risks and genetic information with patients. In addition, one of the respondents who self-rated their knowledge as fair, reported not being comfortable discussing risks and genetic information with patients (Figure 5B).

When the medical specialists from PCH were asked to rate their confidence in their knowledge of genetic tests and testing options, all the respondents (100%: n=6/6) reported being somewhat confident. No respondents were very confident, confident or not confident in their knowledge of genetic tests and testing options (Figure 5C).

All medical specialists from this department were able to spontaneously cite three genetic conditions that would benefit from medical genetic services (100%; n=6/6; Figure 5D). The most commonly cited genetic conditions by medical specialists from this department were osteogenesis imperfecta (50%; n=3/6), followed by haemophilia (33%; n=2/6) and trisomy 21/Down syndrome (33%; n=2/6).

Medical specialists from this department felt that workshops or webinars, more training and access to medical genetic services would help improve their confidence when dealing with such conditions. One respondent specifically mentioned having access to a genetic

counsellor based in Limpopo and being exposed to a genetics clinic at least once a month alongside a genetics team would be of benefit to them and help improve their confidence.

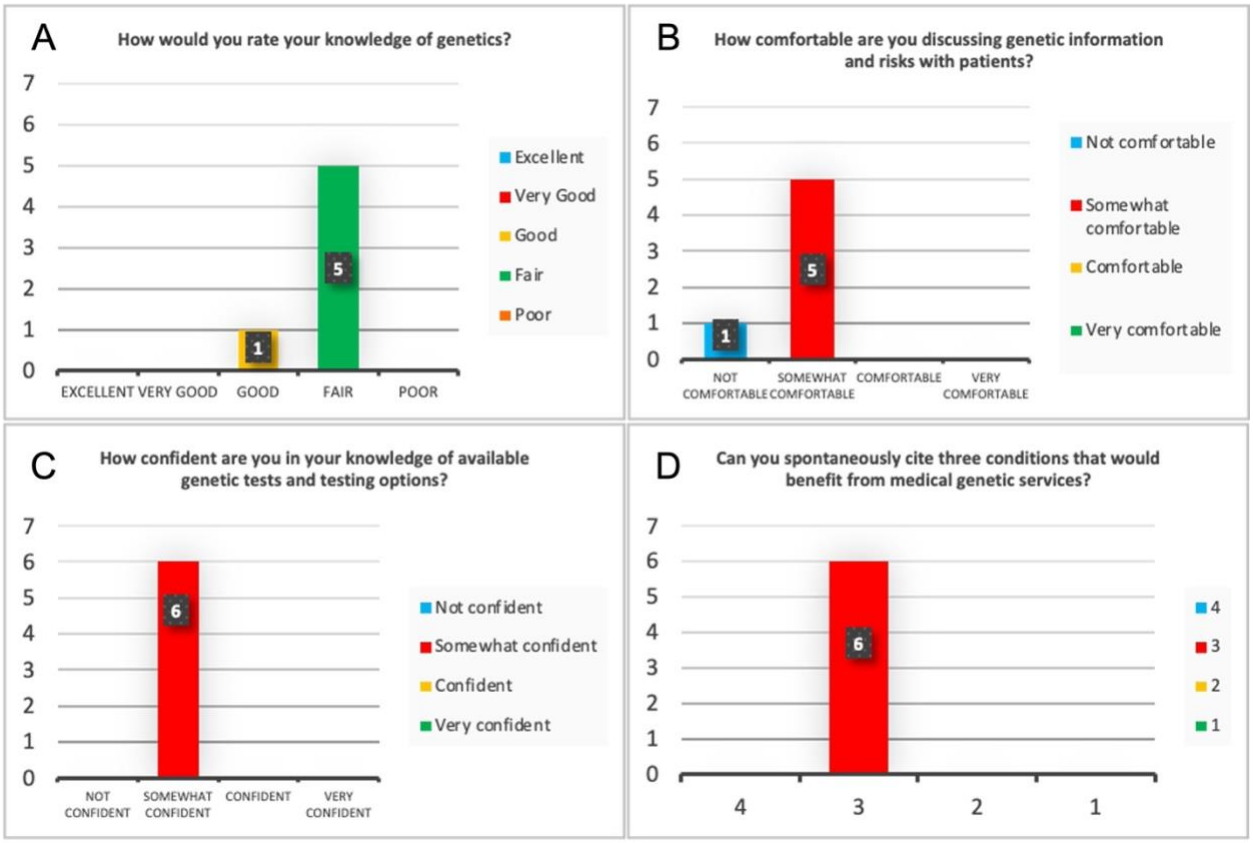

Figure 5: Medical specialists in the Department of Paediatrics and Child Health's self-reported genetics knowledge and confidence in treating patients with genetic conditions. A) Self-reported genetics knowledge. *Five respondents (83%; n=5/6) self-reported their genetics knowledge as fair and one (17%; n=1/6) as good.* ***B) Comfort in discussing genetic information and risk with patients.*** *One respondent (17%; n=1/6) was not comfortable discussing genetic information and risks with patients; while 5 respondents (83%; n=5/6) were somewhat comfortable.* ***C) Confidence in knowledge of available genetic testing options.*** *All 6 respondents (100%; n=6/6) were somewhat confident in their knowledge of genetic tests and available testing options.* ***D) Three conditions that would benefit from medical genetic services.*** *All 6 respondents (100%; n=6/6) were able to spontaneously cite three genetic condition that would benefit from medical genetic services.*

Responses from participants from SG indicated that three quarters of the respondents (67%; n=4/6) felt that their genetics knowledge was fair, while the remaining third (33%; n=2/6) self-rated their knowledge as poor (Figure 6A). This is consistent with the majority of respondents (67%; n=4/6) recalling not receiving any genetics education during specialist training and not including genetics education in their continuing medical education. However, despite this self-rated lack of genetics knowledge, the majority of respondents (83%; n=5/6) were able to spontaneously cite three genetic conditions that would benefit from medical genetic services. Only one respondent could recall only two such conditions (Figure 6D). The majority of genetic conditions cited from this department were cancers, such as breast cancer, colon cancer, thyroid cancer, hereditary colorectal carcinoma, multiple endocrine adenomatosis and multiple endocrine neoplasia type 1 and type 2.

An analysis of the responses from SG on comfort when discussing genetic information and risks with patients revealed that the majority of respondents (50%; n=3/6) felt comfortable, despite self-rating their genetics knowledge as poor or fair. A third of the respondents (33%; n=2/6) reported being somewhat comfortable, while one respondent (17%; n=1/6) reported being not comfortable discussing genetic information and risks with patients (Figure 6B).

Medical specialists from SG reported an overall lack of confidence in their knowledge of available genetic tests and testing options, with half of the respondents (50%; n=3/6) reporting feeling somewhat confident, while the other half (50%; n=3/6) were not confident (Figure 6C).

Respondents felt that their confidence could be improved by exposure to genetic services and genetic-related issues, as well as gaining knowledge and participating in training. One respondent felt that it was not applicable in their context; however, this respondent reported their specific field of interest as trauma surgery and organ transplant surgery.

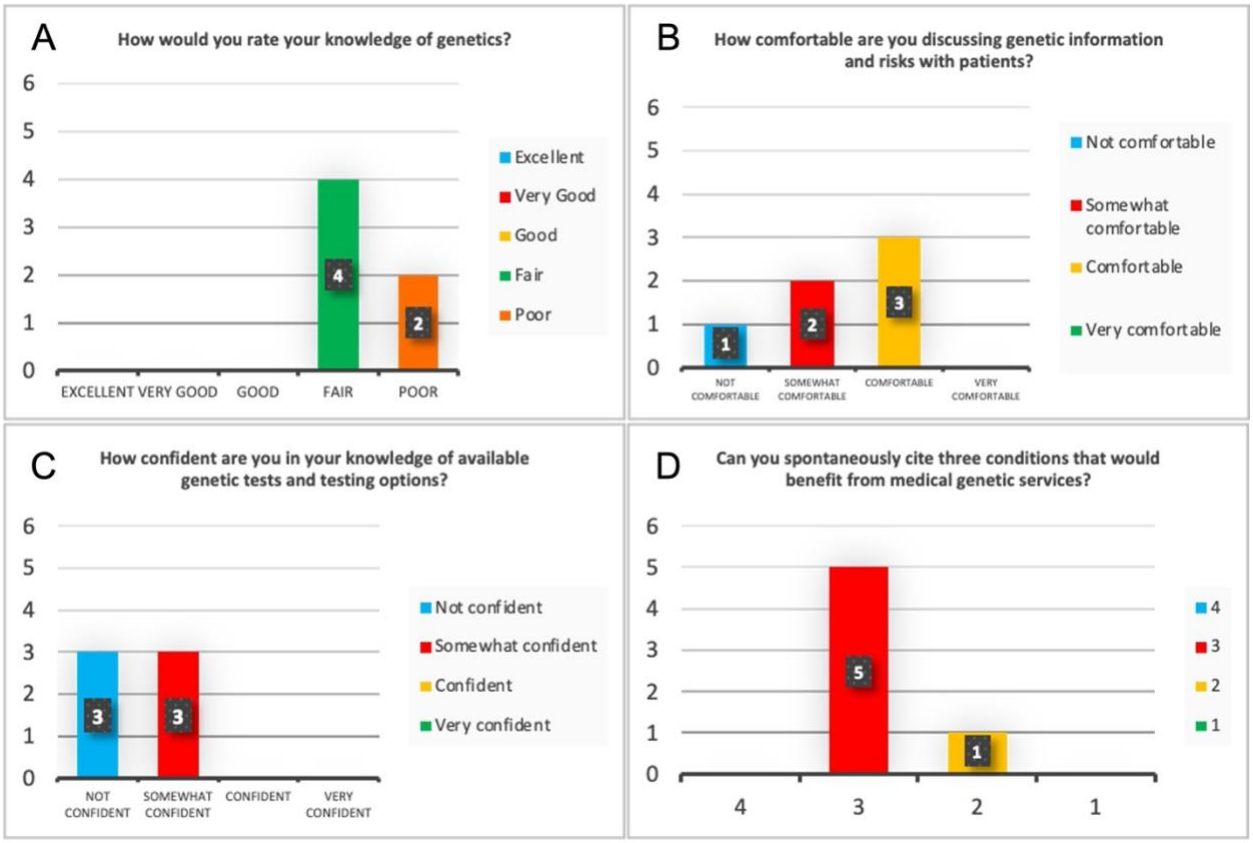

Figure 6: Medical specialists in the Department of Surgery's self-reported genetics knowledge and confidence in treating patients with genetic conditions. A) Self-reported genetics knowledge. *Four respondents (67%; n=4/6) self-reported their genetics knowledge as fair and two (33%; n=2/6) as good.* ***B) Comfort in discussing genetic information and risk with patients.*** *One respondent (17%; n=1/6) was not comfortable discussing genetic information and risks with patients; while 2 respondents (33%; n=2/6) were somewhat comfortable; and three respondents (50%; n=3/6) were comfortable discussing genetic information and risks with patients.* ***C) Confidence in knowledge of available genetic testing options.*** *Three respondents (50%; n=3/6) were not confident in their knowledge of genetic tests and available testing options, while 3 respondents (50%; n=3/6) were somewhat confident.* ***D) Three conditions that would benefit from medical genetic services.*** *Five respondents (83%; n=5/6) were able to spontaneously cite three genetic condition that would benefit from medical genetic services; while one respondent (17%; n=1/6) cited two conditions.*

At the end of this section in the survey questionnaire, respondents were asked an open-ended question on what they thought would help improve their confidence. The responses can be grouped into two main suggestions:

Suggestion 1: Access to information and training

Suggestion 2: Practical exposure and access to expert advice

Suggestion 1: Access to information and training. The majority of specialists (63%; n=20/32) felt that access to information and training on genetics and genetic conditions would improve their confidence in dealing with patients with genetic conditions. This is evident in the following selected responses from participants:

> *"Continuous education, training, contact with genetic team."*
>
> *"More training and practical exposure around genetic services."*
>
> *"Training - webinars, workshops. Something very practical. Hands on – not very theoretical."*

Suggestion 2: Practical exposure and access to expert advice. Half of the specialists (50%; n=16/32) felt that practical exposure to patients with genetic conditions and access to expert advice through consultations, collaborations and medical genetic services would help improve their confidence.

> *"Continuous education, training, contact with genetic team."*
>
> *"Regular meetings with geneticists, conferences related to genetics."*
>
> *"Involvement of geneticists in cases (consultations)."*
>
> *"More practice, more involvement with patients."*
>
> *"Constant exposure, collaborations, consultations."*

"Multidisciplinary teams - discussions and collaborations."

"Time to read and learn more. Telephonic availability of advice."

"Having access to a genetic counsellor that is based in Limpopo. Being exposed to a Genetic clinic at least once a month with them."

3.1.3. Opinion of medical genetic services

In this section of the survey questionnaire, respondents were asked whether they felt that medical geneticists, genetic counsellors and genetic testing services were a luxury, a necessity/indispensable or not applicable to their setting in the context of the Limpopo provincial health system. This section was included in order to ascertain whether respondents felt that comprehensive medical genetic services were a necessary support service for their clinical practice and patients in Limpopo.

The vast majority of medical specialists (97%; n=31/32) felt that access to the services of a medical geneticist was a necessity/indispensable. One respondent (3%; n=1/32) from PCH felt that this service was a luxury in the context of the Limpopo provincial health system (Figure 7A). In response to how medical specialists viewed genetic counselling and genetic testing services, all respondents (100%; n=32/32) indicated that they felt both these services were a necessity/indispensable (Figure 7B and C).

Respondents were then asked which services they thought should be included in comprehensive medical genetic services. The options provided were: medical geneticists; genetic counsellors; and genetic testing services. They were asked to select all the services that they thought should be included in medical genetic services. The majority of respondents (94%; n=30/32) felt that all three services should be included in comprehensive medical genetic services (Figure 7D). Two respondents (6%; n=2/32) from PCH felt that medical geneticists should not be included in comprehensive medical genetic services in the Limpopo provincial health system and felt that only genetic counsellors and genetic testing services were necessary (Figure 7D).

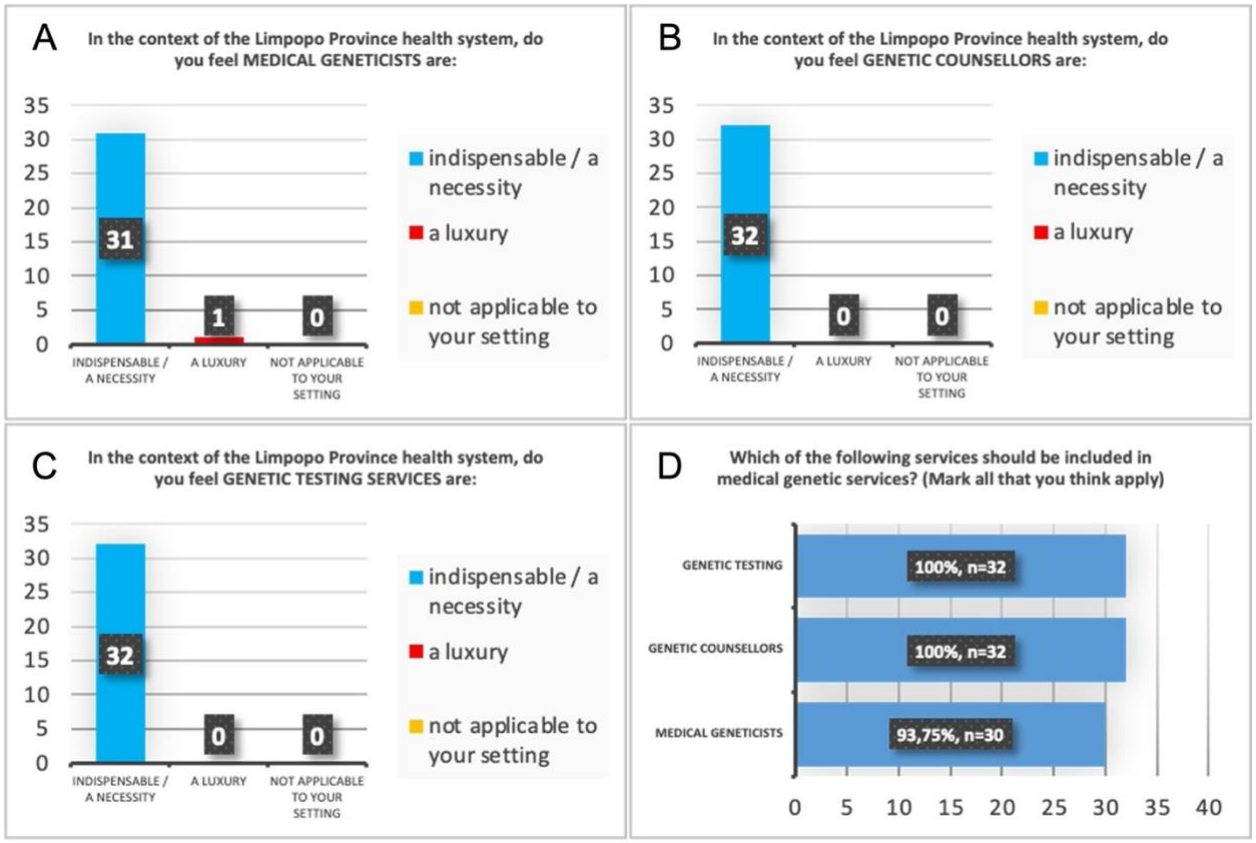

Figure 7: Opinions of medical specialists about comprehensive medical genetic services in the context of the Limpopo health system. A) Medical geneticists. *Thirty-one (97%; n=31/32) respondents felt that medical geneticists were indispensable or a necessity, while one respondent (3%; n=1/32) felt that medical geneticists were a luxury.* ***B) Genetic counsellors.*** *All medical specialists (100%; n=32/32) felt that genetic counsellors were indispensable or a necessity.* ***C) Genetic testing services.*** *All medical specialists (100%; n=32/32) felt that genetic testing was indispensable or a necessity.* ***D) Opinion about which services should be included in medical genetic services.*** *All medical specialists (100%; n=32/32) felt that genetic testing and genetic counsellors should be included in medical genetic services in the Limpopo health system, while 30 medical specialists (94%; n=30/32) felt that medical geneticists should be included.*

3.1.4. Practices

In this section, the medical specialists were asked whether they had seen any patients in the past 12 months who they thought would benefit from seeing a medical geneticist or a genetic counsellor, or who would benefit from genetic testing services. They were then asked to indicate whether they had referred any patients or families for such services in the past 12 months. This section was included in order to better understand the need for medical genetic services in the province and to identify whether patients and families were being referred to out-of-province services in the absence of locally available services.

