A CASE OF KLINEFELTER SYNDROME WITH AN ATYPICAL PRESENTATION

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Klinefelter syndrome, also known as 47 XXY, is a disorder characterized by tall stature, hypogonadism and mental retardation which is caused by nondisjunction events during meiosis and occurs in 500-1000 live male birth. Here we report a patient with Klinefelter syndrome who presented with short stature, in contrast to common tall stature presentation and was diagnosed with growth hormone deficiency.

Case:

A 7-year-old male presented with short stature. He was born to an advanced aged mother with a birth weight of 1430 g at 33 weeks gestation. His height was 104.7 cm (-3.7 SD) and weight 17 kg (-1.5 SD). Anthropometric measurements were otherwise normal. He was prepubertal and had no dysmorphic findings on physical examination. His growth rate was 4 cm/year during the previous year. On laboratory investigations; total blood count, blood chemistry, and thyroid function tests were within normal limits. Autoantibodies for celiac disease were negative. Serum IGF-1 was 101 ng/ml (2.5-25p), and IGFBP-3 was 1480 ng/ml (<2.5 p). Karyotype analysis revealed a 47 XXY genotype. Bone age was 4 years and pituitary MRI was normal. Growth hormone stimulation tests using clonidin and L-dopa revealed peak GH responses of 3.5 ng/ml and 3.92 ng/ml respectively. He achieved 10 cm growth during the first year of growth hormone treatment.

Conclusion:

Although tall stature is a well-known feature of Klinefelter syndrome, patients may present with short stature and can be diagnosed with growth hormone deficiency.

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