

Radiological Findings in Persistent Müllerian Duct Syndrome: Case Report and Review of Literature

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ABSTRACT

This case involved a 36-year-old adult male who presented with an unusual inguinal hernia in which the uterus and fallopian tubes were identified as contents of the inguinal hernia sac. These findings reflected a rare autosomal recessive developmental syndrome known as PMDS (persistent Müllerian duct syndrome). The diagnosis was established and confirmed via radiological-mainly MRI-investigation.

CASE REPORT

CASE REPORT

A 36-year-old man presented with a recurrent inguinal hernia for the third time. Radiological investigations revealed hidden female genitalia that had been missed during two prior surgeries. The patient had a history of right inguinal hernia repair, with the first procedure done laparoscopically and the second performed as an open hernia repair. He also had a history of bilateral cryptorchidism, with orchiopexy performed since that diagnosis. The patient had been under the care of a urologist since childhood, was married, and having fertility problems. The patient was admitted at the time of presentation; he was vitally stable but complaining of right inguinal pain at the site of the previous inguinal hernia. Hormonal assays were within normal limits, and chromosome studies revealed karyotype 46-XY and a normal chromosome complement with an apparent abnormality.

Imaging Findings

A pelvis CT scan with intravenous injection of contrast material was performed, which showed a blind-ending, fluid-filled tubular structure arising just posterior to the urinary bladder and extending into the right inguinal canal (Fig. 1). A

scrotal ultrasound was then performed, the sagittal gray-scale ultrasound of the scrotum demonstrated a uterus with fluid in the endometrial cavity within the scrotal sac (Fig. 2). Both the CT and US findings were then confirmed via an MRI of the pelvis, which revealed a tubular structure with signal characteristics and the zonal anatomy of uterine tissue representing the uterus. It was connected to a fluid-filled structure representing the upper two-thirds of the vagina, which was communicating with the right ejaculatory duct (Fig. 3); notably, the lower third of the vagina was absent (Fig. 4). The uterus was herniated to the right inguinal canal (Fig. 5). Neither seminal vesicles nor ovaries were seen. The prostate was present and unremarkable.

Management

The patient underwent elective exploratory laparotomy, which included resection of the uterus and Müllerian duct remnants and repair of the hernia. The excised gross specimen showed a normal-appearing uterus and endometrium, with no masses identified (Figs. 6, 7).

Follow-up

The patient was discharged four days post-surgery and followed for six months. He had no complaints of pain or swelling, and the only ongoing issue was his fertility problem.

DISCUSSION

Etiology & Demographics:

This case represents a rare form of pseudohermaphroditism in males (persistent Müllerian duct syndrome), which is caused by a deficiency in the intrauterine release of AMH (anti-Müllerian hormone)—also known as Müllerian inhibiting substance (MIS) [1]. PMDS has an autosomal recessive mode of transmission, with few cases reported that had the possibility of familial X-linked inheritance [2]. In this congenital syndrome, there are extra female genital organs present in a normal 46XY genotypical and phenotypical male. These extra organs—the uterus, fallopian tubes, and upper one-third of the vagina—are derived from the Müllerian duct in fetal life [3].

Müllerian and Wolffian ducts are both present in the fetus in the seventh week of gestation. The Wolffian duct, if it develops normally, will form male reproductive organs; subsequently, the Müllerian duct will be regressed and will not develop from the MIS released from the Sertoli cells of the male fetus [1]. Testosterone produced from the male fetal testicles is also involved in this differentiation. In a PMDS patient, the MIS is not released; therefore, he will continue to form both male and female reproductive organs that are derived from the Wolffian and Müllerian duct, respectively [3].

PMDS has two anatomical types: male and female. The male type is most common (in up to 90% of cases) and further classified into two subcategories: hernia uteri inguinalis and transverse testicular ectopia [4]. Most cases present as hernia uteri inguinalis (with unilateral cryptorchidism and contralateral inguinal hernia) [4]. Transverse testicular ectopia is an extremely rare condition that includes herniation of both testes and Müllerian structures on the same hernia sac [5]. Our case was not a classical presentation of PMDS because the patient was an adult, and both testes were present in the scrotum at the time of presentation [4]. The female variant of PMDS, which is even less common, is characterized by the presence of both testes being undescended and fixed in the round ligament [4]. Many cases of PMDS can be hard to diagnose, and they are most often missed due to unfamiliarity with the condition [5, 6]. For both types of PMDS, fewer than 300 cases exist in the literature; notably, most of these cases were diagnosed in the pediatric population in patients younger than 10 years of age [5, 7].

