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The role of IGF-1R gene polymorphisms with regard to susceptibility to Idiopathic short stature risk in the Chinese population of Jiangxi area

YANG Yu[1], HUANG Hui[1],YU Zhen[1],WANG Wei[2],YANG Li[1],HUANG Wei[3],XIE Li-ling[1]
(1 Department of Endocrinology, Genetics and Metabolism，Jiangxi Provincial Children's Hospital,Nanchang 330006;2 Department of Pediatrics, Ruijin Hospital,Shanghai Jiaotong University School of Medicine, Shanghai;3 Key Laboratory of Health and Disease Genomics,Chinese National Human Genome Center at Shanghai, Shanghai)
Corresponding author:Yang Yu

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 v.s GA vs. AA,χ^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 riks for lower IGF-1SDS 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.

Tab.1 Armitage trend test for rs2684788 locus of the IGF-IR gene of ISS in Jiangxi

SNP locus	ISS group(n=295) N	%	Control group(n=314) N	%	χ^2	*P*	OR	95% CI	Boferroni correction
genotype									
GG	184	62.3	239	76.1	15.984	<0.001			
GA	33	11.1	31	9.8					
AA	78	26.4	44	14.0					
Allele									
G	200	67.8	254	80.8	13.724	<0.001	1.685	1.272,2.233	
A	95	77.5	60	72.0			1.00	1.00	<0.001
Dominant mode									
GG+GA	217	73.5	270	85.9	14.641	<0.001	1.887	1.352,2.634	<0.001
AA	78	26.5	44	14.0			1.00	1.00	

Tab 2 Correlation analysis of (GG+GA) genotype of G dominant mode at rs2684788 locus and clinical parameters in ISS group of Jiangxi Area

	GG+GA（n=217）	AA(n=78)	$\bar{x}\pm s$ (n=295)	*P*
HtSDS	-3.12±1.03	-3.08±0.77	-3.11±0.98	0.745
Weight SDS	-1.80±1.26	-1.56±1.28	-1.73±1.36	0.134
BMI	15.71±4.05	15.98±5.30	15.77±4.34	0.575
IGF-1SDS	-1.60±1.50	-0.98±1.80	-1.33±1.70	0.004
IGFBP3SDS	-1.91±1.24	-2.06±1.08	-1.99±1.16	0.145
THSDS	-0.80±0.81	-0.96±1.00	-0.87±0.92	0.252

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