UNDERSTANDING DOCTORS' KNOWLEDGE AND ATTITUDES CONCERNING GENETICS AND GENETIC SERVICES IN SOUTH AFRICA

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DECLARATION

I, Gillian Düsterwald, 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

AD	Autosomal dominant
AR	Autosomal recessive
ASHG	American Society of Human Genetics
BRCA	Breast cancer susceptibility gene
CME	Continuing Medical Education
CPD	Continuing Professional Development
DTC	Direct-to-consumer
FASD	Foetal alcohol spectrum disorder
GC-SA	Genetic Counselling South Africa
GP	General practitioner
GenTEE	Genetic Testing in Emerging Economies
HIV/AIDS	Human immunodeficiency Virus/Acquired Immunodeficiency Syndrome
HPCSA	Health Professionals Council of South Africa
IMR	Infant mortality rate
NSGC	National Society of Genetic Counsellors (United States of America)
SNPs	Single nucleotide polymorphisms
UCT	University of Cape Town
WHO	World Health Organisation
XLD	X-linked dominant
XLR	X-linked recessive

ABSTRACT

The burgeoning knowledge in genetics implies that genetic services (including clinical and counselling) will be in increasing demand in the future.

This study investigated South African doctors' genetic education, knowledge and attitudes towards genetic services and examined whether these factors affect referral to genetic services. Several studies have indicated that health professionals have poor understanding of genetics and genetic conditions, and this lack of insight extends to knowledge of genetic services and how and when to access them, so that those who would benefit from services might not gain access to them because they are not referred by their doctors (Delikurt et al., 2015).

METHODS

A questionnaire was developed based on published research and questions relating to the aims of the project. Forty one questions were asked, covering referral patterns, demographics, education, knowledge and attitudes to genetic services and genetic counselling.

The sample population consisted of 140 GPs attending a family practitioners' conference. Fifty one responses were received.

Results were analysed using descriptive statistics and content analysis of open ended questions.

RESULTS

Results show that 52.9% of general practitioners have referred to genetics in the past, 92.2% think they will refer in future and only 49% know how to access genetic services. Doctors who knew how to access genetic services were more likely to have previously referred patients to these services. Almost half the doctors who had not used genetic services previously felt that genetic services were difficult to access and several attributed this to their lack of knowledge.

Doctors who had the most education were more likely to have previously referred patients to genetic services. Doctors indicated that they would like more education on basic genetics, common genetic conditions and genetic services via forums such as conferences, CME activities and online resources.

More than half of the doctors rated themselves as "not confident" in their genetic knowledge. Down syndrome was the most commonly seen genetic condition in practice,

followed by cystic fibrosis and breast cancer. More than 80% of respondents did not know about direct-to-consumer testing, but 46% thought they might be approached to interpret the results of a direct-to-consumer test in future. Doctors showed poor understanding of the ethics of testing minors for genetic conditions.

Genetic services and genetic counselling were seen as indispensable by 66.7% and 74.5% of doctors respectively.

CONCLUSIONS

Overall, doctors' knowledge of genetics seems poor and most of them indicate they would like more education on basic genetic concepts, referral guidelines for genetic services and how to access genetic services, which agrees with the amount of education being the most important factor relating to previous referral to genetic services.

This study will provide guidance for awareness and education programmes, and inform the future development of genetic services in South Africa.

1 LITERATURE REVIEW

The literature for the review was selected by searching the Google Scholar database using the terms: [genetic counselling referral OR doctor OR specialist]; Articles excluding patents. An alert was created, and abstracts of relevant articles were read to determine whether or not they would add to the literature already selected. Full articles were downloaded when the abstract indicated that they would be relevant.

1.1 Introduction

Genetic services have existed in South Africa since the 1970's and strong policies have been created for their development. However, the expansion of these services has stagnated since the 1990's, when the Human immunodeficiency virus/Acquired Immunodeficiency syndrome (HIV/AIDS) epidemic diverted resources from genetic services into primary health care. Improved care for HIV patients has controlled the epidemic and the infant mortality rate (IMR) has decreased. Demographics in South Africa indicate that genetic services are now essential, and proper use of these services could further reduce the IMR through appropriate management of congenital conditions, but there are still very few funded posts for medical geneticists, genetic counsellors and medical genetic scientists (who work in laboratories) in the public and private sector (Nippert et al., 2013). While public health policy may be the main reason for the lack of posts, use of genetic services by medical doctors working in primary care is of interest. This study was designed to research whether primary care doctors are using genetic services (or would like to use them) and see them as a valuable addition to their practice.

The exponential growth of genomics and molecular biology in the past two decades, including the Human Genome Project, advances in sequencing and information technology, has resulted in almost daily reports of breakthroughs in genetics (Burke et al., 2010). This increase in knowledge could lead to opportunities for improving public health by improving early detection, implementing more effective prevention programmes and targeting personalised treatment. These developments will allow improved use of resources by health professionals (Kurlan & Ford, 2015) e.g. better screening of populations ensuring that treatments get to people who will benefit, new treatments and genetic testing options. Consensus among delegates at a Public Health Genomics meeting held in Suffolk, UK, (Burke et al., 2010) was that "…health professionals had to engage with the genomics agenda and to recognise its potential for disease prevention and health improvement."

However, the rate of expansion of genetic knowledge makes it difficult for medical doctors to keep up to date without continuing education and better use of genetic services will only happen if doctors know how and when to refer patients to these services. There is little known about how doctors working outside genetics use genetic services and what factors are associated with referral to genetic services. Several studies have indicated that health professionals have poor understanding of genetics and genetic conditions, and this lack of insight extends to knowledge of genetic services and how and when to access them, so that those who would benefit from services might not gain access to them because they are not referred by their doctors (Delikurt et al., 2015).

This descriptive survey-based study attempts to describe how medical doctors working in general practice in South Africa understand and use genetic knowledge and genetic services. The study will also investigate what factors have been associated with previous referral to genetic services, and whether the doctors think they may refer in future. Finally, the survey served as an educational tool to raise awareness about genetics and genetic services amongst the group of doctors participating in the survey.

1.2 Health care services in South Africa

Health care services in South Africa are inequitable (Benatar, 2004 cited in Greenberg et al., 2012). There are disparities in medical services between the nine provinces, which implement government programmes and policies at their own discretion and according to their individual needs (Greenberg et al., 2012). Access to medical services is compounded by social dynamics. The country is positioned between first world and third world development, with a rural population of 18.8 million (GeoHive, 2015), nine official languages and several different cultures.

A large number of poor and indigent people are seen in the state sector, where fees are charged according to income, so that the poorest patients are treated for free and wealthier patients pay according to their means. Primary care occurs in district based clinics and more specialised care (including genetic services) requires referral to secondary, tertiary and quaternary state hospitals which may not be easily accessible by these patients. In contrast, a smaller and wealthier proportion has medical aid insurance which pays professionals working in private care (Greenberg et al., 2012). Private patients will usually consult a general practitioner (GP) or family physician for primary care, and rely on these doctors for referral into specialised services if necessary. Private patients can also be seen in state hospitals and their medical aid will be billed accordingly. Since there are currently no medical geneticists

working in private practice and only a few genetic counsellors working privately part time, private patients are generally also referred to state genetic services for consultations with medical geneticists.

1.3 The importance of genetic services in health care

Genetic services comprise medical geneticists, genetic counsellors, genetic nurses and medical scientists working as a collaborative team. Together, these professionals provide comprehensive health services for individuals and families affected by genetic and congenital conditions.

While genetic services are available in South Africa, budget constraints mean the focus is on primary health care, including antenatal care, childbirth and early childhood care. Tertiary services, including genetic services, are not given prominence in local health settings and the development of South Africa's genetic services is hampered by the lack of genetic posts and limited investment in new laboratory technology and equipment (Kromberg & Krause, 2013). In a small study in which doctors responded to a survey on genetic testing, they indicated that there were too few genetic counsellors and medical geneticists in South Africa (Vogel et al., 2011).

The emphasis on common diseases is appropriate, but rare diseases, of which 70% are inherited or congenital, are collectively a great burden on individual families and society as a whole and are often associated with lifelong special needs (Nippert et al., 2013). At least 1 in every 15 births in South Africa is estimated to be affected by a congenital disorder (Malherbe, Christianson & Aldous, 2015). This means that over 18 000 new cases should be reported every year, but in 2012 only just over 2000 cases were reported - an under-reporting of 88% (Malherbe, Christianson & Aldous, 2015). Between 2008 and 2010, the reported percentage of under-5 year old deaths due to congenital and genetic disorders doubled from 4% to 8%, which is similar to figures reported in India (3% to 7%) and the Philippines (6% to 10%). This increase can be attributed to a reduction in deaths from infectious diseases and malnutrition, yet it is still believed that "poor universal clinical diagnostic services and inadequate surveillance and reporting systems" leads to under-reporting of deaths due to congenital anomalies (Nippert et al., 2013).

The burden of genetic conditions as a proportion of morbidity becomes relevant when the IMR reduces to below 40 in 1000 live births (Malherbe, Christianson & Aldous, 2015). In South Africa, with the improved access of the population to antiretroviral therapy, the IMR in 2014 was 33.5 per 1000 births (Malherbe, Christianson & Aldous, 2015).

Up to 70% of congenital diseases can be prevented, treated, or reduced in severity through genetic services, including prenatal screening and, for example, through the introduction of folic acid supplementation reducing the incidence of neural tube defects, but attending doctors need to recognise these conditions and know where to refer patients in order for this potential to be realised. Genetic services are also becoming more important with increased understanding of genetic contributions to common diseases such as cancer, cardiovascular disease, diabetes and dementia (Riesgraf et al., 2014).

1.2.1 Genetic services in South Africa

Genetic services in which medical geneticists and genetic counsellors work as a collaborative team were first officially offered in South Africa in 1974, when 15 publicly funded genetic nurse positions were established in centres across the country (Jenkins, 1990; Greenberg et al., 2012). As early as 1982, Op't Hof stated that facilities in South Africa were too limited to provide genetic counselling to all prospective couples with indications such as family history, a child with a genetic disorder or malformation, intellectual disability, metabolic disease, disorders of sex development, history of miscarriages or stillbirths, teratogens and consanguinity. While all of these indications and psychosocial issues remain relevant today, the options for patients have been profoundly increased and now include carrier testing, preimplantation genetic diagnosis, non-invasive prenatal testing, prenatal diagnosis and next generation sequencing. Recently, debates around the "three-parent embryo" (for mitochondrial conditions) and "embryo editing" (using gene editing technologies to repair or alter mutated genes) have added even more complexity to the field of genetics (Albertini, 2015).

The first masters' programme in genetic counselling in South Africa was introduced in the late 1980's (Greenberg et al., 2012). The number of genetics posts increased gradually through the late 1990's and early 2000's (Kromberg, Sizer & Christianson, 2013) until by 2008 there were five genetic services centres in South Africa. However, the HIV/AIDS and tuberculosis epidemics redirected funds and support from genetic services and thwarted efforts to implement policy guidelines (Kromberg, Wessels & Krause, 2013), so that infrastructure for primary care counselling remains undeveloped (Nippert et al., 2013).

The GenTEE report comments that there are currently sufficient genetic training programmes in South Africa to train staff for medical genetic services. However, "the national and provincial health departments lack(ing) the will, commitment and finances to support this training and make posts available for those trained" (Nippert et al., 2013).

Diagnoses of genetic conditions need to be confirmed by skilled professionals and laboratory services and there is currently a lack of both of these in South Africa, despite policy guidelines published by the National Department of Health in 2004 recommending 20 medical geneticists and 80 genetic counsellors to be in posts by 2010 (Malherbe, Christianson & Aldous, 2015). At present, there are 8 medical geneticists and 30 genetic counsellors registered with the Health Professionals Council of South Africa (HPCSA) (HPCSA, 2015). However, only a maximum of 18 of the listed genetic counsellors are currently practising (J. Greenberg, pers. comm., 2015).

In the developing countries mentioned in the GenTEE report (Argentina, Brazil, China, Egypt, India, Oman, the Philippines and South Africa) (Nippert et al., 2013), the lack of sufficient medical genetic services probably contributes to a lack of awareness by GPs about the availability and utility of these services, and thus there are fewer referrals to these services. Despite this, the WHO states that: "Cuba, Brazil, India, China and South Africa are countries that offer excellent examples of the utilisation of genetic technology and serves to address the health needs of their populations…." (WHO, 2008).

1.2.2 Access to genetic services in South Africa

Fairly comprehensive genetic services offering clinical, counselling and laboratory genetic services as well as academic training and outreach clinics are available in the public health system in the larger cities of Cape Town, Bloemfontein and Johannesburg, with a limited service available in Durban. These services are linked to the academic centres of the universities of Cape Town, Free State, Stellenbosch, KwaZulu Natal and Witwatersrand (Greenberg et al., 2012) and are available through referral from other hospitals and clinics.

Rural communities are served through outreach visits, but these are insufficient for the needs of the large rural population in South Africa. Access to all forms of healthcare in rural areas is significantly more difficult than in urban areas due mainly to distance between clinics and lack of money for transport, and this is true of genetic services too. The limited accessibility and scope of health care facilities in rural areas has an impact on how rural doctors feel about genetics and genetic services. However, the fact that patients in rural areas are more reliant on primary care doctors for referral to genetic services means that it is even more important for these physicians to have the knowledge to be able to refer to genetic services when necessary (Marathe et al., 2015).

A small number of wealthier patients can be seen in the private sector, where several genetic counsellors have small practices with the support of academic centres (HPCSA, 2015,

Kromberg, Wessels & Krause, 2013). However, at a meeting of Genetic Counselling South Africa (GCSA) in 2011, counsellors in private practice reported that they were not very busy. This does seem to be improving slowly with more awareness by some doctors, particularly after efforts are made to include genetics education in professional meetings (pers. comm., Frieda Loubser, 2015).

Bidondo et al. (2015) suggested several strategies for addressing birth defects from the public health perspective in Argentina, a developing country with many similarities to South Africa. Among these were to address the perception that, due to the need for technology, research and genomics, medical genetics is a service for highly developed countries only, and, secondly, to develop genetic training programmes for health professionals at all levels with an emphasis on primary care. Similarly to South Africa, Argentina has insufficient genetic services available for the population and services are unevenly spread through the population and among different disorders e.g. in South Africa special services exist for neural tubes defects but not for Down syndrome (Nippert et al., 2013). A suggestion to attach trained genetic counsellors to groups of GPs or specialist clinics in towns and cities which currently have no genetic services would help to raise awareness of genetics and the unique skills that genetic counsellors possess by both the medical professionals and the lay public (Greenberg et al., 2012).

1.4 Referral to genetic services by doctors

According to the WHO in 2008, a functioning public health care system requires an appropriate referral system to specialised services (e.g. genetic services) if needed. Consequently, patients in South Africa are unable to access genetic services directly, as these services are found in the tertiary healthcare sector. A GP's ability to recognise genetic conditions and refer to appropriate specialists is thus crucial and requires basic knowledge of genetic conditions as well as the benefits and availability of genetic services (Marathe et al., 2015). However, in South Africa, there is little use of genetic services by medical professionals working outside academic hospitals, and patients who would benefit from these services might not gain access to them because they are not referred by their doctors (Greenberg et al., 2012).

Despite the fact that up to 15% of a GP's consultations are said to involve genetic conditions (Hopkins, 2007), several studies worldwide have shown that a large number of GPs have never referred patients to genetic services and that there are several barriers to referral. The

most commonly cited reason for not referring patients to genetic services is lack of knowledge, both of conditions and of availability of services.

In a survey in the Netherlands, Baars, Henneman & ten Kate (2005) found many doctors did not refer patients for genetic counselling due to a lack of knowledge about genetics and genetic testing. A previous study (Aalfs et al., 2003) in the Netherlands indicated that 29% of GPs had never referred a patient for genetic counselling. Of the remainder, 40% of referrals had been initiated by the patient. Similarly, Claybrook et al. (2010), found that only half of oncologists surveyed in Indiana had referred patients with colorectal cancer to genetic services. Reasons cited for not referring patients included lack of appropriate patients, lack of information about genetic services and lack of knowledge about which patients should be referred. These studies are further supported by a recent investigation in Columbia, where doctors working in genetic clinics reported that education and training in genetics in medical schools were inadequate, and that medical practitioners did not know enough to be able to refer appropriately to a genetic service (Rodas-Perez et al., 2015). This report also cited absence of training in communication skills and the influence of religious beliefs as barriers to referral by some colleagues. Tan & Fitzgerald (2014) working in Australia, surveyed GPs and specialists on their referrals to genetic services for Lynch syndrome (hereditary colon cancer) and found three main barriers: 1) the clinical knowledge of the physicians, including which patients are eligible for services and lack of time and skills to collect family histories; 2) the patients' experiences, interest in genetic services and knowledge of family history and 3) organizational issues including cost and access to services, referral guidelines and referral pathways. These authors suggested developing supportive tools for physicians.

The same factors were recognised in a review of nine studies (Delikurt et al., 2015), mostly from America but including Australia and the Netherlands, which described six barriers to referral to genetic services by non-genetic healthcare professionals:

i) lack of awareness of patient risk factors,
ii) failure to obtain family history,
iii) lack of knowledge of genetics and genetic conditions,
iv) lack of awareness of genetic services,
v) inadequate coordination of referral,
vi) lack of genetics workforce.

Another recent review (Mikat-Stevens, Larson & Tarini, 2014) classified barriers across four themes:

i) knowledge and skills,

ii) ethical, legal and social implications (e.g. genetic discrimination),

iii) healthcare systems

iv) scientific evidence.

This review also mentioned patient anxiety and lack of time as barriers to referral.

In South Africa, Kromberg, Sizer & Christianson (2013) included financial, geographical, cultural and political barriers in the list. However, geographical and financial barriers do not necessarily stop people from travelling to use genetic services if they know that they need them and where to find them, and this information would usually be offered by their primary care providers. Furthermore, most genetic services clinics offer an outreach programme for rural areas (J. Greenberg, pers. comm. 2012). Geographical and financial barriers to use of genetic services are also mentioned in a study in Columbia where genetic services are not covered by health insurance (Rodas-Perez et al., 2015).

Sometimes, patients are more aware of their need for genetic services than doctors. In 2010, the National Society of Genetic Counsellors (NSGC) in America (quoted in Riesgraf et al., 2014), claimed that about 50% of genetic counselling clients are self-referred. This implies that doctors do not always recognise when a client should be referred to genetic services and the system of referral to state services, via tertiary institutions, may not be optimal at this point for patient needs. In another example from Pakistan, 53% of couples self-referred for prenatal genetic counselling and only 44% were referred by their doctors (Afroze & Jehan, 2014). The self-referrals were mostly seeking information on family history or risks for future pregnancies. The authors attributed the lack of referral to unawareness by doctors and lack of service integration. Once again, genetic education for doctors is proposed as part of the solution.

In South Africa, the general public's lack of knowledge about genetic services, as well as cultural beliefs regarding genetic conditions, create further barriers to the use of these services and self-referrals are less frequent (Kromberg, Sizer & Christianson, 2013). Anecdotal evidence collected by the author suggests that even people with tertiary education do not understand what genetic counselling entails. Once they are better informed, many of them say that they themselves, or someone that they know, could have used the service. Again, anecdotally, the few people that the author has met who have used counselling services have struggled to find them in the private sector and usually their medical service providers have been unable to help them to locate a genetic counsellor. This

observation is also mentioned in Doyle et al.'s 2015 paper where it was mentioned that "genetic counsellors are a less visible, less explicit component ….simply because, being a relatively new profession, there are not enough practising genetic counsellors to be able to demonstrate broad utility." This is further borne out by Delikurt et al. (2015) who summarised barriers to individual use of genetic services as lack of awareness of personal risk, lack of knowledge and/or awareness of medical history of family members and lack of knowledge of genetic services.

The importance of raising awareness of genetics by doctors was illustrated in a survey done at a conference on genetic testing in South Africa. While fewer than 50% of these doctors had requested a genetic test previously, more than 75% indicated that they intended doing so after the conference (Vogel et al., 2011).

1.5 Medical doctors' knowledge of genetics

Adequate knowledge of genetic conditions is necessary for diagnosis and/or appropriate referral to genetic services by doctors. Furthermore, while specialised genetic services are essential for complete care, genetic and genomic information is increasingly being used across healthcare disciplines and thus all health professionals should have a basic working knowledge of genetics (Seven et al., 2015).

It has been suggested that many GPs are "confidently incompetent" (they do not know what they do not know) in their knowledge of genetics (Hapgood et al., 2002, cited in Hopkins, 2007). However, many other studies have shown that GPs feel they lack the skills and knowledge to deal with patient queries about genetic conditions in practice and are thus fully aware of their incompetence in genetics (Aalfs et al., 2003; Barbero et al., 2003; Bernhard et al., 2005; de Abrew, Dissanayake & Korf, 2014; Klitzman et al., 2013; Marathe et al., 2015; Rodas-Perez et al., 2015; Trivers et al., 2011). These studies have been conducted worldwide on health professionals' knowledge of genetic conditions, with most indicating that their knowledge of genetics is very limited, is perceived as not clinically relevant and does not cover the complex psychosocial and ethical issues raised by genetic conditions, and that this impacts on the quality of care of patients (de Abrew, Dissanayake & Korf, 2014).

For example, in the USA in 2013, Klitzman et al. (2013) found that, of 220 physicians surveyed, 73.7% and 87.1% respectively rated their knowledge of genetics and genetic testing as very or somewhat poor. These figures have remained static over the last decade. GPs in the Netherlands also felt that their level of genetic knowledge was limited (Aalfs et al., 2003). Barbero et al. (2003) found that health professionals in Argentina had little

knowledge of medical genetics. In Maryland, USA only 10% of obstetric patients with at least one indication for genetic counselling were referred to genetic counselling clinics and insufficient information was given to the patients on what to expect from the counselling service (Bernhard et al., 2005). Another study in the United States found that doctors inaccurately estimated risk for *BRCA*1/2 mutations and thus referred too many low risk and too few high risk patients to genetic services (Trivers et al., 2011). Van Wyk (2008) found similar gaps in Johannesburg GPs' understanding of cancer genetics. Finally, research in Tasmania showed that GPs managing genetic cardiac disease are reliant on information from cardiologists for information on the condition itself, as well as whether or not to refer patients for genetic counselling (Marathe et al., 2015).

In contrast, Wonkam, Njamnshi & Angwafo (2006) and Abdolahi et al. (2014) found that doctors in Cameroon and Iran respectively demonstrate an adequate level of clinical genetic knowledge and the need for genetic services although doctors in Cameroon are largely unaware of DNA diagnostic tests.

Despite the majority of studies described above recounting a self-reported lack of knowledge on genetics by medical doctors, Kumar & Gantley (1999) reported that GPs felt that they would be able to integrate genetic technology into their work with little adaptation. However, at the same time, many studies show that doctors themselves feel that they need better training in genetic conditions, how to access genetic services and when to refer patients to services (Baars, Henneman & ten Kate, 2005; Tan & Fitzgerald, 2014; Tan, Spurdle & Obermair, 2014).

1.6 Doctors' education in genetics

Knowledge and education are inextricably linked, and many of the studies investigating the genetic knowledge of doctors recommend that improved education of doctors in genetics is the major factor that will influence better understanding and awareness of genetics, and more appropriate use of genetic services (Baars, Henneman & ten Kate, 2005; de Abrew, Dissanayake & Korf, 2014; Klitzman et al., 2013; Rodas-Perez et al., 2015; Tan & Fitzgerald, 2014; Tan, Spurdle & Obermair, 2014; Trivers et al., 2011; Wonkam, Njamnshi & Angwafo, 2006). Greenberg et al. (2012), discussing the challenges faced by the genetic counselling profession in South Africa, indicate that education is key to both medical and lay people using services. They state: "If the health professionals were better informed, they would refer a wider range of patients, more people would benefit…..and counsellors could expand their

expertise…" i.e. Better understanding of genetic conditions would lead to more demand for services.

More and better structured training in genetics is required at medical schools, as many GPs feel that their genetic education has been insufficient even though they recognise the importance of genetics for their work (Burke et al., 2009; Rodas-Perez et al., 2015). Furthermore, with the amount of genetic knowledge increasing at such a fast rate (Burke et al., 2010), it would be impossible for undergraduate training to provide sufficient genetic education for medical doctors, and this education should be supplemented by continuing medical education (CME) or continuing professional development (CPD) once these doctors are practising. South African doctors are obliged by the HPCSA to maintain their professional status through obtaining CME points and this is an ideal platform for improving their understanding of genetics (Pather, 2006). Postgraduate education in genetics is also important as even medical specialities recognise that they are limited in their knowledge, awareness and understanding of genetics (Pather, 2006; Rodas-Perez et al., 2015).

Even prior to the completion of the Human Genome Project, Hunter et al. (1998) commented that genetic education about services, conditions, testing and implications for the patient and family would become very important in the future with the expansion of genetic knowledge, for both patients and clinicians. This need for "genetically literate" doctors has been emphasized by several authors (Feero et al., 2014 cited in de Abrew, Dissanayake & Korf, 2014; Wonkam, Njamnshi & Angwafo, 2006) discussing how medical education needs to adapt to the rapid rate of knowledge acquisition, especially as more genetic testing becomes available.

In South Africa, basic training of health care professionals, particularly nurses, occurs from primary health care level through to tertiary settings, while other medical professionals such as physiotherapists, occupational therapists and medical doctors receive limited undergraduate teaching, with only four medical schools (University of Cape Town, University of the Witwatersrand, University of the Orange Free State and University of Stellenbosch) employing medical geneticists and integrating medical genetics into undergraduate student curricula (Nippert et al., 2013). Limited medical genetics is also taught at three other universities, but teaching is usually done by non-genetic specialists such as paediatricians. The GenTEE report assumes that this unsatisfactory undergraduate medical training results means that most physicians in South Africa "do not recognize the genetic basis of diseases of their patients, do not know how to refer to genetic services, if available, and do not give due importance to genetic counselling" (Nippert et al., 2013).

A review group in the UK identified several desired outcomes for training of undergraduate doctors in genetics (Burke et al., 2009). These were:

i) the ability to identify patients with genetic conditions, with understanding of inheritance patterns and basic genetics;
ii) the ability to manage patients with genetic conditions;
iii) the ability to appropriately refer patients with genetic conditions;
iv) the ability to access information on genetics
v) the ability to understand the uses and limitations of different genetic tests, and ethical issues associated with genetic testing;
vi) the ability to discuss genetic information with patients.

This group also identified 17 genetic conditions that doctors should be able to identify and manage. These included chromosomal conditions (e.g. Down syndrome), common single gene disorders (e.g. haemophilia,) common disorders with a genetic component (e.g. Alzheimer's disease) and familial cancers. Core competencies in genetics for nurses have also been described and they include outcomes i), iii), v) and vi) above and add the skills of taking family histories and drawing pedigrees, which would be valuable for doctors too (Seven et al., 2015).

Recently, a group in Texas implemented a medical genetics programme for their paediatric residents, since they felt that genetic training had been deficient in primary care residents. This programme had three main outcomes, indicating the effectiveness of a well-structured educational programme (Nguyen et al., 2015):

i) learning opportunities were provided in a variety of settings, and the trainees said it was "the most educational rotation" that they experienced;
ii) an increase in trainee confidence and clinical competence;
iii) increased awareness and appreciation for multidisciplinary relationships, in particular for genetic counsellors.

