Case Report

Persistent Mullerian Duct Syndrome With an Irreducible Inguinal Hernia

Mehrdad Mohammadi Sichani, Mitra Heidarpour, Asghar Dadkhah, Mehran Rezvani

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Azahra Hospital, Isfahan University of Medical Sciences, Isfahan, Iran

Corresponding Author:
Mehrdad Mohammadi Sichani, MD
Department of Urology, Azahra Hospital, Isfahan University of Medical Sciences, Isfahan, Iran
Tel: +98 311 261 4561
E-mail: m_mohammadi@med.mui.ac.ir

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INTRODUCTION

Persistent mullerian duct syndrome (PMDS), or *hernia uteri inguinale*, describes a group of patients with a 46,XY karyotype and normal male external genitalia, but internal mullerian duct structures. Typically, these phenotypic males have unilateral or bilateral undescended testes, bilateral fallopian tubes, a uterus, and an upper vaginal draining into a prostatic utricle. The condition is commonly diagnosed after mullerian tissue is encountered during inguinal herniorrhaphy or orchidopexy.[1] PMDS is a form of male pseudohermaphroditism. When it presents through an inguinal hernia, it is referred as hernia uteri inguinale.[2] Persistent mullerian duct syndrome results from either a deficiency of antimullerian hormone (AMH) activity or by an abnormality in its receptor. Antimullerian hormone, produced by fetal testicular Sertoli cells, is responsible for the involution of embryonic mullerian structures in normal males.[2] Approximately, 150 cases of PMDS have been reported, whereas hernia uteri inguinale is even scarcer.[3] Familial association has been found in some cases.[4] The condition may occur sporadically or be inherited as an X-linked, autosomal dominant, or autosomal recessive pattern.[5] In transverse testicular ectopia, both testes descend through the same inguinal canal into the same scrotal sac. Persistent mullerian duct syndrome associated with transverse testicular ectopia is much rarer.[6] We report a case of PMDS with liomyoma incidentally found during surgery for irreducible inguinal hernia.

CASE REPORT

A 55-year-old man was admitted with the diagnosis of irreducible right inguinal hernia. The patient had got married 20 years earlier. He had no sexual dysfunction and was fertile. He had 3 daughters and 1 son. His son had hypospadiasis. On examination, the secondary sex characters were found to be well developed. The patient had normal masculine features, moustache, beard, pubic and axillary hair, and penis. The right testis was well developed, with hernia descending to the external ring. The left scrotum was normal. The hernia was irreducible, tense, and nontender.

After general anesthesia, an inguinal incision was made. The spermatic cord was delivered. The peritoneal sac was recognized and dissected off from the cord. A standard herniorrhaphy was performed. In addition, we encountered a solid

mass parallel to the cord, approximately 6 × 3 cm in diameter. This structure was tapered in both sides to a fibrotic band; in one side, it had been adhered to the scrotum and had been fixed to the epididymis and in the other side, it had been adhered to the pelvic cavity. After dissection, the typical figure of a uterus and 2 mullerian ducts were found (Figure 1). The mass comprised of mullerian duct derivatives with rudimentary uterus in the center (Figure 2). Postoperative period was uneventful. Histologic examination revealed uterus (with a small liomyoma) and normal fallopian tubes. Karyotyping was 46,XY.

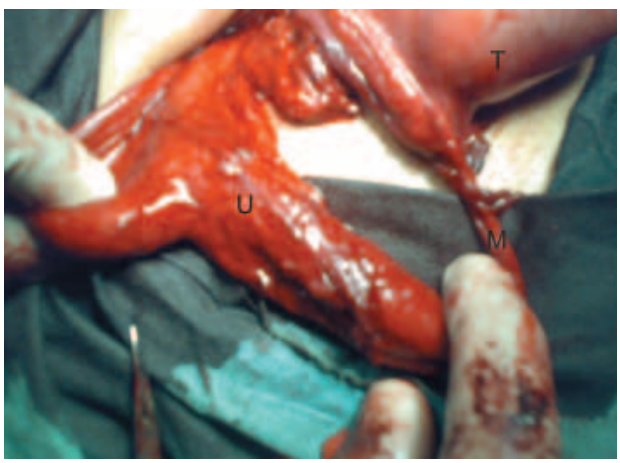

Figure 1. A typical picture of the uterus and mullerian ducts in contact with the testis in persistent mullerian duct syndrome. U indicates uterus; T, testis; and M, mullerian duct.

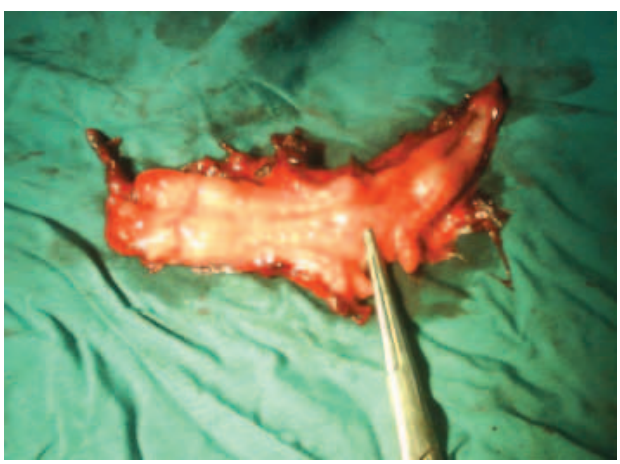

Figure 2. Resected uterus and mullerian ducts. Histopathology examination confirmed the clinical diagnosis.

