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A Deletion in Duchenne Muscular Dystrophy Gene Found Through Whole Exome Sequencing in Iran

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Abstract : Duchenne muscular dystrophy (DMD) is a severe progressive X-linked neuromuscular illness that affects movement through mutations in dystrophin gene. The mutation leads to insufficient, lack of or dysfunction of dystrophin. The cause of DMD was determined in an Iranian family. Exome sequencing was carried out along with a complete physical examination of the family. In silico methods were applied to find the alteration in the protein structure. The homozygous variant in DMD gene (NM-004006.2) was defined as c.2732-2733delTT (p.Phe911CysfsX8) in exon 21. In addition, phylogenetic conservation study of the human dystrophin protein sequence revealed that phenylalanine 911 is one of the evolutionarily conserved amino acids. In conclusion, our study indicated a new deletion in the DMD gene in the affected family. This deletion with an X-linked inheritance pattern is new in Iran. These findings could facilitate genetic counseling for this family and other patients in the future.

Keywords : duchenne muscular dystrophy, whole exome sequencing, iran, metabolic syndrome
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