

Growth hormone treatment and puberty in patient with Pallister-Hall syndrome.

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BACKGROUND

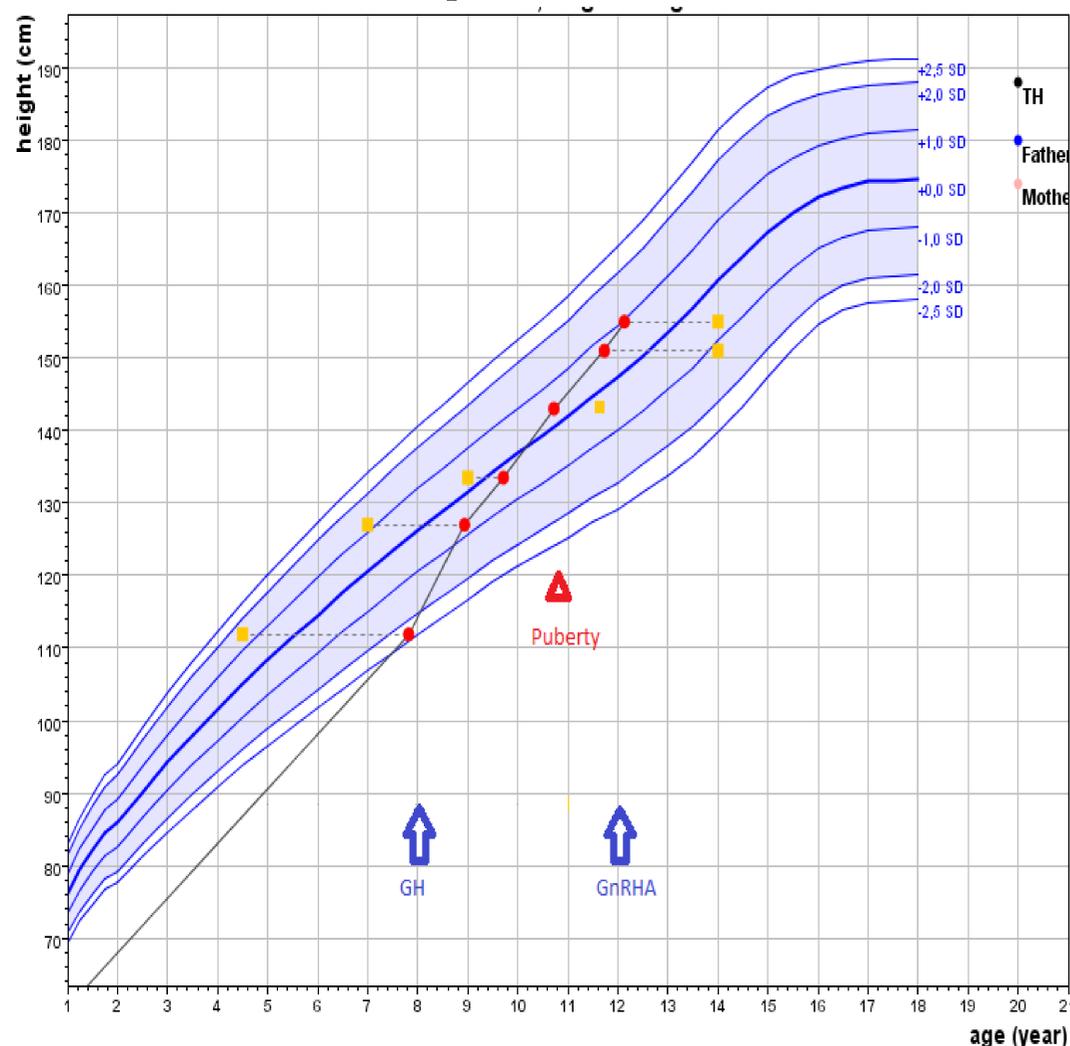
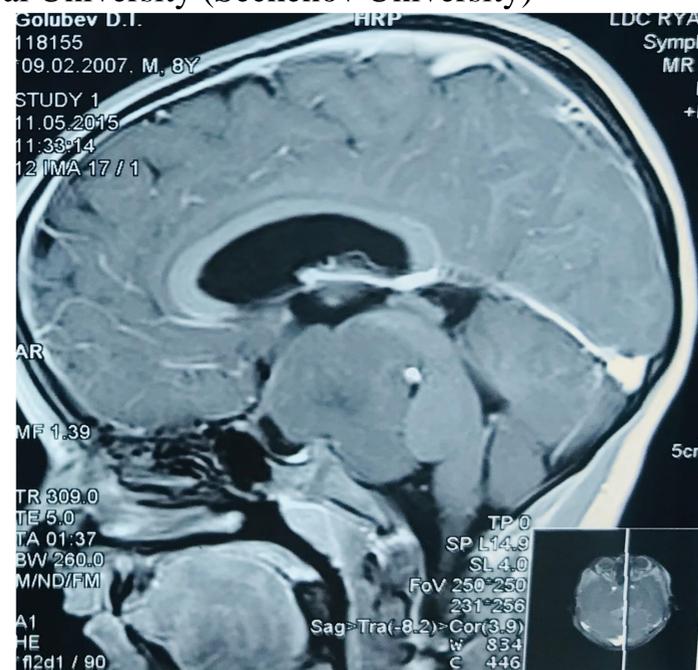
Pallister-Hall syndrome (PHS) is a rare autosomal dominant disorder characterized by a complex of different abnormalities (polydactyly, bifid epiglottis, hypothalamic hamartoma, imperforate anus). Syndrome is caused by mutations in the GLI3 gene. PHS is associated with hypopituitarism, early or precocious puberty.

OBJECTIVE

to demonstrate a patient with PHS treated with growth hormone (GH) and gonadotropin releasing hormone analogue (GnRHA).

RESULTS

Our patient is a 11 years old boy. His parental height is 174 cm mother, 180 cm father. He was born at 39 weeks of gestation, with birth length 53 cm, weight 3900 g, multiple abnormalities (imperforate anus, polydactyly, micropenis, cryptorchidism, bifid epiglottis). MRI of brain performed at the age of 7 years demonstrated hypothalamic mass 5,8*4,1*4,9 cm with signal density suggestive of hamartoma. Considering all components, PHS was suspected.



CONCLUSION

Children with PHS can have GH deficiency and GH therapy can achieve a good result. Precocious or early and rapidly progressing puberty can demand GnRHA therapy.

The first endocrine examination was at 7 years. His height was 111,9 cm (SDS -2,35). The bone age was 2,5 years delayed. IGF-1 was < 25 ng/ml. Thyroid and adrenal function were normal. The GH peak after clonidine stimulation was 3,8 ng/ml. The boy was treated with GH 0,033 mg/kg/day with good effect. At the age of 10 years he had a spontaneous puberty. His height was 143 cm (SDS +0,16). Tanner 2. Bone age was accelerated for 1 year. His hormonal status was LH 3,6 mIU/ml, FSH 2,3 mIU/ml, testosterone 4,4 nmol/l, IGF-1 264 ng/ml. At 11 years his height was 151 cm (SDS +0,58). Tanner 3. For 1 year his bone age was accelerated for 3 years. His hormonal status was LH 4,7 mIU/ml, FSH 1,7 mIU/ml, testosterone 26,7 nmol/l, IGF-1 202 ng/ml. To improve his final height GnRHA therapy was added.

Authors declare no conflicts of interests