



# Identifying the current status and future needs of clinical, educational, and laboratory genetics services in Pakistan: a web-based panel discussion

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## Abstract

While the prevalence of genetic disorders has been well documented in the Muslim-majority, low-socioeconomic country of Pakistan, the provision of medical genetic services remains limited and cost-prohibitive to the masses in the country. With the objective of identifying gaps in the provision of medical genetics services as perceived by the healthcare providers and the general public, the Pakistani Society of Medical Genetics and Genomics (PSMG) organized a needs assessment webinar on December 6, 2020, titled, “A Vibrant Discussion on the Current Status and Future Needs of Medical Genetic Services in Pakistan.” The objectives of the webinar were (1) to explore the current availability of medical genetics services, (2) to identify areas in clinical genetics delivery models needed to improve the state of medical genetics in the country, and (3) to garner the interest in such provisions from the expert and lay audience. The webinar consisted of a moderator-led, structured interview of an expert panel including the following topics: (1) postgraduate clinical genetics and genetic counseling training programs, (2) medical genetics clinics and formal genetic counseling services), (3) clinical genetic testing and (4) patient support and advocacy groups. The webinar was followed by a short, web-based survey completed by 35 of the 60 attendees. The results of this survey indicated overwhelming support for establishing formal genetic counseling educational opportunities (91.6%) and increasing the availability of genetic testing (100%). This report further summarizes the opinions and recommendations of the panelists and the audience survey results.

**Keywords** Pakistan · Karachi · Genetic counseling · Geneticists · Genetic testing · Genetics clinics · Pakistani Society of Medical Genetics and Genomics · PSMG

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## Introduction

While advances in genomic medicine and technology have significantly improved public health outcomes in many countries around the world, there continues to be a dearth of basic medical genetic services and professionals in Pakistan. Pakistan, a Muslim-majority country in South Asia with a population of over 220 million people (Population Fund UN 2022), has a well-documented incidence of genetic and hereditary disorders. Due to strong cultural, socio-economic, and religious reasons, the country has one of the highest rates of consanguinity in the world (~70%) thereby increasing the incidence of recessively inherited disorders, such as hemoglobinopathies, metabolic disorders, and other rare recessive conditions (Ahmed et al. 2022; Ashfaq et al. 2013; Hussain 1999). Additionally, while no official patient registries exist, the estimated carrier frequency for beta-thalassemia is approximately 5–7%, with 9 million carriers in the total population (Ahmed et al. 2022). There is also a relatively high incidence of infant mortality, congenital birth defects, intellectual disability, bilateral hearing loss, and breast cancers (Gustavson 2007; Farooq et al. 2011; Ashfaq et al. 2013).

The World Health Organization (WHO) recognizes the lack of genetic services in developing countries such as Pakistan and states that 70% of birth defects globally could be prevented or treated if clinical genetic services were strengthened in developing countries (World Health Organization & March of Dimes 2006). A multitude of factors play a role in the lack of clinical genetics services in Pakistan, including but not limited to (1) a lack of centralized, government-run prenatal and newborn screening programs for common conditions including hemoglobinopathies and metabolic disorders, (2) lack of genomics infrastructure, patient registries, and population-specific databases, and (3) lack of public awareness programs (Riaz et al. 2019). Additionally, even though a number of physicians in Karachi overwhelmingly supported the inclusion of genetic counseling as an allied workforce in the healthcare system of Pakistan 10 years ago (Ashfaq et al. 2013), no concrete steps have been taken by any major academic institution in the country to establish a postgraduate level clinical genetics or genetic counseling (GC) training program (Riaz et al. 2019).

In order to calculate the clinical workforce required to meet the demands of the country, we have used estimates from the Royal College of Physicians (RCP), United Kingdom (UK). RCP estimates that 3 geneticists and 6–12 genetic counselors are required per 1 million individuals (in the UK). Based on these figures, 660 geneticists and 1320–2460 genetic counselors would be required to meet the needs of the population of 220 million Pakistanis

(Royal College of Physicians 2013; Abacan et al. 2019). It would be an understatement to say that the current medical genetics workforce in the country is understaffed with only two formally trained geneticists practicing in Karachi with a population of 15 million, suggesting a ratio of 1 geneticist to 7.5 million people.

