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Mutational Screening in Congenital Deaf Families of Pakistani Population by Whole Exome Sequencing Analysis

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Abstract

Congenital deafness is one of the most common inherited disorder caused by various causes. It can be originated by acquired as well as genetic factors. Consanguinity is one of the most important factor in developing countries for highest prevalence of the disorder. Inherited deafness can be classified into syndromic and non-syndromic categories. Almost 60 genes are responsible for non-syndromic deafness. Three deaf families were selected on the basis of age onset (Prelingual), congenital type and parents with Consanguinity. In current study three deaf Prelingual affected Pakistani families were selected for whole exome sequencing analysis (WES) for identification of pathogenic variant. Capillary sequencing was used to confirm the mutant variants. In current study a Homozygous missense mutation was found in USH1C (MIM# 605242) c.307C>T (p.Arg103Cys), a homozygous missense mutation P.Ala595Thr c.1783G>A in MYO15A (MIM# 602666) and third was found in TPRN gene MIM# 613354).

Biography:

Sumaira Kanwal has completed he PhD from Kongju national University Korea and postdoctoral studies from Samsung Medical center. After getting her degree she is working as a faculty member in COMSATS University Islamabad, Sahiwal campus. (One of the leading University of Pakistan. She has published more than 25 papers in reputed journals and has been serving as an editorial board member of various journals. Her main field of interest is Neurology and neuromuscular disorders.

Speaker Publications:

1. Adzhubei IA, Schmidt S, Peshkin L, et al. (2010) “A method and server for predicting damaging missense mutations”. Nat Methods 7:248–249.
2. Ahmed ZM, Goodyear R, Riazuddin S, et al. (2006) “The tip-link antigen, a protein associated with the transduction complex of sensory hair cells, is protocadherin-15.” J Neurosci 26:7022–7034
3. Ahmed ZM, Riazuddin S, Aye S, et al. (2008) “Gene structure and mutant alleles of PCDH15: nonsyndromic deafness DFNB23 and type 1 Usher syndrome”. Hum Genet 124:215–223.
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