Khushnooda Ramzan et al., J Mol Genet Med 2017, 11:4 (Suppl)
DOI: 10.4172/1747-0862-C1-021

Genetic characterization of Usher syndrome in Saudi Arabia population

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Hearing impairment is the most common disabling sensory defects in humans with both genetic and environmental etiologies. Severe to profound hearing loss affects 1 in 1,000 newborns, and another 1 in 2,000 children before adulthood. There are many sociological implications of hearing loss, comparatively poor educational achievements, especially for those losing their hearing early in life and problems of isolation and loneliness for those losing their hearing later in life. Usher syndrome (USH) is the most common cause of combined deafness and blindness inherited in an autosomal recessive mode. The identification of disease genes is an important and challenging problem. Proper diagnosis, prevention, as well as care for patients require an understanding of disease pathophysiology, which is best understood when the underlying causative gene(s) or genetic element(s) are identified. Due to high degree of consanguinity in the Saudi Arabian population, incidence of autosomal recessive diseases including hearing loss is higher than worldwide rate. Our study was aims to characterize the genetic basis of this disorder in Saudi Arabia. Consanguineous families are a powerful resource for genetic linkage studies/homozygosity mapping for recessively inherited hearing impairment. Prioritized linkage analysis and homozygosity mapping for was conducted. A next-generation sequencing-based multiplexing assay that encompasses the 120 known hearing loss genes was also used. For genes involved in Usher syndrome, we found mutation in MYO7A (42 families), CDH23 (5 families), PCDH15 (4 families), USH1G (1 family), USH1C (1 family) and USH2A (2 family). The benefit of this study is to reduce the incidence of deafness in Saudi Arabia by providing knowledge and awareness through screening of carrier status and genetic counseling, thereby decreasing socioeconomic burden. Through this study, we have established a detailed KSA specific clinical and molecular database that should improve and accelerate the diagnostic capacity for this disorder in our population.

Biography

Khushnooda Ramzan has completed her PhD in Molecular Biology from Punjab University, Pakistan during her PhD she mapped new loci for hearing loss by whole genome wide scan, which was a first step towards the later identification of a novel gene. Currently, she works as a Scientist in the Department of Genetics, King Faisal Specialist Hospital and Research Centre, Riyadh, Saudi Arabia. After her PhD, she has joined Department of Genetics, KFSH and RC, Riyadh as Postdoc and continued working on the genetics of hearing loss in Saudi families. This led her to an unusual finding that the most common gene GJB2 responsible of hearing loss worldwide does not play a significant role in their population. Her research focus was then focused to investigate the role of other genes and to identify the novel loci/genes within the Saudi population. She has characterized and documented genetic basis of autosomal recessive deafness in more than 200 families of Saudi Arabian origin; their incidence and distribution were also documented.

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