Only 7 specialists (25%; n=7/28) had referred patients to a medical geneticist in the past 12 months (Figure 8A and B). When analysed as individual departments, the department with the highest referral rate was PCH. All (100%;n=6/6) respondents indicated that they had treated patients in the past 12 months who would have benefitted from seeing a medical geneticist, and 50% (n=3/6) had referred patients (Figure 8C and D). Within INTMED and OBGYN, while almost all medical specialists had seen patients who would have benefitted from seeing a medical geneticist (100%; n=4/4 and 92%; n=11/12 respectively), only a quarter of the respondents had referred patients to a medical geneticist in the past 12 months (25%; n=1/4 and 25%; n=3/12; Figure 8C and D). Within FMED and SG, no patients had been referred to a medical geneticist, despite the majority of respondents indicating that they had seen patients in the past 12 months who would have benefitted from seeing a medical geneticist (75%; n=3/4 and 67%; n=4/6 respectively; Figure 8C and D).

Results of participant responses for each specific aspect of comprehensive medical genetic services, medical geneticists, genetic counselling and genetic testing respectively, are presented in the next section.

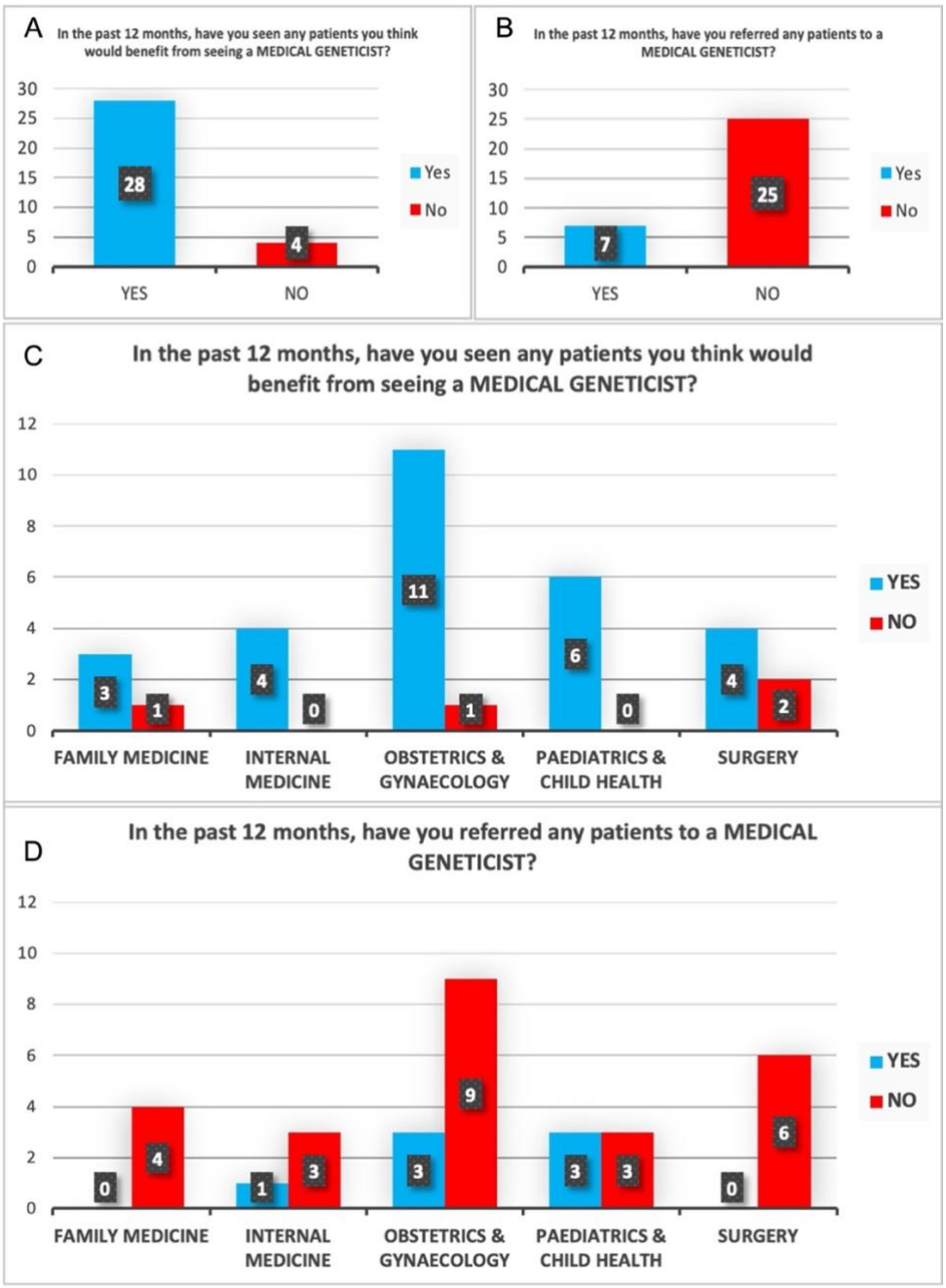

Figure 8: Patient identification and referral practices to medical geneticist services. A) *Twenty-eight medical specialists (88%; n=28/32) had seen patients that would benefit from consulting a medical geneticist, while 4 participants (13%; n=4/32) had not.* ***B)*** *However, only 7 medical specialists (22%; n=7/32) had referred any patients for consultation.* ***C)*** *Seventy-five percent (n=3/4) of medical specialists from the Department of Family Medicine had seen patients that would benefit from consulting a medical geneticist; 100% (n=4/4) from the Department of Internal Medicine; 92% (n=11/12) from the Department of Obstetrics and Gynaecology; 100% (n=6/6) from the Department of Paediatrics and Child Health; and 67% (n=4/6) from the Department of Surgery.* ***B)*** *However, no medical specialists (0%; n=0/4) from the Department of Family Medicine had referred any patients for consultation; 25% (n=1/4) from the Department of Internal Medicine; 25% (n=3/12) from the Department of Obstetrics and Gynaecology; 50% (n=3/6) from the Department of Paediatrics and Child Health; and no medical specialists (0%; n=0/6) from the Department of Surgery.*

When medical specialists were asked to indicate whether they had seen any patients or families in the past 12 months who would benefit from genetic counselling, the vast majority of respondents (81%; n=26/32) indicated that they had, while only 28% (n=9/32) of the respondents reported referring patients or families to genetic counselling services (Figure 9A and B). This translates into a referral rate of 35% (n=9/26).

When analysed per department, PCH once again had the highest referral rate with two thirds (67%; n=4/6) of the medical specialists indicating that they had referred patients or families for genetic counselling in the past 12 months (Figure 9C and D). Within OBGYN, 11 of the respondents (92%; n=11/12) indicated that they had seen patients or families who would have benefitted from seeing a genetic counsellor; however, only four medical specialists (33%; n=4/12) had referred patients or families to these services (Figure 9C and D). This translates into a referral rate of just over a third (36%; n=4/11). Within INTMED, while all respondents (100%; n=4/4) had seen patients or families in the past 12 months who they felt would have benefitted from seeing a genetic counsellor, only one medical specialist (25%; n=1/4) had referred patients or families to these services (Figure 9C and D). In both FMED and SG, half of the respondents (50%; n=2/4 and n=3/6 respectively) reported that they had seen patients or families in the past 12 months who would have benefitted from genetic counselling services; however, none of the medical specialists from these two departments had referred any patients or families (Figure 9C and D).

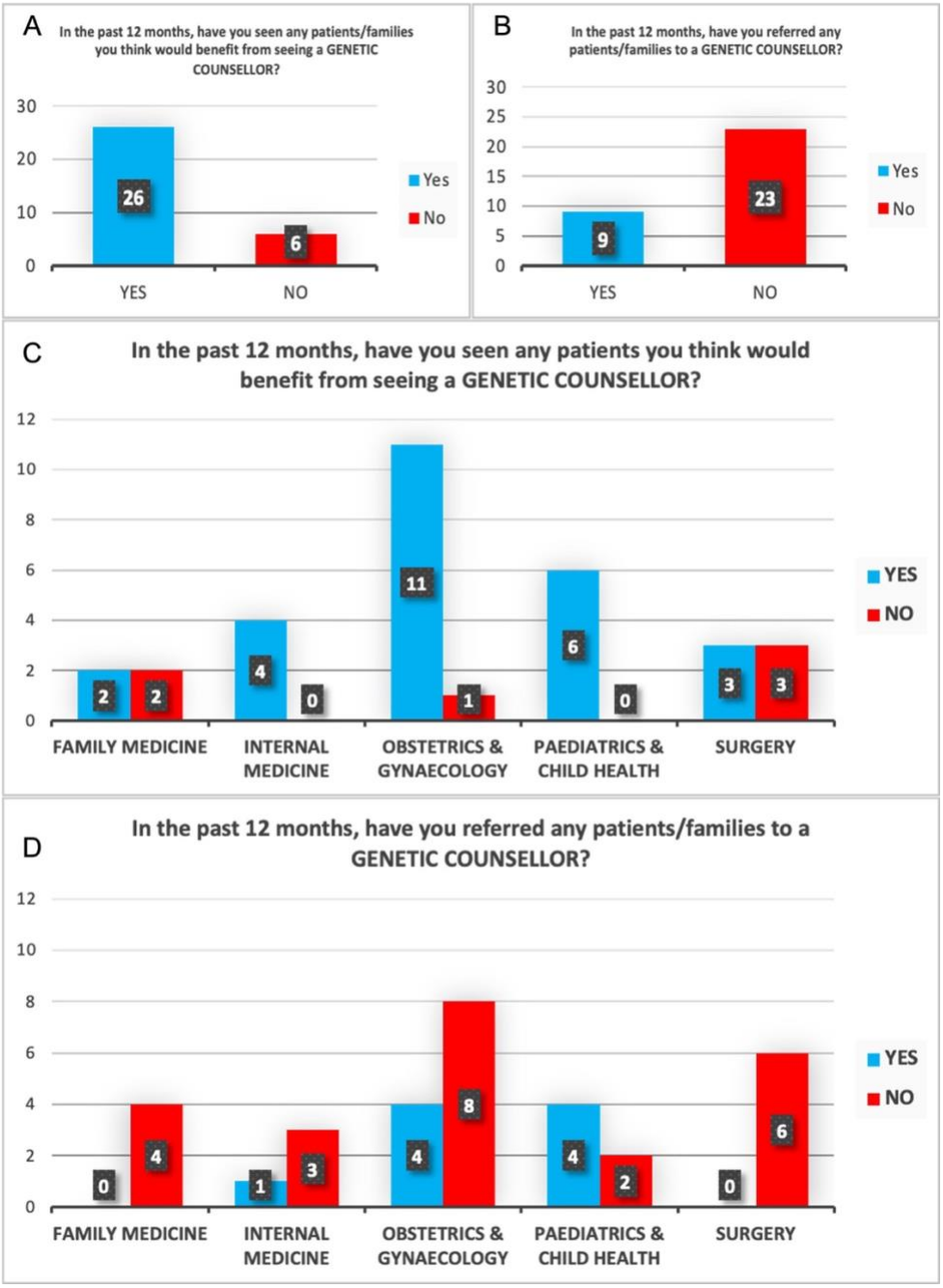

Figure 9: Patient identification and referral practices to genetic counselling services. A) *Twenty-six medical specialists (81%; n=26/32) had seen patients that would benefit from genetic counselling, while 6 participants (19%; n=6/32) had not.* ***B)*** *However, only 9 medical specialists (28%; n=9/32) had referred any patients for consultation.* ***C)*** *Fifty percent (n=2/4) of medical specialists from the Department of Family Medicine had seen patients that would benefit from genetic counselling; 100% (n=4/4) from the Department of Internal Medicine; 92% (n=11/12) from the Department of Obstetrics and Gynaecology; 100% (n=6/6) from the Department of Paediatrics and Child Health; and 50% (n=3/6) from the Department of Surgery.* ***B)*** *However, no medical specialists (0%; n=0/4) from the Department of Family Medicine had referred any patients for genetic counselling; 25% (n=1/4) from the Department of Internal Medicine; 33% (n=4/12) from the Department of Obstetrics and Gynaecology; 67% (n=4/6) from the Department of Paediatrics and Child Health; and no medical specialists (0%; n=0/6) from the Department of Surgery.*

The next service within the scope of comprehensive medical genetics that the medical specialists were asked about was genetic testing. This service was the one that was most utilised, with 30 of the respondents (94%; n=30/32) reporting having seen patients that would have benefitted from genetic testing services and half of the respondents (50%; n=16/32) indicating that they had sent patients samples for genetic testing (Figure 10A and B).

The respondents who indicated that they had sent samples for genetic testing were from INTMED, OBGYN, and PCH. In addition, all respondents from these departments reported having seen patients who would have benefitted from genetic testing services in the past 12 months (Figure 10C and D). PCH was the department that utilised these services the most, with all of the respondents (100%; n=6/6) indicating that they had sent patient samples for genetic testing in the past 12 months (Figure 10C and D). Two thirds of the respondents (67%; n=8/12) from OBGYN indicated that they had sent samples for genetic testing in the past 12 months, while half of the respondents (50%; n=2/4) from INTMED reported having sent samples for testing (Figure 10C and D). The departments that utilised this service the least were FMED and SG, with no respondents reporting sending samples for genetic testing in the past 12 months, despite 75% (n=3/4) and 83% (n=5/6) of respondents, respectively, indicating that they had seen patients that would benefit from such a service (Figure 10C and D).

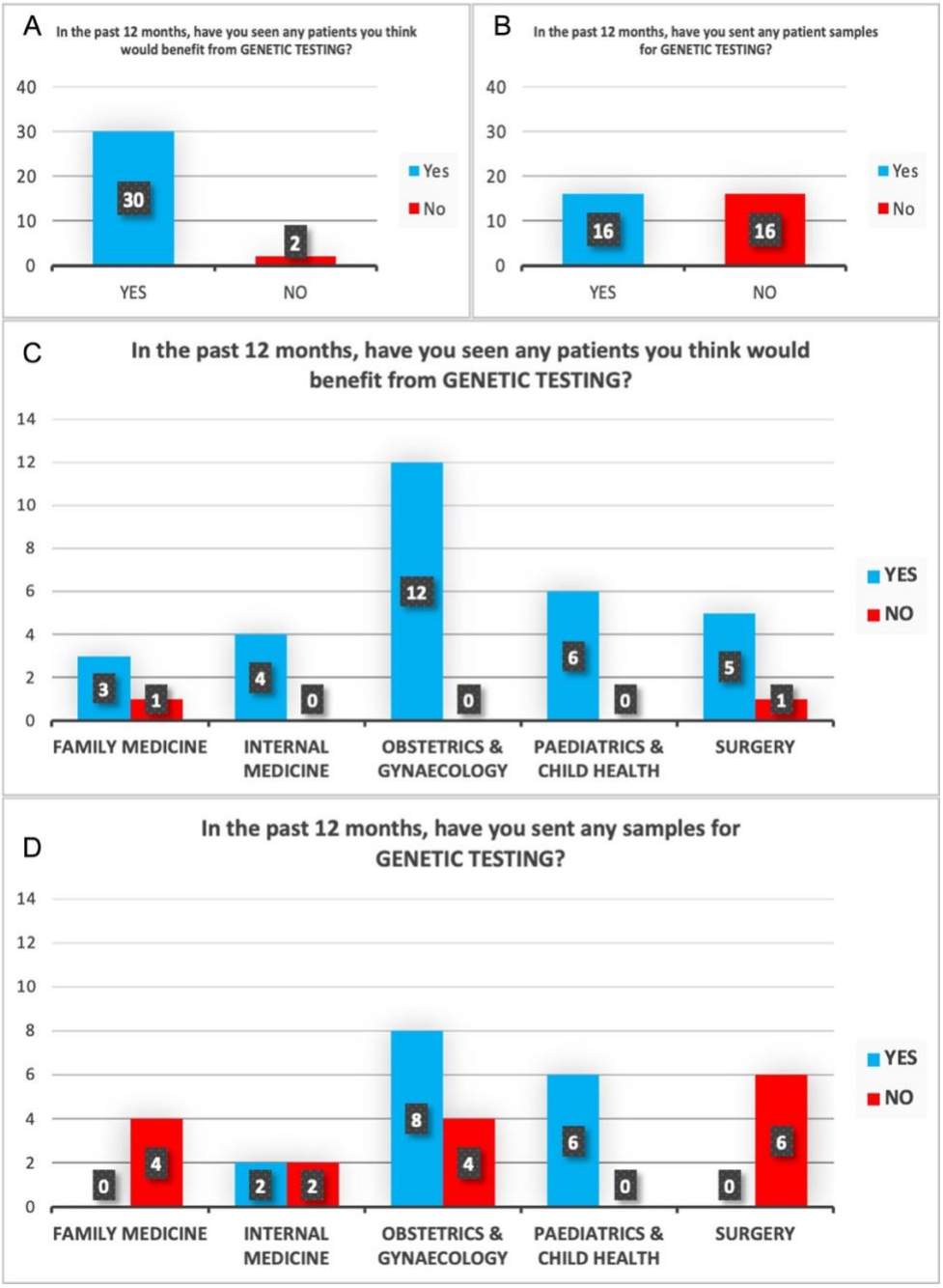

Figure 10: Patient identification and referral practices for genetic testing. A) *Thirty medical specialists (94%; n=30/32) had seen patients that would benefit from genetic testing.* ***B)*** *However, 16 medical specialists (50%; n=16/32) had sent samples for genetic testing.* ***C)*** *Seventy-five percent (n=3/4) of medical specialists from the Department of Family Medicine had seen patients that would benefit from genetic testing; 100% (n=4/4) from the Department of Internal Medicine; 100% (n=12/12) from the Department of Obstetrics and Gynaecology; 100% (n=6/6) from the Department of Paediatrics and Child Health; and 83% (n=5/6) from the Department of Surgery.* ***D)*** *However, no medical specialists (0%; n=0/4) from the Department of Family Medicine had sent any samples for genetic testing; 50% (n=2/4) from the Department of Internal Medicine; 67% (n=8/12) from the Department of Obstetrics and Gynaecology; 100% (n=6/6) from the Department of Paediatrics and Child Health; and no medical specialists (0%; n=0/6) from the Department of Surgery.*

An analysis of the three core services together revealed that a total of six respondents (19%; n=6/32) had referred patients or families to all three services; medical geneticists, genetic counsellors and genetic testing, in the past 12 months. This included one respondent from INTMED, two from OBGYN and three from PCH. Interestingly, the respondent from INTMED was the respondent who had completed the DoH genetics course. Only two respondents (6%; n=2/32) indicated that they had not seen any patients or families who would have benefitted from seeing a medical geneticist, genetic counsellor or genetic testing in the past 12 months, one from SG and one from FMED.

