Association of insulin growth factor-1 receptor gene polymorphisms with genetic susceptibility to idiopathic short stature

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ABSTRACT. The association between single nucleotide polymorphisms (SNPs) in the insulin-like growth factor-1 receptor (IGF-1R) gene and susceptibility to idiopathic short stature (ISS) was investigated. Seven hundred and twelve Chinese children clinically diagnosed with ISS and 575 normal individuals were recruited between 2008 and 2011, and their SNPs were genotyped. Preliminary screening revealed that the rs1976667 and rs2684788 loci were significantly associated with genetic susceptibility to ISS (P = 0.03636 and P = 0.01352, respectively). Stratification by sex revealed that in males, different genotypes at the rs1976667 locus were significantly associated with genetic susceptibility to ISS (P = 0.047), showing G dominant inheritance (P = 0.018). The G allele at the rs2684788 locus was significantly associated with genetic susceptibility to ISS (P = 0.016), showing G dominant inheritance (P < 0.001). In females, different genotypes at the rs1976667 locus were
significantly associated with genetic susceptibility to ISS (P = 0.011), showing G dominant inheritance (P = 0.005). Different genotypes at the rs2684788 locus, the G allele, and the G recessive mode of inheritance were all significantly associated with genetic susceptibility to ISS (P < 0.005). The genotypes at the rs1976667 locus in the female ISS group were significantly correlated to IGF-1 standard deviation integral value (SDS) (P = 0.006). The rs1976667 and rs2684788 loci of the human IGF-1R gene are likely associated with different genetic susceptibilities to ISS in males and females. Different clinical phenotypes of ISS may be associated with SNPs of IGF-1R.

**Keywords:** Idiopathic short stature; Single nucleotide polymorphisms; IGF-1 receptor; Genetic susceptibility