Healthcare practitioners’ knowledge, attitudes and practices of genetics and genetic testing in low- or middle-income countries - A scoping review

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Research Article

Keywords: Knowledge, attitudes, practices, primary healthcare practitioner, low- or middle-income countries, scoping review, education

Posted Date: February 6th, 2023

DOI: https://doi.org/10.21203/rs.3.rs-2077021/v1

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Abstract

Background

It is twenty years since the human genome was published. The role of the primary healthcare practitioner (pHCP) in diagnosing and caring for individuals with genetic conditions is envisioned to increase as this knowledge is applied to enable individualised treatment. It is imperative that their genetics knowledge and practices stay current and in line with local or international guidelines.

Method

This scoping review aimed to identify articles in the peer-reviewed, published literature to understand the knowledge, attitudes and practices of pHCP related to genetics, genetic testing and genetic services. Peer-reviewed, full text, journal articles in English focused on human genetics/genomics knowledge, attitudes and practices in low- or middle-income countries (LMIC) and published between January 1990 to April 2022 were included. Both quantitative and qualitative aspects of the literature were examined.

Results

Twenty-eight articles from 16 LMIC across five World Health Organisation (WHO) -defined regions met the inclusion criteria. The number of articles in LMIC has increased over the years and included articles ranged from 1991 to 2021. The South East Asia Region (SEAR) published the most articles (n=8; 29%) and the Western Pacific Region (WPR) the least (n=2; 7%). Ten countries published only one article each, whereas Brazil published the most (n=6; 22%). Eleven articles reported on single gene disorders (39%) and new technologies of pharmacogenomics and genome editing were reported once each (3%). Nineteen articles included aspects of attitude towards genetics, while eight included practices.

Primary HCP lack of knowledge was evident in genetic diseases, emerging technologies, referral of patients to genetic specialists and clinical guidelines for managing genetic conditions. Attitudes towards genetic services and new technologies were generally positive. Barriers to genetic services identified, included inadequate genetic services and required capacity, financial limitations and religious reasons.

Conclusions

Addressing and implementing education of pHCP is necessary to enable appropriate care for patients and families in need. There is a lack of published literature in LMIC regarding pHCP genetic knowledge, attitudes and practices. More research is required on educational interventions for practising pHCP and how to improve their ability to care for patients and families with genetic conditions.

Background

Since a working draft of the human genome was first made available in 2000 (1), the availability and demand for genetic services has increased, along with the hope of increased personalised, or
individualised medicine (2). Primary healthcare practitioners (pHCP) are generally the first point of contact and source of referral for genetics clinics for most patients (3). With a limited number of genetics healthcare professionals worldwide, there is a global move to incorporate genetics and genomics into primary healthcare. This creates a need for partnership (4) and requires pHCP to have the knowledge and ability to identify genetic conditions, order diagnostic or predictive genetic testing and translate results for the patient and/or family.

The clinical genetics field is expanding rapidly, including for rare, single gene conditions, complex conditions, and genetic predisposition to common diseases, potentially caused by multiple genes. Advances in identifying these conditions have been enabled by an increase in available genomic testing, technology and an array of testing options (5), at decreasing cost. In just over a decade, genetic and genomic testing evolved from linked marker analysis to targeted analysis of specific mutations within a gene/s, to the advent of next-generation sequencing (NGS) technology - where many genes, and even whole exome (WES) and genome testing (WGS) can be conducted.

Knowledge and awareness of genetics-related skills are necessary for optimal implementation of genetics applications into practice. The extended delay in integrating new technology and evidence-based findings into clinical practice is well documented (6). Few non-genetic pHCP feel adequately prepared to discuss, test and perform risk assessments for many genetic conditions, and interpret the genetic test results (7), especially as genetic terminology becomes increasingly complex. Genetics is a rapidly evolving science, and pHCP need to be constantly kept up to speed.

Low- or middle-income countries (LMIC) face severe resource constraints which impact healthcare services. Specific health challenges include low life expectancy at birth, high infant and under-5 mortality rates, and poor educational and health outcomes, due to a lack of access to quality healthcare (8). Extensive work on genetics knowledge, attitude and practices has been conducted and reported in more highly resourced, high-income countries (HIC) but is lacking in LMIC, particularly in Africa. In South Africa, it is unknown how familiar and comfortable pHCPs are with genetics and the appropriate use of genetic tests.

To address this unknown component, a scoping review was conducted to interrogate the published literature for pHCP's genetic knowledge, attitudes and practices towards genetics and genetic testing in LMIC. The aim was to gather as much information about the topic, to identify key articles and/or authors, and to investigate the type of work being performed.

