Case Report

Persistent müllerian duct syndrome: a case report

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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 cryptorchid testes. It is caused by a defect in the mullerian inhibiting factor. 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 45-year-old male presenting with right inguinal hernia.

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Introduction

Persistent mullerian duct syndrome (PMDS) is a rare form of male pseudohermaphroditism, characterized by the presence of a uterus and fallopian tubesin genotypically and phenotypically normal males owingto failure of mullerian duct regression.[1]

The syndrome is caused either by an insufficient amount of mullerian inhibiting factor (MIF) or due to the insensitivity of the target organ to the MIF. The diagnosis of PMDS is often established during operative treatment of associated abnormalities such as inguinal hernia and undescended testis in a genotypically and phenotypically normal male.[2]

Case Report

A 45 year old male patient came with complaints of right inguinal hernia since 4 months. Hernia was repaired and inguinal canal contents were removed.

On dissection, uterus like structure along with a fallopian tube and mass resembling undescended testis were separated and were submitted for further histopathological evaluation. Grossly, Theuterus measures about 4.5 x 4 x2 cm in size and fallopian tube measures 7 cm. The testis measured 2.5x2x1 cm (Fig. 1 and 2)

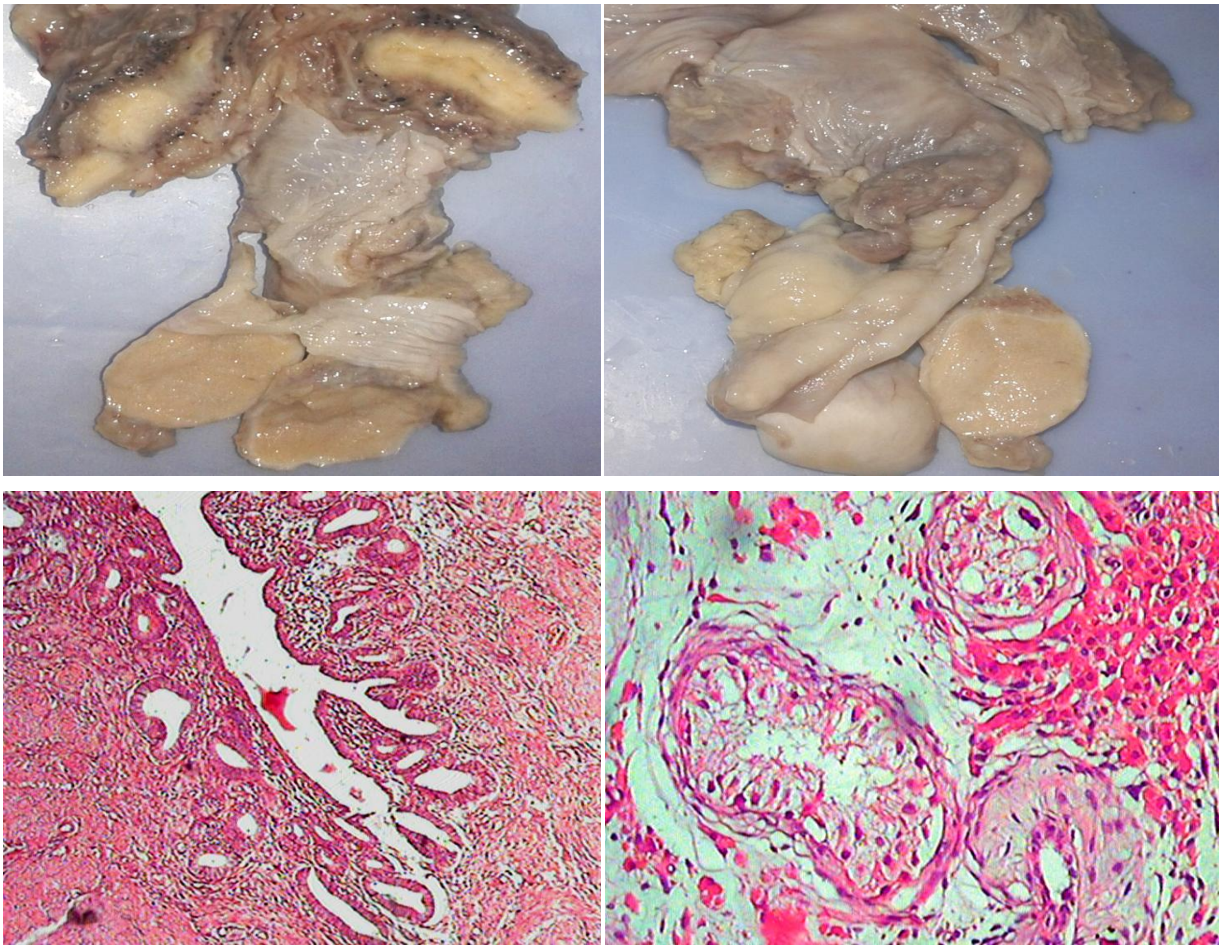

Fig 1-Inguinal hernia contents- Uterus and testis: Fig 2-Inguinal hernia contents- Fallopian tube and testis. Fig 3- Histopathology- Atrophic endometrium and myometrium; Fig 4. Histopathology – undescended testis

The section from the uterus showed atrophic endometrium which measured 0.1 cm in thickness. The myometrium was unremarkable (Fig. 3). The fallopian tube showed normal tubal histology. No ovarian tissue was found. Undescended testis was atrophic with tubular basement membrane thickening and Leydig cell hyperplasia (Fig 4)

Discussion

PMDS is a rare form of male pseudo-hermaphroditism, characterized by the presence of a uterus and fallopian tubes owing to failure of mullerian duct regression in genotypically normal males.[1] Nilson described the condition in 1939 and termed it as hernia uteri inguinale.[3]

American NationalInstitute of Health estimates that there are less than 200000 cases of PMDS in US.Exact incidence in India is not known.[3] In a human foetus, both mullerian and wolffian ducts, the anlagen of the female and male reproductive tracts, respectively, are present at 7-week gestation.

