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In silico analysis of USH1C gene causing deafness autosomal recessive 18 and Usher syndrome type 1C

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Abstract: Hearing loss is known as a most common sensorineural disorder affecting 1.86 in 1000 neonates. Genetic factors are involved in half of the all hearing loss cases. Hearing loss is classified to syndromic and non-syndromic types so that, the autosomal recessive non-syndromic hearing loss contributes nearly 80% of total [1]. Mutation of USH1C gene results to deafness autosomal recessive 18 and Usher syndrome type 1C [2]. At the present study, to investigate evolutionary relationship of USH1C gene in human compared to other organisms, ClustalX, GENEDOC and Tree view softwares and for study of gene network and various promoter elements of this gene, GeneMANIA and Gene2promoter sorfwares were used, respectively. With respect to phylogenetic tree drawed by analysis of USH1C nucleotide sequence, Homo sapiens has the highest and lowest similarity to Gorilla gorilla gorilla and Pteropus vampyrus, respectively. Additionally, study of USH1C gene network revealed its physical interaction with USH1G, USHBP1, CDH23, MYO7B, MYO7A, UBA1, WDR61 and SAAL1 which the mutation in each of them leads to non-syndromic hearing loss. Analysis of USH1C gene promoter showed the presence of 4 matrix families ETSF (specific for protein transporters and effective in T cell activation), RXRF (involved in the regulation of gene expression), SORY (affects developmental processes of organism) and ZFXY (involved in sex determination) (P<0.01) [3, 4]. In conclusion, obtained results emphasize complexity and functional importance of highly conserved USH1C gene.

Key words: Hearing loss; USH1C gene, Phylogenetic; Gene network; Promoter

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