Following the section on patients and referrals, the medical specialists were asked whether they would refer patients to medical genetic services (medical geneticist, genetic counsellor or genetic testing) in the future, if they knew how to access these services in South Africa and whether they would like improved access to these services. All respondents (100%; n=32/32) reported that they would refer patients to these services in the future (Figure 11A), despite more than half of the respondents (63%; n=20/32) reporting that they do not know how to access medical genetic services in South Africa (Figure 11B). In addition, all respondents (100%; n=32/32) indicated that they would like improved access to medical genetic services (Figure 11C).

Despite all respondents from the different departments reporting that they will refer patients to medical genetic services in the future, the majority of medical specialists from FMED (100%; n=4/4), INTMED (75%; n=3/4), OBGYN (58%; n=7/12) and SG (83%; n=5/6) indicated that they did not know how to access medical genetics services in South Africa (Figure 11D). The majority of respondents (83%; n=5/6) from PCH indicated that they did know how to access these services (Figure 11D).

Of those respondents who reported not knowing how to access medical genetic services in South Africa, 65% (n=13/20) had never referred patients to any of the three core services within comprehensive medical genetic services, 30% (n=6/20) had sent patient samples for genetic testing and 5% (n=1/20) had referred patients or families for genetic counselling.

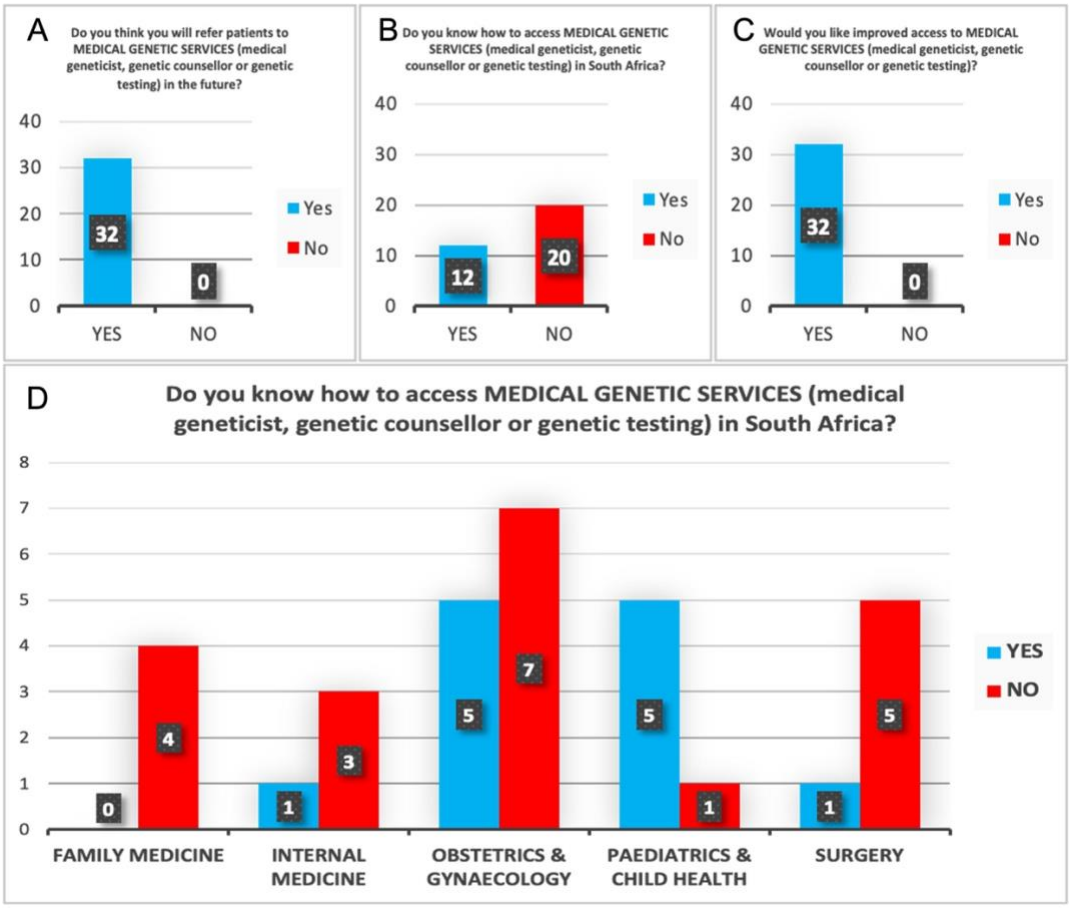

Figure 11: Accessing medical genetic services. A) *All medical specialists (100%; n=32/32) were of the opinion that they will refer patients to comprehensive medical genetic services in the future.* ***B)*** *However, only 12 medical specialists (38%; n=12/32) reported knowing how to access medical genetic service in South Africa.* ***C)*** *All medical specialists (100%; n=32/32) reported wanting improved access to medical genetic services.* ***D)*** *No medical specialists (0%; n=0/4) from the Department of Family Medicine knew how to access medical genetic services in South Africa; 25% (n=1/4) from the Department of Internal Medicine; 42% (n=5/12) from the Department of Obstetrics and Gynaecology; 83% (n=5/6) from the Department of Paediatrics and Child Health; and one medical specialist (17%; n=1/6) from the Department of Surgery reported knowing how to access medical genetic services in South Africa.*

At the end of the survey questionnaire the respondents were asked an open-ended question “Is there anything you would like to add?” The majority of responses reiterated the need for medical genetic services in Limpopo. This is apparent in the selected responses below:

> *“Our provinces has a lot of inherited conditions (especially in neurology) due to consanguinity. We really need these services in our province.”*

> *“Having access to a medical geneticist and genetic counsellor in the province would be great, because that would limit patients being transferred to another province for these services. The testing so far is not a problem.”*

> *“Medical genetic service should be made available in the province to reduce time to intervention as it is delayed by referral outside of the province.”*

> *“Screening for aneuploidies is needed in the province.”*

One response addressed the need for genetic education and training:

> *“More training for doctors in terms of genetic services and counselling; contact with geneticists about testing options.”*

Others referred to barriers experienced in accessing medical genetic services:

> *“For genetic testing we rely on NHLS. However, do not have direct access to genetic counsellor or medical geneticist.”*

> *“Tried referring patient to Pretoria for genetic counselling; samples sent to Pretoria but come back after a long time.”*

> *“AMA, diabetes, hypertension - need for services; do not know how to access medical genetic services in Limpopo Province”*

3.1.5. Inferential statistical analysis

Fisher's exact test was used to determine if there was a significant association between demographic, education and training, genetics education, genetics knowledge and referral practices to medical genetic services. This statistical analysis revealed a statistically significant association between knowing how to access medical genetic services and having referred patients to a medical geneticist in the past 12 months (two-tailed $p<0.001$). A statistically significant association was also shown between knowing how to access medical genetic services and having referred patients or families to a genetic counsellor in the past 12 months (two-tailed $p<0.001$). All other associations tested were not shown to be statistically significant ($p<0.05$). Results obtained from the inferential statistical analysis are reported in Table 5.

Table 5: Results obtained from Inferential Statistical Analysis

	In the past 12 months, have you seen any patients you think would benefit from seeing a MEDICAL GENETICIST?	**In the past 12 months, have you seen any patients/families you think would benefit from seeing a GENETIC COUNSELLOR?**	**In the past 12 months, have you seen any patients you think would benefit from GENETIC TESTING?**	**In the past 12 months, have you referred any patients to a MEDICAL GENETICIST?**	**In the past 12 months, have you referred any patients/families to a GENETIC COUNSELLOR?**	**In the past 12 months, have you sent any patient samples for GENETIC TESTING?**
Department	Chi-square = 0.419 Fischer = 0.331 Cramer's V = 0.378	Chi-square = 0.055 Fischer = 0.054 Cramer's V = 0.547	Chi-square = 0.310 Fischer = 0.310 Cramer's V = 0.394	Chi-square = 0.258 Fischer = 0.217 Cramer's V = 0.421	Chi-square = 0.075 Fischer = 0.082 Cramer's V = 0.515	**Chi-square <0.001** **Fischer <0.001** **Cramer's V = 0.736**
Age	Chi-square = 1.0 Fischer = 1.0 Cramer's V = 0.147	Chi-square = 0.933 Fischer = 1.0 Cramer's V = 0.132	Chi-square = 1.0 Fischer = 1.0 Cramer's V = 0.203	Chi-square = 0.291 Fischer = 0.375 Cramer's V = 0.347	Chi-square = 0.388 Fischer = 0.438 Cramer's V = 0.319	Chi-square = 0.849 Fischer = 0.849 Cramer's V = 0.195
Gender	Chi-square = 0.689 Fischer = 0.689 Cramer's V = 0.026	Chi-square = 0.554 Fischer = 0.554 Cramer's V = 0.056	Chi-square = 0.510 Fischer = 0.510 Cramer's V = 0.162	Chi-square = 0.297 Fischer = 0.297 Cramer's V = 0.173	Chi-square = 0.499 Fischer = 0.499 Cramer's V = 0.072	Chi-square = 0.500 Fischer = 0.500 Cramer's V = 0.070
When did you complete your undergraduate training?	Chi-square = 0.692 Fischer = 0.692 Cramer's V = 0.239	Chi-square = 1.0 Fischer = 1.0 Cramer's V = 0.148	Chi-square = 0.438 Fischer = 0.438 Cramer's V = 0.264	Chi-square = 0.509 Fischer = 0.471 Cramer's V = 0.350	Chi-square = 0.188 Fischer = 0.149 Cramer's V = 0.443	Chi-square = 0.830 Fischer = 0.830 Cramer's V = 0.283
When did you complete your specialist training?	Chi-square = 0.662 Fischer = 0.662 Cramer's V = 0.218	Chi-square = 0.460 Fischer = 0.403 Cramer's V = 0.297	Chi-square = 0.746 Fischer = 0.438 Cramer's V = 0.228	Chi-square = 0.556 Fischer = 0.607 Cramer's V = 0.280	Chi-square = 0.388 Fischer = 0.337 Cramer's V = 0.342	Chi-square = 1.0 Fischer = 1.0 Cramer's V = 0.189
How many years have you worked as a medical specialist?	Chi-square = 0.841 Fischer = 1.0 Cramer's V = 0.183	Chi-square = 0.482 Fischer = 0.557 Cramer's V = 0.265	Chi-square = 0.617 Fischer = 1.0 Cramer's V = 0.221	Chi-square = 0.478 Fischer = 0.529 Cramer's V = 0.293	Chi-square = 0.367 Fischer = 0.367 Cramer's V = 0.333	Chi-square = 1.0 Fischer = 1.0 Cramer's V = 0.194
How much genetics education did you receive in undergraduate training?	Chi-square = 0.186 Fischer = 0.102 Cramer's V = 0.488	Chi-square = 0.009 Fischer = 0.007 Cramer's V = 0.692	Chi-square = 0.339 Fischer = 0.282 Cramer's V = 0.422	Chi-square = 0.649 Fischer = 0.706 Cramer's V = 0.348	Chi-square = 0.297 Fischer = 0.382 Cramer's V = 0.446	Chi-square = 0.169 Fischer = 0.187 Cramer's V = 0.498
How much genetics education did you receive in your specialist training?	Chi-square = 0.325 Fischer = 0.268 Cramer's V = 0.428	Chi-square = 0.333 Fischer = 0.216 Cramer's V = 0.439	Chi-square = 0.218 Fischer = 0.339 Cramer's V = 0.491	Chi-square = 0.221 Fischer = 0.146 Cramer's V = 0.466	Chi-square = 0.146 Fischer = 0.081 Cramer's V = 0.500	Chi-square = 0.016 Fischer = 0.016 Cramer's V = 0.613

How many hours per week do you dedicate to continuing medical education?	Chi-square = 0.744 Fischer = 1.0 Cramer's V = 0.200	Chi-square = 0.422 Fischer = 0.765 Cramer's V = 0.254	Chi-square = 1.0 Fischer = 1.0 Cramer's V = 0.137	Chi-square = 1.0 Fischer = 0.790 Cramer's V = 0.098	Chi-square = 0.108 Fischer = 0.108 Cramer's V = 0.352	Chi-square = 0.100 Fischer = 0.100 Cramer's V = 0.395
Has your continuing education ever included genetic education?	Chi-square = 0.689 Fischer = 0.689 Cramer's V = 0.026	Chi-square = 0.446 Fischer = 0.446 Cramer's V = 0.122	Chi-square = 0.490 Fischer = 0.490 Cramer's V = 0.126	Chi-square = 0.297 Fischer = 0.297 Cramer's V = 0.173	Chi-square = 0.499 Fischer = 0.499 Cramer's V = 1.0	Chi-square = 0.500 Fischer = 0.500 Cramer's V = 0.070
How would you rate your knowledge of genetics?	Chi-square = 0.838 Fischer = 0.838 Cramer's V = 0.237	Chi-square = 0.529 Fischer = 0.619 Cramer's V = 0.290	Chi-square = 1.0 Fischer = 1.0 Cramer's V = 0.195	Chi-square = 0.277 Fischer = 0.167 Cramer's V = 0.367	Chi-square = 0.181 Fischer = 0.092 Cramer's V = 0.415	Chi-square = 0.217 Fischer = 0.235 Cramer's V = 0.375
How comfortable are you discussing genetic information and risks with patients?	Chi-square = 0.216 Fischer = 0.110 Cramer's V = 0.362	Chi-square = 1.0 Fischer = 1.0 Cramer's V = 0.170	Chi-square = 0.216 Fischer = 0.103 Cramer's V = 0.422	Chi-square = 0.346 Fischer = 0.274 Cramer's V = 0.337	Chi-square = 0.240 Fischer = 0.182 Cramer's V = 0.363	Chi-square = 0.480 Fischer = 0.480 Cramer's V = 0.307
How confident are you in your knowledge of available genetic tests and testing options?	Chi-square = 0.848 Fischer = 0.734 Cramer's V = 0.199	Chi-square = 0.199 Fischer = 0.178 Cramer's V = 0.382	Chi-square = 0.718 Fischer = 0.534 Cramer's V = 0.246	Chi-square = 0.015 Fischer = 0.018 Cramer's V = 0.570	Chi-square = 0.016 Fischer = 0.016 Cramer's V = 0.545	Chi-square = 0.460 Fischer = 0.460 Cramer's V = 0.324
Do you know how to access MEDICAL GENETIC SERVICES (medical geneticist, genetic counsellor or genetic testing) in South Africa?	Chi-square = 0.515 Fischer = 0.515 Cramer's V = 0.098	Chi-square = 0.248 Fischer = 0.248 Cramer's V = 0.207	Chi-square = 0.617 Fischer = 0.617 Cramer's V = 0.067	**Chi-square <0.001** **Fischer = <0.001** **Cramer's V = 0.683**	**Chi-square <0.001** **Fischer <0.001** **Cramer's V = 0.664**	Chi-square = 0.033 Fischer = 0.033 Cramer's V = 0.387

3.2. Phase 2: Qualitative, Semi-structured Follow-up Interviews

3.2.1. Theme 1: Current situation in Limpopo

Interviews with participants provided a glimpse into the current situation in Limpopo in terms of the diagnosis, treatment and management of patients with congenital abnormalities and genetic conditions. This theme provides insight into the experiences of medical specialists in the province and highlights how local skills and expertise translates into appropriate care and management for patients with more common genetic conditions. In contrast, despite the availability of the necessary skills and expertise, some services are not provided due to a lack of local resources and capacity. Provision of appropriate patient care locally is further hindered by a lack of in-depth knowledge and understanding of medical genetics, coupled with a lack of local capacity, resources and services. While medical specialists can refer patients to medical genetic services in other provinces, participants felt that there were a number of challenges to consider before making use of these out-of-province referrals. This lack of local knowledge, capacity, resources and services left medical specialists feeling isolated and frustrated with the current situation in the province.

