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一个典型中国a一地中海贫血家系的基因分析

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摘要 本文应用印迹杂交技术和a一珠蛋白基因探针，对临床上发现的一个HbH病人DNA进行了限制性内切酶图谱分析，确定其为a一地贫1与右侧缺失a一地贫2双重杂合子（a-/--）。对其家系的另11个成员进行了a一珠蛋白基因分析，证明他们均为。一地贫基因携带者。其中先证者的父亲及一个女儿的基因型与先证者相同，母亲、妻子、两个弟弟及长子为a-地贫I杂合子（aa/--），两个妹妹及二子、三子均为右侧缺失。一地贫2杂合子（a-/--）。家系的基因分析对采取各种途径控制a-地贫的传延具有重要意义。

关键词
分类号

Gene Analysis of a Typical Chinese a-thalassemia Family

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Abstract

Utilizing Southen blot technique and a-globin gene probe physical maps of restriction endonuclease sites around a-globin genes of an HBH patient found in clinic are fulfilled. The results show that itis a double heterozygote of a-thal 1 and rightward deletion a-thal 2 genotype(a-/--). The arrangement of a-globin genes of another 11 members of his family is analyzed. The results show that they all are carriers of a-thal gene. The genotypes of propositus' father and daughter are the same as propositus'. His mother, wife, two brothers and the eldest son are heterozygotes of a-thal 1 a-thal 2. Gene analysis of families is of important significance for contrlo of a-thalassemia by adopting different preventive measures.

Key words

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