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BMH Med. J. 2017;4(3):103-108 **Case Report**

Persistent Mullerian Duct Syndrome (PMDS) With Large Intraabdominal Seminoma

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Abstract

Persistent Mullerian Duct Syndrome, a form of male pseudohermaphroditism is characterized by the presence of the Mullerian duct derivatives in an otherwise phenotypically as well as genotypically normal male. We report a case of large intra abdominal seminoma in a male patient with cryptorchidism, along with persistence of Mullerian duct derivatives (uterus).

Keywords: Cryptorchidism, anti mullarian hormone, mullerian duct derivatives, MDCT, MRI, pseudohermaphroditism, Seminoma

Introduction

Persistent mullerian duct syndrome is characterized by the presence of uterus, fallopian tubes and or upper vagina in a phenotypically and genotypically normal male [1]. It is caused either by defective synthesis of anti mullerian hormone (AMH) or end organ resistance due to defective AMH 2 receptor [2]. Two anatomic forms are found in persistent mullerian duct syndrome. Commoner is unilateral cryptorchidism and contralateral inguinal hernia. In the other variant, bilateral cryptorchidism is seen with the uterus in the pelvis and the testes embedded in the broad ligament.

Seminoma is the most common testicular neoplasm in the 3rd, 4th decade of life. Risk of tumour in undescended testis is 30 times greater than in an orthotopic gonad. Risk is not decreased by subsequent orchidopexy, even persists in contralateral normal testis of males who have undergone orchidopexy.

Case report

A middle aged man presented with abdominal distension and feeling of fullness which was insidious in onset but progressed to cause enough discomfort and seek medical attention. A father of one, he had no significant past medical history except for a herniorrhaphy for left inguinal hernia at the age of 33. MRI for the present complaint, done elsewhere showed large abdominopelvic mass, which was reported as retroperitoneal sarcoma / GIST.

Before undergoing surgical procedure patient opted for second radiological opinion where upon he was referred to our department. CT scan of abdomen and pelvis with contrast was done in 128 slice MDCT scanner and images were acquired in arterial, venous and excretory phases. Correlative sagittal T2W MR images were also acquired. CT images (**Figures 1, 2** and **3**) showed a large heterogeneously enhancing lobulated abdominopelvic soft tissue mass apparently arising from the recto vesical space. The approximate size of the mass was 20 x 19 x 12cm and showed cranial extension to the level above umbilicus. The mass showed ill-defined fat planes with prostate caudal extension noted to the level of 3rd coccygeal segment. There was compression of urinary bladder, rectum and sigmoid colon. Displacement of pelvic small bowel loop cranially and lateral extension to the level of iliac vessels with maintained fat planes noted. Bilateral ureteric compression and obstructive uropathic changes were present, as was gross ascites. Arterial supply from the right gonadal artery was clearly depicted, suggesting a possible gonadal origin.