Subtheme 1: "Bread and butter": Common genetic conditions within the skills and expertise of local services

Interviews with medical specialists highlighted pockets of skills, knowledge and expertise available in the province for the treatment and management of certain patients with common genetic conditions and the provision of basic medical genetic services. Medical specialists from the PCH reported being confident enough in their genetics knowledge and expertise to provide basic genetic information and genetic counselling to families of patients with the more commonly seen conditions within this discipline, such as haemophilia, trisomy 21/Down syndrome and DMD. One participant from this department referred to trisomy 21/Down syndrome as *"bread and butter"* [P2], implying that it is a commonly seen condition within their discipline which *"we find easy to discuss"* [P2]. Medical genetic services provided for patients with these common genetic conditions

extends to genetic testing to confirm the diagnosis, as described by this participant when discussing genetic testing for trisomy 21/Down syndrome and DMD:

> *"Yes, we do all our testing. We do genetic testing and genetic counselling altogether. Then we definitely have confirmation of our diagnosis."* ***[P8]***

This availability of knowledge and expertise in the context of these common genetic conditions, combined with genetic confirmation of a diagnosis, translates into a holistic approach to care and management for the whole family. This is highlighted by a medical specialist who described a tailored management programme for DMD to screen and provide surveillance for at-risk family members as well as the identification of carriers for informed decision-making for future pregnancies:

> *"We screen other boy siblings, and then we also screen the females and the mother. Then for example, the females know whether they are carriers and then what to expect in their next pregnancies. And the boys, you might find they are still young, but they carry the gene. And then you'll know in future they will be affected".* ***[P8]***

Within the Department of Internal Medicine, this availability of basic skills, knowledge and expertise and the provision of basic medical genetic services was described by a medical specialist in the context of the most common genetic condition seen within their service, polycystic kidney disease. This participant explained how patients diagnosed with this genetic condition receive basic medical genetic services in terms of limited genetic counselling on inheritance patterns and risk to offspring, and ultrasound screening and surveillance for children of affected individuals:

> *"I do explain to that patient that you have a genetic illness, you most likely got it from one of your parents and you also have a 50% chance of passing it on to every child". ... "So in a sense, I do a limited form of genetic counselling myself for that small group of patients". … "And because we can identify the phenotype, if I diagnose polycystic kidney disease and someone tells me they've got children, then I would actually ask them to bring in their children. And I screen their children with an ultrasound, but not with a genetic test".* ***[P5]***

Subtheme 2: "Here we are not": Skills and expertise available but lack of resources and capacity in the province translates into substandard level of care

Within the discipline of obstetrics and gynaecology, a key area where medical genetic services are indicated is for the diagnosis, management and prevention of foetal anomalies, through genetic amniocentesis and advanced foetal anomaly ultrasound scans. In addition, due to the increased risk of aneuploidies with advanced maternal age, the NDoH maternity and antenatal care guidelines stipulate that all pregnant women 37 years or older should receive a foetal anomaly scan at 18–20 weeks and be offered the option of a genetic amniocentesis for aneuploidies accompanied by proper genetic counselling.

The medical specialists interviewed as part of this study have the required skills, knowledge and expertise to do genetic amniocentesis procedures *"I can do an amniocentesis"* said **[P7].** *"I used to do amnios every day as a training doctor"* said P6 *"But here we are not"*.

As this participant highlights, despite having the required skills, medical specialists do not routinely perform these procedures. According to other participants this is due to a lack of resources and a lack of standardised protocols:

> *"Let me tell you, there is not even that gauge needle [in Pietersburg Hospital Campus] for doing amniocentesis". **[P4]***

> *"It's not practice here to routinely test and offer genetic testing for women [of advanced maternal age]." **[P9]***

Similarly, while basic ultrasound services are available at various district and regional hospitals in the province, these are not routinely done for pregnant women of advanced maternal age as no formal screening programme has been implemented in the province, as evident from this interaction with one OBGYN specialist:

> ***"****No, sadly not. We're working hard to try to change that. But at the moment there is no formal program of scanning or looking for chromosomal abnormalities in pregnancies. It just does not exist."* ***[P9]***

Currently, ultrasound scans are only conducted based on medical indication for patients with a poor obstetric history, a small for gestational age foetus, twins or absent foetal movement. This was highlighted by one medical specialists who explained how if any abnormalities, such as an abnormal heart or abnormal brain, are identified via basic ultrasound, patients are then referred to the high-risk clinic at the Pietersburg Hospital Campus for a more advanced foetal anomaly scan, where the patients are then managed and referred to Gauteng:

> *"Ultrasound generally is per indication within the public domain, currently". … "when they see an abnormal heart, an abnormal brain, they then send to us in Polokwane and then we manage the patient further and communicate with Steve Biko and send the patients out that side."* ***[P6]***

However, when it came to the treatment and management of less common and more complicated genetic conditions and the provision of more specialised medical genetic services, medical specialists reported a number of challenges faced, such as a lack of in-depth knowledge and understanding, a lack of available resources and services and a lack of capacity.

Subtheme 3: "It's complicated": Lack of in-depth knowledge and understanding

Medical genetics is a complex and specialised field that requires specialised education, training and in-depth knowledge and understanding in order to properly diagnose, manage and counsel patients and families. Most of the medical specialists interviewed felt that they lacked the appropriate knowledge and expertise to provide proper genetic information and counselling to patients because, as P6 said: *"this is not our area of expertise".* Another participant described how this limited genetics knowledge and understanding makes it difficult for doctors to explain genetics and modes of inheritance to patients:

> *"You know genetics, even doctors don't understand it very much. It is a complicated thing to explain what are genes, what is inheritance, what mode is dominance, what is recessive".* ***[P4]***

Similarly, even the medical specialists with basic genetics knowledge and expertise felt that less common and more complicated genetic conditions require a more in-depth knowledge and understanding to be able to provide comprehensive genetic counselling for patients and families. One medical specialists from PCH explained how this gap in knowledge and expertise leaves them feeling overwhelmed and unprepared to provide comprehensive counselling for parents of patients with more complicated and less common conditions:

> *"But then if it becomes like a complex condition, then it becomes a problem. You have to do a whole lot of work. You have to read and obviously, I'm going to read now about genetic counselling and do it in the next 30 minutes, obviously I'm just browsing through. I'm not going to be fully prepared for that counselling session".* ***[P2]***

This gap in knowledge and expertise was also described in the context of diagnosing and managing congenital anomalies in the province. This lack of knowledge, understanding and expertise was described by participants from PCH in the context of rare genetic syndromes and not being able to diagnose and manage these conditions:

> *"So many of those [dysmorphic babies] that I, personally, can't tie the condition to say 'most likely it could be this syndrome'".* ***[P2]***

> *"It is those patients with the rare syndromes that we don't have clear experience and understanding. And we actually don't even understand the mode of inheritance or anything like that."* ***[P8]***

Similarly, when babies are born with congenital abnormalities, participants from OBGYN described not having the knowledge and expertise to further manage these patients or to

identify which genetic tests to conduct based on phenotype in order to provide answers for the mother:

> *"We deliver abnormal babies, we don't even know what to do with them. The mother needs to know what is wrong with this baby. What tests should be done?"* ***[P6]***

This participant further explained how overwhelming the area of genetic testing is for medical specialists because of the constant change in available testing options and techniques and how challenging it is for them to keep up-to-date with this ever changing field:

> *"There's just continuous change in terms of the tests that are conducted in the [genetics] laboratories. ... There's just so much development in terms of testing and everything"* ***[P6].***

Subtheme 4: "We just don't have the time": Provision of appropriate patient care locally is further hindered by lack of capacity, resources and services

In addition to the self-reported lack of in-depth knowledge and understanding of genetic conditions creating a barrier to comprehensive care for patients in the province, medical specialists highlighted how the lack of local capacity, resources and services contributes to this gap in the provision of appropriate local patient care.

Most medical specialists described how their heavy workload and busy schedules meant that they did not have the time or capacity to dedicate to comprehensive genetic information giving and counselling for patients and their caregivers:

> *"We just do not have the time to sit and go through a three-generation genetic tree and explain in detail to each parent exactly how a condition is transmitted."* ***[P10]***

> *"As I said, clinicians don't have time for that, you know. It's very difficult. We see a new patient with cancer every day. There is no time, really. They are busy operating*

and are not able to sit down and relax and give really in-depth time to the patient and his family." ***[P11]***

Many participants reported that a lack of available services prevented them from providing appropriate care to their patients because there is no one to refer patients to. A participant from OBGYN spoke about how *"the services are non-existent here"* ***[P9]***, which results in a lack of locally available options when caring for a patient who would clearly benefit from medical genetic services:

"You have a patient who you definitely think this one would benefit from a geneticist, but there's none here." ***[P9]***

Participants from OBGYN highlighted another gap in available resources and services related to postnatal investigations of stillbirths and neonatal death. One participant, who had previously worked in Gauteng, was particularly frustrated by a lack of laboratory services to test for a possible genetic cause for stillbirths in Limpopo. This participant's very positive experience of the support received from the local laboratory in Pretoria was contrasted with their experience in Limpopo, where *"Nothing will be done. No tests will be conducted, no counselling for the mother and the family…* [because]…*you can't send a dead baby to another province"* **[P6]**.

The burden that these deficiencies in services can place on patients was highlighted by a specific case mentioned by a participant from OBGYN of a patient *"having recurrent abnormal babies. She was delivering babies with abnormal hands, abnormal legs, very short legs, very small heads."* ***[P6]***. The participant went on to explain how this patient had two previous stillbirths where *"there were no genetic tests conducted on those babies. So you can imagine what a loss she has suffered, and now she's pregnant again"* **[P6].** This specific patient was subsequently referred for comprehensive medical genetic services in Pretoria. However, this case highlights the detrimental consequences that these gaps in services can have for the affected patients, the wider community and the doctors involved:

> *"Those babies were just given birth to and buried, nothing being done. So you can imagine what has been happening. It's unfair to the community, unfair to the patients. It's only now that I'm here, I'm picking these up and I'm trying to correct the whole situation."* ***[P6]***

In addition to the lack of local services, two participants mentioned that, in the public healthcare system, certain tests can only be accessed by specific specialities, which *"becomes a bit of a challenge in that you get limited into some the type of tests you can do sometimes."* ***[P3]***, creating another barrier to providing genetic testing for patients in the absence of locally available medical genetic services. A participant from PCH mentioned cases of dysmorphic children where they were fairly certain of a specific diagnosis but were denied access to genetic testing services to confirm the diagnosis, despite having successfully accessed the same test previously. This creates the added burden for the patient of having to access out-of-province medical genetic services in order to get permission for the genetic test to be conducted to confirm the suspected diagnosis:

> *"We will see dysmorphic children and some of them, we might be fairly confident about the pattern, a good example has been Noonan syndrome. But we've had a variable response where we're told that we can't access the Noonan panel that I know they've got at NHLS because I've accessed it at least once successfully and it can only be accessed through the medical geneticist. So that then puts you in a bit of a catch 22. Because your patient has to go 500 kilometres to get permission to have a test that you're pretty sure they qualified for anyway. So those kind of rules are a little bit difficult."* ***[P10]***

Subtheme 5: "I really need to know that it's going to add substantial value": High threshold for out-of-province referrals

Due to the lack of locally available medical genetic services, medical specialists can refer patients to services. The centres most commonly utilised were those around Pretoria, such as the services at the Dr. George Mukhari Academic Hospital and the Steve Biko Academic Hospital, and, less commonly, the services at the Charlotte Maxeke Academic

Hospital in Johannesburg. However, medical specialists reported being very selective in referring patients to out-of-province services due to the immense financial, social and emotional burden this places on the patients and their families, as well as on the colleagues and services in Gauteng.

The George Mukhari Academic Hospital situated in Ga-Rankuwa, approximately 30 kilometres from the Pretoria city centre, near the border between the Gauteng and North West provinces. This public hospital is approximately 280 kilometres away from the Pietersburg Hospital Campus in Polokwane, Limpopo. Because of the distance that patients and families would need to travel in order to access medical genetic services, and the logistics that such a journey involves, participants felt that such referrals needed to add substantial value to the treatment and management of the patient. This was highlighted by one participant from PCH:

> *"For me, before I send a patient on a 250 kilometre, well 500 kilometre return journey, I really need to know that it's going to add substantial value. That's a big social disruption."* ***[P10]***

This same participant went on to explain that, in their experience, once patients and families are sent to out-of-province services, they remain within that service for all future appointments and follow-ups, resulting in further financial and emotional strain being placed on the patient and families. This participant added that, even though sending patients out-of-province may result in a diagnosis, if it will not change the treatment and management of the patient then there is no value in or benefit to referring patients:

> *"I know once they've done it, they're going to get locked into a sequence of trips. … So that makes me think three times about sending them. I have to ask myself, 'yes, we might get a diagnosis. How will that change the care of this particular patient?' And if I can't see a clear benefit, then I'll say rather I'm not sending them."* ***[P10]***

In Limpopo, patient transport between tertiary healthcare institutions is a government-funded service. While no direct travel-related costs are the responsibility of patients and

families, costs associated with the logistics of having to travel to another province are incurred by individuals, which places a heavy social burden on individuals.

> *"The nearest one [service] is several kilometres away and our patients, some of them are indigent, so they do not have the means to access those via their own means."* ***[P9]***

As one participant from PCH shared, out-of-province referrals often result in patients having to sleep at the local hospitals to be on time for the out-of-province planned patient transport, resulting in them being away from home for up to three days:

> *"It's tiring because they have to sleep [at the local hospital] and go in the morning come back and sleep in the hospital. So, they'll be away from home for an average of three days. The mother is working. They don't have the time. They will opt not to follow up.* ***[P8]***

Another reason provided for this high threshold for referring patients was the emotional burden out-of-province referrals places on patients and families. A number of participants felt that the benefit of accessing medical genetic services did not justify the added trauma that these patients would need to endure. For example, within OBGYN, patients with unexplained foetal losses who would benefit from medical genetic services are not referred because of the immense emotional trauma the patient is already experiencing. One participant felt that it is unfair to subject these patients to the added trauma of having to travel while grieving:

> *"That I've seen mostly are the patient with unexplained foetal losses. I don't send them because its far, and the patients are in mourning, they are grieving. It's just my own thinking that I thought I shouldn't subject them to more trauma of travelling. I just leave it."* ***[P4]***

A number of medical specialists were also cognisant of the added workload for colleagues in Gauteng associated with out-of-province referrals. As one participant from OBGYN

explained, Gauteng colleagues have their own population to serve, which makes them think twice before sending patients to out-of-province medical genetic services:

> *"I think about the workload of our colleagues in Gauteng. They probably have a lot of patients coming and knocking at their doors. … They are not only servicing us, they also service other huge areas."* ***[P7]***

Subtheme 6: "It takes long": Challenges of out-of-province referrals

Despite this high threshold for referring patients to out-of-province medical genetic services, participants did report that some patients are referred. However, this too comes with certain challenges. One such challenge reported by participants is the long waiting list and how it can take up to three months to get an initial appointment, as highlighted by this participant:

> *"It's quite a challenge because the first appointment might be in three months from now and then follow up."* ***[P8]***

Within the context of OBGYN, this delay in accessing required services can have serious consequences in terms of human rights and the Choice on Termination of Pregnancy Act 92 of 1996. This Act stipulates that termination of pregnancy can be offered to pregnant women from the 13th week of gestation up to and including the 20th week for severe foetal anomalies. One participant explained how, even if a women presents to the high-risk clinic at 18 weeks, the delays experienced with accessing medical genetic services means that patients miss the 20 week cut-off for offering termination of pregnancy. This participant went on to explain that patients need to be picked up and referred by 12 to 14 weeks, at least, in order to be afforded the choice to terminate if a foetal anomaly is confirmed:

> *"In my experience, it takes long. If your patient presents at 18 weeks, you know that even if they [specialists in Gauteng] say that a disease is there, you won't be able to terminate because it will be well over 20 weeks. And with patients usually booking late. You should send them at least at about 12 to 14 weeks for you to be able to take a decision. Because it takes about 6 weeks."* ***[P4]***

Despite the burden that out-of-province referrals place on patients and their families, and on the services in Gauteng, the participants did report that colleagues in Gauteng were very helpful and accommodating, and that patients and families do come back with a better understanding and more realistic expectations. A participant from PCH spoke positively about their experience of communicating with colleagues from the out-of-province medical genetic services and how helpful colleagues were:

> *"I would say it's a challenge, but when you call they are very welcoming. I won't say anything about them that they don't want us to call because when you pick up the phone and call they are very helpful, they are very welcoming."* ***[P2]***

This same participant also spoke about the benefit to the patient; how their understanding of the condition is improved by access to comprehensive counselling as well as management of expectations for what the future may hold:

> *"When they come back they do have a better understanding. Like we say, if you've counselled them fully you increase the understanding of what their child has, their expectations, things that they should look out for. So, when they come back they are happy."* ***[P2]***

Subtheme 7: " We know the truth. We should be doing the right thing": Lack of services leaves medical specialists feeling isolated and frustrated

The unavailability of necessary resources, capacity and services to provide patients with appropriate care leaves medical specialists feeling isolated and frustrated.

Some participants felt frustrated about not being able to treat and manage their patients holistically themselves as, once patients are referred for out-of-province medical genetic services, they get locked into that service and usually do not get referred back to the local services. A participant from PCH expressed frustration with the lack of collaboration and interaction between the different services, resulting in the families having to travel extensively between the two provinces and services:

"It is quite vexing because they get a first appointment and then they tend to have a sequence of follow-up appointments and the interface between our service and that service is limited to us doing investigations to clarify cardiac involvement, renal involvement, those kinds of things. So mostly imaging investigations that will be requested of us. But we don't really get given down referred responsibility for counselling or genetic support. And the patient, they can sometimes end up going up and down oh 5, 6, 7 or more times. It's a lot." ***[P10]***

Participants from OBGYN expressed frustration about the lack of postnatal investigations for stillbirths and neonatal deaths in the province. Currently, the lack of local services means that tests are not conducted in order to provide answers for the families and appropriate postnatal counselling:

"So now, because there are no services at all, we just throw away those precious placentas, which are probably holding answers to the questions which we say unexplained." ***[P7]***

"Babies born with abnormalities here, the baby will be just sent to the mortuary. Nothing will be done. No tests will be conducted, no counselling to the mother and the family. … We just send them home and they keep on losing babies and nothing is being done. … You can't send a dead baby to another province" ***[P6]***

One particular participant from OBGYN expressed intense frustration about the lack of available services in the province and the resultant injustice due to the below minimum standard of care provided to patients in the province:

"It's very minimal services. It's terrible. It's very bad. … it's unfair to the community, it's unfair to the patients. … I'm sorry for the overload. I'm just frustrated with what we're experiencing here. Its below standard of care what's being offered to the patients. And the patients, they do not know, that's the thing. They do not know what's supposed to be done, you know. … It's very wrong of us isn't it. We know the truth. We should be doing the right thing." ***[P6]***

The lack of locally available medical genetic services poses a challenge for both medical specialists and their patients in the provision of the appropriate level of care, highlighting a need for a dedicated service in the province.

3.2.2. Theme 2: “It increases the quality of service that you offer”: The need for and benefit of a dedicated service

This theme provides insights into the opinions of medical specialists in terms of the need for and benefit of a locally available medical genetics service in the province. It describes the large population in Limpopo who are serviced by the public healthcare system and the need for the knowledge, skills and expertise of a dedicated medical genetics team to assist in providing comprehensive care to the patients and families, as well as for registrar training in the province. An increase in the quality of the services provided and inclusion of appropriate screening and surveillance programmes could have a positive effect on the economic impact of genetic conditions in the province.

