



## Transverse testicular ectopia with persistent mullerian duct syndrome: Misdiagnosis and surgeon's unexpected discovery: A case report

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### ABSTRACT

Reports on the occurrence of persistent mullerian duct syndrome along with transverse testicular ectopia are extremely rare globally. In this condition, the fallopian tubes, uterus, cervix and upper two-thirds of vagina occurs alongside transverse testicular ectopia. In most cases, the condition is discovered as an incidental finding during surgical procedures. In this report, we present a case of a 29-year-old Somali male that had scrotal swelling, pain around the inguinal area and perceived infertility for over seven years whom we incidentally found to have a persistent mullerian duct syndrome with transverse testicular ectopia and managed accordingly.

### 1. Introduction

Persistent Mullerian duct syndrome (PMDS) is a rare sexual development disorder and even rarer when the condition is accompanied by transverse testicular ectopia (TTE). PMDS is the presence of the uterus, fallopian tubes, and proximal vagina in a normally virilized man with normal male genotype (46XY). The PMDS is caused by anti-mullerian hormone (AMH) or AMHR-2 receptor deficiency.<sup>1</sup> TTE or crossed testicular ectopia is where both the spermatic cords and its testes exit on a single inguinal canal rather than on either side.

Majority of the patients with PMDS and TTE are incidentally found during surgery and treated in early childhood making its discovery extremely low during adulthood.<sup>2</sup> The preoperative accurate diagnosis is mainly uncommon. Sonography which is the first-line imaging frequently reports only inguinoscrotal hernia and undescended testes. We present a case of a young Somali man who presented right scrotal swelling, inguinal area pain, and primary infertility.

### 2. Case presentation

A 29 years-old man came to our urology department with right scrotal swelling since birth, with complaint of inguinal area pain, and

primary infertility for 7 years. The patient was married on three separate occasions for more than 7 years without conception. Upon physical examination, we noticed swelling of the right scrotum that could not be manually reduced. The patient is normally virilized with normal secondary sex characteristics. Scrotal Doppler ultrasound concluded the following findings; right-side scroto-inguinal hernia, right-side moderate hydrocele, and bilateral varicocele. Both testicle was visualized inside the scrotum with normal size. Sperm analysis revealed necrozoospermia.

During the initial operation, we incidentally discovered a fully developed uterine-like structure with broad and round ligaments, fallopian tubes, and normal-looking testicle on either side of the uterus (Fig. 1). Both testicle exited on the right side of the inguinal canal alongside the persistent mullerian duct structures (Fig. 1). We discussed with the patient and his family about the findings and took biopsies for histopathological analysis.

Hormonal testing including; FSH, LH, prolactin, estradiol E2, testosterone, beta-HCG, LDH, AFP, and AMH were all in the normal range. *Trans*-rectal ultrasound showed normal prostate gland and seminal vesicles. Pelvic MRI with contrast failed to recognize PMDS or TTE structures.

First histopathology result indicates fibromuscular tissue from the

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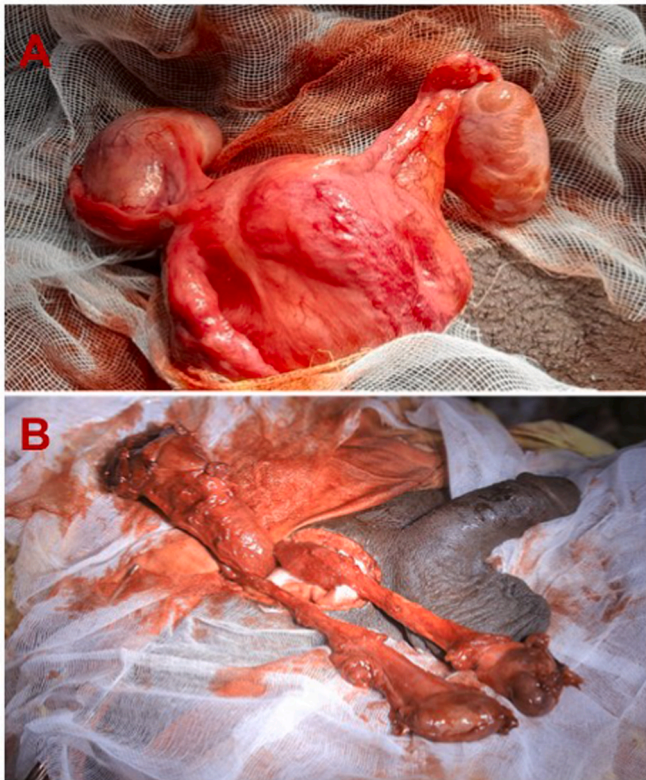
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**Fig. 1.** A: Persistent mullerian duct structures (middle part) and two testes on either side, B: Separated persistent mullerian duct structures from two the spermatic cords with testes.

