Addressing healthcare practitioner genetic knowledge, attitude and practices in Gauteng Province, South Africa

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Abstract

Genetic testing and services are crucial in modern healthcare. Genetic testing is increasingly important in the diagnosis and treatment of more common diseases as well as rare diseases. Primary healthcare practitioners (HCPs), including general practitioners (GPs), are often the first contact point for patients seeking genetic information. However, many HCPs lack adequate genetic expertise to assist patients with genetic-related health concerns.

Currently, no data is available on the genetic knowledge, attitudes, and behaviours of HCPs in the Gauteng province of South Africa. We aimed to investigate HCPs' knowledge, attitude and behaviours towards genetics and genetic testing in the Gauteng province of South Africa.

A tailored pre-validated questionnaire was administered online and in-person to targeted HCPs in the private healthcare sector in Gauteng, South Africa, to investigate genetic knowledge, behaviours and attitudes towards genetics and genetic testing.

Of the 61 HCPs who responded (16.3% response), 95% were trained at South African medical schools. GPs, pathologists, physiotherapists, and specialists answered the questionnaire. Self-reported genetics knowledge was poor/very poor for 70% of respondents, making them ill-prepared to cope with the anticipated increasing demand for genetic information in primary healthcare. However, 93% displayed appropriate behaviour to genetic scenarios presented and showed positive attitudes towards providing and referring to, genetic services. Nevertheless, access to specific genetic information and resources was limited. Eighty-seven percent of respondents were interested in further genetic education.

HCPs are interested in and expressed positive behaviours and attitudes towards genetics and genetic services, but their current knowledge is insufficient to cope with the growing demand.

Introduction

Genetic testing and services are becoming increasingly important in generalised healthcare. As the public is becoming more aware of the effect of genetics on health, general practitioners (GPs) are often seen as a starting point to access information on genetic testing and services. Genetic information is increasingly being used in the diagnosis and treatment of more common diseases and is no longer solely restricted to rare diseases. Access to associated genomic data is also improving. However, many GPs lack the appropriate knowledge and skills to assist patients with genetic-related health concerns.

Genetic knowledge, attitudes and practices in everyday healthcare are lacking. International studies have shown that GPs and other healthcare practitioners (HCP) acknowledge a deficit in their genetic knowledge, attitudes and practices (Baars, Henneman, Ten Kate 2005; Emery, Watson, Rose, Andermann 1999; Suther and Goodson 2003). Data on this subject in low- or middle-income countries (LMIC), including South Africa (SA), is lacking. To date, there is no published data on the knowledge, attitude and practices of GPs and other HCPs based in the Gauteng province of SA.
One potential reason for this worldwide shortfall in genetic knowledge, attitudes and practices is the delay in incorporating scientific developments into the medical curriculum and its translation into general practice. While pharmacogenomic medicine is becoming more widely available, it remains poorly understood (Albitar and Alchamat 2021) and is not commonly used. HCPs lack the necessary skills to handle the increasing demand for genetic information and testing and are often unprepared for implementing genetics and genomics into everyday practice. While genetic testing is becoming part of mainstream medicine globally with the introduction of personalised healthcare – many HCPs are unprepared for their role in this service.

**Genetics of common diseases**

Common genetic diseases include disorders such as congenital heart defects (CHD), heart disease, diabetes and psychiatric conditions, and some single-gene disorders (Collins 1999). Personalised medicine is gaining traction with the introduction of preventative medicine and the identification of individuals at high risk for common inherited conditions (Rosso, Pitini, D’andrea et al. 2020). The focus of genetic testing is evolving to include the effect on an individual’s health directly, instead of focusing primarily on the risk to their offspring (Wang, Gonzalez, Merajver 2004).

With "genomics moving from the research bench to the clinical bedside” (Chow-White, Ha, Laskin 2017), it is beneficial for all HCPs to remain up to date with genetic and genomic knowledge and the health implications for their patients. In doing so, it enables earlier diagnoses and care, decreases overall costs, and promotes the further expansion of knowledge related to the genetic and clinical spectra of genetic conditions (Wang, He, Yin et al. 2016). An increase in genetics knowledge of HCPs would also assist in the prevention of inappropriate, unnecessary testing and reduce the associated costs (Willoughby, Aldous, Patrick et al. 2016). The application of personalised medicine can significantly impact future healthcare, including eradicating conditions such as cancer, heart disease, sickle cell anaemia and other physical and mental health problems (Nerlich, Dingwall, Clarke 2016).

**Genetic testing and the family**

Primary HCPs may argue that specialised genetic testing is not included within their scope of practice and that genetics is specialised, complex, and not easily understood. In reality, HCPs are perfectly positioned to understand family structures, dynamics, relationships, and medical histories that are especially relevant for inherited conditions. Such non-genetic specialists are often involved in the healthcare of a patient or family over an extended period before and after a genetics consultation, making their genetics knowledge and insight critical (Harris and Harris 1999). Many are also able to offer basic genetic investigations and counselling. HCPs may be more comfortable with the basic principles of genetic inheritance as taught in medical school and are aware of potential medicine/medicine interactions and their side effects. Increasingly targeted medicines, with the advent of pharmacogenomics, offer another layer of information based on an individual’s genetic makeup,
including genetic variants and polymorphisms. This is an additional tool for HCPs to use to their patients' advantage, through personalised drug prescription, to reduce adverse drug reactions and increase the efficacy of treatment (Burke 2004).

For specific cases, HCPs may refer patients to specialised genetics centres for further investigations and diagnosis of suspected genetic conditions. However, the patient and/or family will still require continued care and medical management undertaken by the primary HCP.

In the context of this data shortfall in genetics knowledge, this study aims to investigate the knowledge, attitude and behaviours towards genetics and genetic testing in HCPs practising in primary healthcare in the Gauteng province, SA.

