

# 46 XY Ovotesticular Disorder: A Rare Case Report with Review of Literature

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## Abstract

Ovotesticular disorder represents 10% of cases of disorder of sex development characterized by the presence of both ovarian and testicular tissue in the same individual, with karyotype 46 XY being a rare sex chromosomal abnormality. We report the case of a 16-year-old person, who is reared as female, with a complaint of primary amenorrhea along with lack of secondary sexual characteristics, karyotype 46 XY. Prophylactic bilateral gonadectomy was done, and histopathological examination of bilateral gonads revealed ovarian stroma with a few Sertoli cell line tubules suggestive of bilateral ovotestis; hence, we concluded and framed our diagnosis of ovotesticular disorder.

**Keywords:** 46 XY karyotype, ovotesticular disorder, ovotestis

## INTRODUCTION

Disorder of sex development (DSD) is a congenital condition with an incidence rate of 1/4500 live births, and it comprises the defect in chromosomal, gonadal, and anatomical sex development.<sup>[1]</sup> This leads to defects in the external, internal genitalia and defect in pubertal development. Ovotesticular DSD has an incidence of <10% of all DSDs and the overall incidence of 1/100,000 live births.<sup>[1]</sup> In this disorder, both ovarian and testicular tissue are present in the same person.<sup>[2]</sup> The external genitalia is usually ambiguous but can range from normal male to normal female. Out of total diagnosed ovotesticular disorder cases, 46 XX chromosome is seen in 59.5% of cases, and 12.3% have 46 XY chromosome.<sup>[3]</sup> Laparoscopy or laparotomy with gonadal biopsy/gonadectomy allows for histologic confirmation of both ovarian and testicular tissue. These patients have increased the risk to develop germ cell tumor in the future, because of the presence of a nonfunctional testicular component in ovotestis, so removal

of nonfunctional gonads is required. A multidisciplinary approach is emphasized to maximize the potential of these individuals to develop into normal functioning social adults.

## CASE REPORT

A 16-year-old person, brought up as a female, presented to our outpatient department, with complaints of primary amenorrhea along with lack of secondary sexual characteristics development. Her medical and surgical history was unremarkable. No history of pubertal delay was seen in her mother and she was born out of a nonconsanguineous marriage. On physical examination, built and nourishment was average, height 170.18 cm, weight is 61.5 kg and body mass index of 21.2, and normotensive of her age group. The patient was found to have the ill-developed breast of Tanner Stage II and sparse axillary hair and pubic hair of Tanner Stage II. On examination, per abdomen was soft, no organomegaly, no mass at inguinal region noted. The vulvar region was well developed including normal development

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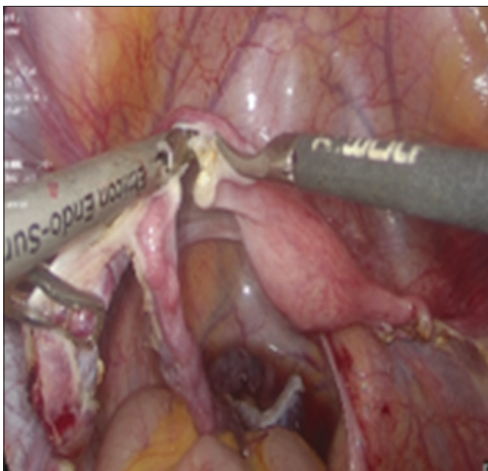
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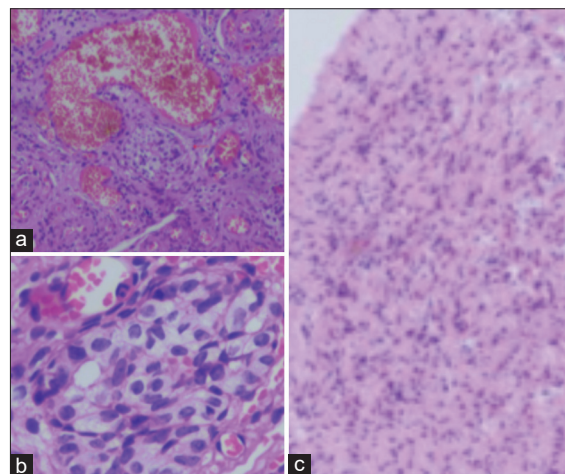
of labia majora, labia minora, clitoris and external opening of urethra and vagina, vaginal introitus was 1.5 cm, vaginal length measured 3 cm. No Turner stigmata were noted. Routine biochemical investigations and hormone levels were assessed. With raised follicle-stimulating hormone level of 128.53 mIU/ml, luteinizing hormone: 37.43 IU/L, and low anti-Mullerian hormone: 0.46 ng/ml. Radiological investigation: Ultrasonography suggestive of small uterus with a thin endometrium. Bilateral ovaries are small in size. Magnetic resonance imaging (MRI) of the pelvis suggestive of perineal vaginal atresia/hypoplasia with poorly visualized fallopian tubes and ovaries, uterine remnant visualized, endometrium atrophic. Karyotype report shows pattern 46 XY normal male chromosome. Phenotype female. Findings are consistent with sex reversal disorder such as gonadal dysgenesis. We reached a diagnosis of that this 16-year-old person who reared as female is having gonadal dysgenesis. The patient was put on conjugated estrogen therapy for initial 3 months on the dosage of 0.325 mg tablet once a day and then shifted to 0.625 mg once a day regimen. Significant secondary sexual characteristic development was noted in 1 year of initiating the therapy, with breast developed to Tanner Stage III, pubic hair Tanner Stage IV, and axillary hair development noted. The decision for the removal of streak gonads was planned, and laparoscopic prophylactic bilateral gonadectomy was done. Intraoperatively, the uterus was found to be normal in size and morphology, fallopian tubes were found well developed. On both sides, streak gonads were noted and removed laparoscopically [Figure 1]. Histopathological examination (HPE) revealed sections from bilateral streak gonads showing fibrous ovarian stroma without any primordial follicles. Several Sertoli cell line tubules were noted [Figure 2]. The patient was administered with estrogen therapy postoperatively and planned for psychological counseling and lifelong hormonal therapy.



