

Case Report

Persistent Mullerian Duct Syndrome in An Elderly Male: A Rare Entity

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Abstract

Persistent mullerian duct syndrome (PMDS), is an extremely rare variant of male pseudo-hermaphroditism, characterized by the persistence of mullerian ductal structures (uterus/fallopian tubes/vagina) in a phenotypically normal male. PMDS was reported earliest by Lenhossek in 1886 and Nilson in 1939 and subsequently there have been few and far reports between. We present an 82-year-old gentleman, who approached us with a left inguinoscrotal swelling diagnosed clinically as hernia, but when taken up for hernioplasty, revealed the presence of a rudimentary uterus. On retrospective questioning as well as histopathological evaluation, the diagnosis of PMDS was made. The importance of being aware of the existence of this disease entity lies in the fact that a surgeon may encounter a case maybe only once in his surgical lifetime, and an incidental intra-operative discovery makes it a mindboggling condition to manage. Thus, the need to maintain a high index of vigil for timely diagnosis and appropriate surgical management.

Keywords: Persistent mullerian duct syndrome; PMDS; Male pseudo hermaphroditism

Introduction

Persistent mullerian duct syndrome (PMDS), is an extremely rare variant of male pseudo-hermaphroditism, characterized by the persistence of mullerian ductal structures (uterus/fallopian tubes/vagina) in a phenotypically normal male [1]. Earliest reports of PMDS were by Lenhossek in 1886 [1] and Nilson in 1939 [2] and there have been few and far between. We report the case of an 82-year-old gentleman, who presented to us with a left inguinoscrotal swelling diagnosed clinically as hernia, but when taken up for hernioplasty, revealed the presence of a rudimentary uterus. On retrospective questioning as well as histopathological and genetic evaluation, the diagnosis of PMDS was made.

Case

An 82-year-old gentleman presented to the outpatient department, with a left inguino scrotal swelling and pain, since the past few days. He was a normally appearing phenotypic male, with normal secondary sexual characters. Inguino scrotal examination revealed a left sided reducible, direct inguinal hernia and an ectopically palpable right testis superior to the left sided hernia. His penis was normally developed. A CT lower abdomen suggested a left inguinal hernia, with small bowel loops as content and a hyperdense 12 x 9 cm SOL (space occupying lesion) within the left scrotal sac, left sided hydrocoele and an ectopic right testis lying superior to the mass. After all pre-operative basic work up, he was taken up for surgery and underwent a left inguinal exploration, excision of the left sided mass lesion with left sided hernioplasty (Figure 1a, 1b).



Figure 1(A): Intraoperative photograph of the left inguinal mass lesion, (B): Immediate post-operative specimen-cut open.

Histopathology report was consistent with a male hermaphrodite, showing the presence of both uterine (non-secretory endometrium and myometrial leiomyoma) and testicular elements (atrophic testis with leydig cell hyperplasia and bilateral vas deferens) in situ. Post-operative course was uneventful (Figure 2a, 2b, 2c, 2d).

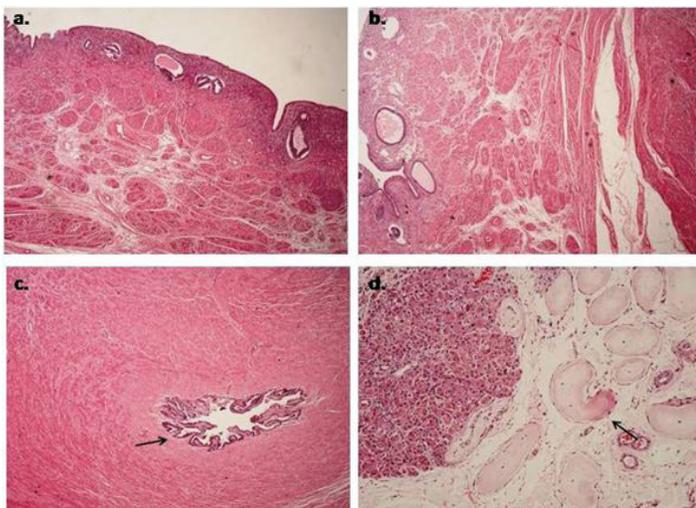


Figure 3a: Endomyometrial elements. b: Endomyometrial elements. c: Fallopian tube (arrow mark). d: Atrophic seminiferous tubules (arrow mark)