**Method**

**Questionnaire tool**

A pre-validated questionnaire developed as part of previous research elsewhere but applicable for use with adaptation, GPGeneQ, was used to capture the knowledge, attitudes and practices in this study (Flouris, Hawthorne, Aitken et al. 2010), with permission from Prof S Metcalfe (Pers Comm, Prof Metcalfe 4/6/2017). This questionnaire was adapted to incorporate genetic conditions relevant to the South African context, as recommended by the 2001 Human Genetics Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities produced by the South African Department of Health (Department of Health 2001). The adaptation of the survey was undertaken before the publication of the new Clinical Guidelines for Genetics Services (National Department of Health 2021) in SA, hence the reliance on the 2001 document.

**Structure of the questionnaire**

The questionnaire was divided into five sections: Section 1: Demographics, self-rated genetics knowledge, and current practice; 2: Genetics knowledge; 3: Self-reported behaviour using vignettes; 4: Attitudes; 5: Personal experience of genetics in the workplace; and 6: Interest in further education. See supplementary file 1.

**Study Setting**

In-person Continuing Professional Development (CPD) registered events held during the study period were targeted to recruit respondents (practising HCPs) to participate in a written, anonymous survey. This included two relevant CPD events implemented in Gauteng Province from October 2019 to May 2020, as detailed in Table 1. Professional HCP groups were also targeted and emailed a link to the survey (using Google Forms) to recruit further respondents.
Table 1
HCP event details

<table>
<thead>
<tr>
<th>Healthcare Practitioners in attendance</th>
<th>Event type</th>
<th>Method of questionnaire responses</th>
<th>Time</th>
<th>Place</th>
</tr>
</thead>
<tbody>
<tr>
<td>GPs &amp; physiotherapists</td>
<td>CPD meeting</td>
<td>In-person - Paper-based</td>
<td>5th October 2019 – available for 2 hours</td>
<td>Bryanston Country Club (BCC), Johannesburg</td>
</tr>
<tr>
<td>Pathologists</td>
<td>Desktop-online request</td>
<td>Email link</td>
<td>December 2019 - available for one month</td>
<td>Online</td>
</tr>
<tr>
<td>Endocrinologists</td>
<td>CPD meeting</td>
<td>In-person - Paper-based</td>
<td>22nd January 2020 -available for one hour</td>
<td>Charlotte Maxeke Academic Hospital (CMAH), Johannesburg</td>
</tr>
<tr>
<td>GPs</td>
<td>Desktop – online request</td>
<td>Email link</td>
<td>May 2020 – available for one month</td>
<td>Online</td>
</tr>
</tbody>
</table>

Study Respondents

HCP groups were targeted using convenience sampling including GPs, pathologists and clinical specialists. All were requested to complete the survey anonymously, either on paper (in person) or via a shared email link.

Measuring and Scoring Knowledge, Behaviour and Attitudes

The scoring scheme for each section was performed following the GPGeneQ questionnaire and assessment (Flouris, Hawthorne, Aitken et al. 2010). Specifically: Section 1 collected demographic data about the participants, self-assessment of genetics knowledge and current practice. The knowledge assessment (Section 2) comprised ten true/false/unsure items. A score of 1 was given for a correct answer, a zero for an "unsure" answer, and a -1 for an incorrect answer. Possible scores ranged between −10 and +10.

In Section 3, the behaviour scale implemented included five open-ended questions. Points were awarded for each response with a maximum score per question of 4 points, and a maximum score (for all questions) of 20 points. A score of 3 or 4 points for a question was considered a very appropriate response, a score of 1 or 2 points was appropriate, and a score of 0 was inappropriate. Across all five questions, a collective score of 0–4 points was considered inappropriate, 5–10 points was considered an appropriate response, and 11–20 points was very appropriate for this section. Specific standards were used for scoring to ensure that a variety of responses were considered appropriate.

In Section 4, the attitude scale was composed of 15 questions on a 5-point Likert-type response, ranging from strongly disagree to strongly agree. A score of 2 points was a very positive response, 1 point for positive, 0 for neutral or not answered, -1 for negative and −2 for a very negative response. Scores were
added for a total attitude score ranging from −30 to +30. Ten questions were posed in a positive-framed manner and five in a negative-framed manner.

**Measuring personal experience of genetics**

Section 5 requested information regarding the participants' personal experience of genetics. The responses related to genetic conditions and testing were recorded as free text and summarised into themes during data analysis.

**Interest in further education**

When asked if they would be interested in a CPD-accredited workshop in Section 6 of the survey, respondents could indicate a yes/no answer.

For the full questionnaire/survey see Supplementary File 1.

**Statistical Analysis**

Descriptive statistics made use of means, medians, proportions, and rates of the quantitative data scores were calculated using Microsoft Excel®.

**Results**

A total of 374 potential HCPs were contacted for participation in the survey. A total of 61 responses were recorded, with an overall response rate of 16.3%, as detailed in Table 2. The highest response rate was seen at the in-person event at the Endocrine Clinic, CMAH and the lowest via the online requests to GPs.