1.7 Family doctors as gatekeepers for genetic services

Family physicians (or GPs) provide comprehensive, holistic care for patients of all ages with all conditions (Hopkins, 2007; Abdolahi et al., 2014) and are often the first point of screening for early signs of serious conditions. The relationship of a family physician with a family, as well as their continued involvement in patient care and long-term management of chronic and complicated conditions means that these clinicians are familiar with health history and

healthcare needs in families (Hopkins, 2007) and may be able to identify individuals and families for whom genetic testing could be helpful (Burke et al., 2009) e.g. detecting single gene disorders such as hypercholesterolaemia or inherited cardiomyopathies could identify asymptomatic family members who would benefit from prophylactic treatment. However, in the public health system in South Africa, particularly in the Community Health Clinics, the role of a GP in continuing health care is less apparent as doctors often rotate duties (Masters, 2010). Up to 15% of GP consultations are believed to involve genetics, but this may be an underestimate when including queries such as stature, familial baldness, family history of cancer and drug responses (Hopkins, 2007).

A directed family history is the first important tool for screening for genetic conditions, and family physicians are in the unique position of being able to take a comprehensive family history which could indicate referral to genetic services. However, these doctors do not have the time or the knowledge for comprehensive genetic consultations and counselling during their short contact times with patients. Recommendations for primary care geneticists, that could reduce the load on overloaded tertiary centres, have been made in the United Kingdom, but this has not been accepted as a viable option as support structures and evidence for cost savings are not available (Hopkins, 2007). Similarly, by the 1990s, the National Department of Health in South Africa had realised the need for more widespread genetic services and set up policy guidelines which supported trying to offer medical genetic services including counselling services to the public through primary care but these policies have also not been implemented (Nippert et al., 2013).

Since South Africa is a developing and diverse country, its genetic services requirements may differ from those of developed countries, but studies done in Johannesburg (Kromberg & Berkowitz, 1986) and in Queensland (Kromberg, Parkes & Taylor, 2006) found that GPs were the second largest referral source of clients to genetic services and recommended further genetic education for them.

1.7 Unique roles and skills of genetic counsellors

A study in Columbia (Rodas-Perez et al., 2015) found that doctors working in medical genetics did not believe that other health professionals could work as genetic counsellors, emphasising the specialist nature of this field. However, as far as this researcher can ascertain, no studies have been done in South Africa on what medical doctors working outside of genetics understand about the roles of genetic counsellors and how they can improve delivery of health services.

Genetic counselling is a relatively new profession, first practiced in the 1970s (Harper, 2010) and gradually gaining in popularity since then. Genetic counsellors' roles have evolved since 1971, and they now see clients in many different areas (Uhlmann, Schuette & Yashar, 2009). Initially, they were supportive of paediatric geneticists and helped with family histories, researching genetic conditions, explaining genetic concepts to families and supporting families emotionally. Nowadays, while the traditional skills are still very important, genetic counsellors also work in a variety of clinical settings, including paediatrics, neurogenetics and cancer genetics, and in diverse fields like pharmacogenomics (looking at individual variations in drug response), bioinformatics, diagnostic laboratories (advising on appropriate use of genetic tests), research development, and public health and policy settings, often as part of a multidisciplinary team in specialist units in these diverse settings (Doyle et al., 2015; Skirton et al., 2014).

Fulfilling these roles requires a unique set of skills, all of which are not usually part of a doctor's skills (Doyle et al., 2015), and which Uhlmann, Schuette & Yashar (2009) classed into four domains:

i) communication skills (written and verbal);
ii) critical thinking skills (evaluating and presenting risks through analysing information);
iii) interpersonal, counselling and psychosocial assessment skills;
iv) professional and ethical values.

A review by Skirton et al. (2014) suggested that genetic counsellors undertake a significant workload in direct patient care and there should be more use of genetic counsellors in countries where they are underutilised.

In South Africa, genetic counsellors play several other roles beyond counselling (Kromberg, Wessels & Krause, 2013). These include teaching, research, marketing, public engagement and administration. Counsellors themselves suggested that more marketing of the profession is needed, more education should be provided and more referrals sought in order for the profession to grow and reach its potential (Kromberg, Wessels & Krause, 2013).

1.8 Medical genetics in the future

New technologies such as next generation sequencing and CRISPR (clustered regularly interspaced short palindromic repeats), as well as the field of pharmacogenomics and the increasing recognition of personalised healthcare, are all easily accessible to the public through the internet and, increasingly, social media (Riesgraf et al., 2014).

Pharmacogenomics, particularly, is a field that will have impact on general practice in future, as more and more drug metabolism pathways are associated with single nucleotide polymorphisms (SNPs), with implications for drug dosage regimens (Walter & Emory, 2012). As the public becomes more aware of these advances, there will be a greater need for genetic services to help the public to understand their utility, creating the need for public health genomics policies and for medical doctors to be able to answer queries about genomics from their patients. The Bellagio statement (Boccia & Zimmern, 2015) defined public health genomics as "the responsible and effective translation of genome-based knowledge and technologies into public policy and health services for the benefit of population health."

Whereas previously, cost and time meant that only one gene could be tested at a time, the advent of massively parallel rapid gene sequencing means that many genes can be sequenced at one time, at much reduced cost. At present, sequencing is of limited clinical utility because of the uncertain implications of the many variants of unknown significance being discovered. However, as this information begins to complement clinical knowledge, future integration into clinical practice is probably inevitable (Vassy et al., 2015). Since June 2013, when Angelina Jolie disclosed her *BRCA* 1 positive status, and when the United States Supreme Court ruled that genes could not be patented (Borzekowski et al., 2013), public awareness and demand for genetic testing has been vastly increased, especially in the First World. A study done in America found that referrals for *BRCA* testing increased by 85% following the release of Jolie's results, with an increase of 107% in the number of identified *BRCA*1/2 carriers, which implied that the referrals were appropriately identified. The authors identified the challenge of meeting increased demand for genetic services including screening, counselling, testing and preventive surgery (Raphael et al., 2015). While it is recommended that multigene panel testing is accompanied by genetic counselling to explain the nature of variants of unknown significance and the implications of the testing itself, in reality the consumer-driven nature of the testing and the fact that testing is offered direct to the consumer via the internet means that these ethical issues are not often covered and the value of these tests in improving risk assessment, early detection and prevention, particularly in oncology, has not been realised. Kurlan & Ford (2015) recommend research on the effectiveness of clinician-patient communication and healthcare delivery systems required for panel testing, with an emphasis on access and ethics and Klitzman et al. (2013) stated that it is crucial to understand knowledge, attitudes and practices of doctors regarding genetic testing as it becomes more extensive and increasingly available.

In 1960, only 700 inherited conditions were recognised, by 1980, 3000 single gene disorders were categorised (Op't Hof, 1982) and now over 10 000 disease causing genes are listed in the gene card database (Weizmann Institute of Science, 2014). In the near future, the ready availability of genomic sequencing will mean that, in order to fulfil the imperative to "do no harm", multigene panels will need to be carefully selected and tailored to each patient (Kurlan & Ford, 2015), so that patients without risk factors aren't screened unnecessarily, patients understand what is being tested, and so that they receive the necessary advice about screening and prevention strategies.

Adding to the availability of clinically useful genetic tests, a range of direct-to-consumer (DTC) genetic tests can now be purchased online via the Internet. Some of these tests offer health-related products including susceptibility testing for common diseases such as cancers and diabetes, as well as pharmacogenomics testing. Some of these tests are available without consultation with a physician, although all recommend pre- and post-test genetic counselling, and some are only available through a medical doctor. In a review article (Goldsmith et al., 2013) up to 65% of primary care doctors were not aware of DTC testing, and up to 85% did not feel qualified to interpret the results of a DTC test. This article also mentioned the need for improved education of doctors, not only in genetics but also in personal genomics.

The current genetic workforce is understaffed, even in developed countries such as the USA, and Kurlan & Ford (2015), commenting on the availability of multigene panels, recommend a "social investment" in training for genomics and social medicine, both of genetic counsellors and clinicians so that recommended referral to expert clinicians for test selection, pre- and post-test counselling can occur "whenever possible."

1.9 Motivation for research

The idea for this study was borne out of frustration with the lack of genetic counselling posts in South Africa. Several studies have indicated that health professionals have poor understanding of genetics and genetic conditions, and this lack of insight extends to knowledge of genetic services and how and when to access them, and means that people may not be referred appropriately to genetic services (Delikurt et al., 2015). No previous studies have addressed these issues in South Africa.

Several studies worldwide (e.g. Aalfs et al., 2003; Baars, Henneman & ten Kate, 2005; Barbero et al., 2003; Claybrook et al., 2010; Freedman et al., 2003; Hunter et al., 1998; Klitzman et al., 2013) and two in South Africa (van Wyk, 2008; Vogel et al., 2011) have

evaluated doctors' knowledge and attitudes[1] to aspects of genetics. This study will cover a broader range of doctors' genetic knowledge and education and also investigate their opinions on genetic services in an attempt to understand what factors affect use of genetic services by doctors in South Africa.

In achieving this goal, it is hoped that awareness of genetic counselling and genetic services is raised among doctors. By addressing issues identified in the study, genetic services could respond to non-genetic doctors' needs and referral to genetic services could become more frequent. Ultimately, this knowledge could be used to gain insight into how genetics can be integrated into medical care by doctors.

The information obtained here could also be used to develop new academic curricula for basic and clinical genetics at medical schools, as well as a CPD programme for further genetics education of doctors.

Finally, the answers obtained from doctors to the survey may encourage the South African Department of Health to implement the policy guidelines that are currently being updated for the broadening of genetic services in South Africa.

However, the study is limited by small numbers and will be cautious in extrapolating findings to the general GP or family practitioner population. Further research will be needed to verify findings.

1.9.1 Aims

The aim of this study was to assess general practitioners' knowledge and attitudes towards genetics and genetic services in South Africa, and to understand what factors play a role in referral to genetic services.

1.9.2 Objectives

1. To determine whether doctors have referred previously to genetic services and whether they intend to do so in future.
2. To determine what factors relate to doctors' use of genetic services.

[1] The word "attitudes" as used in this dissertation refers to the doctor's opinions towards aspects of genetics, genetic services and genomics. This study does not explore attitudes from a psychological or social science perspective.

3. To investigate how much genetics education doctors have received in undergraduate studies and as postgraduates, the form of this education and what sort of education they would like in future.
4. To explore what doctors know about genetics, genetic conditions, genomics, and ethical issues of testing children.
5. To explore doctors' understanding of the value of genetic services.
6. To explore doctors' opinions about the roles of genetic counsellors.
7. To investigate what doctors think they need from genetic services in South Africa.

2 METHODS

The major research questions asked in this study were:

1. What do doctors know about genetics and genetic services in South Africa?
2. What factors affect doctors' referral to genetic services in South Africa?

The study attempts to explore these factors by asking several open-ended questions to inform the quantitative answers obtained from the survey.

This chapter describes the participants and methods used to conduct this research. It includes design, participants, instrumentation, ethics considerations and data analysis of the study.

2.1 Study design

This descriptive cross-sectional survey uses a structured questionnaire with closed- and open-ended questions. This type of survey is described by Sandelowski (2000: 337) as "especially amenable to obtaining straight and largely unadorned (i.e., minimally theorized or otherwise transformed…) answers to questions of special relevance to practitioners and policy makers" and is thus well suited to this study which seeks to understand what GPs know about genetics and genetic services, what factors are associated with the use of genetic services by GPs in South Africa, and how the use of these services by GPs could be improved. The advantages of this type of study include greater validity of data and ensuring that questions are answered from different perspectives (Sandelowski, 2000).

The quantitative component of this study provided possible associations between referral patterns, demographics, education and knowledge of the participants. However, the information gained from quantitative surveys is constrained by what has not been asked as all variables are not known (Kearns, 1992). The addition of open-ended questions to describe the answers to quantitative questions seeks to include some of these unknown variables. Qualitative methods are designed to answer questions from the perspective of the subjects experiencing a particular phenomenon (Vaismoradi, Turenen & Bondas, 2013), and in this study, open ended questions were asked to attempt to understand doctors' attitudes to genetics and to inform the quantitative answers by asking doctors to explain their responses.

2.2 Study participants

The 5-day annual University of Cape Town Division of Family Medicine Conference of General Practitioners, was held from 19 to 23 January in 2015 at the River Club in Cape Town, and is open to all doctors in South Africa. A wide range of interested physicians are invited to attend

every year (www.uct.ac.za/calendar/events). The 2015 Programme included Ophthalmology, Pharmaco-Therapeutics, Bioethics (which included talks about genetics), a section on the Future of Medicine, talks on Cardiology, Rheumatology, Gynaecology, Geriatrics and Dermatology, thus doctors practising in a variety of fields as well as general practice were expected to attend the conference. Doctors could choose which sessions to attend, but most were present for the whole conference.

The sample was chosen for convenience as this was an easy method of obtaining a relatively large and diverse group of GPs in one area at one time. Since all doctors could attend and would obtain CME points for attendance, the sample was assumed to be reasonably representative despite its small size relative to the larger population of doctors practicing in South Africa. In this study, the term "family doctors" will indicate both general practitioners (GPs) and family physicians. In South Africa, family physicians are required to hold a MBChB and a MMed in Family Medicine whereas GPs only need a MBChB in order to practise. However, this study is mainly concerned with physicians (also called clinicians or doctors) working in primary health care, whether they are qualified as family physicians or as GPs and since the conference stipulated "General Practitioners" in its title, it could be assumed the doctors were working as GPs. General practitioners work in a wide range of areas, from private to public, urban to rural (Masters, 2010). The participating doctors came from different regions in South Africa and two indicated that they practised in other countries (Zimbabwe and Ireland) as well as doing limited work in South Africa.

The study population was considered appropriate for answering the research question, as GPs are commonly involved in primary care and are the most likely health professionals to identify conditions in their patients requiring referral to specialist services, including genetics.

As an incentive to participate, they were offered the chance to be included in a lucky draw for a two night stay in a holiday flat in Cape Town.

2.3 Research instrumentation

The research instrument was a structured questionnaire generated on Google Forms, which allowed an Internet-based survey to be available, as well as a printed survey to be distributed at the conference. The internet survey was used by one respondent in the pilot study.

Data was collected using the self-administered structured survey that was designed to investigate GPs' genetic education, knowledge of genetics, and attitudes to genetic services, as well as demographic factors and how these issues relate to patient referral patterns to

genetic services. Survey content was informed by a review of current published literature on knowledge and attitudes of doctors to genetics and genetic services, as well as by questions that would help to answer the research question. Several questions were adapted from previously published papers and these are cited below (section 2.3.2).

The language of the survey was English, and this was appropriate as the conference was conducted in English, most medical information is available in English and it could be assumed that conference delegates were proficient enough in English to complete the survey instrument.

Christensen et al. (2013) listed the following advantages of self-administered, structured surveys:

i) self-administered questionnaires are cost- and time-effective, as large numbers of responses can be obtained without expending time on individual face-to-face or telephonic interviews. However, face-to face interviews are known to have higher response rates, possibly due to the motivating physical presence of an interviewer. This was offset in this case by the economic and time restraints of this project, which had to be completed in a few months with a limited budget. While lower levels of non-response are considered to be an indicator of quality, it is not known what bias is introduced by non-respondents to surveys;

ii) answers to self-administered surveys tend to be of higher quality than answers from face-to-face interviews, particularly with sensitive questions, due to the phenomenon of social desirability bias. In this survey, respondents were more likely to be honest about their assessment of their own knowledge of genetics and whether or not they thought genetic services are valuable, if they believed that their answers were anonymous, particularly if their knowledge was thought to be inadequate or if they held negative stances about genetic services. Factual answers tend to show no difference between self-administered and face-to-face survey modes.

iii) paper based surveys also allow respondents to be more free in their answers and to add comments wherever they feel it is appropriate, thus allowing participants to comment on aspects about which they feel strongly and allowing for a richer analysis of the content;

iv) individuals with higher education levels have been associated with an increase in response rate to all modes of surveys. The participants in this survey were all

medical doctors attending an English conference, so it was expected that they were all highly educated.

Further advantages to using the paper-based survey was the ease of distribution at the conference. This format and venue also made it more likely that respondents would answer from their own knowledge and not try to search online or through other medical references for correct answers to knowledge questions. This survey could be distributed to future cohorts without influencing the quality of the data collected.

Disadvantages of self-administered questionnaires include:

i) a lower response rate, both to the overall survey and to individual items in a questionnaire, probably due to the lack of interviewer motivation;

ii) visualisation of items of scale may influence responders to choose less extreme answers than they would if they were asked face-to-face;

iii) as mentioned above it is also well known that there is an inherent bias in all modes of survey, because it is not known what factors differ between non-respondent and respondent characteristics;

iv) since time available is restricted, recall bias can be a problem with all types of surveys, particularly self-administered surveys, as there is no way of prompting memory with probing questions as is sometimes done in face-to-face or telephone interviews. However, only three questions in this survey relied on recall – two on use of genetic services, and one on conditions seen in practice (Christensen et al., 2013).

2.3.1 Information sheet and informed consent (Appendix A)

An information sheet and informed consent form were attached to the front of the questionnaire (Appendix A). The information sheet explained the purpose of the survey, that it consisted of 41 questions that would take less than 20 minutes to complete, and asked the doctors to complete the questionnaire as fully as possible. It also mentioned that we were interested in the doctors' working knowledge of genetics, as well as their exposure to genetics and experience in practice. Contact details of the researcher and supervisor were included on the cover page, which the respondents were encouraged to remove and keep. It also described the lucky draw.

The informed consent sheet requested participants to sign consent to fill in the survey and asked them to email the researcher if they wanted access to the online version of the

questionnaire. Furthermore, there was a section asking respondents to indicate whether or not they would like feedback on the study results.

2.3.2 Questionnaire

The final survey consisted of five sections containing 41 questions altogether (Appendix B). Questions were asked in various forms, including questions with multiple choice answers, ranked answers using a Likert scale and short answer questions, allowing the collection of both qualitative and quantitative data.

The sections were as follows:

Section 1 consisted of three initial questions relating to patient referral to genetic services. The questions asked whether the doctor had previously referred patients to genetic services, whether they thought they would refer to genetic services in future, and whether they knew how to access genetic services in South Africa.

Section 2 contained eight demographic questions. Demographic information included field of practice, age, educational qualifications and year of graduation, gender, location of practice, years in practice and specific fields of interest. Questions in this section were informed by previously published literature (Baars, Henneman & ten Kate, 2005; Klitzman et al., 2013; Tan, Spurdle & Obermair, 2014).

Section 3 asked questions on education and knowledge. The four questions on education included amount and form of genetics education during undergraduate studies and since graduation. The sub-section on knowledge comprised 12 questions. These were:

i) A Likert scale of self-reported confidence in knowledge of genetics on a scale of 1 to 5 from not confident to very confident. The Likert scale is used as a measure of attitude (Boone & Boone, 2012);
ii) A table asking doctors to fill in genetic conditions that they had seen in their practice, and then answer questions on mode of inheritance, whether or not a genetic test is available and whether or not the condition would have a significant impact on life. This question was adapted from a similar question by Wonkam, Njamnshi & Angwafo (2006).
iii) A table pre-populated with 10 inherited and congenital conditions and asking about the doctors' experience with the conditions, the availability of metabolic, genetic, carrier and predictive testing, and whether or not other family members are at risk).

This question was adapted from similar questions in papers published by Baars, Henneman & ten Kate (2005) and Seven, et al. (2015).

iv) A checklist question on how prenatal conditions are diagnosed.

v) A series of four questions on inherited breast cancer.

vi) To assess knowledge of genomics and new technologies, three questions were asked about DTC testing.

vii) A multiple choice question on the ethics of testing children under the age of 18 was included in this section.

Section 4 asked 14 questions about genetic services and included several open-ended questions which targeted general practitioners' perceptions of and attitudes to genetic services. Questions in this section included:

i) which medical professionals should refer to genetic services;

ii) roles of genetic counsellors;

iii) three questions adapted from Wonkam, Njamnshi & Angwafo (2006) about the value of genetic services and genetic counselling;

iv) a question about further genetic education needs;

v) two questions about access to genetic services;

vi) a question on what needs doctors have from genetic services

Three questions were asked about doctors' knowledge and attitudes about genomics in Section 5. These included:

i) an open-ended question asking what the respondent would do if a patient approached them with results from a DTC genetic test;

ii) how we can make genomics more accessible to clinicians in South Africa;

iii) whether pharmacogenomics will have an impact on prescribing medications in future.

The last question in the questionnaire was an open-ended question asking whether the genetics section on the last day of the conference had changed the respondents' answers in any way.

2.3.3 Pilot study

Prior to the conference, a pilot draft of the survey was sent to 10 family doctors practising in Cape Town. Five paper surveys were delivered to a local family practice and five internet surveys were sent to doctors involved in problem based learning facilitation at the University

of Cape Town Faculty of Health Sciences. Only two replies were received, one paper survey from the family practice and one internet survey from a problem based learning facilitator. The following changes were made to the questionnaire:

i. Typesetting errors were amended;
ii. Question 15 was duplicated in the pilot questionnaire. The duplicate question was removed and all questions after 15 were re-numbered;
iii. Originally, question 17 asked doctors to list 5 genetic conditions they had seen in practice and then refer back to these conditions to answer a series of questions about each. This question was tabulated in the final questionnaire as a more practical way to answer this question.
iv. Similarly, in the pilot survey, question 18 was a series of 10 (18 a. to j.) checkbox questions asking "What experience have you had with (name of condition)?" with 9 possible answers. This was also tabulated for practicality.
v. The final question to be answered after the genetics session at the conference was not included in the pilot survey.

2.4 Ethical considerations

Ethics approval was obtained from the UCT Human Research Ethics Committee (HREC/REF: 812/2014) before starting data collection. An information sheet and consent form were attached to the front of the survey (Appendix A). Participants signed consent to participate in the survey. Since they are medical doctors, it was expected that the participants had a high level of reading comprehension and that they understood the information sheet and consent form. The participants were reassured that all collected data would remain anonymous. The informed consent page and request to participate in the lucky draw was removed and kept separately to the questionnaires before analysing the data, so that the data remained anonymous.

2.4.1 Data safety and monitoring

All written data generated was kept in a locked cabinet to be destroyed after the project's completion. The data was only made available to the researcher and the supervisors. Names were removed as soon as questionnaires and interviews were returned. Participants' anonymity was assured by ascribing a code to each questionnaire and removing all identifying information.

A separate list of names and contact details was kept for the prize draw and destroyed after the winner was announced.

A separate list of names was kept of participating doctors who wanted feedback after completion of the study.

2.5 Data collection

Paper questionnaires were handed out to each doctor attending the conference and the doctors were reminded of the invitation to complete the questionnaires each morning. Ten doctors did not accept questionnaires. Two other surveys were distributed at the conference, which limited time available for completion of the questionnaire.

Surveys were distributed to delegates over the five days of the conference. The participants could choose to answer the questionnaire at any time between the beginning of the conference (19 Jan 2015) and 30 April 2015, but only one respondent completed the questionnaire after the conference and returned it to the researcher by email. Although all participants were offered the opportunity to complete the questionnaire on the internet, no one used this method. A further 50 surveys of the 140 distributed were completed and returned by hand during the conference for a 36.4% response rate, which was similar to the response rate of 36.7% quoted by Wonkam, Njamnshi & Angwafo (2006) but lower than the reported response rate to mail surveys of physicians in America of between 50 and 59% (Asch, Jedzriewski & Christakis,1997). The response rate was, however, much higher than the 15.2% response rate to a mail survey of GPs in South Africa (Masters, 2010).

This low number of responses could theoretically lead to increased response bias, but studies done on response bias in paediatricians in America have shown very low rates of response bias compared to patients and the whole population (Cull et al., 2005).

Not all surveys were completed fully, but sufficient information was obtained from each questionnaire that all were included in data analysis.

The responses were captured into the google docs' survey instrument and a response form similar to an Excel form was generated.

2.6 Data analysis

The raw data generated from the questionnaires was analysed using descriptive statistics, content analysis and chi-squared association analysis. Descriptive statistics were used for quantitative analysis, to summarise the data numerically. Themes were identified from open-ended questions and the numbers of statements relating to those themes were reported. In this way, underlying reasons for basic quantitative observations could be determined,

providing a deeper understanding of the results than would have been obtained from pure statistical analysis.

Data was organised into tables and charts for easy comparison and figures were presented both as frequencies and percentages, which were rounded off to the nearest 0.1%.

Several questions contained a text box next to the answer "Other", in which comments could be made. These comments were noted and content analysed for further insight into the answers supplied by multiple choice or checklist questions.

In the analysis of the results, several questions were not presented in the order of the questionnaire, as some answers were found to be more relevant to other sections. The question asking why doctors hadn't previously used genetic services was moved to the first section on referral patterns of doctors, as the answers were applicable to this segment. A question on what sort of further education on genetics, if any, the doctors would like, was allocated to the section on Education. The question asking what the respondents think about their knowledge of genetics was later allocated to the section on Knowledge, and analysed to inform the respondents' self-assessment on their knowledge of genetics. The genomics questions were also later allocated to the Knowledge section.

2.6.1 Referral patterns of doctors to genetic services

Descriptive statistics were used to quantitatively describe referral to genetic services in the past and predicted referral in future, and this was linked to later answers to an open-ended question on why some doctors had not used genetic services previously.

2.6.2 Demographics

Demographics of the sample were analysed using descriptive statistics.

2.6.3 Doctor's genetic education and knowledge of genetics

Education and knowledge of doctors were both analysed using descriptive statistics, with individual answers being informed by content analysis of open-ended questions.

Answers to the questions on genetics knowledge (questions 17, 18, 19, 20, 21, 22, 23, 24, 25, 26 and 27) were scored as below. Answers given by doctors were discussed with other genetic counsellors and also by consulting Firth, Hurst & Hall (2005) and Gene Reviews.

Question 17: A table of conditions doctors recalled seeing in practice. Each condition noted was given a score out of three, one for correctly identifying a genetic condition, one for correctly identifying the mode of inheritance and one for correctly identifying whether or

not a genetic test is available. No marks were allocated for answering the question about the impact on the patient's life as answers were regarded as subjective, but these answers were noted and analysed.

Question 18*. Table pre-populated with a list of ten genetic conditions. Answers to this table were scored out of 30, with a mark given for correctly identifying whether or not a genetic test is available, a mark for correctly identifying whether predictive or carrier testing is available and a mark for correctly identifying whether other family members are at risk. The section on metabolic testing was removed from the analysis as it was felt that the question was ambiguous. It was also decided that, since Down syndrome can, in some circumstances, be familial, either answer for "Other family members at risk" would be accepted (Appendix C shows a table of correct answers).

Question 19*. Doctors chose from the following list of options for prenatal diagnosis: family history, ultrasound scan, non-invasive prenatal testing, amniocentesis, karyotype, testing maternal DNA, testing paternal DNA and other. All seven of these answers were correct and a mark out of seven was given depending on how many correct answers were chosen.

Questions 20 to 23*. Hereditary breast cancer. This section was marked out of four, with "both" being the correct answer for the first question, "10%" for the second question, "Yes" for the third question and "As someone at higher than population risk" for the fourth question.

Question 26*. The question on the meaning of "VOUS" was added to the overall knowledge score, with one point given for the correct answer: Variant of Unknown Significance.

Question 27*. The question asking when it is appropriate to test a child under the age of 18 for a genetic condition gave seven options and doctors could choose as many as they thought were accurate. Three answers in this section were correct: "When the child requests a test", "When a child is symptomatic" and "When treatment or management is available that could prevent or delay the onset of a genetic condition". A score out of 3 was added to the final knowledge score, but the answers were further analysed to see which incorrect answers were chosen as well.

