CASE REPORT

PERSISTENT MULLERIAN DUCT SYNDROME

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Persistent Mullerian Duct Syndrome is extremely rare. Our patient, a 32 years old male, with history of orchidectomy presented with mass abdomen. He was initially diagnosed with seminoma and subsequently treated with chemotherapy. Biopsy of the mass showed germ cell tumour and MRI abdomen revealed female rudimentary organs confirmed on per operative and later on histopathology. Karyotype was 46 XY.

Keywords: Persistent Mullerian Duct Syndrome; Seminoma; Chemotherapy; Karyotype

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INTRODUCTION

Persistent Mullerian Duct syndrome (PMDS) is a rare autosomal genetic entity caused by mutation in MIS and MISR2 genes.[1] The three forms exist namely the most common female type followed by utriinguinal and transverse testicular ectopia type.[2] We present our case of PMDS which posed as a diagnostic dilemma to surgeons.

CASE PRESENTATION

A 32 years old male presented in May 2018 with abdominal pain and sensation of fullness in lower abdomen for two months associated with weight loss of 5 kg. The pain was dull, intermittent, non-radiating and located in the periumbilical region. Contrast enhanced CT showed a well-defined mass in the abdominal cavity; 20.4 cm by 11.8 cm, with multiple pockets of adjacent cystic areas. Bilateral hydronephrosis was also seen [Figure: 1]. Consequently, bilateral ureteroscopy was done with double J stenting. A year prior to this, the patient had a history of bilateral inguinal orchidectomy due to undescended testis. Biopsy at the time showed fibromuscular tissue with dilated vessels. No malignancy or testicular tissue was seen.

Trucut Biopsy of the abdominal mass showed a stage 2 ($T_4N_3M_0S_1$), classical seminomatous germ cell tumour with a baseline β-HCG of 1300 mIU/mL and an α-Fetoprotein level within normal ranges [Figure: 2]. Chemotherapy was initiated in October 2018 with the standard regimen of Bleomycin, Etoposide and Cisplatin (BEP) for four cycles. Post-treatment Contrast enhanced CT scan showed a soft tissue mass; 7.5cm by 5.8cm, in the midline. Inferiorly, the mass was compressing the urinary bladder while laterally, it was abutting gut loops. A soft tissue satellite nodule was seen in the posterior aspect of the lesion [Figure 3]. An MRI scan of the pelvis confirmed the presence of uterus and ovaries with other rudimentary structures and advised correlation with karyotyping [Figure:4]. At this point, the patient was referred to the General surgery department for the removal of the uterus, ovaries and rudimentary structures. Karyotype was also advised.

Surgery to remove the above-mentioned structures was planned. Total abdominal hysterectomy was carried out. During surgery, a left ovarian mass, right ovary, Fallopian tube, uterus and a blind ending vagina was found [Figure 5]. No metastasis or retropharyngeal lymph node findings were seen. Karyotype also showed 46XY chromosomes]. No residual disease was found. The patient was subsequently discharged on medications and with resolution of abdominal symptoms. Follow up histopathology report confirmed mixed gonadal dysgenesis with seminomatous germ cell tumour (persistent Mullerian Duct Syndrome) and presence of female reproductive organs

