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Case Report A Rare Case of 46, XX Ovotesticular Disorder of Sex Development (DSD)

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ABSTRACT

Background: Ovotesticular disorder of sex development (ovotesticular DSD) is a very rare condition in which an individual is born with internal gonads of both sexes, such as ovaries, testes, or ovotestes. **Objective:** To report a rare case of 46,XX/46,XY ovotesticular DSD in a 16-year-old adolescent presenting with menstruation through the urinary meatus. **Method:** Case report including history taking, physical examination, karyotype analysis, and multidisciplinary management involving urology, gynecology, endocrinology, pediatrics, and psychiatry. **Result:** The patient presented with a penis, undescended left testis, and a perineal opening resembling a vagina. Until the age of 14, the patient was raised as a boy. At puberty, breast development and pubic hair appeared, followed by irregular menstruation and painful hematuria. Physical examination revealed normal sexual hair distribution, incomplete vaginal structure beneath the scrotum, and hypospadias. Gonads were located on the right labioscrotal fold. Karyotype analysis showed 46,XY (85%) / 46,XX (15%). A multidisciplinary team provided medical, surgical, and psychosocial care. **Conclusion:** Gender assignment as male was made by a multidisciplinary team after evaluating gender identity and role, and considering the patient's own wishes. Comprehensive multidisciplinary management is crucial for all DSD patients.

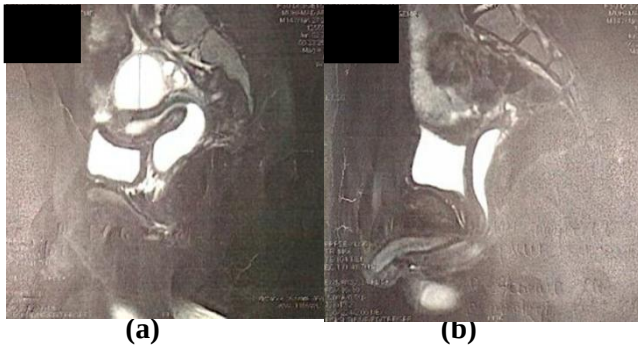
BACKGROUND

Ovotesticular disorder of sex development (OT-DSD) means the simultaneous presence in the same individual of both, histologically proven, testis and ovary. The gonads can be any combination of ovary, testes or combined ovary and testes (ovotestes). The external genitalia are usually ambiguous but can range from normal male to normal female. Most cases have a sporadic distribution, although there are a few documented cases of familial recurrence. Ovotesticular DSD is the rarest disorder of sex development in humans and has an approximate incidence of less than 1/20,000. The disorder may account for less than 3-10% of all DSD. At least 500 affected individuals have been reported.[1-4] *Matsui* reports that the most frequent gonadal combination is ovotestis-ovary (33.9%). The gonadal combination of ovotestis-streak gonad occurs in only 1.2% of the case. Gonadal tumours occur between 2.6 and 4.6% of OT-DSD, more frequently in 46, XY cases.[5]

CASE PRESENTATION

The patient is a 16 years old teenager who complains of bleeding through the urinary meatus every menstruation. He is the 3rd child of 3 siblings, 2 older brothers are normal (31 and 29 years). The patient had a penis, left sided undescended testis since birth, and a hole below the scrotum which looked like a vagina. For more than 14 years he was reared as a boy, with no further complaints, until he reached puberty. The breasts began to enlarge, pubic hair began to grow a year later, accompanied by irregular menstruation. and monthly painful hematuria. The patient was treated on 23 October 2019 for a urethral abscess, a cystostomy operation was performed.

From physical examination, sexual hair distribution was on normal parts of the body. He had an incomplete abnormal vagina on his scrotum. Menstruation blood came from this vagina. Presents with abdominal pain, appears attached cystostomy, clear yellow urine production. On the penis, MUE appears on the penoscrotal. Hypospadias was encountered. He had a normal scrotum which had a labioscrotal fold. Gonads were found on the right side of the labioscrotal folds. Karyotype was 46 XY (Y chromosome is very small, will be confirmed with C-banding): Change to be 46 XY (85%)/46 XX (15%)



(a) **(b)**

Figure 1. Sagittal view of pre-operative magnetic resonance imaging (June 2017), revealing (a) right testis and male urethra connecting the urinary bladder to the penis; (b) a vagina and anteflexed uterus with fluid retention.