Subtheme 1: “There is an extreme need”

When discussing the perceived need for and benefit of comprehensive medical genetic services in Limpopo, participants spoke about the large population in the province serviced by the public healthcare system who would benefit from locally available services. Participants stated that *“the province is big and we see quite huge volumes of patients.”* ***[P7]*** and that *“there’s enough people to justify a dedicated service here”* ***[P5].*** In fact, one participant from PCH felt that Limpopo could be the centre for medical genetics for the whole country, based on the number of genetic conditions and syndromes seen within this province compared to other provinces:

> *“Limpopo can be the hub of genetics for the whole country. I've only been to Gauteng where I did my training, but I haven't seen as many syndromes or genetic problems that I'm seeing now here”.* ***[P2]***

A high prevalence of congenital anomalies and pregnant woman of advanced maternal age seen in the province was highlighted by participants from OBGYN and the need for proper prenatal screening for aneuploidy risk for this cohort of patients locally:

> *"Advanced maternal age, there's lots of more than 40 year olds who are falling pregnant. Certainly those [patients] would need genetic services".* ***[P9]***

Outside of PCH and OBGYN, medical specialists from SG reported a large number of cancer patients in the province who would benefit from comprehensive medical genetics services and how access to these services could assist medical specialists in this discipline to provide genetic answers for cases with a strong family history:

> *"We see an enormous amount of cancer; colon cancer, gastric cancer, breast cancer, thyroid cancer. It will really be a great help I think if geneticists would come on board and discuss these cases. Because, one of the typical things, especially in breast cancers, is how many people in your family have it. I remember last week we had an exam, I think she had four first degree relatives with breast cancer. So for instance, you could have tested for the various genes there."* ***[P11]***

There is, therefore, a clear sense of the need for comprehensive medical genetic services among health professionals working in Limpopo in order to provide the appropriate level of care to patients locally.

Subtheme 2: "Someone with experience"

Discussions with the participants revealed a strong sense that having access to the specialised skills, knowledge and expertise of medical genetics professionals would provide the support and guidance needed to provide patients with the proper level of care in the province. For example, the NDoH guidelines for maternity care in South Africa, stipulate that all pregnant woman 37 years and older should be informed about their increased risk for aneuploidies and be referred to a genetics clinic to be offered genetic counselling, a foetal anomaly scan and genetic amniocentesis. However, these guidelines are not always adhered to currently within the province. Participants from OBGYN felt

that, in order to meet this minimally accepted national standard of care for patients in Limpopo, the support and guidance from an experienced medical genetics team would be required:

> *"Those services need someone with experience. They need someone who's able to counsel them and tell them about the risks, so that we are able to then consent them for testing. … Such counselling needs time, it needs a specialised service".* ***[P6]***

One participant from OBGYN who would like to establish an aneuploidy screening and foetal anomaly clinic at the Pietersburg Hospital Campus, highlighted how such a clinic would still require support and guidance from medical genetic services:

> *I will be establishing an aneuploidy screening and foetal anomaly clinic. So we will definitely be consulting a lot for genetic counselling and prognosticating such patients.* ***[PX]*** [3]

Participants from OBGYN also felt that a medical genetics professional would be required to provide guidance and assistance in the identification and diagnosis of congenital abnormalities and to assist with decision making and termination of pregnancy options. One participant felt that the knowledge and expertise of medical genetic professionals would be invaluable in providing the proper level of care in cases where a congenital abnormality is diagnosed through genetic amniocentesis:

> *"To prognosticate. To tell us must we advise termination, or must we allow the mother to then continue with the pregnancy. Such advice is helpful if someone knows the genetic information around that condition that is picked up during an amniocentesis, for instance. I think the role is very vital for us in foetal medicine".* ***[P6]***

[3] Participant number excluded to maintain anonymity.

Within PCH, participants described the unmet need for the skills, knowledge and expertise of medical genetics professionals in providing assistance with the diagnosis of syndromic babies treated and managed within their service, which could translate into a better level of care for these patients:

> *"So within our discipline it's something that is of vital importance to have, because we've got a whole lot of dysmorphic babies who we may not even know what syndromes they have".* ***...*** *"Having a geneticist, someone with a better knowledge of investigating, analysing, assessing those patients and even counselling them would be a better team member to have in our field [paediatrics].* ***[P2]***

The expertise within a medical genetics team would also benefit the medical specialists from a continuing education and training perspective. Participants felt that such interactions would provide opportunities for exposure and learning, translating into a better and more comprehensive service being provided in the province:

> *"You learn a lot quicker if you have somebody who is very familiar with the system at your side, just walking you through it a couple of times, a dozen times, maybe. It's much easier than trying to look it up or talk on a phone".* ***[P10]***

Having these medical genetics professionals as part of a multi-disciplinary team to provide guidance and assistance to the medical specialists locally would translate into a more holistic approach to patient management and care and an increase in professional satisfaction by being *"able to offer 'world-class medicine'. The type of service that a woman gets in Cape Town. I'd love for them to get it in the public sector here in Limpopo. And I don't think we're asking for a lot. It's just basic things.* ***[P9]***

Subtheme 3: "It's a watershed moment": Managing a genetic diagnosis

Medical specialists also highlighted benefits to patients and their families that locally available comprehensive medical genetics services would provide. In light of the high-threshold for referring patients to out-of-province services, as described in Theme 1, one

clear benefit of having services available locally is that more patients would be referred to these services, as explained by P4:

> *"The willingness to refer will be more than if it's far, because there are no logistics to consider".* ***[P4]***

It would also reduce the burden that having to access out-of-province services places on patients in terms of reducing the waiting time and travelling costs of the patients, which would alleviate some of stress and anxiety experienced by these patients:

> *"It [local services] will reduce the waiting time, the travelling costs as well as anxiety. The moment the patients get anxious, they probably might dip into depression. That is one of the biggest concerns."* ***[P7]***

This is beneficial not only for the patient but also for the rest of the family. Locally available medical genetics services could assist to identify at-risk individuals and family members during comprehensive counselling sessions, and could translate into appropriate surveillance and preventative measures for genetic conditions. This was highlighted by a participant from SG who saw a benefit for comprehensive medical genetic services in terms of identifying and responding to individuals at risk for developing hereditary or familial cancers:

> *"Tremendous [benefit]. And also not only the patient, but also the family, because I'm pretty sure you can trace a lot of other vulnerable people in that family.* ***…*** *Mainly I think for the other people in the family who can be tested and then they can be put on preventative programs".* ***[P11]***

According to participants, medical genetics professionals have a better understanding of genetic conditions and specialised counselling skills, which would mean patients and their families would have a better understanding of their specific condition and, thus, higher levels of patient satisfaction:

> *"I think the genetic counsellor will definitely go an extra mile and go in depth, elaborate and educate these patients. And I believe patient satisfaction will be more*

*than when the patient has been counselled by me compared with the genetic counsellor, because the genetic counsellor should have a lot of information at his or her fingertips". **[P7]***

Medical genetics professionals can address the psychosocial needs and well-being of patients as, with their specialised knowledge, skills and expertise, they could provide proper support and counselling during a time that is often very stressful for patients and families:

*"It can be a bit of a watershed moment and you probably need somebody to take you through that because I think there's potential to trigger you into very negative ways of thinking about what that means. And it's not something you can do in two minutes." **[P10]***

Furthermore, by attending to the psychosocial aspects, medical genetics professionals can support patient autonomy and informed decision-making. Within the discipline of OBGYN, this benefit was specifically mentioned in the context of antenatal care and screening for aneuploidies. One participant described how access to medical genetic services locally could lead to abnormalities being detected early enough so that patients could make informed decisions on whether they wanted to continue with a pregnancy or not, and plan accordingly:

*"Our patients will benefit because that [access to local services] will reduce the time to know the chances that my child would be born with this condition. And that will help them plan going forward with a pregnancy, whether they want to continue or not continue. Unlike to be told at an advanced gestational age that your baby has got this condition and we can't treat it. And then that becomes a problem because now you've missed the period for termination of pregnancy. You're going to sit with the child that is handicapped whilst you could've done a TOP [termination of pregnancy] earlier. Or even if you don't want to do a TOP, you can have processes in place and then you start planning accordingly. And then I think that that will reduce the levels of anxiety for more patients". **[P7]***

This patient autonomy and informed decision-making extends to understanding recurrence risks associated with specific conditions and the risk to future pregnancies. Participants felt that having locally available comprehensive counselling expertise would benefit patients and families in terms of being able to make informed decisions about future pregnancies when equipped with proper knowledge and understanding of inheritance patterns and risk of recurrence:

> *"I see you guys as specialists explaining more in detail and also giving more facts than we do in terms of her risk of having another Down syndrome baby".* ***[P3]***

Subtheme 4: "Training our doctors": Enhanced education and training of registrars

In addition to the perceived need for and benefit to patients and appropriate patient care that locally available medical genetic services would provide, participants saw an unmet need for the education and training of registrars within the departments of PCH and OBGYN that could be bridged through the availability of local medical genetics skills, knowledge and expertise. For one participant from PCH, the presence of medical genetics professionals would enhance the education and training of registrars as they would be able to support registrars in identifying cases and providing basic counselling to patients and families:

> *"The other thing that would be different when we've got them [medical genetic services] here is the training for the registrars. When they're trained better they can even do a better job with the counselling themselves. … It will also help our registrars to pick up those cases, and know that they've got someone who can carry them through and with a better understanding".* ***[P2]***

A participant from OBGYN identified a need for specialised services in the province in terms HPCSA accreditation for the training of doctors in the province. This participant highlighted the possibility that the University of Limpopo could lose accreditation for the specialist training of registrars if registrars are not exposed to and trained in the basic procedures within the specific specialities scope of practice, such as being able to conduct foetal anomaly scans and genetic amniocentesis:

"Unfortunately, it [amniocentesis] is not currently done at all. …. We need to start. We are behind. And the HPCSA might not allow us to continue training doctors if we are unable to do such basic testing. As a university, they won't allow us to train doctors if we are unable to do such tests as per recommendation by medical experts". ***[P6]***

Subtheme 5: "We are paying a big economic price for not having it": Social and economic impact

Finally, some participants felt that locally available medical genetics services would ultimately provide a financial benefit to the LDoH in terms of identifying at-risk individuals and implementing screening and surveillance programmes and informed decision-making about future pregnancies.

A participant from PCH spoke specifically about conditions that are very expensive to treat, such as haemophilia, or conditions with an immense social expense, such as DMD. This participant felt that the absence of proper family history taking and comprehensive genetic counselling about the risk of reoccurrence and informed decision-making resulted in missed opportunities for upstream interventions, which could be avoided if the appropriate medical genetic services were available:

"We just do not have the time to sit and go through a three-generation genetic tree and explain in detail to each parent exactly how a condition is transmitted. And that means, without the genetic counsellor, we are going to miss opportunities for upstream intervention of conditions that can be really, really expensive to treat like haemophilia. Even if you think about social expense; Duchenne muscular dystrophy, many of them. We are paying a big economic price for not having it." ***[P10]***

This participant went on to explain how having a genetic diagnosis, and knowledge of carrier status and inheritance patterns could empower patients and families to make informed decisions about future pregnancies and the associated risks, which could potentially have a positive effect on health spending for genetic conditions:

> *"It empowers them to make decisions that could have huge downstream consequences in terms of health spending. We can't prescribe that to people. We can only give them the tools and then see what happens. But, I'm thinking that given those tools, the potential downstream effects are massive."* ***[P10]***

Similarly, within the context of OBGYN, participants felt that proper antenatal care and services could translate into financial benefit to the LDoH by reducing the costs associated with the treatment and management of babies born with congenital abnormalities and syndromes, such as trisomy 21/Down syndrome:

> *"Prevention is better than cure as they say. So, if you could prevent some of these by doing the necessary antenatally, you are saving the department in the future."* ***[P9]***

While there is both a perceived need and a benefit for locally available comprehensive medical genetic services, it is also clear that Limpopo is severely under-resourced and lacking in critical capacity. In light of this, we need to think very carefully and critically about how to bring medical genetic services to this province and provide patients access to these services.

3.2.3. Theme 3: "The demand side is clear. The issue is the affordability side and that's the problem": Strategies for providing medical genetic services in an under-resourced province

This theme discusses strategies described by participants to provide medical genetic services in Limpopo. These strategies described by the participants included providing discipline-specific genetic education and training to the medical specialists to address self-reported genetics knowledge deficiencies; providing support and guidance through interinstitutional telegenetics collaborations; establishing a core critical mass of genetic counsellors in the province; using research projects as a cost-effective way to provide genetic testing for patients; and training healthcare professionals to extend the reach of the medical genetic team.

Subtheme 1: "This is one area we don't have a lot of information about": Education and training

Both the survey questionnaire and the one-on-one interviews revealed deficiencies in genetic knowledge and understanding, which medical specialists felt could be addressed through the provision of discipline-specific education and training. A platform identified for this are the weekly administrative meetings that medical specialists from each discipline attend. A starting point would be education and training on what services are available and how to access these services:

> *"Every Monday morning, we have an admin meeting. So, it's on platforms like that where you can just come in and maybe introduce what you do to us so that people can be aware and start to understand that there are things that can be done for the patients that they see. … So, it will be very nice to just have you guys maybe visit our departments and just tell us what you do and what we can also do to assist to our patients."* ***[P3]***

Further education and training could then be more discipline-specific and provide information on the genetic conditions and genetic tests relevant to the different departments:

> *"A workshop or maybe in-service training for all the departments, because this is one area that most of us, honestly speaking, don't really have a lot of information about."* ***[P3]***

> *"Even the testing itself. Training that doctors need so that the doctors know this is what needs to be ordered. … And updates as well, as to what's the latest on the market."* ***[P6]***

Subtheme 2: "What do I do, please help": Support and guidance

Another area identified by the medical specialists as a requirement for being able to provide medical genetics care to patients in the province was support and guidance from medical genetics professionals. Some felt that having medical genetics professionals as

part of multidisciplinary teams would assist them to screen and identify at-risk patients. For instance, this participant from SG described the value in this support and guidance for cancer patients:

> *"For instance, in our academic meeting, we could discuss once a week all the cancer cases and screen to see which ones can be taken further [offered genetic testing]."* ***[P11]***

Medical specialists also felt that having the ongoing support and guidance from medical genetics professionals to assist with the diagnosis of patients with complex genetic conditions would be very beneficial, as highlighted by this participant from PCH:

> *"Having a geneticist or someone with a better knowledge of investigating, analysing, assessing those [dysmorphic] patients and even counselling them would be a better team member to have in our field." [p2]*

Because there is currently no medical geneticist positions in Limpopo, patients have to access this speciality outside of the province. One way to address this lack of capacity locally would be through interinstitutional collaboration and telegenetics. A participant from PCH felt that collaboration and support, combined with the potential increased diagnostic power of new genetic testing techniques, could negate the need for out-of-province referrals for medical genetics consultations:

> *"I think that general paediatricians supported by a medical geneticist can actually get quite smart at getting to genetic answers. Especially if these new testing tools are as potentially powerful as I think they are going to turn out to be. If we get smarter to saying, 'okay, well let's do a panel for a dysmorphic child with developmental delay', that panel might give us an answer that negates the need to have a geneticist actually look at the child and guide us with that."* ***[P10]***

In support of interinstitutional telegenetics consultations, one participant from OBGYN described a case where they had to contact colleagues for advice. This highlights how beneficial such support and guidance from medical genetics colleagues can be in the absence of local capacity:

> *"I have had to phone Cape Town for advice on what to do. We've had two anencephaly babies, for example. So, the advice was 'this is a lethal condition, so go forth and offer termination of the pregnancy."* ***[P9]***

However, while the lack of capacity with regard to medical geneticists can be partially bridged through telegenetics and interinstitutional collaboration, support and guidance, this strategy cannot be applied to the lack of capacity for genetic counselling services due to the nature and content of these sessions:

> *"Genetics has got a person-to-person interface … you can't really cut down on dealing with people's intellectual and emotional assimilation of what they're hearing about their genetics."* ***[P10]***

So at a minimum, in order to provide comprehensive genetic counselling services to patients and families in the province, the centralised medical genetics team in the province needs to have genetic counsellors. Speaking from experience with specialist capacity in the province, a participant from PCH suggested starting with a core critical mass of genetic counsellors within the province who would then need to 'think out of the box' and find ways to increase throughput to be able to reach as many patients as possible without compromising the quality of genetic counselling provided:

> *"Preferably you want to start with a core. I think what we've seen with the specialties is if you start up with a core of three specialists, you're more likely to succeed than if you start with one, because one person fatigues, leaves, and you get interruptions in the service. So actually you need to start with a little bit of a critical mass. Let's just say three for now. And I think three people then, they would do what they are trained to do, genetic counsellors, and then they would ask themselves, 'how do we improve our throughput? How do we expand our reach, given that we are only three'."* ***[P10]***

Subtheme 3: "Set it up as a research project": Cost-effective genetic testing

Genetic testing can be very costly and, for an already under-resourced and overburdened province, the feasibility of being able to provide answers to all patients through the

implementation of genetic testing is unrealistic. A suggestion provided for a way to bridge this gap is to establish research projects for specific cohorts of patients in order to get genetic answers for patients, as well as to get a better understanding of the genetic variation within the province. This was highlighted by a participant from PCH when discussing provincial budgets and the feasibility of genetic testing:

> *"Probably not [budget for testing]. So I would see it that we would have to set it up as a research project most of the time."* ***[P10]***

Another participant from INTMED also highlighted the benefit of research in being able to provide information on genetic aetiology and variation in local populations:

> *"Research and academic aspect in terms of finding new mutations in our settings, but also just confirming known mutations, whether our patient has it or not."* ***[P5]***

Subtheme 4: "How do we expand our reach?": Training of healthcare professionals to extend the reach of the medical genetics team

One cost-effective strategy to increase throughput with only a small core critical mass of genetic counsellors would be to strengthen local capacity to assist the team of genetic counsellors. The participant from PCH suggested providing genetics education and training to psychologists in the province, for them to assist with providing the psychosocial aspect of genetic counselling services because these medical practitioners already have the skills and expertise required to manage psychological distress:

> *"In Limpopo for instance, psychologists are actually quite well off [in terms of staff per capita ratios]. So maybe you want to think about that. They've got tools for dealing with people in emotional distress. How about giving psychologists a bit of genetic training?"* ***[P10]***

This strengthening of local capacity was also highlighted by a participant from FMED, who felt that *"a lot can be done in primary health care"* ***[P3].*** This participant felt that, if the medical specialists in family medicine had the necessary knowledge and training as well

as access to genetic testing options, they could diagnose, treat and manage a number of patients at that level, rather than referring patients to an already overwhelmed and overburdened paediatrics service:

> *"We can actually start the process early. I can see the patient and if we need to I can discuss with the paediatrician, but be able to do the investigations and then we take it from there."* ***[P3]***

This participant also suggested that training of nurses in primary health care clinics to take a proper three-generation family history and to identify genetic 'red-flags' could potentially lead to more patients being identified and, thus, more patients receiving comprehensive care:

> *"If we have to strengthen the primary health care system, we need to start by imparting knowledge on the nurses. Nurses are doing deliveries in the clinics, they are seeing pregnant women."* ***[P3]***

The suggestion to upskill nurses to provide limited genetic counselling services was reiterated by the participant from PCH, who spoke about the nurse counsellors trained under Professor Venter who used to provide basic genetic counselling to patients and how this model should perhaps be revisited in order to strengthen genetic counselling capacity and services in the province:

> *"We had that nurse counsellor model that Professor Venter started here. Maybe one would revisit that, and look at lessons learned, tweak it, see what can be done."* ***[P10]***

This project, therefore, indicates that a model of providing cost effective medical genetic services in Limpopo could be to set up by a small team of genetic counsellors who could act as a hub for improving access to medical genetic services by:

- Providing a link between medical specialists in the province and medical genetics services in institutions in other provinces,

- Providing training to medical specialists to provide information on the genetic conditions and genetic tests relevant to the different departments,
- Providing training to other health care professionals to strengthen genetic counselling capacity in the province,
- Facilitating research by providing a link between health services in the province and academia.