Answers to the questions marked above with a * were scored and marks were added together for an overall score out of 45 for each respondent that answered all questions. Ratings on genetic conditions seen in practice were separated from the other knowledge

questions as they did not add information to doctors' overall knowledge score, and the number of conditions seen by each doctor varied. This section gave a quantitative score, as well as some qualitative information where respondents commented on their answers. Answers to questions were also analysed independently to inform future educational presentations for doctors.

2.6.4 Doctors' knowledge and attitudes about genetic services

Attitudes and knowledge of doctors about genetic services were analysed using descriptive statistics, with individual answers being informed by content analysis of open-ended questions.

2.6.5 Content analysis

Answers to open-ended questions dispersed throughout the survey instrument were categorised into conceptually similar responses using content analysis which is a flexible method for analysing text data obtained from open-ended survey questions and other sources such as interviews, print media and focus groups. According to Hsieh and Shannon (2005:1278) "qualitative content analysis is defined as a research method for the subjective interpretation of the content of text data through the systematic classification process of coding and identifying themes or patterns". Conventional content analysis was used in this study due to the limited amount of existing research on doctors' attitudes to genetics and genetic services and how this relates to their use of these services. This method meant that, in most cases, no preconceived categories or theoretical perspectives were imposed on data obtained from the respondents. However, genetic counsellor's roles, as described by respondents, fitted into the four categories of skills mentioned by Uhlmann, Schuette & Yashar (2009) and this guide was used to categorise responses.

Similarly, open-ended answers on the value of genetic services and genetic counselling were put into themes and then categorised according to respondents' choice of whether services were "Indispensable", "A luxury", "Pointless" or "Other".

Initially, all responses were read several times to get an overall sense of the content. The responses were then read word by word to extract key concepts from the content, which were highlighted. Notes were made of this initial analysis before creating codes describing the key concepts. The codes were then grouped into themes (Hsieh & Shannon, 2005).

Supporting comments from open-ended questions are included under themes in the Results section. Many comments were excluded from the Results section for the sake of brevity, but

some were used to illustrate themes and all relevant comments are included in Appendix E with reference made in the relevant results section.

2.6.6 Test of association

Test of association or "goodness of fit" analysis was conducted using a chi-squared (χ^2) distribution in contingency tables (matrices of counts) to determine which variables were significantly associated with patient referral to genetic services by doctors in the past. The χ^2 test statistic is a function of the differences between observed and expected frequencies with a known sampling distribution, and is valid with a sample size of at least 40 (Underhill & Bradfield, 2012).

The Yates' continuity correction (Y) was used to prevent overestimation of significance when at least one cell of the chi-squared data table was smaller than 5, although it has been argued that the Yates' continuity correction overcorrects and is unnecessary even with small sample sizes. A significance value of the chi-square test $p > 0.05$ implies that the data is normally distributed, i.e. that there is no significant difference between observed and expected frequencies (Underhill & Bradfield, 2012).

For the analysis, variables were grouped into above the average and below or equal to the average, so years of practice were divided into less than or equal to 24 and greater than 24. Hours of undergraduate education were grouped into less than or equal to 5 hours or more than 5 hours, and, similarly, hours of genetics education post-graduation were clustered into 3 hours or less, and more than 3 hours. Overall knowledge was grouped into less than or equal to the average score obtained (39%) and more than 39%. The number of conditions seen in practice were divided into 3 or less and 4 or more.

When doing statistical analysis, the acceptable number of respondents depends on the type of research study, and depends on the central limit theory, which states that in a sufficiently large sample, the distribution of the mean of the samples will be normal. A figure of 30 respondents is usually sufficient unless the distribution is very skew (Underhill & Bradfield, 2012) and 10% to 20% of the population is sufficient for a descriptive study (Gay & Diehl, 1992). The 51 responses obtained in this study were thus considered sufficient for associations to be made between referral behaviour of doctors in this sample and other characteristics. There were also sufficient responses (36.4%) for a descriptive study of the conference delegates, but further research will be needed to extrapolate this to the population of general practitioners in South Africa.

Where questions were not answered by all respondents, calculations were based on numbers of participants responding to each question.

3 RESULTS

Results from the survey are presented below.

3.1 Sample demographics

Fifty one (n=51) responses were obtained from the 140 questionnaires distributed at the conference (10 delegates did not accept a questionnaire), a response rate of 36.4%. It was not possible to determine reasons for non-response as the only demographics available for the conference delegates was the number of males and females. The sample demographics are summarised in Table 1. Since not all respondents answered all questions, numbers do not always add up to 51.

3.1.1 Gender

Overall, 60% of respondents were female and 40% were male. One respondent did not answer this question. The conference was attended by 150 delegates, of which 41.3% were female. The SAMA database in 2010 listed 65.4% male doctors, so this sample was not representative of the population of doctors in South Africa.

3.1.2 Age, decade of graduation and years in practice

The majority of respondents were aged between 30 and 69, with a drop in the 40-49 age group corresponding with a dip in graduation from 1981 to 2000, and 2 respondents in both the 20-29 and 70-79 age groups.

Years in practice ranged between 4 and 50 years, with an average of 24.12 years in practice.

Fifty respondents classified their field of practice. Most of them (56%) said they were GPs, and 26% said they worked in family medicine (Table 1).

3.1.3 Practice location

All 51 respondents answered the question on practice location. Several respondents picked more than one practice location, hence numbers add up to more than 51. Doctors were evenly distributed between the private sector and the public sector at 45.1% and 41.2% respectively, with some doctors working in both sectors. 33.3% worked in urban areas and 11.8% in rural settings.

Table 1: Respondents' demographic characteristics

Variable	Number	Percent (%)
Gender		
Male	20	40
Female	30	60
***Total**	**50**	
Age		
20-29	2	3.9
30-39	14	27.5
40-49	5	9.8
50-59	14	27.5
60-69	14	27.5
70-79	2	3.9
***Total**	**51**	
Decade of graduation		
1961-1970	2	4.3
1971-1980	14	29.8
1981-1990	9	19.1
1991-2000	8	17.0
2001-2010	14	29.8
***Total**	**47**	
Qualifications		
MBChB only	21	42.9
MBChB plus one	13	26.5
MBChB plus two	9	18.4
MBChB plus three or more	6	12.2
***Total**	**49**	
Field of practice		
General practice or general medicine	28	56
Family medicine /practice /physician	13	26
Medical officer	2	4
Primary health care	2	4
Family medicine facilitator	1	2
Disability grants	1	2
Academia	1	2
Not practising	1	2
Palliative medicine	1	
***Total**	**50**	
Practice location		
Private sector	23	45.1
Public sector	21	41.2
Urban	17	33.3
Rural	6	11.8
Tertiary hospital	1	2.0
Primary care	20	39.2
Academic	3	5.9
Other	7	13.7
****Total**	**98**	

*Not all respondents answered all questions, so numbers do not add up to 51.

** Some respondents gave more than one answer so answers total more than 51

Seven (13.7%) chose the option of “Other”. Comments made under other included:

- no formal practice at the moment;
- multiple practices;
- NGO – Khayelitsha;
- UCT Student Wellness Service;
- Department of Correctional Services;
- Ireland (Public/Private).

3.1.4 Specific fields of interest of respondents

A wide range of professional interests were described by respondents. Doctors working in in the public sector cited areas such as: Occupational exposure to blood / body fluids; Palliative medicine; Infectious diseases: TB; HIV/AIDS; Trauma and Preventative care. Many of these concerns are commonly raised in public health care in South Africa (Mayosi et al., 2009).

Doctors in private practice had a wide range of interests, encompassing the specialities of Neurology, Psychiatry, Geriatrics, Ophthalmology, Travel medicine, Allergy, Sports and Infertility. Several were also interested in diseases of lifestyle, particularly diabetes mellitus and hypertension. More general interests cited by private doctors included: Integrated medicine; Female health care; Nutrition and Health.

3.2 Referral patterns of doctors to genetic services

Doctors were asked whether they had previously referred to genetic services, whether they thought they might refer in future, and whether they know how to access genetic services in South Africa. Fig. 1 illustrates the doctors’ answers. There was an almost even distribution between doctors who had used genetic services (52.9%) and those who had not (47.1%). Of those who had not referred to genetic services, one commented that Pathcare had been used for testing.

Almost all (92.2%) respondents thought they would refer to genetic services in the future and two of those who said they would not were retired.

In response to the question on whether or not they knew how to access genetic services, 49% said “yes” (with two mentioning Groote Schuur and Red Cross Hospitals), 20 (39.2%) said “no” (one had accessed through Ampath) and six said “Other”, commenting that they were not sure of the referral pathways, would ask at clinics, and were not aware of all services.

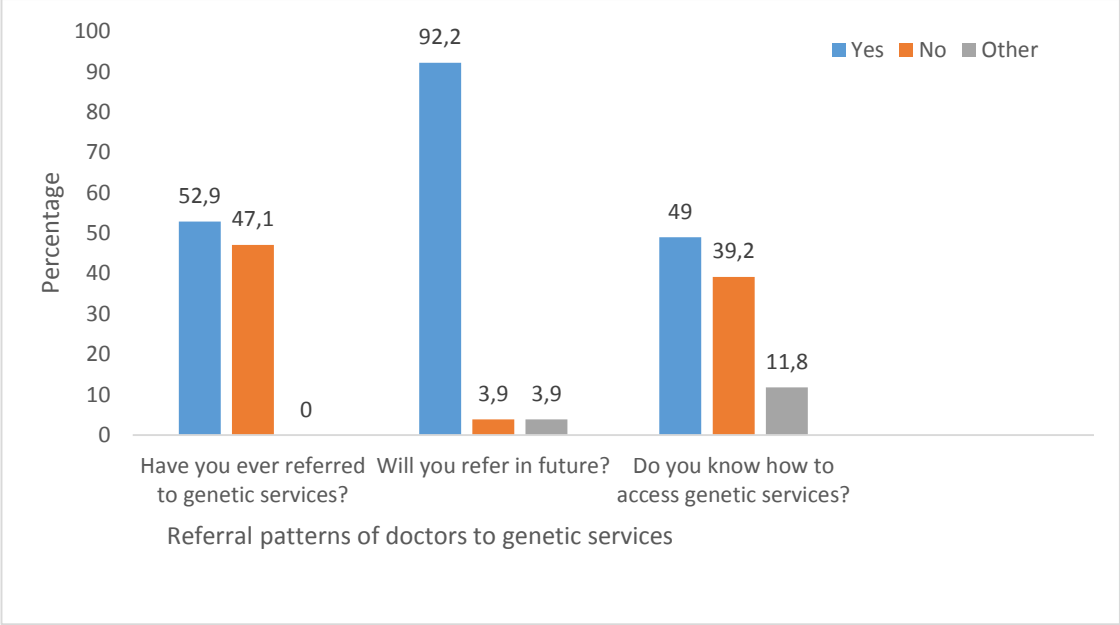

Fig. 1: Respondents' referral patterns to genetic services

Although only 24 doctors said they had not previously referred to genetic services, 43 answered the checkbox question: "If you haven't used genetic services previously, why not?" Their replies are summarised in fig. 2 and a full list of comments appears in Appendix E. This question was placed towards the end of the questionnaire and it is possible that respondents had forgotten their first answer, or that they wanted to comment on their use of genetic services. Many respondents commented on their chosen answer even though only the option "Other" had a comment box. This helped to explain their choices. Almost half of the doctors (44.7%) said that genetic services were "Difficult to access". This is in close agreement with the 39.2% who do not know how to access services (fig. 1). Another third (28.9%) said they had not found it necessary to refer to genetic services, and 26.3% gave "Other" as a reason for not referring.

Lack of familiarity with genetics and genetic services was a common theme across the respondents' comments, and indeed across many of the questions, no matter what choice they made. Comments under "Difficult to access" included:

- "??because my knowledge limited"

Doctors that selected "Not necessary", commented:

- "Patients never requested referrals"
- "Patients already diagnosed when I see them".

Comments under Other included:

- "Didn't know I could"
- "Not sure what they are offering."

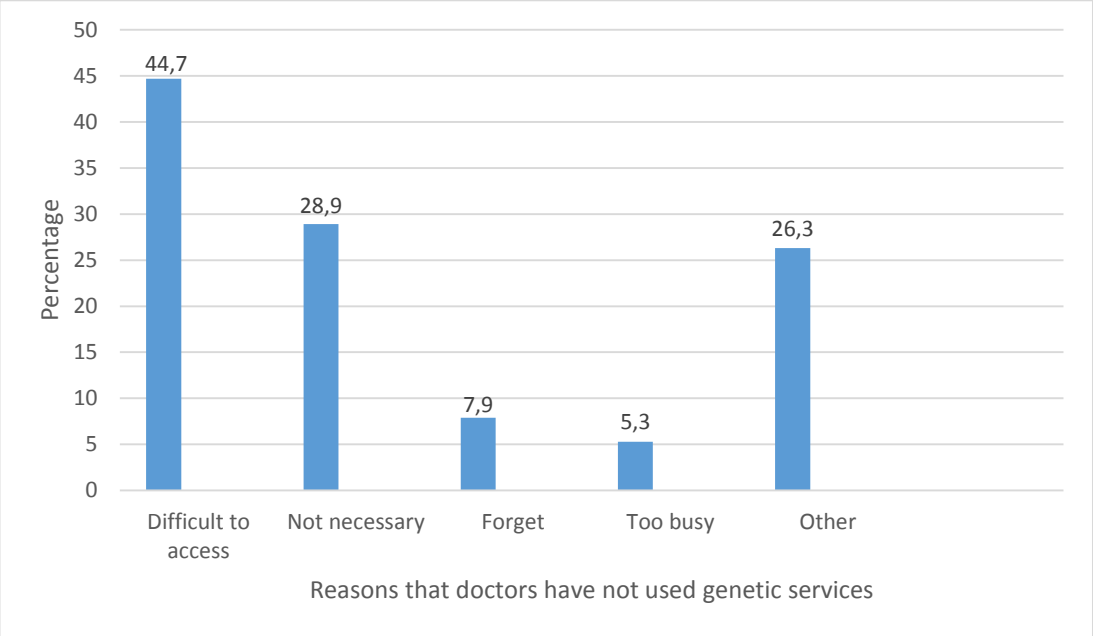

Fig. 2: Doctors' reasons for not using genetic services previously

3.3 Doctors' education in genetics

Table 2 summarises the amount and form of genetics education received by respondents during undergraduate training and since graduation.

3.3.1 Undergraduate genetic education

All 51 respondents answered the question "How much genetics education did you receive in undergraduate training?" with 50.5% indicating they had less than 10 hours' of undergraduate genetic education. Almost 20%, however, said that they had more than 10 hours of undergraduate genetic training. Three respondents commented next to their answer that they could not remember how much education they had received:

- "Cannot remember how much, had some lectures".
- "Difficult to quantify. Incorporated as a short lecture or part of a lecture in each block e.g. O&G (obstetrics and gynaecology), Medicine etc."

It is, however, impossible to know whether participants' answers are accurate when questions require recall of events.

All respondents answered “How was this education offered?” with some choosing more than one option. As expected, since lectures are the most common form of tertiary education, lectures were overwhelmingly the most frequently chosen form of education (90.2%). Interesting and specific comments under “Other”, indicating the importance of education being offered in different forms, were:

- “Consultations”
- “Visit to the genetic counselling centre.”

3.3.2 Postgraduate genetic education

“How much genetics education have you received since graduation?” was answered by all respondents. An almost equal number said they had received no (29.4%) or 0 to 3 hours (33.3%) of genetics education since graduation, so in total 62.7% had fewer than 3 hours of education in genetics since graduation. Only one respondent implied that she had many hours of genetic education, saying under “Other”:

- “I worked in Genetics full-time and part-time in the 80s.”

Once again, respondents reported that most of their postgraduate education to date was in the form of lectures, with almost half (49%) selecting this option. The attractiveness of attending educational activities for continuing professional development (CPD) (also known as continuing medical education - CME) points was illustrated by 37.3% saying they had earned points for genetics education. The relevance of journals and congresses in self-study were mentioned, and one respondent said they had “clinical exposure” to genetics since graduation. This shows that a wide range of educational activities are acceptable for postgraduate education.

Table 2 Genetic education of respondents

Variable	Number	Percent (%) **
Amount of genetics education in undergraduate training		
None	3	5.9
1 or 2 hours	10	19.6
3 to 5 hours	13	25.5
5 to 10 hours	7	13.7
>10 hours	10	19.6
Don't know	6	11.8
Other	2	6.9
Total*	**52**	
Form of genetics education in undergraduate training		
Not applicable	3	5.9
Lectures	46	90.2
Meetings	4	7.8
Self-study	7	13.7
Short course	1	2.0
Other	5	9.8
Total*	**66**	
Amount of genetics education since graduation		
None	15	29.4
0 to 3 hours	17	33.3
3 to 5 hours	9	17.6
>5 hours	7	13.7
Other	4	7.8
Total*	**52**	
Form of genetics education since graduation		
Not applicable	14	27.5
Lectures	25	49.0
Meetings	9	17.6
CPD points	19	37.3
Self-study	10	19.6
Short course	3	5.9
Other	4	7.8
Total*	**84**	

* Some respondents gave more than one answer so answers total more than 51.

**Percentage out of number of respondents to each question, not number of answers

3.3.3 Doctors' needs from genetic education

A variety of answers were obtained to the open-ended question: "What sort of further education on genetics, if any, would you like?" These were grouped into 4 themes. Several doctors made comments that fitted more than one theme, so numbers do not add up. A full list of quotes appears in Appendix E. The four themes were:

1. Forum for education (n=16, 31.4%)
2. Topics (n=10, 19.6%)
3. Resources for self-study (n=10, 19.6%)
4. Not applicable (n=1, 2.0%)

Theme 1: Forum for education. Most doctors (n=16, 31.4%) explained how and where they would like to receive genetic education. Many doctors felt that there was not sufficient opportunity for genetic education, and some realised the importance of updating their genetic knowledge through various platforms, including conferences, tutorials, short courses, CME activities and refresher courses, e.g.

- "Education on genetics does not happen often. Congresses and medical journals should bring more of this".
- "Part-time / distance learning with contact sessions - towards a certificate."
- "Short courses, starting with the basics, including information about indications for referral and contact details of referral centres."
- "Refresher courses which teach about the advances in diagnosis, prognosis and management of genetic diseases"

One respondent remarked that he/she would like more hands-on experience of genetic services.

- "What genetic counsellors do? To sit in on counselling sessions. Common genetic conditions - to attend a genetic clinic and see these conditions."

Theme 2: Topics. Ten respondents (19.6%) mentioned that they would like to know more about genetics and when it would be useful to their practice and many indicated that their understanding of basic concepts in genetics including genetic tests, genetic conditions and the function of genetic services was lacking, e.g.

- "A basic course with presentation of information useful in the GP setting".
- "Reminders of genetic conditions to help us pick them up"

- “Information about indications for referral and contact details of referral centres”.

A lack of understanding of the role of genetic counsellors was mentioned by two respondents, e.g.

- “What do genetic counsellors do?”

A number of participants (n=7) wanted help with patient management as they felt that this would allow them to improve patient care, e.g.

- “GP oriented courses to help me better serve my patients and their communities”.

Finally, 3 wanted current knowledge, e.g.

- “Updates on new developments” .

Theme 3: Resources for self-study. Ten respondents (19.6%) felt that they would like access to materials for self-study, which would be easily available when they needed it. Three people mentioned the internet or technology, e.g.

- “Access to a good website”
- “Phone apps”.

Another 3 requested print media, including journal articles, and one said:

- “Have a booklet in my practice covering "genetics 101".

Theme 4: Not applicable. One doctor was retired and commented:

- “Too late!”

These answers gave insight into how respondents like to learn, as well as what sorts of topics in genetics are of interest to them.

3.4 Knowledge – what doctors know and what they think they know about genetics

Overall, doctors rated their genetic knowledge as very poor, and this was somewhat confirmed by their answers to the practical knowledge questions, with an average score of 39.6% obtained for the 11 questions that were scored, although this may not indicate very poor knowledge of genetics.

The majority of respondents (n=28, 54.9%) were not confident in their knowledge of genetics when measured on a Likert scale. This followed a declining trend, with 14 (29.4%) being slightly confident, 3 (5.9%) somewhat confident and only one feeling confident in their genetic knowledge. No respondents rated themselves as very confident (fig. 3).

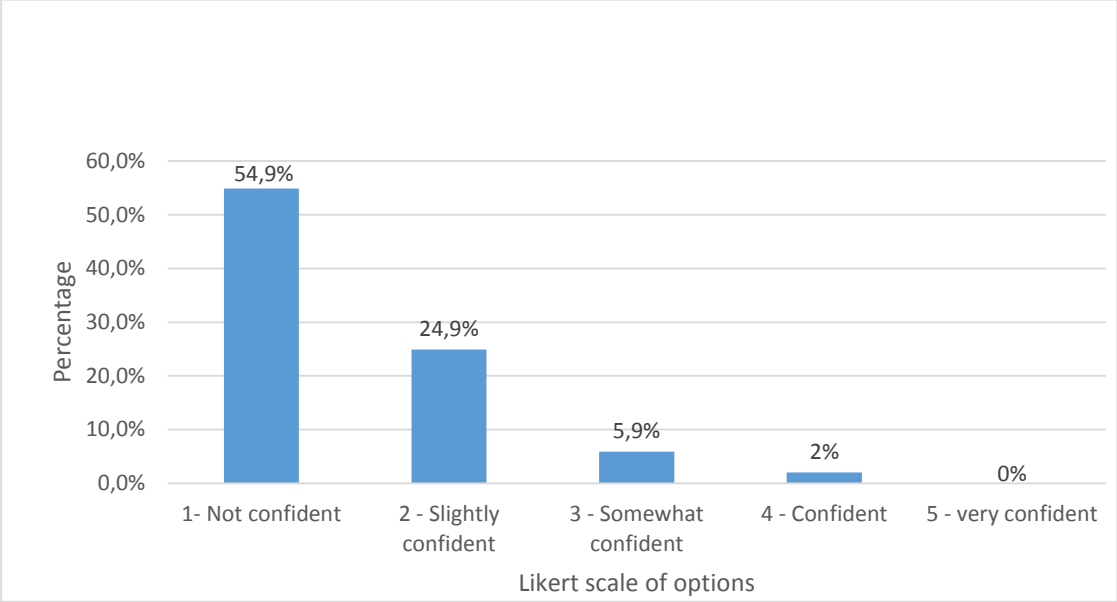

Fig. 3 Respondents' self-rating of genetic knowledge

The doctors' feelings about their knowledge of genetics were further explored in the open-ended question: "What do you think about your knowledge of genetics?" (Full table of comments appears in Appendix E). The resulting answers were rated as:

1. Very poor
2. Could be better
3. Good

Theme 1: Very poor. The majority of doctors (n=36, 70.6%) felt that they did not have enough knowledge to help patients with genetic conditions. As one participant said:

- "I have a basic knowledge of a few conditions and principles of genetics but feel that I do not have adequate knowledge to confidently diagnose and manage all genetic conditions."

Another doctor thought that their paucity of genetic knowledge was unacceptable:

- "Virtually zero knowledge which is not really acceptable"

On a positive note, a few (n=6, 11.8%) thought that although their knowledge was poor, they felt that they could rectify this lack of knowledge with education, and thought that increased knowledge might enable them to diagnose more genetic conditions:

- "I would love to learn and know more. I realise I have many 'gaps' BUT usually would ask for advice/ help from specialists / colleagues. May diagnose more conditions with more awareness of testing / implications / presentation."

There were 2 doctors who felt that their knowledge was so poor that it was:

- "Non-existent"
- "Woeful."

Theme 2: Could be better. Four individuals (7.8%) were reasonably confident in their knowledge, but felt that they could still learn:

- "Could always improve, probably in the context of ongoing developments in the field."

Theme 3: Good. Only two respondents rated their knowledge as good and one attributed this to:

- "Good teaching in undergraduate at UCT."

3.4.1 Knowledge of conditions seen in practice

This tabulated question asked the doctors to list inherited conditions that they had seen in their practices. For each condition they listed, they would then choose whether a genetic test is available, the mode of inheritance of the condition, and whether or not it would have a significant impact on a patient's life (Table 3). A total of 45 different conditions were listed by 37 (72.5%) respondents. Between 1 and 8 conditions were listed per respondent, with an average of 3.62 conditions mentioned per doctor. Knowledge of conditions seen in practice was scored on the basis of 1 mark per recognised condition, 1 mark if the correct answer was given for genetic test availability, and 1 mark for correct mode of inheritance. This was difficult to compare as some respondents only listed 1 condition while others listed several. Overall, participants scored an average mark of 70.6% (SD 21.2), with a mode of 66.7% and a range between 20% and 100%, which indicates that they have a good knowledge of genetic conditions that they have seen in practice.

Fig. 4 shows the 10 most frequently cited conditions reported as being seen in practice by responding doctors. Down syndrome was the most frequently reported condition, followed

by cystic fibrosis, breast cancer, sickle cell anaemia, haemophilia, Huntington disease, albinism, thalassemia, familial hypercholesterolaemia and Marfan syndrome. The other 35 conditions mentioned were seen by less than 10% of respondents.

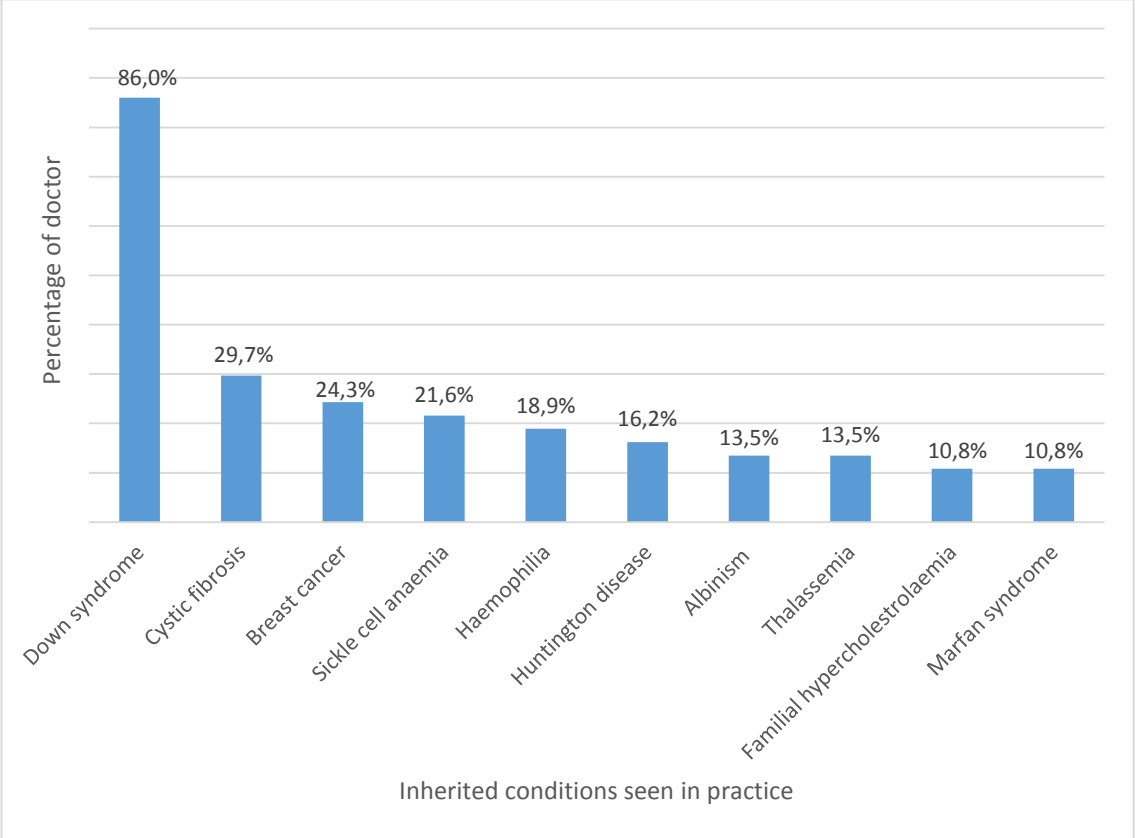

Fig. 4 Ten inherited conditions seen most frequently in practice

Table 3 is a summary of answers obtained for the ten most commonly seen inherited conditions (Appendix E shows a complete table of answers for all 45 conditions).

