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实验研究报道

CITED2基因SGJ序列插入突变与先天性心脏病发病

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摘要

目的 分析先天性心脏病 （congenital heart defects, CHD） 患者的CITED2基因编码链基因突变的情况。 方法 收集101例散发型CHD患者和104例正常健康新生儿血液进行DNA抽提、PCR扩增，应用变性高效液相色谱仪进行CITED2基因全部编码序列的突变检测，对有异常峰型的DNA进行直接测序，并与GeneBank进行比较。 结果 首次在动脉导管未闭的患者发现CITED2基因的一种新的插入突变，在CITED2基因编码链碱基483位起始处插入一个重复9肽（c483_484ins27），导致蛋白的丝氨酸－甘氨酸富含区（SGJ）插入9个氨基酸p.Ser161-Gly162ins9。对照组中未检测到此突变。在CITED2基因的EP300结合基序未发现突变。 结论 中国先心病患者中存在CITED2基因突变，新发现的CITED2基因的重复9肽插入突变c483_484ins27可能是导致动脉导管未闭发生的原因之一。

关键词 先天性心脏病；CITED2基因； 突变

分类号

Insertion mutation in the serine-glycine rich junction of CITED2 as potential molecular cause for congenital heart disease

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Abstract

Objective To detect coding regions mutation of CITED2 gene in patients with congenital heart disease(CHD). Methods The blood cell genomic DNA of 101 patients with CHD and 104 normal neonatal was isolated. Whole coding regions were amplified by PCR. The PCR products were detected by DHPLC and samples with different melting profile shapes were sequenced, compared to GeneBank sequence databases. Results A novel insertion mutation was first identified in patients with patent ductus arteriosus (PDA) compared with 104 healthy controls. Mutation was located between 483C and 484G (c.483_484ins27), which resulted a 9-mer peptides repeat insertion in the serine-glycine rich junction (SGJ) of CITED2 amino acid sequence. Other patients and healthy individuals were normal. Conclusion There was mutation in CITED2 gene of Chinese CHD. The 9-mer peptides insertional mutation newly found may be one of the causes for PDA.

Key words

congenital heart disease CITED2 gene mutation

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