



Establishing the Pakistani Society of Medical Genetics

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Last summer, two genetic counselors and two geneticists connected virtually with the goal of expanding genetic counseling services in their home country of Pakistan. Despite a population of ~220 million, with high rates of consanguinity and infant mortality, the entire country has only two trained medical geneticists and no genetic counselors. Hoping to increase awareness, Dr. S. Ajaz Ahmed and Dr. Rizwan Naeem connected with Dr. Jamal Raza, the Executive Director of the National Institute of Child Health (NICH) in Karachi. Dr. Raza detailed the abject state of genetic testing and the lack of trained specialists to provide counseling to his patients. Through various professional connections, the geneticists joined forces with two genetic counselors, Myla Ashfaq and me, to set up a telehealth genetics clinic at the NICH. This effort led to the formation of the Pakistani Society of Medical Genetics (PSMG).

A small conference room with an antiquated laptop and a capricious internet connection held the first NICH genetics clinic. In attendance were endocrinologist Dr. Mohsina Ibrahim, clinic coordinator Misbah Hanif, the patient, and her mother. Modestly clad, the 18-year-old proband, let's call her Amina, was short-statured and feeble. Dr. Ibrahim diagnosed Amina with Turner syndrome (TS) based on her clinical presentation and karyotype. The family's main concerns were whether Amina could marry and have children, and whether her diagnosis resulted from her mother's poor nutrition during pregnancy. These questions told the backstory that Amina's mother had been blamed for Amina's condition. As earnestly as I could, in my rusty Urdu, I began with, "Amina, with all our training in big American universities and all the years of practicing in big American hospitals, we can confidently say that your condition is not caused by the poor diet in either your mother during her pregnancy or in you as a child. To the best of our understanding, this condition is not inherited and happens by chance." There was an eerie silence in the room. Amina wiped the tears that were silently traveling down her cheek, while her mother embraced her. After, Misbah shared Amina's mother's sentiments: "For the 18 years of my daughter's life, I had felt that I was a bad mother for not feeding my child. Today, I will go home feeling heard by the doctors and my daughter will know that I am a good mother."

Instances like this are the reason most of us chose the genetic counseling profession. Increasing access to services for an underserved population re-affirms that what we do matters and that there is an urgent need for the globalization of genetic counseling services.

Information about PSMG

In just nine months, PSMG has started weekly telehealth genetics clinics staffed by Ambreen Khan, CGC, and Aisha Furqan, CGC in two hospitals, organized a monthly online lecture series on medical genetics and genetic counseling, collaborated with local support groups, and established connections with US- and Europe-based genetic laboratories. Under the PSMG mission statement "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", we continue to seek assistance from the genetic counseling community in establishing a robust genetic counseling training program in Pakistan to help patients like Amina and her mother. Please visit our website at www.PSMG.org for more details. To help in this massive endeavor email GCinPakistan@gmail.com.



AISHA FURQAN, CGC is a board-certified genetic counselor with oncology genetics expertise in clinical and laboratory settings. She received a graduate degree in Genetics and Data-mining from Cal Poly, Pomona in 2011 and a graduate degree in Genetic Counseling from Stanislaus State University/UCSF in 2015. She is passionate about the availability of genetic counseling services in Pakistan. Aisha was born and raised in Karachi. She enjoys hiking the mountains in central California where she now lives.