Clinical & Imaging Findings:

The diagnosis of PMDS is mainly incidental; it is often found during either hernia repair or surgery for undescended testes [8]. Nevertheless, even with surgery for reduction of the

hernia sac, the diagnosis can be missed, as it was in our case. The recognition of PMDS is made through clinical history and genetic study; it can be confirmed via imaging and pathological analysis [4, 9, 10]. US can identify the contents of the hernia sac, the thickness of the uterine wall, and any collection in the endometrial cavity and intra-abdominal undescended testes. Additionally, it can help with visualization of the prostate and seminal vesicles, if present [10, 11]. CT can also help distinguish the Müllerian structures [12]. An MRI is confirmatory in this diagnosis due to its different signal intensities and its greater morphological ability to delineate the pelvic organs and the complex structures in PMDS, which cannot be determined on either US or CT [4, 5, 10, 11].

Our case was unique for the following reasons: 1) late clinical presentation and 2) the cause of primary infertility and the extra-genital organs were missed during the first hernia repair. The recognition of this unusual condition and the diagnosis were established based on the radiological investigation and confirmed via MRI.

Treatment & Prognosis:

Removal of the Müllerian remnants was needed due to an increased risk for malignancy [5, 7]. Per a case series performed by Farikullah et al., between 3.1% and 8.4% of PMDS patients will develop a malignancy of Müllerian origin [7]. Orchidopexy, followed by excision of the Müllerian duct remnants whenever possible, will decrease the risk of malignant transformation [6, 7]. Müllerian malignancies are more likely to develop than testicular cancer [5]. The incidence of testicular malignant transformation in the undescended testes of PMDS patients is estimated to be 5% to 18%, which is similar to the incidence of testicular carcinoma in abdominal testes in patients without PMDS [7]. In all cases of PMDS, the aims of surgical intervention are to both preserve fertility and avoid malignant changes [5, 7]. Laparoscopy is the preferable choice for operations; however, the procedure approach should be discussed with the surgical team [6]. Farikullah et al. advised a long-term follow-up with US or MRI due to the risk of malignant transformation, which often develops after puberty [7].

Differential Diagnosis:

Mixed gonadal dysgenesis (MGD), one of the most common differentials of persistent Müllerian duct syndrome, involves ambiguous genitalia associated with unilateral testes and a contralateral gonadal streak [13]. Because it is a challenge to both visualize and characterize the gonadal streak based on imaging, a biopsy is needed [14]. Streak gonads can appear as low signal intensity stripes on a T2 weighted MRI sequence; however, they are still difficult to see [14]. CT of the pelvis is not advocated for the pediatric population (the age group in which most of the gonadal anomalies present) due to the inherent risks associated with high radiation doses [14, 15].

Inguinal hernia of any sac contents is one of the differential diagnoses of PMDS (along with herniated bowel

loops, bladder, appendix, neoplasms, or metastases) [16]. The most common and characteristic findings on ultrasound of an inguinal hernia are abnormally moving intra-abdominal contents of variable echogenicity “mesenteric fat, bowel, or both” through the deep inguinal ring and inguinal canal [17]. A CT with oral contrast will show a contrast material-filled bowel loop that is herniating through the inguinal canal [16]. Similarly, other herniated sac contents account for the fewest cases of inguinal hernia and are often accompanied by comorbidities of varied origin, such as congenital anomalies, acquired conditions, vascular conditions, iatrogenic conditions, infectious processes, and neoplasms [16, 18]. MRI is not used to either diagnose or evaluate inguinal hernias in general [19]. Miller et al. recommend that patients with clinically suspected inguinal hernia yet atypical physical examination findings undergo an MRI because it is the most sensitive radiological modality for detecting hidden occult hernias [19].

Herniation of the urinary bladder into the scrotum (scrotal cystocele) due to injury or iatrogenic causes is another differential of PMDS [20]. US of the scrotum will show a fluid-filled structure that joins the urinary bladder intra-abdominally, with volume changes and wall thickening of the herniated section after micturition [20]. CT and MRI of the pelvis can reveal bladder hernia with pointing of the herniated portion both anteriorly and inferiorly into the hernia side [20].

TEACHING POINT

Persistent Müllerian duct syndrome (PMDS) is a rare condition that can present at any age. Recurrent inguinal hernia and primary infertility should cause professionals to suspect PMDS. CT and US can distinguish the Müllerian duct derivatives (uterus, fallopian tubes, and the upper part of the vagina). MRI is the best modality in the diagnosis of PMDS. Most common differentials of PMDS are mixed gonadal dysgenesis, herniated small bowel loops, and scrotal cystocele.

