

中国人扩张型心肌病与受磷蛋白基因关系的研究 Association between Mutation of Phospholamban Gene and Dilated Cardiomyopathy

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摘要

受磷蛋白 (phospholamban) 是心肌收缩的一个重要调节因子, 可抑制心肌肌浆网钙ATPase的活性、降低其对钙的亲和力。正常情况下, 受磷蛋白可被不同的蛋白激酶磷酸化从而解除其肌浆网钙ATPase的抑制作用。国外两个DCM家系研究发现受磷蛋白基因突变与DCM的发生有关, 研究目的旨在探讨中国人群心肌特异性受磷蛋白基因突变与特发性扩张型心肌病发病的关系。收集60例确诊的特发性扩张型心肌病患者临床资料, 采集血样本并分离白细胞和提取基因组DNA。应用PCR扩增肌特异性受磷蛋白基因片段, 经测序检测基因突变的存在。结果显示60例扩张型心肌病人受磷蛋白基因开放阅读框未发现突变, 仅有两例患者出现阅读框3'部分非翻译区分别终止密码153和173位单个碱基T的缺失。两例患者终止密码后非翻译区单个碱基T的缺失据分析并无实际意义。因此我们认为绝大部分中国人扩张型心肌病的发病可能与受磷蛋白基因突变无关。Abstract: Phospholamban (PLB) is a prominent regulator of myocardial contractility and a reversible inhibitor of the cardiac sarcoplasmic reticulum Ca²⁺ ATPase activity. In normal cardiac muscles, phospholamban can be phosphorylated at distinct sites by various protein kinases and release its inhibition to sarcoplasmic reticulum Ca²⁺ ATPase. The studies of pedigrees have shown dilated cardiomyopathy (DCM) is related with mutation of PLB. The aim of present study is to investigate the relationship between mutation of PLB gene and DCM. Sixty patients with idiopathic DCM were enrolled in present study. The clinical data were collected, including clinical symptoms, ECG and echocardiography. Peripheral blood samples of all these subjects were collected to extract genome DNA. The fragments of PLB gene were amplified by PCR and PCR fragment sequencing was performed to study whether mutation of phospholamban gene exists. phospholamban gene did not show any mutation in these patients. Most Chinese DCM patients may not be related with mutation of PLB gene.

关键词 [扩张型心肌病](#) [受磷蛋白基因](#) [突变](#) Key words [dilated cardiomyopathy](#) [phospholamban gene mutation](#)

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