Furthermore, at the postgraduate level, the College of Physicians and Surgeons Pakistan (CPSP) has initiated a Molecular Genetics and Genomics group where educational activities are being facilitated through the members of the Association of Molecular Pathology Pakistan; however, no formal degree or postgraduate training in clinical genetics is offered (Riaz et al. 2019). The lack of certified GCs, trained geneticists, and limited access to genetic testing furthers the health disparities in the region and burdens the already overwhelmed healthcare infrastructure.

The scale of the challenge can also be gauged by looking at some of the recently published studies on the relatively higher rate of genetic disease burden reported in the Pakistani population. For instance, Akbar et al. (2022) stated that about 22% of patients in Karachi who met the National Comprehensive Cancer Network (NCCN) eligibility criteria for hereditary cancer germline testing were positive for a pathogenic variant in one of the high-risk genes as compared to 5–10% of the cases that have been documented in other parts of the world. Similarly, Cheema et al. (2020) reported that 61% of 349 index cases who had whole exome/genome testing were found to have a genetic diagnosis with diagnostic yield being higher among consanguineous couples as compared to the previously reported diagnostic yields from 25 to 58% in other populations (Dillon et al. 2018).

To overcome these enormous challenges, like-minded clinical genetics experts of Pakistani origin that are currently based in the United States of America (USA) formed a volunteer, charitable organization by the name PSMG in June 2020. The organization's mission is to enable the provision of affordable medical genetics services to the people of Pakistan and to create awareness of genetic disorders through education, community outreach, and research. One of the first tasks for the group was to perform a needs assessment to improve the provision of clinical genetic services in the country by soliciting the advice of key opinion leaders in a wide range of healthcare specialties and patient advocacy groups working in Pakistan. A needs assessment was necessary to garner the perspectives of Pakistani healthcare providers as current data on the views of medical professionals in Pakistan regarding the current status and needs for genetic services is lacking in wider literature.

The objective of this report is to present the findings of the structured interviews of expert panelists conducted during a webinar and a short survey of the audience that followed the webinar. The overarching aims of the webinar were to (1) explore the current availability of medical

genetics services, as perceived by the healthcare providers and patient advocacy groups, (2) identify areas in clinical genetics delivery models needed to improve the state of medical genetics, and (3) garner the level of interest in such provisions from the expert and lay audience.

## Materials and methods

The webinar was held on Zoom (a video conferencing platform) on December 6, 2020. The event was organized by the members of PSMG in collaboration with partners including Jinnah Sindh Medical University Alumni Association of North America (JSMUAANA), National Institute of Child Health (NICH), and Aga Khan University Hospital (AKUH). The webinar was chosen as the platform for this discussion for three main reasons (1) to bring together a wide range of experts from Pakistan and the USA, (2) to accommodate panelists from different time zones, and (3) to avoid in-public gathering during the COVID-19 pandemic.

### Selection of the expert panelists

Experts from the USA and Pakistan in a wide range of healthcare specialties and patient advocacy groups were invited to participate in a group interview during the webinar. The panelists were selected based on convenience sampling. The medical experts were colleagues and collaborators of the webinar organizers and the patient advocate was approached via social media (Facebook). This helped with getting a more balanced perspective of the status of clinical genetics in the USA as compared to what is available in Pakistan.

The panelists from Karachi included (1) a medical geneticist, (2) a pediatric endocrinologist, (3) an obstetrician and gynecologist, (4) a molecular laboratory director, and (5) a representative from a patient advocacy group. The panelists from the USA included (1) a medical geneticist, (2) a molecular laboratory specialist, and (3) a genetic counselor. All of the panelists and the moderator are of Pakistani origin.

### Development of interview questions for the discussion

The “group interview” methodology was used to ascertain the opinions of leaders and professionals in the healthcare and patient advocacy fields. A list of questions and a consent agreement for recording and reporting were emailed to the eight panelists one week before the webinar. The interview questions were largely divided into (1) the current status of clinical genetics services in the panelists’ fields of expertise and (2) their opinions on what was needed to improve upon the current conditions. Given the diversity of the panelists

with respect to their fields of expertise, we did not identify any pre-existing tools in the literature that could be used for developing the interview questions. The questions were open-ended and based on inductive reasoning.

