

Case Report

PERSISTENT MULLERIAN DUCT SYNDROME- A RARE ASSOCIATION

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ABSTRACT

Persistent Mullerian duct syndrome is a rare form of male pseudohermaphroditism characterized by presence of Mullerian duct structures in a phenotypically and genotypically male. We are reporting a case of 47 year old male having one child who presented as acute abdomen and was clinically suspected to have lower abdominal mass. On subsequent radiological investigation patient was diagnosed to have persistent Mullerian duct syndrome (PMDS), a bicornuate uterus with leiomyoma in the left horn and a prostatic utricle cyst. Patient was treated conservatively.

Keywords: *Persistent Mullerian Duct Syndrome (PMDS), Leiomyoma, Prostatic Utricle Cyst*

INTRODUCTION

PMDS refers to pseudohermaphroditism of internal male gonads and its characterized by presence of Mullerian duct derivatives like uterus, fallopian tube, ovaries (Yuksel *et al.*, 2006; Divya *et al.*, 2010).

They are genetically 46XY with normal male characters, normal male external genitalia and no chromosomal anomalies and normal testosterone levels production and responsiveness (Yuksel *et al.*, 2006; Divya *et al.*, 2010).

The persistent Mullerian duct syndrome is thought because of failure to synthesis or release of MIF, failure of end organ to respond to MIF and effect in timing to release MIF.

It is an autosomal recessive congenital disorder. Patients' presentation may vary according to their symptoms.

CASES

A 47 year old male patient came with complains of abdominal pain since 2 years but increased since 1 week with complains of painful micturition and urinary disturbance. Patient also had suprapubic pain. Patient was married and father for one child of 20yrs age.

On examination patients vitals was stable and CVS, RS, CNS was normal and on P/A examination patient had suprapubic tenderness and P/R showed a palpable mass which lead to suspicion of lower abdominal mass either BPH or Rectal carcinoma. Patients blood routine, urine routine, RFT, LFT were within normal limits.

Patient was then referred for USG, and was found to have a well defined lobulated hypo echoic lesion in the pelvis appearing like a uterus with two separate horns and the right horn contains minimal fluid in the endometrium and small in size, left horn is bulky compared to right horn and shows well defined round shaped small hypo echoic lesion in the left lateral wall, in which there is no fluid within the endometrium and there was another small 2.5X2 cm size well defined anechoic cystic lesion in the region of prostate with no septations or internal echoes/solid elements or increased vascularity within.

USG Scrotum was done and the patient was found to have normal testis bilaterally with normal size shape echo texture and vascularity.

No focal lesion noted. Rest of abdominal organs appears normal. As with USG the possibility of PMDS with prostatic cyst and leiomyoma of left uterine horn was raised and to confirm the diagnosis of PMDS patient was further investigated with CT and MRI and patient was found to have Type1 PMDS with associated prostatic utricle cyst with leiomyoma of left uterine horn.

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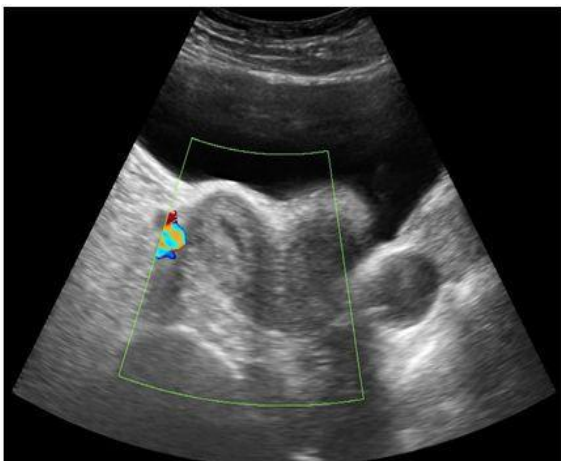


Figure 1: USG image showing uterus like structure posterior to bladder with a cystic lesion

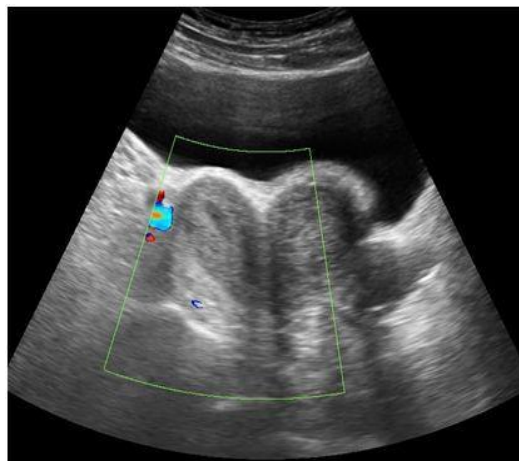


Figure 2: This USG image shows right uterine horn with fluid in the endometrial cavity