<table>
<thead>
<tr>
<th>CPD Event</th>
<th>Number of attendees/potential respondents (n)</th>
<th>Number of responses (n)</th>
<th>% Response rate</th>
</tr>
</thead>
<tbody>
<tr>
<td>BCC</td>
<td>113</td>
<td>20</td>
<td>17.7%</td>
</tr>
<tr>
<td>Email - Pathologists</td>
<td>141</td>
<td>34</td>
<td>24.1%</td>
</tr>
<tr>
<td>Endocrine Clinic, CMAH</td>
<td>5</td>
<td>5</td>
<td>100%</td>
</tr>
<tr>
<td>Email - GPs</td>
<td>115</td>
<td>2</td>
<td>1.7%</td>
</tr>
<tr>
<td>TOTAL</td>
<td>374</td>
<td>61</td>
<td>16.3%</td>
</tr>
</tbody>
</table>

**Section 1: Demographics of Respondents**

Responses in the first section of the questionnaire relating to the respondents' demographics indicated that the majority of the 61 respondents were pathologists (n = 33, 54%), with GPs representing 36% (n =
22) of the cohort, as detailed in Table 3. The remainder of the cohort comprised specialists and physiotherapists. GPs included those self-described as a GP, with a certificate in aesthetics, travel medicine, medical registrar, family medicine, or an interest in pulmonology and geriatrics. Pathologists who responded had sub-specialities in virology, microbiology, histopathology, haematology, chemical and anatomical pathology. Other participating specialist HCPs included physicians, gynaecologists, endocrinologists, and internal medicine physicians.

<table>
<thead>
<tr>
<th>Category of HCPs</th>
<th>Number (n)</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pathologist</td>
<td>33</td>
<td>54.1%</td>
</tr>
<tr>
<td>GP</td>
<td>22</td>
<td>36%</td>
</tr>
<tr>
<td>Specialist</td>
<td>4</td>
<td>6.6%</td>
</tr>
<tr>
<td>Physiotherapist</td>
<td>2</td>
<td>3.3%</td>
</tr>
<tr>
<td>Total</td>
<td>61</td>
<td>100</td>
</tr>
</tbody>
</table>

The age range of respondents:

There was a 50-year age range among the 61 respondents, which included 12 (19%) respondents still practising after retirement age, with five aged between 70–79 years (Fig. 1). Fifteen respondents (25%) were at the beginning of their careers. Most respondents (n = 34; 56%) were aged 40–59 and had some years of experience in their chosen profession.

Training locations of respondents

Only three respondents (5%) received their qualifications at overseas institutions, and 58 respondents (95%) trained at South African medical schools, as detailed in Table 4.
Table 4
Location of institutions where first qualifications were obtained by respondents.

<table>
<thead>
<tr>
<th>Location of medical school</th>
<th>Medical school</th>
<th>Number</th>
</tr>
</thead>
<tbody>
<tr>
<td>International</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Ireland</td>
<td>Trinity College Dublin</td>
<td>1</td>
</tr>
<tr>
<td>Rhodesia (Zimbabwe)</td>
<td>Not reported</td>
<td>1</td>
</tr>
<tr>
<td>United Kingdom</td>
<td>University of Bristol</td>
<td>1</td>
</tr>
<tr>
<td>South Africa</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Medunsa</td>
<td>(Now Sefako Makgatho Health Sciences University)</td>
<td>1</td>
</tr>
<tr>
<td>Stellenbosch University</td>
<td></td>
<td>6</td>
</tr>
<tr>
<td>University of Cape Town</td>
<td></td>
<td>9</td>
</tr>
<tr>
<td>University of the Free State</td>
<td></td>
<td>3</td>
</tr>
<tr>
<td>University of KwaZulu Natal</td>
<td></td>
<td>4</td>
</tr>
<tr>
<td>University of Pretoria</td>
<td></td>
<td>10</td>
</tr>
<tr>
<td>University of the Witwatersrand</td>
<td></td>
<td>25</td>
</tr>
<tr>
<td>Total</td>
<td></td>
<td>61</td>
</tr>
</tbody>
</table>

The year of the first qualification of HCPs ranged from 1966 to 2011. Most HCPs (n = 20; 32%) received their qualifications between 2000 and 2009, with 29% qualifying between 1980 and 1989 (Fig. 2). HCPs had been in practice for between 5 and 50 years, with a mean of 23 years of practice.

Further genetics education

Further genetics education received by HCPs during their post-graduate career was recorded as one or more choices out of five options (Fig. 3). The majority (n = 63; 73%) of further genetics education obtained was via lectures and CPD events. Very few HCPs (1%) reported receiving extra genetics education from conferences.

Genetics knowledge - self-assessment:

HCPs self-reported a deficit in their genetics knowledge (Fig. 4). Forty-three respondents (70.5%) indicated their genetics knowledge to be poor or very poor. Four HCPs (6.6%) (two pathologists and two GPs) reported their knowledge as good or very good. The remaining individuals (23%) felt that their knowledge was "sufficient", although this was not defined.
Discussion of genetics in daily practice, including taking a family history

A third of HCPs (n = 20, 33%) reported that they never discuss genetics with their patients, and 66% of HCPs discuss genetics to varying extents in their practice. Some HCPs only discuss genetics if the patient has already been diagnosed with a genetic condition (n = 13, 21%) or if there is a good reason (n = 17, 28%). Less than 20% of HCPs discussed genetics with their patients often or mostly, as seen in Fig. 5.

Recording of a family history

Figure 6 graphically represents the frequency of different methods for recording family histories. Six HCPs (10%) draw a family pedigree regularly for inpatient consultations. Most HCPs (n = 30, 49%) record family histories as written text only in the patient record/file. Twenty-five respondents (41%) never record a family history.

Comfort level when discussing genetics:

Self-assessment of levels of comfort when discussing genetics issues is represented in Fig. 7.

Just under half of HCPs (n = 27, 45%) surveyed were uncomfortable discussing genetics with patients. Most HCPs (38%, n = 23) were ambivalent and very few (n = 4; 7%) reported that they were comfortable/very comfortable discussing genetics (Fig. 7).

Section 2 results relating to specific genetic/genomic knowledge included:

Section 2: Knowledge Assessment (10 questions)

Section 2 of the questionnaire assessed basic genetic conditions, prenatal testing, and inheritance. The lowest score obtained was -4, and the highest was +4. For example, out of a possible 10 marks, a score of -4 indicated seven incorrect and three correct answers.

