遗传学报 1988 15 (1): 52-60 ISSN: 0379-4172 CN: 11-5450/R

用合成的寡核普酸探针鉴定中国人口一地中海贫血基因突变[1)]

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收稿日期 修回日期 网络版发布日期 接受日期

摘要 用人工合成的六种对13-地中海贫血基因特异的寡核昔酸作为杂交探针，对10例小地中海贫血病人及其父母的份珠蛋白基因进行分析，鉴定出六种价地中海贫血突变：(1)TATABox-28 A--->G; (2)IVS-1n.5 G--+C; (3) Codon 17 A---,PT; (4) Codons 41-42-46P; (5) odons 71--72+A;(的IVS-4 n.654Cr-aT。本文分析的中国人R-地中海贫血患者中，上述六种突变所占的百分比分别为5%p,1U%,10%940%}20%和15%0

关键词 中国人0-地贫突变,寡核普酸杂交

分类号

Use of Synthetic 01igonucleotides in the Detection of --thalassemia Mutation in Chinese*

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Abstract

This article describes the use of six synthetic oligonucleotides specific for six types of point mutations in the analyses of mutant O-globin genes by molecular hybridization to DNA of 10Chinese families of patients with月-thalassemia. The samples .were collected from Shanghai,,Zhejiang, Sichuang, Hunan and Guangxi. Thesix types are (A) A--->G substitution at TATA Bo、一28 bp, (B) G-->C substitution at IVS-1, position 5, (C) A-->T substitution in codon 17,,(D) 4 base pair deletion (TCTT) in codons 41----42, (E) nucleotide insertion of A between codons 71-72 and (F) C--*T substitution at IV-S-2, position 654. Results showed that the per-,centages of these six types of mutation were 5%, 10% 10%, 40%, 2001o and 15% of the日-thala--ssemia genes in these patients, respectively. This is in contrast to the reported data (7, 10) in which most of the mutant genes was caused by type E and F mutation and none of type A ,.was reported. This discrepancy could be due to the difference in the geographic locations of the Chinese patients conducted in these studies. In addition, although previous data showed aclose association of haplotypes and specific mutations in an ethnic group, our data showed'that four types of mutations (C, D, E, F) were found in the most common haplotypes+一一一+一+. A plausible explanation is that the multiple mutations found in this haplotype may have.arisen by gene conversion.

Key words Chinese thalassemia mutation Oligonucleotide hybridization

DOI:

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