REFERENCES

1. Josso N, Lamarre I, Picard J, Berta P, Davies, Morichon N, Peschanski M, Jeny R. Anti-Müllerian Hormone in Early Human Development. *Early Human Development* 1993; 33 (2): 91-99. PMID: 8055780.
2. Belville C, Josso N, Picard J. Persistence of Müllerian Derivatives in Males. *American Journal of Medical Genetics* 1999; 89: 218-223. PMID: 10727997.
3. Rey R, Picard J. Embryology and Endocrinology of Genital Development. *Bailliere's Clinical Endocrinology and Metabolism* 1998; 12: 17-33. PMID: 9890060.
4. Dekker H, Jong I, Sanders J, Wolf R. Persistent Müllerian Duct Syndrome. *Radiographics* 2003; 23: 309-313. PMID: 12640149.

5. Shalaby M, Kurkar A, Zarzour M, Faddan A, Khalil M, Abdelhafez M. The Management of the Persistent Müllerian Duct Syndrome. *Arab Journal of Urology* 2014; 12: 239-244. PMID: PMC4435512.
6. Manjunath B. G., Shenoy V. G., Raj P. Persistent Müllerian duct syndrome: How to deal with the müllerian duct remnants — a review. *Indian Journal of Surgery* 2010; 72: 16-19. PMID: PMC3452545.
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 International* 2012; 110: E1084-E1089. PMID: 22540537.
8. Nayak V. J., Kamath A. S., Krishnappa M. H., Bylappa S. K. Persistent müllerian duct syndrome: A case report and review of the literature. *International Journal of Applied and Basic Medical Research* 2014; 4 (2): 125-127. PMID: PMC4137638.
9. Loeff D, Imbeaud S, Reyes H, Meller J, Rosenthal I. Surgical and Genetic Aspects of Persistent Müllerian Duct Syndrome. *Journal of Pediatric Surgery* 1994; 29(1): 61-65. PMID: 7907140.
10. Renu D, Rao B, Ranganath K, Namitah. Persistent Müllerian Duct Syndrome. *Indian Journal of Radiology and Imaging* 2010; 20(1): 72-74. PMID: PMC2844757.
11. Di Cesare E, Di Bartolo De Vincentiis, Maffey MV, Splendiani A, Masciocchi C. US and MRI in a Case of Persistent Müllerian Duct Syndrome. *Pediatric Radiology* 1998; 28: 865-867. PMID: 9799320.
12. Gujar N, Choudhari R, Choudhari G, Bagali N, Mane H, Awati J, Balachandran V. Male Form of Persistent Müllerian Duct Syndrome Type I (Hernia Uteri Inguinalis) Presenting as an Obstructed Inguinal Hernia: a Case Report. *Journal of Medical Case Reports* 2011; 5:586. PMID: 22185203.
13. Agrawal A. S., Kataria R. Persistent Müllerian Duct Syndrome (PMDS): a Rare Anomaly the General Surgeon Must Know About. *Indian Journal of Surgery* 2015; 77(3): 217–221. PMID: PMC4522266.
14. Chavhan G. B., Parra D. A., Oudjhane K, Miller S. F., Babyn P. S., Pippi Salle F. L. Imaging of Ambiguous Genitalia: Classification and Diagnostic Approach. *RadioGraphics* 2008 Nov-Dec; 28(7):1891-904. PMID: 19001646.
15. Smith-Bindman R., Lipson J., Marcus R., Kim K. P., Mahesh M., Gould R., Gonzales A. B., Miglioretti D. L. Radiation Dose Associated with Common Computed Tomography Examinations and the Associated Lifetime Attributable Risk of Cancer. *Arch Intern Med*. 2009;169(22):2078-2086. PMID: PMC4635397.

16. Bhosale P. R., Patnana M, Viswanathan C., Szklaruk F. The Inguinal Canal: Anatomy and Imaging Features of Common and Uncommon Masses. *RadioGraphics* 2008; 28:819–835. PMID: 18480486.

17. Jacobson J. A., Khoury V., Brandon C. J. Ultrasound of the Groin: Techniques, Pathology, and Pitfalls. *American Journal of Roentgenology* 2015 Sep; 205(3):513-523. PMID: 26102306.

18. Shadbolt C. L., Heinze S. B., Dietrich R. B. Imaging of groin masses: Inguinal anatomy and pathologic conditions revisited. *RadioGraphics* 2001 Oct; 21:S261-S271. PMID: 11598262.

19. Miller J., Cho J., Michael M. J., Saouaf R., Towfigh S. Role of Imaging in the Diagnosis of Occult Hernias. *JAMA Surg.* 2014 Oct;149(10):1077-80. PMID: 25141884.

20. Bacigalupo L. E, Bertolotto M., Barbiera F., Pavlica P., Lagalla R., Mucelli R. S. P., Derchi L. E. Imaging of Urinary Bladder Hernias. *American Journal of Roentgenology* 2005; 184:546-551. PMID: 15671377.

