

综述

家族性高胆固醇血症亚型 ——隐性遗传性高胆固醇血症研究进展

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收稿日期 2005-12-15 修回日期 2006-3-10 网络版发布日期 接受日期

摘要

家族性高胆固醇血症(familial hypercholesterolemia, FH; MIM 143890)是一种常染色体显性遗传性疾病, 是脂质代谢疾病中最严重的一种, 导致早期发生较为严重的冠心病(coronary artery disease, CAD)。FH存在一些亚型, 其中常染色体隐性遗传性高胆固醇血症(autosomal recessive hypercholesterolemia, ARH; MIM 603813)纯合患者, 可表现为胆固醇水平异常升高、皮肤肌腱黄色瘤和早发的冠心病, 临床表现与FH极为相似。

关键词 [常染色体隐性高胆固醇血症](#); [衔接子蛋白](#); [家族性高胆固醇血症](#)

分类号

The subtype of familial hypercholesterolemia—the progression of autosomal recessive hypercholesterolemia

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Abstract

Familial hypercholesterolemia is a disease with autosomal dominant heritage. It is the most severe disease of lipid metabolism. It will induce severely coronary artery disease (CAD) in earlier period. Recently, some subtypes have been found in FH. One of them is the autosomal recessive hypercholesterolemia(ARH). The ARH-homozygotic patients have clinical manifestations such as abnormal cholesterol, xanthoma and premature CAD in earlier period, which are similar to FH.

Key words [autosomal recessive hypercholesterolemia](#) [adaptor protein](#) [familial hypercholesterolemia](#)

DOI:

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