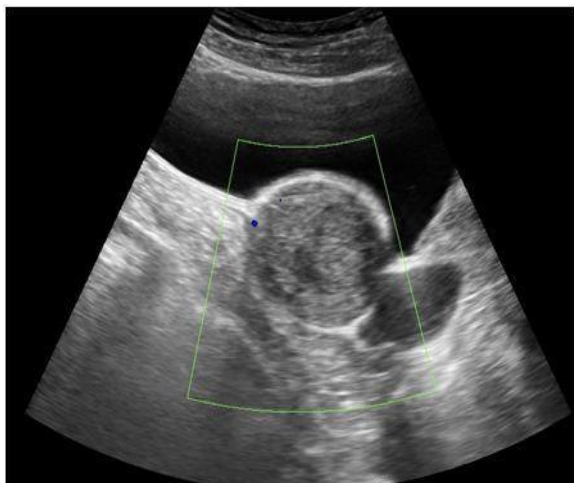


Figure 3: This image shows the left uterine horn with a small well defined hypo echoic lesion in the left lateral wall without vascularity suggesting leiomyoma in the left uterine horn. This image also shows a well defined cystic lesion suggesting prostatic utricle cyst

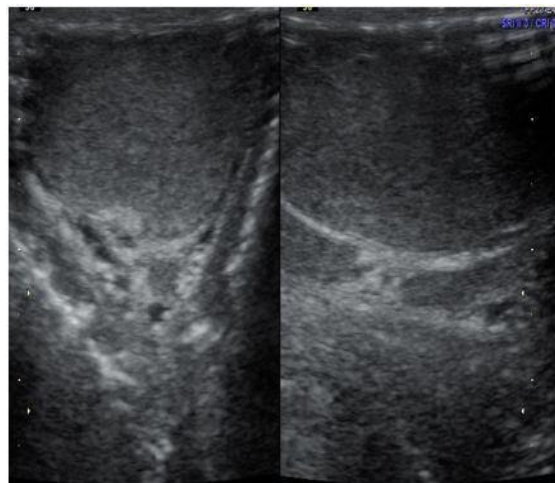


Figure 4: USG Scrotum of the patient showing bilateral normal testis with no focal lesion

Patient then underwent triple contrast CT and the CT findings were A 2.9 x 2.7 x 3.2 cm size well defined non enhancing hypo dense cystic lesion just posterior to the prostate and extending superiorly towards the seminal vesicle on left side S/O Prostatic utricle cyst. And there is a well defined homogenously mildly enhancing iso-dense lesion noted in the pelvis posterior to the bladder, superior to the seminal vesicle. No evidence of internal calcification or necrosis.

Lesion maintained fat plane bladder, rectum, prostate and seminal vesicle. No evidence of communication with the bowel loop seen. Lesion appears lobulated with fusion of two ovoid structures of each size measuring 4.3 x 3.1 cm on right side and on left side measuring 6.3 x 5.1 cm and they are fused in medial aspect.

Tiny linear soft tissue structure seen extending from the above described lesion into the inguinal canal bilaterally. Other abdominal organs were normal.

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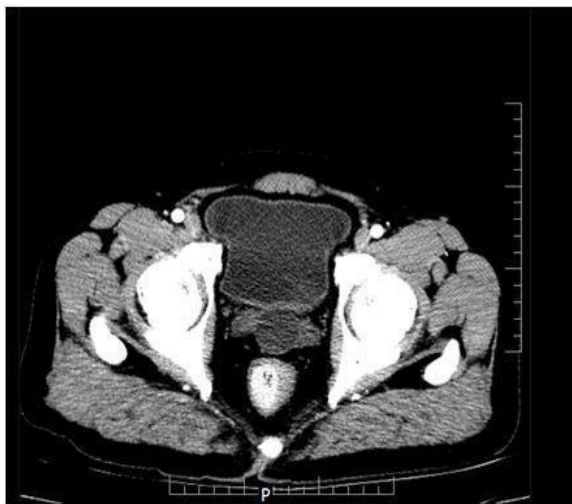


Figure 5: This image shows 2X2.2cm well defined non enhancing cystic region in the prostate



Figure 6: This image shows well defined lobulated, homogeneously enhancing isodense mass posterior to bladder fused in midline not communicating with bowel or bladder



Figure 7: CT coronal section showing well defined hypodense non enhancing cystic lesion posterior to the bladder in the region of prostate. A well defined homogeneously enhancing soft tissue structure seen in the recto-vesical space resembling uterus



Figure 8: CT sagittal reconstructed section showing right uterine horn

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Figure 9: CT sagittal reconstructed section showing left uterine horn