### Current status

For the healthcare providers, the questions were developed with their fields of expertise in mind. For instance, the US-based medical geneticist and the genetic counselor were asked about their experiences of coming from Pakistan to study medical genetics. This was intended for two main reasons (1) to showcase the “brain-drain” of prospective providers who left the country as a direct result of the lack of formal medical genetics education in Pakistan, and (2) to showcase the mission of the PSMG and to garner interest in membership into the society. Furthermore, the Pakistan-based healthcare providers were asked about the current status of the provision of medical genetics services in their respective clinics and hospitals. This included the comparison of the public and private sectors, and the variations among obstetrics and gynecology, pediatric endocrinology, and medical genetics clinics. Similar questions regarding the current status were posted to the patient advocate.

### Future needs

During the second part of the webinar, need-related questions were posted to the panelists to elaborate on the education and services needed to improve upon the current state of medical genetics in the country.

### Analysis of the discussion

English was used as the primary language of communication. The entire session was recorded and transcribed. The transcription was reviewed by two authors and using thematic content analysis, common themes were identified and summarized. While the questions were predetermined and designed to assess the specific needs of the study, none of the themes that emerged from the discussion were coded or pre-determined. Coder 1 (SAA) identified the themes from the transcriptions and coder 2 (MA) reviewed and provided feedback regarding the themes. Only minor disagreements about the themes between the two coders emerged, which were resolved with further discussion.

### Advertisement of the event and audience gathered

The webinar was advertised on professional (LinkedIn) and general public (Facebook, WhatsApp and Instagram) social media platforms. This was intended to garner a wide range of audience of both lay and experts.

## Audience survey design and analysis

Given that the webinar was advertised to a broad audience including medical students, healthcare professionals and the general public, the audience survey questions were based on inductive reasoning to capture the interest of the audience in (1) higher education for genetic counseling and medical geneticists, (2) provisions of medical genetic services via specialty genetics clinics, and (3) establishment of local clinical genetics laboratories in the country. Additionally, two questions were designed to identify the education level of the audience and their interest in membership in the PSMG. The link to the 5-question Google Form survey was disseminated among the attendees, and responses were tabulated and analyzed using descriptive statistics.

## Results

Of the eight panelists, three were from the USA, including two geneticists and one GC and five panelists were from Pakistan, including a US-trained geneticist, an Obstetrician/Gynecologist who is a maternal–fetal medicine specialist, a pediatric endocrinologist, a genetic testing laboratory director, and a patient advocate. The moderator was a US-based GC. All of the panelists and the moderator are of Pakistani origin. The Pakistan-based participants were from Karachi which is the largest and most populous city in Pakistan.

Four main areas of discussion were identified (1) clinical genetics training programs for physicians and GCs, (2) designated genetics clinics with formal genetic counseling services, (3) genetic testing laboratory services, and (4) advocacy/support groups for affected patients and families.

## Postgraduate clinical genetics and genetic counseling training programs

Medical genetics education is at a very rudimentary stage in Pakistan. There are no post-graduate degrees offered in either clinical genetics or genetic counseling. Medical colleges/universities do not focus on genetics in their curriculum and medical students have minimal exposure to medical genetics. Medical genetics is not recognized as a subspecialty in Pakistan and there are no residency training programs due to a lack of trained academic faculty. The absence of any form of post-graduate training in medical genetics has led to a complete void of this essential and much-needed medical specialty.

A pediatric endocrinologist at a public pediatrics hospital, had this to add on the unfortunate state of medical genetics training,

“... what is available [for genetic testing and counseling] is a very sad thing because this is something of a completely naive area, and there are hardly any trained people around, so I mean, of course, talking about this kind of a facility being available in the public sector is out of question... pediatricians and obstetricians have traditionally been filling in for this role of explaining to their patients about the kind of disease that they have and offer some kind of a formal/informal genetic counseling to the parents for the disease and the future outcomes.”

