

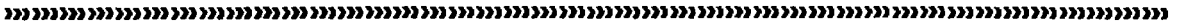
# THE APPLICATION OF PCR/HETERODUPLEX FORMATION ANALYSIS TO $\beta$ THALASSEMIA PRENATAL DIAGNOSIS

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A simple and rapid approach for detecting the  $\beta$  thalassemia gene [ Codon41-42(-CT-TT) ] by heteroduplex analysis after PCR is described. The diagnostic approach can distinguish between the heterozygote and homozygote. Four pregnant women at risk for  $\beta$  thalassemia were also studied by using this approach. The advantages, limitations and vistas of the approach are discussed.

**Key words** PCR; heteroduplex;  $\beta$  thalassemia; mutation; prenatal diagnosis



· 简 报 ·

## 广东汉族人基因组D17S5位点遗传多态性

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**关键词** D17S5位点; 遗传多态性; PCR; 汉族

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本文介绍用二步法 PCR 检测广东汉族人基因组 D17S5 VNTR (数量可变的串联重复序列) 遗传多态性的结果。共发现有12个等位基因。这些等位基因片段的大小在 100bp至 870bp之间 (每一重复单位为70 bp), 等位基因的频率分布在0.005至0.19之间。统计学分析证明D17S5各等位基因频率在本组人群的分布符合哈-温氏平衡 ( $\chi^2 = 74.63, df = 6.6, P > 0.05$ ), 并遵循孟德尔遗传规律。这一遗传多态性系统的杂合

度和父权排除率分别是85.95%和0.5575, 因而对个体识别和医学遗传学是一个很有价值的遗传标记。

Horn等曾提出在扩增 D17S5位点时会出现一些非特异性的扩增带。他们试图增加反应体系中 dNTP 的量来克服这种现象, 但这样会使实验的用费增加。我们提高复性温度至60℃, 使PCR变成二步法, 未再发现非特异扩增带。

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