



Stakeholder knowledge and attitudes toward the use of predictive genetic testing in South Africa

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Abstract

The use of predictive genetic testing, particularly for risk profiling in non-communicable diseases (NCDs), has the potential to benefit public health by decreasing the disease burden and alleviating the pressure on healthcare. It is thus important to assess knowledge and uptake among stakeholders. This study aimed to assess end users' (community, medical practitioners and medical students) knowledge and attitudes regarding the use and support of genomic medicine. A descriptive cross-sectional survey was conducted in Kwazulu-Natal (KZN) among 3 groups of stakeholders ($n = 170$): medical practitioners from both private and public healthcare, medical students from UKZN and community members represented by teachers. Three structured questionnaires using a Likert scale were administered. Responses were stratified by practice type, and a scoring scale was developed. Principal component analysis (PCA) was used to reduce data on all constructs that made up each variable. All three groups showed adequate knowledge and a positive attitude towards use, apart from medical students who were not as keen to support future use in their own practice. Although medical practitioners supported the use of this technology, with necessary guidelines, expertise and affordability, only 18% from private practice reported having used it. PCA reduced data to fewer parsimonious constructs for all 3 groups: common threads included an awareness that genetic testing may improve health and disease outcomes; guidelines for use; and the provision of education to increase awareness, training to bolster expertise and confidence to use these services. Participants in this study attributed a lack of uptake to limited expertise and professional support, and a lack of legislative guidelines. We recommend updating continuing professional development for medical practitioners and promoting community education concomitantly. Provision of guidelines and increased accessibility to resources are important.

Keywords Genomic medicines · Disease risk · Genetic testing · NCDs · PCA

Introduction

While the use of genomic information in diagnostic and therapeutic medicine is growing in the developed world, equitable access and use of this technology is limited in poorer countries. In Africa, health needs such as poverty and malnutrition, infectious diseases and the lack of basic health infrastructure are prioritised over the use of genomic medicine (Séguin et al. 2008). These constraints are also prevalent within the South African (SA) healthcare system, where inequalities in the private and public health sectors

further impede the use of genomic medicine (Wonkam and Mayosi 2014). Apart from the burden of infectious diseases, South Africa is in an epidemiological transition where the burden of non-communicable diseases (NCDs), including hypertension, diabetes mellitus, dyslipidaemia, cardiovascular disease and cancer, has significantly increased over the last 2 decades (Akinyemi et al. 2015). Predictive genetic testing for these outcomes, particularly in the case of high-penetrance genetic variants associated with common types of cancer (breast/ovarian/colon cancer) and hypercholesterolemia, has shown to be both efficacious and cost-effective (McBride et al. 2013). In addition, genomic data may be used to evaluate population interactions between environmental factors and cultural behaviours in relation to disease outcomes (Akinyemi et al. 2015). This may decrease morbidity and mortality and reduce the pressure on an overburdened healthcare system by adopting a preventative, rather

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than curative strategy. Knowledge of risk of NCDs, obtained by predictive genetic testing, may lead to interventions such as behavioural modification to avoid entirely or prolong the onset of such diseases (McBride et al. 2013).

However, the high cost of genomics and lack of technological capacity in Africa are exacerbated by a relatively low level of public and professional understanding of genomics in African countries (Wonkam and Mayosi 2014). This further impedes uptake, and it is necessary to promote education at all levels, beginning at school. Related issues such as biomedical ethics, informed consent, community engagement, privacy and confidentiality, use of genetic information and governance of biorepositories should be included so that the lay public is introduced to genomics (Wonkam and Mayosi 2014). In 2013, Kromberg et al. reported that genetic services available in SA could only meet 10% of the country's genetic needs (based on an estimate of the calculation of the burden of disease). Although there is a provision of genetic services for the predictive testing of NCDs, SA lacks adequate infrastructure to ensure the successful implementation of these services. In addition, the implementation of genomic medicine is dependent on the knowledge and awareness among end users, which includes both the public and medical professionals.

Research pertaining to genomic medicine is advancing rapidly, and it is important that end users are prepared and informed accordingly. While it is essential that the public understands the health and well-being benefits associated with genetic testing, advances will also require health professionals to be equipped with the knowledge and tools in order to optimally implement in clinical practice (Guy et al. 2020). Research conducted in developed countries such as the UK and the USA has shown that medical practitioners do not understand the importance of predictive genetic testing ((Marzuillo et al. 2013; Teng and Spigelman 2014; Avard et al. 2009). Despite increasing evidence supporting the use of genomic in clinical practice, many healthcare professionals have expressed a lack of confidence in implementation (Majid et al. 2011), particularly in relation to interpreting and communicating results to patients (Guy et al. 2020). This indicates the need for educational approaches to help medical practitioners interpret genetic risks correctly (Avard et al. 2009).

Unfortunately, there is a dearth of published research from the African context concerning participation, perspectives and use of genomic medicine and research (De Villiers 2011). It is vital to obtain the opinion of the public specifically regarding non-communicable diseases as they are end users. It is also essential that one identifies and establishes the level of knowledge and perceptions of public health genomics in context, as data has shown that each country varies in their level of knowledge and understanding of genomic medicine. Perceptions also vary according to

their surroundings and what is most influential in the existing healthcare landscape. When the status quo is known regarding the level of knowledge and attitudes towards PHG, the general public can be protected and the medical fraternity educated to avoid the probability of ethical and social discrimination when predictive genetic testing is used. Therefore, this study aims to assess end users' (community, medical practitioners and medical students) knowledge and attitudes regarding the use and support of genomic medicine.