There was a consensus among the panelists regarding the dire need for the provision of comprehensive genetic services in Pakistan as this is no longer a “luxury”, but rather a “necessity”. For this to occur, a post-graduate training program for medical graduates and genetic counseling students would need to be established. The panelists agreed that at this time, compared to training medical geneticists, it would be more practical and feasible to train GCs. They discussed ideas on the best way to establish genetic counseling training programs in Pakistan, such as, genetic counseling certification programs in sub-specialties, such as prenatal or cancer. Additionally, the idea that individuals interested in genetic counseling can be trained on-the-job by medical geneticists currently practicing in Pakistan was discussed. A similar approach has been employed by the Pakistan-based geneticist at AKUH and by the US-based GCs and geneticist at NICH. At AKUH, the medical geneticist (one of the panelists) has trained an individual with a Masters in Genomic Medicine from the UK as a GC by creating a course that provided clinical exposure and training over a two-year period. Additionally, the US-based medical geneticist and GC are remotely training a PhD candidate in Genetics at NICH through virtual classes and observations of telemedicine genetics clinics. While these are excellent initiatives and the training of these two local GCs is most valuable, the group agreed that it would be ideal to establish a Master’s level genetic counseling program that could graduate multiple students with the eventual goal of creating a strong local genetic counseling workforce. Panelists from NICH and AKUH stated that their institutions will be interested in establishing a certificate or diploma program and a Master’s program in genetic counseling in collaboration with PSMG and local university/hospital systems.

## Medical genetics clinic and formal genetic counseling services

By the time of writing this manuscript, there is only one formal medical genetics clinic in the private sector at AKUH in Karachi. This clinic is staffed by two foreign-trained medical geneticists and a locally trained GC. Additionally, in the province of Punjab, a thalassemia screening program is established at the government level and basic recurrence risk

counseling is provided to parents of affected children. More recently, U.S.-based medical geneticists and GCs established pediatric and prenatal telemedicine clinics at the NICH and South City Hospital, respectively. According to the panelists from these two facilities, these telemedicine clinics have been well received by the patients and have assisted in training of the local staff.

The panelists discussed that as medical genetics is not extensively covered in medical school curriculum in Pakistan and physicians often feel stress and discomfort when confronted by genetic indications. For instance, many OB/GYN doctors feel ill-equipped to counsel patients about potential genetic diagnoses secondary to ultrasound abnormalities, multiple miscarriages, and neonatal deaths and would prefer to refer these patients to genetic professionals if available. Similarly, while pediatricians can provide information and counseling on more common genetic conditions, such as Down syndrome and cystic fibrosis, they would feel more comfortable in referring undiagnosed patients and those with rare genetic syndromes to trained genetics professionals.

A maternal fetal specialist, had this to say about the urgent need of genetic counselors in the country,

“the important thing is that in Pakistan there is a high rate of consanguinity and ... there is a high prevalence of genetic conditions that [is why it is important that] we have genetic counselors in place because it ... makes our lives a little bit easy because we have somebody who can work out, have a detailed history, give the risks and then decide then give them the options to the parents if there is a prenatal testing available.”

Also, there is a lack of general awareness of genetic disorders within the physician community and they are not inclined to order genetic testing because of discomfort with the ordering process, interpretation of the test results and subsequent management and counseling of the patients. At this time, limited genetic counseling is provided by non-genetic professionals within their specific specialties.

Moreover, there is a significant disparity between the patients who can afford access to private healthcare and those who seek medical care in the public healthcare sector. Since genetic testing remains expensive and genetic counseling services are almost nonexistent in the public sector, patients lack access to even the basic genetic evaluation. The situation is far worse in rural communities where patients must travel to large urban centers to get specialized medical care.

One of two medical geneticists in Pakistan, had this to say about the state of economic disparities with respect to genetic services in the country,

“So you know the issue with Pakistan in general is that we are a country of contrasts and huge disparities exist within our healthcare systems and there are

massive differences between what is available in the private sector and what you can offer patients and what is available in the public sector... [the endocrinologist in a public pediatrics hospital] is trying to do a lot in the government sector despite all the challenges, all the funding issues, and the fact that the patient population that-uh you know, him and his team are asked to see really cannot afford even very, very simple things which may seem affordable to others. Myself and my team work at the Aga Khan University, which is a private university, over there we do see a lot of unaffordable patients but a number of them, you know, can at least pay the consultation fee and then we can try to figure out, you know, creative ways to subsidize them [for genetic testing].”

Despite recognizing the need for genetics clinics and genetic counseling services, the panelists emphasized that a lack of GCs and medical geneticists in Pakistan is the root cause for the absence of such services. PSMG, on a short-term basis, continues to help establish additional telehealth genetics clinics and in the process provide training to local health experts. The Pakistani medical geneticist in particular, recommended the utility of telehealth when he said,

“... the mere act of traveling from up country down to Karachi, for example, can just exhaust family resources. So, we must try to provide telehealth to a number of genetics problems that are existing in remote parts of the country.”