Of the 139 answers citing conditions seen in practice, 94.2% (n=130) correctly identified conditions with a genetic or congenital component. The other 9 conditions mentioned by respondents are not commonly seen in the genetic clinic, e.g. attention deficit disorder, asthma, diabetes, hypertension and Stevens Johnson syndrome (these can be seen in the full table in Appendix E). Spinal muscular atrophy (SMA) was cited by one doctor, who also correctly identified the inheritance of the condition as autosomal recessive, but was not aware of the existence of a genetic test for the disorder.

Similarly, of the potential 115 correct answers for availability of a genetic test, 69.6% (n=80) chose correctly. Finally, from 129 answers given for inheritance patterns, 58.1% (n=75) selected the correct mode of inheritance. Almost all conditions (except haemophilia and thalassemia, which can be lethal) were considered to significantly impact life.

Table 3: Knowledge of genetic conditions seen by doctors in practice (conditions listed by respondents)

Name of inherited condition cited by doctors as being seen in practice	Number citing condition	Number choosing: Genetic test available	No. choosing: Mode of inheritance (correct answer highlighted)					Number choosing: Significant impact on life	Percent (%) correct answers	
			Autosomal Dominant	Autosomal Recessive	X-Linked	Chromosomal	Other		Testing	Inheritance
Down syndrome	32	28	2	2	1	26	1	27	88	81
Cystic fibrosis	11	8	2	7				10	73	64
Breast cancer	9	8	3		2		2	6	89	33
Sickle Cell Anaemia	8	6	1	4				7	75	50
Haemophilia	7	7		2	4			2	100	57
Huntington disease	6	5	5			1		5	83	83
Albinism	5	4		3		1	2	4	80	60
Thalassemia	5	0	1	3				0	0	60
Familial Hypercholestrolaemia	4	2	2	2		1		3	50	50
Marfan Syndrome	4	1	3					4	25	75

These results indicate that GPs have seen a range of genetic conditions and they have increased knowledge about these disorders, although only 58.1% of respondents knew how these conditions are inherited, implying that their genetic knowledge, even of familiar conditions, could be improved.

3.4.2 Knowledge and awareness of 10 inherited conditions

Only 47 of the 51 respondents (92.2%) attempted the table pre-populated with a list of 10 conditions and not all answers were complete (Table 4). Percentages were calculated based on the 47 that filled in the table, although the remaining 4 respondents may not have known the conditions at all and thus percentages would have been lower. Correct answers are marked with a √ and incorrect answers with a X. The column for "Metabolic testing available" was removed from the final analysis as the interpretation was ambiguous (Appendix 5 shows the full table).

All 47 doctors were aware of breast cancer and all but one knew of Down syndrome, as expected, as these conditions had been seen in practice by more than 80% of respondents. Cystic fibrosis, haemophilia, sickle cell disease and albinism were all known to more than 70% of the doctors but only cystic fibrosis had been seen by more than 50% of doctors. Lynch syndrome was the least recognised condition, known by only 31.9% of respondents and only seen by 8.5%. Thus, awareness of inherited conditions seems to be closely linked to the experience of seeing patients with that condition, although more doctors are aware of all conditions than had actually seen them in practice. This is in agreement with the previous result, where doctors were well informed about conditions they had seen in practice.

Cognisance of availability of genetic tests was variable. Most respondents (82.6%) knew that a genetic test was available for Down syndrome, but only 22.7% knew that there was a genetic test for spinal muscular atrophy. Surprisingly, only 59.6% were aware that a genetic test is available for a *BRCA* mutation. The words "inherited breast cancer" were not specified in the table and it is acknowledged that this may have caused confusion.

Analysis of answers on the availability of predictive and carrier tests indicated that doctors did not know the difference between these tests as the almost 50/50 split between correct (191) and incorrect (189) answers could have been randomly generated. Several respondents wrote comments indicating that they thought either predictive or carrier testing implied prenatal testing e.g. comments such as "antenatal" or "in utero" under "carrier testing available" for Huntington disease and Down syndrome; for cystic fibrosis one respondent wrote "antenatal" under "carrier testing available" and under "predictive testing available"

wrote “antenatal screen”. This indicates that doctors have no real understanding of the meaning of the terms “predictive” and “carrier” testing.

Table 4: Knowledge and awareness by doctors of 10 inherited conditions (table pre-populated with conditions)

Name of condition	*Know the condition	*Seen the condition in practice	Genetic test available	Carrier testing available	Predictive testing available	Other family members at risk
Breast cancer	47(100.0%)	43 (91.5%)	√28 (59.6%)	X21(44.7%)	√19 (40.4%)	√32 (68.1%)
Down syndrome	46 (97.9%)	41 (87.2%)	√38 (82.6%)	X5 (10.9%)	X14 (30.4%)	√5 (10.9%)
Cystic fibrosis	43 (91.5%)	24 (51.1%)	√28(65.1%)	√15 (34.9%)	X9 (20.9%)	√17 (39.5%)
Haemophilia	40 (85.1%)	23 (48.9%)	√29 (72.5%)	√21 (52.5%)	X10 (25.0%)	√19 (47.5%)
Sickle cell disease	36 (76.6%)	18 (38.3%)	√19 (52.8%)	√14 (38.9%)	X4 (11.1%)	√17 (47.2%)
Albinism	35 (74.5%)	21 (44.7%)	√9 (25.7%)	√6 (17.1%)	X4 (11.4%)	√9 (25.7%)
Huntington disease	32 (68.1%)	16 (34.0%)	√21 (65.6%)	X17 (53.1%)	√5 (15.6%)	√23 (71.9%)
Spinal muscular atrophy	22 (46.8%)	8 (17.0%)	√5 (22.7%)	√4 (18.2%)	X1(4.5%)	√5 (22.7%)
Spinocerebellar ataxia	16 (34.0%)	6 (12.8%)	√4 (25.0%)	X1 (6.3%)	√0 (0.0%)	√4 (25.0%)
Lynch syndrome (hereditary non-polyposis colon cancer)	15 (31.9%)	4 (8.5%)	√6 (40.0%)	X3 (20.0%)	√2 (13.3%)	√10 (66.7%)

*Percentages for Know the condition and Seen the condition as a proportion of the 47 respondents who filled in the table. All other percentages as a proportion of the number of people who knew the condition.

There was a general lack of awareness that *all* inherited conditions place family members at risk (apart from chromosomal conditions in most cases). No respondents wrote that all the conditions (except for Down syndrome) would place other family members at risk. One respondent, however, understood translocation forms of Down syndrome – saying "it depends" under the column: "Other family members at risk". One doctor said "maybe" for the risk to other family members of inherited breast cancer.

The results of this question on genetic conditions confirmed the doctors' self-reported comments (section 3.4) that they need basic education on genetic concepts. Pre-populating table 4 with a list of conditions may have acted as a reminder to the doctors, as many more doctors confirmed seeing all the conditions than had cited them from recall in the previous question (table 3). Some doctors may have added these conditions to the previous table later.

3.4.3 Awareness of prenatal diagnosis of genetic conditions

The multiple choice question on what is involved in prenatal diagnosis of genetic conditions was answered by 50 of the 51 doctors. Fig. 5 shows the percentage of doctors choosing each option. All 7 options given were correct, but only 26% (n=13) chose all these possibilities (table 5). The most common combination chosen was family history, amniocentesis and ultrasound scan, with 88% (n=44) picking at least these three options together. Amniocentesis was the most popular choice (n=49, 98%), followed closely by family history (n=48, 96%), indicating a good understanding of the importance of family history in detecting genetic conditions. Just under 60% of respondents ticked non-invasive prenatal testing, meaning knowledge of this new technology is becoming widespread.

Table 5: Number of correct prenatal test options chose by respondents

Answers	Number	(%)
7 correct	13	26.0
6 correct	6	12.0
5 correct	7	14.0
4 correct	11	22.0
3 correct	11	22.0
2 correct	1	2.0
1 correct	1	2.0
Total	50	100

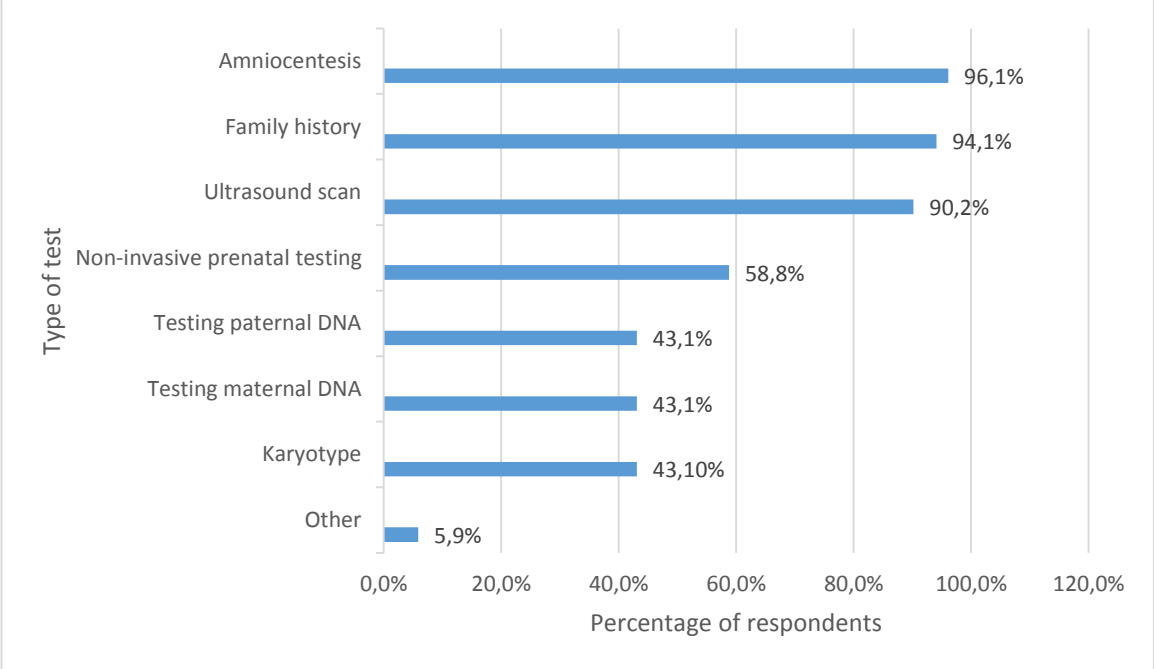

Fig 5. Percentage respondents choosing each prenatal test option

3.4.4 Awareness and knowledge of inherited breast cancer

Four questions were asked about inherited breast cancer:

1. A multiple choice question on whether maternal or paternal history, or neither or both (which is correct) were more important in inherited breast cancer.
2. A multiple choice question asking what percentage of breast cancer is inherited and offering the following options: 1%, 5%, 10% (the correct answer – Nelson et al., 2014), 15%, 25%, 50%, 100% or "Other".
3. A question asking whether a male diagnosed with breast cancer is at risk of a *BRCA* mutation even without family history. Options were "Yes" (which is correct), "No" and "Other".
4. A multiple choice question on how a patient with family history that tests negative for a *BRCA* mutation should be managed with the choices: "As someone at population risk", "As someone at higher than population risk" (correct) and "Other".

Respondents demonstrated limited knowledge about inherited breast cancer. Most respondents (n=35, 70%) chose maternal history as most important in inherited breast cancer, not understanding that, although breast cancer is a disease usually found in women, this does not imply that the gene cannot be passed through the paternal line. Only 30%

(n=15) of respondents chose the correct answer of both paternal and maternal history being important.

Almost half of the doctors (n=22, 43.1%) chose a number greater than the correct answer of 10% for the second question: the percentage of breast cancer that is inherited. Most (n=14, 28.2%) thought that 25% of breast cancer is inherited and 21.7% (n=11) chose the correct answer (fig. 6). No one thought that only 1% of breast cancer is inherited. Comments from those that selected "Other" (n= 9, 17.4%), were "not sure" or "guessing".

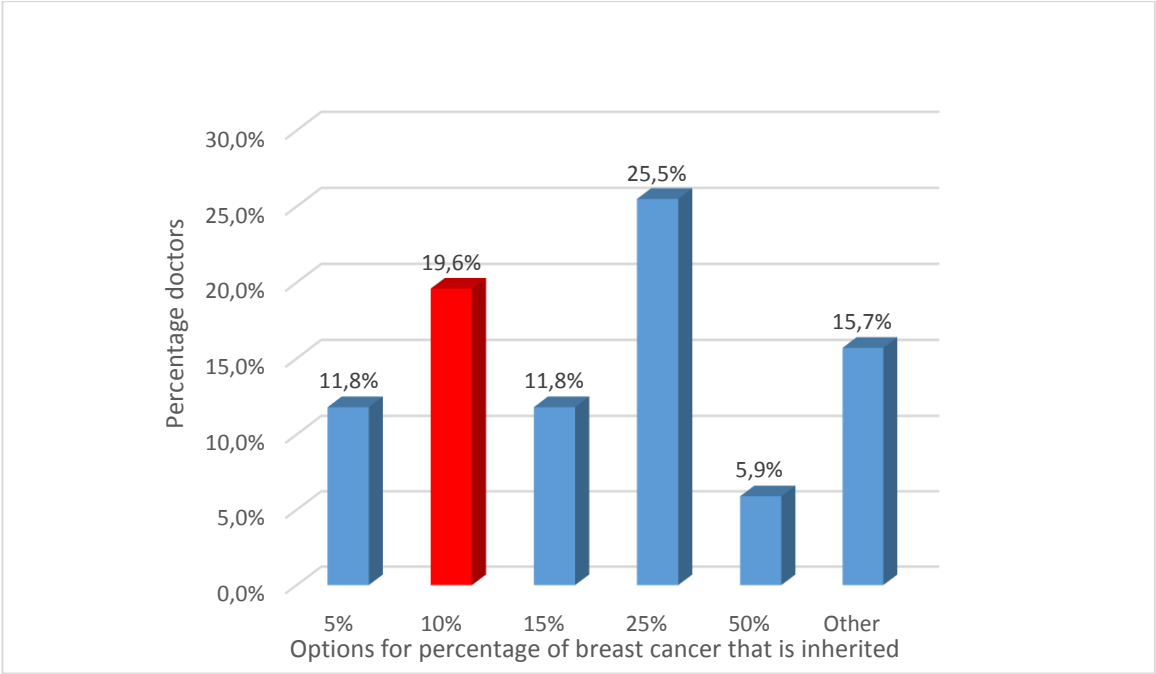

Fig. 6 Respondents' perception of percentage of breast cancer that is inherited

Fifty percent of respondents (n=25) correctly answered the third question. A patient with family history that screens negative for a *BRCA* mutation should be treated "As someone at higher than population risk". This shows understanding that family history alone is a risk factor for breast cancer, possibly due to underlying genetic factors that have not yet been discovered. "As someone at population risk" was chosen by 39.6% (n=20), with 10.4% (n=5) selecting "Other". Several doctors qualified their answers (either under "Other" or, in one case, "As someone at population risk"), commenting: "Depending on other risk factors" and "DO NOT KNOW!" One mentioned "Depends on age presentation. If below 40 higher risk", implying an appreciation of the role of a directed family history in the management of a patient with a family history of breast cancer

Thirty (65%) of the 46 respondents that answered the fourth question correctly said that a male diagnosed with breast cancer is at risk for an inherited *BRCA* mutation even without

family history. Only 13.0% (n=6) answered “No”, but 21.7% (n=10) chose “Other” and said they “did not know”, or were “unsure”.

3.4.5. Overall knowledge score

The total number of correct answers for each participant for questions 18, 19, 20, 21, 22, 23, 26, 27 were scored out of 45. The average knowledge score per participant was 39.9%, with scores ranging between 6.7% and 71.1% and a standard deviation of 15.7%, and 35.6% being the mode (most frequent answer). These scores indicate that the participants’ knowledge may not be very poor, but still illustrate the lack of depth shown by answers to the various questions and highlight the need for more genetic education and awareness programmes for GPs.

3.4.6. Knowledge and attitudes concerning genomics

DTC genetic testing is new technology that 82.4% (n=42) of the respondents do not yet know about. Comments on this innovation in the question “Do you think you might be approached to interpret results of a direct-to-consumer test in future?” indicated that participants were aware that they need to know more and almost half (n=23, 46%) thought they might be approached to interpret these tests in future despite not knowing about them at present, showing an understanding that new technology is usually assimilated over time. Nineteen doctors (38%) did not expect to have to interpret these tests in future, and 2% said they did not know. The remaining 14% (n=7) of doctors chose “Other” and made comments such as “Possibly when I know more”, “You never can tell!”, “Probably” and “Do not know what it is”, again indicating a need for education on this new technology.

A chi-squared test showed no relationship between those who know what DTC testing is and those who thought they might be approached to interpret results ($p=0.134$). However, only 9 people (17.6%) knew about DTC testing, so this association could be skewed. Of the 42 (82.4%) who didn’t know about DTC testing, 18 (42.9%) said they expected to be asked to interpret results in future and another 18 (42.9%) said they did not expect to be asked. Seven (77.8%) of the 9 respondents who did know about DTC testing said they expected to be asked to interpret results and two (22.2%) said they did not expect to be asked, with one commenting “Rare if at all (deep rural area hospital)” and the other saying “Work in the public service”, indicating that the doctors feel that this type of testing is unlikely to be accessed by rural people or people who use public health care.

The abbreviation, VOUS, was only recognised by one third of respondents (n=17) as meaning “Variant of unknown significance”, more than half (n=29) said they did not know what VOUS

means and the remaining 10% chose incorrect answers (fig. 7). This is another indication of poor knowledge of genomics as this abbreviation is very commonly used in DTC testing and genetics research and indicates: “An alteration in the normal sequence of a gene, the significance of which is unclear until further study of the genotype and corresponding phenotype in a sufficiently large population” (Pagon, et al. 2015).

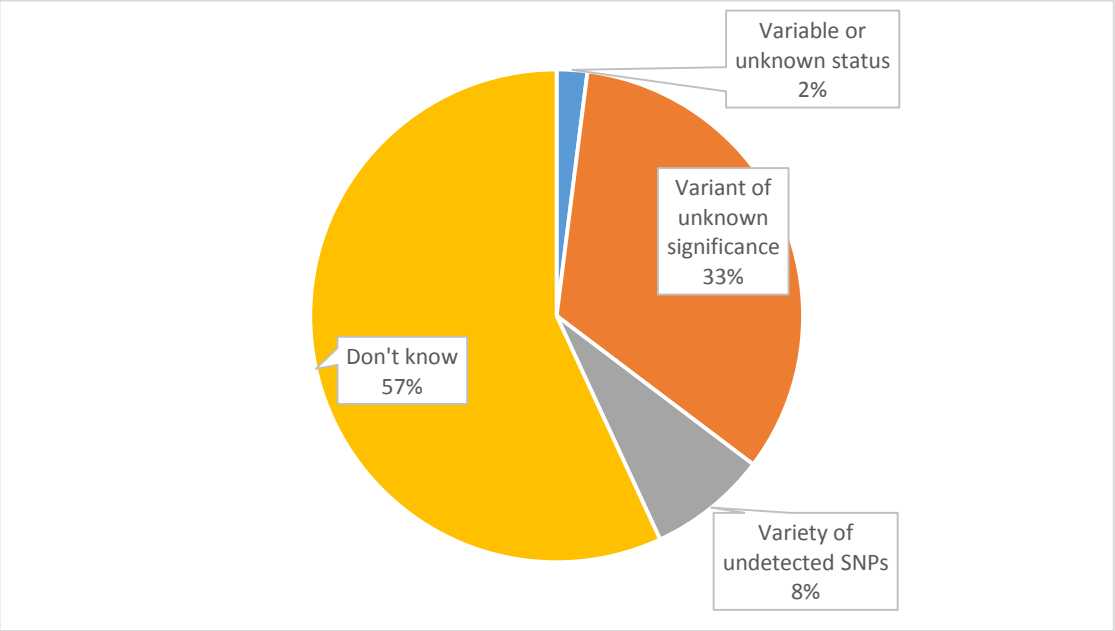

Fig. 7 Percentage of respondents choosing different answers for the meaning of VOUS

Forty five responses were obtained for the open-ended question “What would you do if a patient approached you with results from a direct-to-consumer genetic test?” Four themes were obtained from content analysis of the responses, some which applied to more than one theme, so numbers do not add up to 45.

The themes were:

1. Ask for advice / seek information (n=26, 57.8%)
2. Refer (n=20, 44.4%)
3. Interpret results (n=12, 26,7%)
4. Don’t know (n=4, 8.9%)

Theme 1: Ask for advice /seek information: The majority of participants (57.8%) thought that, with some help from colleagues or genetic services, they would be able to discuss the results from a DTC test with a patient. One doctor said:

- "I would attempt to contact a colleague at the Genetic services to ask for assistance / advice on any further steps to be done / taken to assist the patient."

Two indicated that they were nervous about interpreting these results, and so would consult the internet or colleagues for assistance:

- "Panic - Need to obtain information therefore go to internet."
- "Call for help!!"

Another 4 felt that the company supplying the tests would be able to explain the results:

- "Refer back to direct-to-consumer tester."

Theme 2: Refer. Twenty respondents (44.4%) felt that they did not have enough knowledge or experience to offer their patients a proper consultation on results from a DTC test and said they would refer to either colleagues or specialists, or to genetic services:

- "I would refer them to specialist and genetic services. This information should be given to the patient under proper guidance and explanation."
- "Depends on what the issue is – Refer the problem.

Theme 3: Interpret the results. Twelve participants thought they could interpret the results themselves without mentioning the need for consulting with specialist services or colleagues:

- "Consider my own clinical assessment of patient. It would help if the parameters of the validity of the tests are cited with the results."
- "May ask patient to come back for another appointment to get the best information to give the patient the best answers".

Theme 4: Don't know. Only four doctors acknowledged that they didn't know what they would do if approached with results from a DTC test:

- "I have no knowledge at all."

Genetic services, which would be the most likely place to find good information on DTC testing, were only mentioned by 17 respondents, although only 9 respondents originally said they knew about DTC testing. More doctors said that they would seek advice or information from colleagues (the most common source of information for doctors according to Bennett et al., 2006), the DTC test company or through self-study (often the internet) than would try and call genetic services. Even the doctor who said he/she would "panic" followed this by

saying he/she would search the internet for information. These answers are interesting as the doctors appear to be quite confident when asked what they would do with a DTC test query, in contrast to their stated lack of confidence in their genetic knowledge (section 3.4).

Doctors were then asked whether they think that pharmacogenomics would have an impact on prescribing medications in future, in a multiple choice question with answers "Yes", "No" or "Other". Overall almost 90% (n=45) of the doctors thought pharmacogenomics would have an impact on prescribing medications in future, with the remainder saying they did not know about pharmacogenomics, remarking: "Don't know anything about this" and "Not sure what it is" (see fig. 8). Nobody chose "No", again indicating an understanding that new technology will impact practice eventually.

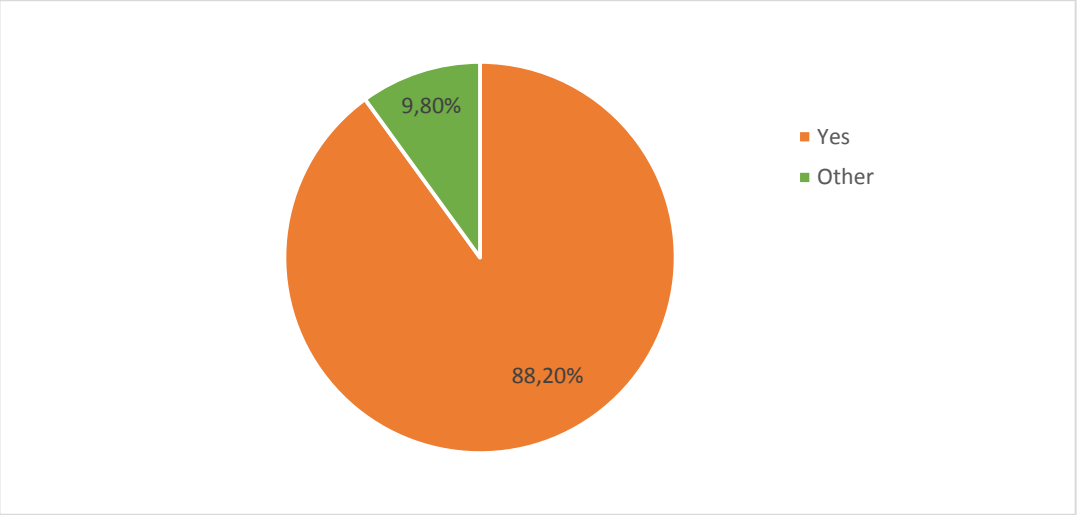

Fig. 8 Percentage of respondents who think pharmacogenomics will have an impact on prescribing medications in future.

Thirty nine doctors responded to the open-ended question: "How can we make genomics more accessible to clinicians in South Africa? Genomics refers to whole genome science and includes genome sequencing that is already available to consumers online." (A full table of responses appears in Appendix E). Responses were similar to those on what sort of education doctors would like to receive in future, with lack of awareness and need for more information included as major themes.

Four themes were generated from respondents' comments:

1. Raising awareness (n=18, 46.2%)
2. Education and information (n=18, 46.2%)
3. Increase accessibility (n=8, 20.5%)
4. Don't know (n=5, 12.8%)

Theme 1: Raising awareness. Five respondents (12.8%) suggested advertising and marketing would increase accessibility to genomics – indicating that they think of genomics as a commercial entity, possibly due to confusion with DTC testing. Comments included:

- "Advertise widely. Reduce price and make it affordable for man in the street."

Six participants (15.4%) suggested the internet as an awareness tool:

- "Internet access promoted by SAMA."

Another 5 (12.8%) mentioned raising awareness through other media such as magazines and telephonic support.

Theme 2: Education and information. Nine comments (23.1%) cited education as the best way of increasing accessibility to genomics, feeling that the subject matter is complex:

- "Educate! Educate! Educate!"
- "Make subject matter more user friendly."
- "Clarify relevance which I understand and which can help me explain to patients".

Six participants (15.4%) suggested lectures, workshops and CPD activities would increase understanding of genomics:

- "Through this kind of lecturing venue."

Other respondents (n=5, 12.8%) commented on the need for more information on genomics:

- "Give us more information re services + cost + availability - via email / post"

Theme 3: Increase accessibility. Eight doctors (20.5%) felt there was insufficient contact with genetics and genomics:

- "1. More visibility 2. Contact numbers to be held in surgeries 3. Genetics contact".
- "Perhaps informing clinicians firstly of this availability and means to access it".

Theme 4: Don't know. Five people had:

- NO IDEA".

