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Characterization of the genes underlying Usher Syndrome in Saudi Arabian population

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Hearing loss is one of the most common sensory disorders in humans with both genetic and environmental etiologies. Genetic causes of hearing loss are extremely heterogeneous and the autosomal recessive inheritance accounts for a substantial part of this disorder. The Usher (USH) syndrome is characterized by congenital sensorineural hearing loss and retinitis pigmentosa (RP). It is the most common form of inherited deaf-blindness with a prevalence of ~ 1/6,000. Three clinical subtypes (USH1-USH3) are defined according to the severity of the hearing impairment, the presence or absence of vestibular dysfunction and the age of onset of RP. So far, ten causative genes have been identified underlying different subtypes of the USH syndrome. Consanguineous families are a powerful resource for genetic linkage studies/homozygosity mapping for recessively inherited disorders. Our study aims to comprehensively delineate the genetic basis and occurrence of USH gene mutations in clinically diagnosed Saudi patients. Whole genome SNP genotyping coupled with targeted sequencing of the candidate gene was utilized. Our results indicate that in the Saudi Arabian population, *MYO7A* is the most common gene for USH syndrome. Mutant alleles in other genes (*PCDH15*, *CDH23*, *USH1C*, *USH1G*, and *USH2A*) were also identified that provided insight in the molecular basis of USH syndrome in this population. The benefit of this study will hopefully provide the foundation for knowledge and awareness through screening of carrier status, genetic counseling; premarital screening, prenatal and PDG opportunities for multiple family members thereby have a major impact upon early intervention and prevention of hereditary hearing loss.

Biography

Dr. Khushnooda Ramzan, PhD in molecular biology works as scientist in the Department of Genetics, King Faisal Specialist Hospital and Research Centre, Riyadh, Saudi Arabia. She got her PhD 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 in the mapped families. Later, she joined department of genetics, KFSC&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 150 families of Saudi Arabian origin; their incidence and distribution were also documented.

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