



# 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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## Purpose

To investigate the current state and future needs of medical genetic services in Pakistan through a discussion with key healthcare professionals in the U.S. and Pakistan.

## Background

Pakistan is a Muslim-majority country in South Asia with a population of over 220 million people. Due to strong cultural, socio-economic, and religious reasons, it has one of the highest rates of consanguinity in the world (~70%) thereby increasing the incidence of recessively inherited disorders, such as hemoglobinopathies<sup>1</sup>, metabolic disorders and other rare recessive conditions<sup>2</sup>. There is also a relatively high incidence of infant mortality<sup>2</sup>, congenital birth defects<sup>2</sup>, intellectual disability<sup>2</sup>, bilateral hearing loss, and breast cancers<sup>3</sup>. Despite the staggering rates of genetic disorders in the country, the provision of Medical genetics services is extremely limited and cost-prohibitive.

The Pakistani Society of Genetics and Genomics (PSMG)<sup>4</sup> was formed in June 2020 by a group of US-trained geneticists and GCs of Pakistani origin in collaboration with Pakistan-based physicians with the goal of increasing awareness and availability of medical genetics services in Pakistan through education, community outreach, and research. One of the initial initiatives by PSMG was to organize a web-based panel discussion titled “A Vibrant Discussion on the Current Status and Future Needs of Medical Genetic Services in Pakistan”<sup>4</sup> to assess the needs of the Pakistani community as perceived by Pakistani healthcare providers and patient advocates.

## Methodology

The webinar was held on Zoom and was organized by the members of PSMG in collaboration with partners including Jinnah Sindh Medical University Alumni Association of North America (JSMUANA), Agha Khan University (AKU), and National Institute of Child Health (NICH) on December 6, 2020. Consent for recording was obtained from the panelists. The event was promoted via social media platforms such as LinkedIn and Facebook.

The moderator of the event asked each of the panelists pre-planned questions regarding the current state of medical genetic services in Pakistan, specifically in Karachi, and the future needs for the country. English was used as the primary language of communication. The entire session was recorded and transcribed. The transcription was analyzed by two authors using thematic content analysis; common themes were identified and summarized.

The link to the 5-question Google Form survey was disseminated among the attendees, and responses were tabulated and analyzed using descriptive statistics.

## Results

The expert panel comprised of eight US- and Pakistan-based health care professionals. From the US: two medical geneticists and one GC. From Pakistan: a U.S.-trained medical geneticist, 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 four main areas of discussion were as follows:

### 1. Postgraduate clinical genetics and genetic counseling training programs

- No post-graduate degrees in clinical genetics or genetic counseling and no genetics residency training programs
- Compared to training medical geneticists, it’s more feasible to train GCs
- Panelists’ institutions showed interest in establishing a GC training program

### 2. Medical genetics clinic and formal genetic counseling services

- Two geneticists and a locally-trained GC staff one of the only medical genetics clinic in the private-sector at AKU in Karachi
- Thalassaemia screening program was established by the Punjab government and provides basic recurrence risk counseling
- General and specialty physicians often feel ill-prepared to order genetic testing and provide subsequent counseling
- US-based medical geneticists and GCs established pediatric and prenatal telemedicine clinics at the NICH and South City Hospital in Karachi, respectively
- A lack of GCs and medical geneticists in Pakistan is at the root of lack of such services

### 3. Clinical genetic testing

- Affordable genetic testing is inaccessible to most of the Pakistani population
- A public-sector laboratory that provides affordable blood and tissue karyotyping is needed
- While the uptake is low, the AKU provides blood and tissue karyotyping and chromosomal microarrays (CMA) in addition to thalassaemia, cystic fibrosis, and muscular dystrophy testing
- Some other private laboratories offer thalassaemia genetic testing only
- Since most advanced genetic testing is unavailable in Pakistan, some private hospitals have contracted with international laboratories that remain cost prohibitive
- Two major areas of improvement for immediate short-term impact on public health were discussed: (1) development of newborn screening programs and (2) establishment of advanced genetic testing laboratories

### 4. Patient support and advocacy groups

- There are a handful of patient support groups in the country including the Karachi Down Syndrome Program (KDSP) that are underfunded and largely underutilized.
- One of the panelists, a parent of a child with Apert syndrome, described her journey from the time her daughter was born to her diagnosis as a period of “tremendous chaos”. She went on to form a patient advocacy group called Special Needs Pakistan (SNP) that has more than 14,000 members. This is a platform that provides resources, 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 sociocultural considerations including anecdotal incidences of infanticide and neglect of affected children.

## Results cont.

### Survey Responses from Participants

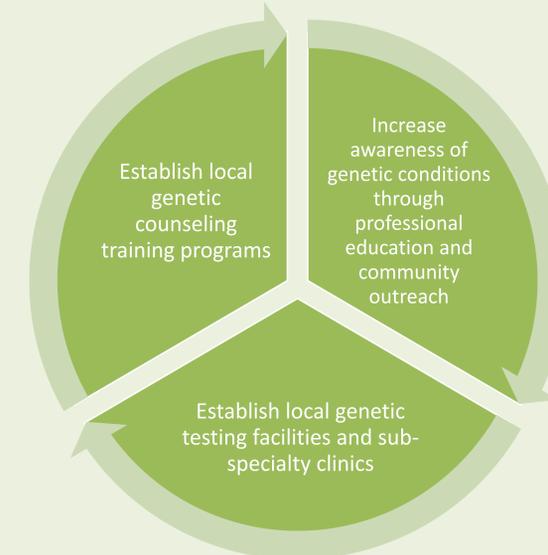
Thirty-five out of 60 attendees (58%) completed the survey. One-third (34%) of the respondents were students (bachelors, masters or medical), 28.5 % were medical doctors, the rest help a masters or PhD degrees.

Question	Response n (%)		
	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

## Discussion

- The PSMG webinar on Medical Genetics provided a platform where professionals from different institutions and countries collaborated towards a common goal of increasing medical genetics services and education in Pakistan.
- The discussion painted a dire picture of the current status of medical genetics services stressing the lack of:
  - 1) Medical genetics services and education
  - 2) Advanced next generation molecular genetics laboratories
  - 3) Population-wide screenings
  - 4) Appropriate patient support groups

## Conclusion and Future Directions



## References

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