Nine respondents (15%) obtained the lowest score (-4) in this series, and six respondents (10%) obtained the highest score (4) (Fig. 8). The overall mean score was 0.56 points. Between the different categories of HCPs, the mean score ranking was highest for physiotherapists with a score of 1 point (n = 2), pathologists = 0.03 points (n = 33); GPs = -1.23 points (n = 22); and specialists = -2.5 points (n = 4) (Fig. 9).

Incorrectly answered questions:

Only five questions were answered incorrectly by all participating HCPs. Scores ranged from −50 (question 5) to -6 points (Question 8). Eighty-seven percent (n = 53) incorrectly answered question 5 regarding haemochromatosis, the genetic result and its clinical implications. Three respondents (5%) responded correctly and five (8%) were unsure (Fig. 10). The sum of the incorrectly answered questions for all HCPs was −147 points.
Correctly answered questions:

On average, five questions were answered correctly. Question 2, regarding diagnostic antenatal testing by chorionic villus sampling (CVS), was answered correctly by most HCPs (n = 53; 87%) with a score of 50 points. There were three (5%) incorrect answers and five (8%) unsure (Fig. 10). The total score for all HCPs’ correct answers was 113.

Section 3: Behaviour Assessment: (5 questions)

Three respondents (two GPs and one physiotherapist) did not answer any of the behaviour-related questions; their responses were excluded from the results for this section. The highest behaviour rating score in Section 3 of the questionnaire was 19 points, and the lowest was 1 (range of scores represented in Fig. 11).

The mean score for the remaining 58 HCPs was 10.8 points. The lowest-scoring question concerned a child with developmental delay (mean score 1.9), and the highest-scoring question was regarding haemochromatosis (mean score 2.5).

In total, across all questions, a generally inappropriate response per respondent was recorded as a score of < 5, an appropriate response recorded as between 5 and 10, and a very appropriate response between 11 and 20. Four (7%) respondents recorded an inappropriate response, 27 (46.5%) had an overall appropriate response, and 27 (46.5%) had a very appropriate response (Table 5).

<table>
<thead>
<tr>
<th>Response</th>
<th>Number of respondents</th>
<th>Proportion (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Inappropriate</td>
<td>4</td>
<td>7</td>
</tr>
<tr>
<td>Appropriate</td>
<td>27</td>
<td>46.5</td>
</tr>
<tr>
<td>Very appropriate</td>
<td>27</td>
<td>46.5</td>
</tr>
<tr>
<td>Total</td>
<td>58</td>
<td>100.00</td>
</tr>
</tbody>
</table>

Of the four HCPs with an overall inappropriate response, two were pathologists, and two were GPs. One of the GPs has been in practice for 30 years, qualifying in 1989, and the other for 53 years, qualifying in 1966. The pathologists had been in practice for a total of 38 and 35 years respectively, obtaining their first qualifications in 1980 and 1983.

On average, GPs scored below the appropriate response range (a score of 2 or more per question) for three of the five behaviour questions (questions 1,2,3). Specialists also scored below the appropriate range for three questions (questions 1,2,5). The remaining pathologists scored appropriately for all five questions.
Section 4: Attitude Assessment (15 questions)

The attitude scale consisted of 15 items, each with a Likert-type scale response from strongly agree to strongly disagree. Possible scores ranged from −30 to +30.

Two GPs did not answer any of the questions on attitudes. They were therefore excluded from this analysis. For the 59 remaining HCPs, the mean score was 15.56 points. Scores range between 6 and 25 points (details in Fig. 12). Question 11 scored the lowest (mean= -1.12) regarding offering prenatal genetic screening only to older women and the highest score (mean = 1.59) was regarding family history-taking being a key component of general practice.

The mean score for GPs was 14.9 points (n = 20; range 7–23), for physiotherapists was 15.5 points (n = 2; range = 12–19), for specialists was 19 points (n = 4; range = 17–20) and for pathologists was 15.55 points (n = 33; range 6–25). Scores were recorded as 0–15 = positive attitude, 16–30 = very positive attitude toward genetics. No HCPs scored an overall negative attitude towards genetics.

Positively-framed question

In general, all HCPs responded positively to nine of the ten scenarios presented in Section 4. However, most (86%) disagreed with the statement that only older women should be offered prenatal screening and believed all women should be offered prenatal screening. Attitudes of the pathologists (Fig. 13) and GPs are displayed graphically (Fig. 14) and show a similarity in the pattern of attitudes towards the different scenarios. Pathologists were more sceptical than GPs, only related to recording partial family history.

Negatively-framed questions

Pathologists and GPs had similar attitudes toward the negatively framed questions (Fig. 15 and Fig. 16). Some GPs preferred that only genetic specialists should be allowed to order genetic tests.

Section 5: Open text questions

Genetic conditions seen in practice:

The number of genetic conditions reported ranged from zero to 31. The group of conditions most frequently reported was haemoglobinopathies (52 times), which included conditions such as sickle cell disease, thalassaemia, haemophilia and von Willebrand's disease. Down syndrome was reported 25 times, and haemochromatosis 20 times. Twelve conditions were reported only once, including Williams syndrome, Wilson's disease, Noonan syndrome, Bartter syndrome, alpha-1-antitrypsin, tuberous sclerosis, a hepatic disorder, MELAS, achondroplasia; cancer-related conditions reported once were von Hippel-Lindau syndrome (VHL), pancreatic cancer, non-Hodgkin's lymphoma. Pregnancy screening was also mentioned once.
Genetic tests requested by HCP:
Twenty-eight respondents (46%) did not answer this question. The remaining respondents reported 15 different genetic tests they had requested in practice. The most reported test request was for haemochromatosis (n = 14), followed by Down syndrome (n = 9) and cystic fibrosis (n = 9). Tests requested only once included complement deficiency, malignant hyperthermia, pharmacogenomics testing, disorders of sex development and galactosemia.

