专论与综述

卵巢早衰候选基因的突变研究进展

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摘要

卵巢早衰病因复杂，多数病例病因不明。在已知的病因中，遗传因素非常重要。卵巢早衰不但可导致不孕症，患者的低雌激素水平还增加了患骨质疏松症和冠心病的危险。目前临床上的主要治疗措施包括激素替代治疗和供卵治疗不孕症等，但效果都不理想。鉴定卵巢早衰的致病基因是从根本上治疗和预防该病的基础。目前已发现在X染色体上和常染色体上多个基因与之相关，文章综述了近年来对卵巢早衰候选基因的突变分析情况，旨在从分子水平分析研究卵巢早衰的发病机制提供基础。

关键词　卵巢早衰　候选基因　突变分析

分类号　Q987

Advance in Mutation Analysis of the Candidate Genes in Premature Ovarian Failure

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Abstract

<P>Premature ovarian failure (POF) is a complicated and heterogeneous disease. In majority of cases the underlying cause is not identified. Among the known causes, genetic aberration plays very important role. POF not only causes infertility, also adds the risk of osteoporosis and coronary heart disease because of the low level estrogen. The major therapy measures in present include hormone replacement therapy and infertility treatment with donated oocytes, but the effect is not ideal. To identify the genes of POF is the basis for treating and preventing this disease. A large number of candidate genes of POF are found on X chromosome and autosomes. The present paper reviewed the advance in mutation analysis on the candidate genes of POF, which is aimed to provide a basis to explore its molecular mechanism.</P>

Key words　premature ovarian failure　candidate genes　mutation analysis

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