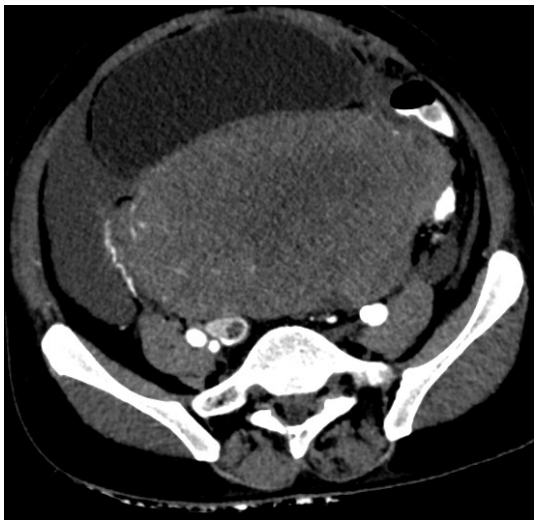

Figure 1

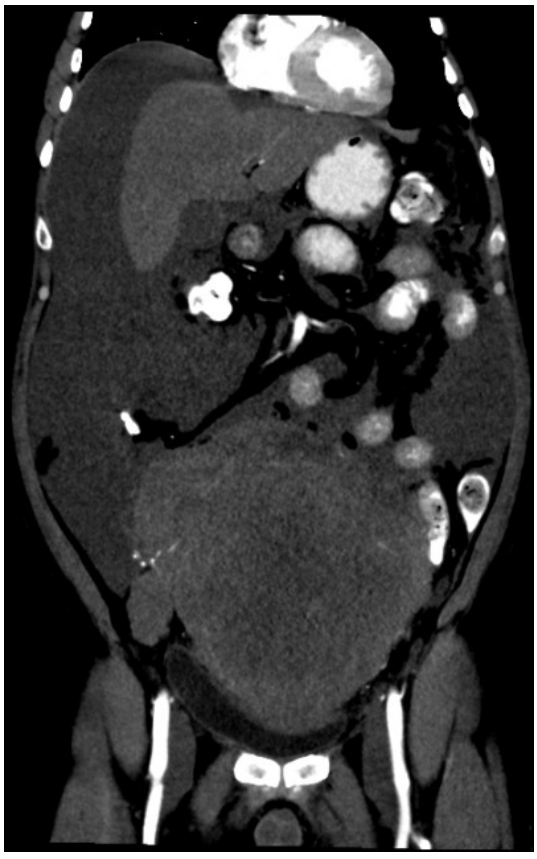

Figure 2

In view of this typical arterial supply, patient was retrospectively interrogated in detail to obtain any missed information. History of left sided orchidopexy along with herniorrhaphy and an empty scrotal sac on right side was obtained, information the patient deemed irrelevant to an intra abdominal problem. This was further confirmed on clinical and scrotal USG examination. Based on the imaging findings and clinical history, diagnostic possibility of intra abdominal testicular neoplasm from un descended right testis was suggested. Sagittal MR images showed heterointense lesion with areas showing whorled appearance (**Figure 3** and **4**)

