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Persistent Mullerian duct syndrome (PMDS): Case report and review of literature

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ABSTRACT

Persistent Mullerian duct syndrome (PMDS) is a rare form of male pseudohermaphroditism (MPH) which characterized by the presence of uterus, fallopian tubes and upper part of vagina in an otherwise normally differentiated 46, XY male. The syndrome is caused by either an insufficient amount of antimüllerian hormone AMH or by insensitivity of the target organ to this factor, It is usually discovered in patient who assessed for infertility, cryptorchidism or during intra-abdominal surgery. Herein We report a rare presentation case of PMDS with intra-abdominal mass which came to be seminoma.

1. Introduction

Persistent Mullerian duct syndrome (PMDS) is a rare form of male pseudohermaphroditism (MPH) which characterized by the presence of uterus, fallopian tubes and upper part of vagina in an otherwise normally differentiated 46, XY male.[1] The syndrome is caused by either an insufficient amount of antimüllerian hormone or by insensitivity of the target organ to this factor.[1] Approximately there are 200 cases reported in the literature.[2] It is usually discovered in patient who assessed for infertility, cryptorchidism or during intra-abdominal surgery.[2] We report a rare case of PMDS with intra-abdominal mass.

2. Case report

A 38-year-old male was referred to our urology clinic from local hospital due to Left side hydronephrosis secondary to abdominal mass.

He's married and has a 5-year-old son a product of IVF. He has no significant medical history.

There was no personal history or family history of cancers.

Upon physical examination, he has a normal pattern of external genitalia, phallus and urethra were normal. However, the patient had empty scrotum, and appeared to be hypoplastic with no palpable testis, no surgical scars.

An abdominal ultrasound scan showed an empty scrotum. While CT scan of an abdominal and pelvic, showed rudimentary uterus, and the left testis is being replaced by a large mass about 8.5 × 8.1 × 8.9 cm (Fig. 1). The mass is inseparable from rudimentary uterus like structure at inferior aspect of side of the mass. With multiple paraaortic lymph nodes at the level of left renal helium, which forming a large mass measuring about 9.3 × 8 × 7.9 cm (Fig. 2).

In MRI findings were suggestive of pelvic germ cell tumor of left testis with left paraaortic lymphadenopathy. The Mullerian remnant representing infertile uterus continues with left seminal vesicle. While the right testis was atrophic. Features overall are suggestive of PMDS.

A chromosomal analysis revealed an apparently normal male karyotype 46, XY, with no numerical or structural aberrations.

Laboratory data showed Alpha-fetoprotein (AFP) Human chorionic gonadotropin (HCG) Lactate dehydrogenase (LDH) were elevated, 3.3 IU/L, 30 IU/L and 490 IU/L respectively. Blood hormonal assay depicted the elevation of FSH 26 IU/L, LH 12.9 IU/L free testosterone 38.38 IU/L, Creatinine 126 μmol/L.

Semen analysis showed low sperm count and accordingly, sperm banking was done for him.

Patient underwent CT guided biopsy to Left para-aortic mass which reported as metastatic lymph node from germ cell tumor consistent with seminoma.

After discussing the case with Oncology team, He initially received standard chemotherapy with bleomycin, etoposide and cisplatin X 4 cycles then underwent a PET CT CAP which demonstrated a pelvic soft tissue mass consistent with seminoma has no significant interval change

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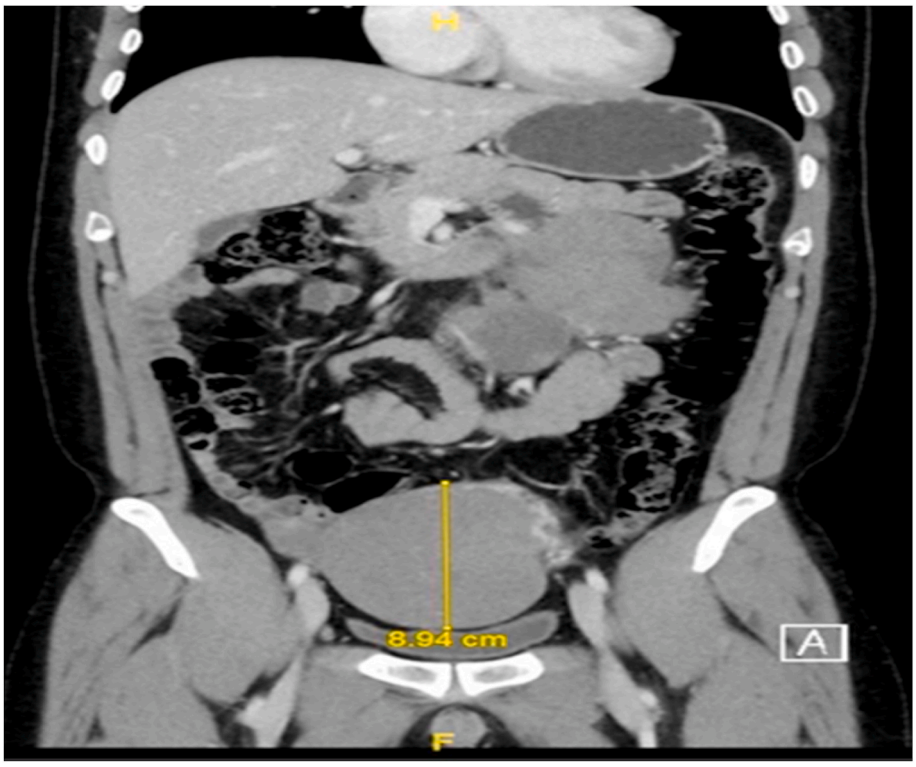

