

## HUMAN GENETICS &amp; GENETIC DISEASES

## MOLECULAR MEDICINE &amp; DIAGNOSTICS

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**Molecular genetics of the Usher syndrome in Saudi Arabia: Identification of known and novel mutations by homozygosity mapping and next generation sequencing****Khushnooda Ramzan, Mohammed Al-Owain, Selwa Alhazzaa and Faiqa Imtiaz**  
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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 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 overall results of this study are highly suggestive as the underlying molecular basis of hearing loss in Saudi Arabia is very genetically heterogeneous. The benefit of this study will hopefully provide 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.

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