

高脂血症患者脂蛋白脂酶基因外显子4区域变异的研究

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摘要 为了探讨广东地区高脂血症患者脂蛋白脂酶(lipoprotein lipase, LPL)基因的分子变异, 从258例高脂血症患者外周血白细胞中提取基因组DNA, 用PCR-SSCP方法分析外显子4及其附近区域, 对SSCP带型异常样品进行克隆和序列测定。在2名高脂血症患者LPL基因内含子3的3'端-6bp处发现C→T转换突变, 252例正常对照中未发现该突变。IVS-3的C→T突变可能与高脂血症有关。

Variants of Exon 4 and Its Flanking Region of LPL Gene in Patients with Hyperlipidemia

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Abstract: To elucidate abnormalities of LPL gene in hyperlipidemia in the Chinese population in Guangdong, genomic DNA was extracted from leukocyte of 258 patients with primary hyperlipidemia. A segment of LPL gene including exon 4 and its flanking sequences was analyzed by PCR-SSCP. The PCR products with abnormal SSCP pattern were cloned and sequenced. As a C→T transition mutation at -6 bp of intron 3 was found in two Chinese with hyperlipidemia and the mutation was not found in 252 normolipidemic controls, the C→T transition in intron 3 may be related to hyperlipidemia.

Key words: lipoprotein lipase; hyperlipidemia; polymerase chain reaction; single strand conformation polymorphism

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