FIGURES

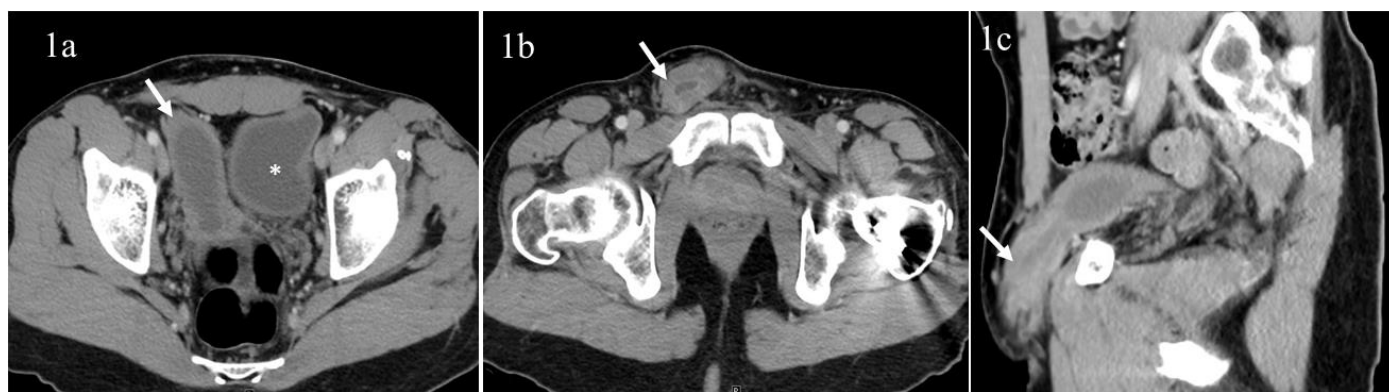


Figure 1: A 36-year-old male with Persistent Müllerian duct syndrome hernia with hernia uteri inguinalis.

Technique: 1a, 1b, 1c. (GE Healthcare Discovery CT750 HD) Pelvis CT scan with intravenous injection of contrast material, 95 ml Xenetix™ (iobitridol), 120 Kvp, 17 mAs, 3.75mm slice thickness.

Findings:

- a) An axial pelvis CT scan with intravenous injection of contrast material demonstrates a blind-ended fluid-filled tubular structure (arrow) arising just posterior to the urinary bladder (asterisk).
- b) An axial pelvis CT scan with intravenous injection of contrast material demonstrates the same blind-ended fluid-filled tubular structure extending into the right inguinal canal (arrow).
- c) A sagittal pelvis CT scan with intravenous injection of contrast material demonstrates a blind-ended fluid-filled tubular structure extending into the right inguinal canal (arrow).

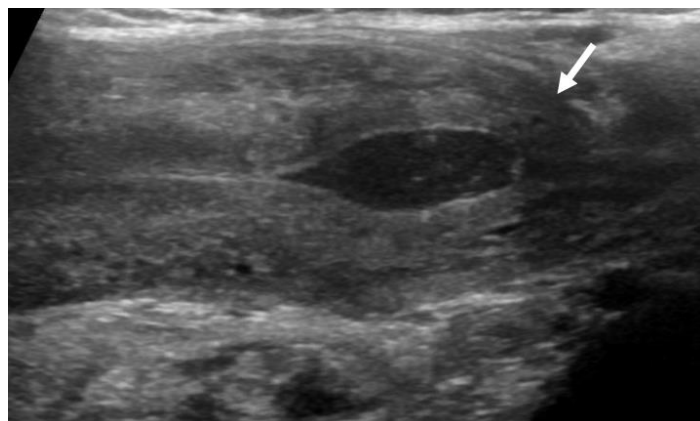


Figure 2 (left): A 36-year-old male with Persistent Müllerian duct syndrome hernia with hernia uteri inguinalis.

Technique: Ultrasound performed on GE LOGIC E9 with ML 6-15 transducer at a frequency of 15.0 MHz.

Findings: A sagittal gray-scale ultrasound of the scrotum demonstrates a uterus with fluid in the endometrial cavity within the scrotal sac (arrow).

