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Persistent Mullerian Duct Syndrome and Hernia Uteri Inguinalis in a Male With Normal Fertility

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ABSTRACT

Nonregression of Mullerian ducts in individuals who are phenotypically male with an XY genotype is a rare occurrence. This condition usually presents as unilateral or bilateral undescended or partially descended testes. Additionally, it is often associated with inguinal swellings. These swellings are caused by an undescended testis and may be bilateral, or they may present as unilateral if both testes are in the same inguinal region (transverse testicular ectopia). Inguinal swellings may also result from embryological remnants of the Mullerian duct. Most males with this disorder have azoospermia. The present report is a very rare case of a 28-year-old male who presented with unilateral inguinal swelling on the left side. Exploration of the inguinal canal on suspicion of an inguinal hernia revealed a hypoplastic uterus, single nodular ovary, and coiled Fallopian tube with an easily discernable fimbrial end. Histology confirmed hernia uteri inguinalis. No testicular tissue could be located in the inguinal canal. The right testis had a normal volume and was descended completely in the scrotum. Quite uniquely, this individual possessed a normal stature, sperm count, and fertility. These characteristics are very rarely documented in association with persistent Mullerian duct systems in men.

INTRODUCTION

Establishment of sexual phenotype in humans begins with the determination of the *chromosomal sex* at the onset of life. The chromosomal sex of a male (ie, the XY genotype) translates into establishment of the *gonadal sex* with formation of the testes. The testes further regulate development of the characteristic *phenotypic sex* in the man [1].

Manifestation of gonadal activity for the development of a particular phenotype is a multi-step process [2]. The testis produces testosterone that regulates development of the Wolffian ducts and male external genitalia. Before the eighth week of gestation, the testis also produces the important Mullerian inhibiting factor (MIF). The MIF restrains development of female internal genitalia, the ovaries at the gonadal ridges, the uterus, and the uterine tubes; it also maintains the kinetics of normal masculine development [3]. MIF functions locally and acts on receptors expressed around the fetal Mullerian tissue [4]. Synthesis of defective MIF molecules or MIF receptors may give rise to varying degrees of virilization of internal or external genitalia in the male [5,6]. Persistence of the Mullerian ducts may manifest as any of the 3 major forms: (1) persistent Mullerian duct syndrome (PMDS); (2) mixed gonadal dysgenesis (MGD); or (3) intersex

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Abbreviations and Acronyms

DMP = dysgenetic male pseudohermaphroditism
DSD = disorder of sexual development
MGD = mixed gonadal dysgenesis
MIF = Mullerian inhibiting factor
PMDS = persistent Mullerian duct syndrome

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case report

disorder of sexual development (DSD), which was previously known as dysgenetic male pseudohermaphroditism (DMP) [7-9]. Infertility is typical, with only 6 reported cases of fertility in males with these conditions to date [7,10].

Male individuals with PMDS present with several anatomical variants. Unilateral or bilateral undescended or partially descended testes with inguinal swellings are typical presentations. Both of the testes may also be located on the same side, at different levels of descent; this condition is called *transverse testicular ectopia*. The Mullerian duct derivatives or remnants may also be pulled inside within the contralateral or ipsilateral inguinal canal; this is termed *hernia uteri inguinalis* [10,11].

Intersex conditions such as PMDS have deep psychological implications. Surgical management in patients with PMDS and allied conditions is dilemmatic and varied, depending on the status of the gonads, degree of development of the Mullerian derivatives, fertility, and age of the patient [12-15]. Although the immature uterus, uterine tubes, and ovaries do not usually turn malignant, they are excised with care to preserve the vas deferens of the undescended testis on that side in order to restore fertility [16,17]. An orchidopexy is simultaneously performed on the undescended testis, if present. Orchidectomy is advised for a testis that cannot be mobilized out of the inguinal canal. It is also recommended for an adult patient when the risk of malignant transformation of the undescended testis is high [18].

CASE REPORT

A 28-year-old male presented with unilateral swelling in his left groin. On physical examination, the swelling felt like a soft, reducible, rounded mass of approximately 5 cm in diameter near the deep inguinal ring. The condition was diagnosed as a left inguinal hernia. The left side of the scrotum felt empty, with a single, well-developed and fully descended testis (volume about 7 mL) on the right side.

The patient had normal stature with a height of 160 cm (63 in) and a weight of 68 kg (150 lbs). He was the father of 2 children. The external genitalia appeared normal and there was no gynecomastia.

Ultrasonography only revealed that the swelling was a single nodular mass near the deep inguinal ring. The examiners suspected that it was an undescended testis.

Surgical exploration of the inguinal canal revealed that the contents were comprised of: (1) a slightly elongated (3 cm x 2 cm) nodular mass (Figure 1), which raised the suspicion of a rudimentary uterus; (2) a coiled cord of about 4 cm attached to the mass at one end; (3) a small, rounded nodule close to the nodular mass. The end of the coiled cord that was located away from the mass resembled the fimbriations at the end of a Fallopian tube. No testis could be detected in or around the explored tissue or within the inguinal canal. The inguinal canal was repaired after the contents of the mass were excised. The deep ring and the part of the abdomen accessible from the deep inguinal ring was thoroughly searched to detect the presence of the missing testis or sequestrated Mullerian remnants.

