

## A Novel GH1 Functional Mutation in a Family with Isolated Growth Hormone Deficiency

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### Introduction

The familial type of isolated growth hormone deficiency (IGHD) is characterized by a variable degree of growth restriction and low but detectable GH serum concentrations. The following forms are recognized: recessive type IA and IB, autosomal-dominant type II and X-linked recessive type III. Phenotype-genotype correlations are notoriously difficult to be established. Herein, we described the variable clinical status of a family with a novel *GH1* mutation which likely lead to GH dysfunction.

### Case Report

The proband was an eight-year- and nine month-old boy who presented with short stature. His height was 108.5 cm

[-4.15 standard deviation score (SDS)] and his weight was 14.5 kg (-5.6 SDS), MPH was 164.9 (-1.8 SDS), bone age was six years. Interestingly, two GH stimulation tests had normal peak GH value of 12.6 ng/mL (with clonidine) and 12.1 ng/mL (with insulin). Other pituitary hormones and magnetic resonance imaging of the pituitary region were normal. The proband received recombinant human GH (rhGH) treatment (30 µg/kg/day) and he grew 5.1 cm in six months.

### Results

Sequencing of the *GH1* gene revealed a novel heterozygous mutation in the patient, his mother and his sister with severe short stature, but no phenotypic characteristics of GHD (p.Q110E).

### Conclusion

Establishing the genetic diagnosis of GHD is a challenge, but clinical feature exceptions have to be considered.