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论著

δ-氨基-γ-酮戊酸脱水酶和Vit D受体基因多态性与不同民族儿童铅中毒的遗传易感性研究

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摘要 背景与目的: 探讨汉族、维族和哈族儿童δ-氨基-γ-酮戊酸脱水酶(ALAD)、维生素D受体(VDR)基因多态性及其与铅中毒遗传易感性的关系。 材料与方法: 采用聚合酶链式反应-限制性片段长度多态性(PCR-RFLP)方法对新疆乌鲁木齐市的489名汉族、499名维族和525名哈族儿童ALAD、VDR基因多态性进行分析。 结果: ALAD、VDR不同基因型在汉、维、哈族中分布的差异均具有统计学意义(P<0.01)。对VDR基因的3个等位基因位点BsmI、Taq I和Apa I单体型分析发现,汉族儿童中单体型Atb、AtB在铅中毒组显著降低(P<0.01),而单体型ATb、aTb在铅中毒组显著增高(P<0.05)。其它民族单体型分析未显示统计学意义(P>0.05)。 结论: ALAD、VDR不同基因型分布具有种族差异;在汉族儿童中,单体型Atb和AtB可能是铅中毒的保护因素,而单体型ATb和aTb可能是铅中毒的危险因素。

关键词 δ-氨基-γ-酮戊酸脱水酶 维生素D受体 基因多态性 单体型 铅中毒

Polymorphisms of δ-Aminole- vulinic Acid Dehydratase; Vit D Receptor and Genetic Suscep- tibility of Lead Posioning in Han; Uygur and Kazak Children

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Abstract BACKGROUND AND AIM:To explore the polymorphisms of δ-aminolevulinic acid dehydratase(ALAD); vitamin D receptor(VDR) and genetic susceptibility of lead posioning in Han; Uygur and Kazak children. MATERIALS AND METHODS: The ALAD and VDR genotypings were determined by PCR-RFLP in 489 Han; 499 Uygur and 525 Kazak individuals from Urumqi city of Xinjiang province. RESULTS: The genotype frequencies of ALAD and VDR showed significant differences in Han; Uygur and Kazak subjects (P<0.01). According to VDR-BsmI、Taq I and Apa I haplotype analysis; haplotype Atb and AtB in Han were considerately decreased in lead poisoning group(P<0.05) while haplotype aTb and ATb significantly increased in lead poisoning group(P<0.01).However; such results were not found in Uygur and Kazak (P>0.05). CONCLUSION: A significant difference was seen in the frequency distribution of ALAD and VDR genotype among the different races. Haplotype Atb and AtB might be protective factors while haplotype ATb and aTb might be risk factors in Han.

Keywords δ-aminolevulinic acid dehydratase vitamin D receptor genetic polymorphism haplotype lead posioning

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