Histological examination confirmed the presence of: (1) endometrial and myometrial tissue in the elongated nodule; (2) Fallopian epithelium lining the tubular elongated cord; and (3) ovarian follicles and corpora albicans in the small excised node (Figure 2). All remnants of the Mullerian ducts were excised without orchidopexy because no testis could be detected in the canal. The patient was advised to return for long-term follow-up.

Figure 1. Endometrium of the Rudimentary Uterus Showing the Epithelium (top) and the Glands (arrows) (magnification 10x).

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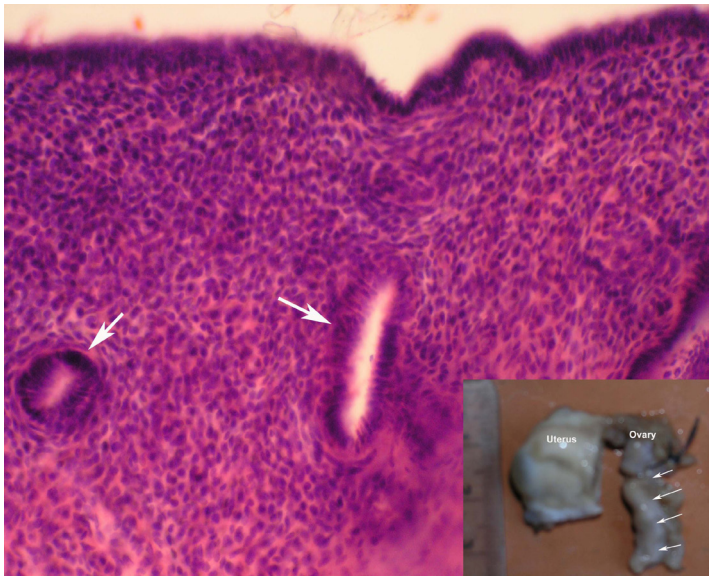

The inset shows the gross appearance of the excised mass with the cord-like Fallopian tube rudiment (arrows).

Figure 2. Section of the Ovoid mass Showing a Corpus Albicans (CA) (within arrows) and Ovarian Follicles (asterisk) (Scanner view 4x).
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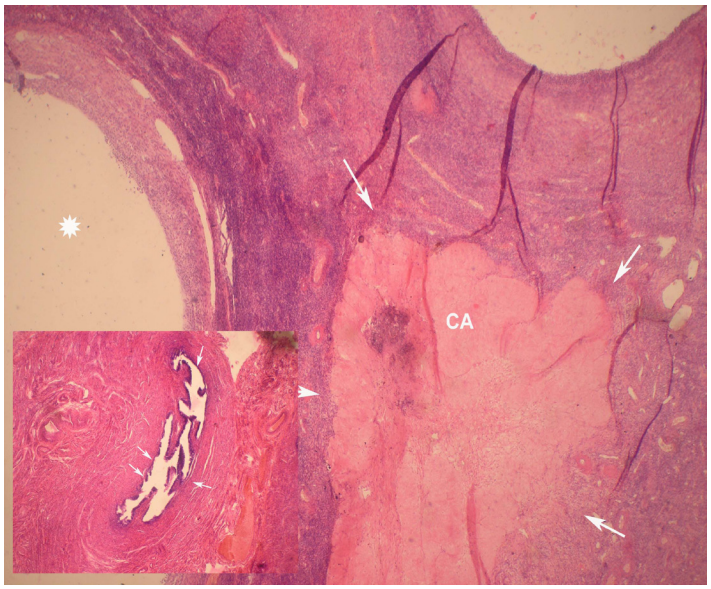

The inset shows the Fallopian tube (demarcated by arrows) with its epithelium, surrounding musculature, and blood vessels (magnification 10x).

DISCUSSION

By definition, PMDS is not associated with malformations of the male external genitalia, as seen in intersex DSD or as streak gonads with MGD. It is difficult to detect PMDS preoperatively, because the condition is associated with a normal penis and scrotum. It is usually detected during surgical management of cryptorchidism or an inguinal enlargement associated with PMDS, as seen in the present case. Reports of fertility in males with persistent Mullerian systems are conflicting, in part because these conditions have been also observed to be familial [18-20]. Azoospermia is the usual feature and may result from dysgenetic testes or chronic cryptorchidism [18]. Evaluation of the patient indicates that these conditions may closely resemble PMDS.

The present case is extremely rare because the patient presented: (1) a well-developed and fully descended testis without any inguinal swelling on the right side; (2) normally developed male external genitalia; (3) well-organized Mullerian derivatives inside the left inguinal canal without the presence of a left testis; (d) normal fertility and sperm count maintained by the right testis. Very few cases of individuals with PMDS and hernia uteri inguinalis and uncompromised fertility have been reported. Exploration of the inguinal region and adjoining abdominal cavity was done to detect the presence of testicular tissue or Mullerian remnants. Because fertility was already established in the patient, there would have been no point to preserving a cryptorchid testis; these structures exhibit greater propensity for malignant transformation when compared with their normal counterpart [11]. Because no testicular tissue could be discovered on the side of the hernia, only the Mullerian tissues were excised completely from the inguinal canal. A recently reported case showed evidence of malignant transformation in the Mullerian remnants of a male [16]. Although a handful of cases of PMDS with uncompromised fertility have been reported in the literature, instances of Mullerian remnants in the male phenotype presenting in middle age as hernia uteri inguinalis with a single normal testis and unimpaired fertility are comparatively rare [21].

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