Homezygous loss-of-function mutations of the growth hormone receptor (GHR) gene result in GH insensitivity due to a dysfunctional receptor protein. Heterozygous mutations may result in a variable clinical spectrum ranging from normal height to severe short stature.

**CASE REPORT**

Age and gender

7.25 years-old male patient

Family history and previous medical history

Unknown (adopted child)

Mid-parental height and birth weight and length not available

Reason for referral

Faltering growth

Height -3.17 SDS (WHO growth charts) upon the time of consultation

**THERAPY**

Treatment with recombinant human GH (rhGH) was undertaken, with a daily starting dose of 0.028 mg/Kg. Excellent auxological response, with an overall height gain of 1.57 SDS in 24 months.

After the first year of treatment, IGF-I levels persistently above +2 SDS were detected, with the maximum value recorded being +10.64 SDS. Increased IGF-I levels were sequentially recorded subsequently, despite a stepwise down-titration to a daily dose of 0.015 mg/kg.

Treatment discontinuation. Timely normalization of IGF-I levels (-1.24 SDS).

**UNEXPLAINED RESPONSIVENESS TO rhGH**

Lab test

- Baseline IGF-I: -1.57 SDS
- Two pathological GH-stimulation tests (GH peaks: 4.7 and 3.4 ng/mL, respectively)

Radiology

- Remarkably delayed bone age (2.7 years versus 7.3 years)
- Brain MRI: pituitary hypoplasia with intrasellar arachnoid diverticulum

**IDIOPATICT GH DEFICIENCY**

**REFERENCES**


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