First report of mutation in COL11A2 gene in an Iranian family with Autosomal recessive non-syndromic hearing loss

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Abstract:

Introduction: Hereditary Hearing loss (HHL) affects one in 2000 newborns and more than 50% of these cases, the loss has a genetic basis. About 70% of HHL is non-syndromic with autosomal recessive mode of inheritance accounting for ~85% of the genetic load and more than hundred loci have been estimated for this kind of deafness.

Materials and Methods: A 4–year-old boy and her sixteen-year- old sister, both suffering from congenital deafness without any other clinical symptoms were referred to Genetics Research Center to be screened for molecular basis of their phenotype. Bases on clinical observation, autosomal recessive non-syndromic hearing loss was dignosed for both of them.

Using linkage analysis their phenotype localized to 6p21-3 chromosome in which COL11A2 has been located Several mutations have been reported in this gene associated with stickler syndrome type 3 and non-syndromic hearing loss, both with autosomal dominant form of inheritance.

Findings: In this study linkage between COL11A2 gene in DFNA13 locus, causative for autosomal dominant hearing loss, and phenotype of autosomal recessive non-syndromic hearing loss in and Iranian family was analyzed and Pro621Thr mutation was found in this family.

Conclusion: Pro 621 Thr mutation found in this study is the first report of COL11A2 mutation associatied with autosomal recessive non-syndromic hearing loss.

Keywords: Autosomal recessive /non-syndromic hearing loss /COL11A2 gene