Genetic tests requested but not available:
Forty-eight HCPs (79%) did not respond to this question. Thirteen HCPs (21%) reported the following genetic tests that were requested but not available; multiple endocrine neoplasia (MEN), VHL, pheochromocytoma, pancreatic cancer, red cell membrane, haemoglobin variants, centronuclear myopathy, carrier status for spinal muscular atrophy (SMA), the SDHB gene (related to pheochromocytoma and Cowden syndrome), some primary immunodeficiencies (PIDs) and inborn errors of metabolism.

Genetic counselling referrals:
Thirty-eight HCPs (62%) reported that they had referred patients to a genetic counsellor, 18 (30%) had not, and five (8%) did not respond. Nine GPs (15%) reported the location of their closest genetic counsellor, as did sixteen pathologists (26%). However, 19 HCPs (31%) did not know the location of a genetic counsellor.

New genetic terminology:
There were two questions on new terminology: 21 HCPs (34%) had some understanding of the array comparative genomic hybridisation (aCGH) test, and 40 HCPs (65%) either did not know or were unsure. When asked about the meaning of variant of uncertain significance (VOUS), 47 HCPs (77%) did not know. Two of these HCPs reported they subsequently investigated the term. Fourteen HCPs (23%) were able to define and/or explain the new terminology.

Section 6: Interest in a CPD workshop:
Five respondents (8%) reported no interest in a genetics CPD-accredited workshop, three (5%) did not answer the question, and 53 (87%) reported interest in attending a CPD workshop to improve their genetics knowledge and skills.

Discussion
This study aimed to investigate genetics knowledge, behaviours and attitudes towards genetic services and testing amongst GPs and other HCPs (non-genetic specialists, pathologists and physiotherapists) in
the Gauteng province of SA. In this study, we have demonstrated through empirical evidence, that genetics knowledge, appropriate attitudes, and practices of genetics in everyday healthcare are lacking. HCPs do not have the necessary knowledge/skills/behaviour/ability to cope with the increasing demand for genetic testing. They appear unprepared for the implementation of new genetics and genomics into everyday practice.

**Genetic Knowledge**

In recent times, societal expectations placed on HCPs concerning understanding, testing, and treatment of genetic conditions have increased. This heightened expectation is likely caused by the public's increased awareness and education about genetics, linked to the increasing availability of diagnostic and direct-to-consumer tests, which can provide insight into their future health. HCPs are expected to interpret and apply genetic information to patient care.

Seventy percent (43/61) of HCPs in this study self-rated their genetics knowledge as either poor or very poor. This was corroborated by the lack of knowledge demonstrated by respondents in the questionnaire. This finding is in keeping with other studies undertaken worldwide, in HIC such as Canada, Italy, Netherlands, and Switzerland, (Baars, Henneman, Ten Kate 2005; Carroll, Allanson, Morrison et al. 2019; Chow-White, Ha, Laskin 2017; Harvey, Fogel, Peyrot et al. 2007; Panic, Leoncini, Di Giannantonio et al. 2014).

Equally in HICs, HCPs have limited genetics knowledge, including carrier rates of common genetic conditions in certain population groups, such as haemoglobinopathies, or cystic fibrosis, how to assess common cancer risks where there is a positive family history, or understanding and communicating genetic risk in reproductive choices (Aalfs, Smets, De Haes, Leschot 2003; Emery, Watson, Rose, Andermann 1999; Harris, Lane, Harris et al. 1999). Factors that contribute towards the lack of genetics knowledge include the low probability of encountering a genetics case in everyday practice, lack of evidence of the usefulness of genetic tests and financial barriers (Acheson, Stange, Zyzanski 2005; Bathurst and Huang 2006).

One additional aspect to consider in the lack of genetics knowledge in SA HCPs is the age range and the exposure to genetics education during undergraduate studies. This cohort of HCPs has a 50-year age range, with many qualifying during the early stages of the development of genetic services in SA.

Cytogenetic services first became available in SA in the 1960s, followed by the private sector in the late 1980s (Jenkins 1990). Relevant techniques were not commonly available in SA, and local molecular genetic services were rudimentary and limited in the country in the early 1980s. Genetic information and technology is a rapidly changing field of healthcare, and a continuous effort is required to keep updated. Opportunities to do so may be limited or may be overwhelming, with the expanse of genetic testing and services now available.
Primary HCPs may be under pressure due to their existing workload and the large number of patients to be seen within a specific timeframe; this time limitation puts pressure on HCPs to assess, diagnose and treat a patient within that limited period. Patients with genetic histories need to be cared for differently. A comprehensive family history can take up to 30 minutes, leaving no time for additional steps during a normal consultation (Bathurst and Huang 2006; Cusack, Hickerton, Nisselle et al. 2021).

Other possible reasons for the lack of comprehensive genetics healthcare at the primary healthcare level include limited genetics knowledge because of rapid advancements in genetic testing, other medical interests, perceived lack of patients with genetic issues, concentration on management and care, being overwhelmed in a state system, perceived lack of access to genetic services.

The challenges of including genetics in a GP medical practice are not limited to LMIC. Cusack, Hickerton, Nisselle et al. (2021) report that a lack of genetic knowledge and skills is reported in Australia, the Netherlands, the USA, Canada, Europe and the UK. Barriers to providing a genetic service in the practice include lack of confidence in genetic skills, long consultations, not keeping updated with knowledge, lack of evidence for the usefulness of genetic testing, and knowing who and where to refer to genetic services. Similar to the findings in this study, there was little genetics education apart from a few lectures in medical school.

