Persistent Mullerian duct syndrome: the hidden normal or abnormal anatomy and the value of laparoscopy

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Persistent Mullerian duct syndrome (PMDS) is a rare disorder of male sexual development. It is characterized by the presence of a uterus, fallopian tubes, and upper vagina in an otherwise phenotypically and genotypically normal male. This malformation is usually an incidental finding during the operative treatment of other more common abnormalities such as inguinal hernia or undescended testes. Not uncommonly, it is seen in association with transverse testicular ectopia. This report describes a case of PMDS in association with transverse testicular ectopia diagnosed at the time of laparoscopy for undescended testes. Physicians caring for these patients should be aware of this and surgeons should be familiar with the different surgical options. PMDS should be considered in all cases of bilateral undescended testes. Aspects of diagnosis and management are also discussed. *Ann Pediatr Surg* **9:37–39 © 2013 Annals of Pediatric Surgery.**

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Introduction

Undescended testes are one of the common congenital anomalies in the pediatric age group and although the majorities are palpable and amenable for surgical correction, about 20% are not palpable [1]. The recent advances in minimal invasive surgery have simplified the management of those with impalpable testes [2,3]. Occasionally, however, the procedure is not without surprises. This report describes a case of persistent Mullerian duct syndrome (PMDS) associated with transverse testicular ectopia (TTE) diagnosed laparoscopically. Aspects of diagnosis and management are also discussed.

Case report

A 14-month-old male infant was referred to our hospital because of bilateral undescended testes. Clinically, he was found to have bilateral undescended testes and subcoronal hypospadias. The right testis was palpable in the inguinal canal but the left one was impalpable. His penile length was normal. MRI of the lower abdomen and pelvis showed the right testis in the inguinal canal, but the left one could not be visualized and no other abnormalities could be detected. The child was planned for laparoscopic-assisted orchidopexy on the left and right side. Laparoscopy indicated a closed left internal inguinal ring. The left testis, vas, and vessels were not visualized. There was, however, a uterus-like structure, 3 cm long on the right side, with tubes extending on both sides. The right gonad was seen coming in and out of the right internal ring. Exploration was performed through the right inguinal region. There was a uterus-like structure with two fallopian tubes and gonads on either side (Fig. 1). It was decided to perform bilateral gonadal biopsies and delay the definitive procedure until further evaluation. Postoperatively, his testosterone level was 0.32 nmol/l (normal 0.00–1.0 nmol/l). His chromosomal analysis showed a 46XY pattern. The pathological examination of biopsies of both the gonads indicated normal testicular tissue with fibrosis. The diagnosis of PMDS with TTE was made. Ten weeks later, he underwent bilateral orchidopexy and excision of the uterus, fallopian tubes, and remnant of the vagina, while preserving both vasa and vessels that were running adherent to the uterus. The pathological examination of the resected specimen revealed a rudimentary uterus and fallopian tubes. Postoperatively, he did well and on a 2-year follow-up, he was found to have a right inguinal hernia that was repaired laparoscopically and both testes were palpable in the scrotums. They were of normal size, but the right one was slightly smaller than the left one.

Discussion

The first laparoscopic diagnosis of impalpable undescended testes was made by Cortesi *et al.* [2]. Since then, several series have been published describing the various laparoscopic techniques for both the diagnosis and the treatment of impalpable testes [3]. Besides its advantages as a minimally invasive approach and better cosmetic results, laparoscopy proved to be valuable in diagnosing other unsuspected abnormalities including the diagnosis and treatment of intersex disorders [4,5]. This was the case in our patient, who had an impalpable undescended left testis and, because of laparoscopy, he was found to have PMDS associated with TTE.

PMDS, which is also called hernia uteri inguinal, is a rare congenital abnormality that results from a mutation in the gene encoding anti-Mullerian hormone or by a mutation in its receptors [6]. Embryologically, the fetal testes secrete two hormones. The Leydig cells secrete testosterone, which is necessary for the development of the Wolffian ducts into the epididymis, vas deferens, and

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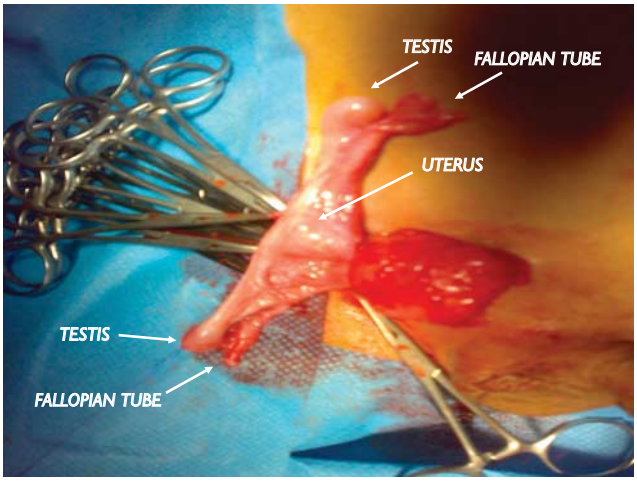
Fig. 1