sample taken from the uterine fundus, while the second tissue from the testes showed normal seminiferous tubules with adjacent Leydig cells with no evidence of malignancy. We discussed treatment options with the patient including, persistent mullerian duct remnant removal and the possibility of bilateral orchiectomy considering the risk of malignancy in which they strongly rejected the latter.

The second operation was performed on the same incision site. Initially, we delivered both testes with its mullerian ducts remnants into the inguinal area. A delicate dissection was carried-out, the two spermatic cords on each side of the uterine cervix was separated from the uterus. The uterus was removed at the level of cervix in the deep inguinal canal. Lastly, inguinal hernia repair and testicular fixation into the scrotum was performed. The second histopathology reported; endometrium tissue, endocervical glands lining the cavity, fallopian tube tissue, and vas deferens tissue (Fig. 2). No evidence of malignant tissues was identified.

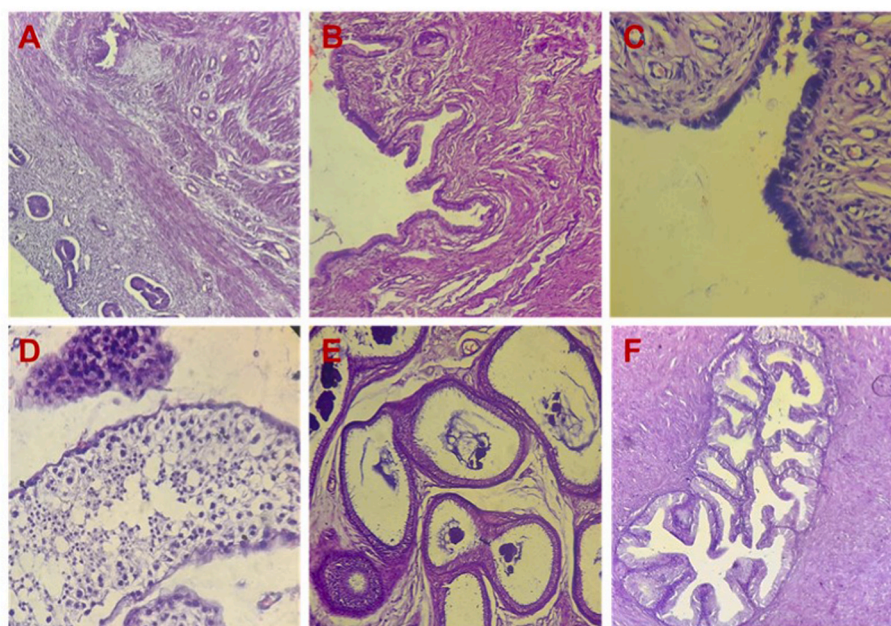
Two months post-operative scrotal Doppler US found; normal left testicle with normal echogenicity and blood supply, however, the right testicle showed abnormal echogenicity. Four-month post-operative sperm analysis revealed necrozoospermia. We explained to the patient the difficulty of normal fertility considering his disorder and recommended him to try assisted-reproductive techniques in the future.

### 3. Discussion

TTE is a rare congenital disorder with an estimated incidence of 1 in 4 million males.<sup>1</sup> To date, there are no more than 160 TTE cases reported in the literature, and only about half of them had TTE accompanied with PMDS.<sup>3</sup> Majority of the TTE with PMDS cases are reported from outside the African continent.