Research methods and design

Study design, site and population

A descriptive cross-sectional survey was conducted in eThekweni in Kwazulu-Natal (KZN) province, among 3 groups of stakeholders considered important in promoting the use of genomic medicine. Medical practitioners from both private and public health sector in the KZN region were sampled ($n = 45$). These practitioners were sampled conveniently from SAMA (South African Medical Association), Ahmed Al-Kadi Hospital, Durban South Practitioners Guild and Osindisweni Hospital and had to be registered with the Health Professions Council of South Africa (HPCSA) and have practiced medicine for at least 2 years. Medical students comprised of second-year students from the UKZN Nelson R Mandela Medical School who had not yet been exposed to the genetic module in their medical curriculum ($n = 79$). They were also conveniently sampled. Representatives of the community included educators from private and public primary schools in Durban and surrounding areas ($n = 46$). Ten teachers from each of the five schools (3 public and 2 private) in the Durban area were selected. We considered educators in this study to be representative of the community as they have tertiary education and access to medical aid and would have the means to access genetic services. Teachers had to be registered with South African Council for Educators (SACE) with private medical aid. This study was approved by the Durban University of Technology's Institutional Research and Ethics Committee (REC 91/16). Permission was sought from KwaZulu-Natal Department of Education, and the registrar of UKZN. All participants signed an informed consent.

Data collection

Three structured questionnaires were designed based on prior studies (De Vos 2011) and an expert focus group in order to gather data relevant for each of the 3 subgroups: medical practitioners, medical students and teachers. All questionnaires were piloted to identify any ambiguities or misinterpretation prior to commencement of the study. The

reliability score (Cronbach's alpha) for knowledge (0.61) and attitude (0.64) indicated an acceptable, consistent scoring for the knowledge and attitude sections for questionnaires (please see attached questionnaires as supplementary material). Questionnaires were self-administered and comprised of Likert-scale questions and evaluated knowledge and attitudes regarding the use of genomic medicine on a 5-point Likert scale: strongly agree, agree, disagree and strongly disagree and undecided.

Data management and analysis

Descriptive and frequency analysis was conducted using SPSS (version 12). Data were collected using a structured questionnaire which was divided into 3 sections. Section A collected demographic data on each of the respondents. Section B elicited information about the respondent's knowledge regarding genomic medicine, and section C collected data about the respondent's attitude to the use of genomic medicine. Knowledge and attitude responses of participants were stratified by practice type for medical practitioners. A scoring scale was used to determine the level of knowledge and type of attitude participants had towards predictive genetic testing. Knowledge levels were divided into three categories: excellent (if participant scored between 32 and 44 points), adequate (if participants scored between 31 and 24 points) and poor (if participants scored 24 points and below). All knowledge-based questions answered correctly were given a point, while incorrect answers were scored as 'zero'. The same criteria applied for scoring of attitudes of participants. Where the response of participant was positive, a point was given, and for a negative answer, no point was given. Participants scoring between 33 and 22 points were categorised with a positive attitude. Participants scoring with points below 21 were identified as having a negative attitude towards predictive genetic testing.

Principal component analysis (PCA) was used to perform dimension reduction on all the constructs that made up each variable. This was performed to determine the highest loading factor, which were the constructs that best represented the variables; knowledge of genomic medicine use and the attitude towards use of genomic medicine. The Kaiser–Meyer–Olkin (KMO) sampling adequacy test and Bartlett's test of sphericity were used to assess the suitability of the data for analysis. The Bartlett's test of sphericity was significant ($p < 0.005$) for all the factors, while a KMO measure value of more than 0.6 was considered acceptable to predict factors. The data analysed was thus suitable for analysis for all three groups for both knowledge and attitudes. The eigenvalue was used to decide which factors to retain. If the eigenvalue was greater than 1, the factor is retained. In addition, varimax rotation was used to rotate retained factors in each application. Rotating factors increases their

interpretability and proportion of variance. The variables with greater than 0.5 loadings and highest loadings were the basis for assigning the new variable's description or 'component' (Kamaruddin et al. 2019).

Results

A total of 169 participants included 45 medical practitioners, 79 medical students and 45 community representatives, and the majority were female (63%). The demographic characteristics of the study sample population from all sample groups are presented in Table 1. There was an almost equal representation of medical practitioners from the public and private sectors with an average of 22 years in practice. More practitioners from the private practice agreed that predictive genetic tests increased prevention of chronic disorders (62% vs 47%), while more practitioners from government practice agreed that genetic testing can contribute to supporting the NHI through health promotion (47% vs 23%). Both groups were in agreement about the necessity for guidelines, expertise and affordability (Fig. 1). More practitioners in private practice (42% in private vs 29% in public) disagreed that testing should only be introduced if it is cost-effective. Although most practitioners in both practice types supported, and would use genetic testing for the

Table 1 Demographic characteristics of general public, medical practitioners and medical students ($n = 170$)

Characteristic	<i>n</i> (%)
General public ($n = 45$)	
Age (mean, SD)	39.3 (11.30)
Gender	
Male	4 (8.70)
Female	42 (91.30)
Medical practitioners ($n = 45$)	
Age (mean, SD)	46.79 (14.65)
Gender	
Male	31 (68.9)
Female	14 (31.1)
Practice type	
Private	14 (31.1)
Government	17 (37.8)
Public	6 (13.3)
Other	4 (8.9)
Number of years in practice (mean, SD)	22. (14.6)
Medical students ($n = 79$)	
Age (mean, SD)	20.22 (2.76)
Gender	
Male	28 (35.4)
Female	51 (64.6)