3.4.7 Knowledge of the ethics of testing children under 18 for genetic conditions

The checkbox question: "When is it appropriate to refer a child under the age of 18 for a genetic test?" (Fig. 9) – had 3 correct answers: "when a child requests a test" (under specific circumstances), "when a child is symptomatic" and "when treatment or management is available that could prevent or delay the onset of a genetic condition". The other 4 options: "when a second degree relative has a genetic condition", "when a sibling has a genetic condition" "when a parent has a genetic condition" and "when parents request a test" are not considered to be appropriate when testing children for genetic conditions that could impact their lives physically, emotionally and socially.

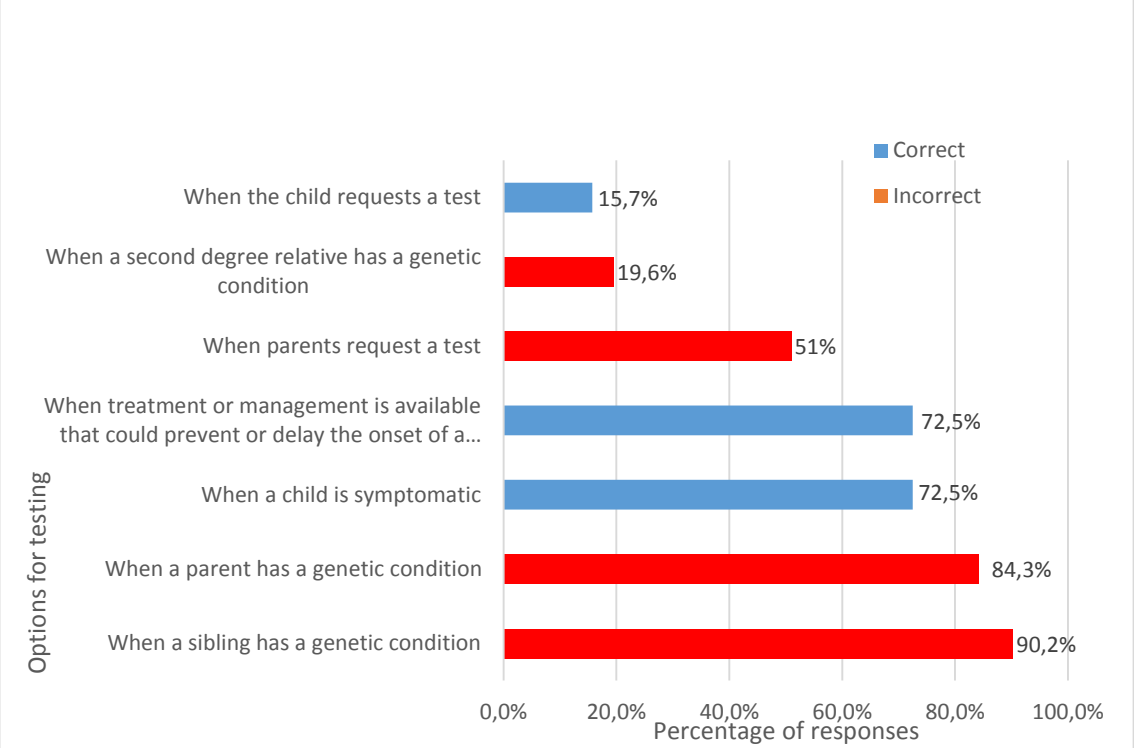

Fig. 9 Doctors' responses to when it is appropriate to test children under the age of 18 for a genetic condition

All respondents answered this question. The most frequently chosen correct answers were "when treatment or management is available that could prevent or delay the onset of a genetic condition" (n=37, 72.5%) and "when a child is symptomatic" (n=37, 72.5%), but just 4% (n=2) chose these only two answers and did not choose any incorrect options.

The most frequently chosen options overall were “when a sibling has a genetic condition” (n= 46, 90.2%), and “when a parent has a genetic condition” (n=43, 84.3%), neither of which are correct.

When analysing which answers were chosen together, 8 out of the 50 respondents (16%) chose all 3 correct answers, but they all chose either 3 or all 4 incorrect options as well, indicating little real understanding of the ethical issues behind genetic testing of minors. There is also a possibility that the question was not properly understood by the doctors.

3.5 Knowledge and attitudes of doctors concerning genetic services

3.5.1 Which medical professionals should refer to genetic services?

All respondents answered this open-ended question and most (n=37, 72.5%) thought that all medical professionals should be able to refer to genetic services. This indicates that the doctors understood that patients with genetic conditions could be seen in many health care settings. Seven groups of medical professionals were recorded from comments about who should be able to refer to genetic services. (A full table of comments appears in Appendix E).

Group 1: All. Thirty seven respondents (72.5%) thought all medical professionals should be able to refer to genetic services:

- “Any medical professional picking up / suspecting a genetic abnormality / condition in any patient. Whether a surgeon, GP, neurologist etc. anyone can diagnose a genetic condition”.

Five (9.8%) remarked that genetic services should be available whenever necessary:

- “Where there is a need or special case”.

Group 2: Specialists. Fifteen (29.4%) felt that specialists would probably diagnose genetic conditions and refer on to genetic services:

- “Specialists should be the medical professionals to refer. Only they have all the details, full diagnosis. They would also decide about further treatment/ prevention/ management”.
- “Those who diagnose genetic conditions most likely specialists rather than GPs”.

Under this category, the following specialisations were named: Paediatricians (9), Gynaecologists/obstetricians (8), Internal medicine physicians (4), Surgeons (2). Neonatologists, Neurologists, Oncologists and Psychiatrists were each mentioned once.

Group 3: General practitioners. Eighteen (25.3%) respondents thought that GPs were particularly likely to see patients with genetic conditions and need to refer them to genetic services:

"GPs especially".

Group 4: Nurses. Nurses were also seen as important sources of referral to genetic services and were cited 9 times (17.6%):

- "Nurses in medical practice"
- "Professional nurses (in discussion with doctors)".

Group 5: Institutions. One person mentioned Tygerberg and Groote Schuur hospitals.

Group 6: Pathology laboratories (1)

Group 7: Allied services. One respondent took a broader view of who should refer, mentioning:

- "Psychologists, OTs (occupational therapists) and Physios".

3.5.2 What doctors think is the role of genetic counsellors

Genetic counsellor's roles were described by all 51 respondents and fitted into the four categories of skills (not roles) mentioned by Uhlmann, Schuette & Yashar (2009). These skills were used as a guide to categorise responses into 4 themes:

1. Theme 1: Critical thinking skills (n=38, 74.5%)
2. Theme 2: Interpersonal, counselling and psychosocial skills (n=30, 58.8%)
3. Theme 3: Communication skills (n=28, 54.9%)
4. Theme 4: Professional ethics and values (n=7, 13.7%)

Theme 1: Critical thinking skills. Most of the respondents (n=38, 74.5%) felt that genetic counsellors are able to critically assess genetic conditions. Thirteen (25.5%) thought that genetic counsellors could use this information to assess and communicate risks to patients and families of inheriting genetic conditions:

- "Very important when planning pregnancy where known genetic condition is in the family or where genetic condition diagnosed and the family members may be affected by latent condition e.g. Huntington's disease".

Several respondents (n=10, 19.6%) remarked that genetic counsellors could use their skills to screen and diagnose patients with genetic conditions:

- “To advise, screen and assist in diagnosis.”
- “To make the diagnosis”

Another 11 (21.6%) respondents felt that, by applying their genetic knowledge, genetic counsellors could advise and guide patients and families on what to do about genetic conditions:

- “……what to do if a result is positive”
- “TO ADVISE THE WHOLE FAMILY”
- “To provide accurate and informative guidance to families with genetic diseases”.

Some respondents made comments which align more closely with the non-directive nature of genetic counselling such as:

- “Someone with genetic knowledge who can advise on likelihood of certain outcomes”
- “To prepare family/patient for test - implications of the results”

Genetic testing and its’ utility were mentioned by 9 (17.6%) respondents and indicated a good understanding of genetic counsellors’ roles:

- “Explain the test, what positive and negative findings mean, and what one may gain by doing the test. To assist with family decision making on basis of test”

Theme 2: Interpersonal, counselling and psychosocial skills. Interpersonal, counselling and psychosocial skills were recognised by more than half (58.8%) of the respondents as important functions of genetic counsellors. This role was aptly described by one respondent as “walk(ing) the journey” at genetic clinic while another mentioned “holding” families’ anxieties.

Skills involved in counselling, such as providing information, and helping to calm patients were mentioned by 18 (35.3%) respondents:

- “Inform, prepare, support. Not to add anxiety + stress”
- “Counselling of doctors and patients”

- "Someone who addresses all fears and concerns and facts of the specific condition with the patient and the rest of the family".

Support, help and assistance (for doctors, patients and families) were mentioned by 12 respondents (23.5%) as skills that genetic counsellors possess:

- "To help with this complicated issue about which I know nothing"
- "….to support families with genetic defects".

Theme 3: Communication skills. The communication skills of genetic counsellors were mentioned by more than half of the doctors (n=28, 54.9%), with their main role being seen as educating and informing patients. The following comment summarises thoughts on this skill:

- "Provides reliable information on the incidences, presentation, prognosis, complications, relevant treatment including rehabilitative actions on various genetic disorders."

Seven doctors (13.7%) also mentioned that genetic counsellors help patients to make informed choices and complex decisions about their genetic conditions:

- "Education (informed choice)"
- "Counselling the family on management plans + options available for their affected child or patient".

Theme 4: Professional ethics and values (13.7%). Finally, eight people commented on the professionalism of genetic counsellors:

- "…specially trained to deal with these complex matters".

The role of a genetic counsellor as a team worker who provides optimal care to patients along with medical geneticists, genetic nurses, laboratory scientists and specialists was recognised by 4 (7.8%) doctors:

- "To form part of the multidisciplinary team managing a patient /family diagnosed with a genetic disease /condition".

Fig 10 is a graphic representation of doctors' perceptions of the roles of genetic counsellors.

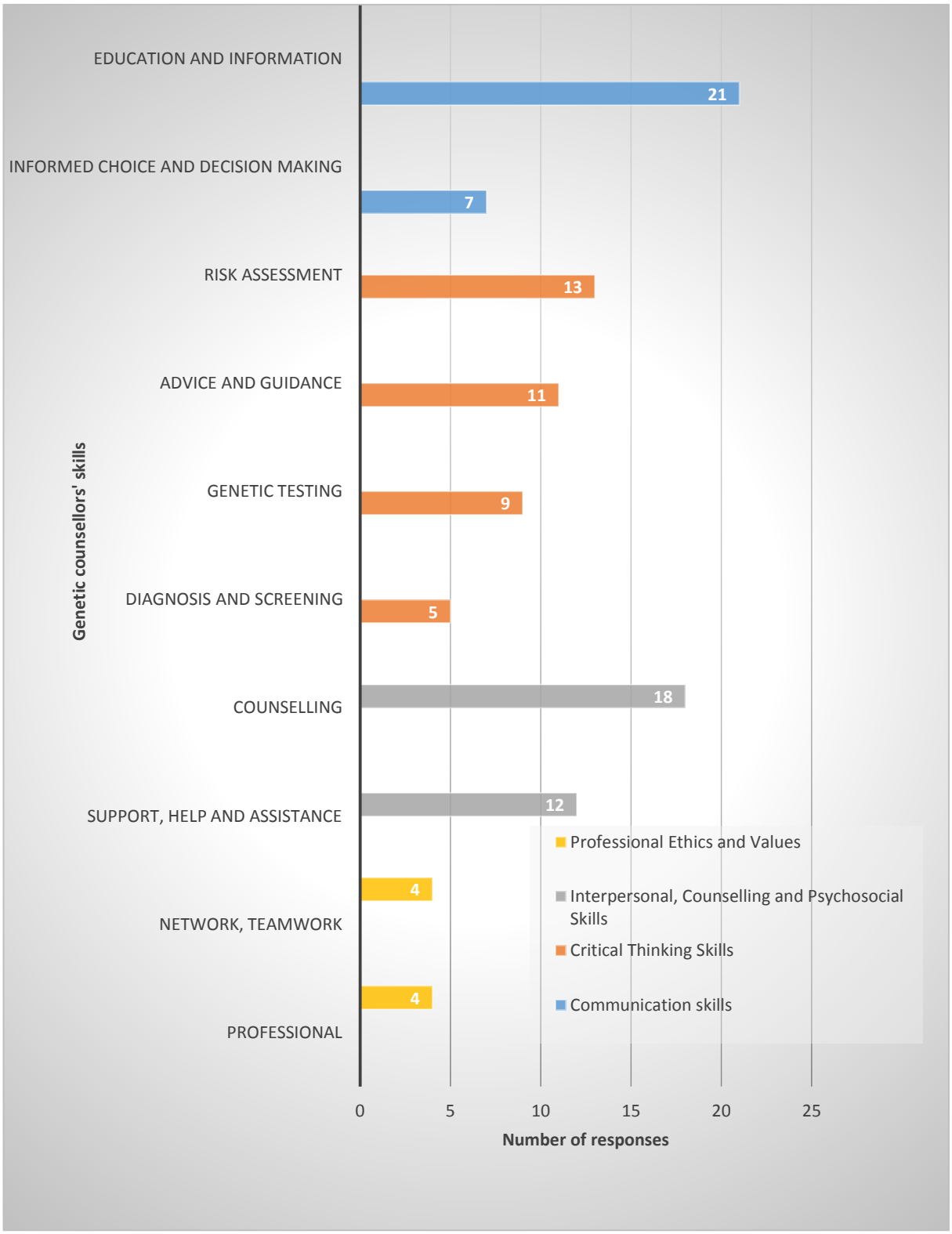

Fig. 10 Doctors' perceptions of genetic counsellors' roles

3.5.3 Doctors' attitudes towards genetic Services and genetic counselling

This was a two-part question, which first asked doctors to choose whether they felt genetic services were "Indispensable", "A luxury", "Pointless" or "Other" (with a comment box.) The same question was asked specifically for genetic counselling (fig. 11). These questions were followed by a question asking the respondents to explain their previous choices.

All respondents completed the question on genetic services, and some who did not choose "Other" added a comment as well. They all saw the value of genetic services and genetic counselling. Slightly more said that genetic counselling is "Indispensable" than chose the option of "Indispensable" for genetic services (74.5% vs 66.7%), but no one chose the option of "Pointless" for either counselling or services.

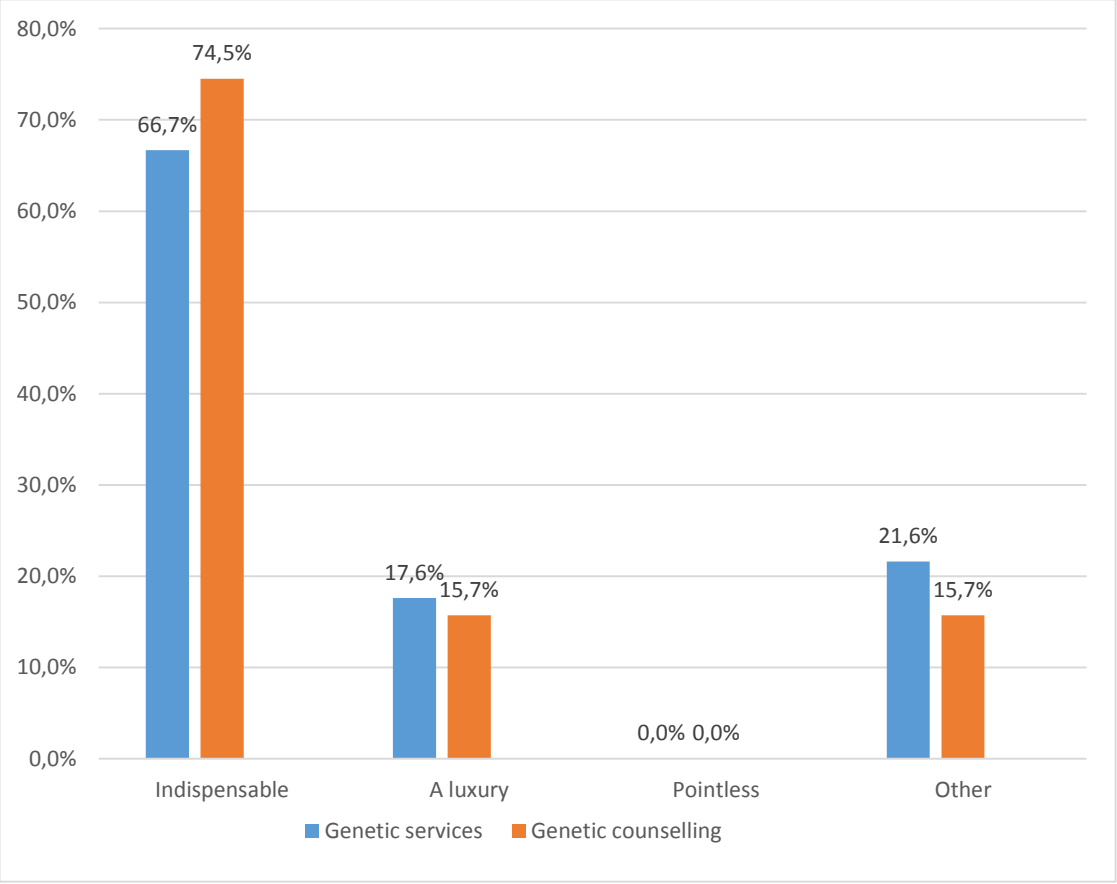

Fig. 11 Doctors' perceptions of the value of genetic services and genetic counselling

Forty seven doctors explained their answers to the previous question and these were divided into remarks about genetic services only, comments about genetic counselling only, and statements about both genetic services and genetic counselling together. (For a complete table of comments, see Appendix E). Comments were categorised under the 3 themes of "Indispensable", "A luxury" and "Other" as follows:

1. Theme 1: Indispensable (Genetic services - n=34, 66.7%)
   (Genetic counselling – n= 38, 74.5%)
   Categories: i. Management (n=16, 34.0%)
   ii. Support (n=10, 21.3%)
   iii. Knowledge (n=10, 21.3%)
   iv. Burden of conditions (n=5, 10.6%)
   v. Complete health service (n=4, 8.5%)
   vi. Access (n=3, 6.4%)
   vii. Essential (n=2, 4.3%)
   viii. Specialist field (n=2, 4.3%)
   ix. Resources (n=1, 2.1%)

2. Theme 2: A luxury (Genetic services – n=9, 17.6%)
   (Genetic counselling n=8, 15.7%)
   Categories: i. Resources (n=6, 12.8%)
   ii. Access (n=4, 8.5%)
   iii. Knowledge (n=1, 2.1%)
   iv. Burden of conditions (n=1, 2.1%)

3. Theme 3: Other (Genetic services "necessary" – n=1, 2.1%)
   (Individual answers for genetic services and genetic counselling were "relevant", "useful", "important" and "needed")
   Categories: i. Support (n=2, 4.3%)
   ii. Burden of conditions (n=2, 4.3%)
   iii. Management (n=1, 2.1%)
   iv. Essential (n=1, 2.1%)
   v. Specialist field (n=1, 2.1%)

Theme 1: Indispensable. Management was seen by most respondents as the reason that genetic services and genetic counselling are Indispensable. Screening, testing, diagnosis, options, treatment and planning were all included in this category. Genetic counselling was seen as "Indispensable" because it helps with managing and adapting to genetic conditions:

- "… essential to help manage other risk factors."

- "Genetic counselling is crucial to help parents and families deal with rare conditions that will have lifelong consequences."

Other reasons that both genetic services and counselling were seen as "Indispensable" pertaining to management included helping patients to understand why they might want to be tested for genetic conditions and what options are available to them:

- "Patients need to understand the implications of testing - the nature of the disease and risk to family."
- "…. give them options about prevention, how to deal with the situation etc., lifestyle."
- "… both diagnosis and potential treatment (physical/psychological) become very important."

Three respondents (6.4%) who said genetic counselling was "Indispensable" because of the role it plays in support made comments such as:

- "Genetic counselling is crucial to help parents and families deal with rare conditions that will have lifelong consequences."

The theme of support, especially in providing information, for both patients and their families as well as medical practitioners was mentioned by 7 respondents (14.9%) who said both services and counselling were "Indispensable":

- "Family and patients need lots of support and need to be informed with facts to help them deal with illness"
- "General practitioners need referral facility for support."

Ten doctors (21.3%) commented that in-depth knowledge of genetics, including both increase in genetic knowledge and lack of genetic knowledge by doctors, made genetic services and genetic counselling "Indispensable":

- "Genetic services are very important….as more conditions are found to be linked to genetic background."
- "Genetic services provide DEPTH around the genetic conditions. That knowledge is not available/present outside of formal genetic services."
- "Recent knowledge growth in genetics is beyond average GPs ability to cover."
- "New field that is going to influence how we treat patients and influence decisions about treatments."

Five GPs (10.6%) noted the increasing burden of genetic conditions as being a reason that genetic services are “Indispensable”:

- “With the current burden of genetic conditions and the risk of increased incidence in future of genetic conditions due to lifestyle changes, increased age of childbearing and adverse environmental factors, Genetic services will become an even more important part of medicine” .

One felt that the psychological and physical burden of genetic conditions made genetic services “Indispensable”.

- “Genetic conditions have a major impact on wellbeing. Thus both diagnosis and potential treatment (physical/psychological) become very important.”

The importance of genetic services as part of a complete health care service which would provide care for everyone was mentioned by 3 respondents (6.4%), although two thought that these services were expensive:

- “An holistic health service needs genetic services.”
- “Why discriminate patients with genetic disorders just because services are EXPENSIVE?”

Genetic counselling was also seen as “Indispensable” to a complete health service as it is complementary to genetic services by a respondent who said:

- “If a genetic service is present it has to go hand-in-hand with counselling.”

Availability of genetic services for everyone was raised by 2 doctors (4.3%) who thought genetic services were “Indispensable” because of increased knowledge and the need for genetic testing:

- “With all the advances in the subject and management of genetic conditions, all should have access to these services.”

One doctor who thought that genetic counselling was “Indispensable” because of the options of genetic testing and counselling for genetic conditions commented:

- “It’s important for patients to have access to counselling and testing, doesn't matter the condition.”

Two participants, who said services and counselling are “Indispensable” simply commented that genetic services and counselling are essential:

- "Common sense tells me this is an essential service."

Two respondents noted that genetics is a:

- "Specialised field and very important."

Theme 2: A luxury. Genetics is perceived as important in a comprehensive health service, but many of those answering "A luxury" drew attention to the lack of resources, the cost of genetic services and the first/third world divide, e.g. under genetic services, one doctor said:

- "Unfortunately in state sector where time with patients is so pressured and budgets are constrained, it is a luxury. Even in private, the cost to many patients is prohibitive".

Four comments (8.5%) related to a belief that rural areas and poor people lack access to good primary health care due to resource and time limitations and that this should be prioritised over tertiary services such as genetics:

- "In a 3rd world setting where basic medical care is appalling, is it relevant. In the first world e.g. Ireland, I see lots of genetic conditions - basic health needs are met therefore more appropriate."
- "Indispensable (in an ideal world), a luxury (in this world): In a country where people still have no running water, genetic services are a luxury!"

Comments about difficulty accessing counselling and services in the public sector were made by 2 respondents who chose "A luxury" in each category. Supporting comments for counselling were:

- "Difficult to access genetic services for majority of the patient population"
- "It seems to be something available in private services and as someone working in the public sector, I don't know the roles of genetic counsellors or how to access them."
- "In 3rd World countries only a fraction of the population have access to the above."

Further statements from respondents who thought services were "A luxury" because of difficulty accessing them in the private sector included:

- "Generally, as far as I am aware, accessing services is difficult and seems unavailable in private sector - expensive if it is"

One respondent, who saw genetic services as “A luxury” (but “eventually indispensable”) again emphasised the need for increased education and awareness of genetics and genetic services:

- “When I know more about this then I would utilise it???”

One respondent said that genetic services are “A Luxury” but nevertheless recognised the impact of these services on the burden of genetic conditions because:

- “Genetic services are poorly accessed especially in rural areas, yet are indispensable due to the number/prevalence of genetic-related diseases”.

Theme 3: Other. One person said genetic services were “necessary”, because of: “psychosocial support”.

Another respondent said services and counselling were “Relevant” in supporting medical professionals as these services:

- “…..reduce the load from other health care practitioners”.

An additional respondent said genetic services and counselling are “relevant” to lowering the burden of disease as:

- “Various healthcare practitioners can be trained to deliver genetic services, but to have specialised Genetic Services centre will significantly lower the burden of genetic disorders…..”

A different participant noted that genetic services and counselling are “important” in reducing the effect of genetic conditions because:

- “Genetic abnormalities have great impact on patients and families.”

Management was also seen as significant by one respondent who said genetic services and counselling were “useful”:

- “… Option of genetic services needs to be available to patients for current and future decisions on health care.”

One said that genetic services and counselling were “needed”:

- “REALLY needed”.

3.5.4 Doctors' attitudes and needs concerning genetic services

Doctors were asked two multiple choice questions about whether they would like improved access to genetic services (fig. 12) and, if so, what form this access should take (fig.3). This was followed by an open-ended question asking what the doctors need from genetic services in South Africa (see Appendix E for a full list of comments).

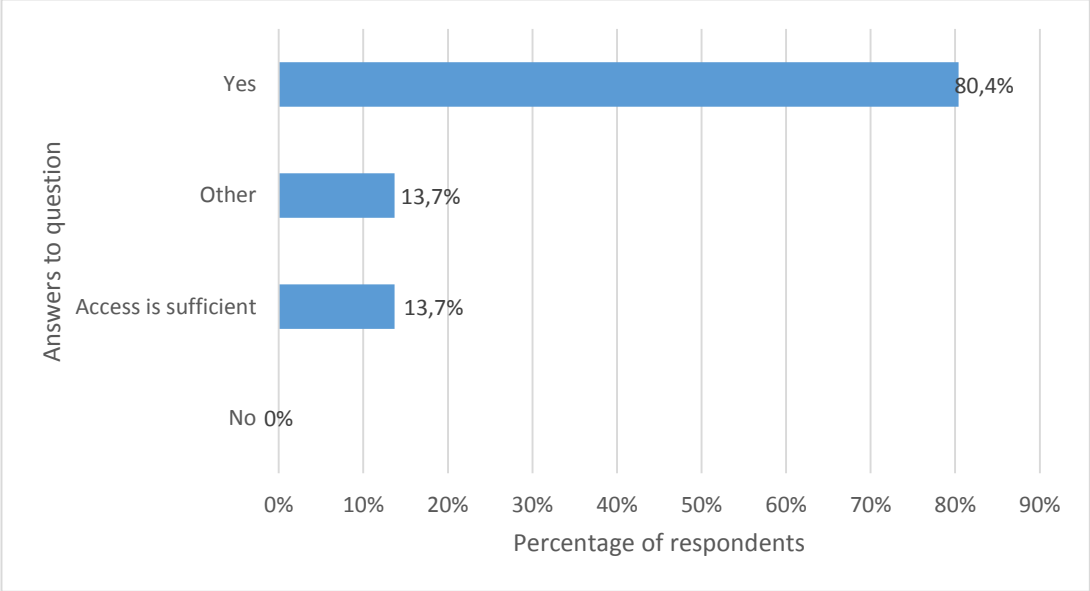

Fig. 12 Doctors' responses to whether or not they would like improved access to genetic services

The theme of increased education in genetics was the most popular choice of what form of improved access to genetics doctors would like (fig. 13). Improved support via the internet or telephonically was also mentioned. One person asked for specialist visits to community health clinics.

