

人类与医学遗传学

## 钙敏感受体基因、白介素6基因与中国人群骨密度相关联

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摘要

钙敏感受体是钙新陈代谢的一个重要因素, 白介素6是参与破骨细胞分化及功能的一种多效细胞因子。因此, 钙敏感受体基因和白介素6基因都为骨矿物质代谢的重要候选基因。本研究旨在利用数量性状传递不平衡检测法, 检测白介素6基因和钙敏感受体基因与腰椎和髌部骨密度的关联和连锁, 以证实它们是否为影响中国人群骨密度的重要候选基因。本研究的样本为来自中国上海的401个中国核心家庭, 共1, 263个个体, 均为汉族。每个核心家庭由父母双亲和至少一个20~45岁的健康绝经前女儿组成。腰椎与髌部的骨密度采用Hologic QDR 2000+ 双能X射线扫描仪进行了检测。用PE377测序仪及相关软件分别对白介素6和钙敏感受体基因中的一个CA重复多态微卫星位点进行了基因分型。分析结果表明钙敏感受体基因(CA) 12等位基因 (P = 0.006) 及(CA) 18等位基因 (P= 0.02) 与股骨颈骨密度之间存在显著的整体关联。白介素6基因的(CA) 14等位基因与整个髌部 (P = 0.021)、股骨颈 (P= 0.041) 以及转子间区 (P = 0.029) 骨密度之间均存在显著的家庭内关联。白介素6基因 (CA) n位点与腰椎BMD之间存在显著的连锁 (P = 0.001)。本研究结果表明白介素6基因和钙敏感受体基因可能为与中国人群骨密度变异相关联的候选基因。

关键词 [关联; 连锁; 骨密度; 钙敏感受体基因; 白介素6基因; QTDT检测](#)

分类号

## The Human Calcium-Sensing Receptor and Interleukin-6 Genes are Associated with Bone Mineral Density in Chinese

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### Abstract

<P> Calcium sensing receptor (CASR) is a central factor involved in calcium metabolism. Interleukin-6 (IL-6) is a pleiotropic cytokine that plays an important role in osteoclast differentiation. Thus, both CASR and IL-6 are important in bone and mineral metabolism and are prominent candidate genes for osteoporosis. The study aimed to test association and/or linkage between the CASR and IL-6 genes with bone mineral density (BMD) variation in a Chinese population. A cytosine-adenine (CA)n repeat polymorphism in the CASR gene and the IL-6 gene was genotyped, respectively, in 1 263 subjects from 402 Chinese nuclear families. Employing tests implemented in the program QTDT (quantitative transmission disequilibrium tests), a significant total association of the CASR (CA)12 allele (P = 0.006) and (CA)18 allele (P = 0.02) with BMD at the femoral neck was found. For the IL-6 gene, significant within-family associations were found between the (CA)14 allele and BMD at the total hip (P = 0.021), the femoral neck (P = 0.041), and the intertrochanteric region (P = 0.029). A significant linkage was also observed between IL-6 CA repeat polymorphism and BMD at the spine (P = 0.001). The results

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suggest that the CASR gene and the IL-6 gene may have effects on BMD variation in Chinese.</P>

**Key words** [association](#); [bone mineral density](#); [CASR gene](#); [IL-6 gene](#); [linkage](#); [QTDT test](#)

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