RECOMBINANT GH TREATMENT IN CHILD WITH PSEUPSEUDOHYPOPARATHYROIDISM ASSOCIATED WITH GROWTH HORMONE DEFICIENCY

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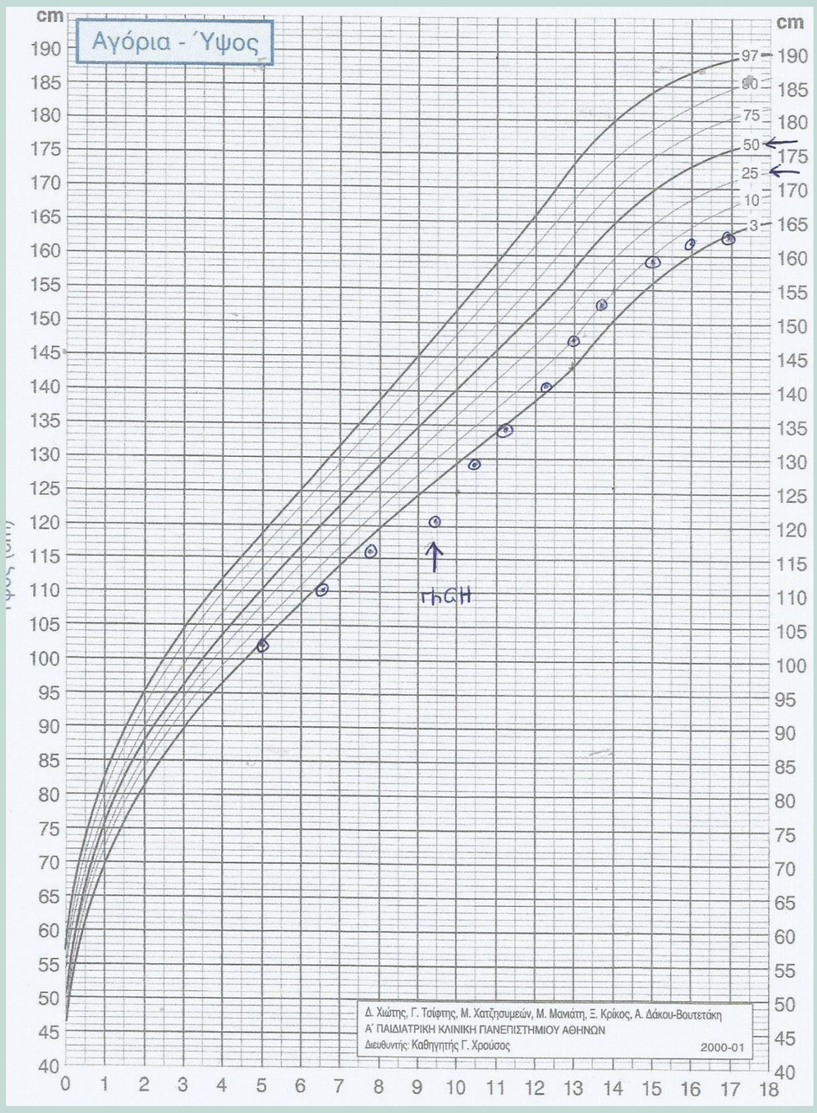
BACKGROUND

- Pseudopseudohypoparathyroidism (PPHP) is a rare variant of pseudohypoparathyroidism (PHP) type I, with typical anatomical abnormalities known as Albright's hereditary osteodystrophy (AHO)(short stature, brachydactyly particularly involving metacarpals and metatarsals, round face, stocky build,ectopic ossifications and a number of possible associated defects), but with normal calcium, phosphate and PTH levels and normal response to exogenous PTH.
- Short stature results from a combination of multiple factors including the premature fusion of growth plates and absence of growth spurt in addition to GH deficiency.
- We report the case of boy with PPHP and GHD, treated with rhGH to final height

CASE PRESENTATION

- A boy with morphologic features characteristic of AHO was followed in our Department because of severe growth retardation. He presented
  - with stocky build, mild obesity,
  - round face and short neck,
  - short pudgy hands and feet with shortening and thickening of third and fourth metacarpals and metatarsals, also demonstrated by radiological examination.
  - Serum and urine calcium and phosphate levels were normal as well as serum ALP, PTH and urinary cAMP concentrations.
- At the age of 9.5 years he was diagnosed with isolated growth hormone deficiency demonstrating an abnormal GH response in standard provocation tests (peak GH 3.9ng/dl) and low IGF-1 levels. Since then the patient has been treated with rhGH for 6 years at a replacement dose (0.18-0.2mg/kg/wk).

Even though in the beginning he presented a significant improvement of growth velocity (7.2 cm/y vs 3.5cm/y), the growth spurt during puberty was limited (22cm) As a result HSDS improved compared to pretreatment values (-1.93 vs -2.30), but final height reached the 3rd centile, far below the target height (50th centile)



DISCUSSION

To our knowledge this is the second case of a patient with PHPP and GHD treated with rhGH to final height.

Similarly to the case of patients with PHP, it seems that rhGH treatment has a significant effect on growth in prepubertal years.

Taking into account the limited clinically useful time window of effective treatment and the underlying skeletal disease, higher doses of rhGH should be considered in patients with PPHP