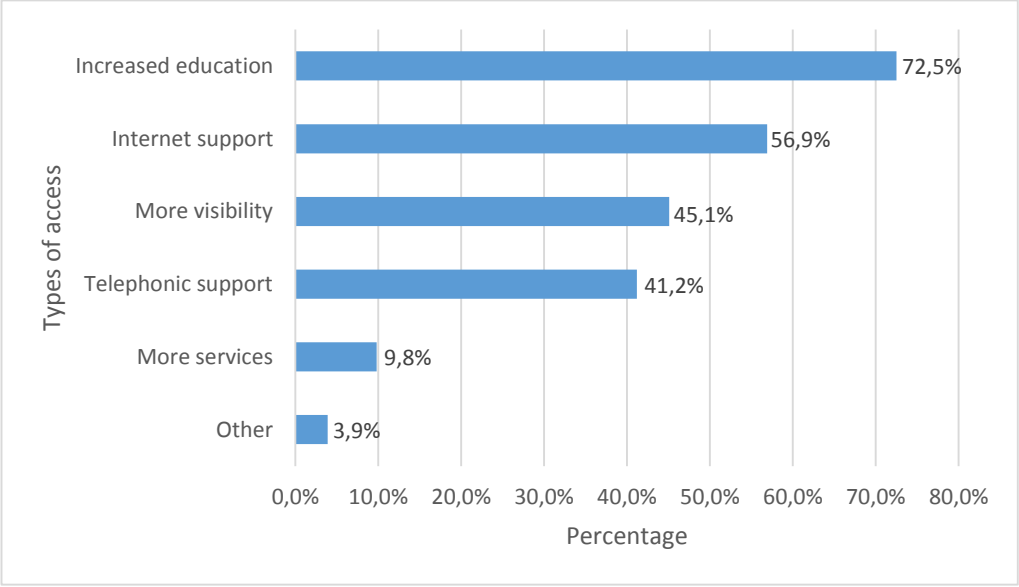

Fig. 13 Types of increased access to genetic services preferred by doctors

Thirty one responses were obtained for the open-ended questions asking what doctors need from genetic services in South Africa. The answers were analysed and sorted into 6 themes as follows:

1. Theme 1: Referral (n=15, 48.4%)
2. Theme 2: Advice (n=11, 35.5%)
3. Theme 3: Access (n=7, 22.6%)
4. Theme 4: Information (n=6, 19.4%)
5. Theme 5: Nothing (n=4, 12.9%)
6. Theme 6: Don't know (n=1, 3.2%)

Theme 1: Referral. Almost half of the respondents (n=15, 48.4%) mentioned referral as their major requirement from genetic services, as they did not know how, when or where to refer patients:

- "Primary care. So I want to be able to refer to a service that can counsel and clinically manage if need be."
- "Improved referral pathways."
- "Referral criteria??At risk patients!!"

Three (9.7%) pointed out the need for assistance with assessment and diagnosis:

- "To assist with support in diagnosis and management of patients with suspected /confirmed genetic conditions."

Another three (9.7%) respondents wanted genetic counselling for their patients:

- "Genetic counselling after diagnosis for patients and family members."

Theme 2: Advice. A further third (n=11) felt that they needed advice on genetic conditions from experts who would be able to help them with management and referrals to support services:

- "To provide comprehensive knowledge on the disease, support, suggestions on management and to provide a network of support services."

Three wanted telephonic advice so that they could have immediate access to genetic services:

- "A helpline such as that offered by Dept. of Pharmacology would be great- one could email or call for advice."

Theme 3: Access. Seven doctors (22.6%) did not know how to access genetic services at all and requested contact information:

- "Where are you? How do I contact you?"

Theme 4: Information on progress in genetics and how to recognise genetic conditions was mentioned by 6 participants (19.4%):

- "Communication on genetic developments"
- "What we can do to pick up families or patients with genetic problems".

Theme 5: Nothing. Four respondents said they did not need anything from genetic services in South Africa:

- "So far I did not need any help from genetic services".

Of these four doctors, one does not work in the country and two are retired, so their lack of need for genetic services in South Africa is understandable.

Theme 6: Don't know. One person did not know what they needed from genetic services.

3.6 Influence of conference genetics session on respondents' answers

Only eight responses were obtained for this question, but the genetics session was held on the last morning of the conference and many of the questionnaires had been returned already. However, the doctors' lack of knowledge and the importance of raising awareness were the recurring themes from these answers too.

Both of the respondents who said their answers had not changed said this was due to the complexity of the information and the need for more education. One respondent felt that the subject matter was too complex:

- "NO - would need to listen to lectures several times over! Simple minded GP :-) (too complicated)."

Three participants said their answers had changed because their awareness had been raised by the genetics session:

- "Yes - a subject that I have never explored or been subject to till Angelina raised the issue!"
- "Awareness +++"
- "Yes. Created awareness."

Another three explained that their knowledge was increased by the genetics session, and one listed what he/she had learnt during the conference:

- ”Yes. The explanation surrounding developments is inspiring/highlights the ever-increasing role of genetics in medical practice.”
- ”YES BETTER UNDERSTANDING AND KNOWLEDGE”
- ”1. Role of CONFIRMING diagnosis - multi factors involved!

2. Costs involved.

3. 3 parent embryo (ovum donor)

4. Non-invasive prenatal testing

5. Pharmacogenomics

6. Role of research (schizophrenia)”.

3.7 Factors influencing previous referral to genetic services by doctors

Chi-squared (χ^2) tests of association were performed to obtain an indication of predictors for referral to genetic services by comparing various aspects with previous referral to services. Factors were assumed to be significant at $p<0.05$, which indicates that there is more than 95% probability that the distribution of interest was not due to chance (table 7).

Only three factors were significant in determining whether or not doctors had previously referred to genetics. The most important feature ($p=0.001$) was that doctors who have had more than 3 hours of postgraduate genetic education are significantly more likely to have referred to genetic services. Two other factors significantly impacting previous referral to genetic services are, not unexpectedly, knowledge of how to access genetic services ($p=0.009$) and more than 5 hours of undergraduate genetic education ($p=0.037$). A fourth factor, whether or not the doctor had extra qualifications (which may relate to the amount of postgraduate education), was significant to a lesser degree ($p=0.063$, i.e. $p<0.1$).

Table 7 Associations between predictors for previous referral to genetic services. Significance highlighted in yellow (p<0.05) or green (p<0.1).

Factor	χ^2	Significance level (p)	Significance level adjusted for observations with n<5 (Yates coefficient Y)
Knowledge of how to access services	6.76	0.009	
Gender	0.12	0.729	
Years in practise (≤24 vs >24)	0.01	0.907	
Public vs private practice	0.38	0.537	
Urban vs rural	2.37	0.123	0.288
Tertiary vs primary care	0.79	0.375	0.882
Undergraduate education (≤5h vs >5h)	4.36	0.037	
Postgraduate education (≤3h vs >3h)	10.31	0.001	0.004
Knowledge scale (self-rated) (1 vs 2 to 4)	1.52	0.218	
Knowledge % (≤39% vs >39%)	1.90	0.168	
Genetic services indispensable vs luxury	0.05	0.817	0.870
Genetic counselling indispensable vs luxury	0.97	0.325	0.566
Need for improved access	0.6109	0.434	0.680
MBChB only vs MBChB plus other qualifications	3.47	0.063	
Number of conditions seen in practice (<3 vs >3)	0.04	0.842	

4 DISCUSSION

In this study, 51 doctors attending a Family Practitioners' Conference in Cape Town completed a survey assessing their knowledge and perceived knowledge of genetics, exposure to genetic conditions and education, perceptions of genetics, genetic education and genetic services and referral patterns to genetic services, both in the past and in the future. This discussion will include findings related to the major research questions, study limitations, practice implications, research recommendations and conclusions.

4.1 Sample demographics

A broad range of GPs attended the conference, with a wide diversity of age, years in practice, interests and education level, making this a relatively good sample approximating the population of GPs in South Africa. However, the 40:60 male: female ratio of survey participants was opposite to that of the conference delegates (58.7:41.3). This female gender response bias has also been noted in a review on response bias for paediatricians' surveys and is particularly relevant in small samples (Cull et al., 2005). On the other hand, the 1:3 ratio of doctors working in rural vs private practice agrees with the ratio in the SAMA database.

Of note was the reduced number of participants in the 40 to 49 year old age group. This may be due to large numbers of doctors leaving South Africa in the 1990s shortly after they graduated, or because the doctors are in their peak earning years and thus do not have time to attend conferences, or it may have been an artefact due to doctors of this age not being motivated to fill in the survey.

A second point of interest was that there were no respondents that graduated after 2010. This may be due to the nature of internship and community service, as these doctors are not yet established in their own practices and are unable to take time off for conferences.

4.2 Referral to genetic services by doctors

The findings indicate that the doctors are aware of the need for genetic services, with almost all (92.2%) indicating they will refer in future, although only slightly over half (52.9%) have referred previously. While few studies have been done on referral to genetic services, these numbers are similar to those found by Claybrook et al. (2010), who found that slightly over half (58%) of surveyed oncologists in Indiana had referred patients to colorectal cancer genetic services previously, but 98% thought they would refer in future. Whereas 83% of the oncologists in Indiana knew how to access genetic services, in the current study, a large

number of respondents (39.2%) are not aware of how to access genetic services. Lack of awareness of how to access genetic services was, as expected, positively associated (p=0.009) with the number of doctors who have not referred previously (47.1%).

The lack of sufficient knowledge about genetics and genetic services was a recurring theme from the survey results. Some participants said that they had no knowledge of what genetic services were offering, where they were or even that they could access services, most felt that their knowledge of genetics was very poor and several said that they did not know what the referral guidelines were for genetic services. This raises the question of whether the lack of posts for genetic professionals, and consequently, the lack of sufficient genetic services in South Africa, has caused reduced awareness by other health professionals of their availability. Alternatively, the insufficient number of genetic services could be caused by the lack of awareness of doctors who are thus not driving the development of these services.

Among the third of doctors who said that they had not previously found it necessary to refer to genetic services, only 2 mentioned that they were retired or not dealing directly with patients and one said that patients had not requested a referral, indicating that the doctor felt it was not their responsibility to decide when to refer a patient to genetic services. According to the National Society of Genetic Counsellors (NSGC) in America, about 50% of genetic counselling clients are self-referred (Riesgraf et al., 2014), implying that doctors do not always recognise when a client should be referred to genetic services and the system of referral to state services, via tertiary institutions, may not be optimal at this point for patient needs.

Other papers by Delikurt et al. (2015) and Claybrook et al. (2010) established similar reasons for doctors not referring to genetic services, including lack of knowledge on where and why to refer patients. Mikat-Stevens, Larson & Tarini (2014) classified these barriers into themes including knowledge and skills, healthcare systems and scientific evidence, as well as lack of time (two doctors in this study ticked “Too busy”). Guidelines on where and when to refer patients to genetic services might help to increase awareness among doctors of these services, resulting in more appropriate use and improved care for patients with genetic conditions.

4.3 Doctors’ genetic education

The wide range of educational qualifications, fields of interest and practice locations indicate that a broad range of doctors responded to the survey.

Most respondents recalled having received some undergraduate education, with the majority of this education being in the form of lectures, although a few said that they had done self-study and seen genetic consultations during undergraduate training. More than half of the respondents remembered less than 5 hours of training in genetics, with only 13.7% recalling 5 to 10 hours of training. Currently, MBChB students at the University of Cape Town (UCT) receive 20 hours of genetic education over their 6 year degree (pers. comm., J. Greenberg, 2015). However, the surveyed doctors had been in practice for an average of 24.1 years and had attended diverse universities, so they would have had different experiences. This concurs with the GenTEE report (Nippert et al., 2013) which commented that medical doctors receive "limited" undergraduate teaching, and may reflect the perception that genetics is not often useful in patient management as a relatively limited number of patients require genetic services. However, with 1 in 15 patients being affected by congenital disorders (Malherbe, Christianson & Aldous, 2015) and since patients with genetic conditions require lifelong management and their families may also be affected, the overall burden of conditions can be relatively high, especially when the conditions require regular medical follow-ups by local doctors. Furthermore, as more and more genetic tests become available, genetics is going to become increasingly important in medical practice and doctors in many disciplines and specialities will need the background to understand when to test patients for conditions and how to interpret the results from the tests (Goldsmith et al., 2013).

A third of doctors had not received any education related to genetics since graduation, and another third had less than 3 hours of post graduate genetic education. Most of this education was through lectures or CPD activities.

In common with studies done in Britain and Columbia (Burke et al., 2009, Rodas-Perez et al., 2015), almost all of the participants wanted more genetic education, recognising that their knowledge of genetics was inadequate and suggesting conferences, online courses, lectures and workshops as environments for educational activities. Over 70% of doctors in the present study felt that they do not even have a basic grasp of topics in genetics and expressed a need for education on basic genetic terminology, what genetic services offer, how and when to refer patients to genetic services, common genetic conditions, screening for conditions, management of conditions and what tests are available. Some wanted to know what genetic counsellors do and others requested simple, easily accessible information. Finally, three were interested in new developments in the field. These answers indicate an interest in genetics and a willingness to learn more about the subject, and provide a

framework for future conference presentations and other educational initiatives. Much of the information that was requested was reflected in the answers to the knowledge section (below).

Other studies also found that doctors were very interested in more education on topics including genetic testing and genetic services, as well as tools such as algorithms to guide genetic referrals (Claybrook et al., 2010; Freedman et al., 2003), which were reflected in doctors' answers to what they need from genetic services in South Africa.

Doctors in this study indicated that they would like basic education that would help them to identify and manage patients who need referral to genetic services, as well as access to web pages and other resources which they could refer to for information whenever necessary. Well-structured genetic education programmes for both undergraduates and for post-graduate medical training would help to meet the need for more knowledge of genetics and genetic services.

4.4 Doctors' knowledge of genetics

The mean knowledge score obtained after adding up answers to several of the knowledge based questions was 39.9%, with a range between 6.7% and 71.1%. This low mean score was reflective of over 70% of the respondents recognising that that their genetic knowledge is very poor or could be better. However, since many of the participants scored over 39.9%, this could indicate that their overall knowledge of genetics is better than they think, although it may still be insufficient for their patients' needs. More education in genetics would help to improve the knowledge of participants. This self-reported lack of genetic knowledge was similar to that reported in other studies worldwide (Klitzman et al., 2013; Aalfs et al., 2003).

4.4.1 Conditions seen in practice

Despite the self-reported lack of genetic knowledge, doctors had a reasonable grasp of conditions that they had seen themselves in practice. This indicates that they are willing to engage with genetic conditions when their patients are affected. Common conditions such as attention deficit disorder, asthma, diabetes and hypertension are not commonly seen in the genetic clinic. Respondents citing these conditions may have recognised that all conditions have an element of genetic predisposition, and may have misunderstood the question, or they may have thought that the disorders have a purely genetic cause. Although these conditions are generally thought to be multifactorial in aetiology, the answers do indicate an understanding of the value of family history as hypertension, particularly, is known to have a hereditary component. Stevens Johnson syndrome, which is a cluster of

symptoms caused by an allergic reaction to medication, may have been cited because of the word “syndrome” indicating that the respondent did not realise that, while a syndrome is a collection of symptoms occurring together, it does not necessarily have a genetic cause.

As expected, the most commonly seen condition in practice was Down syndrome (seen by over 80% of the respondents) as it occurs in about 1 in every 525 live births (Kromberg, Sizer & Christianson, 2013). Doctors also had a good understanding of the chromosomal nature of the condition with almost 90% of them answering this question correctly. Thirty percent of doctors had seen cystic fibrosis, which has a reported incidence of 1 in 3000 among White South Africans (Kromberg, Sizer & Christianson, 2013). Almost two thirds of these doctors knew that cystic fibrosis is inherited as an autosomal recessive condition. One quarter of respondents mentioned breast cancer, which is a condition seen in 1 in 10 women in South Africa, although only up to 10% of all breast cancer cases are inherited (Nelson et al., 2014). This awareness of breast cancer as an inherited disease could be due to the commonly cited “Angelina effect” which has raised awareness of inherited breast cancer among the public via the popular media (Raphael et al., 2015). Sickle cell anaemia, seen by just over 20% of doctors in the study, occurs at a rate of less than 1 in 10 000 in Black immigrants (Kromberg, Sizer & Christianson, 2013), but the severity of the disease and the increasing immigrant population in South Africa, as well as the fact that sickle cell anaemia is often one of the first conditions studied (at secondary and tertiary level) may also have raised awareness among practitioners. Half of the respondents correctly identified the autosomal recessive pattern of inheritance of sickle cell anaemia. Conditions such as Huntington disease, haemophilia, thalassemia and albinism had been seen by between 13 and 20% of doctors and correct answers on their inheritance patterns varied from 57% to 83%. Spinal muscular atrophy (SMA), although a very severe condition that is found in 1 in 2000 blacks (Kromberg, Sizer & Christianson, 2013) was only cited by one doctor, who also correctly identified the inheritance of the condition as autosomal recessive, but was not aware of the existence of a genetic test for the disorder. Albinism is present in 1 in 3900 Blacks (Kromberg, Sizer & Christianson, 2013) and was seen by five doctors. Albinism is a very easily recognisable disorder in dark-skinned races and this may contribute to recall of the condition.

The relatively high numbers of doctors seeing cystic fibrosis, which is more common in White patients, and the low numbers of doctors mentioning SMA and albinism (more commonly present in Black patients) could be related to the racial profile of the patients seen by doctors (particularly in private practice) but this was not assessed, nor were conference delegates racially profiled.

One doctor saw foetal alcohol spectrum disorder (FASD), which is the most common congenital disorder seen in South Africa, having the highest prevalence of all countries and being seen in up to 9% of children (May et al., 2013) in some areas of South Africa. FASD is commonly recognised among communities of mixed ancestry who live and work on farms and so patients with this condition may not be seen by doctors in private or urban settings, who made up nearly half of the respondents. Three respondents mentioned spina bifida which is another relatively common congenital disorder, occurring in up to 1 in 200 live births in South Africa (Fieggen & Stewart, 2014). These disorders are congenital conditions (i.e. they are present at birth) and affected children are often seen in genetic clinics. Similarly to the situation with FASD, the social profiles of patients seen by doctors may have been a factor in the low numbers reported by doctors as the burden of neural tube defects has been reduced by folic acid supplementation and, furthermore, patients receiving more antenatal care may be more likely to terminate a pregnancy where a foetus is found to have spina bifida.

It was interesting that all conditions, except for haemophilia and thalassemia, were seen as having a significant impact on patients' lives by most respondents. This implies that doctors seeing these conditions in general practice may be seeing patients with milder forms and thus may not be aware that both conditions can be lethal and need very frequent follow-ups at haematology clinics. Alternatively, their patients may be well managed and go directly to tertiary centres when they develop severe symptoms rather than reporting to their GP. If these conditions are considered to be serious, however, the doctors may not be referring other family members appropriately to genetic services for risk counselling. Wertz and Knoppers (2002) (cited by Wonkam, Njamnshi & Angwafo, 2006), found, however, that even experienced genetics professionals do not always agree on the relative severity of conditions and it would thus be unreasonable to expect the GPs in this study to rate conditions with confidence.

4.4.2 Knowledge and awareness of 10 inherited conditions

The results of this question on a list of 10 genetic conditions provided in the survey confirmed the doctors' self-reported comments (section 4.3) that they need basic education on genetic concepts, as answers to the questions indicated that they have little in-depth knowledge of the conditions that they recognised. In particular, their understanding of commonly used genetic terminology such as "carrier testing" and "predictive testing" was very poor. Although 6 of the 10 conditions (Down syndrome, haemophilia, breast cancer, cystic fibrosis, albinism and sickle cell anaemia) were recognised by 75% or more respondents, analysis of answers on the availability of predictive and carrier tests indicated that doctors did not know

the difference between these tests, and several seemed to think that these tests were mainly done on pregnancies, thinking that the word ”predictive” meant the test would predict whether the foetus would carry the gene (when these tests are commonly done to predict whether at risk adults carry a dominant gene for a late onset condition) and, similarly, that a “carrier” test means that a foetus “carries” a mutated gene (or a whole chromosome) when this test is usually performed on an adult at risk of carrying a mutated gene for an autosomal or X-linked recessive condition. This implies that education on basic genetic terminology should be improved at all levels. Awareness of basic genetic terminology would give doctors better understanding of genetic testing and when to order genetic tests appropriately, i.e. a predictive test for an autosomal dominant late onset condition and a carrier test for a couple at risk of having a child with an autosomal recessive condition.

The doctors had a good basic understanding of the practicalities of prenatal testing, but their answers to this question (pre-populated knowledge table) indicate that they would not know when to use the various techniques available, or what they would need to test in a family with a genetic condition.

The general lack of awareness that all of the listed genetic conditions (apart from chromosomal conditions) place family members at risk also implies a lack of basic understanding of genetics and mechanisms of inheritance which could easily be rectified by improved education and awareness programmes. Likewise, educating the doctors on these basic concepts could raise their awareness of when to refer to genetic services.

4.4.3 Knowledge of the ethics of testing children under 18 for genetic conditions

The lack of knowledge of the meaning of carrier and predictive tests is reflected in the respondents’ answers about testing minors for genetic conditions.

No one answered this question correctly, indicating little real understanding of the ethical issues behind genetic testing of minors (or that the question was difficult to understand). Doctors’ basic understanding was similar to what they would do with a non-genetic condition, i.e.

1. 72.5% chose to test if the child is symptomatic and there is treatment available (these are correct for genetic conditions as well),
2. Test if a sibling (90.2%) or parent (84.3%) has a condition (in the case of an infectious disease, a child could be at risk of developing the condition). Over 90% of respondents thought it was appropriate to test a child for a genetic condition if a

sibling is affected, showing that, when prompted, respondents do recognise that all genetic conditions place family members at risk in contrast to the answers given in 4.4.2 (above).

These answers would at first appear logical, as they imply an understanding that if a condition is present in the family, the children are at risk of inheriting that disorder. However, the responses do not take into account the ethical principles of beneficence, autonomy, justice and non-maleficence for the child. The American Society of Human Genetics (ASHG) has taken the position that, unless medical intervention is available, children (usually above the age of 18) should be allowed to make their own decisions about whether and when to be tested for genetic conditions when they are old enough to make considered judgements with full information and understanding of the implications of results from genetic tests (Botkin et al., 2015). Genetic counselling would usually be recommended for informed decision making.

Only 15.7% of doctors recognised that a child under the age of 18 has a right to request a genetic test (under certain circumstances). This is another area in which doctors need education, specifically because genetic testing is different from other medical testing as the consequences of testing are lifelong and have implications for future decisions on childbearing and, in some cases, life choices. The right for a child not to know their genetic status, or to decide for themselves when they want to know, should be protected. The ethics of genetic testing will become increasingly important as more tests and DTC testing become available, and proper policies will need to be implemented to prevent harm being done to the public through inappropriate use of genetic tests (Skirton, 2015).

4.4.4 Awareness and knowledge of inherited breast cancer

Despite the awareness of inherited breast cancer generated by media exposure since Angelina Jolie disclosed her *BRCA* status to the media, doctors had limited understanding of the mechanisms of inheritance of the *BRCA* gene, with few understanding that breast cancer can be inherited through both paternal and maternal lines. The “Angelina effect”, first mentioned in Time magazine in 2013 (Borzekowski et al., 2013) is credited with raising awareness of inherited breast cancer and *BRCA* testing among the public and this increased consciousness could cause doctors to assume that more than 10% of breast cancer is inherited. In the current study, 19.6% of doctors chose 10%, but 25.5% thought that 25% of breast cancer is hereditary and 5.9% chose 50% as the correct answer. A study done in the USA, prior to the disclosure of Angelina Jolie’s status, also found that doctors inaccurately

estimated risk for *BRCA*1/2, mutations and thus referred too many low risk and too few high risk patients to genetic services (Trivers et al., 2011). Consequently it seems that doctors are superficially aware of the *BRCA* genes and their tendency to cause inherited breast cancer, but they do not have in-depth knowledge of the risks it confers, or how it is inherited.

Education programmes with genetic referral guidelines and information about risk assessment models for inherited breast cancer (e.g. Manchester, Boadicea) would be valuable for doctors working with families who may be at risk for inherited cancers (Nelson et al., 2014)

4.4.5 Doctors' knowledge and attitudes concerning genomics

Over 80% of respondents did not know about DTC testing, which is higher than the 65% mentioned in a review article by Goldsmith et al. (2013). However, unlike the doctors mentioned in this systematic review, where 85% did not feel qualified to interpret the results of a DTC test, only 10% of doctors in the current study said they did not know what they would do if approached to interpret a DTC test result. Almost 27% said they would interpret the results themselves, while the rest would either refer or gather information before discussing the results with a patient. This indicates that the doctors may be assuming their existing skills in dealing with new phenomena may be adequate for DTC testing as well. Similar responses were obtained in 1999 by Kumar & Gantley, prior to the existence of DTC tests, who found that GPs felt that they would be able to integrate genetic technology into their work with little adaptation, and the results from this study may simply reflect lack of familiarity with this new technology. However, in the current study, this confidence in their ability to manage DTC testing is not present in doctors' feelings on their knowledge of other genetic issues and may be related to the lack of understanding about what DTC tests offer and how easily accessible they are to the public.

Currently, reporting on the results of DTC testing is unregulated and the lack of basic genetics knowledge displayed by physicians means that doctors may "mishandle, misinterpret, and misadvise these patients on what is one of the most important pieces of medical information they will ever receive" (Matloff & Caplan, 2008, cited in Harris, Kelly & Wyatt, 2013).

South Africa's relative isolation from the larger markets of Europe and America may have shielded the doctors from this technology which is becoming increasingly popular elsewhere, but DTC testing is now being advertised in popular media in South Africa too and GPs may be faced with requests from patients to order or interpret tests soon. Despite their

unexpected confidence in working with genomics, respondents felt that awareness of genomics needed to be raised through marketing, education, information and CME activities.

Ninety percent of respondents to this study believed that genetic profiles might influence pharmaceutical therapy, similarly to 98% of physicians in a study by Stanek et al. (2012). This indicates an awareness of the importance of genetics for medicine in the future, although pharmacogenomics tests for medications such as warfarin, tamoxifen and some anti-depressants are available but not yet widely used by physicians (Klitzman et al., 2013).

In common with studies done in Argentina (Bidondo et al., 2015), there was a perception that genomics is not relevant to rural practice or people using public health care. This is probably true for now, as most patients using these services are from lower socioeconomic groups and would not be able to afford these tests. However, as one respondent noted: "You never can tell."

Answers to these questions again highlight the need for more comprehensive education of GPs, especially those in the private sector, who may, in future, be approached to interpret or request DTC tests. The field of pharmacogenomics is rapidly developing and will be available to GPs soon. Genetic services will not be able to manage large numbers of private patients with queries, so laboratories offering DTC tests, as well as pharmaceutical companies, may need to employ genetic counsellors to help GPs and other medical professionals with interpreting results from tests and helping to explain how a patient's genetic profile could affect their medication regime.

4.5 Doctors' knowledge and attitudes concerning genetic services

4.5.1 Which medical professionals should refer to genetic services?

Most of the doctors (72.5%) said that all medical practitioners should be able to refer to genetic services, which was inclusive of the other categories mentioned, i.e. specialists, GPs, nurses, institutions and pathology laboratories. This reflects the understanding that genetic conditions can be recognised in any health care setting, from primary care through to tertiary and quaternary institutions, e.g. a community nurse working in a rural setting could diagnose a new born baby with Down syndrome, and an oncologist in a tertiary hospital could identify a family with a strong history of early onset cancer.

