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Persistent Müllerian Duct Syndrome with Inguinal Uterine Hernia: A Case Report and Literature Review

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Abstract

Inguinal uterine hernia is a rare male pseudo androgynous syndrome characterized by the presence of Müllerian ducts and the uterus, cervix, and fallopian tubes in a male with a 46, XY karyotype; it is a type of persistent Müllerian duct syndrome.

Case Presentation: A 40-year-old male presented with a left inguinal mass, and an inguinal ultrasound was suggestive of a left-sided incarcerated hernia. On laparoscopic exploration, the patient was found to have a uterus, cervix and vagina in the abdominal cavity.

Discussion: Persistent Müllerian Duct Syndrome (PMDS) is caused by defects in the Anti-Müllerian Hormone (AMH); it is not easily diagnosed, and abdominal CT, MRI and laparoscopy should be combined to achieve a comprehensive diagnosis. The main goal of treatment is to prevent cancer and to preserve the reproductive function of the patient as much as possible.

Conclusion: PMDS should be strongly suspected in patients with inguinal hernias combined with cryptorchidism, and patient reproductive function, the surgical risks and the possible complications should be fully evaluated to develop a comprehensive treatment plan before performing surgery.

Keywords: Persistent Müllerian duct syndrome; Inguinal uterine hernia; Laparoscopic exploration; Literature review

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Introduction

Persistent Müllerian Duct Syndrome (PMDS) is a rare congenital form of male pseudo hermaphroditism; it is a dysplastic reproductive sexual development disorder found in biological males (46, XY), and its pathophysiology can be explained by a defect in the action of the Anti-Müllerian duct Hormone (AMH) [1]. It is characterized by the simultaneous development of the Wolffian and Müllerian ducts, which give rise to the uterus and fallopian tubes. The diagnosis of PMDS is usually incidental; it is discovered during treatment for an inguinal hernia or cryptorchidism. However, persistent Müllerian duct syndrome combined with an inguinal uterine sliding hernia is extremely rare, according to national and international reports. The author reports a case of a patient with male Müllerian duct syndrome combined with inguinal uterine sliding hernia admitted for gastrointestinal surgery at the Sichuan Provincial People's Hospital.

Case Presentation

The patient was a 40-year-yld male. In January 2022, the patient was admitted to the hospital for "a year-long history of a reproducible mass in the left inguinal region with vague pain for 2 weeks". On admission, a mass approximately 6 cm × 5 cm × 4 cm in size was discovered on palpation of the left inguinal region. The mass could enter the scrotum and was soft in texture. The area was mildly tender to the touch. The mass was clearly visible in the standing position and could be reduced in the lying position; the patient had normal external genital morphology. A recurrent inguinal hernia on the left side was considered. The patient said that his sex life was normal. With regard to the patient's history, hernia repair and right crypt orchidectomy were performed in our hospital 3 years ago due to a bilateral inguinal hernia.

After admission, haematological parameters, liver and kidney function, coagulation function, electrocardiogram and chest X-ray results were normal. The preoperative diagnosis was a recurrent inguinal hernia on the left side and left testicular hydrocele; laparoscopic exploration was performed

Figure 1: Intraoperative image.
a: General shape of the uterus under laparoscopy; b: Laparoscopic adjacency of the bladder, uterus and bladder-uterine trap; C: Laparoscopic view of the uterine horn slipping into the hernia sac through the hernia ring opening.

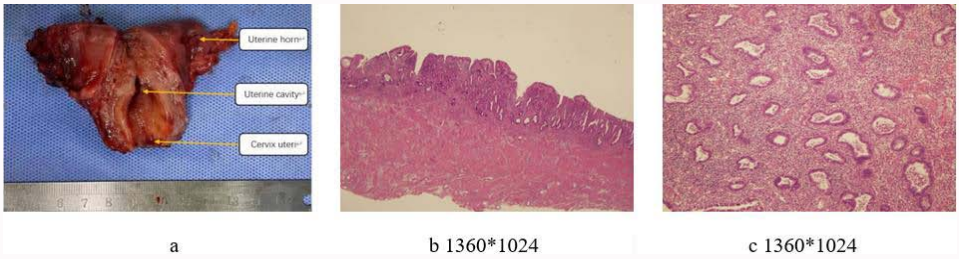

Figure 2: Gross uterine specimen under direct observation.
a: Pathological sections of the uterus; b: Typical endometrial and myometrial tissues; c: Glandular structures in the endothelial tissue.