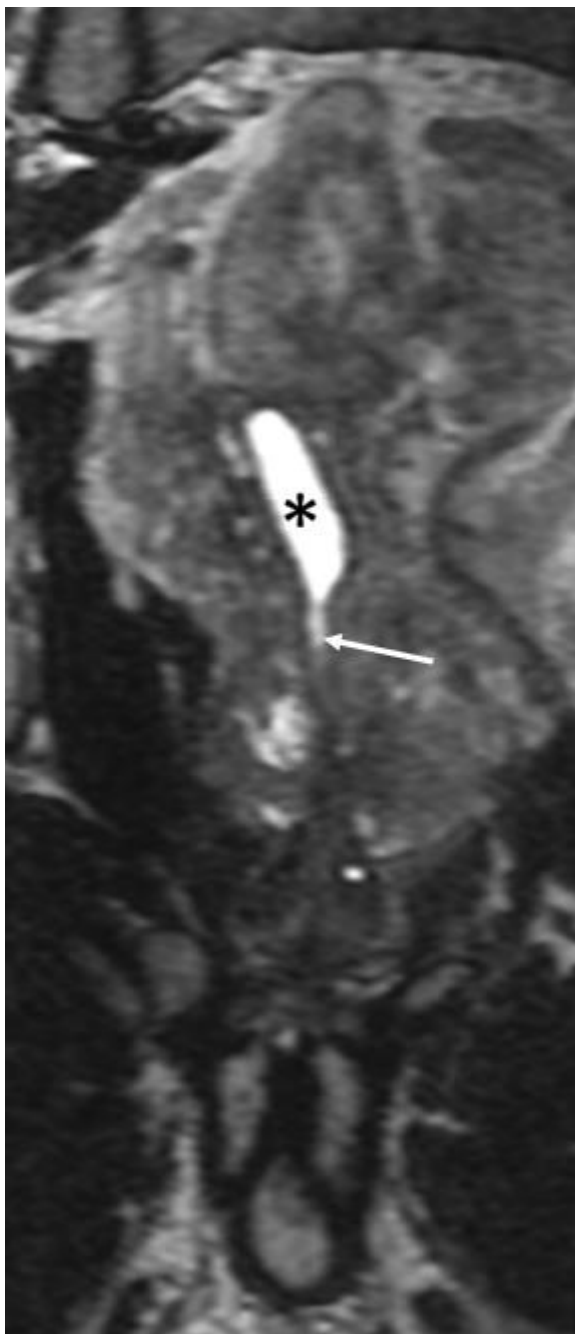


Figure 3: A 36-year-old male with Persistent Müllerian duct syndrome hernia with hernia uteri inguinalis.

Technique: MRI 1,5T; T2W coronal oblique (TE = 111ms, TR= 2500ms); 0.4 mm slice thickness.

Findings: A coronal oblique reformatted MRI of the pelvis demonstrates a fluid-filled structure representing the upper two-thirds of the vagina (star) and communicates with the right ejaculatory duct (arrow). The lower third of the vagina is absent. Neither seminal vesicles nor ovaries are seen.

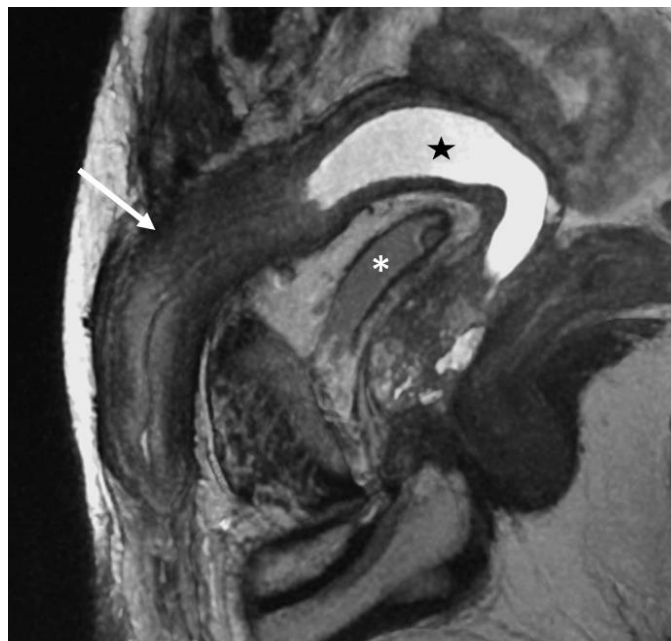


Figure 4: A 36-year-old male with Persistent Müllerian duct syndrome hernia with hernia uteri inguinalis.

Technique: MRI 1,5T; T2W sagittal (TE = 111ms, TR= 2500ms); 1.6 mm slice thickness.

Findings: A sagittal reformatted MRI of the pelvis demonstrates a tubular structure with a signal characteristic and the zonal anatomy of uterine tissue representing the uterus (arrow), which is herniated to the right inguinal canal. It is connected to a fluid-filled structure representing the upper two-thirds of the vagina (star) and communicates with the right ejaculatory duct. The lower third of the vagina is absent. Neither seminal vesicles nor ovaries are seen. Note its relation to the urinary bladder (asterisk).