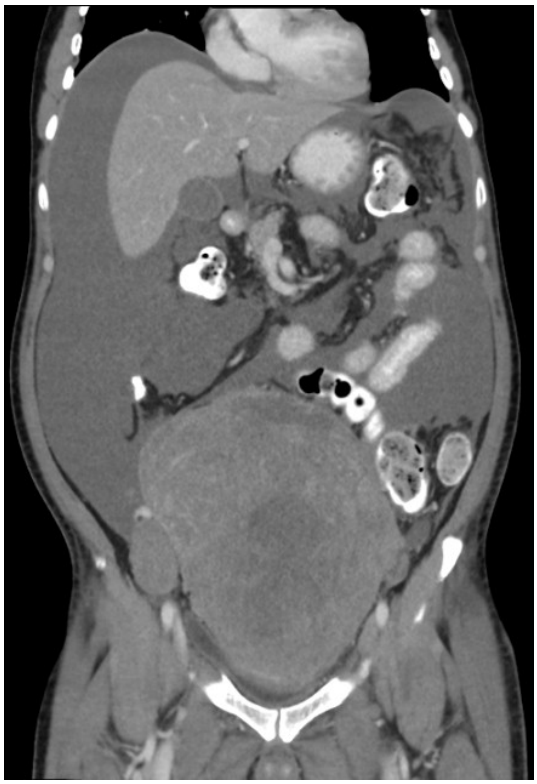

Figure 3

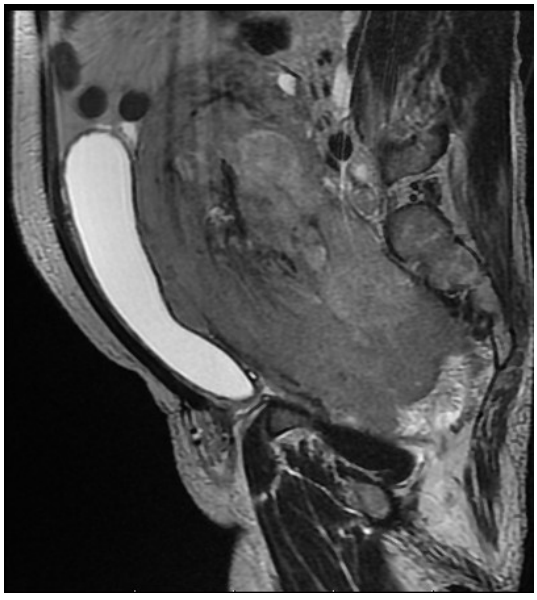

Figure 4

Shortly afterward patient underwent debulking surgery. Imaging findings were confirmed intra operatively and the histopathological examination revealed neoplasm composed of sheaths and lobules of cells having uniform prominent eosinophilic nuclei and clear cytoplasm. Areas of necrosis

were seen. (**Figure 5** and **6**) Tumour invaded serosa of the colon. Also seen was a rudimentary uterus with endometrial gland, stroma and myometrium. Tumour cells were positive for SALL4, negative for CK and AFP. Final histopathological diagnosis was seminoma (classic type, with rudimentary uterus).

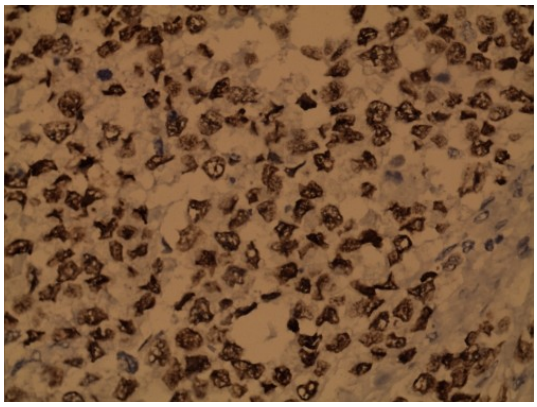

Figure 5

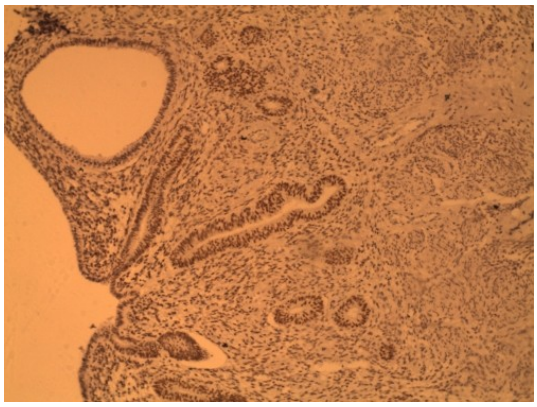

Figure 6

Discussion

PMDS is a rare form of male pseudohermaphroditism in which mullerian duct derivatives are present in a phenotypically and genotypically normal male. It results from a failure or mistiming in the synthesis and release, or end organ resistance to AMH. The subjects have normal levels of testosterone with normal male secondary sexual characters.

In a human fetus the Mullerian and Wolffian ducts are both present at 7 weeks of gestation. In a male fetus, the testis differentiates by the end of the 7th gestational week. Normal sex differentiation is controlled by testosterone, dihydrotestosterone, and AMH. Sertoli cells secrete AMH, which leads to regression of the Mullerian ducts. Testosterone has a direct effect on the Wolffian ducts, and promotes their differentiation into the epididymis, vas deferens, and seminal vesicles [3-5]. Dihydrotestosterone induces male differentiation of external genitalia. PMDS patients have both Wolffian and Mullerian duct structures due to a deficiency of AMH.

Persistent Mullerian duct syndrome is encountered in two variants. The more common male form, also called hernia uteri inguinale [6] in which one testis is usually normally descended in the scrotal sac. The contralateral testis and tube, are either in the inguinal canal or can be brought into the inguinal canal along with the uterus and ipsilateral Fallopian tube by gentle traction on the presenting testis. The other, less common, form, the female type, [7] is characterized by bilateral cryptorchidism with the testes embedded in the broad ligament in an "ovarian" position with respect to the uterus, which is fixed in the pelvis.

