The Expression of NKX2.5 Gene in Congenital Heart Disease

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Background: Congenital heart diseases (CHD) are the most common heart developmental anomaly disease.Nevertheless, the aetiology of CHD in the majority of cases remains unknown. Abnormal cardiac development appears to occur through a process that is heterogeneous and complex, with both environmental and genetic risk factors. Heart formation is a complex process regulated by many transcription factors. Although most of CHD cases are sporadic, there are several genes which were found to be associated with CHD, including NKX2.5, TBX5, GATA4, etc.

Objective: To analyze the changes of NKX2.5 mRNA in human myocardium of congenital heart diseases (CHD) and to explore the relationship between NKX2.5 and congenital heart disease.

Method: The total mRNA were extracted from 10 cases with atrial septal defect (ASD) myocardium and 10 cases with ventricular septal defect (VSD) and 8 cases with non-CHD myocardium. 10 ASD cases are l2.71¡À8.12 (1.6~25) years old, 10 VSD cases are l2.44¡À4.98 (2¡«18) years old, 8 non-CHD cases are 22.92¡À12.33 (5-38) years old. The levels of NKX2.5 gene mRNA expression in the myocardium of CHD and the control group myocardium were detected using the RT-PCR (Reverse Transcription-Polymerase Chain Reaction) technique.

Results: The NKX2.5 gene was expressed in both the non-CHD and CHD myocardium. The levels of NKX2.5 gene mRNA expression were (73.72¡À6.21)£¥in the non-CHD myocardium, and (49.39¡À4.95)£¥in the ASD myocardium, and (29.69¡À3.48)£¥in the VSD myocardium. The levels of NKX2.5 gene mRNA expression in the ASD and VSD myocardium were significantly less than those in the non-CHD myocardium (t=9.26, 19.06, respectively; P <0.01)£®

Conclusion: The levels of NKX2.5 gene mRNA expression in the ASD and VSD myocardium were significantly less than those in the non-CHD myocardium. The results suggest that the abnormal expression of NKX2.5 may be involved in the pathogenesis of ASD and VSD.