

What is your diagnosis?

A 29-year-old well-adjusted male presented with complaints of cyclical hematuria and pain for 6 months and a loss of libido and erectile dysfunction for 2 years. Although he had a predominantly male phenotype as evidenced by the male body habitus and normal virilization, he had gynecomastia. Hormonal analysis revealed low levels of serum testosterone (170 ng/dL) and elevated levels of luteinizing hormone (13.1 mIU/mL), follicle-stimulating hormone (FSH) (12.3 mIU/mL) and estradiol (85.5 pg/mL). On ultrasound (Figure 1a, b) and magnetic resonance imaging (MRI), an undescended testis was observed in the left inguinal canal. The right testis was not visualized, but an ovary with multiple follicles was observed on the right side. A rudimentary blood-filled uterus was observed in the right hemipelvis alongside the right ovary with abnormal communication with the prostatic urethra (Figure 2a, 2b). Intravenous pyelogram was normal. Chromosomal analysis revealed a 46, XX karyotype.

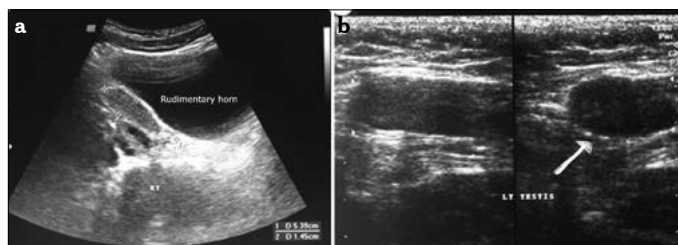


Figure 1. a, b. Ultrasound image showing a uterus-like structure with an ovary on the right side (a); ultrasound image showing testis in the inguinal region on the left side (b)

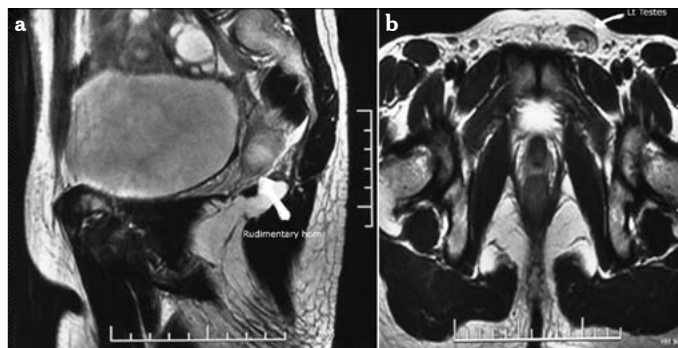


Figure 2. a, b. T2-weighted MRI images showing a rudimentary blood-filled horn with ovary on the right side (a); T2-weighted MRI images showing testis on the left side in the inguinal region (b)



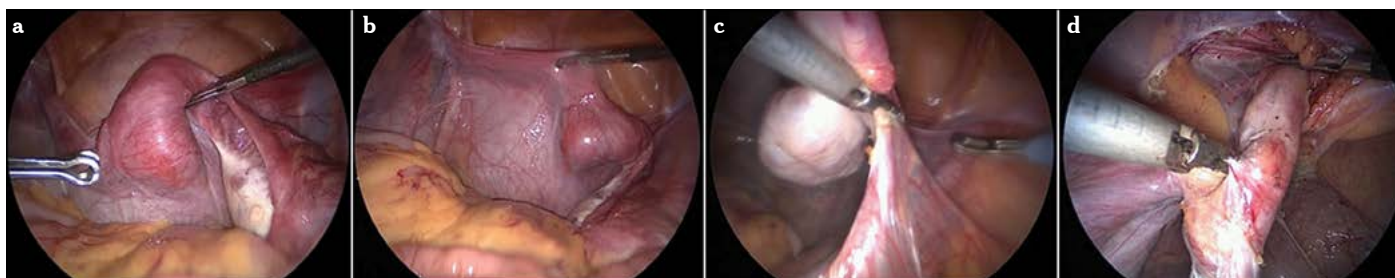


Figure 3. a-d. Laparoscopic findings showing rudimentary horn with the right fallopian tube and ovary (a); laparoscopic findings showing left testis in the inguinal canal (b), right salpingo-ovariectomy (c), and left orchidectomy (d)

Answer

Laparoscopy revealed an ovary and a rudimentary uterine horn on the right side and inguinal testes on the left side (Figure 3a, 3b). The patient underwent laparoscopic rudimentary uterine horn excision with right salpingo-ovariectomy (Figure 3c) and left orchidectomy (Figure 3d). Histopathological examination revealed a small uterus with an ovary on one side with normal ovarian parenchyma and testis on the other with testicular parenchyma with marked tubular atrophy and hyalinization with Leydig cell hyperplasia.