Table 1: Hormone assay table.

Hormone category	Detection value	Normal range
T (nmol/L)	11.8	4.94-32.01
E2(pmol/L)	135.6	40.4-161.5
LH (mIU/ml)	25.51	0.57-12.07
FSH (mIU/ml)	36.94	0.95-11.95
AMH (ng/mL)	0.23	1.45-18.77
PGN (nmol/L)	135.6	Less than 0.64
PRL (mIU/mL)	292.72	86-324
GN (ng/mL)	0.087	Less than 3.0
ACTH (pg/mL)	20.9	Less than 46.0
Chromosome karyotype	46, XY	-

T: Testosterone; E2: Estradiol; LH: Luteinizing Hormone; FSH: Follicle Stimulating Hormone; AMH: Anti-Müllerian Hormone; PGN: Progesterone; PRL: Prolactin; GH: Growth Hormone; ACTH: Adrenocorticotropic Hormone.

after preoperative preparation. During the intraoperative exploration, muscle tissue with a size of approximately 8 cm × 7 cm × 5 cm was observed behind the bladder and anterior to the rectum, a hernia ring opening was observed lateral to the left inguinal inferior abdominal wall artery with a size of approximately 4 cm × 3 cm, and part of the intestinal canal and muscle tissue was observed to have slipped into the hernia sac. The muscle tissue was rich in blood supply, and the tissue was connected to the pelvic wall on both sides by what appeared to be the umbrella of the fallopian tube, with the outer shape resembling a uterus, as shown in Figure 1a, 1b and 1c. Given the patient's sex, the muscle tissue was removed after explaining the patient's condition to the family and obtaining consent. The tissue was removed, and the gross specimen is shown in Figure 2a. The hernia ring opening was closed with sutures, a patch was placed, and the abdomen was closed layer by layer.

Postoperative pathological examination revealed that the pelvic mass was uterus-like tissue. Postoperatively, the relevant tests were performed. Tests of the sex hormones and adrenocorticotropic hormones and the chromosome karyotype results were normal, but the anti-Müllerian hormone level was far below the normal range, as shown in Table 1. Postoperative histopathological sections showed clear evidence of the presence of endometrial tissue, muscle wall tissue, and glandular structures in the endometrial tissue, as shown in Figure 2b and 2c.

PMDS was first described by Nilson as an inguinal uterine hernia in 1939. Clarence classified PMDS into three categories: Type I is characterized by the presence of bilateral intra-abdominal testes relatively similar to the ovaries (approximately 60% to 70%).Type II is characterized by the presence of a unilateral testis with contralateral inguinal hernia with contents including a testis, the uterus and the fallopian tubes, which is also called an inguino-uterine hernia (approximately 20% to 30%) and Type III is characterized by the presence of two testes located in the same inguinal sac (approximately 10%) [2]. A time-bound search of the English language literature (2000-2021) was conducted to identify the relevant studies, and the main clinical features of the syndrome are summarized in Table 2.

Discussion

The incidence and prevalence of PMDS are uncertain. PMDS is usually familial and is an autosomal recessive disorder occurring in individuals with a 46 XY karyotype [19]. It mainly manifests as a result of chromosomal, gonadal or external sex organ discordance and is predominantly female dominant. It has been suggested in the literature that the location of the testes or the doxorubular complex in PMDS patients may differ even between brothers with the same genotype [20,21]. However, approximately 41%of the cases reported in the literature involved bilateral cryptorchidism, approximately 32% involved inguinal uterine hernias, and approximately 27% involved transverse testicular ectopia [6]. In addition, one study highlighted that male sex differentiation is driven by two different hormones, each of which is produced by a different cellular compartment of the fetal testes [6]. Testosterone is produced by fetal mesenchymal cells and acts to maintain the Wolffian duct and promote the differentiation of the external genitalia; at 8 weeks of gestation of a male fetus, testicular support cells produce Anti-Müllerian Hormone

Table 2: Characteristics of cases reported between 2000 and 2021.