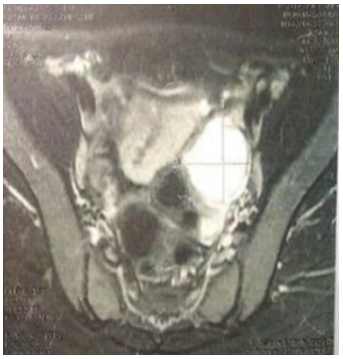


Figure 2. Coronary view of pre-operative magnetic resonance imaging (June 2017), revealing left ovary cyst.

Magnetic resonance imaging showed vaginal and uterine formation with an endometrial line in an anteflexed position. Fluid retention in the vagina was observed. A left single ovary was seen, which measured 3.7x3.6x3.4 cm. Urethroscopy revealed a urethra opening located sub-coronally with rough mucosa and a narrow penoscrotal fistula.

Laparoscopy done by an OBGYN revealed that the uterus and left adnexa was attached to the left pelvic side wall. Identification of the internal reproductive organs: visible ovary and left tube with left hemi uterine. Not found right internal genitalia. The uterus is the size of a chicken egg, adhesions to the left abdominal wall and sigmoid. The ovaries were about 3 cm, there are also ovulation spots on the ovary.

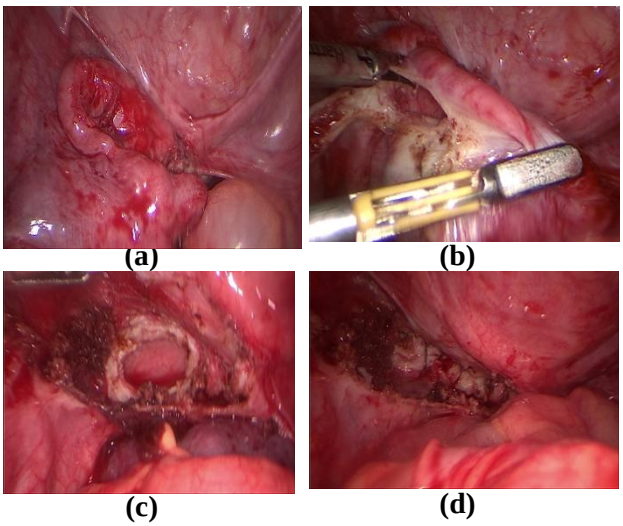


Figure 3. No testicles visible (a). Adhesiolysis left adnexal from the pelvic side wall, and because the patient has decided to be male, we perform to do left salpingooophorectomy and totally hysterectomy (b). After hysterectomy, when the vaginal stump is opened, the impression of the right vaginal wall fuses with bladder wall (c). Performed vaginal removal, stump and bladder wall (cystotomy) was closed in 2 layers with a barbed suture (d). Anatomical pathologic examination revealed normal uterus, tube and ovary, there was no sign of malignancy.

DISCUSSION

46,XX ovotesticular DSD commonly refers to hermaphroditism. The term hermaphrodite is derived from the Greek mythological god Hermaphroditos, son of Hermes and Aphrodite, whose body after being merged with that of the nymph Salmacis assumed a perfect form with both male and female attributes.[1] The causes of true hermaphroditism remain enigmatic, and the most common presentation is abnormal external genitalia ranging from normal male to normal female. The sex chromosomal mosaicisms that have been reported in 33% of OT-DSD cases include : 46, XX/46, XY; 46, XY/47, XXY; 45, X0/46, XY; 46, XX/45, X0; and 46, XX/47, XXY.[6] Successful fertility has been reported in an OT-DSD individual with a male-predominant mosaic karyotype.[2,4,7,14] Proper gender assignment to a neonate born with DSD is a social emergency of the newborn period. Infants and children born with DSD pose a diagnostic and therapeutic challenge to clinicians. Success depends upon rapid and precise diagnosis, appropriate gender assignment, proper medical therapy, and meticulous surgical technique. [1,3,11,15]