### Clinical genetic testing

While a select few private institutions offer limited in-house genetic testing, affordable genetic testing is inaccessible to most of the Pakistani population. The Association of Physicians of Pakistani Descent in North America (APPNA) in association with the Child Aid Foundation and Rotary Club established the first public-sector genetics laboratory at NICH in the late 1990s. This laboratory provides basic services such as blood and tissue karyotyping at an affordable price. Some private laboratories offer thalassemia genetic testing only, both pre- and post-natal. AKUH provides blood and tissue karyotyping in addition to thalassemia, cystic fibrosis, and muscular dystrophy testing. They also provide constitutional microarray; however, uptake is very low due to both cost of the microarrays and also the limited awareness of its utility with general physicians. Advanced genetic testing such as noninvasive prenatal testing, prenatal-microarray, next-generation sequencing panels, and whole exome/genome sequencing are unavailable in Pakistan. Some private hospitals have contracted with European, North American, and Malaysian laboratories, but this is highly cost prohibitive for most patients. A small minority of the population

who is affording, travel internationally to Dubai for prenatal genetic testing while others travel to Europe and North America for postnatal genetic testing and treatment.

A molecular laboratory director at AKUH, echoed the sentiments about lack of awareness and financial difficulties as a barrier to genetic testing when she said,

“laboratory testing is quite limited, it’s both awareness and just healthcare access, this is all part of that paradigm of access, both to physicians who can provide the correct advice and, of course, the support, financial support for the testing.”

While the panel delineated the current status of limited genetic laboratory services in the country, they provided two major areas of improvement for immediate short-term impact on public health (1) development of newborn screening programs to help identify and treat metabolic conditions in neonates and (2) establishment of advanced genetic testing laboratories for variety of genetic and hereditary conditions. Both of these areas would require assistance and funding from the government of Pakistan which would then enable organizations such as the Association of Molecular Pathologists of Pakistan (AMPP) to provide training for laboratorians and the PSMG to provide training for genetic counselors.

### Patient support and advocacy groups

Patient support and advocacy groups are an integral part of genetic service models around the world. While there are a handful of patient support groups in the country, including Karachi Down Syndrome Program (KDSP) and Special Needs Pakistan (SNP), they are underfunded and largely underutilized by the larger medical community. One of the panelists, a parent of a child with Apert syndrome, is the founder of SNP. She described her journey from the time her daughter was born to her diagnosis as a period of “tremendous chaos.” Given the rarity of her daughter’s condition and lack of a trained geneticist at the hospital where she delivered her daughter, she was given a grim prognosis on her survival and no official diagnosis. Given her high socioeconomic status, she was eventually able to see one of the two geneticists in Karachi and get accurate information and anticipatory guidance about her

daughter's condition. Having gone through this difficult diagnostic odyssey with her daughter with no support, she made it a mission to help other families in similar situations. In 2013, she formed an online patient advocacy group that has more than 14,000 members. This is a platform that provides resources and psychosocial and educational support to its members, many of whom have undiagnosed genetic disorders. The discussion of stigma associated with having a genetic disorder and lack of culturally sensitive care was discussed along with other socio-cultural considerations including anecdotal incidences of infanticide and neglect of affected children.

### Survey responses from participants

The webinar recorded 60 attendees. Of these, 35 completed the attendee survey that was disseminated through the chat feature of Zoom, with a response rate of (58%). The majority (34%) of the respondents were students (bachelors, masters or medical), followed by medical doctors (28.5%), the remainder, having a masters or PhD degree. Typically, only MD/MBBS physicians have direct patient contact for healthcare needs while PhDs and master’s level individuals work in research, laboratory or educational settings.

When asked if formal genetic counseling and medical genetics training should be made available in Pakistan, the attendees overwhelmingly agreed and 100% of the respondents believed that there is a need for additional laboratory services in Pakistan (Table 1).

In a free space for optional questions or comments some respondents commented on the need for genetics educations and laboratory services:

“There are huge needed expanded Genetic laboratories services”

“We need medical laboratories in Pakistan and also we have [need] the medical counselor in our hospital to identify the genetic diseases.”

“Graduates from Genetics seek such opportunities. I [would] just like to ask about the initiation of the proper certification program in Pakistan.”

