

CASE REPORT

PERSISTENT MULLERIAN DUCT SYNDROME: A CASE REPORT

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ABSTRACT: A 3-year-old boy presenting with right inguinal hernia was found to have uterus, fallopian tube and testes in a position analogous to ovaries in hernial sac, warranting a diagnosis of persistent Mullerian duct syndrome (PMDS). PMDS is an extremely rare form of internal male pseudo-hermaphroditism in which female internal sex organs, including the uterus, cervix and proximal vagina, persist in a 46XY male with normal external genitalia. The condition results from a congenital insensitivity to anti-Mullerian hormone, or lack of anti-Mullerian hormone, leading to persistence of the female internal sex organs in a male. Clinically, this condition is associated with cryptorchidism. Controversy persists regarding the appropriate treatment of PMDS, since resection of the remnant structures is associated with potential morbidity, but retention risks development of occasional malignancies. We review the literature and discuss various aspects of pathophysiology, diagnosis, and management of PMDS.

KEYWORDS: Persistent Mullerian duct syndrome, Anti-Mullerian hormone, Mullerian inhibiting factor, Cryptorchidism.

INTRODUCTION: Persistent Mullerian duct syndrome (PMDS) is a rare form of internal male pseudohermaphroditism in which Mullerian duct derivatives are seen in a male patient.^[1] The syndrome is caused either by an insufficient amount of Mullerian inhibiting factor (MIF) or due to insensitivity of the target organ to MIF.^[2] The diagnosis of PMDS is often established during operative treatment of associated abnormalities such as inguinal hernia and undescended testis, when a uterus and/or fallopian tube is found along with undescended testis in a genotypically and phenotypically normal male.^[3]

CASE REPORT: A 3-year-old boy was referred to the general surgery clinic for right groin swelling. Physical examination demonstrated reducible swelling in groin and well-appearing male external genitalia with empty scrotum. The abdominal examination was entirely unremarkable. The patient was diagnosed with right-sided inguinal hernia, and cryptorchidism. General routine laboratory investigations done were found to be normal.

Following informed consent from the parents, surgery was done. A 2-cm surgical incision was made at the right groin crease. On exploration, hernial sac containing uterus with fallopian tubes, upper vagina, and testes were present. Total excision of uterus & fallopian tube was done, biopsy from testis was taken, then it was mobilized and orchidopexy was done.

Biopsy from gonad showed seminiferous tubules and epididymal structures were seen with pseudostratified epithelium. No morphologic evidence of intraepithelial germ cell neoplasia or testicular neoplasm was noted.

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Figure 1: Intraoperative photograph showing hernial sac containing uterus, fallopian tubes, and testes.



Figure 1

Figure 2: Biopsy of testes showing seminiferous tubules.

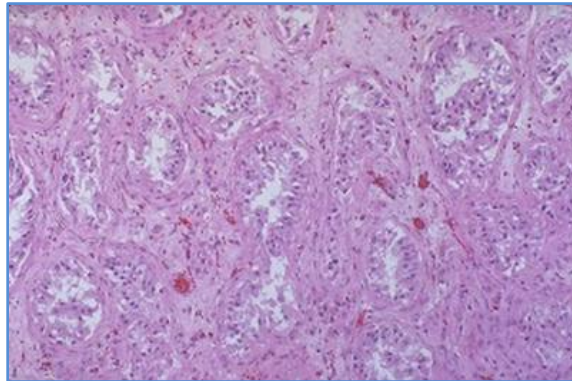


Figure 2

DISCUSSION: PMDS is a rare condition, with only about 150 cases being described in the literature. [4] In a human fetus the Mullerian and Wolffian ducts are both present at 7 weeks of gestation. In a male fetus, the testis differentiates by the end of the 7th gestational week. Normal sex differentiation is controlled by testosterone, dihydrotestosterone, and MIF. Sertoli cells secrete MIF, which leads to regression of the Mullerian ducts. Testosterone has a direct effect on the Wolffian ducts, and promotes their differentiation into the epididymis, vas deferens, and seminal vesicles. [3] Dihydrotestosterone induces male differentiation of external genitalia. PMDS patients have both Wolffian and Mullerian duct structures due to a deficiency of MIF.

Since the secretion and action of testosterone is not affected, the Wolffian duct derivatives and the external genitalia of the fetus progress in the normal male direction. An intersex condition is therefore not usually suspected. But the malformation is incidentally detected during operative treatment of associated abnormalities such as an inguinal hernia or an undescended testis, generally in the first year of life. Henceforth, the diagnosis of PMDS is often established when a uterus and/or

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fallopian tube is found along with undescended testis in a genotypically and phenotypically normal male child.^[3]

Clinically, PMDS cases are divided into three categories;

- The most common is the male type in which one testis is usually found within the scrotum; the ipsilateral uterus and fallopian tube are either in the inguinal canal or can be brought into it by gentle traction on the presenting testis.
- The second type is crossed testicular ectopia, which is characterized by herniation of the testes, the entire uterus and both the fallopian tubes.
- The least common form, or female type, is characterized by bilateral cryptorchidism with testes embedded in the broad ligaments in an ovarian position with respect to the uterus, which is fixed in the pelvis. It is seen in only 10-20% of the cases.^[5]

A review of the literature shows that in patients with intraabdominal testes (as in our case), both the gonads maybe located in a position analogous to the ovaries, with a rudimentary uterus in the centre and the mullerian remnants preventing the mobilization of the testes.^[6]

Manjunath et al. ^[6] reported two cases of PMDS (familial), one with bilateral intraabdominal testes and the other having hernia uteri inguinale with Transverse Testicular Ectopia (TTE).

In PMDS, the testes are usually histologically normal, apart from lesions due to longstanding cryptorchidism. The overall incidence of malignant transformation in these testes is 18%, similar to the rate in abdominal testes in otherwise healthy men.^[7] Germ cell tumors have been reported in the testis, whereas tumors of the Mullerian duct derivatives are very rare. ^[8] Intraoperative methods of diagnosis, especially the gonadal biopsy, can be performed to rule out mixed gonadal dysgenesis and developing malignancy.^[6,9]

In cases of abdominal undescended testis, where a two-stage Stephen Fowler procedure is contemplated, excision of mullerian remnants may be hazardous for the collateral circulation of the testes. Midline splitting of the mullerian remnants and excision of the mucosa are advocated to allow orchidopexy. However, as no malignancy occurs in the retained mullerian ducts, hysterectomy should not be performed.^[6,9] A conflict exists whether orchidectomy should be performed as orchidopexy offers only limited protection against future malignancy.

In our case, the PMDS was of the crossed testicular ectopia, with bilateral cryptorchid testis located in a position analogous to the ovaries, with a rudimentary uterus in the center and bilateral fallopian tubes and mullerian remnants.

To conclude, PMDS is a rare form of male pseudohermaphroditism often encountered unexpectedly at surgery for cryptorchidism or inguinal hernia, diagnosis is enhanced by a high index of suspicion.

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