Case Presentation

SHORT HEIGHT DUE TO GROWTH HORMONE DEFICIENCY IN A SUBJECT WITH KLINEFELTER SYNDROME

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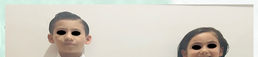
INTRODUCTION:

The clinical phenotype of Klinefelter syndrome described since 1942 by the American physician Dr. Harry Klinefelter. It describes men with tall stature, small testicles, gynecomastia and azoospermia. The precise etiology of supernumerary X chromosomes (47,XXY), the most common genotype identified in 1959. Extra Height at birth is normal, growth acceleration is most rapid in early and middle childhood, with greater than average parental height rarely extremely tall[1]. Very few cases of Klinefelter syndrome with short stature have been reported, most due to the presence of isochromosome Klinefelter; On the contrary, the presence of any associated sign (brain maturation, delayed puberty) requires a karyotype for proper diagnosis and management due to the association with GH deficiency, or the presence of some variant (isochromosome Xq, 49,XXXXY , etc.)[2].



CASE

Phenotype male patient, 9 years 8 months old, with the following history: healthy parents and brother, white family size 166 cm. Product of a normal evolutionary pregnancy, born by eutocic birth, 38.2 SDG, without complications, weight 3100 gr, height 50 cm. Fed with breast milk for 6 months, adequate neurodevelopment. History of recurrent respiratory symptoms requiring inhaled treatment. Allergies and surgeries denied. At 9 months of age, in a pediatric consultation, growth deceleration, low weight and height were identified. Therefore, he was evaluated by Pediatric Endocrinology, for the first time, at 13 months of age: weight -3.68 z, height -3.67 z. head circumference -1.9 z. lower segment 26.5 cm, segment ratio 1.52, stroke 65 cm, length/stroke ratio -2, clinodactyly, no facial dysmorphisms, both testicles descended. Newborn bone age. Laboratory tests initial assessment: Hb 9.8 mg/dl, EGO: density 1015, ph 7.0. Normal thyroid profile, IGF-1 132.4ng/ml. +2DS, normal venous gasometry. Growth speed monitoring began, with persistence of height below p3, it is evaluated by Genetics; At 2 years and 1 month of age, a karyotype was performed with GTG bands, results of 47 XXY. At 4 years 6 months, a growth hormone stimulation test (insulin-induced hypoglycemia) is performed: basal glucose: 87 mg/dl, GH 0.36 ng/ml, 15 min 34mg/dl - 0.34 ng/ml, 30 min 31 mg/dl – 4.0 ng/ml, 45 min 51mg/dl – 7.49 ng/dl respectively.

Partial growth hormone deficiency is confirmed; in February 2018, treatment was started with somatropin 0.16 mg/kg/week up to 0.3 mg/day (0.02 mg/kg/day), adjusting according to his weight.

9 years 8 months old; Body composition: fat mass 13%, 2.9 kg and lean mass 19.2 kg.
10 years 4 months old, Tanner pubic 1, testicles: right 4 cc, left 3 cc. Body composition fat mass 13.8% (3.4 kg), lean mass 21.6 kg. laboratory tests: Glucose 95.2, mg/dl, hbA1c 5.1%. IGF 1 245.8. ng/L. IGFBP3 5.2 mg/L, FSH 1.65 IU/L, LH <0.20 IU/L. 0.1 ng/ml, total testosterone 0.1ng/dl.
10 years 7 months old: fat mass 13.4% (3.4 kg) lean mass 21.9 kg

Image 1. Growth curve.

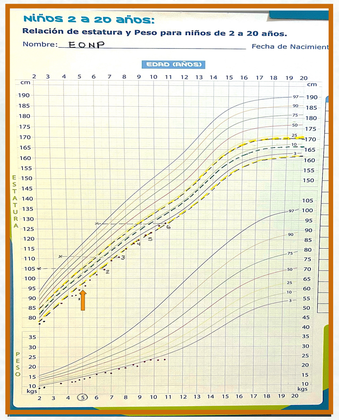


Table 1: Evolution of treatment with growth hormone.

GH treatment time. (years)	Chronological age (years)	Dose GH mg/kg/day	Growth speed (cm/year)	Bone age years
1	5 years	0.03	8.1 (p97)	
2	6 years 7 m	0.024	5 (p10)	2
3	7 years 7m	0.032	3.9 (p3)	3.5
5	9 years 8 m	0.039	4.4 (p25)	
6	10 years 7 m	0.036	5.3 (p50)	6

DISCUSSION:

The short stature in this case was studied according to protocols, however within the genetic study a 47XXY karyotype was found, a rare clinical presentation. Treatment with growth hormone was started and follow-up has been carried out to date.

CONCLUSION:

An unusual relationship between Klinefelter Syndrome and growth hormone deficiency has been reported; in our case it was confirmed by karyotype and had a favorable response with growth hormone treatment.

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