

Original Paper

# Informativeness of D7S2456 marker for molecular diagnosis of autosomal recessive non syndromic hearing loss in five Iranian ethnic groups

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## Abstract

**Background and Objective:** *SLC26A4* gene mutations after *GJB2* mutations are the second currently identifiable genetic cause of autosomal recessive non syndromic hearing loss (ARNSHL) which currently is used in molecular diagnosis of ARNSHL. Several potential STR markers related to this region have been reported. This study was carried out to identify the informativeness of D7S2456 CA repeat STR marker in *SLC26A4* gene region in five ethnic groups of the Iranian population.

**Methods:** In this descriptive study, The locus was genotyped in 165 unrelated healthy individuals of five different ethnics including Fars, Azari, Turkmen, Gilaki and Arabs ethnic groups using polymerase chain reaction (PCR) followed by polyacrylamide gel electrophoresis (PAGE) and fluorescent capillary electrophoresis. Data was analyzed by Gene Marker HID Human STR Identity software, Gene Pop program and Microsatellite Tools software.

**Results:** Analysis of the allelic frequency revealed the presence of 9 alleles for D7S2456 marker in the Iranian population, which allele 5 at the D7S2456 locus with 55% frequency was the most frequent. The most frequent heterozygosity with rate of 81.8% belongs to Azari ethnic group. Analysis of deviations from Hardy-Weinberg equilibrium demonstrated that all the ethnics except Fars were in equilibrium for D7S2456 locus. D7S2456 marker is a moderately informative marker in Iranian ethnic population (PIC value within 0.44 and 0.7).

**Conclusion:** D7S2456 is a moderately informative marker in diagnosis of *SLC26A4* based autosomal recessive non syndromic hearing loss in Iranian population by linkage analysis.

**Keywords:** Short Tandem Repeat, *SLC26A4* gene, D7S2456 marker, Autosomal recessive non syndromic hearing loss, Ethnicity, Iran

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