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Autosomal Recessive and Sporadic Non Syndromic Hearing Loss and the Incidence of Cx26 Mutations in a Province of Iran

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Abstract:

Despite the enormous heterogeneity of genetic hearing loss, mutations in the GJB2 (connexin 26) gene located on " DFNB1" locus (13q12) account for up to 50% of cases of autosomal recessive non-syndromic hearing loss (ARNSHL) in some populations. This study describes the analysis of 100 autosomal recessive and sporadic nonsyndromic hearing loss individuals from 79 families each having at least one deaf child in Chehar Mahal va Bakhtiari province in west of Iran. We have investigated the prevalence of the connexin 26 gene mutations using nested PCR strategy to screen the predominant 35delG mutation and subsequent direct sequencing to detect other Cx26 mutations. Seven different genetic variants were detected from which one novel variant was including 363delC. The 35delG was the most common mutation found in 5 of 79 families (6.3%). Cx26 related deafness mutations (35delG, [V27I; E114G]) and R127H) were found in 12 of 158 chromosomes studied (7.8%). We conclude that the association of Cx26 mutations with deafness in Chehar Mahal va Bakhtiari province is low and looks like most other populations of Iran.

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