Figure 10: CT coronal section showing both right and left uterine horn

The patient underwent MRI after this to confirm the presence of uterus

MRI imaging showed a well defined hypo intense right horn of uterus with hyper intense signal within, left uterine horn showed fibroid within it and a well defined cystic structure was seen posterior to bladder suggesting prostatic utricle cyst

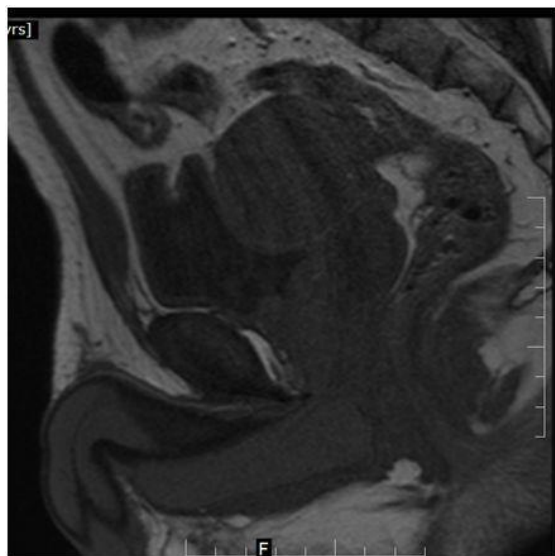


Figure 11: T1 coronal image at the level of mid sagittal plane showing a well defined hypo intense lesion in the prostate posterior to bladder

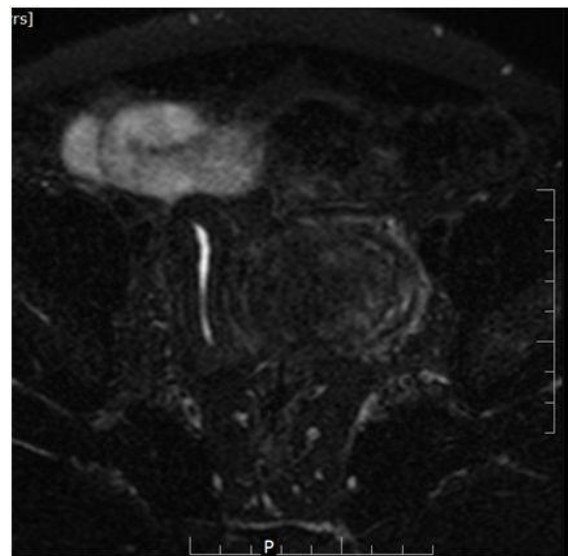


Figure 12: T2 coronal hyper intense signal with right uterine horn with endometrial cavity

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Figure 13: T2 sagittal sequence showing left uterine horn with leiomyoma in it

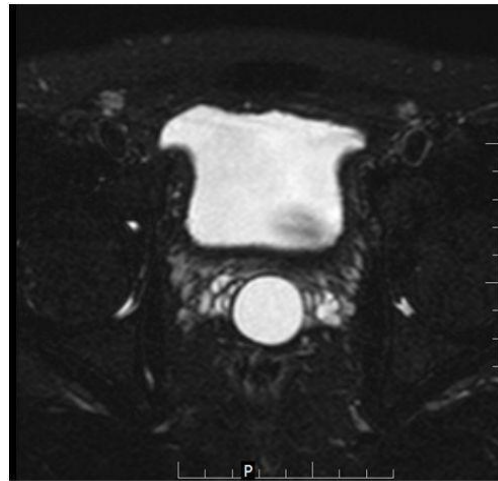


Figure 14: T2 sequence showing prostatic utricle cyst in the region of prostate

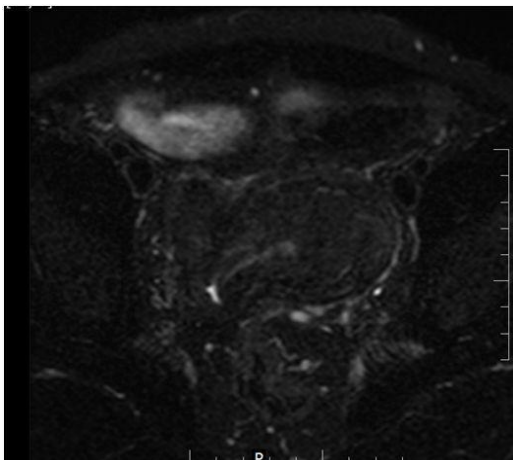


Figure 15: STIR coronal showing hyper intense signal with endometrial cavity of the left uterine horn



Figure 16: T2 CORONAL image demonstrating well defined midline cyst posterior to bladder - prostatic utricle cyst



Figure 17: T2 CORONAL section showing bicornuate uterus behind the bladder

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DISCUSSION

PMDS was first described by Nilson in 1939 (Acikalin, 2004). PMD syndrome is a rare disorder have normal reproductive organs though they have a Mullerian duct derivatives like uterus, fallopian tube also ovaries. Prevalence is not known as this disorder is very rare (Prakash *et al.*, 2009).

