

广东少数民族Morquio综合征StuI位点的RFLP分析 RFLP of a StuI Site in the GALNS Gene in Morquio Syndrome of Guangdong National Minority Population

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摘要 研究广东少数民族群体GALNS基因StuI位点的遗传多态性以及该位点等位基因片段传递的规律, 为今后的连锁分析打下基础。采用PCR-RFLP方法, 对72例无血缘关系的健康广东少数民族个体的144条染色体和3个家系9位成员的18条染色体进行检测, 然后用 χ^2 检验进行统计学处理。等位基因片段D1的频率为0.70, D2为0.30, 杂合率为29%, D1、D2的传递规律与理论上预计的完全符合。广东少数民族群体中StuI位点具有多态性, 其基因频率(D1和D2)与国外高加索群体的有显著差别, 与日本群体及中国南方汉族群体的则无显著差别; 而杂合率与高加索群体及日本群体的均有显著差异, 但与中国南方汉族群体的则无显著差异。

Abstract: To investigate the genetic polymorphism of the StuI site in the GALNS gene from a national minority population in Guangdong and to study the mode of transmission of alleles, PCR-RFLP was used to analyze 144 chromosomes from 72 Guangdong unrelated healthy national minority individuals, and the genotypes of members in three families. To compare the frequencies and heterozygosity between Guangdong national minority people and Caucasians, Japanese and Chinese Han people by using χ^2 test. The frequency of allele D1 (295bp) was 0.70, allele D2 (138 plus 157 bp) 0.30, the heterozygosity was 29%. The genotypes of each member of all families detected were completely agreement with the theoretical assessment. The site of StuI in the GALNS gene from national minority population in Guangdong has polymorphism. There is significant difference between Guangdong national minority population and Caucasians in Western countries, but no significant difference was found between Guangdong national minority population and Japanese and Chinese Han population. In addition, there is significant difference between Guangdong national minority population and Caucasians and Japanese in the heterozygosity, but no significant difference between Guangdong national minority population and Chinese Han population. The transmission of alleles was completely in agreement with the Mendelian genetic law.

关键词 [Morquio综合征](#) [GALNS基因](#) [多态性](#) [限制性片段长度](#) **Key words** [Morquio syndrome](#) [GALNS gene](#) [polymorphism](#) [restriction fragment length](#)

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