Investigation of phylogenetic, protein network and cis regulatory elements of *TPRN*: A key gene causing deafness, autosomal recessive 79

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Abstract: Hearing loss (HL) is the most prevalent sensorineural defect affecting 1 in 500 neonates. Genetic factors are involved in half of the cases. HL is classified to syndromic and non-syndromic. Autosomal recessive non-syndromic HL contributes nearly 80% of total [1]. *TPRN* encodes a sensory epithelial protein. Mutations at this gene have been associated with autosomal recessive deafness 79 [2]. In this research, to explore evolutionary relationship of *TPRN* gene in human in comparison to other organisms, ClustalX, GENEDOC and Tree view softwares and to study of gene network and various cis elements of the promoter of this gene, GeneMANIA and Gene2promoter sorfwares were used, respectively. Regarding to phylogenetic tree acheived by nucleotide sequence analysis of *TPRN*, *Homo sapiens* has the highest similarity to *Gorilla gorilla gorilla* and *Pan Paniscus* and lowest similarity to *Cebus capusinus*. In addition, investigation of *TPRN* gene network showed its physical interaction with CLIC4, CLIC5, DNAAF5, PPP1CA, PPP1CC, and PPP1R18 proteins. Promoter analysis of *TPRN* gene clearly showed the presence of 3 matrix families ETSF (related to protein transporters and effective in T cell activation), RXRF (involved in the regulation of gene expression) and SORY (affects developmental processes of organism) ($P<0.01$) [3, 4]. Totally, obtained results showed the complexity and functional importance of highly conserved *TPRN* gene
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Key words: TPRN gene, Hearing loss, Phylogenetic, Gene network, Cis elements.

References:

[1] N. *Mahdieh*, B. Rabbani, S. Wiley, M. T. Akbari, S. Zeinali. Genetic causes of nonsyndromic hearing loss in Iran in comparison with other populations. J. Hum. Genet. (2010) 1-10.

[2] A. U. Rehman, et al. argeted capture and next-generation sequencing identifies C9orf75, encoding taperin, as the mutated gene in nonsyndromic deafness DFNB79. Am J Hum Genet. 86(3) (2010) 378-388.

[3] D. M. Mutch, P. Anderle, et al. Regional variations in ABC transporter expression along the mouse intestinal tract. Physiol. Genomics. 17 (2004) 11–20.

[4] https://www.genomatix.de/solutions/genomatix-software-suite.html