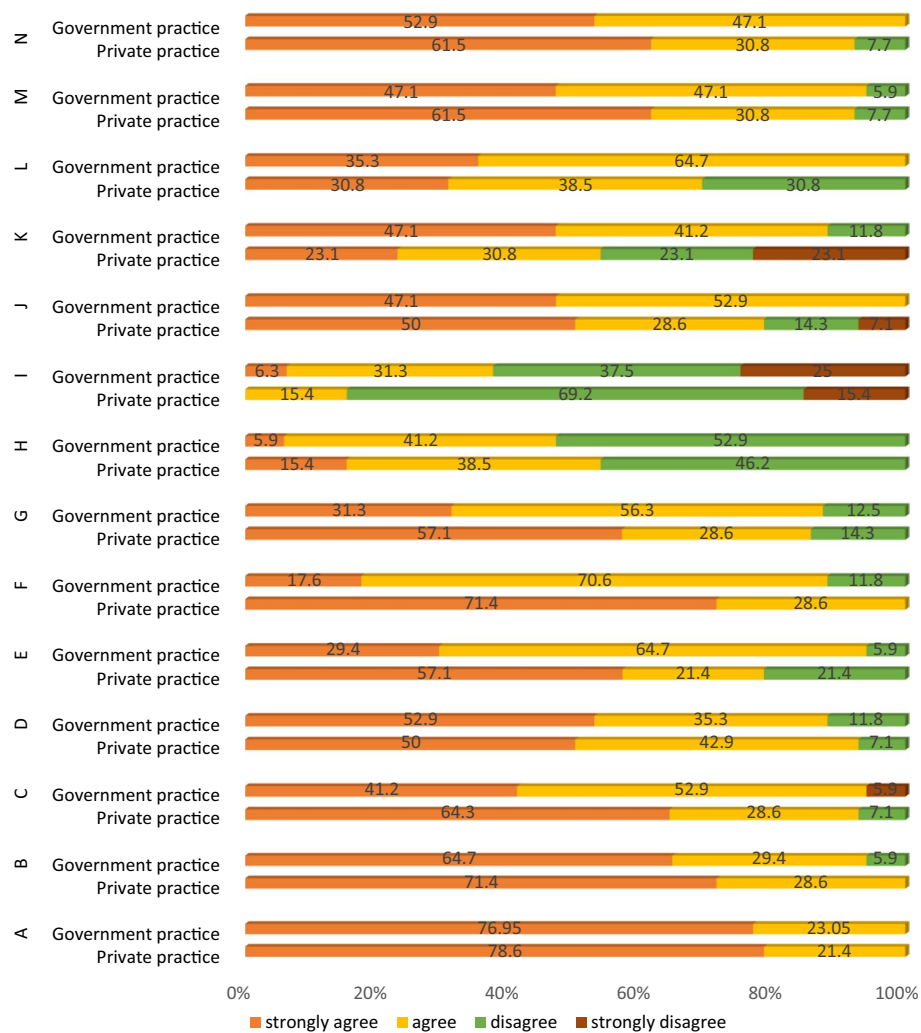


Fig. 1 Knowledge of medical practitioners on genetic testing stratified by practice type ($n=45$). Keys (statement: knowledge of medical practitioners towards genetic testing): (A) performing genetics should be associated with genetic testing; (B) genetic testing can be used to identify a patients susceptibility towards disease; (C) clinical use of predictive genetic testing is to improve the health status of the patient; (D) exposures to various factors can influence a patients risk of disease; (E) there aren't many ethical guidelines in S.A. that govern the use of predictive genetic testing; (F) predictive genetic tests are valid and reliable as long as genetic characteristics are identified in the lab;

(G) there are not many laws in place in SA which protects the patients personal genetic information; (H) I have had exposure to genetic testing as a student; (I) genetic counsellors are needed for patients to consult when they get genetic test results; (J) the use of genetic testing doesn't take into account SLE implications; (K) predictive genetic testing can contribute positively towards health promotion which can assist in the launch of the NHI; (L) guidelines from DOH is needed for the use of predictive genetic testing; (M) predictive genetic tests increase prevention opportunities for chronic disorders

diagnosis and treatment of patients, only 31% (government) and 18% (private) had actually used the service in their practice, which indicated an extremely poor uptake, especially in private care (Fig. 2). Scoring for assessment of knowledge comprised of three categories, i.e., excellent, adequate and poor (Table 2). A similar scoring system was used for attitudes; a positive attitude was linked to a higher score. Overall, knowledge levels of the community representatives with respect to genomic medicine were adequate, and 89% of respondents displayed a positive attitude towards genetic testing. Approximately 78% of medical practitioners from

government and private practice had an excellent knowledge and understanding of genetics, and 82% displayed a positive attitude towards the use of genetic testing. While the majority of medical students (67.1%) had an adequate knowledge of genetic testing, 79% had a negative attitude towards using predictive genetic testing.

