



## Functional Medicine

# Persistent Mullerian Duct Syndrome Presenting in an Incarcerated Recurrent Inguinal Hernia with Hydrocele



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## ABSTRACT

Hernia uteri inguinalis (HUI) is one of the rarest causes of male pseudo-hermaphroditism worldwide. We report the case of a 49-year-old male with discovery of this anomaly during inguinal hernia repair.

A 49-year-old man presented to the clinic for recurrent inguinal hernia with enlarging left scrotum consistent with hydrocele on imaging. Upon exploration of the left groin, the left testis was pulled up into the abdomen, revealing a uterus, fallopian tube, and a second atrophic testis.

Despite the rarity of HUI, the differential diagnosis for inguinal hernia with associated cryptorchidism and/or hydrocele should include this rare form of pseudohermaphroditism.

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## Introduction

Hernia uteri inguinalis (HUI) is one of the rarest causes of 46 X, Y Disorder of Sexual Development (DSD) worldwide. To have a recurrent inguinal hernia containing such a rarity is an opportunity to review the DSD and the proper management of such disorders.

## Case presentation

A 47-year-old male, with three children, presented to the general surgery clinic for evaluation of a bulge in his left groin. Patient was referred for surgical evaluation where history and physical exam were consistent with left incarcerated inguinal hernia. He subsequently underwent uneventful robotic transabdominal preperitoneal (TAPP) repair with mesh. Two years later, the left groin bulge returned, this time with associated scrotal swelling. Emergency department computed tomography (CT) scan revealed left inguinal hernia containing soft tissue density material. An additional soft tissue density was seen anterior to the hernia sac and it was unclear whether this was related to the testes or was a separate pathology. Patient was referred to general surgery where upon physical exam, no right testis was palpable. A large right hydrocele displacing both

testes leftward was visualized on formal testicular ultrasound. Urology was consulted for evaluation and management of the hydrocele, and was elected to be addressed at time of hernia repair.

The patient underwent open repair of recurrent left inguinal hernia. A left oblique groin incision was made and the dissection was carried down to the inguinal ligament. A normal appearing left testis was pulled into the groin incision for evaluation of hydrocele. In conjunction with the testis, a mass was seen associated with the spermatic cord, between normal appearing left testis and separate appearing atrophic testis (Fig. 1). This medial mass appeared congruent with a small uterus, fallopian tube, and smaller ovarian/atrophic testis appearing structure. The mass was excised and the specimen was sent for frozen consultation. Atrophic testis (pathology of the testis consistent with seminiferous tubules and sertoli cells, but lacking characteristic cells of spermatogenesis), benign endometrial-like cavity consistent with uterus, and fallopian tube were histologically identified. The inguinal hernia repair was performed in the typical Bassini fashion with mesh without complications. Further discussion regarding the atypical findings was had with the patient who reported a similar presentation in his brother who was reportedly infertile. Our patient's karyotype was noted to be 46, XY.

## Discussion

## Embryologic development

In normal embryologic development, sexual dimorphism occurs until week seven where the differentiation and inhibition of

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**Figure 1.** Photo taken from the right side of the patient looking caudally. Intraoperative findings revealing a mass associated with the spermatic cord, between normal appearing left testis and separate appearing atrophic testis. This mass appeared congruent with a small uterus, fallopian tube, and smaller ovarian/atrophic testis appearing structure.

Wolffian (male) or Mullerian (female) structures occurs. The presence of SRY on the Y chromosome leads to differentiation of gonad into testis. The presence of Sertoli cells allows for release of MIS aka AMH (Mullerian Inhibiting Substance or Anti-Mullerian Hormone). This hormone leads to degeneration of Mullerian duct derivatives while the presence of Leydig cells, secreting testosterone, allows for differentiation of Wolffian duct structures including epididymis, vas deferens, seminal vesicles, and ejaculatory ducts.

It is this cascade that is altered in individuals with Persistent Mullerian Duct Syndrome, a rare genetic condition leading to the presence of Mullerian duct derivatives in phenotypically male individuals. It is hypothesized that PMDS is the result of MIS/AMH deficiency, receptor mutation, or pure lack of MIS expression in utero.<sup>1</sup>

#### Gonadal dysgenesis

The umbrella term gonadal dysgenesis is used to describe pathology associated with variation in development of sexual differentiation. Persistent Mullerian Duct Syndrome (PMDS), Transverse Testicular Ectopia, and Mixed Gonadal Dysgenesis, among others, are included in this class of pathology.

Three etiologies of PMDS have been described in previous literature. Type one (45%), hernia uteri inguinalis, presents as cryptorchidism with associated inguinal hernia and is attributed to AMH deficiency. Type two (40%), is attributed to AMH receptor defect. The remaining 15% present with PMDS due to unidentifiable cause.

A separate and rare congenital anomaly first described by Von Lenhossek in 1886, Transverse Testicular Ectopia or testicular pseudoduplication, is the presentation of both testicles in the same hemiscrotum. TTE is classified as type 1, accompanied by hernia, type 2, accompanied by Mullerian duct structures, or type 3, associated with other anomalies.<sup>2</sup> Our patient presented as a type 2 with the presence of uterus and fallopian tube along spermatic cord.

Mixed Gonadal Dysgenesis (MGD) is a syndrome also characterized by persistent Mullerian duct structures resulting in an abnormal testis and a contralateral streak gonad, typically in the setting of 45 X/46 XY, or 46 XY karyotype. Incomplete and abnormal testicular descent typically results in asymmetric internal and external genitalia. Other presentations of bilateral streak gonads (46 XY pure gonadal dysgenesis) and bilateral abnormal testes (dysgenetic male pseudohermaphroditism, now termed 46 X, Y Disorder of Sexual Development) have been proposed to be incorporated into one term of MGD.<sup>3</sup> The diagnosis of MGD is often made in the neonatal period, as the abnormal external genitalia can be easily identified. However, in the cases of PMDS, external genitalia can appear seemingly normal and it is only until the abdominal cavity is entered or abnormal structures are incidentally found on imaging for other reasons that this diagnosis is considered.

#### Appropriate follow-up

Two topics regarding management of PMDS arise. First, what is the appropriate follow-up for any individual with history of abnormal testicular descent to ensure adequate screening for increased risk of malignancy? The United States Preventative Services Task Force (USPSTF) does not currently recommend screening for testicular cancer in adolescent or adult men; however, it has not reviewed the evidence for routine screening in males with the history of cryptorchidism. Though the risk of malignancy in cryptorchid testes is 2.75–8 times the normal risk, some studies have shown no increase in malignancy in patients who chose orchipexy over orchiectomy at mean follow-up of 61.4 months.

Second, what is the appropriate surgical management in patients found to have Mullerian duct structures intraoperatively? Shalaby et al studied 19 phenotypically male, 46 XY patients with PMDS. This small study concluded Mullerian duct structures should be prophylactically removed to avoid the risk of malignant transformation.<sup>4</sup>

#### Conclusions

Albeit a rare finding, surgeons should be aware of PMDS when managing phenotypically male patients with unilateral or bilateral cryptorchidism.

#### Conflict of interest

The authors have no conflict of interest.

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