

PERSISTENT MÜLLERIAN DUCT SYNDROME IN A CHILD: SURGICAL MANAGEMENT

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Persistent müllerian duct syndrome (PMDS) is a rare form of male pseudohermaphroditism in which uterus and fallopian tubes are present in a genotypic and phenotypic male. Over 150 cases have been reported in the literature, mostly in adults.^{1,2} PMDS is likely to be encountered during surgery for undescended testis and inguinal hernia, and surgeons should be aware of this disorder and the options of surgical management. We report a case of PMDS and discuss the etiology, pathogenesis, anatomic types and surgical management.

Case Report

A three-month-old Saudi male infant was referred to the Pediatric Surgery clinic for bilateral cryptorchidism. His parents were not related, and there was no family history of undescended testis. On examination, both testes were undescended and impalpable. The penis and scrotum were normal in appearance. A pelvic ultrasound (US) had shown absence of testes and no other abnormality. Human chorionic gonadotrophin stimulation (HCG) test confirmed the presence of testicular tissue.

At seven months of age, there was a palpable left testis within an inguinal hernia. A month later at surgery, the uterus, fallopian tubes and both testes were unexpectedly found within the hernia sac (Figure 1). Testicular biopsies were performed and the testes, uterus and fallopian tubes returned into the pelvis, followed by herniorrhaphy. Postoperatively, chromosome analysis confirmed a normal male 46, XY karyotype, and a pelvic US showed a uterus. Histology of the testicular biopsies revealed immature testes consistent with cryptorchid testes.

At the age of 15 months, the patient underwent bilateral proximal salpingectomies, leaving the fimbria with each epididymis, corporeal hysterectomy and pedicles of myometrium left with the vasa deferentia. The cervix was split longitudinally in the midline to achieve successful

bilateral orchidopexy after mobilizing the internal spermatic vessels. Postoperative follow-up after one year showed both testes in the scrotum to be normal.

Discussion

PMDS is a rare form of male pseudohermaphroditism in which uterus and fallopian tubes are present in a normal male, XY karyotype. Over 150 mostly adult cases have been reported in the literature, nevertheless, with early surgery for undescended testis and inguinal hernia in children, the incidence is most likely to rise.

By the 8th week, the fetal testis produces two hormones which are necessary for sexual differentiation in the male—testosterone and müllerian-inhibiting factor (MIF). Testosterone produced by Leydig cells has a direct effect on the Wolffian ducts to differentiate into epididymes, vasa deferentia, and seminal vesicles and also regression of the lower vagina. MIF, also known as anti-müllerian hormone, is a glycoprotein produced by the Sertoli cells and causes regression of the müllerian duct structures—uterus, fallopian tubes and upper vagina.³⁻⁵ The proposed etiologies for PMDS are deficiency of MIF, abnormality in its receptor, or failure to produce MIF before the 8th fetal week.^{5,6}

PMDS is considered an inherited disorder with a gene mapped on the short arm of chromosome 19.⁷ The mode of transmission is considered genetically heterogenous.⁸ MIF levels may be normal in the serum or testes (MIF positive) or nondetectable or low (MIF negative). In the former, receptor defects may occur, and this also explains the reports of typical pedigrees of X-linked inheritance.^{9,10} The presence of consanguinity in some cases and the occurrence of PMDS in several pairs of brothers suggest autosomal recessive male-restricted transmission.⁸

PMDS is usually discovered incidentally at the time of orchidopexy or inguinal hernia repair in children. The anatomic types are classified as either “male” or “female.” The “male” type is the most common and is also known as hernia uteri inguinalis. One testis is usually in the scrotum, the uterus and fallopian tube being pulled into the canal by traction on the undescended testis. The contralateral testis and the fallopian tube may also appear in the hernia sac, as in our patient. The “female” type is associated with both

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FIGURE 1. Findings at operation: uterus, fallopian tubes and testes in left inguinal hernial sac.

testes embedded in the broad ligament in a position analogous to the ovaries. In both types, the vasa deferentia are firmly tethered to the lateral walls of the uterus. PMDS is present in 30%-50% of all cases of transverse testicular ectopia.^{3,11}

When PMDS is suspected, karyotyping and HCG stimulation test should be done to confirm chromosome 46, XY and testicular tissue, respectively. Pelvic US, although associated with false-negative results, as in our case, may be performed to detect the presence of a uterus. An unusual hard mass in a hernia found on clinical examination should be evaluated by US before hernia repair. A homogeneous well-defined echogenic pyriform mass larger than a testis may be the uterus.¹² Since there is usually a cystic structure on the anti-mesenteric side of the testis, testicular biopsies are necessary to differentiate PMDS from ovotestis.

The surgical management of PMDS is still controversial. PMDS is usually discovered incidentally during surgery for undescended testes and inguinal hernia repair, therefore, a staged procedure is the most viable option. The first procedure should be testicular biopsies, replacement of the testis, uterus and fallopian tubes in the pelvis, and herniorrhaphy. After PMDS has been confirmed by investigations, definitive surgery can then be performed. The surgical approach by Guerrier et al. of bilateral proximal salpingectomies leaving fimbriae with the epididymes, corporeal hysterectomy and bilateral orchidopexy is our preferred procedure.⁶ Although there is a definite risk of injury to the vasa deferentia and the testes during the removal of the uterus, leaving pedicles of myometrium and the fimbriae attached to the epididymes decreases this risk. In fact, removal of the uterus helps in a successful orchidopexy. Although no malignancy of the müllerian structures has been reported, uterine hypertrophy may cause abdominal mass and discomfort.⁸ Orchidopexy requires mobilization of the internal spermatic vessels, which may be short in some patients. Successful orchidopexy may require division of these vessels in some cases.¹ In situations where the testis cannot be mobilized

enough to reach the scrotum, orchidectomy should be performed.² Cross-orchidopexy is required in transverse testicular ectopia.⁶

There have been recent reports of the use of laparoscopy in PMDS.^{1,11} Laparoscopy has a role in the management of PMDS and may be used for testicular biopsies, orchidopexy and herniorrhaphy. To date, there has not been a report of laparoscopic hysterectomy in PMDS.

Preservation of the testis in these children is important because testicular biopsies often show features consistent with undescended testis without PMDS. In contrast, adult PMDS testis reveals hypoplasia.¹ The risk of testicular malignancy in PMDS is 5%-15%, and no different from patients with cryptorchidism.² There have been reports of different types of malignancy, such as seminoma, teratoma, yolk sac carcinoma, embryonal carcinoma and choriocarcinoma in postpubertal patients.¹⁰ Infertility is frequent in PMDS because most are azoospermic.²

In conclusion, PMDS is a rare form of male pseudohermaphroditism which is usually discovered incidentally during an inguinal hernia repair or orchidopexy. The investigations include karyotyping, HCG stimulation test, pelvic US and testicular biopsies. The most preferred surgical procedures are bilateral proximal salpingectomies, leaving fimbriae attached to the epididymes, corporeal hysterectomy and bilateral orchidopexy. Orchidectomy is reserved for the patient whose testis cannot be mobilized into the scrotum. The patient's family should be aware of the possible risk of testicular malignancy and infertility. PMDS is a genetic disorder, and genetic counselling should be offered to the parents of affected patients.

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