**Figure 1:** Laparoscopic view of the left gonad and normal looking uterus

## DISCUSSION

This case of a 16-year-old person who reared as a female having hormone levels suggestive of hypergonadotropic hypogonadism with radiological findings suggestive of anatomical disorder and karyotype suggestive of a 46 XY chromosome framed our diagnosis that it is a case of DSD. As the patient is brought up as a female, a combined decision involving the patient party and gynecologist was made to remove the nonfunctional gonads and assign female sex (phenotype) to her. HPE of the gonadal tissue revealed it to have both ill-developed testicular tissue (Sertoli cell line tissue) and ovarian stroma, favored it to be a case of ovotesticular disorder. However, the karyotype of 46 XY is difficult to distinguish between mixed gonadal dysgenesis and ovotesticular disorder. It is differentiated from it as only testicular (seminiferous tubules) tissue is noted on HPE.<sup>[4]</sup> For the development of normal testis, SRY gene activation is primarily required for further development of the primordial gonads. The ovarian testicular disorder is generally caused by mosaicism, chimerism, and single-gene mutation.<sup>[5]</sup> In this, both ovarian and testicular tissue, also called ovotestis, will be present in the same individual. In most of the reported cases of ovotesticular disorder, ambiguous genitalia is seen, varying from almost normal female with or without clitoromegaly to normal male external genitalia.<sup>[6]</sup> These patients have increased risk to develop germ cell tumor in the future, because of the presence of testicular component in ovotestis, so the removal of nonfunctional gonads is required. In ovotesticular disorder with 46 XX chromosome carries a risk of malignancy (3%), and is higher (25%) in cases of 46 XY chromosome.<sup>[5]</sup> Many germ cell tumors have been reported till date such as gonadoblastoma, seminoma, dysgerminoma, cystadenoma, Sertoli cell tumor, and teratoma.<sup>[6]</sup> Such



**Figure 2:** (a) H3836/2018, 40X, Sertoli cell line tubules. (b) H3836/2018 10X Sertoli cell line tubules (c) H3836/2018, 10X, unremarkable ovarian stroma, no definite follicles seen

patients of ovotesticular disorder require a multidisciplinary approach of management, including psychological counseling and lifelong hormonal therapy, with fertility approaches and functional improvement.

## CONCLUSION

Ovotesticular disorder is diagnosed with a combination of tests including chromosome and genetic analysis, hormone testing, ultrasound or MRI, and gonadal biopsy. Radiological investigations are helpful in anatomical disorder, but confirmatory diagnosis for it is based on histopathological appearance, to differentiate it from mixed gonadal dysgenesis and other DSDs. Considering the potential of fertility depending on the degree of gonadal differentiation and genital development for a preferred gender, the decision for particular gender rearing should be taken.<sup>[7]</sup> Looking at the future risk of gonadal malignancies, prophylactic gonadectomy of dysgenetic gonads was suggested. Such patients are managed by a multidisciplinary approach, including psychological counseling and lifelong hormonal therapy, with fertility approaches and functional improvement.

## Ethical statement

This study was exempted by Independent Ethics Committee of All India Institute of Medical Sciences, reference number: AIIMS/RES/2019/4149.

## Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the legal guardian has given the consent for images and other clinical information

to be reported in the journal. The guardian understands that the patient's name and initials will not be published, and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

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Nil.

## Conflicts of interest

There are no conflicts of interest.

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