**Methodology**

The scoping review was conducted in April 2022, guided by the Arksey and O’Malley framework (9) and in compliance with PRISMA-ScR guidelines (10).

Eligibility Criteria
The search period reviewed spanned January 1990 to April 2022. This period was selected to ensure a ten-year period prior to the publication of the human genome project (working draft) when genetics practice was introduced into mainstream healthcare. Inclusion requirements for the study, included:

- Original, peer-reviewed research;
- Surveys and questionnaires on the knowledge, attitudes and practices of primary related to human genetics and genomics;
- Full text articles available in English, and
- Focus on LMIC as defined by the World Bank (11).

Articles were excluded that were:

- Unrelated to human genetics/genomics, knowledge, attitudes, and practices
- Focused /performed in HIC;
- Surveys involving super-specialists only (e.g., fetal medicine specialists, clinical geneticist/genetic counsellors etc);
- Focused on treatment, ethics, counselling perspectives, personal perspectives, or solely patient perceptions;
- For education purposes only;
- Grey literature, clinical audits, case studies, training programmes, policy articles, scale assessments or tool validation, and
- Unavailable in English or full text versions.

Information Sources and Search Strategy

Comprehensive literature searches were undertaken in three electronic databases selected due to their coverage of both medical and educational papers and accessibility via the online library at the University of KwaZulu Natal (UKZN). These included Web of Science, PubMed and EBSCO.

A Boolean search string was developed for use in these databases, specifically: ("health* practitioner" OR "health* professional" OR "health* provider" OR doctor* OR specialist* OR consultant*) AND (Attitudes OR Knowledge OR educat* OR competenc*) AND (Genetic* OR Genomic* OR inherit* OR herit* OR congenital). Terminology to specify LMIC was not included in the Boolean string, to maximize and capture the full scope of the literature.

Study Selection

Search results were imported into the reference manager software EndNote 20 (12) where duplicates were removed, and obviously irrelevant articles were excluded. The first round of screening was undertaken independently by all three team members (SW, HM and CA) based on title and abstract. Discrepancies were resolved via discussion. The second round of screening of full text papers was undertaken by SW,
following pilot testing by the team on a random set of articles to determine the final inclusion/exclusion criteria. Eligible papers were imported into NVivo® (13), a qualitative data software tool for data collection, organisation, extraction, coding, and analysis.

Data abstraction and data synthesis

A data abstraction template was developed and piloted to enable data abstraction by SW, including country of study, date, focus, methodology, conditions studied etc. The articles were coded and analysed by SW using NVivo®, from which a qualitative framework was developed.

Results

A total of 3274 citations were identified. Following the removal of 650 duplicates and 840 irrelevant articles, 1784 remaining articles were evaluated on title and abstract (first-round screening) to exclude those not meeting the inclusion criteria. Full text of 360 eligible articles was screened (second round screening), resulting in exclusion of a further 332 articles, leaving 28 eligible articles for inclusion and data abstraction (See Figure 1).

Study scope/characteristics

Geography of articles:

Figure 2 summarises the geographical distribution of articles across World Health Organization (WHO) regions. The 28 included articles were published in 16 countries and across five WHO regions (Supplementary Table 1). Regionally, most were published in the South East Asia Region (SEAR) (n=8; 29%). These emanated from four countries from a possible 11 LMIC in the region (36%); India (2), Indonesia (1), Jordan (1), Sri Lanka (4).

There were four articles included from four countries in the Africa region (AFR), all from Sub-Saharan Africa (SSA). This included two (7%) from Western Africa (Ghana and Nigeria), and one each (4%) from Central Africa (Cameroon) and Eastern Africa (Ethiopia). No articles were found from Northern or Southern Africa sub-regions. In the Region of the Americas (AMR) two countries - Brazil and Mexico - published seven articles collectively (out of 25 LMIC in the region; 8%). In the Eastern Mediterranean region (EMR) articles were included from four countries (out of 16 LMIC; 25%); Lebanon, Pakistan, Iran and Syria. Two countries (out of 23 LMIC; 8.7%) published articles in the Western Pacific region (WPR); China and Malaysia (Figure 2).

At a country level, Brazil (AMR) published the most articles (n=6; 22%). Sri Lanka (SEAR) published four articles (14%) and there were two each from Lebanon (EMR), India (SEAR), Pakistan (EMR) and Iran (EMR). The remaining countries had only one article each.

Timing of publications
The first included article was published in 1999 in Mexico (AMR). Nine articles were published between 2000-2009, a further 16 articles between 2010-2019 and two articles from 2021-2022 (Figure 3). While the number of articles increased in the past decade, no articles were published in the EMR after 2013 until 2021. Articles from AFR first appeared in 2007 (Nigeria), and in the WPR, the first article was published in 2013, from Malaysia.