4. DISCUSSION

In this study, 32 medical specialists completed an online Google Form survey questionnaire to gather information on the knowledge, opinions and practices of medical specialist and whether medical specialists in Limpopo refer patients to medical genetic services. Medical specialists recruited for this study were from FMED, INTMED, OBGYN, PCH, and SG working at the PMHC. Eleven medical specialists were subsequently interviewed in order to strengthen the findings from the survey questionnaire and to gain a more in-depth understanding of how specialists in Limpopo view medical genetic services in their context. This chapter discusses the findings of this study related to the main research question: “How do medical specialists at the tertiary hospitals in Limpopo view medical genetic services?” Recommendations for implementation of medical genetics services in Limpopo are also described in this chapter, as well as the limitations of the study and further research recommendations.

4.1. Sample Demographics

Sample demographics from the medical specialists that responded to the survey questionnaire indicated diversity in terms of the age of participants, years since completion of undergraduate training, years since completing specialists training and years in specialist practice.

Of the 56 eligible participants from the five departments targeted, a total of 32 responses were received, giving an overall response rate of 57% (n=32/56). In general, surveys aimed at physicians have a much lower response rate than the general population because of their heavy work schedules and scarcity of time (Hunter et al., 1998; Flanigan, McFarlane & Cook, 2008). In spite of this, the overall response rate for the current study is considerably higher than response rates reported by other researchers for physician surveys (Hunter et al., 1998; Chambers et al., 2015; Cunningham et al., 2015; Cook et al., 2016). Participant recruitment in the current study was enhanced through the use of a combination of recruitment methods, such as the involvement of the HoDs of each

department to facilitate the recruitment process and the use of personalised correspondence in the form of personalised email invitations and telephonic methods.

PCH had the lowest response rate of 38% (n=6/16); however, this was the only department where telephone numbers were not included in the contact details list and as such, participant recruitment relied entirely on emails and reminders. The response rate for PCH is, however, still within the anticipated range for surveys aimed at physicians (Hunter et al., 1998).

A systematic review of methodologies for the improvement of response rates in surveys of physicians highlighted the effectiveness of telephonic surveys as a method to increase physician response rates. This could account for the high response rates from SG (85%; n=6/7), FMED (80%; n=4/5) and OBGYN (63%; n=12/19). In addition, VanGeest, Johnson & Welch (2007) described a link between the perceived value of a study and physician response rates which could account for the low response rate received from INTMED (44%; n=4/9). Alternatively, this could be due to the heavy work schedules and lack of time that often results in low response rates for surveys aimed at physicians (Flanigan, McFarlane & Cook, 2008).

The majority of participants (n=13/32; 41%) were in the 41–50 years of age range, with the majority (n=18/32; 56%) having completed undergraduate medical training in the decade between 2001 and 2011, and specialist training after 2012. Overall, 72% (n=23/32) of the respondents were male and 28% (n=9/32) were female. Although Tiwari et al. (2021), reported that 59% (n=27 387/46 420) of the total of 46 420 medical doctors registered with the HPCSA in 2019 were male and 41% (n=19 033/46 420) were female, a report from the Health Systems Trust indicates that in Limpopo there were 158 male specialists (72%; n=158/220) and 62 female specialists (28%; n=62/220) (Massyn, Pillay and Padarath, 2019). According to this available data, the study sample is representative of the population of medical specialists in Limpopo.

4.2. Genetics Education and Knowledge

4.2.1. Genetics education

The majority of medical specialists reported receiving limited genetics education during their undergraduate training. This is consistent with the GenTEE report published in 2013 which highlighted a lack of integration of medical genetics into undergraduate curricula in South Africa (Nippert et al., 2013). Limited medical genetics education was reported by medical specialists who completed their undergraduate training at the four medical schools in South Africa which had medical genetics professionals on their staff who reported integration of medical genetics into the undergraduate curricula (Nippert et al., 2013). There were inconsistencies in the reported amount of genetics education received that suggests unreliability of the information gathered on genetics education during undergraduate training. According to the National Cancer Institute's Dictionary of Cancer Terms, recall bias is most likely to occur when an event has occurred a long time ago (National Cancer Institute, 2023). Most medical specialists reported completing undergraduate training more than 10 years ago. A study by Burke et al. (2006) also reported a difference in recall of the amount of time devoted to genetics topics during undergraduate training of doctors in the United Kingdom (UK).

The results obtained on the medical genetics education received during specialist training revealed that almost 41% (n=13/32) of participants recalled receiving no genetics education, with the vast majority being from FMED, INTMED and SG falling into this category. This is consistent with a study by Burke et al. (2006) in which 46% of non-geneticist medical trainees from two regions in England (West Midlands and South Western) reported no genetics education scheduled in their specialists training. Following this research, Burke et al. (2006) urged medical curriculum planners globally to integrate genetic education into all years of medical school as well as specialist training programmes specifically for primary care physicians, such as family physicians, internists, obstetricians and gynaecologists, and paediatricians (Burke et al., 2006). Within the South African context, this plea for inclusion of genetics education into medical education

has been made since 1962, voiced by Prof PV Tobias (Christianson, 1997) and echoed by experts in the field over the past few decades (Jenkins, 1990; Christianson, 1997). However, results from this current study highlight that this call to action may not have been adequately met yet within these specialities.

In addition to the specialist reported lack of genetics education during undergraduate and specialist training, in the current study, only 28% (n=9/32) of participants reported continuing medical education that included genetics; with most of these participants (n=4/9) being from OBGYN. This lack of genetics education raises considerable concern with regard to non-geneticist physicians' ability to manage, diagnose and provide comprehensive care for patients with genetic conditions.

4.2.2. Genetics knowledge

With the continuous advancement in genetics and related technologies, the National Coalition for Health Professional Education in Genetics (NCHPEG) devised minimum core competencies that all health-care professionals must have in order to meet the growing need for specialised healthcare services for patients and families with genetic conditions. These core competencies include (but are not limited to) having a basic understanding of genetics and inheritance patterns; understanding the importance of a three-generational family history; understanding the physical and psychosocial benefits and limitations of genetic information; the ability to identify and refer patients and families who may benefit from genetic services; and ensuring that consent for genetic testing is informed (Jenkins et al., 2001).

In keeping with the lack of genetics education reported by participants in this current study, the majority of medical specialists (44%, n=14/32) self-reported their genetics knowledge as fair (on a scale of poor, fair, good and very good). A study by Hofman et al. (1993) on physicians' knowledge of genetics and genetic tests highlighted that non-geneticist physicians who are most likely to encounter patients with specific genetic conditions had the greatest knowledge. However, in the current study, while it could be expected that specialists from OBGYN and PCH would self-report a better knowledge of

genetics than FMED, INTMED or SG, only within OBGYN did the majority of participants self-report their genetics knowledge as good. In PCH, the majority of participants self-reported their genetics knowledge as fair. This lower than expected self-reported genetics knowledge from participants from PCH may be because these medical specialists would see patients and families with genetic conditions more regularly than other specialists and are thus more conscious of what they do not know (being consciously incompetent). According to Houwink et al. (2011), as healthcare professionals become more aware of the impact that genetics can have on their discipline, they become more conscious of their knowledge deficiencies. This could certainly account for the overall lack of self-reported knowledge reported by medical specialists from PCH, because interviews with paediatric specialists revealed adequate genetics knowledge in the context of the more common genetic conditions such as trisomy 21/Down syndrome, DMD and haemophilia.

The majority of specialists from INTMED and SG self-reported their genetics knowledge as fair, while all FMED specialists self-reported their genetics knowledge as poor. This lack of genetics knowledge in healthcare professionals is in keeping with a recent study conducted in Gauteng, South Africa (Walters, Aldous & Malherbe, 2022). In addition, deficiencies in genetics knowledge in non-geneticist physicians have been reported in many countries globally over the past few decades, with little improvement in more recent studies, despite the global movement to increase the integration of genetics education into medical training (Hunter et al., 1998; Suther & Goodson, 2003; Baars, Henneman & ten Kate, 2005; Burke et al., 2006; Houwink et al., 2011).

To gain a better understanding of medical specialists perceived genetics knowledge, participants were asked to spontaneously cite three genetics conditions that would benefit from medical genetic services. The vast majority of all participants (84%, n=27/32) were able to cite three or more conditions. As expected, the condition most commonly cited amongst all medical specialists was trisomy 21/Down syndrome, with one participant reporting seeing a new case almost every week. This is not unexpected as trisomy 21/Down syndrome is the most common chromosomal aneuploidy that occurs in

approximately 1 in 525 live births in South Africa (Kromberg, Sizer & Christianson, 2013) and 1 in 475 live births in Limpopo (Venter et al., 1995).

By discipline, it seemed that the genetic conditions cited are common genetic conditions seen and managed within the specific disciplines. For example, PCH specialists most commonly cited osteogenesis imperfecta, followed by haemophilia; FMED specialists most commonly cited trisomy 21/Down syndrome and trisomy 18/Edwards syndrome; INTMED specialists cited neurodegenerative disorders; OBGYN specialists cited chromosomal aneuploidies; and SG specialists cited various types of cancers. The diseases cited per discipline may provide an insight into the genetic conditions that need specific attention in the province in terms of medical genetic services.

4.2.3. Comfort discussing genetic information with patients

In the absence of locally available genetic services, medical specialists were asked to report their comfort with discussing genetic information and risks with patients. Consistent with previous research (Suther & Goodson, 2003; Burke et al., 2006; Klitzman et al., 2013; Rinke et al., 2014; Chambers et al., 2015; Düsterwald, 2015; Diamonstein et al., 2018), the majority of medical specialists (41%, n=13/32) reported being somewhat comfortable (on a scale of not comfortable, somewhat comfortable, comfortable, to very comfortable), indicating a low level of confidence in their ability to provide this level of care to patients with genetic conditions. The majority of participants from OBGYN reported a lack of comfort in discussing genetic information and risks with patients. This was echoed in the interviews with medical specialists from this discipline, who felt that their lack of understanding of the complexity of genetics and inheritance patterns made it difficult for them to discuss genetics with patients.

Medical specialists from INTMED reported an overall lack of comfort in discussing genetic information and risks with patients, while participants from PCH reported being somewhat comfortable. During the one-on-one interviews, medical specialists from PCH and INTMED described having the skills, knowledge and confidence to provide genetic information and counselling to patients and families with more common conditions, such

as trisomy 21/Down syndrome, DMD, haemophilia (PCH) and adult onset polycystic kidney disease (INTMED). However, when faced with more complex cases, such as dysmorphic or syndromic babies, congenital anomalies and rare genetic conditions, they described deficiencies in their knowledge and ability to counsel patients and families.

In a study by Diamonstein et al. (2018), a small subset of the physicians reported providing genetic counselling to their patients themselves. However, the authors cautioned that the genetic counselling provided by some physicians could be incomplete or below standard, which has also been described in previous research (Klitzman et al., 2013). It is important to note that genetic counselling is not merely the provision of genetic information to patients: it is aimed at "helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates the following: (1) interpretation of family and medical history to assess the chance of disease occurrence or recurrence; (2) education about inheritance, testing, management, prevention, resources and research; (3) [psychosocial] counselling to promote informed choices and adaptation to the risk or condition" (Resta et al., 2006).

A comprehensive genetic counselling session is a lengthy process. While Ormond (2009) indicates that this kind of comprehensive genetic counselling is outside the scope of practice for physicians, if physicians are providing genetic counselling to patients, then it is important to ensure that it is comprehensive and appropriate. In the qualitative phase of the current study, medical specialists, consistent with previous studies (Rinke et al., 2014; Chambers et al., 2015; Diamonstein et al., 2018), reported a lack of time and a busy schedule as barriers to obtaining a proper family and medical history, and providing comprehensive genetic counselling to patients and families. Interviews with medical specialists also revealed that even those medical specialists who feel confident providing some form of genetic information and counselling to patients are not consistently taking a three-generation family history due to the lack of time and heavy workload described. This is consistent with previous research in which appropriate family histories are not obtained by self-reported confident physicians (Rinke et al., 2014). This highlights both a

need for education on the importance of a three-generation family history as well as a need for increased support and capacity in the province.

Of concern, is the fact that, while all the medical specialists from FMED self-reported their genetics knowledge as poor, half of these participants (50%, n=2/4) felt comfortable/very comfortable discussing genetic information and risks with patients. This was also the case for specialists from SG, who self-reported their genetics knowledge as fair/poor, while half of the participants (50%, n=3/6) reported being comfortable discussing genetic information and risks with patients. This raises a concern that patients may be receiving inaccurate or incomplete information from these medical specialists; a concern that has been highlighted in previous research (Klitzman et al., 2013; Diamonstein et al., 2018; Walters, Aldous & Malherbe, 2022).

In keeping with the overall lack of genetics knowledge and comfort in discussing genetic information and risks with patients, the majority of participants (44%, n=14/32) reported being somewhat confident in their knowledge (on a scale of not confident, somewhat confident, confident to very confident). Deficiencies in non-geneticist physicians' knowledge about genetic testing has been reported previously in other studies globally (Baars, Henneman & ten Kate, 2005; Klitzman et al., 2013; Diamonstein et al., 2018).

A lack of appropriate and adequate knowledge about genetics and medical genetic services can lead to missed opportunities for the provision of comprehensive patient care because of poor recognition of a possible genetic aetiology of a disease and a need for referral to medical genetic services (Houwink et al., 2011). Participants identified two main areas lacking: access to genetic information and training and practical exposure as well as access to expert support and advice. This perceived need for training and access to expert support and advice was also described by medical participants during the one-on-one interviews. Access to genetic information and training could be achieved through continuous medical education in the form of workshops, webinars, short courses and reading material. In addition, a number of participants felt that having access to the support and guidance of a multidisciplinary team that includes genetic professionals

would provide them with the opportunity to learn through case-specific and practical exposure. Access to expert support and guidance from medical genetics professionals was described by at least one medical specialists from each discipline, highlighting a perceived need for this expert knowledge and skill in the province. Interestingly, one participant from PCH specifically mentioned having access to telephonic availability of advice (telegenetic services) and another mentioned how having access to a genetic counsellor based in Limpopo and a monthly genetics clinic would improve their knowledge and confidence. This need was also described in a number of studies on non-geneticist physicians' (Burke et al., 2006; Houwink et al., 2011; Rinke et al., 2014; Chow-White, Ha & Laskin, 2017; Diamonstein et al., 2018; Harding et al., 2019). Burke et al. (2006) highlighted a specific need for education and training that is discipline-specific and relevant to specialists' daily practices and needs by focusing on the more common genetic conditions that would be seen within specialities.

4.3. Referral Practices

The vast majority of medical specialists from all disciplines had seen patients or families in the past 12 months who would benefit from consultation with a medical geneticist (88%, n=28/32), genetic counselling (81%, n=26/32) or genetic testing (94%, n=30/32). However, there were very few reported referrals to medical genetic services: only 13% of medical specialists (n=7/32) had referred any patients for consultations with medical geneticists, while 28% (n=9/32) had referred patients or families for genetic counselling. All of the patients referred for consultations with a medical geneticist (n=3/12 for OBGYN and n=3/6 for PCH) or for genetic counselling (n=4/12 for OBGYN and n=4/6 for PCH) had been referred by specialists in PCH and OBGYN, with the exception of one medical specialist from INTMED who reported having referred a patient or family to a medical geneticist or for genetic counselling in the past 12 months. None were referred by specialists from FMED or SG. This low referral rate to genetic specialists and genetic counsellors is consistent with results reported in previous research (Hunter et al., 1998; Chambers et al., 2015; Diamonstein et al., 2018). The study by Hunter et al. (1998) also reported a higher rate of referral by paediatricians, and obstetricians and gynaecologists.

Interestingly, half of the participants (n=16/32) reported that they had sent patient samples for genetic testing in the past 12 months. These were mostly from PCH (n=6/6) and OBGYN (n=8/12). A study by Harding et al. (2019) highlighted that primary care physicians' comfort with the ordering of genetic tests was associated with their personal experience and training. This could account for the differences seen between specialities. However, in a study by Klitzman et al. (2013), this trend of ordering genetic tests in spite of a self-reported lack of genetic knowledge and confidence in providing genetic counselling to patients was also described. They felt that this suggested that physicians could be ordering tests in a suboptimal way, highlighting a need for targeted and speciality-specific education and training (Klitzman et al., 2013).