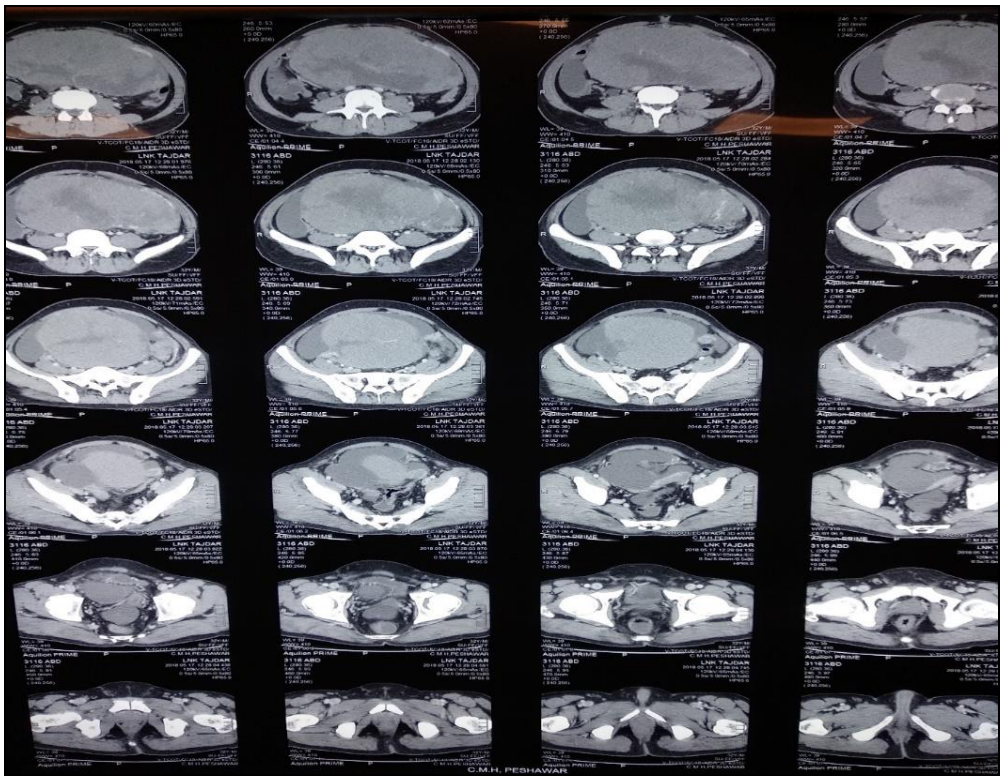

Figure-1: CECT Abdomen films showing mass and Hydronephrosis

Spc Nature: TRUCUT BIOPSY

Spc Site: LOWER ABDOMEN

History: Not stated.

Gross: Specimen container is labeled with the patient's name and medical record number. Received in formalin are multiple cores ranging in size from 0.2 cm to 0.6 cm. The entire specimen is submitted in single block.

Micro: See final diagnosis.

IMMUNOHISTOCHEMICAL STAINS:

Cytokeratin:	Negative
S100:	Negative
Desmin:	Negative
Myogenin:	Negative
CD30:	Negative
OCT3/4:	Positive
SALL4:	Positive
Calretinin:	Negative
CD45:	Positive

Diagnosis: LOWER ABDOMEN, TRUCUT BIOPSY:
Malignant germ cell tumor, consistent with seminoma (see note).

Note: Please correlate with serum tumor marker studies.