**Scope of Genetics described in the study**

Most of the specific genetic conditions identified during the review were included as a part of broader concepts, e.g. prenatal diagnosis and termination of pregnancy (TOP) were grouped as one concept (n=5, 18%) and related to a variety of genetic conditions. Eleven of the 28 articles (39%) researched single gene disorders/group of disorders (sickle cell anaemia/disease (SCD), primary immune deficiencies (14), thalassaemia, Huntington’s disease, bleeding disorders and Brugada syndrome). General genetics and/or biochemistry was included in four articles (14%), and new reproductive technologies were discussed in three (11%). Diagnosis and treatment of genetic conditions were discussed in two articles (7%), and genetic counselling was the focus of one article (3%). The newer technologies of genome editing, and pharmacogenomics were reported in only one article each (Figure 4).

Sickle cell anaemia was the individual genetic disorder with the greatest number of articles (n=3, 11%), and as a collective, the inherited blood conditions were the most frequent (SCD, thalassaemia, bleeding disorders) (n=6, 21%).

**Study types**

The majority (96%) of studies used quantitative methodologies and a few incorporated a qualitative component, including the use of an interview tool (15). Cross-sectional studies, using convenience sampling and a survey or questionnaire tool, were the most common methodologies used in the included articles.

For a detailed overview of characteristics of the included articles, see Supplementary Table 1.

**Topics**

All articles incorporated a combination of genetics/genomics knowledge, attitudes, and practice of healthcare workers, with the aim of improving the provision of genetic healthcare services in their countries (Figure 5). Articles incorporate either one, two or all three aspects of knowledge, attitudes and practices, but the majority focused on attitude only (36%, n=10). Six (21%) focused on attitude and knowledge and only one article (4%) on attitude and practice.

The coding of the full text of the articles using NVivo®, was used to develop a thematic framework (Figure 6).

**Discussion**
This review aimed to interrogate the published literature for pHCP's genetic knowledge, attitudes and practices towards genetics and genetic testing in LMIC. To our knowledge, this is the first review of this kind. Over the 32-year period of study (1990-2022) only 28 articles met the inclusion criteria from 16 LMIC countries.

The low number of articles on the knowledge, attitudes and practices of genetics/genetic testing found in this review may be indicative of the limited implementation of genetic services in LMIC. High income countries have been the forerunners in introducing and offering genetic testing for diagnosis and prenatal diagnosis, with research into genetics knowledge of pHCP dating back to 1989 in these countries (16). In comparison, many LMIC lack comprehensive genetic services, reflecting the ongoing epidemiological transition in these countries – a process that was completed in most HIC countries decades ago (17).

Publications overview

**Geography**

South East Asia Region

Geographically, the highest number of articles meeting the inclusion criteria were identified in SEAR (n=8), emanating from only four of the 11 countries. This may be accounted for, in part, by a group of HCPs with an interest in reproductive and genetic technologies, including TOP, in Sri Lanka, who collectively published three articles over four years (2002-2005) (18-20). The first IVF clinics emerged in the country in 1999, but without accompanying laws or guidelines governing their regulation (18). The authors of these three timeous publications were interested in the effects of these new technologies on reproductive options, especially since Sri Lanka is a multi-cultural and multi-religion country, with various worldviews and ethical concerns. This was followed up by an article by de Silva, Jayawardana (21) who investigated TOP for four genetic conditions in Sri Lanka.

India was another SEAR country with articles published; one on late TOP for fetal anomalies (22) and the other on Huntington's disease (23). There was then an 8-year gap until Alfaqih, Khader (24) investigated attitudes towards genetics and biochemistry in Jordan. The most recent publication from this region was from Indonesia on attitudes towards genome editing (25). The lack of publications from other countries in the region may be due to a high rate of consanguinity and the general lack of physicians and inadequate genetic education of physicians, who are not trained or empowered to identify and diagnose genetic conditions (26). It may also be indicative of the early stages of epidemiological transition experienced by the countries during this time period, when the diagnosis and management of infectious diseases would have been a greater priority. For example, India was reported to have neither a social healthcare system nor a compulsory healthcare insurance system at the time. Although WHO has frequently emphasised the need for genetic services, with proposed guidelines in all member states (27) the continued lack of financial and human resources and supporting evidence base prevents implementation – particularly in LMIC.
The Region of the Americas

The AMR and EMR were the next most productive regions, with seven articles published in each. In AMR, the majority of articles (n=6) were published in Brazil, plus one in Mexico, which was also the earliest article included in this review in 1999 (28), focusing on predictive and prenatal diagnosis of Huntington’s disease. In that article, they reported that a genetics course was only considered mandatory for completing a medical degree since 1996 at the Medical School of the National Autonomous University of Mexico. They also suggested that specific genetic conditions be managed by genetics units and specialists with greater disease-specific knowledge. This provides context for the lack of genetics-related knowledge/services in the country. No subsequent articles were identified regarding knowledge, attitudes and practices of HCP in Mexico, published in English.