PCA was used to reduce the constructs for the variables' relating to knowledge of genomic medicine and attitudes towards use into a few parsimonious constructs for all 3 groups. In the first PCA application of this study, three new factors were retained for knowledge among community

Fig. 2 Attitudes of medical practitioners towards genetic testing stratified by practice type ($n=45$). Keys (statement: attitudes of medical practitioners towards genetic testing): (A) medical and health practitioners require training on predictive genetic testing/genomic medicine; (B) predictive genetic testing should be introduced only if it is proven to be cost effective in SA; (C) I have started using predictive genetic testing for diagnosis and treatment of patients; (D) I will use predictive genetic testing in diagnosis and treatment of patients

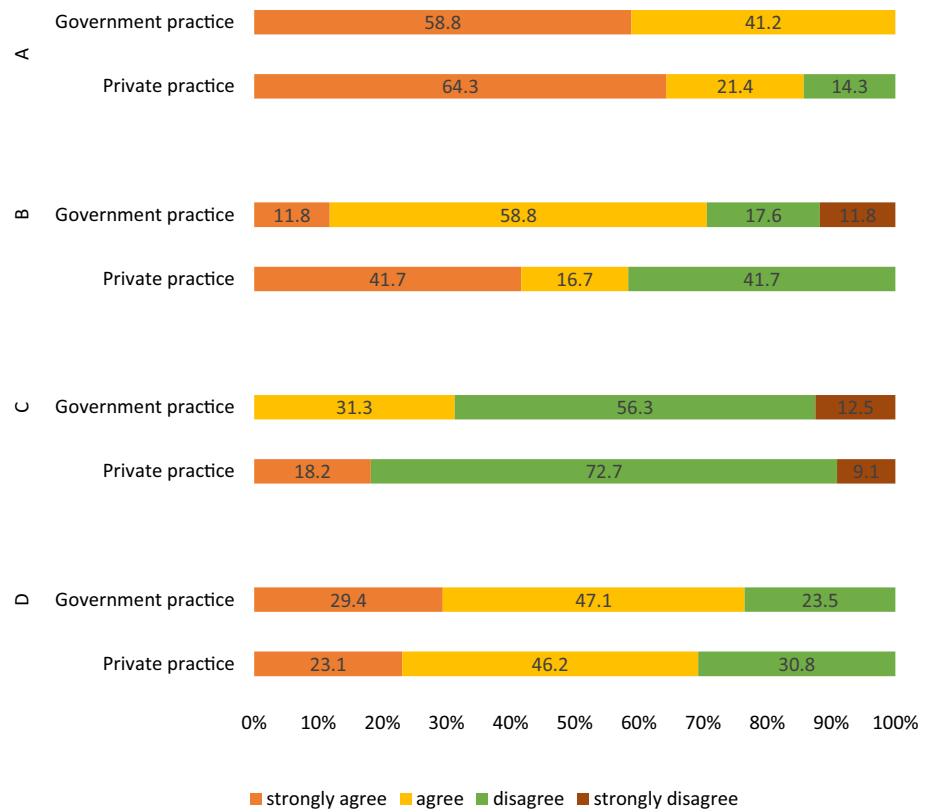


Table 2 Knowledge and attitude scores of participating stakeholders with respect to the use genetic testing

Knowledge	Poor n (%)	Adequate n (%)	Excellent n (%)
Medical students	0	53 (67.1)	25 (32.9)
Medical practitioners	0	10 (22.2)	35 (77.8)
Community representatives	1 (0.47)	36 (76)	9 (4.23)
Attitude	Positive n(%)	Negative n(%)	
Medical students	17 (21.5)	62 (78.5)	
Medical practitioners	37 (82.2)	8 (17.8)	
Community representatives	44 (93.5)	3 (4.3)	

representatives (eigenvalue > 1). They are labeled *disease prevention, lack of guidelines and regulations and availability and access to genetic testing* (Table 3). Four knowledge-related statements loaded strongly against the component disease prevention which indicated that they understood the concept, the use and the advantages of genetic testing. Component 2 relating to the lack of an ethical and legal framework for genetic testing indicated that participants were aware of this limitation. The third component highlighted awareness of genetic services offered in SA, which is important in promoting successful and optimal use of these services. The variance of the three components accounted for 62.6%, p value = 0.000 (Bartlett’s test). Ten variables related to attitude yielded four factors which included *willingness to use predictive genetic testing, confidence in use of genetic testing, emotional and mental attitude and*

importance of improving public knowledge of genetic testing. Even though participants showed a fear of discrimination related to genetic testing results (59%), more than 60% showed a willingness to use it as they perceived that the benefits outweighed the disadvantages. While many would not object to their genetic data used for research purposes, 44% felt that patient consent was mandatory. In terms of use, they do not anticipate that it would be expensive and are comfortable with finding out about their possible health risks. More than 46% supported community exposure to genetic testing (factor loading = 0.89). The variance in these 4 factors accounted for 66.7%, p value < 0.005).

In the second PCA application, 3 new factors were derived from 14 variables evaluating knowledge, i.e. *genetic testing can improve health and disease outcomes, perceived challenges associated with using genetic testing and lack*

Table 3 Principal component analysis of knowledge and attitudes of community representatives