The Mullerian duct usually disappears during early development in males but it is retained in those with Mullerian duct syndrome. They have normal 46XY with normal male external genitalia.

Usually it is associated with cryptorchidism or inguinal hernia and during surgery it is found incidentally or during radiological examination.

Sometimes when the patients have associate prostatic utricle cyst as in our case patient may present with difficulty in micturition and mimic lower abdominal mass symptoms.

Usually one testis will be descended and it will pull the fallopian tube and uterus into the track through which it has descended. This is called HERNIA UTERI INGUINALIS or the undescended testis from the other side of the body is also pulled into same track. It is called TRANSVERSE TESTICULAR ECTOPIA (Jaka and Shankar, 2007; Dekker *et al.*, 2003).

They may also present with acute abdomen, lower abdominal pain, urinary disturbance, infertility or hematospermia. Undescended testis if not treated it may lead to seminoma.

Genetics: It is autosomal recessive condition. The gene associated with PMD syndrome will have mutation of MIF gene or MIFR2 gene which is situated in the short arm of chromosome 19. MIF gene is responsible for MIF and MIFR2 is the activator of this protein. The MIF protein and MIFR2 protein is responsible for male sex differentiation. All fetus will develop Mullerian duct as a precursor of female reproductive organ. During development the male fetus the proteins will work together and cause disappearance of Mullerian duct. But mutation of MIF or MIFR2 receptor gene may cause failure of Mullerian duct regression (Campbell *et al.*, 2007; Ulu *et al.*, 2009).

45% is due to MIF gene- Type I

40% is due to MIFR2 gene- Type II

15% is neither due to MIF or MIFR2 gene but cause is unknown.

Anatomical Types:

- 1) Testis in scrotum and uterus and fallopian tube in the inguinal canal.
- 2) In some cases, contralateral testis and fallopian tube are also in hernia sac.
- 3) They may present with Mullerian duct cyst.
- 4) Prostatic utricle cyst
- 5) Intersex and pseudo hermaphrodite.

Female type is very rare characterized bilateral cryptorchidism with testis in broad ligament (Ulu *et al.*, 2009).

In this case that we have described it is anatomical type but is associated with prostatic utricle cyst

It is important to differentiate between prostatic utricle cyst and Mullerian duct cyst. Usually the cranial end of Mullerian duct remains as the appendix of testis and the caudal end forms the prostatic utricle in normal males (Campbell *et al.*, 2007; Josso *et al.*, 2005).

Prostatic utricle cyst will arise away from the veromontenum and away from midline. But in case of Mullerian duct cyst the cyst communicates with veromontenum by thin stalk. Prostatic utricle cyst may be associated with other congenital urinary tract abnormalities (Ulu *et al.*, 2009).

Patients with prostatic utricle cyst may present with lower abdominal pain, difficulty in passing urine, dribbling of urine, bladder outlet obstruction and also as hemospermia (Devaraju *et al.*, 2012). In imaging studies it will be seen as a well defined cystic lesion behind prostatic urethra and bladder (Devaraju *et al.*, 2012).

Chances of malignancy in case of prostatic utricle cyst is 3%

Treatment includes surgical deroofting or frontal excision or endoscopic excision

PMDS also must be differentiated from other intersex disorders. Karyotyping assessment of testicular response to chorionic gonadotrophin stimulation is essential to verify both sex and existence of functional testicular tissue.

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Diagnosis: it is done by USG, CT, MRI or during Exploratory laparotomy. Most of the structural abnormalities can be identified by USG but further imaging like MR imaging will help in differentiation of structures on basis of signal intensity characteristic and morphologic features. Before puberty even AMH levels may help in diagnosis.

Treatment: The Mullerian duct derivatives must be surgically excised and orchiopexy is done in children, in adults high inguinal orchidectomy is done with supplementation of testosterone. Dissection should be done superior to the cavernosal neurovascular bundle to preserve the continence and erectile function.

Conclusion

PMDS recognition is important for diagnosis since it is a rare type of congenital anomaly high index of suspicion is necessary in diagnosis. Use of imaging modalities like USG, MRI is used to diagnose Mullerian duct syndrome and prostatic utricle cyst. The uniqueness in our case is a 47yr old male patient having a child presenting with bicornuate uterus and associated prostatic utricle cyst presenting as lower abdominal pain with leiomyoma in the left uterine horn which is utmost a rare combination.

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