Brazil as an individual country had the highest number of published articles (n=6, 22%) in the review. The earliest article, in 2008, focused on a specific cardiac diagnosis of Brugada syndrome (29), however, three articles were published by different departments of a specific institution, the Federal University of São Paulo (30-32). These focused on HCP awareness of primary immunodeficiencies and competencies in prevention and diagnosis of birth defects. More recent, studies were published by different groups across Brazil (15, 33) with a focus upon the provision of genetic healthcare services. This may be linked to a co-ordinated health strategy in the country in response to the Zika outbreak in the region, which also benefited other conditions, including congenital disorders.

Eastern Mediterranean Region

Seven articles from four of the 16 LMIC countries in the EMR were included. These seven articles included Lebanon (34, 35), Pakistan (36, 37), Iran (38, 39) and the most recent from Syria (40). The topics covered were diverse; one article on attitudes towards termination of pregnancy (34), two on haematological disorders (36, 39), two on the provision of clinical and counselling genetic services (35, 37), and one on HCP’s knowledge of PIDs (38). The most recent article from this region focused on the newer developments of pharmacogenomics (40). Countries in the EMR have a higher rate of consanguinity in Arab communities, which has led to a higher frequency of congenital disorders (CD), particularly autosomal recessive conditions (41). Coupled with the views and laws around TOP, research into autosomal recessive and haematological disorders, TOP and the provision of genetic services, provides insight into healthcare services in this region.

Africa

While there are 47 countries in the AFR region, only four produced articles included in this review. Three of the four articles focused on pregnancy and SCD (42-44). SCD is the most common monogenic disorder (45) with a high birth prevalence in SSA(46). In countries such as Cameroon, where there is no universal medical insurance, non-communicable diseases (NCD) such as SCD represent an increasing health burden (43). The carrier rates for SCD are estimated to be around 1 in 4 people (42). Although national control programmes may be partially implemented, care for affected individuals is still lacking (42, 43).
Timings of Publications

During the study period (January 1990 to April 2022), only one article (28) preceded the publication of the human genome in 2000. In the following decade, nine studies were published and a further 18 from 2010 to April 2022. This increase in relevant publications across LMIC is encouraging, particularly individual publications from India, Iran, Lebanon and Malaysia (22, 23, 34, 35, 38, 39, 48), suggesting recognition of the need for clinical genetic services. Many African countries, where there is a high burden of SCD (42-44), have begun to assess the need for HCP and genetic services in primary healthcare, which is ideal for screening, antenatal care and early childhood treatment of genetic conditions.

Qualitative Framework

The thematic framework (Figure 6) developed from the articles was used to contextualise and evaluate the qualitative component of this review.

Lack of knowledge:

With over half of the articles (n=15) incorporating aspects of pHCP knowledge, most reported on the insufficient genetic knowledge of HCP. This lack of knowledge leads to barriers in services due to uncertainty and lack of confidence in treating and referring patients with genetic problems (49). The results of self-rating and/or assessment of knowledge scores implemented in these articles ranged from very knowledgeable, satisfactory and adequate/fair to insufficient (15, 24, 28-31, 37-39, 44). This reported deficit supports the call for continuous educational interventions for the improvement of pHCP’s knowledge (50).

Shortfalls in several areas of genetic knowledge were identified:

Lack of basic genetic knowledge

Both pHCP and nurses were reported as lacking specific genetics knowledge (i.e., as taught in medical school or nursing college). This included: not knowing that genetic conditions could be “prevented”; the availability/option of prenatal screening, and; the inheritance pattern of certain conditions, including how to collect information via a family history (28, 31, 32, 51). A systematic review by Talwar (52) focused on
furthering the education of non-genetic specialist HCP found that most studies and interventions on basic genetics knowledge were conducted in the USA, and others were similarly undertaken in Canada, Europe and HICs in Asia. A recent review by Chou, Duncan (53) highlighted strategies to facilitate delivery of genetic testing and services through primary healthcare, again with most studies from HIC only. One article focusing on LMIC was a needs assessment undertaken in Ethiopia by Quinonez, Yeshidinber (51), to evaluate HCP experiences with genetic services, and suggested tools to assist with data collection for epidemiological studies. This study highlighted how clinical genetic services may be supported in LMIC by the introduction of tools to assist pHCP.

