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用DNA扩增法检测镰状细胞基因 *

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摘要 本文报道应用DNA扩增技术对国内首例镰状细胞特征患者（Hb s杂合子）进行基因诊断。方 法是从患者干血标本中微量抽提基因组DNA，通过聚合酶链反应（PCR）扩增其β珠蛋白基因 ，经限制性内切酶MstII消化后作电泳分析直接检测Hb s基因。本文介绍的DNA诊断技术快速 、灵敏、简便，它不需要放射性同位素标记的探针，可以采用干血抽提的DNA，因此，对遗 传病基因诊断和携带者的筛查具有重要价值。

关键词 中国人Hb s基因,DNA扩增,聚合酶链反应

分类号

Detection of Sickle Cell Gene by Analysis of Amplified DNA Sequences

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Abstract

This paper describes a technique of DNA amplification in vitro and its applicati on on detection of sickle cell(Hb S)gene.Genomic DNA was microextracted from dri ed blood specimen of the first patient with sickle cell trait in China.Target DN A sequence was amplified by the polymerase chain reaction(PCR)with the primers β1 (5’-ACACAACTGTGTTCACTAGC-3’)and β2(5’-CAACTTCATCCACGTTCACC-3’) that primed a mplification of an 110-base -pair(bp)segment of βglobin gene.The amplified DNA was digested with a restriction endonuclease Mst II,which has a recegnition site at codon 6 in the normal βglobin gene,and cleaved the normal amplified βglobi n DNA into two fragments of 54bp and 56bp which was as an overlap band in agaros e gel electrophoresis,while the 110bp fragment ampified from DNA of sickle cell mutaion remained uncleaved owing to a single base substitution(A→T)at codon 6 i n the mutation. DNA amplification method is rapid,sensitive and simple,and does not require radi oactive probes.Besides,the PCR amplification can be carried out on the DNA extra cted from dried blood samples.So the technique is ver useful for gene diagnosis and carier screening of genetic disease.

Key words Chinese HbS gene DNA amplification Polymerase chain reaction

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