Knowledge-related factors	Loadings
Component 1: disease prevention (Variance 33.17%, eigen 3.32)	
• Genetic testing can be used to determine future susceptibility to certain diseases	0.826
• Genetic testing can be used to also find genetic conditions and treat them	0.721
• Genetic testing can be used to prevent diseases such as diabetes, cancer	0.705
• Having a genetic test is valid and accurate	0.585
Component 2: lack of guidelines and regulations (Variance 15.11%, eigen 1.51)	
• There are few ethical guidelines for human genetic tests in South Africa and these guidelines protect us	0.857
• There are no laws in South Africa for the protection of personal genetic information	0.802
• Genetic testing is useful in helping to use the right medication for treatment	0.667
Component 3: availability and access to genetic testing (Variance 14.3%, eigen 1.41)	
• Genetic testing is being offered in South Africa	0.835
• I know where to have a genetic test done in KwaZulu-Natal	0.816
Component 4: prevention of communicable diseases (Variance 10.24%, eigen 1.02)	
• Genetic testing can be used to prevent HIV, AIDS, T.B	0.929
Attitude-related factors	
Component 1: willingness to use predictive genetic testing (Variance 26.98%, eigen 2.69)	
• I would like to learn more about genetic testing	0.761
• I would like to find out if I am at risk for any chronic disease	0.733
• I would like for my genetic data to be used in scientific research	0.549
• Genetic testing will not be too expensive to use	0.529
Component 2: confidence in the use of predictive genetic testing (Variance 15.84%, eigen 1.58)	
• I am not concerned about my genetic data being used for further studies without my consent	0.766
• I will not be worried about what my genetic test results will be like if I had to have a test done	0.755
Component 3: emotional and mental attitude (Variance 13.76%, eigen 1.37)	
• If my genetic test result is positive for a disease, it will not cause me to be stressed	0.732
• I will not be discriminated against if my genetic test results are positive	0.667
Component 4: importance of improving public knowledge of genetic testing (Variance 10.12%, eigen 1.01)	
• Genetic testing is n important topic for the community to know about	0.897

of legislation for genetic testing in SA (Table 4). While most practitioners (73%) knew of genetic testing, only 50% were aware of it being offered in SA. Component 1 (which accounted for 36% of variance) showed strong support and advocacy for the use of genetic testing. They agreed that genetic testing may improve health outcomes, particularly for chronic diseases, and acknowledged the gene-environment nexus in terms of genetic predisposition (loadings from 0.61 to 0.86). Component 2 highlighted perceived challenges such as the need for genetic counsellors to provide a supportive role and guidelines from the Department of Health to facilitate the appropriate use of genetic testing. This is

aligned to component 3, where the lack of legislation for such testing was highlighted. Variables for attitudes towards the use of genetic testing in their own practice were reduced to 2 components, i.e. *use in clinical practice* and the *need for training in genetic testing*. Only 25% of practitioners reported that they were presently using genetic testing in their practice, while 20% reported that they would not use genetic testing in diagnoses and treatment of patients. This could be related to perceptions that predictive genetic tests should only be introduced if proven to be cost-effective, which presently has not been proven and the lack of appropriate training which is highlighted by component 2.

Table 4 Principal component analysis of knowledge and attitudes of medical practitioners

Knowledge-related factors	Loadings
Component 1: genetic testing can improve health and disease outcomes (Variance 36.44%, eigen 4.00)	
• Genetic testing can be used to identify a patient's susceptibility towards a genetic disorder and NCD	0.858
• Performing genetics should be associated with genetic testing	0.869
• Predictive genetic tests increase prevention opportunities for chronic diseases	0.682
• The clinical use of a predictive genetic test is to improve the health status of the patient	0.671
• Exposures to various factors such as socioeconomic status, lifestyle and environment can influence a patient's risk of disease due to their predisposition	0.619
Component 2: perceived challenges associated with using genetic testing (Variance 19.18%, eigen 2.11)	
• Genetic counsellors are needed for patients to consult with once they have their genetic test results	0.871
• Guidelines form DOH is needed for the appropriate use of predictive genetic testing	0.651
Component 3: lack of legislation for genetic testing in SA (Variance 10.77%, eigen 1.18)	
• Predictive genetic testing is being offered to individuals in S.A	0.798
• There are not many ethical guidelines in South Africa which govern the use of predictive genetic testing among patients	0.787
• There are not many laws in place in S.A. which protects the patient's personal genetic information	0.716
Attitude-related factors	Loadings
Component 1: use in clinical practice (Variance 34.01%, eigen 1.36)	
• I have started using predictive genetic testing for diagnosis and treatment of patients	0.832
• Predictive genetic tests should only be introduced to clinical and public health practice only if it is proven to be cost-effective in S.A	0.765
Component 2: need for training for predictive genetic testing (Variance 30.19%, eigen 1.21)	
• I will use predictive genetic testing in diagnosis and treatment of patients, if trained appropriately	0.793
• Medical and health practitioners require training on predictive genetic testing/genomic medicine	0.749

We derived 4 components from 12 variables relating to knowledge among medical students, i.e. *awareness of the use of genetic testing, understanding of need for legislative framework, benefits of genetic testing and correct use of genetic testing*. Awareness of the use of genetic testing yielded a strong eigen value of 3.27, which indicated that students were aware that genetic testing could be used to prevent disease when used in conjunction with proper laboratory and counselling support. More than half of the students interviewed were aware of predictive genetic testing being offered in South Africa, but conceded that there is a need for understanding the legislative and ethical guidelines for all stakeholders. Students agreed that the use of genetic testing could contribute positively towards disease prevention and encourage health promotion (component 3, eigen = 1.39); however, they agreed that genetic counsellors were essential to facilitate the process of patients receiving their genetic results. These 3 components related to knowledge among students accounted for 52% of the variance. Two components were derived for attitudes of students, i.e. *uptake and use of genetic testing and need for training on*

genetic testing. While students showed a willingness to use predictive genetic testing after they have graduated, they felt that practical training in this field was essential. The factor loading for the need for training on genetic testing is high. Once again, the need for guidelines was highlighted (Table 5).

