A new mutation of FRMD7 gene in X-linked congenital nystagmus in an Iranian family

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ABSTRACT
Background & Aims: Congenital nystagmus is one of the most common eye diseases characterized by involuntary eye movements. A large family from West Azerbaijan province (mostly living in the city of Khoy) was referred to medical genetics department of Sarem hospital which congenital nystagmus has been detected in 12 of them with X-linked dominant inheritance pattern. Two X-linked genes on the short and long arms of the X chromosome had been reported that linkage analysis had been performed on them at the medical genetics department of Sarem hospital and no positive results were found. X-linked mutations in a third gene which is called FRMD7 have been reported recently that its related characteristics were same as what has been observed in the family members of this family. Therefore, we decided to investigate the FRMD7 gene in this family.

Materials & Methods: Methods of indirect (linkage) and direct sequencing (sequencing) were used to assess gene mutations of FRMD7.

Results: This study led to the identification of mutations c.37 C>T. The observed variation has not previously been reported in patients with congenital nystagmus. Mutation of cytosine to thymine base and deletion of glutamine amino acid, that results in premature truncation of the protein

Conclusion: The results of this study emphasize the heterogeneity of the disease. Therefore, study of this gene as a cause of congenital nystagmus in the Iranian society should be considered.

Keywords: nystagmus, FRMD7, X-linked gene, Sanger method, direct DNA sequencing

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