

中国汉蒙两族人群MTHFR基因热敏感性多态性分布的比较 Comparing the Distribution of Genetic Polymorphism of MTHFR Thermolabile between Mongolian Population and Hans of China

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摘要 为比较中国蒙汉两族人群MTHFR基因第677位核苷酸多态性的分布情况, 获得该位点多态性的群体遗传学数据, 本研究应用PCR扩增技术, 其扩增产物用限制性核酸内切酶Hinf I消化后进行非变性聚丙烯酰胺凝胶电泳, 分析蒙汉族人群中MTHFR基因第677位核苷酸基因型(野生型、杂合型和突变纯合型)的分布频率。结果表明, 蒙族人群基因型构成以野生型为主, 占45.6%, 突变杂合型占39.2%, 突变纯合型仅占15.2%, 汉族人群基因型构成以突变杂合型为主, 占55.7%, 野生型仅占17.9%, 突变纯合型占26.4%, 明显高于蒙族人群。经 χ^2 检验, 两组基因型构成比具有显著性差异($P < 0.001$); 蒙族人群MTHFR 677T等位基因频率为34.8%, 经u检验显著低于汉族人群(54.2%)的频率。据此认为, 中国蒙族人群MTHFR热敏感性基因突变频率显著低于汉族人群, 提示该基因多态性分布在中国不同民族人群中存在差异。

Abstract: The purpose of this study is to compare the genetic polymorphism distribution of the 677th nucleotide of MTHFR between the Mongolian population and the Hans of China, and to obtain the population genetic data of this polymorphism. Using PCR-RFLP method, the authors analyzed the genotypes of the 677th nucleotide of MTHFR in Mongolians and Hans. Results show that in Mongolian population, the proportion of wild type is 45.6%, proportion of heterozygotes is 39.2% and that of homozygotes is 15.2%; While in Hans, proportions are wild type 17.9%, heterozygotes 55.7% and homozygotes 26.4%. The ratios of genotypes are significantly different between Mongolian and Han populations (χ^2 -test, $P < 0.001$). The 677th allele frequency in Mongolians is 34.8%, lower than that in Hans (54.2%, u-test, $P < 0.001$). This suggests that the mutant MTHFR gene frequency is significantly higher in the Han population than in the Mongolian population in China.

关键词 [蒙汉族人群](#) [聚合酶链式反应](#) [MTHFR](#) [基因多态性](#) **Key words** [Mongolian population and Hans polymerase chain reaction\(PCR\)](#) [MTHFR](#) [genetic polymorphism](#)

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Abstract

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