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人类单纯性先天性心脏病中TBX5基因的突变及表达研究 Studies on the Mutation and Expression of TBX5 Gene in Human Simple Congenital Heart Disease

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摘要　应用PCR-DGGE方法在61个单纯性先天性心脏病核心家系共216位成员中检测TBX5基因8个外显子的突变情况；以β-actin作为内对照对单纯性先天性心脏病患者（房间隔缺损12例、室间隔缺损16例、法洛四联症6例）和3个非先天性心脏病患者（正常对照）心肌标本进行RT-PCR扩增和定量分析，观察TBX5基因在mRNA水平有无差异。研究了人类单纯性先天性心脏病患者中TBX5基因突变及表达情况。在所有被检成员中，未发现突变；与非先天性心脏病患者（正常对照）相比，单纯性先天性心脏病患者TBX5基因mRNA表达呈下降趋势。通过本研究认为TBX5基因编码区突变不是人类单纯性先天性心脏病的致病原因，但其转录水平的异常可能是单纯性先天性心脏病的一种潜在致病机制。

Abstract:This work is to investigate the mutation and expression of TBX5 gene in human simple congenital heart disease.The mutations of eight exons of TBX5 gene in 61 CHD family members(a total of 216 individuals including 65 patients and 151 normal relatives)were examined by PCR-DGGE.Using β-actin as internal control,the differential expression between 34 myocardium samples from simple congenital heart disease patients and three normal controls was conducted by RT-PCR.There is no mutation detected in all samples;The mRNA expression levels of TBX5 gene show descent tendency in samples of simple congenital heart disease compared with normal controls.The mutations in coding region of TBX5 gene do not cause human simple congenital heart disease,but the abnormality in transcription level of TBX5 gene maybe a kind of mechanism causing human simple congenital heart disease.

关键词　TBX5基因　心脏发育　突变　表达 Key words　TBX5 gene　cardiac development　mutation　expression

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