According to potential fertility, the medical and surgical decisions could be difficult in 46 XY OT-DSD. It seems that males report greater satisfaction with their gender and sexual lives.[9,16] In OT-DSD, the gonad may contain ovarian and testicular tissue. The developing ovary may lead to a functional ovarian tissue which causes menstruation in 50% of cases of OT-DSD.5 The production of ovarian steroids can also suppress expression of gonadotropins in testicular tissue via a negative feedback effect, resulting in tubular atrophy, poor germ cell development, Leydig cell

hyperplasia, and sclerosis that finally causes degeneration of the testicular tissue.[15] As the testosterone production may decline to inadequate levels because of the testicular regression, the consideration of substitutive hormonal treatment should be initiated.[14,15]

Conservative treatment is the goal of the multidisciplinary treatment of DSD, especially when the sex assignment is required during neonatal period.[5,12,16] The aims of surgery are to restore functional genital anatomy, facilitate future reproduction, reduce urological hazards related to abnormal genitourinary anatomy, avoid fluid or blood retention in vaginal or uterine cavities, avoid late virilization at puberty in individual raised as girls or breast development in individuals raised as boys, reduce risk of gonadal cancers, foster development of *individual* and *social identities*, avoid stigmatization related to atypical anatomy, and respond to the parent's desire to bring up a child in the best possible conditions. [3,11-13]

The timing of surgery remains controversial.[11-14,16] Our surgical interventions will not preclude the future possibility of a female-to male genitoplasty. The female intraabdominal gonad macroscopically looked like a streak gonad, despite the histological presence of follicles. No evidence exists about prophylactic removal of asymptomatic discordant structures, such as *Müllarian* remnant.[1] A clinical surveillance is mandatory. Annual ultrasound surveillance is recommended for gonadal cancer risk even when the gonad is in the scrotum. If precancerous lesions are suspected, more invasive procedures (MRI, serum tumor markers, biopsy) are mandatory. This patient is apparently and expectedly happy to remain male. It is therefore planned to remove ovary and uterus on the left side. Removal of the vagina is also indicated. In other countries (such as Australia), it is now mandatory to preserve all tissue in case the boy might want to use the ovarian part later; this may not be the case in Indonesia but is the patient choice. The chance of a gender change in this child is negligible as he has enough testosterone. He will start testosterone replacement therapy as the bone age is very advanced. Bilateral mastectomy will be considered after 6 months-the tissue will shrink by about 80% so a smaller procedure will be better for him. [2,5,15]

A comprehensive multidisciplinary approach by clinicians is also important for the management of any DSD patients. [9] The expert multidisciplinary team for DSD should include a pediatrician, obstetrician/gynecologist, urologist, geneticist, surgeon, endocrinologist, psychologist and a peer counselor. These disciplines are all important for the comprehensive management of DSD patients. [2,14]

CONCLUSION

In this patient, gender assignment as male was made based on evaluation of gender identity and gender role by the multidisciplinary team in our center, taking into account the patient's wish. Since the patient has not reached the legal age to make a medical decision, parental consent and patient assent were obtained as part of this process, and parental support for the transfer of information and the strategies greatly helped the clinicians to deliver the best possible treatment for the patient. Serial measurement of estradiol should also be carried out to confirm absence of residuary ovarian tissue. Due to the potential risk of the testicular regression, hormonal evaluation

and hormone replacement may be given to maintain secondary sexual characteristics. Longitudinal follow- up is required to avoid adverse effects that may occur due to gonadal insufficiency and inherent risk of malignancy.[3,14-16] 46,XX ovotesticular DSD is a rare case. Only a few cases have been found and documented. Sexual assignment is a challenge and the treatment debated. Treatment consists of medical treatment, surgical care, and consultation of expertise in urology, gynecology, endocrinology, pediatrics, and psychiatry. It is important to accommodate the requests of the patient, if he or she can express the preference about the sex of rearing. However, after the Consensus Conference of Chicago, most authors suggest not to proceed with a surgical treatment if it is not considered life-saving. A constant psychological support to the family and a multidisciplinary follow-up are the keys for an optimal outcome.