Ovotesticular disorder of sex development (ovotesticular DSD) (earlier known as true hermaphroditism) is a very rare disorder in which an infant is born with the internal reproductive organs (gonads) of both sexes (female ovaries and male testes) (1, 2). The gonads can be of any combination, ovary, testes, or combined ovary and testes (ovotestes). The external genitalia can either be ambiguous or can range from normal male to normal female. Here we describe a case of ovotesticular DSD who was raised as a male and who presented in adulthood because of failing testicular function.

In true hermaphrodites, it is rare to witness a well-adjusted adult male presenting so late with hematuria. In this case, gynecomastia at puberty is indicative of a functional ovary. The male external genitalia, sexual orientation, and intact erectile function till the early twenties are suggestive of a well-functioning testis. Testicular failure due to dysgenetic changes has been reported in true hermaphrodites (3). In our case, there is a possibility of failing testicular function and a dominance of ovarian function resulting in loss of libido and initiation of menstruation. Most of the reported cases have ambiguous genitalia with a uterus, cervix, or vagina; however, complete male phenotype is scarcely observed in patients with ovotesticular DSD. In one of the reports by Dutta et al. (4), a 15-year-old phenotypic male presented with three episodes of cyclical hematuria and biochemically normal serum testosterone levels with elevated gonadotropins levels. In another report (5), an 18-year-old male patient presented with bilateral scrotal pain with scrotal swelling and bilateral gynecomastia and a 46, XX karyotype.

Because of the rarity of the condition, the most appropriate management remains uncertain. In our case, the patient was raised as a male and presented unusually late at the age of 29 years. The decision for bilateral gonadectomy was taken in view of subnormal testicular function as evidenced by low testosterone and raised gonadotropin levels. This was later confirmed by the histopathology of the testes. Furthermore, the patient was very apprehensive regarding even 4%–5% risk of malignancy in the inguinal testes and preferred to have it removed.

Kandala Aparna Sharma¹, Vatsla Dadhwal¹, Ashish Kumar Saini², Sumita Agarwal¹, Shobha Kandpal¹

¹Department of Obstetrics and Gynecology, All India Institute of Medical Sciences, New Delhi, India

²Department of Urology, All India Institute of Medical Sciences, New Delhi, India

References

1. Krob G, Braun A, Kuhnle U. True hermaphroditism: geographical distribution, clinical findings, chromosomes and gonadal histology. *Eur J Pediatr* 1994; 153: 2-10. [\[CrossRef\]](#)
2. Yordam N, Alikasifoglu A, Kandemir N, Caglar M, Balci S. True hermaphroditism: clinical features, genetic variants and gonadal histology. *J Pediatr Endocrinol Metab* 2001; 149: 421-7. [\[CrossRef\]](#)
3. Blackless M, Charuvastra A, Derrtyck A, Fausto-Sterling A, Lauzanne KLee E. How sexually dimorphic are we? Review and synthesis. *Am J Hum Biol* 2000; 12: 151-66. [\[CrossRef\]](#)
4. Dutta D, Shivaprasad KS, Das RN, Ghosh S, Chatterjee U, Chowdhury S, Dasgupta R. Ovotesticular disorder of sexual development due to 47, XYY/46,XY/45,X mixed gonadal dysgenesis in a phenotypic male presenting as cyclical haematuria: clinical presentation and assessment of long-term outcomes. *Andrologia* 2014; 46: 191-3. [\[CrossRef\]](#)
5. Irkilata HC, Basal S, Taslipinar A, Gur A, Piskin MM, Kurt B, et al. Ovotesticular disorder of sex development with a prostatic gland and review of literature. *Andrologia* 2009; 41: 387-91. [\[CrossRef\]](#)