SNOMED: T-Y4230 M-80103

Figure 2: Histopathology of Trucut biopsy showing Seminoma

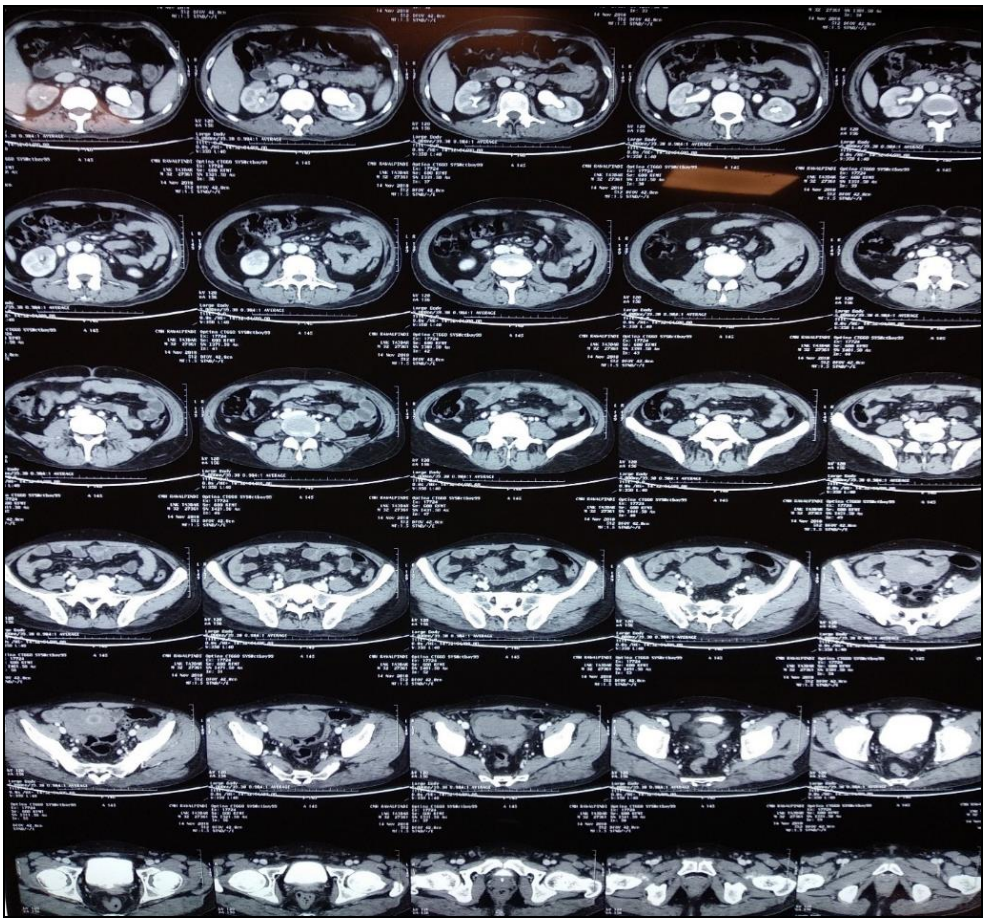

Figure 3: CECT Abdomen films post treatment and interfaces

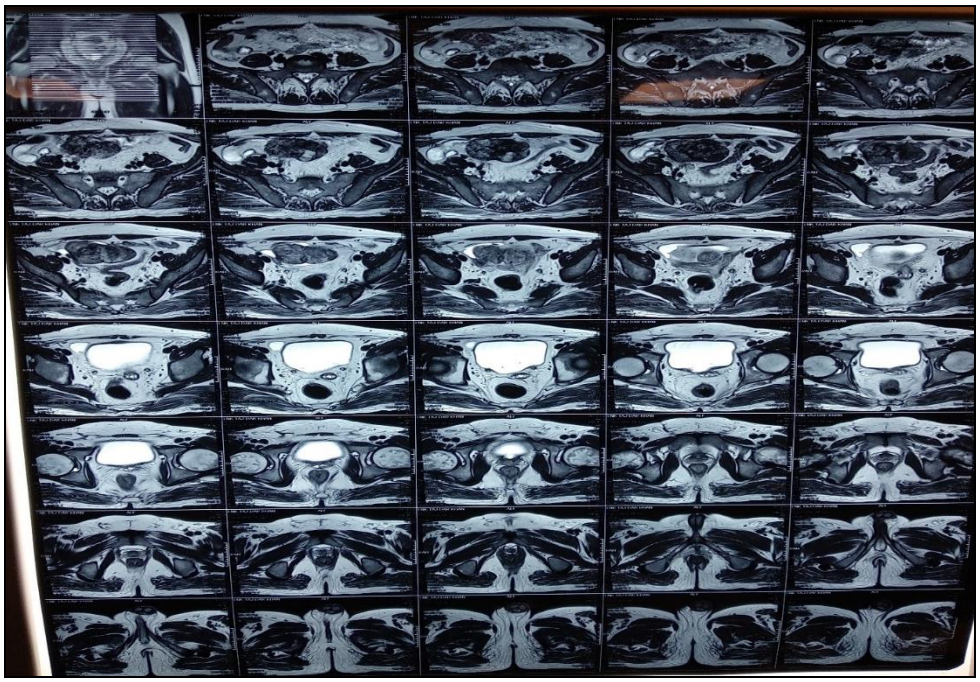

Figure 4: MRI Abdomen films showing rudimentary structures suggesting laparotomy and karyotype