Emerging technologies

Applying existing genetics knowledge in primary practice is already a challenge for many pHCP. Genetics and genomics are rapidly developing fields, and pHCP are at risk of being left behind in a genomics era that is becoming increasingly reliant on this knowledge. Staying abreast of newer developments in genetics includes the application of genome editing in newborns, WGS, whole genome association studies and epigenetics (24, 25, 30, 38). Keeping up to date with these and other emerging technologies and the scope and methodologies of available tests is challenging for pHCP, as they are infrequently exposed to genetic conditions in their practices/clinics.

Educational interventions to inform pHCP about new genetic developments will enhance their knowledge, and this may be incentivised by accrediting such interventions as continued professional development (CPD) events.

Referral to Genetic Services

The referral of patients in existing health systems in LMIC is challenging. HCP may be unaware of existing facilities and/or the travelling distances and time required by patients to access these services may be prohibitive. Antoun, Zgheib (35) reported that up to 15% of HCP were unaware of genetic services in their area of practice. It has also been shown that HCP are less likely to order genetic tests if they are unaware of the availability of genetic services (54). This lack of knowledge of services leads to a lack of appropriate care for patients and their families. In Brazil, Iriart, Nucci (15) reported a lack of referral from inland, rural Brazil to the capital cities and urban centres, where genetic services are offered. Transportation challenges and the cost of travel in a resource poor country is a major obstacle for patients and may result in a reluctance by HCP to refer. Decentralisation and expansion of genetic services may contribute to increased access to these services in LMIC.

Clinical Guidelines

Knowledge of and attitudes towards recommendations of the Center for Disease Control (CDC) and other international and national guidelines or a local protocol was found to be poor. Ferreira, Russo Akiba (31) reported that only half of pHCP prescribed folic acid for pregnant women, and only 10% of HCP reported knowledge of the international guidelines for testing for Huntington’s disease (28). This deficit highlights the need for education related to relevant guidelines and recommendations for the management and care
of CDs. Such recommendations and guidelines may also be country-specific, for example in Cameroon and central African countries for SCD, or the international guidelines for diagnostic and predictive testing for Huntington's disease.

Attitudes of Primary Health Care Practitioners

Towards genetic services

Of the 28 included articles, 19 (70%) included a component on attitudes in general, indicating a belief that genetic testing and counselling are important (35) - especially among general practitioners (GPs) and primary care physicians (36). A positive attitude was also noted towards genetic counsellors being incorporated into the healthcare system, with five articles discussing positivity towards obtaining a genetic diagnosis and genetic services in general (24, 28, 32, 36, 37). Supportive attitudes towards genetic counselling, prenatal diagnosis and selective termination of affected pregnancies were also highlighted (44), depending on the specific legislation related to TOP. Overall, positive attitudes were generally related to the potential offered by genetics and genetic testing for the future development of genetic services in LMIC.

In contrast, the findings of Wonkam and Hurst (43) emphasised that genetic services challenge the “power to cure” by HCP, which may create an overall negative attitude towards such services. This suggests that pHCP may feel helpless and uncomfortable caring for patients with incurable genetic conditions, thus impacting on their confidence. Education and psychosocial interventions for HCP are proposed to assist with the perceived emotional burden of caring for affected individuals. HCP should be made aware that parents see HCP's offering to care for the patient (and their family) may be better than no treatment at all. Other reasons for HCP not accepting genetic services include a mistaken belief that other preventable diseases (mainly infectious) should be treated first, the fear of discrimination, an increase in abortion rates and individual stress (36).

Towards patients with genetic conditions

Insecurity of pHCP related to prescribing medication for patients with genetic conditions was expressed (15). HCP face bureaucratic challenges in accessing appropriate care (medication and nutrition) for their patients (15). This may leave patients and families feeling further stigmatized and unable to access appropriate treatment, together with a reluctance by HCP to care for patients where they are not able to treat appropriately.

Towards new technologies

In general, the articles indicated that most pHCP are positive about the impact of new technologies on genetics and care. They understood the role of genetics in designing better targeted therapies and how the role of specific variants may determine disease susceptibility (24). A similar attitude was noted towards preimplantation genetic diagnosis (PGD). The use of new technologies to overcome infertility was viewed in a positive light (18). A positive attitude towards the implementation of non-invasive
pregnancy testing (NIPT) in China was also reported (47), as well as towards genome editing for the purposes of treating fatal or debilitating diseases, at both the embryonic and somatic levels in Indonesia (25).