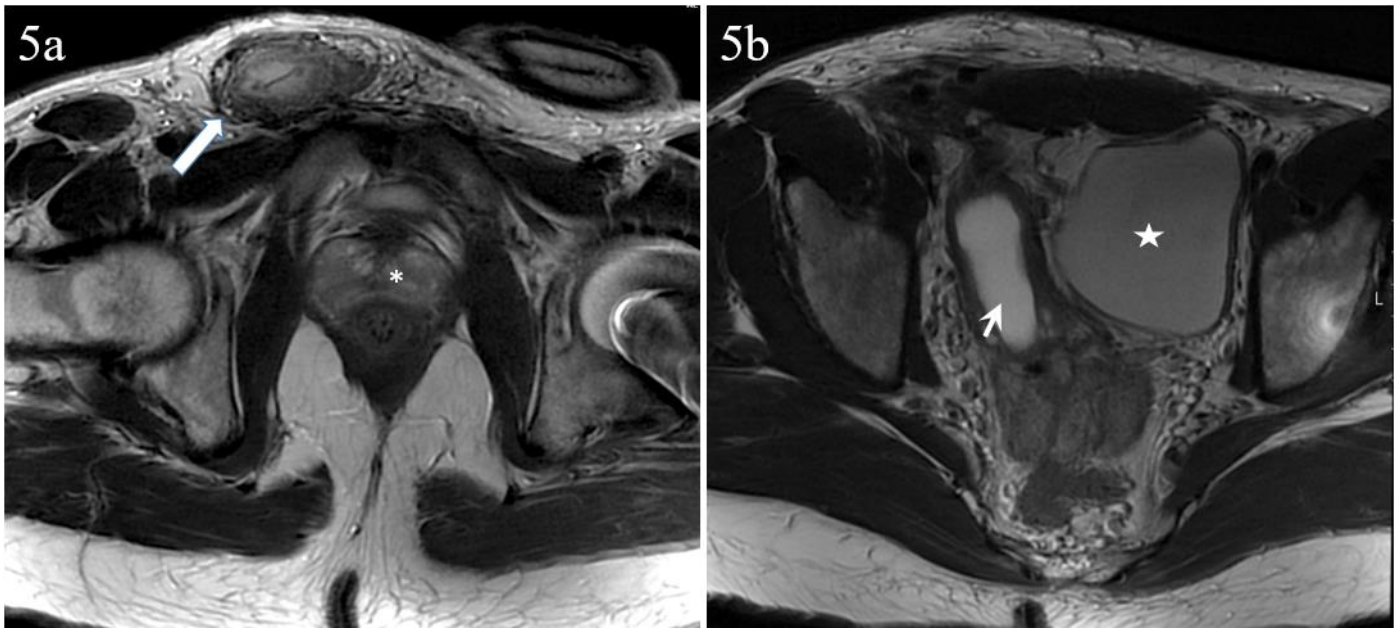


Figure 5: A 36-year-old male with Persistent Müllerian duct syndrome hernia with hernia uteri inguinalis.

Technique: 5a, 5b; MRI 1,5T; T2W axial (TE=100ms, TR=1529); 5mm slice thickness.

Findings:

- a) An axial T2-weighted reformatted MRI of the pelvis demonstrates a tubular structure with a signal characteristic and zonal anatomy of uterine tissue representing the uterus (arrow), which is herniated to the right inguinal canal. Neither seminal vesicles nor ovaries are seen. Note the normal prostate (asterisk).
- b) An axial T2-weighted reformatted MRI of the pelvis demonstrates a fluid-filled structure representing the upper two-thirds of the vagina (short arrow) adjacent to the bladder (star).