There was no correlation between the number of years the HCP worked and their level of genetics knowledge, as genetics knowledge is likely more related to specific genetic education and training, with no investigation into the relationship between experience and gain of genetics knowledge over time (Haga, Kim, Myers, Ginsburg 2019; Harding, Webber, Ruhland et al. 2019). (Baars, Henneman, Ten Kate 2005)

The GPs' mean knowledge (-1.23 points) was more than that of the specialists (-2.5 points) but less than that of the pathologists (0.03 points) and physiotherapists (1 point). It is not meaningful to attach any statistical significance to these findings, due to the small number of specialists and physiotherapists in the sample population.

An interesting finding was that the majority (57%) of HCPs expressed some level of comfort or were not uncomfortable about discussing genetics with their patients, despite lacking the necessary genetics knowledge. This is upheld by Douma, Smets, Allain (2016); Harding, Webber, Ruhland et al. (2019), whose studies reported concerns about inaccuracy or misleading genetic information being shared with patients, sub-optimal use of genetic testing and missed referrals. Lapham, Kozma, Weiss et al. (2000), reported that few HCPs were confident in offering genetic services, especially if they lacked knowledge. However, in this study, lack of knowledge did not prevent appropriate behaviour associated with this knowledge, meaning that patients were referred appropriately regardless of their HCPs' knowledge. Truong, Kenneson, Rosen, Singh (2021) reported that HCPs were motivated to refer patients because they did not have the genetic knowledge. They further report that it is unclear when genetic referrals or testing are required. More research is needed in this area to understand the impact on patients and the consequences thereof. Potential risks for patients when HCPs have insufficient genetic knowledge include a misinterpretation of
genetic results, inadequate appropriate counselling, inappropriate or lack of requests for genetic testing and incorrect management and treatment decisions.

**Behaviour:**

Assessing the behaviour of HCPs is challenging, and different approaches have been taken in the genetic environment, as indicated by the published literature on this topic. Flouris, Hawthorne, Aitken et al. (2010) measured behaviour as an assessment of skills. A literature review of core competencies required for HCPs by Tognetto, Michelazzo, Ricciardi et al. (2019) described skills as "abilities". These skills include gathering family histories using an internationally standardised format for a pedigree, identifying families with potential genetic conditions, explaining genetic risks, referring to genetics clinics, obtaining current information regarding the genetic condition, using new genetic technologies, and educating families and fellow HCPs about genetics (Jenkins, Blitzer, Boehm et al. 2001).

The specific concepts explored in the current study included the next steps required for appropriate management, including taking a family history, requesting genetic testing and how and when to refer to specialists and relevant genetic services. The survey questions related to these concepts were framed as open-ended scenarios to prevent information from being missed which may occur with closed questions, and prevent prompting responses (Flouris, Hawthorne, Aitken et al. 2010). When initially developed in Australia, this type of questioning was the first known example of using pre-coded, open-ended items to measure behaviour. (Flouris, Hawthorne, Aitken et al. 2010). To date, no other published studies have used this approach to assess the behaviour of HCPs in SA.

It is encouraging to observe that 93% of respondents answered with appropriate or very appropriate responses to further case management in the scenarios presented. Two pathologists and two GPs (6.9%) had an overall inappropriate response (inappropriate advice or lack of referral) to the scenarios posed. Three of the four qualified in the 1980s and one in the 1960s. While the data set is too small to make a direct correlation, older HCPs may be less familiar with behaviour associated with autosomal recessive conditions or with pregnancy screening. Three respondents did not answer any of the behaviour-related questions. It is unknown whether they were unsure how to respond, had not previously treated a patient with these conditions, or were unable to respond to the questions for other reasons (e.g., time constraints, lack of interest etc).

Responses to the behaviour question (question 2) regarding developmental delay were associated with the lowest overall scores (mean 1.9 points). Nevertheless, twelve of the 20 (60%) GPs recommended an appropriate referral (2 points) either to a paediatrician, developmental paediatrician, or neurologist. The term developmental delay encompasses a broad umbrella of conditions which include amongst others, physiological development, motor development, cognitive development and psychological development (Khan I 2023) and therefore appropriate referral to specialists who care for affected children is an encouraging finding.
Jenkins (1990) refers to unpublished research conducted by Op't Hof, regarding human genetics training at the seven medical schools in SA in the 1980s. One medical school is reported to offer no specific course in human genetics, five schools reported an average of 35 hours of training (ranging from 20 to 48 hours), and one school only provided two hours of human genetics training. It is unsurprising that with such limited focus on undergraduate education around genetics and genetic counselling, and minimal exposure of doctors to genetics training decades prior, would not perform well in either knowledge or behaviours towards genetic-based scenarios.

Interestingly, the behaviour response towards treatment for haemochromatosis was the most appropriate response for behaviour (mean score = 2.25 points for GPs, 2.59 points for pathologists and 2.75 points for specialists) and was the knowledge question with the poorest response. Many of the HCPs participating would likely have experience in managing a patient with haemochromatosis in their practice. It is therefore unsurprising that HCPs were aware of further testing steps for this common genetic condition, despite being unaware of the underlying genetic cause.

**Attitude:**

Fifteen questions scored using a Likert-scale response of strongly disagree to strongly agree, from −2 to +2 points per question, were used to assess attitudes. Six questions were framed in a negative manner (i.e., support groups are of little benefit to individuals/families), which were mixed with nine positively framed questions. This mix of question framing is to control for bias, to ensure a more comprehensive understanding of responses, and to check for consistency of responses, ensuring validity.

Total attitude scores ranged from 6 to 25 points from a possible range of -30 to 30 points, indicating, in general, a positive attitude towards genetics and testing. This survey used both positively framed (nine questions) and negatively framed (six questions) questions to provide more meaningful responses and reduce bias.

