

# APOA1/C3/A4/A5基因簇的新载脂蛋白基因：APOA5及其研究进展

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**摘要** 应用人和鼠的比较基因组学和功能基因组学方法, Pennaccio等和Vliet等分别在APOA1/C3/A4基因簇中发现新的载脂蛋白基因APOA5。人的APOA5基因编码366个氨基酸, 与人APOA4、小鼠Apoa5高度同源。APOA5转基因小鼠其甘油三酯(TG)减少至野生型的1/3, 而Apoa5基因敲除小鼠其TG却增加4倍。APOA5多态位点SNP3(-1131T>C)和S19W及单倍型APOA5\*3有显著升高TG的作用。APOA5调节血浆TG水平的作用与APOC3作用相反, 为冠心病等心血管疾病的易感因素。

**关键词** [载脂蛋白APOA5](#) [APOA5基因](#) [甘油三酯](#) [冠心病](#)

分类号

## A Novel Gene in APOA1/C3/A4/A5 cluster : Apolipoprotein A5

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### Abstract

Using methods of comparative and functional genomics, a new gene coding for apolipoprotein A5 was identified in the vicinity of APOA1/C3/A4 cluster on human chromosome 11q23 by Pennaccio team and Vliet team. The open reading frame of human APOA5 encoded a 366-amino acid protein with high sequence homology to mouse ApoA5 and human APOA4. Mice expressing a human APOA5 transgene showed a decrease in plasma triglyceride concentrations to one-third of those in control mice; conversely, knockout mice lacking ApoA5 had four times as much plasma triglycerides as controls. Single nucleotide polymorphisms (SNPs) in APOA5 (S19W, -1131T>C) and APOA5 haplotype (APOA5\*3) were independently associated with high plasma triglyceride levels. These findings indicate that APOA5 is an important determinant of plasma triglyceride levels, a major risk factor for coronary artery disease.

**Key words** [Apolipoprotein A5](#) [APOA5 gene](#) [Triglycerides](#) [Coronary heart disease](#)

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