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

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.