The one outlier response in the positively-posed questions specified that only older women should be offered prenatal genetic screening. Most HCPs strongly disagreed (39%) or disagreed (47%) with this, indicating that pregnant women of all ages should be offered prenatal screening. Since the original questionnaire was devised (Flouris, Hawthorne, Aitken et al. 2010), non-invasive prenatal screening (NIPT) has become more widely and commonly used globally, which enables prenatal screening for those women and families that can afford it. The 2020 American College of Gynecologists (ACOG) Practice Bulletin 226 recommends that all women should have access to prenatal screening and diagnostic testing for fetal chromosome abnormalities regardless of age. Our questionnaire, however, was distributed before the new American guidelines were released, with which local gynaecologists attempt to keep updated. HCPs that work with medical aid funding may be updated by the funders through their communication. Nevertheless, in the private healthcare setting in Gauteng, SA, which was the setting for the current study, where the majority of HCPs included in this study work, best practice screening methods would have been available, but possibly not affordable. Therefore, the responses toward
prenatal screening that all women should be offered the option are more in keeping with the updated guidelines.

The outlier of the negatively phrased questions was regarding taking only a partial family history, where most HCPs agreed (34%) or strongly agreed (27%) with this. The reasons for this are unknown. Possible explanations include time constraints on HCPs and a lack of insight into the reasons for requesting a complete family history.

Other findings

Genetic conditions reported:

Haemochromatosis and Down syndrome were the genetic conditions indicated as requiring genetic testing most often. This is unsurprising since the incidences of these conditions in the SA population are reported as 1 in 300 (De Villiers, Hillermann, Loubser, Kotze 1999) and 1 in 525 respectively (Kromberg, Sizer, Christianson 2013). Twelve conditions were mentioned only once, indicating their rarity (Orphanet: an online rare disease and orphan drug database). Of the conditions reported, genetic testing for Wilson's disease and Bartter syndrome are the only ones not routinely offered in SA. It is difficult to comment on the availability of testing for “hepatic disorder” due to a lack of specificity. The remaining conditions reported in this questionnaire have genetic testing available in SA-based genetic laboratories. This is encouraging, indicating that appropriate genetic tests are available locally. However, currently, the associated costs may be prohibitive. Therefore, genetic testing is available locally, but HCPs are unaware of its availability. A comprehensive, accessible information site would enable HCPs to search for and find which genetic tests are available locally or overseas.

Genetic conditions where no genetic test is available:

This question was answered by 21% of respondents. However, of the eleven conditions reported not to be available, “inborn errors of metabolism” is too broad a category, and enzymatic testing for many of the conditions is available locally, particularly in the private sector. Genetic testing within SA is however available for MEN, VHL, pheochromocytoma, red cell membrane, haemoglobin variants, centronuclear myopathy, SMA carrier testing and the SDHB gene. Some genetic testing for subsections of the PIDs is also available. The responses to this survey question indicate that HCPs do not know where to access information on specific genetic tests, even though contact information for the genetics laboratories is available on pathology company websites. To fill this information gap efforts are currently underway by Rare Diseases South Africa NPC to develop a platform for genetic testing for HCPs (M Gomes, pers comm 19/9/2022).

Referral to a genetic counsellor:

It is encouraging to note that nearly two-thirds of GPs have referred patients to a genetic counsellor. However, it is concerning that many are unaware of the closest locations of genetic counsellors. This information is available online in various formats, including the Health Professionals Council of South
Africa (Hpcs) website and medical directories such as Medpages®. An online search would be sufficient to locate a genetic counsellor in the major cities in SA. In a geographically small province such as Gauteng, SA, the major centres of Johannesburg, Midrand, Centurion and Pretoria have at least 10 genetic counsellors available. The challenge lies in the overall lack of the number of trained genetic counsellors, the lack of genetic counsellors employed by the state sector (who only work in urban locations) as no new posts are being created and the lack of genetic counsellors in rural areas, where patients and families would face greater challenges including by travel costs and other related expenses and associated time away from employment.

New terminology:

The two questions on new terminology were included in this study as an updated addendum to the knowledge section of the original questionnaire. Genetic nomenclature has undergone extensive updating in the past decade. HCPs need to understand the newer terminology when reading the latest genetics literature and being able to interpret genetic reports. The "unsure" or "do not know" responses of 65% and 77% confirm that HCPs are not au fait with the latest terminology. Efforts to provide genetic education are suggested to overcome this knowledge gap.

Interest in a CPD workshop:

The majority (87%) of HCPs expressed an interest in attending a CPD workshop to enhance their genetic knowledge and skills. It is accepted that this may be a biased response since the focus of the questionnaire was to delineate the need for these aspects. However, the questionnaire could also have primed the awareness of the HCPs in how their genetics knowledge and skills were lacking in a rapidly moving medical field. Opportunities, especially in the genetics sphere, to attend CPD-accredited workshops are also attractive to maintain accreditation status as an HCP. Burke (2004) states that primary care providers will “benefit greatly” from genetic specialists developing educational tools and practice guidelines so that genetic information will be used appropriately in primary healthcare.

Limitations

The pre-developed survey used in this study was created in Australia, a HIC, and may be skewed towards the Australian public healthcare system in terms of genetic services and testing available. In comparison, this study was implemented in SA, an LMIC, which has a dual healthcare system with approximately 15.8% of the population belonging to medical aid schemes (Cowling 2023). Most of the SA population (>84%) relies on state healthcare services (Department of Health 2017), which may differ significantly from healthcare services offered in Australia. The HCPs included in this study practice primarily in the private sector in SA.

There was a poor response rate from the HCPs approached, possibly indicating a lack of interest in the topic. This resulted in a small sample size, limiting the wider applicability of the findings from this study. The completeness and quality of responses were also a limitation, suggesting the length of the
questionnaire may have been daunting, with some HCPs answering more superficially, possibly due to time constraints. Some sections of the survey remained uncompleted for three HCPs (genetic tests previously requested and genetic tests available in SA: Section 5).