**Table 1** Responses from attendees regarding views on the inclusion of genetic education and testing

Question	Response <i>n</i> (%)		
	Yes	Maybe	No
Should formal genetic counseling training be available in Pakistan?	32 (91.6)	3 (8.5)	0
Should formal medical genetics training be available in Pakistan?	30 (85.7)	5 (14.3)	0
Is there a need for expanded genetic laboratory services in Pakistan?	35 (100)	0	0
Are you interested in joining the Pakistani Society of Medical Genetics?	28 (80)	6 (17)	1 (3)



“It’s a great webinar, it would be great if medical genetics short courses or certificates will be introduced by universities.”

## Discussion

A group of healthcare professionals who recognized the need for improving the state of medical genetics in their country of origin established a formal, first-of-its-kind Pakistani Society of Medical Genetics and Genomics. Their first challenge was to connect with the local healthcare providers facing similar challenges in the provision of medical genetics in their respective institutions. Secondly, the group needed to identify the specific needs and challenges limiting the provision of life-saving medical genetic interventions to their patient population. Not finding much success in the current literature, the group organized a needs assessment webinar and invited key opinion leaders in the healthcare, laboratory, and patient advocacy fields to gain insights and develop a road map toward the provision of medical genetics in the country.

The discussion painted a dire picture of the current status of medical genetics services. Specifically, the panelists brought to light a complete lack of professional genetic counseling training programs, limited access to local and affordable molecular genetic testing, an absence of population-wide prenatal and newborn screening, and a dearth of patient support groups for affected individuals and their families. Despite this, the panelists proposed possible working models to increase genetics services in the country and demonstrated enthusiasm and drive to continue to work to achieve these goals.

Moreover, interest in the development of genetic counseling training and formal medical genetics training programs was expressed by the majority of the attendees through a survey. All of the survey respondents also expressed the need to expand local genetic laboratory services to make testing more affordable and readily available.

Similar sentiments regarding lack of laboratory molecular genetic testing were expressed in a recent letter to the editor of the *Journal of the College of Physicians (CPSP) and Surgeons Pakistan Journal* titled, “Initiation of Molecular Pathology FCPS in Pakistan Is Urgent!” (Aijaz and Jamal 2022). The authors cite that as the cost of genomic sequencing technology is reducing, the limiting factor to using molecular diagnostic techniques in Pakistan is a lack of technical expertise and importune the College to initiate a fellowship program in Molecular Pathology. Similarly, trained medical and clinical genetics professionals are also a limiting factor in the communication of accurate and tailored genetic information to the population.

Furthermore, the need for increasing medical genetics education in medical school was highlighted by Arshad et al.

(2022) who reported on the self-perceived genetics knowledge and clinical comfort of about 500 medical students from 25 different institutions in Pakistan. Approximately 74% of students expressed desire for additional genetics education during medical school to aid in their role as future physicians, specifically relating to genetic testing methodologies, prenatal and cancer genetics and treatment of genetic disorders. Moreover, Barakzai et al. (2022) assessed genomic literacy among 214 medical professionals in Pakistan and reported that while genomic literacy may be considered ‘satisfactory’ for ‘classical genetics’, there is a need for further training in molecular biology concepts in the medical community. Similar to the previous survey, about 71% of the participants demonstrated interest in additional genomics education stressing the need and desire for increased medical genetics education for healthcare professionals.

In addition to healthcare professionals, the general public has also expressed interest in genetic testing and subsequent diagnoses. Jafri et al. (2015) surveyed 400 Pakistani parents, 200 that had a child previously diagnosed with a genetic condition and 200 parents with unaffected children to assess their views on prenatal testing for 30 specific genetic conditions. Over 80% of the participants with an affected child and over 70% of participants with an unaffected child were interested in prenatal testing and identification of all or most of the conditions (Jafri et al. 2015).

While the interest in education and clinical service is evident, there are several challenges in the implementation of public health genomics in Pakistan. With a population of over 220 million, the manpower and infrastructure required to address the needs of the population poses a monumental challenge and would ideally require the involvement of large-scale government funded projects. Among others, the panelists touched on the lack of funding being a significant barrier to the creation of a large-scale clinical and laboratory model for the provision of robust clinical genetic services. Other prominent barriers to the lack of population-wide genetics screening and basic genetic laboratory and bioinformatics infrastructure are (1) a strained and limited public health budget, (2) absence of a public health system or network of services, (3) prioritization of non-communicable diseases and hospital emergency services, (4) majority of home births (80%), and (5) shortage of trained genetics professionals (Riaz et al. 2019).

