

研究论文

## 中国人群脂蛋白脂肪酶基因突变与高脂血症的相关性研究

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### 摘要

为进行脂蛋白脂肪酶基因突变与中国人群高脂血症的相关性研究, 采用单链构象多态性分析结合DNA序列测定的方法, 对386例(其中108例高脂血症患者, 278例正常对照)中国人群进行突变筛查。结果发现1个新的沉默突变L103L, 1个错义突变P207L, 3个剪接突变Int3/3' -ass/C(-6)→T和普遍存在的S447X多态性, 其中发生在高脂血症组的P207L杂合子为亚洲首报, 并对先证者的家系进行了研究, 认为P207L是家族性高脂血症的病因之一, 而在正常对照组中也有发现的Int3/3' -ass/C(-6)→T, 对以往研究认为其是高脂血症易患因素的观点提出了相反的报告, 对于普遍认为有益的多态性位点S447X, 进一步研究认为其对于正常人群, 特别是健康男性的保护作用更强。结论: 脂蛋白脂肪酶基因变异与高脂血症的相关性十分复杂多样, 大规模的人群筛查具有重要意义。

关键词 [脂蛋白脂肪酶; 基因突变; 中国人群; 高脂血症](#)

分类号

## Genetic Screening of the Lipoprotein Lipase Gene for Mutations in Chinese Subjects with or without Hypertriglyceridemia

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### Abstract

**Objective:** To investigate the association between the mutations in lipoprotein lipase gene and hypertriglyceridemia (HTG). **Methods:** The lipoprotein lipase (LPL) gene was screened for mutations in 386 Chinese subjects with (108 cases in the HTG group) or without HTG (278 cases in the control group), by single-strand conformation polymorphism (SSCP) analysis and DNA sequencing. **Results:** One novel silent mutation L103L, one missense mutation P207L, three splicing mutations Int3/3' -ass/C(-6)→T, and the common S447X polymorphism has been identified in the whole coding region and exon-intron junctions of the LPL gene were examined. Heterozygous P207L found in the HTG group was the first case reported in Asia and subsequently another P207L heterozygote was found in the proband's family, all of which suggested that P207L was one of the causes of familial combined hyperlipidemia, but was not so prevalent as that in French Canadian. Int3/3' -ass/C(-6)→T was found in both groups in the present study although it was regarded as a pathogenic variant to HTG earlier on. Moreover about the beneficial polymorphism S447X, there was also some supportive evidence that the levels of triglycerides (TG) in S447X carriers were significantly lower than non-carriers in the subjects without HTG. **Conclusions:** The association between the LPL variants and HTG is quite complicated and versatile, genotyping of LPL in a larger-scale screening should be necessary and justifiable.

**Key words** [lipoprotein lipase](#) [mutations](#) [Chinese](#) [hypertriglyceridemia](#)

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