

中国南方汉族群体MPSI型Kpn I酶切位点的遗传多态性 RFLP of a KpnI Site in the α -L-iduronidase(IDUA) Gene in Mucopolysaccharidosis Type I of Chinese Southern Han Population

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摘要 为研究中国汉族群体IDUA 基因Kpn I 酶切位点的遗传多态性以及该位点等位基因片段传递的规律, 采用PCR-RFLP技术, 对162例无血缘关系的健康中国汉人的324条 染色体进行检测, 另又对5个家系16位成员进行同样的检测, 然后用 χ^2 检验进行统计学处理。结果表明, 等位基因A1 频率为0.17, 等位基因A2 频率为0.83, 杂合率为29%; A1、A2 的传递规律与理论上预计的完全符合。认为中国汉族群体IDUA 基因 Kpn I 酶切位点也具有遗传多态性, 并且与国外报道的无显著性差异; A1、A2 在世代中的传递完全符合孟德尔遗传规律。

Abstract: To investigate the genetic polymorphism of the Kpn I site in the α -L-iduronidase(IDUA) gene from a Han population in southern China and to study the mode of transmission of alleles, PCR-RFLP was used to analyze 324 chromosomes from 162 Chinese unrelated healthy Han individuals, and the analysis of the genotypes of 16 members in five families. To compare the frequencies and heterozygosity between Chinese Han population and Caucasians in Western by using χ^2 test. The frequency of allele 1 (450bp) was 0.17, allele 2 (390 plus 60 bp) 0.83, the heterozygosity was 29%. The genotypes of each member of all families detected was completely agreement with the theoretical assessment. The locus of Kpn I in the IDUA gene from Han population has polymorphism. There is no significant difference between Chinese Han population and Caucasians in Western countries. The transmission of alleles was agreement with the Mendelian genetic law.

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