A few (29.4%) felt that specialists were more likely to diagnose genetic conditions than other doctors and so they should be the ones to refer. While this may be true in some cases, e.g. a cardiac surgeon may be more likely than a GP to detect that a cardiac condition in a patient

is hereditary, if more GPs understood the indications for referrals, more of them might be comfortable with referring directly to genetic services in certain instances, e.g. when a child is healthy but dysmorphic, or when a patient is worried about a family history of a condition but is not symptomatic.

Basic guidelines on when it is appropriate to refer to genetic services would help GPs to be more confident in referring in future, as "Recognizing signals that are potentially indicative of a hereditary component of a disease" was rated the highest need for genetic education of GPs by Houwink et al. (2012), since a lack of knowledge can lead to poor recognition of genetic problems and a reduction the quality of patient care.

4.5.2 Roles of genetic counsellors

Respondents recognised that genetic counsellors possess many skills valuable to the medical profession. Doctors commonly saw genetic counsellors as being skilled in critical thinking, including being able to critically evaluate conditions to accurately assess risks for future pregnancies, and whether or not genetic testing would be an option for a patient. Their specialised training was also recognised, as was their role in supporting both doctors and patients with genetic concerns. Communication skills, which are the crux of all genetic counselling interactions, were mentioned by more than half of the doctors, with the main role being seen as educating and informing patients. Communication skills included facilitating patients' decision making through informed choice, which is aligned with genetic counsellors' roles (McAllister et al., 2015).

However, the need for more information concerning what genetic counsellors do in practice was emphasised by the mention that genetic counsellors screen, diagnose conditions and advise patients. While genetic counsellors may be involved in screening patients who might need genetic services, this is more frequently done by medical doctors. Genetic counsellors may add information in diagnosing genetic conditions but they are usually confirmed by medical geneticists (i.e. medical doctors that specialise in genetics), specialists or GPs. Finally, genetic counsellors do not offer advice, but, using information, communication and supporting skills, help people to make their own decisions about managing genetic issues in their lives (McAllister et al., 2015).

4.5.3 Doctors' attitudes towards genetic services and genetic counselling

In common with previous studies (Burke et al., 2009; Wonkam, Njamnshi & Angwafo, 2006), all respondents realised the value of genetic services and genetic counselling. Slightly more (74.5%) said that genetic counselling is Indispensable than said that genetic services are

Indispensable (66.7%), but no one chose the option of “Pointless” for either counselling or services. One said: “An holistic health service needs genetic services.” A comment on genetic counselling was that it is part of a complete genetic service: “If a genetic service is present it has to go hand-in-hand with counselling.” These findings are in contrast to the assumptions of the GenTEE report (Nippert et al., 2013) that most doctors in South Africa “do not give due importance to genetic counselling” due to lack of genetic education.

As with genomics, there was a perception by some respondents that rural areas have more important needs than genetic services and that these services are very expensive. Tan & Fitzgerald (2014) also mentioned the perceived high cost of genetic services as a barrier to referrals in Australia. Similarly, the perception that medical genetics is a service only for highly developed countries is mentioned by Bidondo et al. (2015) in a study on genetic services in Argentina, and this perception should be corrected, as one respondent noted: “Why discriminate (against) patients with genetic disorders just because services are EXPENSIVE?” Other comments such as: “A necessary luxury”, “Genetic services are poorly accessed especially in rural areas, yet are indispensable due to the number/prevalence of genetic-related diseases” indicate support for genetic services despite the lack of resources in South Africa. As mentioned in the first part of this study, almost 40% of respondents did not know how to access genetic services, so they are probably unaware of the availability of services in the main centres as well as the regular outreach programmes to community health clinics and hospitals in rural areas. Raising awareness of the accessibility of genetic services in the state might change perceptions that in South Africa genetic services are only for wealthy, private patients. Furthermore, there are no medical geneticists and only a few genetic counsellors working in the private sector, so services may in fact be more readily accessible by state patients.

The burden of genetic conditions was also recognised as a reason for genetic services to be indispensable, in agreement with the GenTee report (Nippert et al., 2013), with one respondent commenting: “With the current burden of genetic conditions and the risk of increased incidence in future of genetic conditions due to lifestyle changes, increased age of childbearing and adverse environmental factors, genetic services will become an even more important part of medicine.”

Once again, the overwhelming theme from this question is that, while doctors recognise the need for genetic services, they were not sure how and when to access these services.

4.5.4 Doctors' needs from genetic services in South Africa

Doctors identified their major need from genetic services as how, where and when to refer patients, implying that doctors do not feel confident about referring patients to genetic services appropriately. This concurs with the GenTEE report's assumption that lack of genetic education means that physicians in the GenTEE countries "do not recognize the genetic basis of diseases of their patients, (and) do not know how to refer to genetic services" (Nippert et al., 2013). This also reflects the continuing theme of lack of knowledge of genetics and genetic services by doctors.

4.6 Influence of the conference genetics session on respondents' answers

Although only 8 doctors answered this question, they were mostly enthusiastic about the increase in awareness and knowledge that the conference genetics session had created, and their answers indicated the value of including genetics sessions in conferences such as this one. An earlier study by Vogel (2011) also indicated that attending a conference about genetics and genetic testing was instrumental in raising awareness of genetics among a group of doctors.

It may have been interesting to ask whether simply completing this questionnaire had changed how the doctors thought about genetics and genetic services. This would have addressed a minor aim of the study, which was to raise awareness of genetics among participants.

4.7 Factors influencing previous referrals to genetic services

Four factors were significant in determining whether or not doctors had previously referred to genetics:

i. more than 3 hours of post-graduate genetic education
ii. knowledge of how to access genetic services
iii. more than 5 hours of undergraduate education
iv. extra qualifications apart from MBChB (less significant)

These factors correlate with the barriers to referral to genetic services mentioned by other authors (Baars, Henneman & ten Kate, 2005, Claybrook et al., 2010, Delikurt et al., 2015; Kromberg, Sizer & Christianson, 2013, Marathe et al., 2015, Mikat-Stevens, Larson & Tarini, 2014, Rodas-Perez et al., 2015, Tan & Fitzgerald, 2014): lack of awareness of patient risk factors, lack of knowledge of genetics and genetic conditions and lack of awareness of genetic services, all of which could be addressed by more education.

Education seems to raise awareness of genetics and genetic services, regardless of any other demographic factors. More education implies greater familiarity with subject matter and, therefore, increased likelihood of recognising the need for genetic referrals. More post graduate education also indicates more interest in the subject matter and doctors with more qualifications were also more likely to have referred to genetic services. This could also be due to increased education being associated with more time at tertiary institutions where genetic services are based. The final, and obvious, association is that doctors who know where to access genetic services are more likely to have referred to them. This could be related to the fact that they have had to find genetic services previously, having recognised a need for a patient, or that they knew where services were and so were more likely to use them.

4.8 Study strengths and limitations

This study has a small sample of 51, however, despite the small sample size, respondents' backgrounds and demographics varied widely. The low response rate could reflect disinterest in the topic or unwillingness to complete the survey. According to Asch, Jedzriewski & Christakis (1997) low response rates do not necessarily increase bias, but in this case, selection bias may have occurred because participants had a greater interest in genetics and genetic services. If this is the case, it strengthens the need for improved genetic education for clinicians as those with less interest would probably have even less knowledge of genetics. However, further studies with more respondents are important to validate the answers obtained in this survey.

The study included more women (60%) than men, compared with 65.4% male doctors in the SAMA report of 2010. Further studies with more participants and, possibly, including focus groups and face-to-face interviews are needed to verify information and conclusions made in this study.

The study covered a broad range of topics and each of these could be individually studied in greater depth.

The response rate of 36.4% was lower than the reported survey response rate of physicians of between 50 and 59% (Asch, Jedzriewski & Christakis, 1997), but this could be explained by the fact that two other surveys were distributed at the conference and time was limited for completing surveys. The paper survey was also quite bulky and respondents may have thought it would take too long to complete.

The conference was held in Cape Town, which has well-developed genetic services at Groote Schuur Hospital, Red Cross War Memorial Children's Hospital and at Tygerberg Hospital, and the delegates may have had more exposure to genetic services than other doctors in South Africa. However, doctors attended the conference from all over South Africa and the demographics questions did not include practice location beyond asking whether it was rural or urban.

Failure to identify all concepts may have resulted in conclusions that do not accurately represent the data. The addition of open-ended questions means that the doctors could more accurately express their thoughts and thus the conclusions may be more representative than what would have been learnt from closed-ended questions only.

Limitations affecting findings and their generalisability include the fact that the doctors were all at a conference, which may indicate greater interest in CME and updating knowledge relative to the overall population. There is, however, no way of knowing whether the answers obtained from respondents differ substantially from those that would have been obtained from non-respondents. Furthermore, many of the questions required recall and it is impossible to determine accuracy of answers given from memory.

The assessment of intention to use genetic services in future may not generalise to actual uptake of services.

4.9 Future implications

Despite the preliminary nature of this study, the findings have implications for interventions with doctors. The self-reported willingness to use genetic services and the desire to learn more about genetic conditions and services suggest the benefits of developing educational initiatives targeted towards increasing medical doctors' awareness of genetics and when and how to refer patients to genetic services.

The results also highlight the importance of awareness initiatives so that doctors understand where to find information on genetic services and where to obtain advice on conditions that they may see in practice, as there are several websites with this information, as well as telephonic advice available in Cape Town, Johannesburg, Bloemfontein and KwaZulu-Natal.

5 RESEARCH RECOMMENDATIONS

More research with larger and more diverse groups of medical practitioners (including specialists and allied services) is needed to confirm the findings of this research, as the sample was small and a larger sample with more representatives could lead to different conclusions.

Deeper insight could be obtained by further developing many of the questions in the survey.

Research into how many patients doctors have referred to genetic services would be valuable. It would also be of interest would also be whether to determine the appropriateness of these referrals.

Studies are necessary to further understand the role of significant predictors in determining doctors' willingness to refer to genetic services.

Qualitative studies, including in-depth interviews with doctors, would provide additional elaboration of why/how the factors identified in this study are important.

A study on patient needs from genetic services in South Africa, including how patients accessed these services, would add clarity to whether patient referrals are appropriate to their requirements.

Further studies on educational topics for genetics training in South Africa, including experts' views on what should be taught, is needed to develop genetics training programmes in medical schools and for CME for practising doctors.

6 CONCLUSIONS

To my knowledge, this is the first study in South Africa to investigate knowledge and attitudes of general practitioners to genetics and genetic services. A variety of questions were asked, and answers were obtained to all of them. This study shows that the use of genetic services could improve by, in particular, increasing genetic education for doctors in order to help them to recognise how, when and where to access genetic services for their patients. The doctors are willing to learn more about genetics and this could be achieved through several channels, including seminars and talks for CPD points, journal articles and internet sites. Participants also understand the value of genetic services as being essential as they do not know enough about genetics to manage patients appropriately. This study has helped to define two of the four pre-requisites for development of genetics literacy (Gaff et al., 2007, cited in de Abrew, Dissanayake & Korf, 2014): 1) recognition of the need; and 2) defining the

knowledge and skills required. The third and fourth pre-requisites: developing and implementing education programmes and evaluating these programmes, are beyond the scope of this study.

Importantly, this study has determined some indications as to what can be done to make genetic services more appropriate to doctors' needs. Of special interest is fundamental education, for instance on basic terminology in genetics, in directed family history-taking for genetic conditions such as inherited cancers, and in recognising dysmorphology in young children and when this would warrant referral to genetic services. Furthermore, information on where to find genetic services and what they can offer for patients is also needed. Annual updates on new developments in genetics are important too. The topics mentioned by Burke et al. (2009) for undergraduate medical students were also mentioned by GPs inthis study. These topics are:

i) the ability to identify patients with genetic conditions, with understanding of inheritance patterns and basic genetics;

ii) the ability to manage patients with genetic conditions;

iii) the ability to appropriately refer patients with genetic conditions;

iv) the ability to access information on genetics

v) the ability to understand the uses and limitations of different genetic tests, and ethical issues associated with genetic testing;

vi) the ability to discuss genetic information with patients.

The development of guidelines for referral to genetic services is also aligned with the doctors' perceived needs. Results from the study indicate that the presence of genetic services does not imply that doctors will automatically make use of them, but that other factors, particularly the amount of education that doctors have had and whether or not they know how to access these services, influence referral to genetic services. The services need to be easily integrated into the doctors' practices and should have a positive influence on patient care. Lack of skills and knowledge in genetics was one of the most commonly cited reasons for not using genetic services and this could also be addressed through education.

Additional research is needed to understand whether educational interventions will play a role in referring patients to genetic services, but the generally positive attitudes of GPs towards genetics and genetic services shown in this study indicate that, with increased knowledge, GPs will be more likely to refer patients appropriately to genetic services.

Increasing the awareness of genetics and genetic services in South Africa among GPs could also influence the Department of Health to implement the policies and frameworks that are already in place for genetic services, thus improving care for patients and families who are affected by genetic conditions.

The major findings of this study confirm the findings in other countries by Afroze & Jehan (2014), Mikat-Stevens, Larson & Tarini (2014), Tan & Fitzgerald (2014), Delikurt et al. (2015) and Rodas-Perez et al. (2015) that doctors feel that they do not know enough about genetics and genetic services, and that more education in genetics is the most important factor influencing previous referral to genetic services.

In the words of Nelson Mandela: “Education is the most powerful weapon which you can use to change the world” (Baggaley et al., 2013).

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APPENDIX A: Information Sheet and Informed Consent

QUESTIONNAIRE: An Investigation into the knowledge and attitudes of General Practitioners to Genomics, Genetics and Genetic Counselling

Survey Information

You are invited to take part in a survey investigating doctors' knowledge of genetics and their attitudes to genetic services in South Africa. This is part of my research for a mini-dissertation for a MSc (Genetic Counselling) at UCT.

The survey is designed to take less than 20 min to answer 40 questions. You do not have to answer all the questions but it would be very useful for data analysis if you could answer as many questions as possible. Please answer using your working knowledge. There are no right or wrong answers as we are interested in your exposure to genetics and your experiences in practice.

The results of this survey will be used to gain insight into how GPs use genetic services in South Africa, and how they would like to see them develop in the future. The responses will also be used to generate CPD courses in Genetics that are aligned to GPs' needs. A report on the answers will be made to all participants if requested.

All participants who complete the survey will be entered into a lucky draw for a two night stay in a one-bedroom apartment in Century City, valued at up to R3000. (http://www.myleisuregroup.com/property/Majorca-219/968).

CONFIDENTIALITY The information obtained will be used in a student's dissertation, in congress presentations and for publication without revealing your identity.

Contact details: Researcher: Gillian Dusterwald

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All identifiers will be removed from the competed questionnaire. PLEASE ANSWER QUESTIONS AS FULLY AS POSSIBLE

Informed Consent

INFORMED CONSENT

I, _____, (full name) consent to participate in the survey titled: An Investigation into the knowledge and attitudes of General Practitioners to Genomics, Genetics and Genetic Counselling.

I understand that I do not have to answer all questions in the survey.

_____ (Signature)

_____ (Date)

I would prefer an electronic version (please provide email address):

PARTICIPATION IN LUCKY DRAW

This will be removed and kept separately from the questionnaire and destroyed after the draw.

Name:

Email address:

Telephone number:

Would you like feedback on the results of the study?

☐ Yes

☐ No

APPENDIX B The Questionnaire

QUESTIONNAIRE

1. Have you ever referred a patient to genetic services?

☐ Yes

☐ No

2. Do you think you will refer patients to genetic services in future?

☐ Yes

☐ No

☐ Other **:** ☐

3. Do you know how to access genetic services in South Africa?

☐ Yes

☐ No

☐ Other: ☐

DEMOGRAPHICS

4. Field of practise

☐

5. Age (years)

- ☐ 20 to 29
- ☐ 30 to 39
- ☐ 40 to 49
- ☐ 50 to 59
- ☐ 60 to 69
- ☐ 70 to 79
- ☐ Other

6. Gender

- ☐ Male
- ☐ Female

7. When did you qualify?

8. What qualifications do you hold?

9. How many years have you worked as a doctor?

10. Do you have a specific field of interest? If so, what?

11. Where is your practice located?

Fill in as many as apply

- ☐ Private sector
- ☐ Public sector
- ☐ Urban
- ☐ Rural
- ☐ Tertiary hospital
- ☐ Primary care
- ☐ Academic
- ☐ Not applicable
- ☐ Other: _____

EDUCATION AND KNOWLEDGE

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

- ☐ None
- ☐ One or two hours
- ☐ Three to five hours
- ☐ Five to ten hours
- ☐ More than ten hours
- ☐ Don't know
- ☐ Other: _____

13. What form did the education take? (Choose as many as apply)

- ☐ Not applicable
- ☐ Lectures
- ☐ Meetings
- ☐ Self-study
- ☐ Short course
- ☐ Other: _____

14. How much genetics education have you received since graduation?

- ☐ None
- ☐ 0 to 3 hours
- ☐ 3 to 5 hours
- ☐ More than 5 hours
- ☐ Other: _____

15. What form has this education taken? (Choose as many as apply)

- ☐ Not applicable
- ☐ Lectures
- ☐ Meetings
- ☐ CPD points
- ☐ Self-study
- ☐ Short course
- ☐ Other: _____

16. On a scale of 1 to 5, how confident are you in your knowledge of genetics?

1- not confident, 2- slightly confident, 3- somewhat confident, 4- confident, 5- very confident

17. Please fill in the table, listing genetic conditions that you have seen in your practice. Tick for yes, leave blank for no,? for don't know. An example has been listed in the first row.

Name of inherited condition	Genetic test available	Form of inheritance					Significant impact on life
		Autosomal dominant	Autosomal recessive	X-linked	Chromosomal	Other	?
Turner's syndrome	√				√		

18. Please fill in the following table, listing specific inherited conditions and what you know about them. Tick for yes, leave blank for no, ? for don't know. Comments in "other". An example has been listed in the first row.

Name of condition	Know the condition	Seen the condition in practice	Metabolic test available	Genetic test available	Carrier testing available	Predictive testing available	Other family members at risk	Other
Turners syndrome	√	√		√				
Huntington Disease								
Down Syndrome								
Haemophilia								
Spinal Muscular Atrophy								
Spinocere-bellar Ataxia								
Breast cancer								
Lynch syndrome (hereditary non-polyposis colon cancer)								
Cystic fibrosis								
Albinism								
Sickle cell disease								

19. Prenatal diagnosis of genetic conditions involves:

Tick as many as apply

- ☐ Family history
- ☐ Ultrasound scan
- ☐ Non-invasive prenatal testing
- ☐ Amniocentesis
- ☐ Karyotype
- ☐ Testing maternal DNA
- ☐ Testing paternal DNA
- ☐ Other: [ ]

20. In inherited breast cancer, which is more important:

- ☐ Maternal history
- ☐ Paternal history
- ☐ Neither
- ☐ Both

21. Approximately what percentage of breast cancer is inherited?

- ☐ 1%
- ☐ 5%
- ☐ 10%
- ☐ 15%
- ☐ 25%
- ☐ 50%
- ☐ 100%

☐ Other:

22. Is a male diagnosed with breast cancer at risk for an inherited mutation even without family history?

☐ Yes

☐ No

☐ Other:

23. If a patient with family history screens negative for a BrCa mutation, how should she be managed?

☐ As someone at population risk

☐ As someone at higher than population risk

☐ Other:

24. Do you know about direct-to-consumer genetic testing?

☐ Yes

☐ No

☐ Other:

25. Do you think you may be approached to interpret results of a direct-to-consumer genetic test in future?

☐ Yes

☐ No

☐ Other:

26. What does the term "VOUS" indicate in a direct-to consumer genetic test result?

☐ Various outstanding samples

☐ Variable or unknown status

☐ Variant of unknown significance

☐ Variety of undetected SNPs

27. When is it appropriate to refer a child under the age of 18 for a genetic test?

Choose as many as necessary

☐ When parents request a test

☐ When the child requests the test

☐ When a child is symptomatic

☐ When a parent has a genetic condition

☐ When a sibling has a genetic condition

☐ When a second degree relative has a genetic condition

☐ When treatment or management is available that could prevent or delay the onset of a genetic condition

☐ None of the above

GENETIC SERVICES

Please take time to think about these questions and answer as fully as possible. Your responses will help us to develop education **programmes** for medical professionals.

28. Which medical professionals should refer to genetic services?

29. What, in your view, is the role of a genetic counsellor?

30. Are genetic services:

☐ Indispensable

☐ A luxury

☐ Pointless

☐ Other:

31. Is genetic counselling:

☐ Indispensable

☐ A luxury

☐ Pointless

☐ Other:

32. Please explain your answers to 30 and 31 above:

33. What do you think about your knowledge of genetics?

34. What sort of further education on genetics, if any, would you like?

35. Would you like improved access to genetic services?

- ☐ Yes
- ☐ No
- ☐ Access is sufficient
- ☐ Other:

35a. If your answer to the above is "yes", what form should this improved access take?

- ☐ More visibility
- ☐ Telephonic support
- ☐ Internet support
- ☐ Increased education (e.g. for CPD points)
- ☐ More services
- ☐ Other

36. What needs do you have from genetic services in South Africa?

37. What would you do if a patient approached you with results from a direct-to-consumer genetic test?

38. How can we make genomics more accessible to clinicians in South Africa? Genomics refers to whole genome science and includes genome sequencing that is already available to consumers online.

39. Do you think that pharmacogenomics will have an impact on prescribing medications in the future?

☐ Yes

☐ No

☐ Other:

40. If you haven't used genetic services previously, why not?

- ☐ Not necessary
- ☐ Too busy
- ☐ Forget
- ☐ Difficult to access
- ☐ Other:

If this questionnaire is completed after the genetics session, have your answers changed because of this session? If so, which questions and why?

PLEASE LEAVE COMPLETED QUESTIONNAIRES IN MARKED BOX AT DOOR

THANK YOU FOR YOUR PARTICIPATION

APPENDIX C Completed table of correct answers for listed inherited conditions

18. Please fill in the following table, listing specific inherited conditions and what you know about them. Tick for yes, leave blank for no,? for don't know. Comments in "other". An example has been listed in the first row.

Name of condition	Know the condition	Seen the condition in practice	Metabolic test available	Genetic test available	Carrier testing available	Predictive testing available	Other family members at risk	Other
Turners syndrome	√	√		√				
Huntington's Disease				√		√	√	
Down Syndrome				√			Sometimes	
Haemophilia			√	√	√		√	
Spinal Muscular Atrophy				√	√		√	
Spinocerebellar Ataxia				√		√	√	
Breast cancer				√		√	√	
Lynch syndrome (hereditary non-polyposis colon cancer)				√		√	√	
Cystic fibrosis			√	√	√		√	
Albinism				√	√		√	
Sickle cell disease				√	√		√	

APPENDIX D Table of doctors' qualifications

Qualification	MBChB.or equivalent (%)	MBChB or equivalent plus 1 other qualification no. (%)	MBChB plus 2 other qualifications no. (%)	MBChB plus 3 or more other qualifications no. (%)	Totals
	21 (42.9)	13 (26.5)	9 (18.4)	6 (12.2)	49
Dip Child Health			**2**	**3**	**5 (10.2)**
M Fam Med		**1**	**2**	**2**	**5 (10.2)**
Dip Anaesthetics			**1**	**3**	**4 (8.2)**
Fellow of College of Family Physicians		**1**	**2**	**1**	**4 (8.2)**
Dip Fam Med		**2**	**1**	**1**	**4 (8.2)**
Dip Pall Med		**1**	**1**	**1**	**3 (6.1)**
Dip Obstets		**1**	**1**		**2 (4.1)**
MSc			**1**	**1**	**2 (4.1)**
MCPCPZ		**2**			**2 (4.1)**
M Maternal and Child Health		**1**			**1 (2.0)**
HIV/AIDS management dipl				**1**	**1 (2.0)**
Dip Primary Emergency Care			**1**		**1 (2.0)**
MP Ax Med			**1**		**1 (2.0)**
BSc Nutrition				**1**	**1 (2.0)**
Exec MBA		**1**			**1 (2.0)**
PG Dip Business Management			**1**		**1 (2.0)**
PDD				**1**	**1 (2.0)**
Mem Royal College of General Practitioners		**1**			**1 (2.0)**
DCh			**1**	**1**	**2 (4.1)**
MFHomeopathy				**1**	**1 (2.0)**
COACH Conflict coaching international				**1**	**1 (2.0)**
MFGP			**1**		**1 (2.0)**
Certificate in Travel Health			**1**		**1 (2.0)**
Mem College of Family Physicians		**1**			**1 (2.0)**

NBA			**1**		**1 (2.0)**
BVetSc		**1**			**1 (2.0)**

APPENDIX E Complete tables of comments

Reasons for not previously using genetic services

Option	Numbers (%)	*Comments
Difficult to access	17 (44.7)	?? because my knowledge limited. Thus indirect referrals. Expensive.
Not necessary	11 (28.9)	I did not need them. I am unemployed for a while already. If there were cases I would refer patients first to the specialists to avoid misinformation. Patients already diagnosed when I see them. Not relevant in my practice - only do disability grant applications. But have friends / contacts who ask me for advice and direction. Work overseas, when in S.A. work in day hospitals. ?relevant when std of care is so poor. Not recognised a need. Patients never requested referral.
Other	10 (26.3)	Didn't know I could. No knowledge. First must refer to specialist – HOSPITAL PUBLIC. Not sure what they are offering. I found my colleagues were not aware of services.
Forget	3 (7.9)	Dealing with subeconomic patients, so reliant on state referrals. Ignorance of non-mainstream awareness of possibilities.

Too busy	2 (5.3)	

What doctors want from further genetic education

Theme and Category	No. of responses*	Illustrative examples
Theme 1: Forum for education	**16**	
Category :Conferences	6	•annual UCT GP conference •Education on genetics does not happen often. Congresses and medical journals should bring more of this. •Also a section (maybe morning or afternoon, 2 - 3 lectures) at a conference like this. •Part of regular lecture / conference. Been absent in the PAST.
Lectures	5	•Small group study lectures •REGULAR LECTURES •Workshops
Seminars to public hospitals	*1*	*•Part-time / distance learning with contact sessions - towards a certificate*
Outreach tutorials	*1*	
Short courses	3	•Short courses, starting with the basics, including information about indications for referral and contact details of referral centres.
CME	3	•Seminars at CME events •Ongoing CME lectures at eg Constantiaberg and Kingsbury hospitals - already have CME programs in place.
Refresher courses	2	•Refresher courses on genetics. •Refresher courses which teach about the advances in diagnosis, prognosis and management of genetic diseases.
Counselling sessions	1	•What genetic counsellors do? To sit in on counselling sessions. Common genetic conditions - to attend a genetic clinic and see these conditions.
Theme 2: Topics	**10**	
Basics	8	• Going back to basics • A basic course with presentation of information useful in the GP setting.

		•Basic and new developments and testing •Basic information about TERMINOLOGY used in Genetics
Services	2	•what genetics clinics offer. •Basic services available •Genetics in primary health care • what is available, where and when to use
Tests available	5	• Info re tests available • New tests and how easy they are to do.
Genetic conditions	5	• Common genetic conditions • Reminders of genetic conditions to help us pick them up • Prioritized to important common conditions /problems
Referral guidelines	5	• Referral guidelines • Need to know what is available, where and when to use / refer • information about indications for referral and contact details of referral centres
Genetic counselling	2	• To acquire knowledge to be able to provide further genetic counselling to patients in the community (particularly after they've been discharged from the clinic) •What genetic counsellors do?
Screening	3	• Clinically orientated screening •Assistance in early detection and screening
Management	7	•GP oriented courses to help me better serve my patients and their communities • what interventions are available to minimise morbidity
New developments	3	• What new developments there are. •.Updates on new developments
Theme 3: Resources for self-study	**10**	
Internet/technology	7	• Short online course • Access to a good website • Email • Phone apps?
Print media	3	• Have a booklet in my practice covering "genetics 101" • Advice sheets

		• Journal articles about what genetics clinics offer.
Theme 4: Not applicable Retired	**1** 1	 •Too late!

