Case Report

Persistent mullerian duct syndrome: A case report and review of the literature

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Abstract

Persistent mullerian duct syndrome (PMDS) is usually an accidental finding either during orchipexy or during routine inguinal hernia repair in male patients presenting with maldescended or crytorchid testes. It is caused by a defect in the mullerian inhibiting substance system. Intraoperatively, mullerian remnants consisting of an infantile uterus and fallopian tubes are usually found. Familiarity with PMDS is necessary to diagnose the condition. We report a case of PMDS in a 14-year-old male presenting with bilateral undescended testes.

Key words: Hernia uteri inguinale, mullerian inhibiting substance, mullerian remnants, persistent mullerian duct syndrome

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Introduction

Intersexual disorders are very important clinical issues with their different aspects relating to diagnosis, treatment and sex of rearing.[1] They are broadly classified into disorders associated with a normal chromosome constitution and disorders associated with an abnormal chromosome constitution.

Persistent mullerian duct syndrome (PMDS) is a form of male intersex caused by a defect in the mullerian inhibiting substance (MIS) system. Patients are phenotypically male and usually present when young with unilateral or bilateral cryptorchid testes and an inguinal hernia into which prolapses an infantile uterus and fallopian tubes.[2] Familial cases have been reported with a probability of sex-limited autosomal recessive or X-linked recessive inheritance. An incidence of PMDS in identical twins has also been reported.[3]

PMDS is often misdiagnosed due to a lack of familiarity with the condition. We report our case to stress the intraoperative diagnosis of this condition with a high index of suspicion on the operating surgeon's part and because of its rarity. A review of the literature showed only about 20 case reports from India including familial cases.

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Case Report

A 14-year-old boy presented with bilateral undescended testes and was diagnosed with intraabdominal testes. He underwent the Fowler Stephen stage I procedure with cauterization of the testicular vessels and, after 6 months, was admitted for the stage II procedure. Preoperative ultrasound examination showed both testes to be of normal size and echo texture, with intact cord vascularity located in the pelvis. Intraoperatively, a diagnosis of mixed gonadal dysgenesis with bilateral oval-shaped gonads and a uterus-like tubular structure was made.

Tissue was obtained on intraoperative consult. Crush smears performed from the tissue showed immature germ cells and few tubular structures but no maturing spermatozoa. The gonads were excised and submitted for histopathological study.

On microscopy, the right gonad showed normal-looking seminiferous tubules with spermatogonia at its various

maturation levels for the age of the patient and a normal-looking epididymis. The left gonad showed the structure of the uterus comprising normal-looking endometrium and myometrium Figure 1: Immature uterus with endometrial lining, inset shows endometrial lining at 450X. Seminiferous tubules and epididymal structures were seen in other bits Figure 2: Seminiferous tubules in the left gonad; Figure 3: Epididymis with pseudostratified epithelium.

No morphologic evidence of intraepithelial germ cell neoplasia or any evidence of testicular neoplasm was noted.

In view of the detailed clinical history and presentation, and in conjunction with the histomorphological features, a possibility of PMDS (hernia uteri inguinalis) was considered.

Genetic testing for MIS system gene mutation, mutations in receptor for MIS gene and sexual karyotyping of the patient was advised.

Discussion

PMDS or hernia uteri inguinale is a rare form of male pseudohermaphroditism characterized by the presence of mullerian duct structures in 46 XY phenotypic males. It is caused by a defect in the MIS system.[4]

The MIS is a large glycoprotein that sertoli cells produce early in fetal life. The gene responsible for the substance is on chromosome 19. MIS is also known as anti-mullerian hormone. The primary function of MIS is to cause regression of the mullerian (paramesonephric) ducts in the male fetus. MIS is first secreted in effective amounts 56-62 days after fertilization, and the process of mullerian regression is normally completed by about Day 77, after which the mullerian tissue is no longer sensitive to MIS.

Another important function of MIS is to initiate testicular descent, principally by its postulated regulatory control over the gubernaculum testis.[2-4] These two functions of the MIS explain the clinical findings of PMDS.

Clinically, PMDS cases are divided into three categories.

- Majority (60-70%) with bilateral intraabdominal testis in apposition analogous to ovaries
- Smaller group (20-30%) with one testis in the scrotum associated 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 hernia sac along with the mullerian structures (transverse testicular ectopia-TTE).

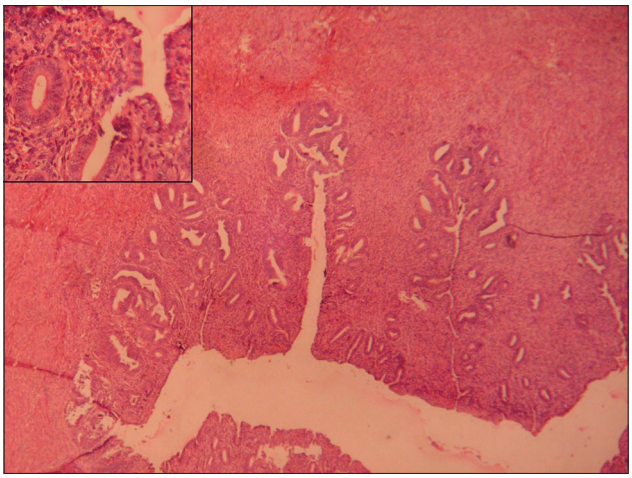

Figure 1: Immature uterus with endometrial lining, inset shows endometrial lining at 450X