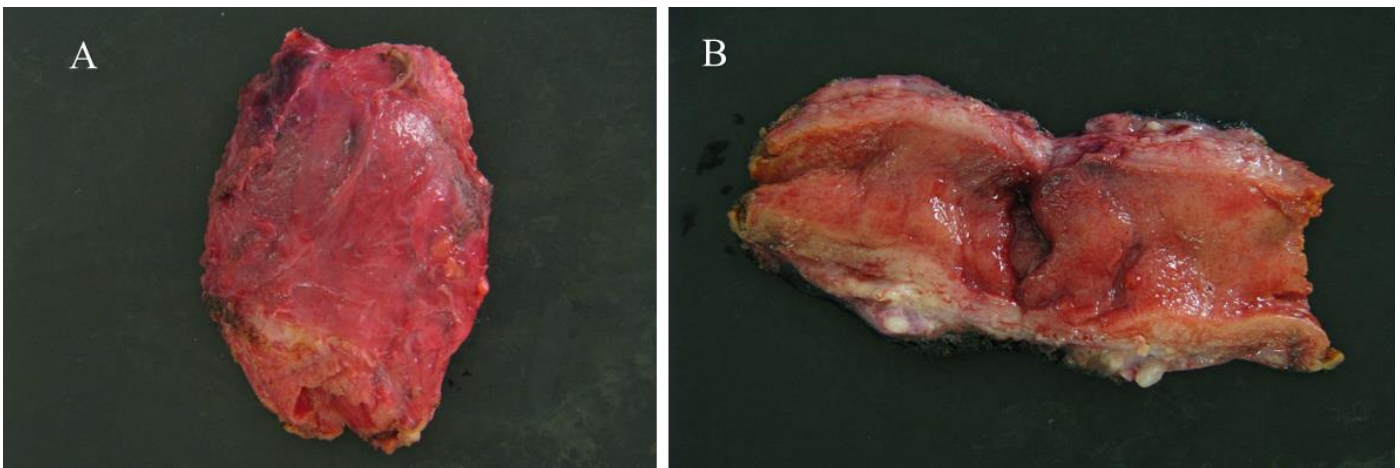


Figure 6: A 36-year-old male with Persistent Müllerian duct syndrome hernia with hernia uteri inguinalis.

Findings: A and B. A gross specimen shows a normal-appearing uterus and endometrium, with no masses identified.

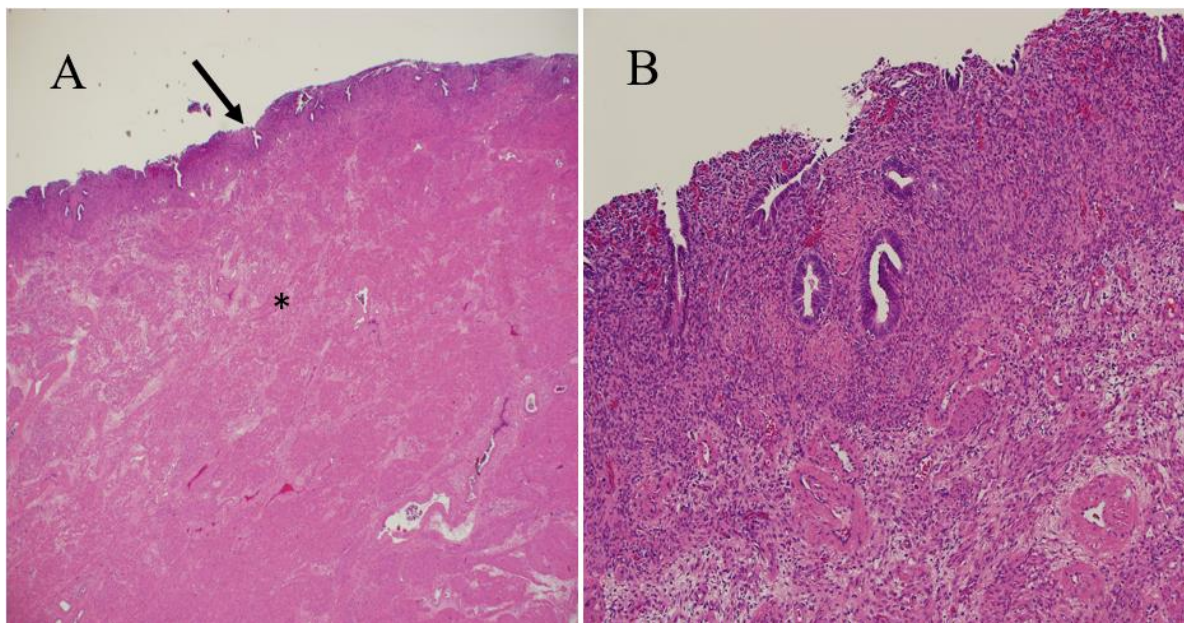


Figure 7: A 36-year-old male with Persistent Müllerian duct syndrome hernia with hernia uteri inguinalis.

Findings:

A. A low-magnification image (hematoxylin & eosin stain; original magnification x4) revealing the uterine wall, which is composed of the endometrial lining (arrow) and a thick myometrium (asterisk).

B. Higher magnification photomicrographs (hematoxylin & eosin stain; original magnification x10) depicting the endometrium, which is composed of endometrial glands surrounded by endometrial stroma.

