

### **POSTER PRESENTATION**

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# Influence of growth hormone receptor exon 3 polymorphism on growth response in children with growth hormone deficiency

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From 8th APPES Biennial Scientific Meeting

Darwin, Australia. 29 October – 1 November 2014

#### **Aims**

Pharmacogenetic effects of recombinant human growth hormone according to growth hormone receptor (GHR) exon 3 polymorphism (fl vs. d3) were controversial. We investigated growth hormone response in children with growth hormone deficiency (GHD).

#### **Methods**

Total 58 prepubertal children (31 boys and 27 girls) with GHD were enrolled in this study. Subjects were divided to 2 groups according polymorphism (fl/fl, n=48; fl/d3 and d3/d3, n=10), and compared baseline phenotypes and the first year growth response to growth hormone treatment.

#### Results

The distribution of GHR exon 3 isoforms in children with GHD demonstrated that the frequency of fl/fl (82.8%) is higher than that in most of European studies. There was no significant difference in baseline height SDS between 2 groups. Height velocity during the first year of growth hormone replacement therapy tended to be higher in subjects who have d3 allele (fl/d3 and d3/d3), but there was no statistical difference according to genotype.

#### Conclusion

It seemed that d3 allele of GHR exon 3 had no impact on the baseline phenotype and growth hormone response in patients with GHD. Relationship between GH dose and IGF-1% to help fully elucidate the value of IGF-1 testing in GH treatment.

Published: 28 April 2015

doi:10.1186/1687-9856-2015-S1-P34

Cite this article as: Yang and Hwang: Influence of growth hormone receptor exon 3 polymorphism on growth response in children with growth hormone deficiency. *International Journal of Pediatric Endocrinology* 2015 2015(Suppl 1):P34.

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