The low referral rate seems to be linked to knowledge of how to access medical genetic services with an overall rate of 38% (n=12/32), the majority of whom were from PCH (n=5/6) and OBGYN (n=5/12). This lack of knowledge of available medical genetic services as a barrier to the provision of medical genetic services to patients is not only an issue in Limpopo, but has been described both nationally (Walters, Aldous & Malherbe, 2022) and internationally (Burke et al., 2006; Diamonstein et al., 2018). All medical specialists in the current study reported the need for improved access to comprehensive medical genetics services in the province.

Underutilisation of medical genetic services has been reported to be due to a lack of adequate genetics knowledge and education (Suther & Goodson, 2003); however, in the current study, a compounding barrier reported by medical specialists was the fact that the closest medical genetics services are in Gauteng, approximately 250 km away from the tertiary hospital in Limpopo. This places an immense burden on patients and families in terms of long travel times, finances and missed employment. It creates what one medical specialist described as a 'high-threshold' for the referral of patients to these out-of-province medical genetic services. This perceived burden influences medical specialists decision whether or not to refer. This barrier to patient referral related to the ability of patients to travel for specialised services has been described by primary health care

physicians based in rural areas in Canada (Harding et al., 2019) and in Romania (Ciucă, Moldovan & Băban, 2021).

4.4. Opinion of Medical Genetic Services

The need for local expertise and knowledge was further emphasised by the fact that all medical specialists felt that access to both genetic counsellors and genetic testing services was a necessity/indispensable in the province and that the majority of medical specialists (97%, n=31/32) felt that access to a medical geneticist was a necessity/indispensable. Participants described that the large population in the province, primarily serviced by the public healthcare system, would justify having a dedicated medical genetics service in the province. According to Statistics South Africa (2022), Limpopo has the fifth largest population in South Africa with just under 6 million people living in the province, and the highest proportion of children under the age of 15 years in the country (Statistics South Africa, 2022). A total of 92.8% of the provincial population relies on the public healthcare system (Massyn, Pillay & Padarath, 2019). Participants also reported a high number of patients in the province with genetic conditions that would benefit from medical genetic services, including a number of patients with dysmorphology and unexplained syndromes, a high number of pregnant women of advanced maternal age and an ‘enormous amount of cancer’ patients in the province presenting at a young age or with a strong family history. In keeping with the overall lack of genetics knowledge and confidence in diagnosing, treating and managing patients with a genetic condition evident from the results of this study, interviews with the medical specialist identified a need for the expert knowledge, skills and experience of a medical genetic team to provide the support and guidance required for the provision of comprehensive care for these patients in the province.

Medical specialists also highlighted the need for expert skills and services in terms of being able to provide comprehensive genetic counselling to patients and families that includes psychosocial support and the identification of at-risk individuals and family members. Comprehensive medical genetic care and support would translate into

increased patient autonomy and informed decision-making about future pregnancies, which may ultimately provide a financial benefit to the Limpopo Department of Health (LDoH) in terms of the treatment and management of genetic conditions that are very expensive and those that can potentially be prevented through comprehensive antenatal care and surveillance programmes.

According to Klitzman et al. (2013) having access to genetic information and genetic services can enhance the prevention and the treatment of many genetic conditions. A study conducted by Gonzaludo et al. (2019) in the United States of America (USA) estimating the burden and economic impact of paediatric genetic conditions reported that the 'lengthy diagnostic odyssey' in paediatric patients with genetic conditions has a significant economic impact on the national healthcare system. Improved genetic knowledge and physician awareness, combined with appropriate genetic testing, could significantly reduce the cost associated with obtaining a diagnosis in patients with genetic conditions (Gonzaludo et al., 2019).

4.5. Models and Strategies for Providing Medical Genetic Services in an Under-resourced Province

An interesting review of the literature by (Gonzaludo et al., 2019), describing the past, present and future of service delivery in genetic counselling in America, provides various strategies and models for keeping up with the growing demand for medical genetic services in the era of precision medicine in every area of medicine, and not just within the historical and traditional context of monogenic and chromosomal disorders within paediatrics and antenatal care. This article highlights how developments in the field of genetic testing, making it less expensive and with higher utility, and applicable to almost every area of medicine, combined with the growing need for education and research, has placed tremendous pressure on the genetic counselling workforce in the USA. In this article, Stoll, Kubendran and Cohen (2018) describe a number of strategies and models that have been developed by genetic counsellors in order to expand their reach by providing services with greater efficiency to enable them to see more families, often with

the goal of increasing access to services in underserviced communities. It is of the researchers' opinion that these strategies and models can be drawn from and build on in order to 'think out of the box'. This is further supported by the statement made by one participant that ways to increase reach and throughput with only a 'core critical mass' of genetic counsellors in Limpopo could be developed. Most of these strategies and models were identified and suggested by the medical specialists during the one-on-one interviews.

According to the Department of Public Service and Administration (2018), medical specialists in public health earn on average R1 051 368 to R1 744 191 per annum in South Africa, depending on their level of experience and years in practice. Speciality nurses earn between R362 559 to R548 436 per annum, depending on their level of experience and years in service (Department of Public Service and Administration, 2018), while genetic counsellors earn between R574 753 to R746 519 per annum (National Health Laboratory Service, 2015). It therefore makes financial sense to motivate for the creation of genetic counselling posts in the province as a starting point.

This 'core critical mass' of genetic counsellors would then form part of multidisciplinary teams to provide support and/or guidance to medical specialists in the province, would provide counselling for more complex cases and would be the link between medical specialists in the province and medical geneticists outside of the province, making use of telegenetics and possible outreach visits.

In collaboration with the Human Genetics Unit at the University of Limpopo, the genetic counsellors could meet the educational needs of the medical specialists and possibly nursing staff. This may ensure discipline-specific knowledge to ensure appropriate genetic counselling and referrals. Stoll, Kubendran & Cohen (2018), in their review, described barriers to the provision of comprehensive medical genetics and suggested a number of different service delivery models. Some of these models may be applicable to Limpopo:

1) Supportive education: In this model, ongoing education and training are provided to medical specialists on genetics, the assessment of risks, basic genetic counselling and genetic testing. Medical specialists are also provided with continuous support and guidance from genetic counsellors or medical geneticists (Blazer, Slavin, & Weizel, 2006; Blazer et al., 2005; MacDonald, Blazer, & Weitzel, 2010 as cited in Stoll, Kubendran & Cohen, 2018).

By establishing a medical genetics hub in the tertiary academic hospitals in the province, the medical specialists in the province could have access to the support and guidance needed to provide comprehensive care to their patients with genetic conditions. This medical genetic team could provide education and training on an ongoing basis through presentations during academic meetings. This education and training could focus on the importance of appropriate family history taking as an invaluable tool to assist in genetic risk assessment. This could potentially assist to reduce the number of missed cases and the identification of at-risk individuals and family members. This education and training could also focus on genetic conditions and tests relevant to each speciality.

This model could also address the lack of screening and surveillance for the increased risk of aneuploidies associated with advanced maternal age. Interviews with medical specialists from OBGYN have identified that the necessary skills and expertise to perform genetic amniocenteses are available in the province and that the required infrastructure to perform foetal anomaly ultrasounds is available at the high-risk pregnancy clinic at the Pietersburg Hospital. Through proper education and training, the genetic counsellors, in collaboration with the medical specialists from OBGYN, could implement the appropriate screening and surveillance of pregnant women of advance maternal age, as stipulated in the NDoH Maternity and Antenatal Care guidelines (Department of Health, 2016).

2) Consultative support: Medical specialists consult with genetic counsellors about the need for patient referrals or the appropriateness of a specific genetic test. In this model, the genetic counsellor provides patient care indirectly through a medical specialist (Dugan et al., 2017; Mathias et al., 2016; O'Leary et al., 2017; Suarez, Yu, Downs, Costa, & Stevenson, 2017 as cited in Stoll, Kubendran &

Cohen, 2018). According to Stoll, Kubendran & Cohen (2018), this model has the potential to save money by ensuring that the correct tests are ordered.

According to scope of practice for genetic counsellors in South Africa, genetic counsellors can request appropriate genetic tests in consultation with a medical practitioner (Genetic Counsellors South Africa, 2013). The genetic counsellors could provide guidance and support to medical specialists in terms of identifying and ordering the most appropriate and up-to-date genetic tests, as well as assisting to identify patients who would benefit from consultation with a medical geneticist. In cases where the diagnosis is confirmed, patients and families can then be referred for genetic counselling.

3) Triage telegenetics: In this model, the genetic counsellor evaluates patients with the local paediatrician, for instance. A medical geneticist is then consulted through telegenetics, either telephonically or through video conferencing, in cases such as complex dysmorphology or when a diagnosis cannot be made. The medical geneticist then advices the genetic counsellor/paediatric team on how to proceed in terms of tests to be ordered and management of the patient (Kubendran et al., 2017 as cited in Stoll, Kubendran & Cohen, 2018).

By establishing interinstitutional collaborations, medical specialists could access the specialised knowledge and expertise of medical geneticists via telegenetics. This model would be beneficial for the large number of dysmorphology cases and stillbirths described in the province.

4) Genetic counsellor clinics: In this model, patients are seen by the genetic counsellors, independent of a medical geneticist, based on a triage referral system from genetic or paediatric services. Such a clinic would be for cases of medically low complexity, when a diagnosis has already been established or for the interpretation of genetic tests that have been ordered by a non-geneticist physician (Hannig et al., 2014; Heald et al., 2013 as cited in Stoll, Kubendran & Cohen, 2018).

The establishment of genetic counsellor clinics in the province could provide the opportunity for genetic counselling for cancer patients, for instance, who currently receive no genetic information or counselling. It could also provide the option of more comprehensive genetic counselling for patients and families with trisomy 21, DMD, haemophilia and adult on-set polycystic kidney disease if the patients themselves or the medical specialists deem it necessary. This model could also be combined with the group education with individual counselling model described below for pregnant women of advanced maternal age.

5) Group education with individual counselling: In this model, unrelated patients are seen together for an introductory genetic counselling session based on their similar situation. One-on-one individual counselling sessions are then offered to those who identify a need for more personalised counselling (Cloutier et al., 2017; Gammon, Otto, Wick, Borowski & Allyse, 2017 as cited in Stoll, Kubendran & Cohen, 2018).

As mentioned above, this model could be used to provide basic genetic information to pregnant women of advance maternal age about the increased risk of aneuploidies. Those patients who self-identify a need for more comprehensive counselling can then be counselled individually by the genetic counsellors.

6) Collaborative telegenetics: In this model, the genetic counsellors work with genetic counsellor extenders (GCEs) placed at remote sites to triage patients based on the complexity of conditions identified. GCEs are usually nurses or other healthcare professionals who receive ongoing support, guidance and education from the genetic counsellors. The GCEs have sufficient knowledge and expertise to provide risk assessment and counselling for the less complex cases, while the more complex cases receive genetic counselling from the genetic counsellor through telegenetics (Cohen & McIlvried, 2013; Cohen & Nixon, 2016 as cited in Stoll, Kubendran & Cohen, 2018).

This model could be used to extend the reach of the medical genetic services to include all districts in the province, through the training of genetic counsellor extenders (GCEs). The Medical Genetics Education Programme (MGEP) is a post-graduate education programme that was developed by the South African Department of Health in response to the Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities (Department of Health, 2001). The aim of this post-graduate education programme is to provide the necessary genetics knowledge, skills and expertise to nurses in primary healthcare. As described by Malherbe et al. (2017), “If used appropriately, widespread MGEP training could swiftly build up a nursing workforce with improved knowledge and skills in medical genetics”. The MGEP programme has been shown to be successful in the training of healthcare providers (Malherbe et al., 2017), and can potentially be used to train genetic counsellor extenders in Limpopo.

5. CONCLUSION

In conclusion, at the time this study was conducted, it seemed to be the first study to explore the genetic knowledge of medical specialists in Limpopo, referral practices and opinions of medical genetic services in the context of the Limpopo provincial healthcare system. In view of the self-reported genetic knowledge deficiencies by medical specialists in Limpopo, the current study highlights the possible continuous lack of comprehensive and contextually relevant genetics education integration into both undergraduate and postgraduate medical curricula, as was highly recommended by various research and policies within the country. While some basic genetic knowledge and skills were described in a subset of the participants, overall, medical specialists reported lack of knowledge, confidence, time, capacity and resources as barriers to the provision of comprehensive care for patients and families with genetic conditions in Limpopo. Patients and families from Limpopo can access medical genetic services in Gauteng; however, the results from this study indicate low referral rates due to the burden out-of-province referral places on patients and families in terms of long waiting lists, long travel times, finances and missed employment. Interviews with medical specialists highlight a clear sense of need for and benefit of the specialised knowledge, skills and expertise of a medical genetic team to assist in providing comprehensive care to the large population in Limpopo reliant on the public healthcare system.

An increase in the quality of services provided and inclusion of appropriate screening and surveillance programmes could have a significant effect on the economic impact of genetic conditions in the province. However, it is clear that Limpopo is under-resourced and lacking in critical capacity and we need to think carefully and critically about how to implement medical genetic services in the province. This project indicates that a model for providing cost effective medical genetic services in Limpopo could be to set up a small team of genetic counsellors who would act as a hub to improve access to medical genetic services by providing a link between local medical specialists and medical genetic services in other provinces; providing discipline-specific education and training to medical specialists to address genetic knowledge deficiencies; providing education and training

to other healthcare professionals to strengthen the genetic counselling capacity in the province and extend the reach of the medical genetic team; and facilitating research in the province.

5.1. Study Strengths and Limitations

5.1.1. Strengths of the study

- The high response rate obtained in the quantitative phase of the study.
- The use of open-ended questions in the interviews allowed for the medical specialists to report their experiences in their own words, providing better insight into the current situation in Limpopo.
- This study has provided useful insight into the current situation in Limpopo and highlighted the need for locally available medical genetic services.
- The study offered some recommendations on how a comprehensive genetics service could be incorporated into the province

5.1.2. Limitations of the study

- Scoring and analysis of the Likert-type responses could have been performed in order to perform statistical analysis on the self-reported genetic knowledge, comfort discussing genetic information and risks with patients and confidence in knowledge of available genetic tests.
- More comprehensive interferential statistics could have been performed on the quantitative data obtained from the survey questionnaire.
- The results of the study are based on perceived knowledge, comfort and confidence in managing patients and families with genetic conditions and not empirical evidence, and may or not reflect actual practices of the participants.
- This study did not assess the use of a three-generational family history as an important tool for identification of at-risk individuals and family members.
- This study did not include the patients perspective or that of the provincial decision makers and other stakeholders.

5.2. Research Recommendations

Drawing from the models and strategies described in the review by Stoll, Kubendran and Cohen (2018), some of these could be implemented in Limpopo as feasibility studies, with pre- and post-evaluations, as a strategy to provide evidence to support motivation to LDoH for medical genetic services in Limpopo.

A study on the experiences of patients and families when accessing out-of-province medical genetic services would provide the patients' perspective on the perceived burden described by medical specialists in the current study.

There is a lack of epidemiology data on congenital disorders in Limpopo. Research on the incidence, prevalence, mortality rate and morbidity of congenital disorders are necessary in order to understand the health needs of the population in the province.

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LEGISLATION

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APPENDIX A: GOOGLE FORMS SURVEY QUESTIONNAIRE

KNOWLEDGE, ATTITUDES AND PRACTICES OF MEDICAL SPECIALIST'S CONCERNING MEDICAL GENETIC SERVICES AT THE PIETERSBURG AND MANKWENG ACADEMIC TEACHING HOSPITALS IN THE LIMPOPO PROVINCE

Information and Purpose:
The survey questionnaire you are being asked to participate in, is for a research study in partial fulfilment for the degree MMedSci in Genetic Counselling at the University of Cape Town. The research study is focused on examining the knowledge, attitudes and practices of medical specialist's concerning medical genetic services at the Pietersburg and Mankweng academic teaching hospitals in the Limpopo Province. The purpose of this study is to explore your understanding and experiences of medical genetic services, more specifically genetic counsellor, medical geneticist and genetic testing services. Medical specialists from the Department of Surgery, Department of Internal Medicine, Department of Family Medicine, Department of Paediatrics and Child Health and the Department of Obstetrics and Gynaecology at the Pietersburg academic teaching hospital or the Mankweng academic teaching hospital will be recruited for participation in this study.

Your Participation:
This survey questionnaire consists of five (5) sections with forty-one (41) multiple choice and short answer questions on demographic information, education and training, and knowledge, opinion and practices concerning medical genetic services in your context.

The survey will take no more than ten (10) minutes to complete.

Confidentiality:
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 of your information and responses will be kept confidential. The researcher will not share your individual responses with anyone other than the research supervisors.

Dependent on responses received in the survey questionnaire, select respondents will be contacted privately to request an in-depth online interview lasting approximately one (1) hour.

Benefits and Risks:
While there is no direct benefit to you for participating in this study, it is intended to determine if there is a perceived need for medical genetic services in the Limpopo Province. There are no risks associated with participating in the study.

Thank you so much for taking the time to complete this questionnaire.

* Required

Informed Consent

Consent to take part in research:

• I voluntarily 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 question without any consequences of any kind.
• I understand that by completing and submitting this Google Form, I am consenting to participation in this portion of the study.
• I understand that I can withdraw permission to use data from my interview prior to 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 in writing and I have had the opportunity to ask questions about the study.
• I understand that participation involves, completing a Google docs questionnaire consisting of forty-one (41) questions that should take around ten (10) minutes.
• I understand that I will not benefit directly from participating in this research.
• I understand that all information I provide for this study will be treated confidentially.
• 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.
• I understand that under freedom of information legalisation 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.
• I understand that by completing and submitting this Google Form I am agreeing to allow the researcher to contact me for a further in-depth interview on medical genetic services in my discipline.

Researcher MMedSci Genetic Counselling degree candidate:
Professor Kathrine Scholtz: 082 453 7978 / Email:
SCHKAT011@myuct.ac.za

Supervisors:
Dr Tina-Marié Wessels: 021 406 6373 / Email: tina.wessels@uct.ac.za
Dr Chris Sutton: 015 287 5432/ Email: chris.sutton@ul.ac.za

Personal Information

1. Full Name: *

2. Department: *

3. Discipline/Field of Practise: *

_____

4. Telephone Number: *

_____

5. Email: *

_____

Demographics and Professional Characteristics

6. Age (years): *

Mark only one oval.

