High Efficacy Growth Hormone Therapy in Patient with Homozygous Mutation in Growth Hormone Gene (*GH-1*) During 3 Years

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Background

Mutations in *GH-1* gene is a rare cause of isolated growth hormone deficiency. Main features of this condition include markedly reduced secretion of GH combined with low concentrations of IGF-I leading to short stature.

Clinical case:

1.56 years old girl with short stature

Height 59 cm (SDS: −7.25)
Weight 4.6 kg, BMI SDS −3.69

Closely related healthy parents

Failure to thrive and psycho-motor delay were noted

Birth length 48 cm (SDS: −1.07)

Birth weight 3670 g (SDS: 0.75)

Phenotype: prominent forehead, saddle nose and blue sclera

Laboratory testing revealed:

IGF-1 **3** ng/ml

Bone age: 8 month

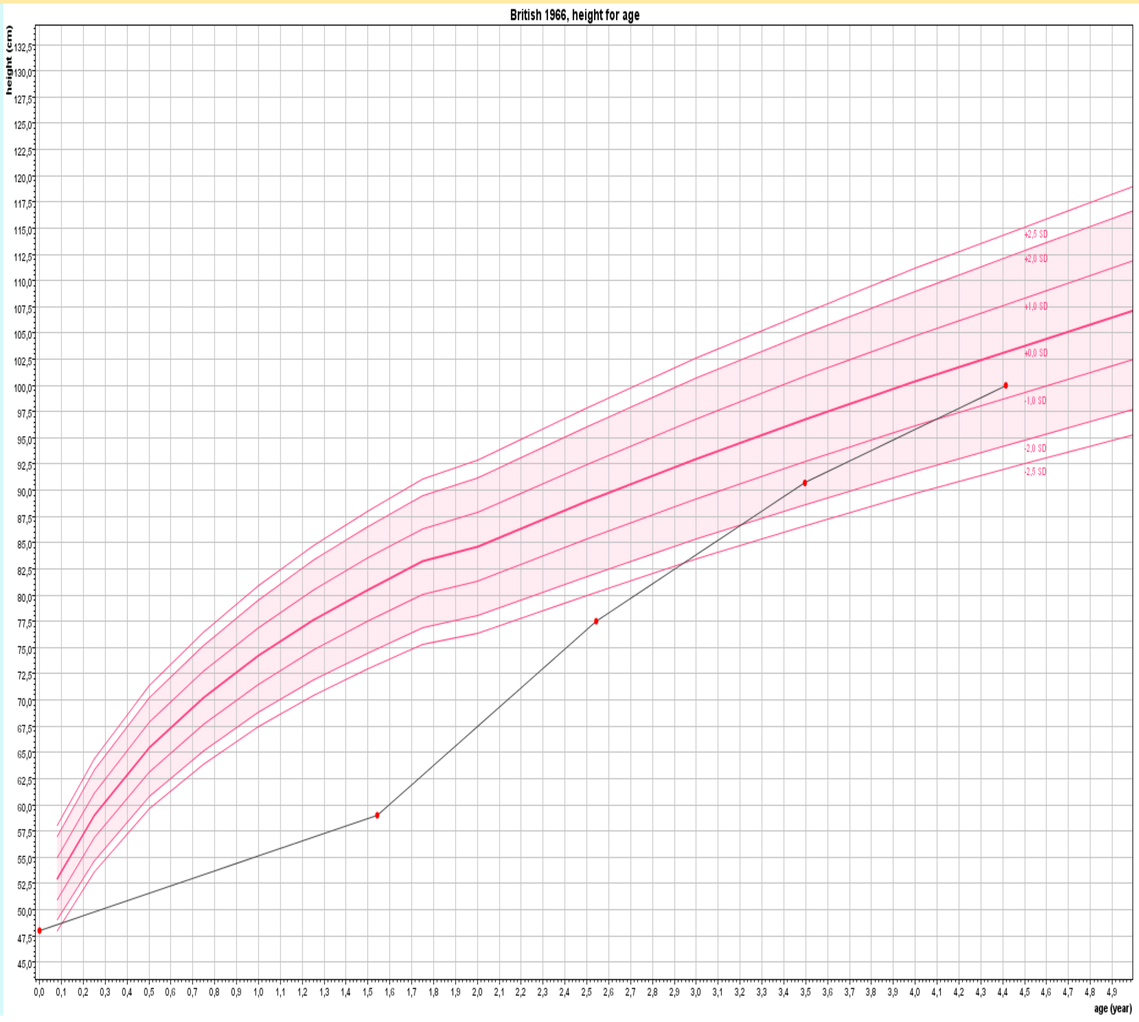
The karyotype is 46 XX

Cortisol, TSH, prolactine levels were normal

Genetic analysis revealed homozygous GH-1 gene deletions

The girl was started on GH therapy and protein rich diet

For the first 3 Years of GH therapy



Conclusion:

Mutations in *GH-1* gene is a rare cause of isolated growth hormone deficiency, which may present with extremely short stature and showing a good response to GH therapy.

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