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Challenges and rewards of Usher syndrome genetics research in Saudi Arabia

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Hearing loss is one of the most common sensory disorders in humans with both genetic and environmental etiologies. Usher syndrome (USH) is the most common cause of combined deafness and blindness inherited in an autosomal recessive mode. Molecular diagnosis is of great significance in revealing the molecular pathogenesis and aiding the clinical diagnosis of this disease. However, the molecular diagnosis remains a challenge due to the phenotypic and genetic heterogeneity in USH. Our study aims to comprehensively delineate 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 were 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 a mutation in *MYO7A* (42 families), *CDH23* (5 families), *PCDH15* (4 families), *USH1G (*1 family), *USH1C* (1 family) and *USH2A* (2 families). The overall results of this study are highly suggestive that the underlying molecular basis of hearing loss in Saudi Arabia is very genetically heterogeneous. The benefit of this study will hopefully provide the foundation for knowledge and awareness through screening of carrier status and genetic counseling, thereby having a major impact upon early intervention for and prevention of Usher syndrome in our population.

Biography

Khushnooda Ramzan, PhD in molecular biology works as Scientist in the Department of Genetics, King Faisal Specialist Hospital and Research Centre, Riyadh. She got her PhD from the Punjab University; during which she discovered new genes for hearing loss. After her PhD she joined the department of genetics at KFSHRC, and continued working mainly on the genetics of deafness in the Saudi population. Her research focus was to investigate the role of different genes in hearing loss within the Saudi families. So far, she has characterized and documented genetic basis of deafness in more than 500 families of Saudi Arabian origin; their incidence and distribution were also documented..

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