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Mutations in the GJB2 Gene Are the Cause of an Important Number of Non-Syndromic Deafness Cases

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Abstract : Deafness is the most common sensory disorder with the frequency of 1/1000 in many populations. Mutations in the GJB2 (CX26) gene at the DFNB1 locus on chromosome 13q12 are associated with congenital hearing loss. Approximately 80% of congenital hearing loss cases are recessively inherited and 15% dominantly inherited. Mutations of the GJB2 gene, encoding gap junction protein Connexin 26 (Cx26), are the most common cause of hereditary congenital hearing loss in many countries. This report presents two cases of different mutations from Iranian patients with bilateral hearing loss. DNA studies were performed for the GJB2 gene by PCR and sequencing methods. In one of them, direct sequencing of the gene showed a heterozygous T→C transition at nucleotide 604 resulting in a cysteine to arginine amino acid substitution at codon 202 (C202R) in the fourth extracellular domain (TM4) of the protein. The analyses indicate that the C202R mutation appeared de novo in the proband with a possible dominant effect (GenBank: KF 638275). In the other one, DNA sequencing revealed a compound heterozygous mutation (35delG, 363delC) in the Cx26 gene that is strongly associated with congenital non-syndromic hearing loss (NSHL). So screening the mutations for hearing loss individuals referring to genetics counseling centers before marriage and or pregnancy is recommended.

Keywords : CX26, deafness, GJB2, mutation
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