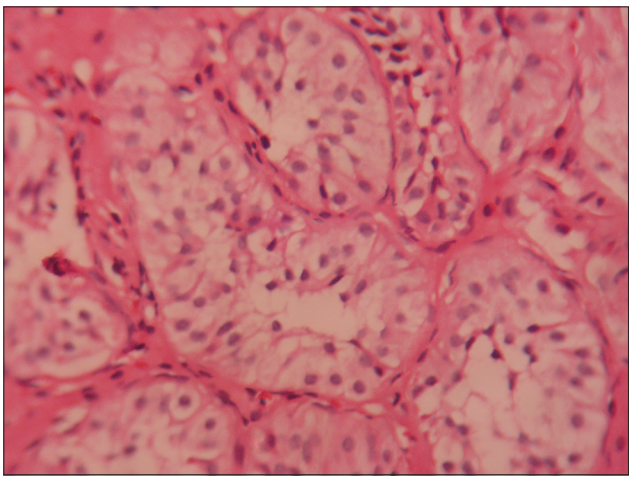

Figure 2: Seminiferous tubules in the left gonad

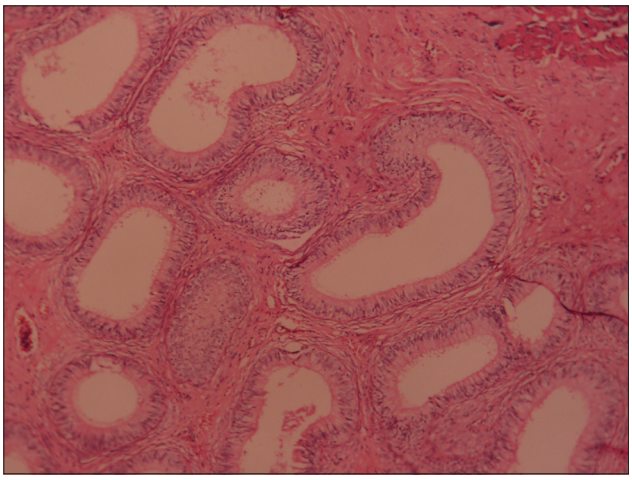

Figure 3: Epididymis with pseudostratified epithelium

A review of the literature shows that in patients with intraabdominal testes (as in our case), both the gonads maybe located in a position analogous to the ovaries, with a

rudimentary uterus in the center and the mullerian remnants preventing the mobilization of the testes.[3]

Manjunath *et al*.[3] reported two cases of PMDS (familial), one with bilateral intraabdominal testes (as in our case) and the other having hernia uteri inguinale with TTE.

Our case did not show any dysplastic changes or malignancy. However, Berkmen[4] reported three cases with PMDS associated with testicular malignancy: Two cases were of testicular seminoma and the remaining one case showed both testicular seminoma and teratoma. Compared with a normally descended testis, the crytorchid testis has a 7-35% increased risk of developing a malignant tumor, especially seminoma. Moreover, the risk of developing a malignancy is greater in an abdominal than in an inguinal testis. Seminoma is the most common histological type in crytorchid patients or in PMDS.

The diagnosis of PMDS is made incidentally during surgical exploration for cryptorchidism or herniorraphy as the mullerian remnants are not palpable on abdominal, rectal or scrotal examination. Intraoperative methods of diagnosis, especially the gonadal biopsy, can be performed to rule out mixed gonadal dysgenesis and developing malignancy.[3,4]

Mixed gonadal dysgenesis is a disorder associated with an abnormal sex chromosome constitution, characterized by ambiguous genitalia with unilateral testis and a streak gonad contralaterally, with persistence of mullerian duct structures on the side of the streak.[2]

Another clinical presentation is that of crossed testicular ectopia or transverse testicular ectopia. It is usually seen to be associated with PMDS. It is a rare congenital anomaly in which both the gonads migrate toward the same hemiscrotum.[5]

In cases of abdominal undescended testis, where a two-stage Stephen Fowler procedure is contemplated, excision of mullerian remnants may be hazardous for the collateral circulation of the testes. Midline splitting of the mullerian remnants and excision of the mucosa are advocated to allow orchidopexy. However, as no malignancy occurs in the retained mullerian ducts, hysterectomy should not be performed.[3,4] A conflict exists whether orchidectomy should be performed as orchidopexy offers only limited protection against future malignancy.

Conclusion

PMDS has an autosomal recessive inheritance. Screening of siblings and second-degree relatives is necessary. Although ultrasonography and magnetic resonance imaging are reported to play a role in locating the mullerian remnants, laparoscopy has a distinctive advantage in diagnosing PMDS. Familiarity of the operating surgeon with this disease condition would increase the chances of correctly diagnosing and appropriately dealing with the mullerian remnants.

References

1. Öçal G. Current concepts in disorders of sexual development. J Clin Res Pediatr Endocrinol 2011;3:105-14.
2. Robboy SJ, Bentley RC, Russell P, Anderson P. Pathology of abnormal sexual development. In: Fox H, Wells M. Haines and Taylor-Obstetrical and Gynecological Pathology. 5th ed. United Kingdom: Churchill Livingstone, Elsevier; 2003. p. 1209-32.
3. Manjunath BG, Shenoy VG, Raj P. Persistent müllerian duct syndrome: How to deal with the müllerian duct remnants- A review. Indian J Surg 2010;72:16-9.
4. Berkmen F. Persistent müllerian duct syndrome with or without transverse testicular ectopia and testis tumours. Br J Urol 1997;79:122-6.
5. Moslemi MK, Ebadzadeh MR, Al-Mousawi S. Transverse testicular ectopia, a case report and review of literature. Ger Med Sci 2011;9:Doc15.

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