Introduction

- 47,XYY syndrome is characterized by an extra copy of the Y chromosome in each cell of a male. It occurs in about 1/1,000 of newborn boys and most males with this syndrome have normal sexual development and fertility. They tend to have tall stature and mild motor and language developmental problems. Testosterone levels are normal. Increased rate of criminal activity in XYY males was related to a lack of judgement and lower socioeconomic status due to a lower mean IQ score.

Case

- A 21-year-old man applied to our clinic with complaints of small testis and penis and lack of beard and ejaculation. He had normal libido and erection. There was no family history of infertility. He hadn’t committed any crime. In physical examination, he had eunuchoid habitus, his height was 178 cm, weight was 66.6 kg, and BMI was 21 kg/m2. His testes were palpable in the scrotum. Axillary and pubic hair development was consistent with Tanner stage 4. Penis length was 6.5 cm. His neurocognitive development and functions were normal. Hormonal tests revealed hypogonadotropic hypogonadism. Other hypophyseal hormones were normal. Bone age was compatible with 14 years and epiphyseal plates were open. In testicular ultrasonography, volumes were 8 ml on the right and 7.5 ml on the left. Hypophyseal MRI showed partially empty sella. In chromosomal analysis, 47,XYY karyotype was detected. After administration of human chorionic gonadotropin treatment, androgen levels increased and ejaculation started although in small amounts (<0.5 ml).

Conclusion

- Men with 47,XYY syndrome have a diverse spectrum of clinical presentation and because of the heterogeneous phenotype and lack of specific symptoms, its diagnosis may be difficult. As presented in our case, hypogonadotropic hypogonadism might be a presenting feature in patients with 47,XYY genotype.

References