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受体型蛋白酪氨酸磷酸酶R基因多态性与重性抑郁障碍的关联分析

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Association between Protein Tyrosine Phosphatase Receptor Type R Gene and Major Depressive Disorder

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摘要 **目的**探讨受体型蛋白酪氨酸磷酸酶R (PTPRR)基因多态性与重性抑郁障碍 (MDD)及其内表型的关联性。 **方法** 收集中国北方汉族MDD患者517例（MDD组）和健康志愿者455人（对照组），采用时间飞行质谱技术检测研究对象PTPRR基因的11个单核苷酸多态性 (SNPs)位点的多态性，采用UNPHASED软件进行等位基因、基因型、单倍型及数量性状分析。 **结果** 单位点分析显示，等位基因频率和基因型分布在MDD组和对照组间差异无统计学意义（校正后$P>0.05$）；单倍型分析显示，三位点单倍型rs1398599 (C)-rs2175711 (A)-rs4489789 (T)($P=0.0023$，OR=1.334, 95%CI=1.104～1.612)和四位点单倍型rs11178391 (C)-rs1398599 (C)-rs2175711 (A)-rs4489789 (T)($P=0.0063$, OR=1.281, 95%CI=1.059～1.549)均显著增加MDD发病风险；数量性状分析显示，SNP rs2203231的等位基因和基因型分别与 韦氏记忆量表 (WMS-R) 的心智原始分($P=0.0038$，$P=0.0024$) 和心智量表分 ($P=0.0057$，$P=0.0038$) 以及图片原始分($P=0.0027$，$P=0.0015$)和图片量表分 ($P=0.0035$，$P=0.002$) 显著相关。 **结论** PTPRR基因rs2203231多态性可能与MDD患者的长时记忆和短时记忆功能障碍相关联。

关键词： 重性抑郁障碍 受体型蛋白酪氨酸磷酸酶R基因 单核苷酸多态性 关联分析 内表型

Abstract： Objective To explore the genetic association between protein tyrosine phosphatase receptor type R (PTPRR) gene polymorphism and major depressive disorder (MDD) and its endophenotype. Methods A total of 517 unrelated MDD patients and 455 unrelated healthy subjects were recruited in this study to detect 11 single nucleotide polymorphisms (SNPs) in the PTPRR locus. They all were of the Chinese Han origin. Genotyping of SNPs was performed by matrix assisted laser desorption ionisation time-of-flight mass spectrometry (MALDI-TOF-MS)-based genotyping approach. The UNPHASED program was applied to analyze the genotyping data. Results Of the 11 selected SNPs, no significant allelic and genotypic association was found between MDD patients and the normal controls (corrected $P>0.05$). However, analysis of haplotypes showed that the three SNPs haplotype rs1398599(C)-rs2175711(A)- rs4489789(T)($P=0.0023$, OR=1.334, 95%CI=1.104-1.612) and four SNPs haplotype rs11178391(C)-rs1398599(C)-rs2175711(A)-rs4489789(T)($P=0.0063$, OR=1.281, 95%CI=1.059-1.549) were associated with increased risk of MDD. Quantitative trait analysis revealed that rs2203231 in the PTPRR locus had strong allelic and genotypic association with the raw score of long-term memory ($P=0.0038$ for allelic association, $P=0.0024$ for genotypic association), the scaled score of long-term memory ($P=0.0057$ for allelic association, $P=0.0038$ for genotypic association),the raw score of short-term memory ($P=0.0027$ for allelic association, $P=0.0015$ for genotypic association), and the scaled score of short-term memory ($P=0.0035$ for allelic association, $P=0.002$ for genotypic association) in MDD patients. Conclusion The polymorphism of PTPRR gene rs2203231 may be associated with the impairment of long-term and short-term memories in MDD patients.

Keywords： major depressive disorder protein tyrosine phosphatase receptor type R single nucleotide polymorphism association study endophenotype

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