World Academy of Science, Engineering and Technology
International Journal of Mathematical and Computational Sciences
Vol:14, No:12, 2020

In silico Analysis of a Causative Mutation in Cadherin-23 Gene Identified in an Omani Family with Hearing Loss

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Abstract : Hereditary hearing loss is a heterogeneous group of complex disorders with an overall incidence of one in every five hundred newborns presented as syndromic and non-syndromic forms. Cadherin-related 23 (CDH23) is one of the listed deafness causative genes. CDH23 is found to be expressed in the stereocilia of hair cells and the retina photoreceptor cells. Defective CDH23 has been associated mostly with prelingual severe-to-profound sensorineural hearing loss (SNHL) in either syndromic (USH1D) or non-syndromic SNHL (DFNB12). An Omani family diagnosed clinically with severe-profound sensorineural hearing loss was genetically analysed by whole exome sequencing technique. A novel homozygous missense variant, c.A7451C (p.D2484A), in exon 53 of CDH23 was detected. One hundred and thirty control samples were analysed where all were negative for the detected variant. The variant was analysed in silico for pathogenicity verification using several mutation prediction software. The variant proved to be a pathogenic mutation and is reported for the first time in Oman and worldwide. It is concluded that in silico mutation prediction analysis might be used as a useful molecular diagnostics tool benefiting both genetic counseling and mutation verification. The aspartic acid 2484 alanine missense substitution might be the main disease-causing mutation that damages CDH23 function and could be used as a genetic hearing loss marker for this particular Omani family.

Keywords : Cdh23, d2484a, in silico, Oman
Conference Title : ICSRD 2020 : International Conference on Scientific Research and Development
Conference Location : Chicago, United States
Conference Dates : December 12-13, 2020