Fig. 1. The left testis is being replaced by a large mass about 8.5 × 8.1 × 8.9 cm, The mass is inseparable from rudimentary uterus like structure at inferior aspect of side of the mass.

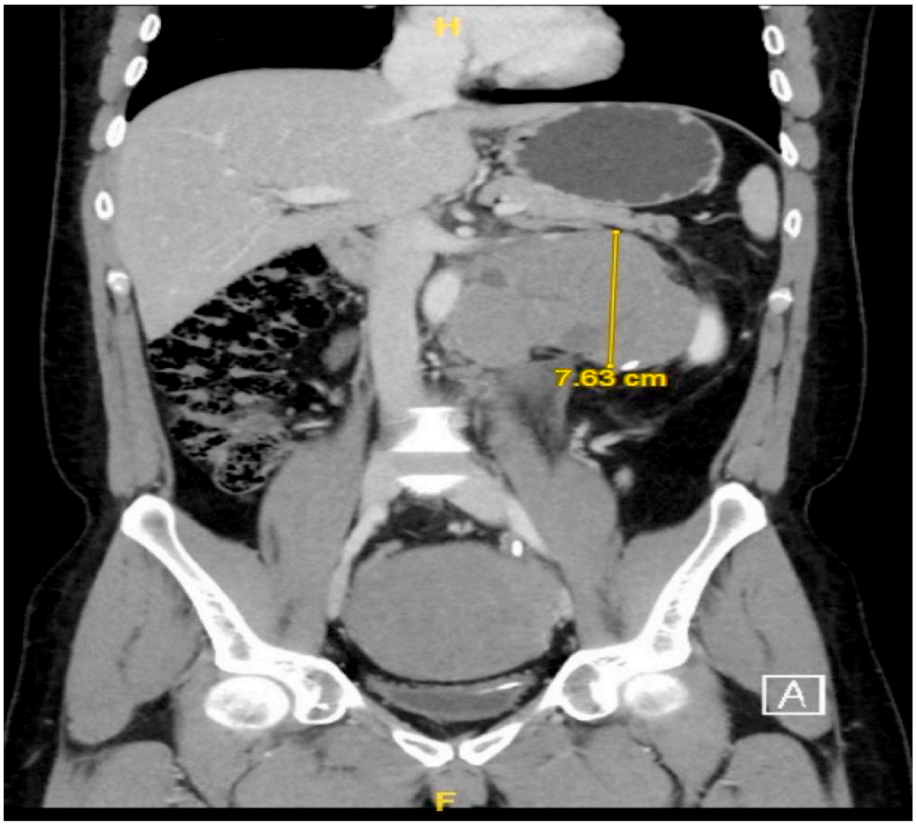

Fig. 2. Multiple paraaortic lymph nodes at the level of left renal helium, which forming a large mass measuring about 9.3 × 8 × 7.9 cm in transverse, anteroposterior and craniocaudal dimension respectively causing compression of left renal collecting system that causing moderate hydronephrosis despite the presence of the left double J stent.

