

Case Report

Persistent Mullerian Duct Syndrome (PMDS) Presenting as a Malignant Tumour

Islam MR¹, Ali MS², Azam SMG³

Abstract

Persistent Mullerian duct syndrome is a condition in which there is presence of Mullerian duct structures (uterus, fallopian tube, vagina etc.) in an otherwise phenotypically, as well as genotypically, normal man. This patient usually presents with unilateral or bilateral cryptorchidism associated with inguinal hernia and ectopic testis. There is the chance of developing malignancy in ectopic testis (incidence being 15%), as well as infertility in case of bilateral cryptorchidism. Our patient suffers from PMDS presents with malignant tumour. The aim of the presentation of this case is to draw the attention in case of unilateral or bilateral cryptorchidism associated with or without inguinal hernia; the possibility of PMDS should be kept in mind to prevent infertility as well as malignancy.

Keywords: Persistent Mullerian Duct Syndrome (PMDS).

Number of Figures: 03

Number of References: 05

Number of Correspondences: 03

1. Corresponding Author:
Dr. Md. Rafiqul Islam
FCPS (Surgery)
Senior Consultant
Department of Surgery
General Hospital, Khulna.
e-mail: dr.rafiqrmc23@gmail.com
Mobile: 01711838481
2. Dr. Md. Showkat Ali
FCPS(Surgery)
Junior Consultant
Department of Surgery
General Hospital, Khulna.
3. Dr. S M Golam Azam
MS (Surgery)
Assistant Registrar
General Hospital, Khulna.

Introduction

Persistent Mullerian duct syndrome is a rare form of male pseudo-hermaphroditism characterized by the presence of Mullerian duct structures in an otherwise phenotypically, as well as genotypically, normal man; only a few cases have been reported in the world wide literature. We report

the case of a 22 year old man with bilateral cryptorchidism, presents with a lump at lower abdomen. After laparotomy there was a well developed uterus, both Fallopin tubes, apparently normal left gonad with a large tumour arising from right gonad. He was diagnosed as a case of PMDS.

Case Report

Mr. Sumon, age 22 years was admitted to a private clinic at Khulna city with a complaint of intra abdominal mass. According to the statement of the patient he was alright one month back. Then he developed a mass in the lower abdomen. This is about 15 cm in diameter, smooth surface, freely mobile, non tender. No abnormality in bowel and bladder habit.

On examination the boy was good looking, healthy, masculine in get up (Fig-1).



Figure-1: Facial appearance of the patient.

(Images are taken by mobile phone camera during operation.)

Secondary sex characters are well developed including male genitalia. His scrotum contains soft tissue without definitive testis on either site. All investigations are found within normal limit except Ultrasonography. Ultrasonography shows a large intra abdominal mass at the lower abdomen with mixed echogenicity. He was diagnosed clinically as a case of mesenteric tumour. He was decided for laparotomy. Abdomen was opened through a paramedian incision. A large mass was detected arising from right gonad, uterus and both fallopian tubes (Fig- 2) were found normal, left testis was atrophied and embedded in broad ligament. Upper part of vagina ends on apparently prostatic tissues.



Figure-2: Uterus at the lower abdomen.

Total abdominal hysterectomy with excision of the tumour (Fig-3) was done. Abdomen was closed with a drain. His external genitalia were further examined. Penis and scrotum were well developed, scrotum contains soft tissue like structure, no testis was found. Specimen sent for histopathological examination. He was diagnosed as a case of PMDS.



Figure-3: Specimen of tumour after resection.

Histopathological report shows uterine muscular tissue with cavity lined by atrophied endometrial tissue. Left testis showing atrophy with hyalinised seminiferous tubules with complete arrest of maturation, tumour at right testis shows malignant testicular tumour (germ cell tumour).

Discussion

In true hermaphrodites, both ovarian and testicular tissue is present in one or both gonads⁴. In female pseudo-hermaphroditism, the gonads are the ovaries, but male tendencies are seen in the organ of reproduction⁵. Conversely, male pseudo-hermaphroditism is a condition in which the gonads are the testes but the internal genitalia are not completely virilized. Male intersex may present (1) with masculine external genitalia with fully developed uterus (as in our patient's), (2) with purely feminine external genitalia, or (3) With external genitalia of equivocal sexuality.

The exact cause of PMDS is not known, however it is thought to rest from a defect of the synthesis or release of

MIF, or from defects in the MIF receptor (2). MIF is released by the Sertoli cells in fetal tissue from seven weeks of gestation onwards, and is responsible for the regression of the Mullerian duct in the male fetus.

The female form, seen in 10% to 20 % of cases, is characterized by bilateral cryptorchidism. The gonads are fixed within the pelvis, with the testes fixed within the round ligament in the ovarian position with respect to the uterus³.

PMDS is usually coincidentally detected during surgical operation, as in our patient's case. However pre-operative Ultrasonography, computerized tomography and MRI allow possible pre-operative diagnosis². The prognosis depends upon the integrity of the testicular tissue and successful correction of cryptorchidism.

The risk of malignancy in an ectopic testis in a case of PMDS is similar to that in a healthy male, with the incidence being 15%. There have been case reports of embryonal carcinoma, seminoma, yolk sac tumor and teratoma in patients with PMDS, whereas tumors of the Mullerian duct derivatives are very rare^{1,3}. Infertility is common, with an absence of spermatozoa observed during semen analysis³.

Excision of tumour along with total excision of the uterus with bilateral fallopian tubes and left testis was performed and the operation was completed with a drain in pelvic cavity.

Our patient recovered well post-operatively. The patient was referred to oncologist for further management of cancer and long-term follow-up.

Conclusion

PMDS is a rare form of male pseudo-hermaphroditism characterized by the presence of Mullerian duct structures in an otherwise phenotypically, as well as genotypically, normal man. The patient with PMDS has unilateral or bilateral cryptorchidism and is usually assigned to the male sex at birth without hesitation. Since patients are phenotypically male, the diagnosis is usually not suspected until surgery is performed for cryptorchidism or hernia repair. In cases of unilateral or bilateral cryptorchidism possibility of persistence Mullerian duct syndrome should be kept in mind in order to prevent further complications such as infertility and malignant change.

Acknowledgement

The authors gratefully acknowledge Dr. Dilruba Parvin MBBS, DMU for helping with valuable suggestions and documentation.

References

1. Prakash N, Khurana A, Narula B. Persistent Mullerian duct syndrome. *Indian J Pathol Microbiol.* 2009; 52: 546-548, 10.4103/0377-56160. [View Article PubMed](#) [Google Scholar](#)
2. Yuskel B, Saygun O, Hengirmen S. Persistent Mullerian duct syndrome associated with irreducible inguinal hernia, bilateral cryptorchidism and testicular neoplasia: a case report. *Acta Chir Belg.* 2006; 106: 119-120. [View Article Google Scholar](#) [Google Scholar](#)
3. Renu D, Rao BG, Ranganath K, Namitha. Persistent Mullerian duct syndrome. *Indian J Radio Imaging.* 2010; 20: 72-74. [10.4103/0971-3026.59761](#). [View Article PubMed](#) [Central Google Scholar](#)
4. Ceylan K, Algun E, Genus M, Gonulalan H. True hermaphroditism presenting as an inguinalhernia. *Int Braz J Urol.* 2007; 33: 72-73. [View Article PubMed](#) [Google Scholar](#)
5. Dekke HM, DeJong IJ, Sanders J, K Wolf RF. Persistent Mullerian duct syndrome. *Radiographics.* 2003; 23: 309-313. [10.1148/rg.232025141](#). [View Article PubMed](#) [Google Scholar](#)