Negative attitudes were expressed towards genome editing when it was applied to non-health related aspects such as enhancing human ability or performance i.e. eugenic (25). Alfaqih, Khader (24) reported on the diverse views around establishing a DNA database to enhance personalised medicine, and Albitar and Alchamat (40) reported that physicians were uncertain about requesting pharmacogenomics testing before prescribing medication. It is anticipated that as related knowledge increases and application of these genetics tests improves, the attitudes of HCP may change and improve as a result of the expanding evidence base.

**Barriers to Genetic Services:**

Barriers to developing and accessing genetic services globally have been reported for decades (49). More recently, key barriers to obtaining these services have been outlined as: 1) Lack of knowledge and skills (the focus of this article); 2) Challenges in national healthcare systems; 3) Ethical, legal and social issues (ELSI), and; 4) Lack of an evidence base (7). Additional barriers include poor skills in taking family histories, non-existent referral guidelines and other tools, and a lack of confidence in delivering genetic services (attitudes and skills), time constraints and cost of tests (32, 33, 35, 48).

Specific barriers to genetic services identified in this review include:

**Inadequate genetic services**

Although the availability of formal genetic services available remains undocumented in many LMIC, an overview of 34 LMIC by Kaur, Hadley (55), indicates that limited genetic services exist in these countries. While anecdotal evidence indicates a lack of clinical genetic services in many LMIC, there is little empiric evidence to support this, particularly in Africa. The Society for the Advancement of Sciences in Africa (SASA) (56) states that clinical genetic services are either non-existent or rudimentary across Africa, except for in South Africa which has a formal training programme to build capacity.

Other, specific challenges in providing genetic services highlighted by this review include:

**Capacity:**

Many LMIC lack the necessary personnel, technology, infrastructure and capabilities to be able to offer a comprehensive clinical genetics service. (43, 51). For example, in Pakistan there are no genetic counselling services offered by the healthcare system (57), preventing patients with single gene and other congenital disorders from receiving the required counselling to accompany a diagnosis (37, 47). Further service bottlenecks may also be due to a lack of adequate genetic specialists in the country, as reported by Ashfaq, Amanullah (37) in Pakistan.
Capacity issues are compounded by the overall scarcity of relevant clinical specialists, including clinical geneticists, and other relevant specialities. In some LMIC such as Ethiopia, there are no clinical geneticists or genetic counsellors (58). Encouragingly, Abacan, Alsubaie (59) reports on several LMIC that have genetic services provided by genetic counsellors. These include Cuba with 900 genetic counsellors equating to 82 per million, Romania (Europe) with 4 per million, Malaysia has 5 equating to 0.2 per million, the Philippines 0.1 per million (SEAR) and South Africa with 0.4 per million (AFR). A recent article from Brazil reported that 332 medical geneticists for approximately 30% of the 211 million population of (1.6 per million) (60). South Africa is the only country in AFR with both clinical geneticists and genetic counsellors. However, with only 14 medical geneticists (0.2 per million) and 20 genetic counsellors (0.4 per million), capacity remains far below the recommended 120 clinical geneticists (2 per million) (61) for 60 million people. This lack of capacity is emphasised by comparison with the UK, with a similar population size (60 million) where 5 genetic counsellors per million are available. This is a far cry from HIC such as the USA with 12, Canada with 9, and Australia/New Zealand with 7 genetic counsellors per million of the respective populations (59).

To maximise capacity, several alternative, innovative options have been proposed by Mikat-Stevens, Larson (7) to increase access to genetic services, including telehealth and virtual consults. The recent COVID19 global pandemic has made virtual consults a reality for many patients and HCP, including genetic services.

The collective challenges of limited resources, high costs, and a lack of demand for genetic testing (due to a continued focus on infectious diseases in many LMIC), results in many genetic tests being sent abroad. Recently, the focus is being shifted to building local expertise and testing capacity, but this remains an uphill struggle for LMIC when international companies offer equivalent testing at a lower price. In response to this need, the African Society of Human Genetics (AfSHG) and the Human Heredity & Health in Africa (H3Africa) aim to create and build genetic capacity by facilitating genetic research laboratory training at all levels to encourage genetic services and to contribute to patient care (www.afshg.org).

**Political Will**

For genetic services to be implemented, legislation must first be developed and implemented using appropriately allocated resources. Wonkam and Hurst (43) postulate that a high rate of termination of SCD-affected pregnancies is indicative of the failure of professional stakeholders to provide adequate care in Cameroon. In contrast, in Brazil, national policy has required the integration of genetic services into the primary healthcare (32). This political commitment to genetic services is demonstrated by the number of medical geneticists in the country, as indicated above.