| | |
|----------------------------|---|
| Etiology | <ul style="list-style-type: none"> • Persistence of Müllerian derivatives (uterus, fallopian tubes, and upper one-third of the vagina) in a normal virilized 46XY male [3]. • Caused by defect in intrauterine release of AMH (anti-Müllerian hormone) [1]. • Transmitted in most cases as autosomal recessive trait [2]. |
| Incidence | Rare condition, incidence is unknown. Fewer than 300 cases reported in literature [3, 5, 7]. |
| Gender ratio | Unique condition affecting males only [3]. |
| Age predilection | PMDS is a surprise finding during orchiopexy or inguinal hernia repair. Most of the cases are discovered during childhood [8]. |
| Classification | <p>There are two anatomic variants of PMDS:</p> <p>A. Male form: unilateral cryptorchidism with contralateral inguinal hernia (80-90%), which is further divided into two sub-categories [4]:</p> <ol style="list-style-type: none"> Hernia uteri inguinalis: descended testes with herniation of Müllerian structures through the ipsilateral inguinal canal [4]. Transverse testicular ectopia: herniation of both testes and of Müllerian structures on the same hernia sac [5]. <p>B. Female form (10–20%): uterus is present in the pelvic cavity, with bilateral cryptorchidism, and the testes are fixed in the round ligament [4].</p> |
| Risk factors | Family history of PMDS. [7] |
| Treatment | Surgical intervention, orchiopexy, and excision of Müllerian derivatives (if possible) to preserve fertility and prevent malignant transformation [6, 7]. |
| Prognosis | If untreated, there is possible risk of malignant transformation of undescended testes and retained Müllerian structures [7]. |
| Findings on imaging | <p><u>Scrotal ultrasound:</u> Ultrasound of the scrotum demonstrates a blind-ending fluid-filled tubular structure seen within the scrotum.</p> <p><u>Abdomen and pelvis CT:</u> A blind-ending fluid-filled tubular structure arising just posterior to the urinary bladder and extending into the right inguinal canal; it has the zonal anatomy of the uterus.</p> <p><u>Abdomen and pelvis MRI:</u> Tubular structure with signal characteristic and zonal anatomy of uterine tissue representing the uterus, and fluid-filled structure representing part of the vagina. The uterus is herniated to the right inguinal canal, and the lower third of the vagina is absent.</p> |

Table 1: Summary table of persistent Müllerian duct syndrome (PMDS).

| Differential diagnosis | Cause | US | CT | MRI |
|---|--|--|---|---|
| Persistent Müllerian duct syndrome – Male form (Hernia uteri inguinalis) | Caused by defect in intrauterine release of AMH (anti-Müllerian hormone) [1]. | Blind-ending fluid-filled tubular structure seen within the scrotum. | Blind-ending fluid-filled tubular structure arising just posterior to the urinary bladder and extending into the inguinal canal; with zonal anatomy of a uterus. | Tubular structure with signal characteristic and zonal anatomy of uterine tissue representing the uterus, and fluid-filled structure representing part of the vagina. The uterus is herniated into the inguinal canal, and the lower third of the vagina is absent. |
| Inguinal hernia (Herniated small bowel loops) | Indirect inguinal hernia: Failure of obliteration (persistence) of <i>processus vaginalis</i> causes it to pass laterally into the inferior epigastric vessels. Direct inguinal hernia: Due to a defect in the Hesselbach triangle, it passes medially into the inferior epigastric vessels [18]. | Abnormally moving intra-abdominal contents of variable echogenicity “mesenteric fat, bowel, or both” through the deep inguinal ring and inguinal canal [17]. | Contrast material-filled bowel loop herniated through the inguinal canal [16]. | MRI is not used to diagnose inguinal hernias; however, it is recommended that patients with a clinically suspected inguinal hernia, which was not revealed by US or CT, undergo MRI [19]. |
| Mixed gonadal dysgenesis (MGD) | Abnormal Chromosomes: 45XY/XO –Mosaic. Presence of testes and streak gonads [14]. | A rudimentary uterus or fallopian tube can be seen, with the streak gonad on one side and testes on the contra-lateral side [14]. | CT of the pelvis is not advocated for the pediatric population (the age group at which most of the gonadal anomalies present) due to inherent risks associated with high doses of radiation [15]. | Although streak gonads can appear as low signal intensity stripes on a T2 weighted MRI sequence, they can still be difficult to detect [14]. |
| Scrotal cystocele | Herniation of the urinary bladder into the scrotum (scrotal cystocele) due to injury [20]. | Presence of a fluid-filled structure that joins the urinary bladder intra-abdominally, with volume changes and wall thickening of the herniated part after micturition [20]. | Pointing of the herniated portion of the urinary bladder, both anteriorly and inferiorly, toward the hernia side [20]. | Pointing of the herniated portion of the urinary bladder, both anteriorly and inferiorly, toward the hernia side [20]. |

Table 2: Differential diagnosis for persistent Müllerian duct syndrome (PMDS).

ABBREVIATIONS

AMH: Anti-Mullerian Hormone
CT: Computed Tomography
MGD: Mixed gonadal dysgenesis
MIS: Mullerian Inhibiting Substance
MRI: Magnetic Resonance Imaging
PMDS: Persistent Müllerian Duct Syndrome
US: Ultrasound

KEYWORDS

PMDS; Persistent Müllerian duct syndrome; Hernia uteri inguinalis; Müllerian duct; Wolffian duct; Scrotal ultrasound; Abdominal CT; Abdominal MRI

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