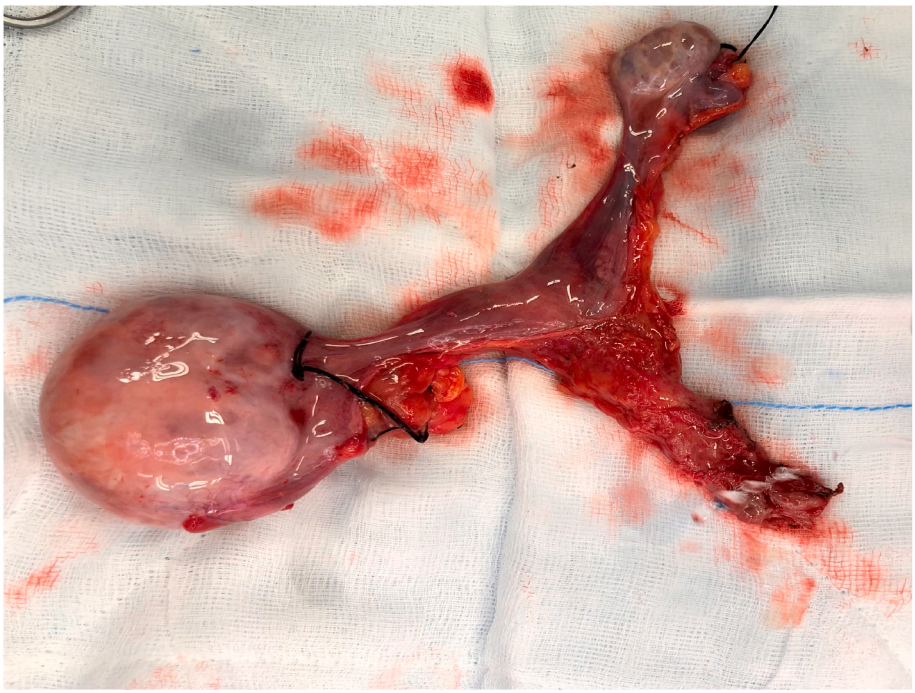

Fig. 3. Rudimentary uterus, and the left testicular mass.

in size compared to previous CT scan (about 5.5 × 4.5 cm). The left paraaortic lymph nodes are measuring about (3.6 × 2.5 × 3.9 cm) with increased heterogeneous metabolic activity. A tumor marker was undetectable, but LDH still remained elevated about 196 IU/L.

A decision was taken to do exploration laparotomy with bilateral orchidectomy, hysterectomy, excision of Left para-aortic lymph node and left nephrectomy (Fig. 3).

Patient tolerated the procedure well and referred to endocrinology for testosterone replacement.

He was discharged home after few days in stable condition.

The pathologic findings reported a diffuse fibrosis of left testis with no residual malignancy. The right testis revealed atrophic seminiferous tubules. Mullerian ducts remnants showed negative for malignancy. Also, Left para-aortic lymph nodes showed no residual of malignancy.

On follow up, abdominal CT scan showed no gross evidence of residual tumor. Tumor markers were also negative.

3. Discussion

PMDS is karyotypically (46, XY) and phenotypically is male who has testis and characterized by the persistence of the uterus, the fallopian tubes, and the upper vagina which are known as Mullerian duct derivatives and results from a defect in the synthesis of AMH or a defect in AMH receptor function.[2,3]

Diagnosis of persistent Müllerian duct syndrome is based on a combination of anatomic and clinical findings and ideally the karyotyping is complementary.[3] In some reports they mentioned that the diagnosis of PMDS depends on clinical, radiological and operative evaluation.[4]

In our case, physical examination revealed that he has a normal pattern of external genitalia However, the patient had bilateral cryptorchidism with hypoplastic scrotum. The chromosomal analysis was done and showed an apparently normal male karyotype 46, XY with no numerical or structural aberrations.

Infertility was a frequently mentioned complication of PMDS despite the presence of normal spermatogenesis in some of the cases.[5] As observed in our case, the patient had secondary infertility and one child outcome of IVF. PMDS patients are at high risk of developing testicular malignancy as in patients with isolated undescended testis. So far, 20 cases are reported of PMDS with testicular malignancy.[5]

The most common type of testicular malignancies is Seminoma, followed by embryonal carcinoma, teratocarcinoma and choriocarcinoma. However, some reports stated that Mullerian malignancies are much less frequent than testicular cancer. Farikullah et al. have reported many cases of malignant transformation of the Mullerian derivatives in PMDS and the rate was between 3.1% and 8.4%.[4,5]

The main goals in treatment of PMDS patient is to manage the undescended testes and Mullerian duct derivatives, to prevent of Mullerian remnants malignancy, and to protect the fertility.[5]

Surgery is the treatment of patients with PMDS. Locating ectopic testes to normal position in PMDS is difficult due to spermatic cord shortening and abnormal position of vasa deferentia.[5] Treatments of choice in case of infertility are Orchidopexy, herniorrhaphy, hysterectomy, and bilateral salpingectomy. On the other hand, if malignancy is suspected, orchidectomy is the treatment of choice in immobilized, small testis[4]

4. Conclusion

The aim of this article is to present a rare case of PMDS with Germ cell tumor, PMDS should be on the differential diagnosis of patient with Bilateral undescended testis, the diagnosis is mainly based on clinical, radiological or intraoperative finding, the management should be directed to remove the malignant tumor and to preserve the patient fertility. Excision of the Mullerian duct remnant is encouraged.

Consent

Informed consent was taken from the patient.

Declaration of competing interest

No conflict of interest

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