Feature	Findings
Age	Most cases are diagnosed in childhood [3-5].
Diagnostic method	Found during laparoscopy or hernia repair [3,6,7].
Clinical symptoms	Inguinal hernia, cryptorchidism, infertility [4,8-10,11-13].
External genitalia	Bilateral, less commonly unilateral, cryptorchidism, normal penis. Male dominant [4,9-10,12-13].
Internal genitalia	Hypoplastic tests, uterus, two fallopian tubes and upper third of vagina [14,15].
Chromosome karyotype	46, XY, sometimes mosaicism [3,4,16].
Pathological examination	Uterus-like tissue, vascularized smooth muscle tissue [4,11].
Surgical protocol	Preservation of reproductive function with testicular fixation and hysterectomy and tubal resection [4,16,17].
AMH	Low or lacking [14,18].
Operation complication	Occasional hernia recurrence or vas deferens injury [19].
Sex	Mainly male dominant type [15].
Sex hormone, pituitary hormone	Basically normal [3,5,11,16].
Sexual life and secondary sexual characteristics	Basically normal [9,15].
Sperm motility	Low or no vitality [8].

Summary of articles published in English from 2000-2021.

(AMH), leading to degeneration of the Müllerian duct [22]. AMH is a member of the TGF-β family [23] that is synthesized by fetal support cells [24] and drives the degeneration of the fetal Müllerian duct. If the fetal Müllerian duct does not degenerate, it differentiates into the uterus, fallopian tubes and upper vagina. It has also been shown that in approximately 85% of cases, the pathophysiology of this syndrome can be explained by AMH deficiency [25]. The AMH level in the patient described in this case was well below the normal range, which is essentially consistent with the above reports.

This case of PMDS was found accidentally mainly due to recurrent inguinal hernia repair, and the contents of the hernia were one side of the uterine horn and the fallopian tube. This patient also had right cryptorchidism, which put the patient at risk of malignancy, indicating that he needed to undergo surgery to prevent the development of life-threatening testicular cancer [7]. One study suggested that the overall incidence of malignancy in patients with cryptorchidism is approximately 18% [26] therefore, the right retained testicle was also removed.

Male persistent Müllerian duct syndrome combined with recurrent inguinal sliding hernia is a rare clinical condition, and there are few relevant reports in the national and international literature. However, the vast majority of PMDS findings are incidental and are usually found during surgery to correct cryptorchidism or an inguinal hernia. Male pseudo hermaphroditism is relatively difficult to diagnose, and PMDS is even more difficult to diagnose. When making a diagnosis of PMDS, patient history and physical signs appear to be crucial as the first considerations; factors such as a history of consanguineous marriage in the family and features on examination of the external genitalia that can be very intuitively distinguished from those of true hermaphroditism should be considered. Second, laboratory examinations play a central role, and karyotype examination is critical for the determination of the patient's sex these factors need to be considered together with the levels of AMH, estrogenic androgen, etc. Third, laparoscopic exploration can not only clearly and intuitively reveal the development of the uterus, fallopian tubes, and ovaries but can also allow the tissue to be biopsied, which can clarify the nature of the observed tissue [27]. Finally, abdominal and genitourinary system ultrasound, CT, and MRI play certain roles in the diagnosis of pseudo hermaphroditism, but the accuracy of diagnoses based on these imaging modalities is low.