While Riaz et al. (2019) outlined both short-term and long-term goals for public health genomics in Pakistan, the authors of this paper were unable to identify any significant large-scale government initiatives that have been taken to address genomic health in the country. One noticeable exception is the provincial Punjab Thalassemia Program run by the Government of Punjab which provides pre-marital carrier screening, genetic counseling and prenatal diagnosis for thalassemia in the province of Punjab (Government of

Punjab 2015). The Program provides genetic counseling by healthcare professionals trained in the genetics of inherited blood disorders. Such a government led program does not exist for other provinces and the Ministry of Health does not appear to have genomic health on its current docket as evident by the The National Health Vision of Pakistan 2016–2025. While focusing heavily on women and child health, the healthcare initiatives in this report put forth by the Ministry of National Health Services do not make any reference to genetic services or population-based newborn screening programs for the country as a whole (Ministry of National Health Services Regulations & Coordination Government of Pakistan 2016).

Although a national approach is lacking, local initiatives in private sector have manifested, such as the establishment of a hereditary breast cancer clinic involving a multidisciplinary team at AKUH in Karachi and a genetics clinic at the Children's Hospital of Lahore that implemented a genetics diagnostic program that provides genetic testing in collaboration with a genetic testing laboratory based out of Rostock, German (Cheema et al. 2020; Ehsan et al. 2022). Similar to these initiatives, the panelists discussed the possibility of the establishment of additional programs in which local and international collaborates work together to create new clinical and educational opportunities.

Additionally, grassroots patient advocacy and social support programs such as KDSP and SNP (among others) exist. These are privately funded, non-profit organizations launched by parents of children with genetic conditions working to provide resources and services to other parents of children with special needs.

While genetic services in Pakistan are limited, other Muslim-majority and neighboring countries, such as Saudi Arabia, Bangladesh and India, respectively, have taken major strides towards increasing genetics awareness, services, and education. Similar to Pakistan, Saudi Arabia has high rates of consanguinity and subsequently high rates of autosomal recessive conditions (Qari et al. 2013). In 2005, a diploma program in genetic counseling at King Faisal Specialist Hospital & Research Center in Riyadh was initiated with the help of a Canadian genetic counselor, and in 2015, genetic counseling was recognized as a Master's program by the Saudi Commission for Health Specialties. A similar approach can be considered in Pakistan where genetic counselors from other countries could be recruited to develop a hybrid synchronous and asynchronous genetic counseling training program with some of the basic science instruction provided by local instructors. India offers three parallel tracks for genetic counselors. A genetic counseling degree can be obtained through a 2-year master's program or a shorter 1-year diploma or certificate program (Abacan et al. 2019). A similar approach could be considered for Pakistan, where a shorter diploma program in a subspecialty could

be made available to increase the genetics expertise in certain areas, such as cancer, or prenatal. Hosen et al. (2021), proposes a framework for the implementation of genetic services for Bangladesh and other low to middle-income countries that relies on collaboration between local and international medical professionals, laboratorians, researchers and counselors. The proposed framework is similar to the current situation in Pakistan, where, in a more informal manner, various healthcare providers, researchers and laboratorians are working together in an attempt to provide clinical genetics services when possible.

Several goals were outlined by the panelists to increase the awareness and availability of genetic services in Pakistan in the areas of education, clinical outreach and advocacy. Anticipated first steps for each of the goals are described in Fig. 1.

## Strengths and limitations

Hosting the panel discussion in a webinar format compared to an in-person meeting made it logistically and economically feasible for experts from the USA and Pakistan to participate. However, a limitation of this format could be the possibility of a lack of engagement with the audience and also among the panelists. However, a reading of the transcription of the webinar makes it clear that consensus, as well as disagreements among the panelists, were registered and a robust, meaningful discussion was held. Ideas that could have come up organically during an in-person discussion may not have emerged during a webinar setting where panelists were asked to unmute their mics and speak only during their turn. While the panelists provided a comprehensive overview of the provisions of the medical genetic services in the entire country, one major limitation of the webinar could be a potential bias toward the city of Karachi, as all the Pakistan-based panelists were Karachi based. Further, the education level of the audience was relatively high compared to average Pakistani population, where literacy rates are approximately 60%. This gap could be considered a limitation of this study as this audience does not represent the population of the country. Lastly, the limitations of the short, web-based survey include the small sample size (i.e., 35 out of 60) and the potential to amplify the opinion of the educated population of the country who had access to the internet and could comprehend and communicate in the English language.

