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# 新发现的蒙古族ABCA1M233V基因和R219K基因多态性

# A novel polymorphism in ABCA1 gene (M233V and R219K) in Mongolian nationality population

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**中文关键词:** 冠心病; 三磷酸腺苷结合盒转运子A1; 多态性; 蒙古族

**英文关键词:**coronary heart disease; ATP binding cassette transporter 1; polymorphism; Mongolian nationality

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**中文摘要:**

目的 探讨ATP结合盒转运子A1基因（ABCA1）M233V和R219K多态性在内蒙古地区蒙古族人群中的分布及其与血脂和冠心病的关系。方法 采用聚合酶链反应-限制片段长度多态性（PCR-RFLP）方法检测115例蒙古族冠心病患者和对照组ABCA1基因相应片段的多态性。结果 内蒙古地区蒙古族人群中ABCA1基因M233V多态性位点存在MM, MV和VV三种基因型, 其在冠心病患者和对照组人群的基因多态性分布差异无统计学意义（P＞0.05）。MM组和MV+VV组总胆固醇（TC）, 甘油三酯（TG）, 高密度脂蛋白胆固醇（HDL-C）和低密度脂蛋白胆固醇（LDL-C）比较, 差异无统计学意义（P＞0.05）。对照组KK基因型的频率显著高于CHD组（P＜0.05)。KK基因型的HDL-C水平显著高于RR型（P＜0.05）, KK型TG水平明显低于RR型, 差异有统计学意义（P＜0.05)。结论 内蒙古地区蒙古族人群中存在新发现的ABCA1基因M233V多态性, ABCAl基因R219K多态性与内蒙古地区蒙古族人群冠心病的遗传易感性相关, KK基因型产生有益的临床血脂谱, 可能是冠心病患者的低危遗传标记。

**英文摘要:**

Objective To investigate ATP-binding cassette transporter 1(ABCA1) M233V and R219K gene polymorphism in Mongolian nationality population in Inner Mongolia region and its correlationship with blood lipids and coronary heart disease(CHD). Methods The target fragments of ABCA1gene were amplified and analyzed by polymerase chain reaction-restriction fragments length polymorphism(PCR-RFLP) technique in 115 Mongolian control subjects without CHD and patients with CHD. Results The subjects presented ABCA1 gene M233V polymorphism, which had three genotypes: MM genotype, MV genotype and VV genotype. There was no significant difference in frequency of allele and genotype in M233V polymorphism between controls and CHD patients(P＞0.05). No significant difference was found in levels of total cholesterol(TC), triglyceride(TG), high density lipoprotein-cholesterol(HDL-C) and low density lipoprotein-cholesterol(LDL-C) between MM genotype and MV+VV genotype(P＞0.05).The frequency of KK genotype was significantly higher in controls than in CHD patients (P＜0.05). KK genotype had higher HDL-C level(P＜0.05) and lower TG level than RR genotype (P＜0.05).Conclusion In Mongolian population in Inner Mongolia region, M233V is a novel polymorphism in ABCAl gene. ABCAl gene R219K polymorphism is associated with CHD risk. The KK genotype results in a beneficial profile of blood lipids, which may be a novel genetic marker for low risk of CHD.

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