DISCUSSION

Mullerian duct derivatives are present in male fetuses until the 8th week of gestation. Thereafter, it regresses by AMH, a glycoprotein produced by fetal Sertoli cells. The human gene for AMH has been mapped to chromosome 19.[3] Absent of AMH or abnormal AMH, as well as defects in its receptor, causes persistence of mullerian duct derivatives in male fetuses. Persistent mullerian duct syndrome is characterized by a normal 46,XY karyotype and normal masculinization of the external genitalia.[1,3] Antimullerian hormone does not have a direct role in the descent of the testes. Serum levels of AMH remain fairly high until 2 years of age; measurable levels persist until puberty and then become undetectable.[5] In this case, its level could not be assessed as the patient was 55 years old. 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.[5] There have been reports of embryonal carcinoma, seminoma, yolk sac tumor, and teratoma in patients who have PMDS.[4] Malignancy arising from the mullerian remnants is also reported (2 cases of adenocarcinoma arising from the remnant uterus in English language literature).[7]

There are three anatomic variants: (1) in the most common male type, one testis is usually found within the scrotum; the uterus and ipsilateral fallopian tube are either in the inguinal canal or can be brought into it by gentle traction on the presenting testis.[3] (2) In some cases, the contralateral testis and tube are also in the hernia sac; transverse testicular ectopia can also occur. [6] (3) 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.[1]

Correct management of PMDS requires recognition of the condition by the surgeon and confirmation with testicular biopsies and chromosomal studies. Distinguishing PMDS from other intersex disorders is critical. A karyotyping and assessment of testicular response to chorionic gonadotropin stimulation are essential to verify both genetic sex and the existence of functional testicular tissue.[8] As in this case, the diagnosis

is often made incidentally during surgery for an inguinal hernia or during exploration for cryptorchidism.[2] Transverse testicular ectopia should be suspected preoperatively in patients who have unilateral inguinal hernia associated with a contralateral nonpalpable testis.[9] In suspected cases, ultrasonography, computerized tomography, magnetic resonance imaging, and laparoscopy may be helpful in diagnosis. [1,10] Before puberty in patients with bilateral cryptorchidism, serum AMH levels also helps in diagnosis. The initial procedure consists of testicular biopsies, herniorraphy, and replacement of the gonads and mullerian remnants within the pelvis.[3]

After confirmation of the diagnosis, definitive surgery consists of removal of the mullerian remnants with orchiopexy or orchiectomy.[3] In cases with transverse testicular ectopia, crossed orchiopexy gives good results, particularly if it is performed with minimal delay.[4] Every effort should be made to preserve fertility and hormonal function, in patients less than 2 years of age.[11] Although orchiopexy, even if performed early in life, does not reduce the risk of malignancy, it provides a palpable location for the early detection of malignancy. Preservation of the mullerian derivatives is incompatible with successful orchiopexies because with sexual maturation, the uterus may become hypertrophic and cause discomfort, or may present as a mass whose origin is unknown.[3] In this case, the characteristic macroscopic appearance gives sufficient evidence of the PMDS .We found 123 case reports on PMDS. The most common presenting symptoms were inguinal hernia, undescended testis, testis tumor, and abdominal mass. Surgeons dealing with hernia should consider the possibility of PMDS, especially when it is associated with cryptorchidism.

CONFLICT OF INTEREST

None declared.

REFERENCES

1. David AD. Sexual differentiation: normal and abnormal. In: Wein AJ, Kavoussi LR, Novick AC, Partin AW, Peters CA, editors. Campbell-Walsh urology. 9th ed. Philadelphia: Saunders; 2007. p. 3826-7.
2. Turaga KK, St Peter SD, Calkins CM, Holcomb GW 3rd, Ostlie DJ, Snyder CL. Hernia uterus inguinale: a proposed algorithm using the laparoscopic approach. Surg Laparosc Endosc Percutan Tech. 2006;16:366-7.
3. di Clemente N, Belville C. Anti-Müllerian hormone receptor defect. Best Pract Res Clin Endocrinol Metab. 2006;20:599-610.
4. Berkmen F. Persistent mullerian duct syndrome with or without transverse testicular ectopia and testis tumours. Br J Urol. 1997;79:122-6.
5. Shamim M. Persistent Mullerian duct syndrome with transverse testicular ectopia presenting in an irreducible recurrent inguinal hernia. J Pak Med Assoc. 2007;57:421-3.
6. Boleken ME, Kaya M, Guran S, Memetoglu ME, Kanmaz T, Yucesan S. Persistent mullerian duct syndrome with transverse testicular ectopia. Int Urol Nephrol. 2007;39:1173-5.
7. Romero FR, Fucs M, Castro MG, Garcia CR, Fernandes Rde C, Perez MD. Adenocarcinoma of persistent mullerian duct remnants: case report and differential diagnosis. Urology. 2005;66:194-5.
8. Dekker HM, de Jong IJ, Sanders J, Wolf RF. Persistent Mullerian duct syndrome. Radiographics. 2003;23:309-13.
9. Chandrasekera SK, Barber NJ, Sheriffdeen AH. Persistent Mullerian duct syndrome with transverse testicular ectopia. Urology. 2003;62:1120.
10. Otsuka T, Nakazono T, Matsuo Y, Kanou T, Masaki Z, Kudo S. Persistent Mullerian duct syndrome suggested by magnetic resonance imaging. Radiat Med. 2003;21:277-9.
11. Brandli DW, Akbal C, Eugsster E, Hadad N, Havlik RJ, Kaefer M. Persistent Mullerian duct syndrome with bilateral abdominal testis: Surgical approach and review of the literature. J Pediatr Urol. 2005;1:423-7.