Evaluation of GATA4, NKX2.5, TBX5, CRELD1 and BMP4 genes with MLPA technique in patients with congenital heart disease

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Background and objective: Congenital heart diseases (CHD) are the most common birth defects. Nevertheless, the etiology of the majority of these illnesses remains unknown. Genetic and environmental factors are important in the etiology of CHD. Cardiac development is a complex and multifactorial biological process. Heterozygous mutations in some genes were defined in CHD.

Methods: In this study, we investigated the presence and frequency of mutations in the NKX2.5, GATA4, BMP4, TBX5 and CRELD1 genes on 255 patients with CHD. Mutational analysis was performed using multiplex ligation- dependent probe amplification -MLPA- technique.

Results: There was ventricular septal defect (VSD) in 144 patients, atrial septal defect (ASD) in 60 patients, complete atrioventricular septal defect (AVSD) 14 the patients and cyanotic congenital heart disease in 37 patients. We identified total 24 mutations in the 20 patients with CHD. We did not find any mutations NKX2.5 and CRELD1 genes in the patients with CHD. Mutations of the GATA4, BMP4 and TBX5 genes were detected in the five patients (VSD:3 and ASD:2), five patients (VSD:4, ASD:1) and a patient with VSD, respectively. There was MSR1 gene mutation in the six patients (Tetralogy of Fallot: 3, VSD: 3).

Conclusions: GATA4, BMP4, TBX5, MSR1 gene mutations could be important in congenital heart disease pathogenesis. However there was need the studies performed in larger numbers of patients used with CHD used the MLPA technique in this issue.