Discussion

Persistent mullerian duct syndrome (PMDS) or hernia uteri inguinale, an atypical and rare variant of male pseudohermaphroditism may be inherited as an X linked or autosomal dominant trait or may be sporadic [1-3]. It has been reported in conjunction with other syndromes such as Klinefelter's, Turner's and Meyer-Rokitansky-Kuster-Hauser syndrome [4]. In a male foetus, masculinisation occurs between 7 to 8 weeks of embryonic life [1]. Leydig cells of the testis secrete testosterone which aids differentiation of the Wolffian duct into epididymis, vas deferens and seminal vesicle, whereas the Sertoli cells secrete mullerian inhibiting substance (MIS) to cause regression of the mullerian duct by the 10th week of gestation [1,3,5]. Manjunath et al [3] have classified PMDS cases into three clinical categories:

- Majority (60 to 70%) with bilateral intra-abdominal testes in a position analogous to ovaries.
- Smaller group (20 to 30%), with one testis in the scrotum, associated with contralateral inguinal hernia whose contents are testis, uterus and tubes (classical presentation of hernia uteri inguinale).
- Smallest group (10%) where both the testes are located in the same hernial sac along with the müllerian structures (transverse testicular ectopia - TTE).

PMDS patients are phenotypically normal, have normal puberty and even normal fertility if gonads are in the normal position within the scrotal sacs [5]. In case the testes are abdominal, they may seem in an analogous position to the ovaries, increasing the surgical confusion [4,5]. The confusion stems from the fact that abdominal testes need to be repositioned in a palpably suitable place on account of the increased risk of malignancy [5,6]. Identification of the epididymis and vas deferens would unequivocally confirm the gonad to be a testis and a decision to perform an orchidopexy can be taken during initial exploration itself [1]. However, Loeff et al advocate that a testicular biopsy is needed to rule out mixed gonadal dysgenesis or carcinoma in situ [1,6]. Contrasting surgical views elucidate either a two staged (with gonadal biopsy in the first stage) or a single staged testicular repositioning [6,7]. Testicular autotransplantation was another alternative approach advocated in the past [8].

Berkeman et al [9] had initially opined that there was no need to remove the mullerian remnants as till then there was no case of malignancy reported, however, this has been refuted and malignancies have been reported in isolated cases thereafter, mandating remnant excision. Diagnosis of PMDS is usually as a surprise en-

tity, discovered incidentally during inguinoscrotal surgeries [4,5]. Ultrasound and an MRI have been reported as useful adjuncts to locate the mullerian remnants [7]. However, since most diagnoses are intraoperative or retrospective, their role remains speculative. Whether or not all infants with an undescended testis mandate a diagnostic laparoscopy, remains food for thought [1]. Screening of the siblings and second degree relatives is necessary to aid the diagnosis [1,6]. A retrospective history and clinical examination may yield surprises for the clinician. The importance of identifying PMDS lies in the fact that such patients are prone for various malignancies and a sudden intra-operative discovery enhances the confusion as regards their appropriate management. In this context, case reports discussing about PMDS, with expert management opinions would serve as a timely reminder about its existence as well as would aid to fill up the lacunae in literature.

Conclusion

PMDS is an extremely rare disease entity that needs appropriate and timely diagnosis for apt surgical management. Being so infrequent, this may be encountered only once in a surgical lifetime. Herein lay the importance of being aware of the very existence of this entity. As is rightly said, **“What the mind knows is what the eyes see”**

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