Laparoscopic exploration has the advantages of reduced trauma, better visualization and fewer complications, and with the continuous development of laparoscopic techniques, this form of exploratory surgery has come to play a major role in the diagnosis and treatment of PMDS, not only through the acquisition of biopsy specimens, but also through the removal of tissue for pathological examination based on the situation observed during the operation these benefits lead to a more accurate diagnoses and resolution of the problem [28]. In addition, the aim of PMDS management should be to detect inguinal uterine hernias and cryptorchidism in a timely manner, to select the surgical plan according to the patient's chosen sex, to preserve reproductive function and to reduce the risk of Mueller's duct malignancy as much as possible. The goal of treatment is to prevent two major complications of PMDS: infertility and cancer [6]. Malignant transformation of Müllerian duct structures has been reported in adult males [29]. In this case, the patient's chosen gender was male; he was married and infertile, with normal external genitalia, a normal sexual life, and inactive sperm Therefore, in combination with the patient's chosen gender, removal of the uterus and confirmation of the nature of the removed tissue by pathological examination was the most appropriate and meaningful treatment for this patient.

In summary, PMDS is clinically rare, with a complex etiology and pathogenesis, and it may be related to chromosomal abnormalities, genetic mutations, inbreeding, etc. Therefore, screening of patients with cryptorchidism, infertility, etc. Should be emphasized. In addition, PMDS is not easy to diagnose, and the diagnosis should be made based on a combination of the patient's clinical history, an external genital examination, a karyotype examination, detection of the level of AMH, and other factors. In addition, the development of the uterus and fallopian tubes as internal genitalia should be clarified under direct laparoscopic observation to achieve a more accurate diagnosis. Finally, the most meaningful treatment plan should be formulated according to the patient's sex. The diagnosis and

treatment of PMDS combined with recurrent inguinal sliding hernia is rarely reported, and since this case involved the gastrointestinal and genitourinary systems, the patient was treated in collaboration with the departments of gynecology and urology to solve the problems of pseudohermaphroditic PMDS, inguinal cryptorchidism and repair of inguinal sliding hernia. It is important to note that for male patients with PMDS, multidisciplinary collaboration is needed.