TTE is classified into three subtypes based on the accompanying anomaly; Type 1: TTE with inguinal hernia (40–50%), Type 2: TTE with PDMS (20–30%), and Type 3: TTE with scrotal, kidneys, and some other anomalies.<sup>4</sup> In contrast, a literature review of 129 TTE cases conducted between 1982 and 2020 showed 27.9%, 49.6%, 22.5% of type 1, 2, and 3, respectively.<sup>3</sup> As these figures has shown, TTE with PMDS is extremely rare.

Preoperative diagnosis is crucial for surgical planning, patient counselling, and consent. However, it is very challenging if the imaging examinations overlooks the accuracy of the diagnosis due to the rarity of the disease or its close resemblance to other familiar conditions. For instance, the initial scrotal Doppler US examination in our case did not report PMDS or the TTE leading to our unexpected discovery of the disorders during the surgery. This is in conformity with the 129 TTE cases reviewed by Zhou et al. where only 55 (42.6%) were preoperatively diagnosed while the remaining 74 (57.4%) cases were incidentally



**Fig. 2.** A: Endometrial tissue, B: Endocervical glands, C: Fallopian tube tissue, D: Seminiferous tubules, E: Epididymis tissue, F: Vas Deferens.

identified during the surgery.<sup>3</sup> Laparoscopy is another tool used for accurate diagnosis of PMDS. Unfortunately, laparoscopy is an invasive procedure and may not be available in every clinical setting.

Testicular malignancy and infertility are two major long-term concerns for the patients diagnosed with PMDS and TTE. The incidence of testicular cancer in patients with PMDS ranges between 18 and 33%.<sup>5</sup> Moreover, seminoma is the most commonly reported tumor.<sup>5</sup> Besides, a metastatic adenocarcinoma of the müllerian duct structures in males with PMDS has also been described.<sup>5</sup> On the other hand, fertility in patients with PMDS and TTE is a rare entity. The reported fertility in the literature ranges between 11 and 19%.<sup>5</sup>

Our case limitation includes; the lack of karyotyping analysis, and AMH mutations study which are not currently available in the country. However, the short follow-up period can also have been seen as a shortcoming.

#### 4. Conclusion

Though the disorders are very rare, it's important to highlight the existence of PMDS with TTE in African continent and not only limited to other regions. Early detection and treatment is essential for better outcomes. Surgeons and radiologists should be aware of these infrequent disorders.

#### Ethics

Informed consent was obtained from the patient.

#### Declaration of competing interest

None.

#### References

1. Abdullayev T, Korkmaz M. Transvers testicular ectopia: a case report and literature review. *Int J Surg Case Rep.* 2019;65:361–364. <https://doi.org/10.1016/j.ijscr.2019.11.007>.
2. Sigdel PR, Dhital P, Kulung Rai BD, Poudyal S, Luitel B, Sharma UK. Persistent Müllerian duct syndrome with transverse testicular ectopia: a case report. *Urol Case Reports.* 2019;25, 100888. <https://doi.org/10.1016/j.eucr.2019.100888>.
3. Zhou G, Yin J, Jiang M, Yang Z, Li S. Clinical characteristics, ultrasonographic findings, and treatment of pediatric transverse testicular ectopia: a 10-year retrospective review. *Urology.* 2021;154:249–254. <https://doi.org/10.1016/j.urology.2021.01.006>.
4. Gauderer MWL, Grisoni ER, Stellato TA, Ponsky JL, Izant RJ. Transverse testicular ectopia. *J Pediatr Surg.* 1982;17(1):43–47. [https://doi.org/10.1016/S0022-3468\(82\)80323-0](https://doi.org/10.1016/S0022-3468(82)80323-0).
5. Picard JY, Cate RL, Racine C, Josso N. The persistent müllerian duct syndrome: an update based upon a personal experience of 157 cases. *Sex Dev.* 2017;11(3):109–125. <https://doi.org/10.1159/000475516>.