

PCR-RFLP检测LDL受体基因Taq I 多态性位点的研究 Detection of the Taq I Polymorphism at the Human LDL Receptor Gene in Chinese by PCR-RFLP Technique

周天鸿1, 李月琴1, 孙宏2, 朱志惠2 ZHOU Tian-hong1, LI Yue-qin1, SUN Hong2, ZHU Zhi-hui2
1.广东暨南大学生物工程系, 广州 510632 2.广东暨南大学华侨医院, 广州 510630 1.Department of Biotechnology, Jinan University, Guangzhou 510632 2.Affiliated Hospital of Medical Collage, Jinan University, Guangzhou 510630

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摘要 应用聚合酶链反应(PCR)扩增人类LDL受体基因外显子4-内含子4-外显子5片段, PCR产物为1.55kb, DNA片段经序列鉴定后, 进行TaqI酶切位点的RFLP分析。结果显示: 中国汉族人群LDL受体基因中存在着Taq I 酶切位点多态性; 200个LDL受体等位基因中Taq I 酶切位点出现的频率为0.515, 该点频率较为适中, 可作为中国汉族人群LDL受体基因的遗传标志来进行家族性高胆固醇血症(FH)的基因诊断。所建立起的LDL受体基因Taq I 位点的PCR-RFLP方法具有快速、简便的特点, 在FH的基因诊断上有应用价值。

Abstract: To develop rapid and sensitive technique for detectin the TaqI polymorphism at the human LDL receptor gene in Chinese, the exon4-intron4-exon5 of the human LDL receptor gene was amplified by polymerase chain reaction(PCR). The PCR products were directly analysed by restriction fragment length polymorphisms(RFLP). The results showed that the TaqI polymorphism is associated with the LDL receptor gene in Chinese of Han nationality; The frequency of T= allele (presence of TaqI cutting site) is 0.515 in 200 LDL receptor alleles. This technique may be used for rapid and sensitive screening of the LDL receptor gene for the TaqI polymorphism.

关键词 [低密度脂蛋白受体基因](#) [中国汉民族](#) [聚合酶链反应](#) [限制性片段长度多态性](#) **Key words** [Low-density lipoprotein\(LDL\)receptor gene](#) [Chinese Han nationality](#) [PCR](#) [RFLP](#)

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