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CONFLICT OF INTEREST

The authors declare that none of them has any conflict of interest with any private, public, or academic party related to the information contained in this manuscript.

REFERENCES

1. Cools M, Köhler B. Disorders of Sex Development. In: Brook CGD, Clayton PE, Brown RS, editors. Brook's Clinical Pediatric Endocrinology. 7th ed. Hoboken: Wiley Blackwell; 2019. p. 105–31.
2. Warne GL. Long-term outcome of disorders of sex development (DSD): A world view. In: Lee PA, Nordenström A, Houk CP, editors. Disorders of Sex Development. Berlin: Springer; 2012. p. 279–90.
3. Balsamo A, Cicognani A, Ghirri P, Scaramuzzo RT, D'Alberton F, Bertelloni S, et al. Disorders of sexual development. In: Buonocore G, Bracci R, Weindling M, editors. Neonatology. Berlin: Springer; 2012. p. 1004–17.
4. Arboleda VA, Sandberg DE, Vilain E. DSDs: genetics, underlying pathologies and psychosexual differentiation. Nat Rev Endocrinol. 2014;10(10):603.
5. Matsui F, Shimada K, Matsumoto F, Itesako T, Nara K, Ida S, et al. Long- term outcome of ovotesticular disorder of sex development: A single center experience. Int J Urol. 2011;18(3):231–6.
6. Undre S, Cherian A. Paediatric urology. Surgery (Oxford). 2020;38(4):224–30.
7. Osaka A, Ide H, Matsuoka K, Iwahata T, Kobori Y, Ban S, et al. SRY-Positive 46, XX Testicular Disorder of Sexual Development With Leydig Cell Tumor. Am J Mens Health.

2020;14(5):1557988320970071.

8. Hersmus R, de Leeuw BH, Wolffenbuttel KP, Drop SL, Oosterhuis JW, Cools M, et al. New insights into type II germ cell tumor pathogenesis based on studies of patients with various forms of disorders of sex development (DSD). Mol Cell Endocrinol. 2008;291(1-2):1–10.
9. Hewitt JK, Warne GL. 46, XY DSD. In: Lee PA, Nordenström A, Houk CP, editors. Disorders of Sex Development. Berlin: Springer; 2012. p. 63–80.
10. Bouty A, Grover SR, Hutson JM. Outcomes: A World View of Individuals with DSD. In: Lee PA, Nordenström A, Houk CP, editors. Disorders | Differences of Sex Development: An Integrated Approach to Management. Berlin: Springer; 2020. p. 319.
11. Delforge X, Brachet C, Damry N, Segers V, Luyckx S, Heinrichs C, et al. A novel approach in the intraoperative management of ovotesticular DSD. J Pediatr Urol. 2020;16(6):768–70.
12. Scarpa M-G, Di Grazia M, Tornese G. 46, XY ovotesticular disorders of sex development: A therapeutic challenge. Pediatr Rep. 2017;9(4):50–2.
13. Mouriquand PD, Gorduza DB, Gay C-L, Meyer-Bahlburg HFL, Baker L, Baskin LS, et al. Surgery in disorders of sex development (DSD) with a gender issue: If (why), when, and how? J Pediatr Urol. 2016;12(3):139–49.
14. Raza J, Zaidi SZ, Warne GL. Management of disorders of sex development–With a focus on development of the child and adolescent through the pubertal years. Best Pract Res Clin Endocrinol Metab. 2019;33(3):101297.
15. Hiort O, Birnbaum W, Marshall L, Wünsch L, Werner R, Schröder T, et al. Management of disorders of sex development. Nat Rev Endocrinol. 2014;10(9):520.
16. O'Connell MA, Hutson JM, Grover SR. Medical Management of DSD. In: Lee PA, Nordenström A, Houk CP, editors. Disorders | Differences of Sex Development: Springer; 2020. p. 193–219.