Discussion

The results of this study supports existing literature, where favourable attitudes to the use of genomic medicine in practice have been identified (Ikeda 2008; Teng and Spielman 2014; Grant et al. 2009; Scheuner et al. 2008). However, despite their positive attitude, they acknowledged that they were limited in the use of this technology. Factor analysis showed that a lack of legislative guidelines and professional training, a paucity of genetic counselling services and inherent cost-effectiveness contribute to this limitation. This is consistent with a study conducted in Italy where the medical fraternity acknowledged the benefits of predictive

Table 5 Principal component analysis of knowledge and attitudes of medical students

Knowledge-related factors	Loadings
Component 1: awareness of the use of genetic testing (Variance 27.32%, eigen 3.27)	
• Predictive genetic testing is being offered to individual in South Africa	0.804
• Performing genetic tests should be associated with genetic counselling	0.650
• Predictive genetic tests are valid and reliable as long as a specific genetic characteristic is identified accurately in the laboratory	0.575
• Predictive genetic tests increase prevention opportunities for chronic diseases	0.566
Component 2 understanding of need for legislative framework (Variance 13.10%, eigen 1.57)	
• There are ethical guidelines in South Africa which govern the use of predictive genetic testing among patients	0.824
• The clinical use of a predictive genetic test is to ultimately improve the health status of the patient	0.760
• There are laws in place in South Africa which protects the patient's personal genetic information	0.549
Component 3: benefits of genetic testing (Variance 11.61%, eigen 1.39)	
• Exposure to various factors such as socioeconomic status, lifestyle and environment can influence a patients' risk of disease due to their genetic disposition	0.824
• Predictive genetic testing can contribute positively towards health promotion and disease prevention which can assist in the launch of NHI	0.693
Component 4: correct use of genetic testing (Variance 8.84%, eigen 1.06)	
• Genetic counsellors are needed for patients to consult with once they have their genetic test results	0.853
• Genetic testing can be used to identify a patients' susceptibility towards a genetic disorder and a non-communicable disease	0.483
Attitude-related factors	
Component 1: uptake and use of genetic testing (Variance 39.14%, eigen 1.95)	
• The curriculum should be extended to capacitate the medical students regarding predictive genetic testing	0.728
• Predictive genetic tests should only be introduced to clinical and public health practice only if it is proven to be cost effective in South Africa	0.719
• I will use predictive genetic testing in diagnosis and treatment of patients when I qualify as a medical practitioner	0.661
Component 2: need for training on genetic testing (Variance 24.25%, eigen 1.21)	
• Guidelines from the department of health is needed for the appropriate use of the predictive genetic testing	0.838
• Medical and health students require practical training on predictive genetic testing/genomic medicine	0.738

genetic testing despite their limited use (Marzuillo et al. 2013), while a study in Japan revealed that support for using genomic medicine was related to experience in practice, where older practitioners were more likely to explore newer medical technology to improve the quality of health care (Ikeda 2008).

This study determined the knowledge and attitudes of medical practitioners, medical students and community representatives towards the use of predictive genetic testing, particularly for chronic diseases. All three groups showed adequate knowledge and positive attitude towards use, apart from medical students who were not as keen to support future use in their own practice. While medical practitioners supported the use of this technology, with necessary guidelines, expertise and affordability, only 18% from private practice reported having used it. PCA reduced data to fewer parsimonious constructs for all 3 groups: common

threads among the groups included an awareness that genetic testing can improve health and disease outcomes; a need to improve guidelines for use; and the provision of education to increase awareness and training to bolster expertise and confidence to use these services. Although 78% of medical practitioners in this study showed an excellent knowledge of genomic medicine, this knowledge was attributed to self-learning rather than specific training. Most medical practitioners from both the public and private sectors revealed that they either attended conferences, workshops or completed online courses to increase understanding. While increased knowledge may improve confidence, it may not relate to use in practice, as knowledge alone does not translate to the skills set required to optimize use of genetic testing. Only 18% in private and 31% in government practice have reported using it in the diagnosis and treatment of their patients, which was associated with numerous challenges as

discussed previously. Use in the public healthcare sectors probably related to oncology and prenatal diagnoses. The use of genetic testing in early diagnosis and disease risk profiling of patients, particularly for chronic diseases, has the potential to significantly improve prognosis and treatment, while decreasing morbidity and mortality (Diaz et al. 2014). This is particularly relevant in the SA context, where the growing burden of NCDs has placed increased pressure on healthcare resources. However, there is limited research in the South African context to evaluate current advocacy and uptake for genetic testing.

Other studies have supported the requirement of a comprehensive skill set and understanding prior to implementation (Houwink et al. 2014 and Guy et al. 2020). It was disconcerting that most of the 50% of medical practitioners who indicated that they were not exposed to genetics are in the public sector, which underpins the need for training which includes counselling and treatment post results. While it would be optimal to have genetic counsellors, it is not always economically feasible in SA public healthcare, so the medical practitioner may need to step into this additional role. The practitioners are aware of the possibility of patients being discriminated against and their test results being used for research purposes without consent. This necessitates the development of ethical and legal guidelines to promote uptake. Contrary to previous studies (Rahma et al. 2020, Wonkam et al. 2006), we found that medical students (79%) were not keen on adopting the use of predictive genetic testing once they qualified as medical practitioners, even though half the students felt that genetic testing should be implemented in South Africa, if it is proven to be cost-effective. It should be noted that these second-year students were not exposed to the genetic modules within the curriculum at the point of data collection. Prior studies among healthcare students have reported a positive attitude and an interest in use (Lanuale et al. 2014), so this attitude among our students may change once they are educated regarding the potential of using the technology. Notwithstanding their attitude, students showed good knowledge of genomics, which alludes to prior learning in the high school Life Sciences curriculum. Factor analysis indicated that students were aware of testing; they agreed that use of genetic testing could contribute positively towards disease prevention and encourage health promotion (benefits), but were cautious with respect to correct use, proper training, additional services and guiding legislation. As with medical practitioners, knowledge did not equate to increased potential uptake.

