

## Case Report

# Persistent Mullerian duct syndrome with transverse testicular ectopia presenting in an irreducible recurrent inguinal hernia

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### Abstract

Persistent Mullerian duct syndrome (PMDS), a form of male pseudohermaphroditism, is rare. This is a case report of 40-years-old, an otherwise normal male, presenting with Mullerian duct derivatives and transverse testicular ectopia (TTE) in the sac of irreducible recurrent inguinal hernia.

### Introduction

Persistent Mullerian duct syndrome (PMDS) is a rare syndrome and is a form of male pseudohermaphroditism; the Mullerian duct derivatives (uterus, fallopian tubes and upper vagina) are present in an otherwise normally differentiated male with a 46,XY karyotype.<sup>1</sup> When it presents through an inguinal hernia, it is referred as hernia uteri inguinale (HUI).<sup>2</sup> PMDS results from either a deficiency of antimullerian hormone (AMH) activity or by an abnormality in its receptor; AMH, produced by foetal testicular Sertoli's cells, is responsible for the involution of embryonic mullerian structures in normal males.<sup>2</sup> Approximately 150 cases of PMDS were reported, whereas HUI is even rarer.<sup>3</sup> Familial association has been found in some cases.<sup>4</sup> Cases are usually discovered during surgery for an inguinal hernia or cryptorchidism, or by the presence of transverse testicular ectopia (one of the rarest form of testicular ectopia). In transverse testicular eclampsia (TTE), both testes descend through the same inguinal canal into the same scrotal sac; PMDS associated with TTE is much rarer.<sup>5</sup> A case of PMDS with TTE, incidentally found during surgery of recurrent irreducible inguinal hernia, is presented here with a review of literature.

### Case report

A 40 years old male, was admitted with the diagnosis of irreducible recurrent left inguinal hernia. He was operated 7 years back for reducible left inguinal hernia. The patient was married for 11 years. He had no sexual dysfunction. He had primary sterility. His 2 elder brothers had also primary infertility, but his youngest brother was fertile.

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 scrotum was well developed on the left side, with hernia descending upto the bottom of scrotum. The right scrotum was poorly developed with non-palpable, undescended testes. The hernia was irreducible, tense and tender, but the bowel sounds were present.

After spinal anaesthesia, an inguinoscrotal incision was made. The sac and its contents were delivered into the wound, and external oblique aponeurosis was opened. The sac was incised and a triangular mass, 8 cm x 5 cm in size was seen (Figure 1). The mass comprised of Mullerian duct derivatives with rudimentary uterus in the centre, fallopian

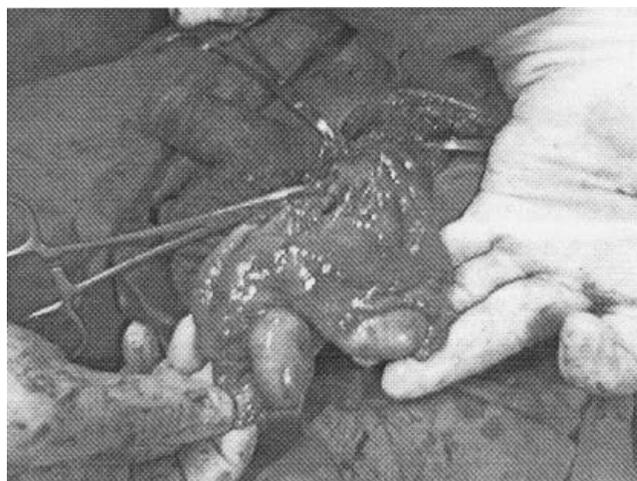


Figure 1. Peroperative view showing PMDS with TTE in hernia uteri inguinale.



Figure 2. Resected specimen showing PMDS with transverse testicular ectopia.

tubes at its upper lateral angles, testis at the lateral end of tubes and left round ligament. The whole mass was removed enbloc (Figure 2). Factors favouring decision of orchidectomy were bilateral cryptorchidism, tumour age of patient and infertility; any hormonal deficiency could safely be replaced. Hernioplasty was performed, and the post-operative period was uneventful. Virilization was maintained in the 8-months follow-up period, without any hormonal replacement.

