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COMT基因多态性与儿童精神发育迟滞及儿童认知能力的相关性研究

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摘要

儿茶酚-O-甲基转移酶(catechol-O-methyl transferase，COMT)是儿茶酚胺类神经递质的主要代谢酶。COMT是儿茶酚-O-甲基转移酶的编码基因，其第4外显子的一个G/A转换可产生不同活性的等位基因。许多遗传学研究报道，这种功能多态性与人类精神类疾病相关。文章利用PCR扩增技术和限制性片段长度多态(Restriction fragment length polymorphism, RFLP)方法，研究中国秦巴山区精神发育迟滞(Mental Retardation, MR)儿童与正常对照儿童的COMT基因功能多态情况，探讨COMT基因功能多态性与儿童认知能力水平的相关性。病例-对照分析结果显示，COMT基因的不同活性等位基因型与秦巴山区儿童MR无相关性 (χ^2=0.776, P>0.05)，其等位基因频率与儿童MR也未呈现出相关性(χ^2=0.335, P>0.05)。但是，在研究中还发现，该地区智商分(IQ)不小于55分的儿童群体中，COMT基因的多态性分布与儿童的智力呈现出相关的趋势。在智力正常组儿童中(IQ≥85)，COMT高活性等位基因纯合体(COMTHH)频率及其等位基因(COMT^H)频率较高，分别为60.98%、79.28%。在智力边缘组儿童(70≤IQ＜85)中，其频率分别为46.67%、70.67%，相应地也低于正常组的频率(0.10>P>0.05)。结果提示，在中国秦巴山区人群中，COMT基因的功能多态性与儿童MR的形成无明显相关性，但它对正常儿童的认知能力可能有一定影响。

关键词

儿茶酚胺-O-甲基转移酶；功能多态性；精神发育迟滞；认知能力

分类号

Association Between a Functional COMT Polymorphism, Mental Retardation and Cognition in Qinba Area Children

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Abstract

<P>Catechol-O-methyl transferase (COMT) plays an important role in the metabolism of neurotransmitters. Two alleles of the COMT gene as a result of a G/A transition in the exon 4 can lead to different COMT enzymatic activities. Much genetic research has revealed that this COMT functional polymorphism was related to human psychiatric disorders. Polymerase chain reaction (PCR) and restriction fragment length polymorphism (RFLP) methods were used to discern the relationships among the functional polymorphism of COMT, mental retardation (MR),

and general cognitive ability of children. The results of the case-control analysis showed that there was no association between the frequencies of genotypes of COMT and MR (χ²=0.776, P>0.05) or between the frequency of COMT alleles and MR (χ²=0.335, P>0.05). COMT polymorphism was found in children whose intelligence quotient (IQ) was above 55. In normal children (IQ≥85), the frequencies of high-activity allele <EM>COMT^H</EM> and the homozygote genotype <EM>COMT^{HH} </EM>were 60.98% and 79.28%, respectively. Both were higher than those of the borderline group (46.67% and 70.67%, 0.10 > P>0.05). Therefore, the result of this study suggests that this functional polymorphism is not an important risk factor for MR, but the COMT^{HH} genotype may have a positive effect on cognitive performance in normal children in the Qinba area.</P>

Key words catechol-O-methyl transferase (COMT)；functional polymorphism；mental retardation；cognitive ability

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