- ◯ < 30
- ◯ 30-40
- ◯ 41-50
- ◯ 51-60
- ◯ > 60

7. Gender: *

Mark only one oval.

- ◯ Female
- ◯ Male
- ◯ Prefer not to say
- ◯ Other: _____

8. When did you complete your undergraduate training? *

Mark only one oval.

- ◯ < 10 years ago
- ◯ 10-20 years ago
- ◯ 21-30 years ago
- ◯ 31-40 years ago
- ◯ > 41 years ago

9. Where did you complete your undergraduate training? *

_____

10. When did you complete your specialist training? *

Mark only one oval.

- ◯ < 10 years ago
- ◯ 10-20 years ago
- ◯ 21-30 years ago
- ◯ 31-40 years ago
- ◯ > 41 years ago

11. Where did you complete your specialist training? *

_____

12. What qualifications do you hold? *

_____

_____

_____

_____

_____

13. How many years have you worked as a doctor? *

Mark only one oval.

- ◯ < 10 years
- ◯ 10-20 years
- ◯ 21-30 years
- ◯ 31-40 years
- ◯ > 41 years

14. How many years have you worked as a medical specialist? *

Mark only one oval.

- ◯ < 10 years
- ◯ 10-20 years
- ◯ 21-30 years
- ◯ 31-40 years
- ◯ > 41 years

15. Do you have a specific field of interest? *

Mark only one oval.

- ◯ Yes
- ◯ No

16. If yes, then what? *

_____

17. How much genetics education did you receive in undergraduate training? *

Mark only one oval.

- None
- 1-3 lectures
- 4-6 lectures
- 7-10 lectures
- > 10 lectures
- Don't know

18. How much genetics education did you receive in your specialist training? *

Mark only one oval.

- None
- 1-3 lectures
- 4-6 lectures
- 7-10 lectures
- > 10 lectures
- Don't know

19. How many hours per week do you dedicate to continuing medical education? *

Mark only one oval.

- 0
- 1
- 2
- 3
- > 3

20. Has your continuing education ever included genetic education? *

Mark only one oval.

- Yes
- No

21. If yes, what courses have you done and through which platforms/institutions/service providers? (In-house program, external program, MOOC such as Coursera, Journal Course, Webinar, Conference etc.) *

Knowledge about medical genetics and medical genetic services

22. How would you rate your knowledge of genetics? *

Mark only one oval.

- Excellent
- Very good
- Good
- Fair
- Poor

23. Can you spontaneously cite three conditions that would benefit from medical genetic services? *

24. How comfortable are you discussing genetic information and risks with patients? *

Mark only one oval.

- Not comfortable
- Somewhat comfortable
- Comfortable
- Very comfortable

25. How confident are you in your knowledge of available genetic tests and testing options? *

Mark only one oval.

- ◯ Not confident
- ◯ Somewhat confident
- ◯ Confident
- ◯ Very confident

26. What would help improve your confidence? *

Opinion of medical genetic services

27. A medical geneticist is a doctor who specialises in diagnosing and treating genetic disorders or conditions. In the context of the Limpopo Province health system, do you feel MEDICAL GENETICISTS are: *

Mark only one oval.

- ◯ a luxury
- ◯ not applicable to your setting
- ◯ indispensable / a necessity

28. Genetic counsellors are health professionals, with specialised education, training and experience in medical genetics and counselling, who help people understand and adapt to the implications of the genetic contributions to disease. In the context of the Limpopo Province health system, do you feel GENETIC COUNSELLORS are: *

Mark only one oval.

- ◯ a luxury
- ◯ not applicable to your setting
- ◯ indispensable / a necessity

29. Genetic testing is a type of medical test that identifies changes in genes or chromosomes. The results of a genetic test can confirm or rule out a suspected genetic condition or help determine a person's chance of developing or passing on a genetic disorder. In the context of the Limpopo Province health system, do you feel GENETIC TESTING SERVICES are: *

Mark only one oval.

- ◯ a luxury
- ◯ not applicable to your context
- ◯ indispensable / a necessity

30. Which of the following services should be included in medical genetic services? (Mark all that you think apply) *

Check all that apply.

- ☐ Medical geneticists
- ☐ Genetic counselling
- ☐ Genetic testing

Practices

31. In the past 12 months, have you seen any patients you think would benefit from seeing a MEDICAL GENETICIST? *

Mark only one oval.

- ◯ Yes
- ◯ No

32. In the past 12 months, have you referred any patients to a MEDICAL GENETICIST? *

Mark only one oval.

- ◯ Yes
- ◯ No

33. In the past 12 months, have you seen any patients/families you think would benefit from seeing a GENETIC COUNSELLOR? *

Mark only one oval.

- ◯ Yes
- ◯ No

34. In the past 12 months, have you referred any patients/families to a GENETIC COUNSELLOR? *

Mark only one oval.

- ◯ Yes
- ◯ No

35. In the past 12 months, have you seen any patients you think would benefit from GENETIC TESTING? *

Mark only one oval.

- ◯ Yes
- ◯ No

36. In the past 12 months, have you sent any patient samples for GENETIC TESTING? *

Mark only one oval.

- ◯ Yes
- ◯ No

37. Do you think you will refer patients to MEDICAL GENETIC SERVICES (medical geneticist, genetic counsellor or genetic testing) in the future? *

Mark only one oval.

- ◯ Yes
- ◯ No

38. If you answered no to the previous question, could you please elaborate? (Type NA if you answered yes to the previous question) *

_____

_____

_____

_____

_____

39. Do you know how to access MEDICAL GENETIC SERVICES (medical geneticist, genetic counsellor or genetic testing) in South Africa? *

Mark only one oval.

- ◯ Yes
- ◯ No

40. Would you like improved access to MEDICAL GENETIC SERVICES (medical geneticist, genetic counsellor or genetic testing)? *

Mark only one oval.

- ◯ Yes
- ◯ No

41. Is there anything you would like to add?

APPENDIX B: INTERVIEW GUIDE

How long have you been working in the Limpopo Province?

Have you ever had any experiences with medical genetic services/outreaches/genetic projects during your time working in the province?

If so, what was your experience of this service/outreach/project?

Have you had any experience of medical genetic services outside of this province? During your training or before coming to this province? If so, can you describe this experience for me?

What are your experiences with medical genetic services in your discipline?

What do you think is the need for comprehensive medical genetic services (medical geneticists, genetic counsellors, genetic testing) in your discipline within this province?

How would you describe the role of a medical geneticist?

Do you think there is a role for medical geneticists within your discipline?

If not, why not?

If yes, what do you think this role is?

In the survey questionnaire you mentioned that you had seen patients that you thought would benefit from seeing a medical geneticist. Could you tell me more about that?

In the survey questionnaire you mentioned that you had referred a patient/s to a medical geneticist. Could you tell me more about that?

How would you describe the role of a genetic counsellor?

Do you think there is a role for genetic counsellors within your discipline?

If not, why not?

If yes, what do you think this role is?

In the survey questionnaire you mentioned that you had seen patients/families that you thought would benefit from seeing a genetic counsellor. Can you tell me more about that? Why do you feel they would have benefitted from seeing a genetic counsellor?

In the survey questionnaire you mentioned that you had referred patients/families to a genetic counsellor. Can you tell me more about that?

What genetic tests are available specifically for patients within your discipline?

Have you ever used any genetic testing services in South Africa?

If so, can you tell me about your experience of this service?

If not, what is the reason you do not consider genetic testing?

How do you feel about the clinical utility of genetic testing within your discipline? And within your context in the province?

Is there anything else you would like to say that you feel might be relevant for my project?

APPENDIX C: UNIVERSITY OF CAPE TOWN HUMAN RESEARCH ETHICS COMMITTEE APPROVAL

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

Room G50- Old Main Building
Groote Schuur Hospital
Observatory 7925
Telephone [021] 406 6492
Email: hrec-enquiries@uct.ac.za
Website: www.health.uct.ac.za/fhs/research/humanethics/forms

25 January 2021

HREC REF: 654/2020

Dr T Wessels
Division of Human Genetics
FHS
Email: Tina.wessels@uct.ac.za
Student: schkat001@myuct.ac.za

Dear Dr Wessels

PROJECT TITLE: OPINIONS AND PRACTICES OF MEDICAL SPECIALISTS CONCERNING MEDICAL GENETIC SERVICES AT THE PIETERSBURG AND MANKWENG ACADEMIC TEACHING HOSPITALS IN THE LIMPOPO PROVINCE-MMEDSC CANDIDATE-PROF KATHERINE SCHOLTZ

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

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

This approval is subject to strict adherence to the HREC recommendations regarding research involving human participants during COVID -19, dated 17 March 2020 & 06 July 2020.

Approval is granted for one year until the 30 January 2022.

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

The HREC acknowledge that the student: - Prof Katherine Scholtz will also be involved in this study.

Please quote the HREC REF 654/2020 in all your correspondence.

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

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

Yours sincerely

PROFESSOR M BLOCKMAN
CHAIRPERSON, FACULTY OF HEALTH SCIENCES HUMAN RESEARCH ETHICS COMMITTEE
Federal Wide Assurance Number: FWA00001637.
Institutional Review Board (IRB) number: IRB00001938
NHREC-registration number: REC-210208-007

This serves to confirm that the University of Cape Town Human Research Ethics Committee complies to the Ethics Standards for Clinical Research with a new drug in patients, based on the Medical Research Council (MRC-SA), Food and Drug Administration (FDA-USA), International Council for Harmonisation of Technical Requirements for Pharmaceuticals for Human Use: Good Clinical Practice (ICH GCP), South African Good Clinical Practice Guidelines (DoH 2006), based on the Association of the British Pharmaceutical Industry Guidelines (ABPI), and Declaration of Helsinki (2013) guidelines. The Human Research Ethics Committee granting this approval is in compliance with the ICH Harmonised Tripartite Guidelines E6: Note for Guidance on Good Clinical Practice (CPMP/ICH/135/95) and FDA Code Federal Regulation Part 50, 56 and 312.

APPENDIX D: PIETERSBURG/MANKWENG RESEARCH ETHICS COMMITTEE APPROVAL

LIMPOPO
PROVINCIAL GOVERNMENT
REPUBLIC OF SOUTH AFRICA

DEPARTMENT OF HEALTH
PIETERSBURG/MANKWENG RESEARCH ETHICS COMMITTEE (PMREC)

ENQUIRIES: Mr MA POOPEDI **DATE: 02 JUNE 2021**

MANAGER: CLINICAL RESEARCH

ananiaspoopedi@gmail.com

REFERENCE	:	**PMREC 17 MARCH UL 2021/C**
DATE	:	**02 JUNE 2021**
RESEARCHER	:	**Prof K Scholtz**
		(PRINCIPAL INVESTIGATOR)
RESEARCH	:	**POST-GRADUATE RESEARCH**
DEPARTMENT	:	**Human Genetics - University of Cape Town**

Protocol Title : Opinions and Practices of Medical Specialists Concerning Medical Genetic Services at the Pietersburg and Mankweng Academic Teaching Hospital in the Limpopo Province.

CANDIDATE	:	**Prof K Scholtz**
APPROVAL STATUS	:	**APPROVED**

SIGNED:

PROF TAB MASHEGO

Prof TAB Mashego, PhD
Chairperson: Pietersburg/Mankweng Complex Research Ethics Committee
School of Medicine
University of Limpopo
REC 300408-006

APPENDIX E: LIMPOPO DEPARTMENT OF HEALTH PERMISSION TO CONDUCT RESEARCH

LIMPOPO
PROVINCIAL GOVERNMENT
REPUBLIC OF SOUTH AFRICA

Department of Health

Ref : LP_2021-03-016
Enquires : Ms PF Mahlokwane
Tel : 015-293 6028
Email : Phoebe.Mahlokwane@dhsd.limpopo.gov.za

Katie Scholtz

PERMISSION TO CONDUCT RESEARCH IN DEPARTMENTAL FACILITIES

Your Study Topic as indicated below;

Opinions and practices of medical specialists concerning medical Genetic services at the Pietersburg and Mankweng academic teaching hospitals in the Limpopo province.

1. Permission to conduct research study as per your research proposal is hereby Granted.
2. Kindly note the following:
    a. Present this letter of permission to the institution supervisor/s a week before the study is conducted.
    b. In the course of your study, there should be no action that disrupts the routine services, or incur any cost on the Department.
    c. After completion of study, it is mandatory that the findings should be submitted to the Department to serve as a resource.
    d. The researcher should be prepared to assist in the interpretation and implementation of the study recommendation where possible.
    e. The approval is only valid for a 1-year period.
    f. If the proposal has been amended, a new approval should be sought from the Department of Health
    g. Kindly note that, the Department can withdraw the approval at any time.

Your cooperation will be highly appreciated

pp **Head of Department** **Date** 19/04/2021

Private Bag X9302 Polokwane
Fidel Castro Ruz House, 18 College Street. Polokwane 0700. Tel: 015 293 6000/12. Fax: 015 293 6211.
Website: http/www.limpopo.gov.za

The heartland of Southern Africa – Development is about people!

APPENDIX F: RESEARCH PROJECT INFORMATION SHEET AND CONSENT FORM

University of Cape Town, Department of Pathology, Division of Human Genetics

Research Project Information Sheet and Consent Form

For any questions about the project please contact:

Kathrine Scholtz: 015 268 4056 / 082 453 7978 / Email: SCHKAT011@myuct.ac.za / kathrine.scholtz@ul.ac.za

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

Dr Chris Sutton: 015 287 5432/ Email: chris.sutton@ul.ac.za

Title: OPINIONS AND PRACTICES OF MEDICAL SPECIALISTS CONCERNING MEDICAL GENETIC SERVICES AT THE PIETERSBURG AND MANKWENG ACADEMIC TEACHING HOSPITALS IN THE LIMPOPO PROVINCE

Information and Purpose: The interview for which you are being asked to participate in, is for a research study in partial fulfilment for the degree MMedSci in Genetic Counselling at the University of Cape Town. The research study is focused on examining the knowledge, attitudes and practices of medical specialist's concerning medical genetic services at the Pietersburg and Mankweng academic teaching hospitals in the Limpopo Province. The purpose of this study is to explore your understanding and experiences of medical genetic services, more specifically genetic counsellor, medical geneticist and genetic testing services. Medical specialists from the Department of Surgery, Department of Internal Medicine, Department of Family Medicine, Department of Paediatrics and Child Health and the Department of Obstetrics and Gynaecology at the Pietersburg academic teaching hospital or the Mankweng academic teaching hospital will be recruited for participation in this study. The initial survey questionnaire will be circulated to all members of staff from the abovementioned departments for completion. Interviews will be conducted with approximately 10 – 15 participants.

Your Participation: Your participation in this study will consist of a completing a Google docs questionnaire consisting of twenty-nine (29) questions that should take around ten (10) minutes and thereafter, an interview lasting approximately one hour. During the interview session, you will be asked a series of questions about medical genetics services in your discipline. 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. There is no penalty

for discontinuing participation. 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 result in any penalty or loss of benefits to which you are entitled. In the event you choose to withdraw from the study all information you provide (including recordings) will be destroyed and omitted from the final research report and any future publications.

Benefits and Risks: While there is no direct benefit to you for participating in this study, it is intended to determine if there is a perceived need for medical genetic services in the Limpopo Province. 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 of your information and interview responses will be kept confidential. The researcher will not share your individual responses with anyone other than the research supervisor.

Consent to take part in research

- I,…….. voluntarily 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 question without any consequences of any kind.
- I understand that I can withdraw permission to use data from my interview prior to 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 in writing and I have had the opportunity to ask questions about the study.
- I understand that participation involves, completing a Google docs questionnaire consisting of twenty-nine (29) questions that should take around ten (10) minutes and thereafter, 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 treated confidentially.
- 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 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 Limpopo and original audio recordings will be retained in a password protected cellphone and/or password protected laptop 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 final examination.
- I understand that under freedom of information legalisation 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.

Researcher MMedSci Genetic Counselling degree candidate:

Professor Kathrine Scholtz: 082 453 7978 / Email: SCHKAT011@myuct.ac.za

Supervisors:

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

Dr Chris Sutton: 015 287 5432/ Email: chris.sutton@ul.ac.za

Signature of research participant

---------- ----------

Signature of participant Date

Signature of researcher

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

---------- ----------

Signature of researcher Date

APPENDIX G: LIST OF CONDITIONS CITED THAT WOULD BENEFIT FROM MEDICAL GENETICS SERVICES

Condition Cited	Frequency
Trisomy 21/Down Syndrome	16
Breast Cancer	6
Trisomy 18/Edwards Syndrome	6
Colon Cancer	5
Cystic Fibrosis	4
Trisomy 13/Patau Syndrome	4
Osteogenesis Imperfecta	3
Recurrent Miscarriages	2
Marfan Syndrome	2
Multiple Foetal Anomalies/Congenital anomalies	2
Turner Syndrome	2
Haemophilia	2
Muscular Dystrophy (no specific type)	2
BRCA1	1
BRCA2	1
Thyroid Cancer	1
Multiple Endocrine Neoplasia Syndromes	1
Multiple Endocrine Adenomatoses	1
Micrognathia	1
Spina Bifida	1
Congenital Heart Disease	1
Diabetes	1

Neurofibromatosis	1
Hereditary Ataxia	1
Wilsons Disease	1
Alport Syndrome	1
Spinocerebellar Ataxia (SCA)	1
Huntington Disease	1
Duchenne Muscular Dystrophy (DMD)	1
Prenatal Genetic Counselling	1
Anencephaly	1
Prada-Willi Syndrome	1
Teacher Collins Syndrome	1
Ovarian Cancer	1
Endometrial Cancer	1
Chromosomal Anomalies	1
Primary Amenorrhoea	1
Albinism	1
Achondroplasia	1
Congenital Adrenal Hypoplasia	1
Neonatal Diabetes Mellitus	1
Congenital Myopathies	1
Myelomenningocele	1
Mucopolysarcharidosis (MPS)	1
Skeletal Dysplasia	1
Thalassaemia	1