Preoperative and postoperative semen analysis gave the same results: of azoospermia. Histopathology revealed atrophic uterus with normal fallopian tubes. Both the masses in the right and left sides of the uterus were underdeveloped testes, having no sign of malignancy. The Barr body count was 17 (border line); Karyotyping was 46XY.

### Discussion

Mullerian duct derivatives are present in male foetuses until 8th week of gestation. Thereafter, its regression occurs mediated by AMH, a glycoprotein produced by foetal Sertoli's cells. The human gene for AMH has been mapped to chromosome 19.<sup>3</sup> Absent or abnormal AMH as well as defects in its receptor causes persistence of mullerian duct derivatives in male foetuses; PMDS is characterized by a normal 46 XY karyotype and normal masculinization of external genitalia.<sup>3,6</sup> AMH does not have a direct role in the descent of the testes. Therefore, it is likely that the mechanical effect of the persistent mullerian duct structures produces cryptorchidism by preventing normal testicular descent.<sup>7</sup> It seems possible that this mechanical effect also leads to both testicles being located on the same inguinal side, producing TTE, as in this case.<sup>4</sup> Serum levels of AMH remain fairly high until 2 years of age; measurable levels persist until puberty and then become undetectable. In this case, its level could not be assessed as the patient was 40 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 normal men; there have been reports of embryonal carcinoma, seminoma, yolk sac tumor, and teratoma in patients who have PMDS.<sup>4</sup> Malignancy arising from the mullerian remnants is also reported; clear cell adenocarcinoma arising from the remnant uterus.<sup>8</sup>

There are three anatomic variants; vasa deferentia are intimately adherent to the lateral walls of the uterus and course along the cervix in all three 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.<sup>3</sup>

2. In some cases, the contralateral testis and tube are also in the hernia sac; transverse testicular ectopia can also occur.<sup>5</sup>
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.<sup>9</sup>

Correct management of PMDS requires recognition of the condition by the surgeon and confirmation with testicular biopsies and chromosomal studies. As in this case, the diagnosis is often made incidentally during surgery for an inguinal hernia or during exploration for cryptorchidism.<sup>6</sup> TTE should be suspected preoperatively in patients who have unilateral inguinal hernia associated with a contralateral nonpalpable testis.<sup>7</sup> In suspected cases, ultrasonography, computerized tomography, magnetic resonance imaging and laparoscopy may be helpful in diagnosis.<sup>1,10</sup> Before puberty in patients with bilateral cryptorchidism, serum AMH levels also helps in diagnosis.

The initial procedure consists of testicular biopsies, herniorrhaphy/hernioplasty, and replacement of the gonads and mullerian remnants within the pelvis.<sup>3</sup> After confirmation of the diagnosis, definitive surgery consists of removal of the mullerian remnants with orchiopexy or orchiectomy.<sup>3</sup> In cases with TTE, crossed orchiopexy gives good results, particularly if it is performed with minimal delay.<sup>4</sup> Every effort should be made to preserve fertility and hormone function, in patients less than 2 years of age. Although orchiopexy, even if performed early in life, does not reduce the risk of malignancy, but it provides a palpable location for the early detection of malignancy. Before puberty, orchiectomy is indicated only for testes that cannot be mobilized to a palpable location.<sup>4</sup> Preservation of the mullerian derivatives is incompatible with successful orchiopexies because with sexual maturation, the uterus may hypertrophy and cause discomfort, or may present as a mass whose origin is unknown.<sup>3</sup> In this case, the characteristic macroscopic appearance gives sufficient evidence of the PMDS with TTE. A decision to enbloc removal of the mullerian remnants and TTE (bilateral orchiectomy) was made on following grounds: the patient is 40 years old with proven infertility, increased risk of malignancy, recurrent hernia and the availability of hormonal replacement therapy. Androgen replacement therapy should be given to patients who have undergone orchiectomy and to those with low levels of testosterone.<sup>10</sup>

## Conclusion

The surgeons dealing with hernia should consider the possibility of PMDS and TTE, especially when it is associated with bilateral cryptorchidism.

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