**Note* Some responses were complex and therefore classified multiple times into themes and/or categories, so numbers do not add up

Self-reported thoughts on genetics knowledge*

Theme and category	Number of responses	Illustrative examples
Theme 1: Very poor	36	
Little knowledge	29	• Virtually zero knowledge which is not really acceptable. •My knowledge is very little - definitely sub-optimal! •Very little! I'd like to know more. • My personal knowledge is very limited. • My knowledge is very little. I personally don't know much besides breast cancer, bowel cancer, which was found with neighbours and family members. • Poor - inadequate •MUCH too little knowledge •Way inadequate • Very sketchy •Very, very marginal • Not sufficient at present • lots of gaps • I have a basic knowledge of a few conditions and principles of genetics but feel that I do not have adequate knowledge to confidently diagnose and manage all genetic conditions
Need education	6	• Poor. Needs updating • Nil. Need education • I need to know a lot more. •I would love to learn and know more. I realise I have many "gaps" BUT usually would ask for advice/ help from specialists / colleagues. May diagnose more conditions with more awareness of testing / implications / presentation.
Non-existent	2	• Non-existent. Woeful!
Theme 2: Could be better	4	• Could always improve, probably in the context of ongoing developments in the field. • Middle of the range
Theme 3: Good	2	• Fair to good • Good teaching in undergraduate at UCT.

Complete list of conditions seen in practice

Name of inherited condition	No. citing condition	Genetic test• √available (no. of responses)	Mode of inheritance/ (no. of respondents)/ √correct mode of inheritance					Significant impact on life (no. of respondents)	%correct
			AD	AR	XL	Chromosomal	Other[ɣ]		Testing/inheritance
Achondroplasia	1	√	√					Y (1)	0/0
Albinism	5	√(4)		√(3)		(1)	(2)	Y(4)	80/60
***Asthma**	1						√(i)	Y(1)	-
***Attention deficit disorder**	1						√(i)1	Y(1)	-/100
Barth Syndrome	1	(1)			√(1)			Y(1)	0/100
Breast cancer (BRCA)	9	√(8)	√(3)		(2)		(2)	Y(6)	89/33
Cleft lip/palate	3		√				√(i)2	Y(2)	-/67
Cystic fibrosis	11	√(8)	(2)	√(7)				Y(10)	73/64
***Diabetes**	3	(1)					√(i)3	Y(3)	-/100
Down syndrome	32	√(28)	(2)	(2)	(1)	√(26)	(1)	Y(27)	88/81
Duchenne's muscular dystrophy	1	√			√			Y(1)	0/0
Epiphyseal dysplasia	1	√	√	√					0/0
Factor V Leiden	1	√(1)	√						100/0
Familial hypercholestrolaemia	4	√(2)	(2)	√(2)		(1)		Y(3)	50/50
Familial adenomatous polyposis	2	√(1)	√(2)	(1)				Y(2)	50/100
Fanconi's anaemia	1	√		√	√		(1)	Y(1)	0/0
Foetal alcohol syndrome	1	(1)					√(i)1	Y(1)	0/100
Haemophilia	7	√(7)		(2)	√(4)			Y(2)	100/57

Huntington's Disease	6	√(5)	√(5)			(1)		Y(5)	83/83
Hypertension	1		(1)				√ (i)	Y(1)	

Name of inherited condition	No. citing condition	Genetic test• √available (no. of responses)	Mode of inheritance/ (no. of respondents)/ √correct mode of inheritance					Significant impact on life (no. of respondents)	%correct
			AD	AR	XL	Chromosomal	Other[ɣ]		Testing/inheritance
Klinefelter's syndrome	2	√(2)		(2)	(1)	√			100/0
Marfan's Syndrome	4	√(1)	√(3)					Y(4)	25/75
Metabolic syndrome	1						√ (i)	Y(1)	-
Muscular dystrophy	1	√(1)	(1)		√				100/0
Myotonic dystrophy	1	√	√	(1)		(1)		Y(1)	0/0
Neurofibromatosis	1	√(1)	√(1)					Y(1)	100/100
***Pancreatic cancer**	1	(1)						Y(1)	-
***Pierre Robin Sequence**	1	(1)				(1)		Y(1)	-
Porphyria	1	√	√(1)						0/100
Prader-Willi Syndrome	1	√(1)				√(1)		Y(1)	100/100
Recessive disorders	2							Y(2)	
Retinitis pigmentosa	2	√	√(1)	√(1)	√	1	√ (ii)	Y(2)	0/100
Schizencephaly	1						√ (i)	Y(1)	
Schizophrenia	1						√ (i)(2)	Y(2)	0/100
Sickle Cell Anaemia	8	√(6)	(1)	√(4)				Y(7)	75/50
Spina bifida	3	(2)					√(i)1	Y(3)	-
Spinal muscular atrophy	1	√		√(1)				Y(1)	-/100
Spinocerebellar ataxia	2	√						Y(2)	0/0
***Stevens-Johnson Syndrome**	1		(1)				√ (i)		-
Sturge-Weber syndrome	1		(1)					Y(1)	-
Thalassemia	5	√	(1)	√(3)					0/60
Triple X	1	√				√			0/0
Tuberous sclerosis	1	√	√(1)					Y(1)	0/100
Turners Syndrome	2	√(2)				√(2)		Y(2)	100/100
Von Willebrand's disease	1	√(1)	√	√	√				100/0

*not commonly seen in genetic clinics. While all conditions are to some extent genetic, only those that are seen in genetic clinics are accepted as “correct” answers here

• genetic tests said to be available if they are mentioned on Gene reviews.

[ɣ] i) multifactorial, ii) mitochondrial

Knowledge and awareness by doctors of 10 inherited conditions. √ correct answer X incorrect answer

Name of condition	*Know the condition	*Seen the condition in practice	Metabolic test available	Genetic test available	Carrier testing available	Predictive testing available	Other family members at risk
Huntington's Disease	32 (68.1%)	16 (34.0%)	X1 (3.1%)	√ 21 (65.6%)	X 17 (53.1%)	√ 5 (15.6%)	√23 (71.9%)
Down Syndrome	46 (97.9%)	41 (87.2%)	X1(2.2%)	√38 (82.6%)	X5 (10.9%)	X14 (30.4%)	√ in certain circum-stances 5 (10.9%)
Haemophilia	40 (85.1%)	23 (48.9%)	5 (12.5%)	√29 (72.5%)	√21 (52.5%)	X10 (25.0%)	√19 (47.5%)
Spinal Muscular Atrophy	22 (46.8%)	8 (17.0%)	1 (4.5%)	√5 (22.7%)	√4 (18.2%)	X1(4.5%)	√5 (22.7%)
Spinocerebellar Ataxia	16 (34.0%)	6 (12.8%)	0 (0.0%)	√4 (25.0%)	X1 (6.3%)	√0 (0.0%)	√4 (25.0%)
Breast cancer	47 (100.0%)	43 (91.5%)	2 (4.3%)	√28 (59.6%)	X21 (44.7%)	√19 (40.4%)	√32 (68.1%)
Lynch syndrome (hereditary non-polyposis colon cancer)	15 (31.9%)	4 (8.5%)	0 (0.0%)	√6 (40.0%)	X3 (20.0%)	√2 (13.3%)	√10 (66.7%)
Cystic fibrosis	43 (91.5%)	24 (51.1%)	13 (30.2%)	√28 (65.1%)	√15 (34.9%)	X9 (20.9%)	√17 (39.5%)
Albinism	35 (74.5%)	21 (44.7%)	1 (2.9%)	√9 (25.7%)	√6 (17.1%)	X4 (11.4%)	√9 (25.7%)
Sickle cell disease	36 (76.6%)	18 (38.3%)	5 (13.9%)	√19 (52.8%)	√14 (38.9%)	X4 (11.1%)	√17 (47.2%)

*Percentages for Know the condition and Seen the condition as a proportion of the 47 respondents who filled in the table. All other percentages as a proportion of the number of people who knew the condition.

What doctors would do if a patient approached them with results from a direct-to-consumer genetic test

Themes and Categories	Number of responses*	Illustrative examples †
Theme 1: Refer	20	
Refer to colleagues/specialists ‡	4	•Refer to a more knowledgeable colleague. •Depending on condition would refer if confirmation needed and intervention warranted (eg BrCa) •Currently would probably refer them to a specialist
Refer to genetic services	8	•Refer to a genetic counsellor •Will probably refer to a Genetic unit •I would refer them to specialist and genetic services. This information should be given to the patient under proper guidance and explanation. •1. Liaise with genetic counselling services 2. Ensure that patient undergo aforementioned (counselling service) 3. Explain the importance of discussion of condition with a specialist service (i.e. face-to-face)
Theme 2: Ask for advice/seek information	26	•Call for help!! •Phone an expert
Genetic services	9	•Seek assistance from the genetic services in SA •Call a genetic expert to assist with interpretation. •Phone genetics clinic at GSH and ask for advice. •needs advice and support from SA gen. services •I would attempt to contact a colleague at the Genetic services to ask for assistance / advice on my further steps to be done / taken to assist the patient.
Direct to consumer test company	4	•First check the reliability of the company doing the test •Refer back to direct to consumer genetic tester •Most likely call the lab that did the test and speak to a consultant

		•Phone the company who did the test for help. Look up on internet?
Colleague	6	•Ask advice from colleague with training •I would ask a colleague if they have heard of it?
Self-study	6	•? learn more on the condition. •I would research the results to learn how to help them interpret the results •Get onto www! •To research on internet and also can phone a tertiary level hospital to help me understand and to explain to the patient. •Panic - Need to obtain information therefore go to internet.
Theme 3 Interpret results	12	•Help the patient to interpret the results. •Consider my own clinical assessment of patient. It would help if the parameters of the validity of the tests are cited with the results. •I would research the results to learn how to help them interpret the results •May ask patient to come back for another appointment to get the best information to give the patient the best answers. •Interpret them. •Try to figure out the result. •Provide support until info obtained, then counsel.
Theme 4 Don't know	4	•I have no idea! •? •Don't know. Depends on what the issue is. - Refer the problem. I have no knowledge at all.

**Note* Some responses were complex and therefore classified multiple times into themes and/or categories, so numbers do not add up

†only a few illustrative comments are quoted here. A full list appears in Appendix 5.2

Themes, categories and examples of responses on how to make genomics more accessible to clinicians in South Africa.

Themes and Categories	Number of responses*	Illustrative examples†
Theme 1: Raising awareness	18	
Advertising and marketing	5	•Advertise widely. Reduce price and make it affordable for man in the street •more marketing of Medicine + of science (as whole) to the people with resources. •More visibility •Direct marketing of information? Reps? •Advertise online services •There could be more awareness and information. •One can always check the internet, but medical professionals should know where exactly to look for information.
Internet	6	•Internet access promoted by SAMA •On-line •Website – interactive. •Internet support •website + apps?
Other media	5	•Magazines – CPD •Telephonic support •Advice sheets
Theme 2: Education/information	18	
Lectures, workshops, CPD activities	6	•Through CPD activities •Through this kind of lecturing venue. •Write articles in SAMJ esp •CME section of SAMJ
Information	5	•Give us more information re services + cost +

		availability - via email / post •Perhaps informing clinicians firstly of this availability and means to access it. •Clarify relevance which I understand and which can help me explain to patients. •medical professionals should know where exactly to look for information. •Provide information to health care providers, who can pass it on to the patients.
Education	9	•Teach us to interpret the results and reassure patients if no problems with results. •-educate! educate! educate! - make subject matter more user friendly!!! •provide training on the rational use of these genomics and safe use as well as how to assist / advise patients they referred to it with queries / interpretation thereof. •Provide a short course and market it well.
Theme 3: Increase accessibility	8	•I was not aware that it is available to consumers online. •Need to know where to access information. •Easy approachability •1.More visibility 2. Contact numbers to be held in surgeries 3. Genetics contact

		•Perhaps informing clinicians firstly of this availability and means to access it. •Make it accessible as a routine blood test. •Make them practical
Theme 4: Don't know	5	NO IDEA I'm not sure. I don't know

*Some responses were complex and therefore classified multiple times into themes and/or categories, so numbers do not add up

†only a few illustrative comments are quoted here. A full list appears in Appendix 5.2

Medical professionals who should refer to genetic services

Medical professional	Number of responses* (%)	Illustrative comments†
Theme 1 Specialists Paediatricians Neonatalogist Gynaecologists/ obstetricians Internal medicine physicians Neurologists Surgeons Oncologists Psychiatrists	15 (29.4) 9 (17.6) 1 (2.0) 8 (15.7) 4 (7.8) 1 (2.0) 2 (3.9) 1 (2.0) 1 (2.0)	•Specialist should be the medical professionals to refer. Only they have all the details, full diagnosis. They would also decide about further treatment/ prevention/ management. •Those who diagnose genetic conditions most likely specialists rather than GPs.
Theme 2 All	37 (72.5)	•All should be able to. •Concerned medical professionals of the whole spectrum. •All health professionals should be aware and refer. •Any medical professional picking up / suspecting a genetic abnormality / condition in any patient. Whether a surgeon, GP, neurologist etc. anyone can diagnose a genetic condition.
Needs basis	5 (9.8)	•Where is the need or special case. •Anyone to assist with diagnosis - provided understand what services offer and where appropriate. •All doctors suspecting a genetic condition or where counselling is needed
Theme 3 General Practitioners	18 (35.2)	•GPs especially
Theme 4 Nurses	9 (17.6)	•Nurse practitioners •Nurses in medical practice •Registered nurses •Professional nurses (in discussion with doctors)
Theme 4 Institutions	1 (2.0)	•GSH (Groote Schuur Hospital) •TBH (Tygerberg Hospital)
Theme 5 Laboratories	1 (2.0)	•Pathology lab
Theme 6 Allied services	1 (2.0)	•Psychologists, OTs (occupational therapists) and Physios

*Some responses were complex and therefore classified multiple times into themes and/or categories, so numbers do not add up †only a few illustrative comments are quoted here. A full list appears in Appendix 5.2

Doctors' responses to roles of genetic counsellors

Themes	Numbers (%)*	Illustrative examples†
Theme 1 Critical thinking skills	38 (74.5)	
Risk assessment	13 (25.5)	•Assess risk of genetic condition.
Patients	4 (7.8)	•Explaining the risk of genetic conditions to families and for people who have the condition already.
Families/ pregnancy planning	10 (19.6)	•…recurrence of cases in a family. •To inform parents of risks of inheriting conditions. •Chances of children inheriting the disease. Other family members at risk. •…risk of passing deformity / carrier gene to offspring. •Very important when planning pregnancy where known genetic condition is in the family or where genetic condition diagnosed and the family members may be affected by latent condition eg Huntington's disease. •… educate them on prognosis, management options and future risk.
Advice and guidance	11 (21.6)	•Someone with genetic knowledge who can advise on likelihood of certain outcomes. •….suggestions on management. •…..advising about choices •…..direct further management
Patients	3 (5.9)	•……what to do if a result is positive. Counsel patient (and family) post-test and plan way forward. •TO ADVISE THE WHOLE FAMILY •To provide accurate and informative guidance to families with genetic
Families/ pregnancy planning	7 (13.7)	

		diseases. •To prepare family/pt for test - implications of the results. •Advice about if they should or should not do the test in the first place. •…advise re planning for children.
Genetic testing	9 (17.6)	•Very important and essential prior to any genetic testing
Patients Families	4 (7.8)	•Counsel patient (and family) post-test. •…..counsel patients, test those possibly affected. •Explain the test, what positive and negative findings mean, and what one may gain by doing the test. To assist ith family decision making on basis of test. •To prepare family/patient for test - implications of the results. •To help the patient and family understand the implications of testing and the implications of results.
Diagnosis and screening	5 (9.8)	•To make the diagnosis. •…guide for suitable screening in genetic conditions. •To advise, screen and assist in diagnosis.
Patients Families	8 (15.7) 2 (3.9)	•Counsel parents and patients on possible diagnosis of genetic condition. •Very important in terms of communicating with and ”holding” families' anxieties and formulating a diagnostic plan.
Theme 2 Interpersonal, counselling and psychosocial skills	30 (58.8)	

Counselling	18 (35.3)	•Counselling for genetic conditions •Explain and counsel genetic conditions •Inform, prepare, support. Not to add anxiety + stress •To understand the nature of the disease. •Discuss the problem with the disorder as to outcome, future treatment
Doctors	1 (2.0)	•Counselling of doctors and patients
Patients	8 (15.7)	•Counsel patient (and family)
Families	7 (13.7)	•Assist patient and/or family to ”walk the journey” whilst attending genetic clinic. •To discuss with parents or patient the risks and management of genetic disorders. •Someone who addresses all fears and concerns and facts of the specific condition with the patient and the rest of the family. •Very important in terms of communicating with and ”holding” families' anxieties and formulating a diagnostic plan. •Counselling individuals, couples and families.
Support, help and assistance	12 (23.5)	•To help in management of burden •Long-term support •Offer support (emotionally)
Doctors	2 (3.9)	•To offer support patient and doctor. •To help with this complicated issue about which I know nothing.
Patients	4 (7.8)	•This assists with the social and financial welfare of the patient. •To support high risk patients

Families	2 (3.9)	•….to support families with genetic defects. •expert knowledge to assist families.
Theme 3 Communication skills	28 (54.9)	
Education and information	21 (41.2)	•Provides reliable information on the incidences, presentation, prognosis, complications, relevant treatment including rehabilitative actions on various genetic disorders. •To supply information and guide for suitable screening in genetic conditions and possible (uncertain) genetic inheritance. •Very important - to explain process and what is about. •To provide comprehensive knowledge.
Patients	7 (13.7)	•Provide information which is understandable by the patients.
Families	8 (15.7)	•educate patient about condition. •He/she needs to explain to the family/patient the possibilities/complications of a disease, preventive measurements like contraception/recurrence of cases in a family. •To educate patient and family. •Profile and needs of genetic condition. Effects on rest of family. •Explain the condition AND its mode of transmission to family.
Options, informed choice, decision making	7 (13.7)	•Someone with genetic knowledge who can advise on likelihood of certain

Patients	2 (3.9)	outcomes. •Treatment options / interventions •Education (informed choice) •Inform patient. Discuss possible outcomes.
Families	4 (7.8)	•Counselling the family on management plans + options available for their affected child or patient. •To assist with family decision making on basis of test.
Theme 4 Professional Ethics and Values	4 (13.7)	
Network, multidisciplinary team	4 (7.8)	•… provide a network of support services
Patients Family	1(2.0) 2 (3.9)	•To form part of the multidisciplinary team managing a patient/family diagnosed with a genetic disease/condition. •…network with allied professionals to support families with genetic defects.
Professional	4 (5.9)	•…..specially trained to deal with these complex matters •Professionals •Expert knowledge to assist families

*Some responses were complex and therefore classified multiple times into themes and/or categories, so numbers do not add up

†only a few illustrative comments are quoted here. A full list appears in Appendix 5.2

Themes pertaining to answers around value of genetic services

Themes *	Number (%) †	Illustrative quotes‡
Theme 1 Complete health service Indispensable	**3 (6.4)**	•An holistic health service needs genetic services. •Why discriminate patients with genetic disorders just because services are EXPENSIVE? •A necessary luxury.
Theme 2 Resources Luxury	**2.1**	•Unfortunately in state sector where time with patients is so pressured and budgets are constrained, it is a luxury. Even in private, the cost to many patients is prohibitive
Theme 3 Support Necessary	**2.1**	•…especially with psychosocial support
Theme 4 Knowledge - increasing Indispensable	**2.1**	•Genetic services are very important and as more conditions are found to be linked to genetic background eg. Breast cancer

Themes pertaining to answers around value of genetic counselling

Themes	Number (%)	Illustrative quotes
Theme 3 Support Indispensable	**6.4**	•Psychosocial support. •Genetic counselling is crucial to help parents and families deal with rare conditions that will have lifelong consequences.
Theme 5 Management Indispensable	**6.4**	•… essential to help manage other risk factors. •…. vital - patients need full information and instructions. •… helps prepare parents in the event of a child with potential medical/ social/ physical difficulties
Theme 6 Access Indispensable Luxury	**6.4** 2.1 4.3	•It's important for patients to have access to counselling and testing, doesn't matter the condition. •Difficult to access genetic services for majority of the patient population. •It seems to be something available in private services and as someone working in the public sector, I don't know the roles of genetic counsellors or how to access them.
Theme 7 Information Indispensable	**2.1**	•…. patients need full information and instructions.
Theme 1 Complete health service Indispensable	**2.1**	•If a genetic service is present it has to go hand in hand with counselling.

Themes pertaining to answers around value of both genetic services and genetic counselling

Themes	Numbers (%)	Illustrative quotes
Theme 5 Management - of conditions (screening, testing, diagnosis, options, treatment, planning) – patient centred	**29.8**	
Indispensable	13 (27.1)	•Diagnosis needs to be made. •Interventions available in certain conditions. Ultimately family can decide / plan re having children. •…. to enable planning of pregnancy or antenatal diagnosis. •Patients need to understand the implications of testing - the nature of the disease and risk to family. •Helps to screen diagnosis. •Treatment and prevention. •Family and patients need lots of support and need to be informed with facts to help them deal with illness. •Needs for diagnosis - how to TREAT OR CURE without diagnosis How to prevent either. •…. give them options about prevention, how to deal with the situation etc., lifestyle. •… both diagnosis and potential treatment (physical /psychological) become very important.
Useful	1 (2.1)	•… Useful: Option of genetic services needs to be available to patients for current and future decisions on health care.
Theme 4 Knowledge - increasing and lack of	**19.2**	
Indispensable	17.0	•Essential, as knowledge is generally lacking (concerning health care providers). •… all the advances in the subject and management of genetic conditions. •Genetic services provided DEPTH around the genetic conditions. That knowledge is not

		available/present outside of formal genetic services. •Recent knowledge growth in genetics is beyond average GPs ability to cover. •Not enough is known about genetics in the general population and clearly medical practitioners do not know enough about the subject matter. •New field that is going to influence how we treat patients and influence decisions about treatments.
Luxury	2.1	•…. (eventually indispensable): When I know more about this then I would utilise it???
Theme 3 Support - effect of condition on patient and family/ support for GPs	**14.9**	
Indispensable	12.8	•Reassure patients. •Family and patients need lots of support and need to be informed with facts to help them deal with illness. •General practitioners need referral facility for support. •They can improve the situation for the family of a patient.
Relevant	2.1	•…..reduce the load from other health care practitioners.
Theme 2 Resources	**12.8**	
Indispensable	2.1	•Expensive
Luxury	8.5	•In a 3rd world setting where basic medical care is appalling, is it relevant. In the first world eg Ireland, I see lots of genetic conditions - basic health needs are met therefore more appropriate. •In 3rd World countries only a fraction of the population have access to the above. •Genetic services are poorly accessed especially in rural areas, yet are indispensable due to the number/prevalence of genetic-related diseases.

		•Indispensable (in an ideal world), a luxury (in this world): In a country where people still have no running water, genetic services are a luxury!
Theme 8 Burden of conditions	**10.6**	
Indispensable	4.3	•With the current burden of genetic conditions and the risk of increased incidence in future of genetic conditions due to lifestyle changes, increased age of childbearing and adverse environmental factors, Genetic services will become an even more important part of medicine. •Genetic conditions have a major impact on wellbeing. Thus both diagnosis and potential treatment(physical/psychological) become very important
Luxury	2.1	•Genetic services are poorly accessed especially in rural areas, yet are indispensable due to the number/prevalence of genetic-related diseases.
Relevant	2.1	•Various healthcare practitioners can be trained to deliver genetic services, but to have specialised Genetic Services centre will significantly lower the burden of genetic disorders, as well as reduce the load from other health care practitioners.
Important	2.1	•Genetic abnormalities have great impact on patients and families
Theme 9 Essential	**10.6**	
Indispensable	8.5	•Common sense tells me this is an essential service. •Should be done on all prenatal patients and breast / colon cancer family members. •Services should be made known to all even unsuspecting (or) at that time uninterested prospective parents.

Needed	2.1	•Essential as genetic testing and counselling not only has implications for the individual being tested but also for family members. •REALLY needed.
Theme 6 Access	**8.5**	
Indispensable	4.3	•Increasing access to genetic testing - imperative for patients to know. •With all the advances in the subject and management of genetic conditions, all should have access to these services.
Luxury	4.3	•Generally, as far as I am aware, accessing services is difficult and seems unavailable in private sector - expensive if it is. •In 3rd World countries only a fraction of the population have access to the above.
Theme 10 Specialist field	**6.4**	
Indispensable	4.3	•Specialised field and very important.
Relevant	2.1	•Various healthcare practitioners can be trained to deliver genetic services, but to have specialised Genetic Services centre will significantly lower the burden of genetic disorders, as well as reduce the load from other health care practitioners.

*Where themes are repeated, the first number allocated is used

†Some responses were complex and therefore classified multiple times into themes and/or categories, so numbers do not add up

‡only a few illustrative comments are quoted here. A full list appears in Appendix 5.2

Themes describing doctors' perceived needs from genetic services in South Africa

Themes	Number (%)*	Illustrative examples†
Theme 1: Referral	48.4	•Genetic referral centres. •Tertiary/University hospitals. •Primary care. So I want to be able to refer to a service that can counsel and clinically manage if need be. •Improved referral pathways. •Down's syndrome; cystic fibrosis, Huntington's and colon polyposis all needs referral for investigation and also to help with counselling. •Guidance in treatment and referral to specific units. •Referral criteria??@risk patients!!
Assessment/diagnosis	9.7	•Confirmation of diagnosis •Screening patients when applicable •To assist with support in diagnosis and management of patients with suspected / confirmed genetic conditions.
Counselling	9.7	•To consult / assess patients with suspected genetic conditions. •Genetic counselling after diagnosis for patients and family members.
Theme 2: Advice	35.5	•To provide comprehensive knowledge on the disease, support, suggestions on management and to provide a network of support services. •Guiding me to important, valuable management and counselling. •How to manage and treat genetic diseases. •Advice re - whom to screen, how to screen, when to screen patients. •Guidance in treatment and referral to specific units. •Advice about referral.
Telephonic	9.7	•Telephonic advice. •Telephonic support. •A helpline such as that offered by Dept of Pharmacology would be

		great - one could email or call for advice.
Theme 3: Access	22.6	•More visibility. •Access details. •Easier access. •Education to know availability. •Do not live in SA, but often we need access via laboratories such as Lancet. •Where are you? How do I contact you?
Theme 4: Information	19.4	•Information. Not much else. •Basic information. •Increased education (e.g. for CPD points). •Communication on genetic developments. •What we can do to pick up families or patients with genetic problems. •More information and training.
Theme 5: Nothing	12.9	•So far I did not need any help from genetic services. •None. Was never an issue which I had to deal with.
Theme 6: Don't know	3.2	•Don't know

*Some responses were complex and therefore classified multiple times into themes and/or categories, so numbers do not add up

†only a few illustrative comments are quoted here. A full list appears in Appendix 5.2