[1]杨俊慧,王小明,王朝永,等.过氧化氢酶基因389C>T多态位点在重庆汉族人群中的分布及其与噪音性耳聋的关联研究[J].第三军医大学学报,2014,36(05):473-477.

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过氧化氢酶基因389C>T多态位点在重庆汉族人群噪音性耳聋的关联研究(PDF)

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Title: Relationship between 389 C>T polymorphism of catalase gene and noise-induced hearing loss in Han population of Chongqing

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关键词: 噪音性耳聋; rs769217; 过氧化氢酶; 单核苷酸多态性

Keywords: noise-induced hearing loss; catalase; 389 C>; T polymorphism (rs769217); single nucleotide polymorphism

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摘要: 目的　探讨过氧化氢酶(catalase, CAT)基因外显子区第9外显子的同义突变位点389C>T（rs769217）多态性在重庆汉族人群中的分布及其与噪音性耳聋的关联研究。

方法　以中国重庆地区汉族无血缘关系的健康志愿者225例，噪声接触者427例作为研究对象。噪音接触者根据接触噪声后是否出现噪音性听力丧失（noise-induced hearing loss，NIHL）又分为非耳聋组，即NIHL(-)组(*n*=303)和耳聋组，即NIHL(+)组(*n*=124)。采用上海天昊公司的专利技术iMLDR^{TM}分型技术，分别检测各组CAT基因389C>T多态位点的基因型，并比较组间基因型、等位基因频率分布及临床变量间的差异。　结果　在研究人群中检测到CAT基因389C>T多态位点的3种基因型CC、CT

和TT，2组人群的T等位基因频率均为0.46，C等位基因频率为0.54，健康对照组和噪音接触组基因型频率分布符合Hardy-Weinberg遗传平衡定律(P>0.05)。健康对照组与噪音接触组、健康对照组与NIHL(-)组间CAT基因389C＞T多态位点的3种基因型(CC、CT和TT)和等位基因(C、T)频率分布均无显性差异(P>0.05)；但健康对照组与NIHL(+)组及NIHL(-)组与NIHL(+)组间3种基因型(CC、CT和TT)和等位基因(C、T)频率分布均有显著性差异(P<0.05),且显性模型分析中即TT+CC *vs* CC均有显著性差异（P<0.05）。　　结论　　389C＞T位点（rs769217）与重庆汉族人群中NIHL遗传易感性有关联，在重庆汉族人群中可作为噪音性耳聋易感性的生物标志。

Abstract:　Objective　To explore the distribution of 389 C>T (rs769217) polymorphism of catalase (CAT) gene in Han population of Chongqing, China, and the relationship between the polymorphism and noise-induced hearing loss (NIHL). Methods　In Chongqing, China, 427 unrelated adult Chinese Han people who were exposed to noise (noise exposure group) and 225 healthy volunteers without exposure (control group) were included in the study. The noise exposure group was further divided into NIHL (-) group (without NIHL, n=303) and NIHL (+) group (with NIHL, n=124). An improved multiplex ligation detection reaction (iMLDR) technique was employed to detect genotypes of 389 C>T polymorphism of CAT gene. Allelic frequencies and clinical characteristics were compared among these groups. Results　Three genotypes, CC, CT and TT, were detected. The frequencies of C and T allele of the CAT gene 389 C>T polymorphism were 0.46 and 0.54 respectively. The genotype distribution in the control group and noise exposure group was in agreement with the Hardy-Weinberg equilibrium (P>0.05). There was no significant difference in genotypic (CC, CT, TT) and allelic frequencies (C, T) between the control group and noise exposure group and between the control group and NIHL (-) group (P>0.05). However, significant differences were observed between the control group and NIHL (+) group and between the NIHL (-) group and NIHL (+) group (P<0.05), especially in the case of dominant association effect (TT+CT *vs* CC, P<0.05). Conclusion　The person who is exposed to noise with the T variant allele has significantly higher NIHL morbidity than those with homozygous C allele. CAT gene 389 C>T polymorphism may be used as a biomarker for the assessment of NIHL morbidity in Chongqing Han population .

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