Letter to Editor

46XX Testicular Disorder of Sex Development

Dear Editor,

We would like to add to the sparse literature on 46XX testicular disorder of sexual development due to the sex-determining region of Y-chromosome (SRY) translocation, wherein there are only 38 papers thus far, including the one reported by Dr. Mantravadi and Dr. Rao.[1] These patients typically seek medical attention for primary infertility or delayed puberty, while a few present in the newborn period with genital ambiguity or hypospadias.[2,3] Although the varying degree of gynaecomastia occurs in around 10%–30% of patients,[2,3] gynaecomastia as presenting complaint has not been seen in any of the reports. A 26-year-old man had presented to our clinic with the only complaint of gynaecomastia of 10 years. He had sparse facial hair growth, with a sexual maturity rating of grade three breasts bilaterally (B3), Tanner stage five pubic hair (P5), the testicular volume of 2 ml bilaterally, and stretch penile length of 9 cm. He had no issues with spontaneous erections or libido. The hormonal evaluation was suggestive of hypergonadotrophic hypogonadism, with karyotype analysis demonstrating 46XX chromosomes, and fluorescent *in situ* hybridisation confirming SRY gene translocation to the short arm of chromosome X (Xp). Low-volume azoospermia was reported on semen analysis. The patient was initiated on testosterone replacement therapy, and reduction mammoplasty was advised for gynaecomastia.

Dr. Mantravadi and Dr. Rao have rightly emphasised the need for multi-speciality approach, including infertility expert, urologist, endocrinologist, psychiatrist and geneticist.[1] We would like to add that testosterone replacement therapy is necessary to correct hypogonadism, and for the long-term physical and sexual well-being of the patient. Furthermore, these patients are at an increased risk of gonadoblastoma;[4] therefore, regular self-examination and ultrasound of testes should be encouraged.

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Conflicts of interest

There are no conflicts of interest.

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