

INTERNATIONAL GENETIC COUNSELING SPOTLIGHTJUNE 17, 2021 JULY 6, 2021 PAKISTAN

Genetics Counseling in a Pakistani Patient Population – by Aisha Furqan, MS, CGC

Briefly about my personal background, I was born and raised in Karachi, Pakistan, and migrated to Northern California at the age of 16 with my parents and two sisters. While I did not understand the concepts of heredity, I was acquainted with hereditary disorders at a young age as my paternal grandmother and all seven of her siblings had severe adult-onset hearing loss and two of my father's male paternal first cousins had an undiagnosed form of muscular dystrophy (likely Becker's muscular dystrophy). More recently, two of my maternal first cousins got married in Pakistan and despite our best efforts, were not able to find pre-marital genetic counseling services in Karachi. Since graduation from the genetic counseling program at California State University, Stanislaus in 2015, I had hoped to start a genetic counseling training program in Pakistan and to create a platform for the exchange of ideas and knowledge about clinical genetics and psychosocial issues between the two countries I call home.

With eagerness to pursue this long-term goal and serendipitously finding the support of like-minded genetics experts in the US, I founded the Pakistani Society of Medical Genetics in the Fall of 2020. With the help of the co-founders Dr. Rizwan Naeem, Dr. Ajaz Ahmed and Texas-based genetic counselor, Myla Ashfaq, the PSMG has established two weekly telehealth genetics clinics in Karachi (one in pediatrics and another in prenatal) and has successfully organized a monthly online lecture series on genetic counseling-related topics. For more information, please visit <http://www.PSMG.org> (<http://www.PSMG.org>).

Herein, I provide two noteworthy case vignettes to shed light on some of the psychosocial issues that are commonly seen in the Pakistani patient population.

During a clinic at NICH, I co-counseled a family with two sons affected with X-linked congenital adrenal hypoplasia (CAH) with Utah-based genetic counselor, Ambreen Khan. While the family was proactive in completing genetic testing for their affected sons, they were hesitant to pursue genetic testing for their daughter even though the testing for daughters was covered under Invitae's complimentary family variant testing program. The hesitance for testing in the daughters resulted from concerns that finding this information may jeopardize marital prospects for them in the future. To add to this was the mother's guilt for 'causing' her sons' condition. We were reassuring that the mother would have no way of knowing that she was a carrier for this condition and the fact that both parents are so involved and dedicated to their sons' health is a testament to their resilience and strength. As a result of culturally sensitive, non-directive genetic counseling, the family felt a sense of relief that was evident in their demeanor over the video and their expression of gratitude at the end of the session.

At the prenatal clinic, I have seen expectant parents with previously affected children with diagnosed and undiagnosed hereditary disorders; six out of seven of whom are in consanguineous marriages. Of note is the latest case of a 30-year-old G2P1 female seen at 13-week gestation age for a previous child with homozygous pathogenic variants in the HADHA gene that is associated with Mitochondrial Trifunctional Protein Deficiency.

Despite numerous logistical and technical challenges, with assistance from the US-based genetic counselor, the team at the South City Hospital managed to get a fetal diagnosis for the parents to make an informed choice by 18-week gestation age (i.e., within the time for a therapeutic abortion). While perusing a prenatal diagnosis of a known familial variant, the question of future relationships between the consanguineous couples is often a tricky one. This couple in particular was determined to stay together and try to complete their family regardless of their knowledge and understanding of the reproductive risks. Most of the couples seen at the prenatal clinic showed similar desires likely due to cultural factors including the strength of relationships between the two families and also perhaps due to the stigmatization of divorces.

In detailing the case vignettes above, I hope to raise awareness about some of the psychosocial aspects of genetic counseling in a developing, predominately consanguineous country like Pakistan. With the help of our US and internally based genetic counselors, we hope to create a genetic counseling training program in Pakistan that can help patients like my grandmother, cousins, and many others.

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