

α2-HS 糖蛋白基因与中国人群的髋部骨大小的相关性研究

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摘要 骨大小是一种独立于骨密度(BMD)的骨质疏松性骨折的重要风险因子。由于其高遗传率, 充分了解控制骨大小的遗传因素有很重要的临床意义。本研究目的为检测中国人群中α2-HS糖蛋白基因(AHSG)多态性和腰椎及髋部骨大小变异之间的关联。我们总共征集了来自中国401个核心家庭(包括父母亲及至少一个女儿)的1 260个研究样本, 并且分型了AHSG基因第7个外显子的Sac I位点多态性。该位点核苷酸的替换(C→G)引起第238号丝氨酸被苏氨酸取代, 因此可能对基因功能有影响。在任何骨骼位点, 没有发现显著的群体分层。发现AHSG基因Sac I位点多态性和转子间(P= 0.019)以及全髋的(P=0.035)骨大小呈显著性相关。该多态性位点能分别解释转子间和全髋3.74%和3.16%的骨大小变异。连锁分析没有检测到显著性结果, 可能的主要原因是样本中同胞对的数目较少, 统计效力较低, 以及Sac I位点多态相对于微卫星标记对连锁分析提供的信息量少。结果表明, AHSG基因多态性可能和中国人群中髋部骨大小变异有关。

关键词 [骨大小](#); [α2-HS糖蛋白\(AHSG\)](#); [关联](#); [连锁](#); [传递不平衡检测](#)

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Alpha2-HS Glycoprotein Gene is Associated with Bone Size at the Hip in Chinese

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Abstract

Bone size is an important risk factor, independent of bone mineral density (BMD), for osteoporotic fracture. Bone size has a high heritability. A better understanding of genetic factors regulating bone size will have important clinical implications. In this study, we explored the relationship between the alpha2-HS glycoprotein (AHSG) gene and bone size variation at the spine and hip in a Chinese population. The study sample comprised 1 260 subjects from 401 Chinese nuclear families (each including both parents and at least one female child). The Sac I polymorphism inside the exon 7 of the AHSG gene was genotyped and analyzed. This variant represents a nucleotide substitution of C to G at amino acid position 238 resulting in a translation polymorphism of threonine to serine and thus making a potential impact on gene function. We assessed population stratification but did not find significant evidence at any skeletal sites. We found significant association between the AHSG Sac I polymorphism and bone size at the intertrochanteric region (P = 0.019) and the total hip (P = 0.035). The polymorphisms explained 3.74% and 3.16% variations in bone size at the intertrochanteric region and total hip respectively. No significant evidence of linkage was detected, largely due to the limited number of sibpairs in this data set and less informative marker (AHSG Sac I polymorphism) (compared with microsatellite markers) for linkage analysis. Our results suggested that the AHSG gene may contribute to bone size variation at the hip in this Chinese population.

Key words [bone size](#) [alpha2-HS glycoprotein \(AHSG\)](#) [association](#) [linkage](#) [transmission disequilibrium test](#)

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