OBJECTIVES
Accumulated evidence indicates that the GH-IGF-1 pathway might be one of the crucial mechanisms of ISS. Insulin-like growth factor-1 receptor (IGF-1R) is the effector molecule that regulates the cascade reaction of hormone receptors in the GH-IGF-1 axis. To investigate the role of IGF-1R gene polymorphisms with regard to susceptibility to idiopathic short stature risk in the Chinese population of Jiangxi area.

METHODS
A total of 609 samples (ISS=295, control=314) from Jiangxi area were controls were enrolled in this study. The possible associations between 46 tag SNPs and progression risk among 295 patients were investigated using a two-step case-control study with a discovery cohort (n=100) and a validation cohort (n=195). SNPs (rs2684788) were genotyped using the SNaPshot Multiplex System.

RESULTS
We found that the rs2684788 in the IGF1R gene is associated with ISS in population of Jiangxi area among allelic model (G vs. A, OR=1.685, 95% CI=1.272-2.233, \( P < 0.001 \)), genotypes (GG vs GA vs AA, \( \chi^2 = 13.724 \), \( P < 0.001 \)), dominant model (GG+GA vs AA: OR=1.887, 95% CI=1.352-2.634, \( P < 0.001 \)). Notably, for individuals having the rs2684788 with the GG/GA genotype, the magnitude of increased ISS risk for lower IGF-1 SDS was significantly elevated (\( P < 0.004 \)).

CONCLUSIONS
The results suggested that the human IGF1R gene SNP rs2684788 might be associated with ISS genetic susceptibility in population of Jiangxi area, and might be associated with ISS clinical phenotype.