The normal sex differentiation in males is controlled by testosterone and MIF. Testosterone has a direct local effect on the wolffian ducts, including differentiation into the epididymides, vas deferens, and seminal vesicles. Also the formation of the urogenital sinus and male external genitalia requires in situ conversion of testosterone into dihydrotestosterone. Despite the normal male genotype and the subsequent normal development of foetal testis, if there is a failure in production of MIF or insensitivity of the target organ to MIF, Mullerian structures do not regress.

Since the secretion and action of testosterone is not affected, the Wolffian (mesonephric) duct derivatives and the external genitalia of the foetus progress in the normal male direction. An intersex condition is therefore not usually suspected. But the malformation is incidentally detected during operative treatment of associated abnormalities such as an inguinal hernia or an undescended testis, generally in the first year of life. Henceforth, the diagnosis of PMDS is often established when a uterus and/or fallopian tube is found along with undescended testis in a genotypically and phenotypically normal male child.

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 healthy men.[4]

Clinically, PMDS cases are divided into three categories:

1. Majority (60–70%) with bilateral intra-abdominal testisin a position analogous to ovaries.

2.Smaller group (20–30%) with one testis in thescrotum, associated with contralateral inguinal herniawhose contents are testis, uterus and tubes (classicalpresentation of hernia uteri inguinale).

3. Smallest group (10%) where both the testes are located inthe same hernial sac along with the müllerian structures(transverse testicular ectopia - TTE). PMDS accountsfor 30–50% of all cases of TTE.[3]

Our case is a classical presentation of PMDS- hernia uteri inguinalewith one testis in thescrotum, associated with contralateral inguinal herniawhose contents are testis, uterus and fallopian tube.

Conclusion

In cases of unilateral or bilateral cryptorchidism associated with inguinal hernia, as in our patient's case, the possibility of persistent Mullerian duct syndrome should be kept in mind in order to prevent further complications such as infertility and malignant change.

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Competing Interests

None declared.

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