Aside from those professionals who advocate use, end users are critical considerations in promoting this technology. Previous data from other countries evaluating community attitudes and knowledge towards predictive genetic testing has revealed a lack of knowledge and misinterpretation of use (Gollust et al. 2015). In contrast, 76% of community

members in this study showed an adequate knowledge, and 93% had a positive attitude to testing. Although many were aware of predictive genetic testing being offered in South Africa, only 24% of participants knew of facilities available to have this type of testing done, while 14 participants knew where to access a genetic test in the KZN region. We assumed that their access to education and private healthcare would translate to increased awareness and use, which was not evident. PCA indicated that community representatives were aware of both the lack of ethical guidelines/legislative framework from a user perspective and were concerned about protection of privacy and possible discrimination. This was also indicated by the factor related to emotional and mental well-being. While many would not object to the use of their genetic information for other purposes, they felt that personal consent was mandatory. This is not aligned with current ethical procedures; usually, a researcher will seek gatekeeper permission and consent from the Department of Health (SA) and the health facility itself and block all personal identifiers, assuming that patient consent is then unnecessary if prior conditions are met. Research consortiums such as H3Africa draw increased attention to a number of longstanding and emerging issues in genetic and genomic research, such as informed consent, community engagement, privacy and confidentiality, use of genetic information and governance of biorepositories (Mitropoulos et al. 2015). While a study in the UK showed that only 1 in 20 of 4050 participants would support use given the high cost (Cherkas et al. 2010), our participants were positive towards uptake despite costs, as they perceived that the benefits outweighed the disadvantages. The factor relating to improving public knowledge of genetic testing is important as it may significantly influence uptake. A web-based genetic application was developed at the Witwatersrand University (SA) to create awareness and educate the community (University of the Witwatersrand 2019). Such initiatives could play a vital role in educating individuals about the fundamentals of genetics prior to undergoing genetic testing, provide unbiased information about benefits and challenges and details of various service providers. The COVID-19 pandemic has sensitized people to genomics (given the use of mRNA-based vaccines) so it is an optimal space to build on this knowledge. It is likely that the poor vaccine uptake is related to misconceptions about the use of genomics (Mitropoulos et al. 2015), and improving public knowledge through various platforms is essential.

It is important to support and promote the use of predictive genetic testing, particularly for early risk profiling of NCDs. Once users are aware of their predisposition, they may be advised on behaviour modification and lifestyle changes to either delay the onset of disease or to avoid contraction of the disease entirely (Grant et al. 2009). These factors could include alcohol consumption, smoking, diet,

exercise, stress and adverse environments. A short-term investment could result in a long-term reward, with respect to decreasing the burden of disease and costs of healthcare. This speaks directly to the National Health Insurance strategy of highlighting preventative healthcare, so that curative care is decreased. All participants in this study were knowledgeable of these services and understood the potential in healthcare, but they also recognized the need for additional training and guidelines for use. Costs and accessibility are mitigating factors, but many agreed that these limitations should not affect uptake. This is the first study to collectively assess the knowledge and attitudes of both advocates and end users of this technology. This is essential in constructing a roadmap for the successful implementation of genomic medicine and testing, particularly with the NHI framework, so that gaps in knowledge, barriers and challenges may be addressed.

Limitations of the study

We intended to gain a ‘snapshot’ understanding of different groups of stakeholders, and the sample sizes were small in each stratum. The community was represented by teachers, who are educated and have access to private medical aid, which does not represent the majority of South African citizens. This was done on the premise that they would have greater opportunity to access these services.

Conclusion

Data from this study suggests that the knowledge and attitudes towards the use and acceptance of predictive genetic testing for NCDs are still in its infancy in SA, even though there was good knowledge and support among stakeholders. This study reveals the gaps and potential measures to address these gaps. Participants in this study attributed a lack of uptake to a lack of expertise and professional support, high costs and a lack of legislative guidelines. It is also important to consider ethical and legal issues. We recommend updating continuing professional development for medical practitioners in this field and promoting community education concomitantly. Academic support should be underpinned by governmental support to provide guidelines for practice and increase accessibility to resources. Future research should be conducted to collect empirical data from representative stakeholders (service providers, genetic counsellors, etc.), community representatives accessing public healthcare and young medical graduates, which may lead to conclusive deductions and generalisations. This may

ensure development of effective guidelines and educational programs.

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Data availability The data that supports the findings of this study are available upon request from the corresponding author.

Declarations

Conflict of interest The authors declare no competing interests.

