

Mutational Screening in prelingual deaf families of Pakistani Population

Deafness is the condition of decreased or absent ability to perceive auditory information. Environment plays great role in its variation and complexity. Sometimes accidents and head injuries or traumas aggravate the situation. Causes of HL are very diverse and long lasting. Congenital HL is occurred at the time of birth it is due to genetic reasons mainly. Other factors are mainly contributing in acquired HL. In Current study three families with familial history were selected for the mutational screening for causative mutation. Pakistan is one the developing countries where consanguineous marriages are encouraged which ultimately ends up in some clinical complications and inherited disorders. . In Pakistan, it is estimated that 80% marriages are first cousin marriages.40% pathogenic mutations and 130 loci are identified in Pakistan. Families were selected on the basis of history and congenital criteria. Later whole exome sequencing was done and causative pathogenic variants were identified. To confirm the causative variants automated sequencing was performed for the forward strand. Three missense mutations were found in MYO15A p.Ala595Thr, USH1C p.Arg103Cys and TPRN p.Pro419Leu. Polyphen score was calculated by online web server. These variations were not present in other healthy members of the families. Both alleles were inherited from the carrier patients.

I. Biography:

Sumaira Kanwal has completed his PhD at the age of 27 years from Kongju National University, South Korea and Postdoctoral She is working as Assistant professor at COMSATS University Islamabad, Sahiwal Campus since 2012., a prestigious University of Pakistan. She has published more than 25 papers in reputed journals and has been serving as an editorial board member of repute. She is motivational scientist who has the ability to drive the colleagues and students in a friendly way. Her basic aim of the research is to benefit the humanity from her knowledge and skills, which the basic essence of research.

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