References

1. Lavergne O, Troisfontaines E, Verstraete A, Demarche M, Nicolas H. Une cause rare de cryptorchidie le syndrome de persistance des structures müllériennes [A rare cause of cryptorchidism, the persistence of müllerian ducts syndrome]. Rev Med Liege. 2018;73(7-8):376-79.
2. Clarnette TD, Sugita Y, Hutson JM. Genital anomalies in human and animal models reveal the mechanisms and hormones governing testicular descent. Br J Urol. 1997;79(1):99-112.
3. Sancar S, Özçakır E, Kaya M. Management of the patients with persistent Müllerian duct syndrome: Is the ultimate goal testicular descent? Turk J Urol. 2018;44(2):166-171.
4. Saleem M, Ather U, Mirza B, Iqbal S, Sheikh A, Shaukat M, et al. Persistent mullerian duct syndrome: A 24-year experience. J Pediatr Surg. 2016;51(10):1721-24.
5. Ren X, Wu D, Gong C. Persistent Müllerian duct syndrome: A case report and review. Exp Ther Med. 2017;14(6):5779-84.
6. 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.
7. Farikullah J, Ehtisham S, Nappo S, Patel L, Hennayake S. Persistent Müllerian duct syndrome: lessons learned from managing a series of eight patients over a 10-year period and review of literature regarding malignant risk from the Müllerian remnants. BJU Int. 2012;110(11 Pt C):E1084-E1089.
8. Alp BF, Demirer Z, Gürağaç A, Babacan O, Sarı E, Sarı S, et al. Persistent Mullerian duct syndrome with transverse testicular ectopia and seminoma. Int Urol Nephrol. 2014;46(8):1557-62.
9. Mukhtar MU, Niazi SAK, Sarwar MZ, Naqi SA. Transverse testicular ectopia with persistent Mullerian duct syndrome: an operative eureka. Int J Surg Case Rep. 2020;71:338-40.
10. Deepika, Kumar A. Persistent mullerian duct syndrome with transverse testicular ectopia: rare entity. J Clin Diagn Res. 2014;8(3):162-63.
11. Sigdel PR, Dhital P, Kulung Rai BD, Poudyal S, Luitel B, Sharma UK. Persistent Mullerian duct syndrome with transverse testicular ectopia: A case report. Urol Case Rep. 2019;25:100888.
12. Kumar S, Mohan A. Persistent Mullerian duct syndrome with transverse testicular ectopia. J Surg Case Rep. 2012;2012(5):9.
13. Ark JT, Moses KA. Operative considerations for late-presenting persistent Müllerian duct syndrome. Urol Ann. 2016;8(3):363-65.
14. Marcus KA, Halbertsma FJ, Picard JY, Otten BJ. A visual pitfall: persistent Müllerian duct syndrome (PMDS). Acta Paediatr. 2008;97(1):129-132.
15. Renu D, Rao BG, Ranganath K, Namitha. Persistent mullerian duct syndrome. Indian J Radiol Imaging. 2010;20(1):72-4.
16. Alharbi KN, Khushaim AO, Alrasheed M, Akhtar M, Neimatallah M. Radiological Findings in Persistent Müllerian Duct Syndrome: Case Report and Review of Literature. J Radiol Case Rep. 2017;11(3):7-14.
17. Agrawal AS, Kataria R. Persistent Müllerian Duct Syndrome (PMDS): a Rare Anomaly the General Surgeon Must Know About. Indian J Surg. 2015;77(3):217-21.
18. Singh R, Kumar SD, Aggarwal N. MRI findings of Persistent Mullerian Duct Syndrome: A Rare Case Report. J Clin Diagn Res. 2017;11(6):TD05-TD06.
19. Natarajan S, Periasamy M, Rangasamy S, Mohan S, Sundararajan P. Persistent Mullerian Duct Syndrome: A Single-Center Experience. J Indian Assoc Pediatr Surg. 2018;23(4):203-5.
20. Abduljabbar M, Taheini K, Picard JY, Cate RL, Josso N. Mutations of the AMH type II receptor in two extended families with persistent Müllerian duct syndrome: lack of phenotype/genotype correlation. Horm Res Paediatr. 2012;77(5):291-97.
21. Nalbantoğlu Ö, Demir K, Korkmaz HA, et al. A novel mutation of AMH in three siblings with persistent Mullerian duct syndrome. J Pediatr Endocrinol Metab. 2015;28(11-12):1379-82.
22. Wongprasert H, Somanunt S, De Filippo R, Picard JY, Pitukcheewanont P. A novel mutation of anti-Mullerian hormone gene in Persistent Mullerian Duct Syndrome presented with bilateral cryptorchidism: a case report. J Pediatr Urol. 2013;9(4):147-49.
23. Cate RL, Mattaliano RJ, Hession C, Tizard R, Farber NM, Cheung A, et al. Isolation of the bovine and human genes for Müllerian inhibiting substance and expression of the human gene in animal cells. Cell. 1986;45(5):685-98.
24. Armendares S, Buentello L, Frenk S: Two male sibs with uterus and Fallopian tubes. A rare, probably inherited disorder. Clin Genet. 1973;4(3):291-96.
25. Çakır AD, Turan H, Onay H, Emir H, Emre S, Comunoglu N, et al. A Novel Mutation of AMHR2 In Two Siblings with Persistent Müllerian Duct Syndrome. Sex Dev. 2017;11(5-6):289-92.
26. Telli O, Gökçe MI, Haciyev P, Soygür T, Burgu B. Transverse testicular ectopia: a rare presentation with persistent Müllerian duct syndrome. J Clin Res Pediatr Endocrinol. 2014;6(3):180-82.
27. Chertin B, Koulikov D, Alberton J, Hadas-Halpern I, Reissman P, Farkas A. The use of laparoscopy in intersex patients. Pediatr Surg Int. 2006;22(5):405-8.
28. Ma D, Ye MZ, Deng XG, Su H. The application of lumpectomy in the diagnosis and treatment of hermaphroditism with 33 case reports. Chinese J Minimally Invasive Surg. 2016;16(10):913-16.
29. Viart L, Peltier J, Forzini T, Page C, Foulon P, Saint F, et al. Syndrome de persistance des canaux de Müller de type féminin: un cas de diagnostic tardif dans le cadre d'une infertilité [Persistent Müllerian ducts syndrome: one case of late hypofertility]. Morphologie. 2015;99(324):23-28.