遗传学报 2005 32 (7): 770-777 ISSN: 0379-4172 CN: 11-5450/R

综述

家族性高胆固醇血症样表型遗传异质性的分子基础

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收稿日期 2004-8-27 修回日期 2004-12-21 网络版发布日期 接受日期

摘要 家族性高胆固醇血症（FH）是由于低密度脂蛋白受体（LDL-R）基因突变，致使细胞表面LDL-R蛋白功能缺陷，导致血浆低密度脂蛋白（LDL）大幅度增高，并可导致早发冠心病。“FH”已经成为携带LDL-R基因突变患者的同意词，但日益增多的研究证实，其他6种基因突变也可通过不同机制导致FH样表型。这些致病基因的发现，促进胆固醇代谢的研究进入新领域，有助于深入探讨胆固醇代谢的调节机制，并将为FH样表型的诊断和治疗提供新的理论依据。本文就有关FH样表型遗传异质性的分子基础研究的近况作一简要综述， 以引起人们的关注。

关键词 家族性高胆固醇血症样表型；基因突变；异质性

分类号

Molecular Basis of Familial Hypercholesterolemia-like Phenotype Heterogeneity

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Abstract

Familial hypercholesterolemia (FH), which is caused by low-density lipoprotein (LDL) receptor mutation, leads to LDL-R dysfunction and high plasma LDL level and early onset of cardiovascular disease. LDL-r mutation has been regarded as the only cause of FH phenotype. However, evidences from recent studies showed that another six gene mutations can also result in FH like phenotype through different mechanism. Further studies on these genes will clarify the mechanism of plasma LDL regulation and provide the molecular basis for the diagnosis and treatment of patients with FH-like phenotype. This review summarizes recent studies on the molecular basis of FH-like phenotype heterogeneity in a new perspective in hoping of drawing attention to the disease.

Key words Familial hypercholesterolemia like phenotype gene mutation heterogeneity

DOI:

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