

## 中国人群的 $\beta$ 地贫的可诊断率与产前诊断中遗传标记的选择策略

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**摘要** 利用与 $\beta$ 地中海贫血致病基因连锁的遗传标记进行产前诊断已成为可能, 但群体中遗传标记与致病基因的不完全连锁决定了产前诊断的局限性。根据中国人群中7个遗传标记的多态性分布数据, 我们计算了各遗传标记及其所有组合的可诊断率。又据各遗传标记及其组合的可诊断率, 我们可推对: (1) 所作出产前诊断可诊断率进行预先评价; (2) 选择适合于中国人群  $\beta$ 地中海贫血产前诊断的最佳策略。本文提出了中国人群中选择遗传标记进行 $\beta$ 地中海贫血产前诊断的最佳路线。同时将基因的连锁分析法与寡核苷酸法进行了比较。

**关键词** [可诊断率,遗传标记,产前诊断, \$\beta\$ 地中海贫血](#)

分类号

## The Diagnosable Rate of $\beta$ -Thalassemia in Chinese Population and the Strategy for Selecting Genetic Markers in Prenatal Diagnosis of the Disease

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### Abstract

It is possible to use some  $\beta$ -thalassemia gene linkage markers for prenatal diagnosis of the disease currently. But there are some limitations due to incomplete linkage between these genetic markers and  $\beta$ -thalassemia gene in population. The diagnosable rate of the genetic markers and their combinations have been calculated according to the polymorphism distribution data of the seven genetic markers in Chinese population. Then according to the diagnosable rate of each genetic marker and their combinations, the following is deduced: (1) Precedent estimation to the diagnosable rate of prenatal diagnosis; (2) Select the optimum strategy for prenatal diagnosis of  $\beta$ -thalassemia suitable for Chinese population. In this paper, an optimum line is presented, regarding to select genetic markers for prenatal diagnosis of  $\beta$ -thalassemia in Chinese population. Meanwhile, the method of gene linkage analysis and that of using oligonucleotide probe are compared.

**Key words** [Diagnosable rate](#) [Genetic marker](#) [Prenatal diagnosis](#)  [\$\beta\$ -thalassemia](#)

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