

父母MTHFR基因型对后代发生神经管畸形的影响 Effect of Parental MTHFR Genotypes on Offspring NTD Risk

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摘要 本研究旨在探讨父母双方MTHFR基因型与其后代发生神经管畸形风险性的关系。采用成组匹配的病例对照研究方案,用纸片法采集病例和对照外周血标本,用PCR-RFLP方法确定基因型(C677T)。结果显示,分别比较两组父亲或母亲基因型频率时,差别不显著;将父母基因型联合起来分析则发现,病例组父母等位突变基因频率(T)高于对照组父母,提示后代是纯合突变(TT)概率越大,发生NTD的风险性也越大。结论是,父母双方的基因型对后代发生NTD的风险性有同等重要的作用,父母双方传递给胎儿的一对突变等位基因(T/T)是NTD发生的风险因素之一。

Abstract:The effect of parental MTHFR genotypes on fetal phenotype of NTD is studied.It was designed as group matched case-control study.Venous blood cases and controls were collected by dry blood spots.Genotype(C677T)of each sample was decided by PCR-RFLP method.When the genotypes of father and mother are compared separately,no significant difference is discovered between cases and controls.On the other hand,when the mutant allele frequency(T)is computed for father and mother together,it is significantly higher in cases than in controls.Higher probabilities of the offspring being homozygotes(TT),higher the risk for NTD occurrence has been observed.It is concluded that genotypes of MTHFR for both father and mother have the same impact on NTD occurrence of their offspring.It is the pair of mutant allele(T/T)which transmitted to the baby from its parents that increases the risk.

关键词 [MTHFR](#) [神经管畸形](#) [病例对照研究](#) **Key words** [MTHFR](#) [neural tube defects\(NTD\)](#) [case-control study](#)

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