

عنوان مقاله:

The identification of Mutations in exon1 of NKX2.6 gene in patients with Congenital Heart Defects

محل انتشار:

سومین کنگره بین المللی پزشکی شخصی ایران (سال: 1397)

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خلاصه مقاله:

Introduction: CHD results from perturbation of the normal program of cardiac development. it is the most common congenital anomaly in newborn babies. CHD is multifactorial, due to both genetic predisposition and environmental influences. NKX2-6 gene encodes a homeobox-containing protein that belongs to the NK-2 homeobox family. This gene plays an important role in the development of the cardiovascular system. Mutations in this gene are associated with CHDs, especially conotruncal defects such as persistent truncus arteriosus. **Method:** we analyzed the Nucleotide changes in exon 1 of NKX2.6 gene in patients suffering from congenital heart diseases in the province of YAZD. The PCR-SSCP and DNA sequencing methods used to detect the mutation of NKX2.6 gene in 110 pediatric patients with CHDs. We also used Polyphen-2 and Psipred databases and Pymol software to predict the effect of mutation detected on the structure and function of the NKX2.6 protein. **Results:** One heterozygous nucleotide replacement at c.109A> T position with amino acid change (S37C) were observed in one pediatric patient. Pymol results show that this mutation change the number and length of hydrogen bonds. Polyphen-2 result show that S37C amino acid change is benign and the results obtained from Psipred website illustrate that c.109A> T mutation causes a change in the secondary structure of the protein. **Conclusions:** Since this heterozygous mutation of the NKX2.6 gene have been observed in conserved region and not detected in any control subjects, and according to the results obtained from bioinformatics, it is expected to affect protein function

کلمات کلیدی:

NKX2.6, Pymol, Polyphen-2, Psipred, mutation

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