**Financial constraints**

**Inadequate Funding**
Seven articles in this review highlighted the challenge of inadequate funding allocated for genetic testing in LMIC (30, 33-35, 37, 44, 47). Most of these countries cannot afford to implement comparable genetic services to those in HIC (51). In LMIC, even basic genetic services are seen as an unnecessary expenditure, and HCP do not consider CDs a priority (33), since the underlying genetic cause cannot be changed and these conditions are considered untreatable (20). Such attitudes, combined with a preference to fund more obvious and “treatable” infectious diseases, results in inadequate budget allocations for genetic services, including genetic testing.

Care for patients and individuals with rare genetic conditions is often costly, and treatment options, if available, are often limited and unaffordable (15). Most state healthcare services in LMIC cannot afford to provide treatment for patients with rare diseases (15, 20). In Cameroon and other African countries, families are expected to pay for hospital bills due to lack of universal health insurance coverage (43).

**Out of pocket expenses**

Due to a lack of state funding for genetic testing and related services, the financial burden falls upon the patient and their families. For patients with no private medical insurance, the patient or family is expected to pay for the services out of their own pocket. Ashfaq, Amanullah (37) reported that the cost of genetic testing in Pakistan is prohibitive, given the *per capita* income, unless a new cost model is developed. The example of carrier screening and prenatal screening/testing for specific conditions was highlighted in five countries: Ghana, Lebanon, Pakistan, Brazil and China (15, 35, 37, 44, 47). Pregnant women in Ghana may not be able to access testing for the sickle cell trait, if they are unable to pay for laboratory testing (44). HCP in Lebanon are hesitant to offer genetic counselling due to the cost (35) and in another study in China, 72.5% of HCP believed that women would not undergo NIPT if they had to pay (47). Such studies undertaken in LMIC highlight the additional financial burden of genetic testing placed upon the patient and family, who are rarely able to afford these costs, as well as transport and additional living expenses. These challenges, in part, contributes to the higher burden of congenital disorders and associated mortality and morbidity in LMIC (62).

**Religious and cultural beliefs**

This review revealed that the diverse cultural and belief systems in LMIC may affect access to specific genetic services, depending on the dominant religion in the country of residence (37). This was specifically highlighted in two topics:

1. Termination of pregnancy

TOP for foetal anomaly is a contentious topic. TOP is not permitted by some religions, and related laws and regulations vary within and between LMIC, limiting options for addressing affected pregnancies (19, 20, 34, 36, 48). de Silva, Jayawardana (21) reported that religious affiliation was the only variable that influenced decisions regarding aspects of TOP in their study. Even following amendments to relevant TOP laws and regulations in some countries, such as in Sri Lanka in 1995 (where TOP was strictly
prohibited, but is now allowed when the mother's life is in danger with supporting signatures from three doctors), TOP for CDs remains controversial, with religious leaders opposing revised legislation to make it permissible (18). The decreased uptake of this secondary prevention measure in both LMIC and HIC results in a higher rate of affected births, requiring lifelong care and considerable socioeconomic impacts.

Five articles in this review highlighted issues related to TOP for foetal abnormality and willingness or reluctance to terminate an affected fetus. Both HCP (21) and patient perspectives (i.e. acceptance of fate and God's will) were reported (37). The common denominator regarding decisions related to TOP for affected pregnancies by patients/families was identified as religious affiliation (21). The greater acceptance by HCP may be attributed to their awareness of the significant financial and social burden of caring for an affected child.

2. Emerging Technologies

Reservations towards the use of some specific genetic technologies e.g., PGD, gene therapy and gene editing, was noted amongst HCP. However, religious beliefs are not always a barrier to genetic services, as some doctors from all religious backgrounds are supportive of the newer genetic techniques to assist with reproductive difficulties (18). Beliefs will differ between countries, cultures, religions and individuals. As education of HCP and the public improves around these issues, there may be a greater willingness to consider the benefits of these technologies.

Limitations:

The inclusion criteria of this review included restriction to publications in English only. Many LMIC are multi-linguistic, with numerous languages and dialects within some countries, and relevant studies published in other languages may have been excluded. Additionally, the high-quality English required by many high-ranking, peer reviewed journals may have prevented the publication of some LMIC manuscripts. Publication bias may be an issue, as positive outcomes of studies performed in LMIC may have a higher chance of being published than those with negative findings (Begg, 1989; (63), or negative findings are not written up by the researchers. While this scoping review focused on non-genetic specialist HCP, some articles covered specialist and non-specialist audiences, and these were retained to so as not to omit important information.