## Future research

Future investigators could consider surveying a wider audience that is more representative of the general population. One potential example could be to design paper-based



**Fig. 1** Goals and anticipated first steps to increase the awareness and availability of genetic services in Pakistan

Education	Awareness and Outreach	Clinical Genetic Services
<ul style="list-style-type: none"> <li>• <i>Establish local genetic counseling training programs</i></li> <li>• Recruit local and foreign experts to develop a culturally competent GC curriculum</li> <li>• Reach out to graduate and medical universities to identify interested institutes with infrastructure for hosting a GC training program</li> <li>• <i>Bolster medical genetics training provided in the medical school and to healthcare providers</i></li> <li>• Provide synchronous and asynchronous learning opportunities for practicing healthcare professionals</li> <li>• Identify and address the gaps in medical school genetics curriculum set forth by the Pakistan Medical Commission to meet international standards</li> <li>• Provide scholarships to recent medical school graduates to do fellowships in medical genetics</li> </ul>	<ul style="list-style-type: none"> <li>• <i>Increase awareness of genetic conditions through professional education and community outreach</i></li> <li>• Provide genetics educational seminars to the general public. For example, PSMG offers bi-monthly educational web-based seminars on a variety of genetics topics that are free and open to the general public</li> <li>• Create social media accounts to spread awareness to the general population in Urdu and other local languages</li> <li>• Collaborate with already established patient advocacy groups and assist with education and counseling aspects of support</li> </ul>	<ul style="list-style-type: none"> <li>• <i>Access to genetics clinics initially through development of telehealth genetics clinics with the help of local and foreign experts</i></li> <li>• Develop telehealth capabilities to offer GC to larger patient population and to help address concerns for privacy</li> <li>• Establish sub-specialty clinics in oncology, prenatal and pediatrics</li> <li>• <i>Affordable genetic testing and screening</i></li> <li>• Establish local genetic testing facilities</li> <li>• Work with international labs to establish relationships with local hospitals and clinics</li> <li>• <i>Work with the public health policy makers to establish population-specific databases and population-wide screenings</i></li> <li>• Establish infrastructure for (a) prenatal screening for at risk pregnancies and (b) population wide newborn screening for common genetic conditions</li> </ul>

needs-assessment surveys in Urdu and other regional languages. Another potential research question to explore is the attitudes of the patient population towards genetic testing and genetic counseling by conducting focus groups of affected and unaffected individuals who did not receive genetic testing and counseling. This could provide insights not only for culturally and socially appropriate genetic counseling techniques but also provide policymakers with valuable information on the utility of investing in genomic technologies at a population level. Lastly, educators could consider comparative studies for curriculum by comparing the genetic counseling courses already existing in genetic counseling training programs. In addition, a compilation of courses currently being taught at local universities in public health policy, sociology, psychology, and religious and cultural studies could help facilitate a

rich psychosocial didactic curriculum for a Pakistani genetic counseling training program.

## Conclusions

The PSMG organized webinar on Medical Genetics provided a platform where professionals with different affiliations from Pakistan and the USA were able to collaborate and brainstorm ways in which the medical genetics needs of the population could be met. The discussion highlighted the lack of clinical genetic services and education in Pakistan and identified the immediate need for implementation of these services through establishing genetic counseling training programs, formal genetic clinics and advanced laboratory

testing facilities. These views were endorsed by the attendees based on the post-session survey results. The webinar participants agreed to work towards these common goals through continued dialogue and collaboration.

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## Declarations

**Ethics approval** This article does not contain any studies with human or animal subjects performed by the any of the authors.

**Conflict of interest** Aisha Furqan is a full-time employee of and owns stock in Natera, Inc. Myla Ashfaq, Syed Ajaz Ahmed, Rabia Aziz-Rizvi, Zahra Hasan, Salman Kirmani, Shama Munim, Rizwan Naeem and Jamal Raza declare that they have no conflict of interest.

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