Like in other cryptorchid testes the PMDS patients are at an increased risk of developing testicular neoplasms. [8] Seminoma is the most frequent carcinoma of testicle in 4th decade of life and

constitutes 60-65% of germ cell neoplasia. Several histopathological characteristics of the tumor has been evaluated and three types of pure seminomas described - classic, anaplastic and spermatocytic types.

Diagnosis of intra abdominal masses in the setting of undescended testes is easy and straight forward. But this important history was lacking at the initial presentation in our case. It was on careful analysis of the CT images, the arterial supply to the mass from the right gonadal artery was noticed and that turned out to be the crux in our case. MDCT with multiplanar image reconstruction capabilities and MIP images are helpful in many situations where the organ of origin of a mass is in doubt, successfully demonstrating the vascular supply of the parent organ.

Even though a clear cut evidence as to which category of PMDS our case belongs is lacking, with the previous history of left sided orchiopexy at the sitting of herniorraphy and empty right srotal sac at the time of presentation, we presume it to be a case of the rarer variant of bilateral cryptorchidism or a mixed anatomical variant.

PMDS is a rare anomaly with nearly 200 cases reported so far out of which less than 40 cases have been associated with testicular neoplasms.

Seminomas are very sensitive to chemotherapy and radiotherapy. Our patient is now being treated by the oncology department with chemotherapy. The prognosis is excellent in cases of seminomatous histology with 5 year survival rates > 90% with platinum based chemotherapy.

Conclusion

PMDS is a rare form of male pseudohermaphroditism. There are two anatomical variants of which the one presenting as bilateral cryptorchidism is rarer. As in other undescended testes PMDS also carries a high risk of malignant testicular tumors the commonest being seminoma. Incidence of cancer, ranges from 3.5-14.5% among patients with cryptorchidism. with highest incidence in 3rd and 4th decades of life, usually identified incidentally by imaging. The risk is not found to be higher in PMDS.

While imaging, the importance of determining the arterial segment supply to a mass cannot be emphasized enough, as manifested by this case. Although parasitic feeders from aberrant sources can often supply a large mass, most often, the feeding vessel can help steer the investigation in the right course by providing a clue to the tissue of origin.

The predominant histological type is pure seminoma (43%) followed by embryonic (28%), teratocarcinoma (27%) and choriocarcinoma (2%). Surgery is mandatory, with chemotherapy an alternative, depending on stage and histological type of malignant transformation. It is advised to remove the Mullerian duct structures in a case of PMDS because of the risk of malignancies they carry.

References

1. Renu D, Rao BG, Ranganath K, Namitha. Persistent mullerian duct syndrome. Indian J Radiol Imaging 2010;20:72-4.

2. Josso N, Belville C, di Clemente N, Picard JY. AMH and AMH receptor defects in persistent mullerian duct syndrome. Hum Reprod Update 2005;11:351-6.

3. Rey R, Picard JY. Embryology and endocrinology of genital development. Baillieres Clin

Endocrinol Metab 1998; 12:17-33.

4. Robboy SJ, Bentley RC, Russell P. Embryology of female genital tract and disorders of abnormal sexual development. In, Blaustein's Pathology of the female genital tract. 5th ed. New York: Springer Verlag; 2002. p. 19.

5. Guerrier D, Tran D, Vanderwinden JM, et al. The persistent mullerian duct syndrome: a molecular approach. J Clin Endocrinol Metab 1989; 68:48-52.

6. Nilson O. Hernia uteris inguinalis beim manne. Acta Chir Scand 1939; 83:231-239.

7. Clemente A, Macchi V, Berretta M, Morra A. Female form of persistent mullerian duct syndrome: MDCT findings. Clin Imaging. 2008 Jul-Aug;32(4):314-7.

8. Kazim E. Intra-abdominal seminomas in persistent mullerian duct syndrome. Urology 1985;26:290-292**.**