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Original Article

Molecular Pathology of 6 Novel GJB2 Allelic Variants Detected in Familial and Sporadic Iranian Non Syndromic Hearing Loss Cases

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Abstract:

Background: Mutations of GJB2 gene encoding connexion 26 are the most common cause of hearing loss in many populations. A very wide spectrum of GJB2 gene mutations associated with hearing loss have been detected but pathogenic role has been tested only for a part of them. In this study, we have provided genetic evidence on the pathogenicity of our previously reported novel GJB2 allelic variants.

Methods: The pathogenic role of GJB2 allelic variants were assessed using co segregation of each allelic variant with hearing loss in family members, absence of the allelic variants in control populations, coexistence with a second GJB2 mutation, nature of the amino acid substitution and evolutionary conservation of the appropriate amino acid.

Results: The GJB2 allelic variants including 363delC, 327delGGinsA, H16R and G200R have been co segregated with autosomal recessive non syndromic hearing loss in five families and are not found in control subjects. The G130V and K102Q were found in heterozygous state in two deaf individuals. G130V results in an exchange a residue highly conserved among all the connexins but was found with a rate of 1% in control subjects and K102Q results in an exchange a residue not conserved among all the connexins and not identified in control subjects.

Conclusion: We conclude that, 363delC, 327delGGinsA, H16R and G200R may be pathogenic. However, the pathogenicity and inheritance of K102Q and G130V can not be assessed clearly and remains to be identified.

Keywords:

Deafness , *Connexin 26* , *GJB2 gene* , *Iran*

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