



Relationship between vitamin D receptor (VDR) polymorphisms and the efficacy of recombinant human growth hormone (rhGH) treatment in children with idiopathic short stature

W. Wang^{1*}, X.P. Luo^{2*}, L.X. Cai^{1*}, Z.R. Cui¹, X.Y. Luo¹ and R.K. Luo³

¹Pediatric Endocrinology Clinic,
Third Affiliated Hospital of Zhengzhou University, Zhengzhou, Henan, China
²Department of Pediatrics, Tongji Hospital, Tongji Medical College,
Huazhong University of Science and Technology, Wuhan, Hubei, China
³Zhengzhou Foreign Language School, Zhengzhou, Henan, China

*These authors contributed equally to this study.
Corresponding author: W. Wang
E-mail: weiwang169@126.com

Genet. Mol. Res. 14 (3): 10507-10514 (2015)
Received February 3, 2015
Accepted May 18, 2015
Published September 8, 2015
DOI <http://dx.doi.org/10.4238/2015.September.8.12>

ABSTRACT. Polymorphisms in the vitamin D receptor (VDR) gene are associated with idiopathic short stature (ISS) in several countries. This study aimed to identify a possible correlation between polymorphisms in the VDR promoter in Chinese children with ISS and the efficacy of the recombinant human growth hormone (rhGH) treatment. Pre-pubertal children with ISS and healthy age- and gender-matched children (N = 95 each) were enrolled in this study. Two single nucleotide polymorphisms (SNPs) in the VDR promoter (rs11568820 at the Cdx-2-binding site upstream of exon 1e and rs4516035 at -1012 upstream of exon 1a) were typed. The growth velocity, standard

deviation score (SDS) of height for chronological age, height SDS for bone age, predicted adult height, and serum insulin-like growth factor 1 (IGF-1) and IGF-binding protein 3 (IGFBP-3) levels of the ISS patients were determined before and 6 months after rhGH treatment. No significant differences were observed in the genotype frequencies between the ISS cases and controls. After rhGH treatment, the growth velocity of the A/G genotype at the Cdx-2-binding site SNP locus was significantly higher than that of the G/G genotype; the IGF-1 and IGFBP-3 levels were also higher in the treated group than the untreated group. However, these changes were independent of the VDR-promoter genotype. Polymorphisms in the VDR promoter may not result in the pathogenesis of ISS in Chinese children. The A/G genotype showed a significantly higher growth velocity than the G/G genotype, and may represent a short-term marker of growth potential.

Key words: Idiopathic short stature; VDR; Insulin-like growth factor 1; rhGH; Insulin-like growth factor-binding protein 3