The way forward:

Much can still be done for the care and prevention of CD, particularly in LMIC. Suggestions from this review include the incorporation of genetic services by pHCP into primary healthcare, including primary health clinics - with the potential for more accessible, efficient and less expensive services. In-service training and online educational programmes (via short courses and CPD -accredited events) would empower HCP with relevant genetics knowledge, skills and an incentive to remain updated on emerging technologies.
Ultimately, better educated pHCP should improve the identification and management of patients affected by congenital disorders and enable integration into routine clinical practice. Educational strategies for genomics must include basic genetic principles and inheritance, and skills development for clinical management. This will be largely dependent on governmental policies, and the individual willingness and commitment of pHCP to incorporate genetics and genomics into daily practice.

Studies in Africa on genetic knowledge, attitudes, skills and practices is an unresearched/unpublished field. Nevertheless, South Africa appears to be in the unique position on the continent to be able to offer genetic services in the form of medical genetic services and genetic testing. Therefore, HCP have an obligation to improve their genetic knowledge, and know how to initially manage, access, and refer families with genetic conditions to available genetic services.

This review highlights the need for further research on the knowledge, attitudes and practices related to genetics and genomics. Such increased awareness will contribute to an improvement of overall healthcare and improve lives of people and families living with congenital disorders and contribute to no one being left behind (27).

Conclusion

Twenty years after publication of the human genome there is still a lag in knowledge and skills of pHCP in LMIC. While many LMIC pHCP are willing, the continuing burden of infectious diseases also constrains them. We have shown from the limited research available in LMIC, there are several avenues that need to be addressed including identified barriers to learning and implementation of genetic services. Strategies to overcome these is necessary, including education at all levels, from undergraduate to experienced HCP, is necessary to continuously update knowledge, teach and improve skills and provide at least the minimum of care for all patients and families. If we do not address the lack of knowledge and skills, patients will not have access to healthcare that should be available and will be effectively left behind.

Abbreviations

<table>
<thead>
<tr>
<th>Abbreviation</th>
<th>Description</th>
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<tbody>
<tr>
<td>AFR</td>
<td>Africa Region</td>
</tr>
<tr>
<td>AfSHG</td>
<td>African Society of Human Genetics</td>
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<tr>
<td>AMR</td>
<td>Region of the Americas</td>
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<tr>
<td>CD</td>
<td>Congenital disorders</td>
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<td>CDC</td>
<td>Center for Disease Control</td>
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<tr>
<td>COVID-19</td>
<td>Infectious disease caused by the SARS-CoV-2 virus</td>
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<tr>
<td>ELSI</td>
<td>Ethical, legal and social issues</td>
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EMR  Eastern Mediterranean Region
GP   General practitioner
HIC  High income countries
LMIC Low- or middle- income countries
NGS  Next generation sequencing
NIPT Non-invasive prenatal testing
PGD  Pre-implantation genetic diagnosis
pHCP primary Healthcare Practitioner
SASA Society for the Advancement of Sciences in Africa
SEAR South East Asia Region
SSA  Sub-Saharan Africa
TOP  Termination of pregnancy
UKZN University of kwaZulu Natal
WES  Whole exome sequencing
WGS  Whole genome sequencing
WHO  World Health Organisation
WPR  Western Pacific Region

Declarations

• Ethics approval and consent to participate
  ○ Not applicable
• Consent for publication
  ○ Not applicable
• Availability of data and materials
  ○ All data generated or analysed during this study are included in this published article
• Competing interests
  ○ The authors declare that they have no competing interests
Funding

- Not applicable

Authors' contributions

- The article was prepared by SW. All three authors undertook the screening of articles in the scoping review. SW and HM completed the thematic framework. CA and HM provided feedback on drafts, and collaborated with SW on interpretation, and writing of the discussion and conclusion.

Acknowledgements

- Not applicable

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43. Wonkam A, Hurst S. A call for policy action in sub-Saharan Africa to rethink diagnostics for pregnancy affected by sickle cell disease: differential views of medical doctors, parents and adult


Figures

**Figure 1**

PRISMA Flow diagram of literature review and study selection process.
**Figure 2**

Proportion and number of articles published per WHO defined region

**Figure 3**

Article publication timeline
Figure 4

Types of genetic conditions/subjects surveyed in the articles
Figure 5

Topics assessed in the articles (n=28)
<table>
<thead>
<tr>
<th>Lack of Knowledge</th>
<th>Attitudes</th>
<th>Barriers to practices</th>
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<tbody>
<tr>
<td>• Basic genetic concepts &amp; conditions</td>
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<td>• Emerging technologies (eg. genome editing)</td>
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<td>• Referral to Clinical genetic services</td>
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**Figure 6**

The structure of the thematic framework identified in scoping review

**Supplementary Files**

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