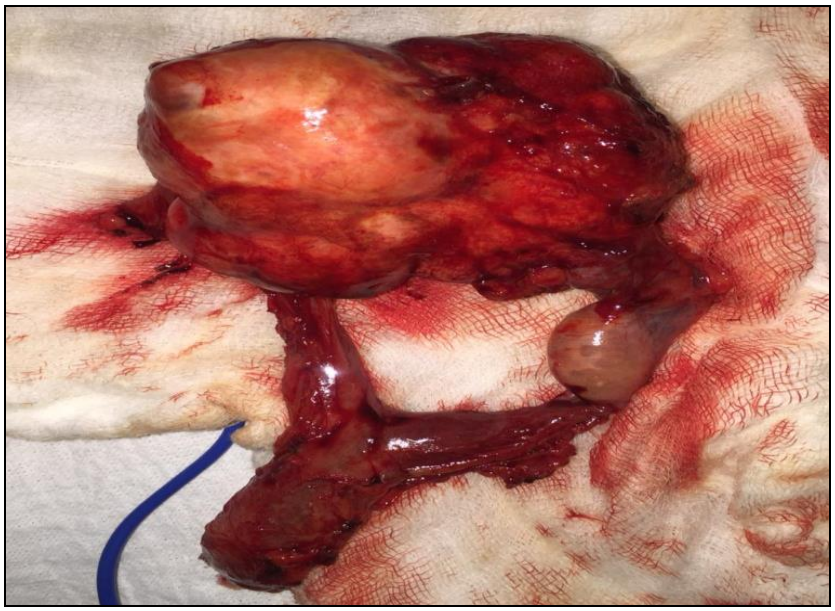

Figure 5: Per Operative picture showing uterus, ovaries and fallopian tube

DISCUSSION

This case was a diagnostic challenge from the start. What started as a simple bilateral inguinal orchidectomy for undescending testis ended up with total abdominal hysterectomy. Testicular cancer is not very common and young men are the most commonly affected demographic.[3,4] Seminoma is the most frequently occurring type of testicular cancer.[5] The average age for seminoma was 34 years as observed in a study conducted in Pakistan. Cryptorchidism increases the risk of germ cell tumours of the testis.[6] These tumours commonly present as a painless swelling.[7] The patient under discussion also presented at 32 years of age and with an abdominal mass. He fits the demographic profile for this type of tumours.

Although there is scarcity of data regarding germ cell tumours in Pakistan, a study done in Pakistan found that Seminomatous germ cell tumours were less prevalent than non-seminomatous ones.[8] The present case is one of seminomatous germ cell tumour. The astonishing findings of female internal reproductive organs raised the suspicion of chromosomal abnormalities. However, karyotype of the patient showed a normal male chromosomal pattern of 46XY. The patient was thus genotypically male. External genitalia included a penis and an empty scrotal sac due to undescended testes.

Tumour markers are generally elevated in germ cell tumours.[9] In our case, β-HCG was elevated while α-Fetoprotein was normal. Although the rates of testicular cancer have been increasing especially in western countries,[10] good survival has been reported for patients with germ cell tumours being treated with the standard regimen of Bleomycin, Etoposide and Cisplatin-otherwise known as BEP[1)]. In the present case, a reduction in the size of the abdominal mass and along with no retroperitoneal lymph node findings suggested good response of the tumour to the chemotherapy regimen.

CONCLUSION

This case has been documented to draw attention to an unusual presentation of seminomatous germ cell tumour in a genotypically male patient. The location and nature of presenting symptoms were also vague, and could not have raised suspicion of underlying tumour and female internal reproductive structures. It would probably make sense to rule out germ cell tumours in patients with cryptorchidism, as this increases the risk of the above-mentioned tumours.

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