**Conclusion and the way forward**

This study is the first known investigation to assess HCPs' genetic knowledge, attitudes and practices (behaviour) in Gauteng, SA. The self-assessed knowledge of genetics was poor, as borne out by the mean knowledge score in this study. Despite this, the behaviour towards dealing with genetic scenarios was fitting. Attitudes towards genetics testing and services were mainly, although not necessarily strongly, positive.

This study was the first step in assessing knowledge, behaviours and attitudes toward genetics and genetic testing. Future research in the genetic test requests of different medical specialities would provide further information on what testing is required in the local setting, thus providing information on the epidemiology of the population who may not be referred to genetic services. This research also prompted the development of an educational intervention to address the gaps and concerns regarding genetics and genetic testing. It is envisaged that the SA genetic community has much work ahead to create awareness and education regarding genetic conditions for HCPs. A two-pronged approach may also help create more awareness and education about genetics in communities. This would create a population-based demand for genetic services for which HCPs would need to ensure that they were continuously learning and updating their knowledge regarding the availability of genetic services, including testing and counselling. This could be led by patient support groups such as Rare Diseases South Africa NPC in association with relevant professional bodies.

With continuous developments in medical genetics, the hours of undergraduate teaching dedicated to genetics have undoubtedly increased since the report by Jenkins (1990). As genetics continues to infiltrate all aspects of medicine, more attention and time will need to be dedicated to ensuring that HCPs are educated appropriately during their undergraduate years. Equally, HCPs need to continue to expand their genetics knowledge and implementation throughout their careers. The HPCSA, to which all practising HCPs must belong, has already mandated obtaining continuing professional development (CPD) points. The genetics community can offer more opportunities such as focused workshops, meetings and online teaching for HCPs to encourage continuous up-to-date genetics knowledge and practices. Investigating the most effective or engaging genetic educational intervention would bolster the education and confidence of HCPs when faced with a genetic issue. In parallel, a patient-centric educational programme would empower individuals and families to challenge their HCP to ensure the best, appropriate care possible.

More dedicated research questions into discovering which genetic tests are requested or needed is an area for future research. This would inform genetic laboratory service providers where to focus on
developing and implementing new tests ordered by HCPs. Linked with this is a publicly available database of genetic tests that are available in SA so that HCPs can be better informed.

A parallel survey of HCPs in the state sector would give an improved view of all HCPs in SA. Equally, the same survey could be administered to HCPs in all provinces in the country. This would enhance the view of the level of education and whether medical schools or post-graduate courses/or education would be more appropriate.

Further surveys could be performed on investigating the age or experience, linked to a speciality or lack of one in the HCP and their genetics knowledge, to better understand where educational interventions could be focussed.

**Abbreviations**

- ACOG: American College of Gynecologists
- aCGH: array comparative genomic hybridisation
- CHD: congenital heart defects
- CPD: continuing professional development
- CVS: chorionic villus sample
- GP: general practitioner
- HCP: healthcare practitioner
- HPCSA: Health Professionals Council of South Africa
- LMIC: low-middle-income country
- MELAS: Mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes
- MEN: multiple endocrine neoplasia
- PID: primary immune deficiency
- SA: South Africa
- SMA: spinal muscular atrophy
- VHL: von Hippel-Lindau syndrome
- VOUS: variant of uncertain significance
Declarations

Contributions:

Material preparation, data collection and analysis were performed by Sarah Walters. The first draft of the manuscript was written by Sarah Walters and all authors commented on revised versions of the manuscript. All authors read and approved the final manuscript.

Ethics approval and consent to participation

Ethics approval for this study was obtained from the Biomedical Research Ethics Committee of the University of KwaZulu Natal (BREC ref No. BFC222/18). Methods were carried out in accordance with relevant guidelines and regulations. Informed consent was obtained from all participants. Online-completed questionnaires were anonymised before being received by the researcher.

Consent for publication

N/A

Availability of data and materials

The dataset used and analysed during this study is available from the corresponding author upon reasonable request.

Competing interests

The authors declare no competing interests as defined by the Journal of Community Genetics.

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Authors’ contributions

SW drafted the main manuscript text, with editing and reviews from both HM and CA. SW and HM prepared the figures. All authors reviewed the manuscript.

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References


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Figures
Figure 1

Age range of respondents
Figure 2
Number of respondents by date of first qualification

Figure 3
The type of extra genetics education received
Figure 4

Self-rated genetics knowledge
Figure 5
The frequency with which genetics is discussed in daily practice

Figure 6
Frequency of method for recording family history

Figure 7
Comfort level of HCPs discussing genetics issues with patients
Figure 8

Frequency of knowledge score of respondents
Figure 9

The median knowledge scores and ranges of the HCP categories
Figure 10

Total knowledge scores of all HCPs per question

Figure 11
Total scores of behaviours

Figure 12

Scores of HCPs’ attitudes towards genetics
Figure 13

Pathologists’ attitudes towards genetics in positive-framed questions
There is a role for all healthcare practitioners to provide counselling about genetic issues.

- Family history taking should be a key component of general practice.
- Only older women should be offered prenatal genetic screening.
- People benefit from talking to/interacting with other people with the same/similar condition to themselves.
- Support groups provide useful information for individuals and families.
- It is important to discuss the psychosocial implications of genetics with a healthcare practitioner.
- A full family history should be taken for every patient.
- Genetic counselling is very beneficial for individuals and families.
- A partial family history (only 1st degree relatives) should be taken for every patient.
- If a person is a carrier for a recessive condition (e.g. Cystic fibrosis), their partner should also be tested for...

**Figure 14**

GPs' attitude to genetics in positive-framed questions
**Figure 15**

Pathologists’ attitude to genetics in negative-framed questions

**Figure 16**
GPs attitudes to genetics in negative-framed questions

Supplementary Files

This is a list of supplementary files associated with this preprint. Click to download.

- Supplementaryfile1Questionnaire.doc