Intraoperative photograph showing features of persistent Mullerian duct syndrome

seminal vesicles. The Sertoli cells, however, secrete the Mullerian-inhibiting hormone, which causes regression of the Mullerian ducts that usually develop into the uterus, fallopian tubes, and upper third of the vagina [7]. PMDS can be caused by deficiency or failure in the production of the Mullerian-inhibiting hormone or abnormality in its receptors. As a result, the Mullerian ducts fail to regress and develop into a uterus, fallopian tubes, and upper vagina in an otherwise normal male with testicular gonads and 46XY chromosomes [6,8]. The presence of consanguinity in some of the reported cases and its occurrence in several pairs of brothers support an autosomal male-restricted mode of inheritance [8]. Others have suggested an x-linked mode of inheritance [9].

Classically, PMDS is seen in an otherwise normal male with normal external genitalia who presents with unilateral or more commonly bilateral undescended testes and or inguinal hernia. It is also called hernia uteri inguinal because, at the time of hernia repair, a uterus and fallopian tubes may be found in the hernia sac. An association between PMDS and hypospadias has been reported before [10,11]. This was the case in our patient, who also had subcoronal hypospadias. The reason for this association is not known. Most cases of PMDS are diagnosed incidentally at the time of surgery for an inguinal hernia or undescended testes. Rarely, the diagnosis is suspected preoperatively during evaluation of undescended testes. There is, however, a strong association between PMDS and TTE. PMDS is present in 30–50% of all cases of TTE and in these cases, cross-orchidopexy becomes a necessity [12,13]. Our patient had PMDS associated with TTE.

The surgical management of PMDS is still controversial. As these cases are discovered incidentally, a staged procedure is the most commonly accepted option. During the initial surgery, bilateral testicular biopsies are performed, followed by relocation of the uterus, fallopian tubes, and testes to the pelvis and herniotomy. Once the diagnosis is confirmed, definitive surgery is planned. The confirmation of the diagnosis includes chromosomal analysis, hormonal assay including an HCG stimulation test, and the result of testicular biopsies. There is, however, still a controversy on whether to remove the Mullerian remnants or not. There are those who advocate leaving the Mullerian remnants to avoid injury to the vas deferens and testicular vessels at the time of their resection [11]. In contrast, there are those who strongly recommend their removal [6,14]. Although very rare, there have been two reports of clear cell adenocarcinoma of the remnant uterus in PMDS [15,16]. In addition, it should be kept in mind that the remnant uterus can hypertrophy, causing pain and discomfort, and removal of the uterus facilitates orchidopexy. Expansion of the urinary bladder while being filled with urine can be markedly embarrassed by the pressing uterus from the back and the embracing tubes/vas from the sides after a 'forced' orchidopexy, leading to the eventual ascent of the 'fixed' gonads during follow-up.

The most commonly performed procedure is bilateral proximal salpingectomy, leaving the fimbriae with the epididymis, hysterectomy, and bilateral orchidopexy [6]. It is important to avoid injury to the vas and vessels at the time of hysterectomy. One way to achieve this is to leave a pedicle of the myometrium and the fimbriae attached to the epididymis. Others advocate splitting the uterus in the midline, bringing the testis with the vas deferens and attaching uterine tissue to the scrotum [17]. In our patient, it was possible to separate the vas deferens from the rudimentary uterus and vagina. We, like others, advocate removing the Mullerian duct remnants. With the recent advances in minimal invasive surgery, laparoscopy is being increasingly used both for the diagnosis and management of PMDS including testicular biopsy, orchidopexy, and herniotomy [10,12,18]. Follow-up of these patients is also important. There is a 5–15% risk of testicular malignancy in these patients, which is not different from that in patients with undescended testes [19,20]. Most of these patients, however, are infertile because of azoospermia, low motility index, or ductal obstruction [21]. It is also important to check the result of testicular biopsy as these usually show testicular tissue with variable degrees of fibrosis that may necessitate testosterone replacement at the time of puberty in those with hypoplastic or fibrotic testes.

Acknowledgements

Conflicts of interest

There are no conflicts of interest.

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