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References

- Akinyemi RO, Ovbiagele B, Akpalu A, Jenkins C, Sagoe K, Owolabi L, Sarfo F, Obiako R, Gebreziabher M, Melikam E, Warth S, Arulogun O, Lackland D, Ogunniyi A, Tiwari H, Kalaria RN, Arnett D, Owolabi MO (2015) Stroke genomics in people of African ancestry: charting new paths. *Cardiovasc J Afr* 26(21):S39-49
- Avard D, Bucci LM, Burgess MM, Kaye J, Heeney C, Farmer Y (2009) Public Health Genomics (PHG) and public participation: points to consider. *J Public Deliberation* 5(1):1–21
- Cherkas LF, Harris JM, Levinson E, Spector TD, Prainsack B (2010) A survey of UK public interest in internet-based personal genome testing. *PLoS ONE* 5(10):1–6
- De Villiers PJT (2011) Are you ready for genomic medicine? *S Afr Fam Pract* 53(2):153
- De Vos AS (2011) *Research at grass roots*. Van Shaik Publishers, Pretoria
- Diaz VA, Mainous AG, Gavin JK, Wilson D (2014) Racial differences in attitudes toward personalized medicine. *Public Health Genomics* 17(1):1–6
- Gollust SE, Gordon ES, Zayac C, Griffin G, Christman MF, Pyeritz RE, Wawak L, Bernhardt BA (2015) Motivations and perceptions of early adopters of personalized genomics: perspectives from research participants. *Public Health Genomics* 15(1):22–30
- Grant RW, Hivert M, Pandiscio JC, Florez JC, Nathan DM, Meigs JB (2009) The clinical application of genetic testing in type 2 diabetes: a patient and physician survey. *Diabetologia* 52(11):2299–2305
- Guy JW, Patel I, Oestreich JH (2020) Clinical application and educational training for pharmacogenomics. *Pharmacy* 8(3):163
- Houwink EJF, Teeffelen SRV, Muijtjens AMM, Henneman L, Jacobi F, Luijk SJV, Dinant GJ, Vleuten CVD, Cornel MC (2014) Sustained effects of online genetics education: a randomized controlled trial on oncogenetics. *Eur J Hum Gen* 22(3):6
- Ikeda W (2008) The public's attitudes towards the use of genetic information for medical purposes and its related factors in Japan. *Community Genet* 11(1):18–25

- Kamaruddin SM, Misni A, Ahmad P, Yaman R (2019) Application of principal component analysis: deriving influencing factors of teenagers' recycling practice. *IOP Conf. Ser: Earth Environ Sci*, 305
- Kromberg JGR, Sizer E, Christianson AL (2013) Genetic services and testing in South Africa. *J Community Genet* 4(3):413–423
- Lanuale C, Leoncini E, Mazzucco W, Marzuillo C, Villari P, Ricciardi W, Boccia S (2014) Public Health Genomics education in post-graduate schools of hygiene and preventive medicine: a cross-sectional survey. *BMC Med Educ* 14(213):1–4
- Majid S, Foo S, Luyt B, Zhang X, Theng Y, Chang Y, Mokhtar IA (2011) Adopting evidence-based practice in clinical decision making: nurses' perceptions, knowledge, and barriers. *J Med Libr Assoc*, 99(3)
- Marzuillo C, De Vito C, Boccia S, D'Addario M, D'Andrea E, Santini P, Boccia A, Villari P (2013) Knowledge, attitudes and behavior of physicians regarding predictive genetic tests for breast and colorectal cancer. *Prev Med* 57(5):477–482
- McBride CM, Bowen D, Brody LC, Condit CM, Croyle RT, Gwinn M, Khoury MJ, Koehly LM, Korf BR, Marteau TM, McLeroy K, Patrick K, Valente TW (2013) Future health applications of genomics: priorities for communication, behavioral, and social sciences research. *Am J Prev Med* 38(5):556–565
- Mitropoulos K, Al Jaibaji H, Forero DA, Laissue P, Wonkam A, Lopez-Correa C, Mohamed Z, Chantratita W, Lee MTM, Llerena A, Brand A (2015) Success stories in genomic medicine from resource-limited countries. *Hum Genomics* 9(1):1–7
- Rahma AT, Elsheik M, Elbarazi I, Ali BR, Patrinos GP, Kazim MA, Alfalasi SS, Ahmed LA, Al Maskari F (2020) Knowledge and attitudes of medical and health science students in the United Arab Emirates toward genomic medicine and pharmacogenomics: a cross-sectional study. *J Personalized Med* 10(4):191
- Scheuner MT, Sieverding P, Shekelle PG (2008) Delivery of genomic medicine for common chronic adult diseases: a systematic review. *Jama* 299(11):1320–1334
- Séguin B, Hardy BJ, Singer PA, Daar AS (2008) Genomics, public health and developing countries: the case of the Mexican National Institute of Genomic Medicine (INMEGEN). *Nat Rev Genet* 9(1):S5–S9
- Teng I, Spigelman A (2014) Attitudes and knowledge of medical practitioners to hereditary cancer clinics and cancer genetic testing. *Fam Cancer* 13(2):311–324
- Wits students create genetics app: Wits science students have created an app to teach people the basics about genetics (2019). <https://www.wits.ac.za/news/latest-news/research-news/2019/2019-03/wits-students-creategenetics-app.html>. Accessed 6 Sep 2022
- Wonkam A, Mayosi BM (2014) Genomic medicine in Africa: promise, problems and prospects. *Genome Med* 6(2):1–3
- Wonkam A, Njamnshi AK, Angwafo FF (2006) Knowledge and attitudes concerning medical genetics amongst physicians and medical students in Cameroon (sub-Saharan Africa). *Genet Med* 8(6):331–338

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