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Case Report

PERSISTENT MULLERIAN DUCT SYNDROME (PMDS) WITH GERM CELL TUMOUR AND ITS ANAESTHETIC IMPLICATION PLANNED FOR EXPLORATORY LAPAROTOMY.

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ABSTRACT:

Persistent Mullerian duct syndrome is a rare condition occasionally encountered in men with normal phenotype but with presence of Mullerian duct structures. In India, owing to neglect and lack of facilities, we encounter this condition in adult males. A patient came with abdominal pain and subsequent work up done and they found a mass retroperitoneally and did a biopsy for the same. Biopsy proved it as a malignant pathology and the patient was started on chemotherapy to reduce the size of tumour. The patient undergone surgery after 6 months as it is a proven case of germ cell tumour. Though rare, every surgeon operating upon inguinal hernia or undescended testes or cryptorchidism needs to know about the presence of the uterus in a phenotypic male patient at any age. High degree of suspicion and awareness is needed to diagnose this condition. We report successful anaesthetic management of a 29-year-old male with persistent mullerian duct syndrome scheduled for exploratory laparotomy under general anaesthesia with epidural catheter insertion for intraoperative and postoperative pain management.

Keywords: Persistent Mullerian duct syndrome. Gell cell tumour. Chemotherapy. Uterus . Exploratory laparotomy with excision of intraperitoneal malignancy. Phenotypic male . Undescended testis.

1. INTRODUCTION

Persistent Mullerian duct syndrome (PMDS) is a rare disease that occurs in men with a completely normal phenotype and is characterised by the presence of Mullerian duct structures. Organogenesis of the male external genitalia is not affected. There is failure of regression of the mullerian structures, resulting in the presence of a uterus, fallopian tubes and the upper third of the vagina in males with a 46XY karyotype. Persons affected with this syndrome show the presence of male and female reproductive systems together.

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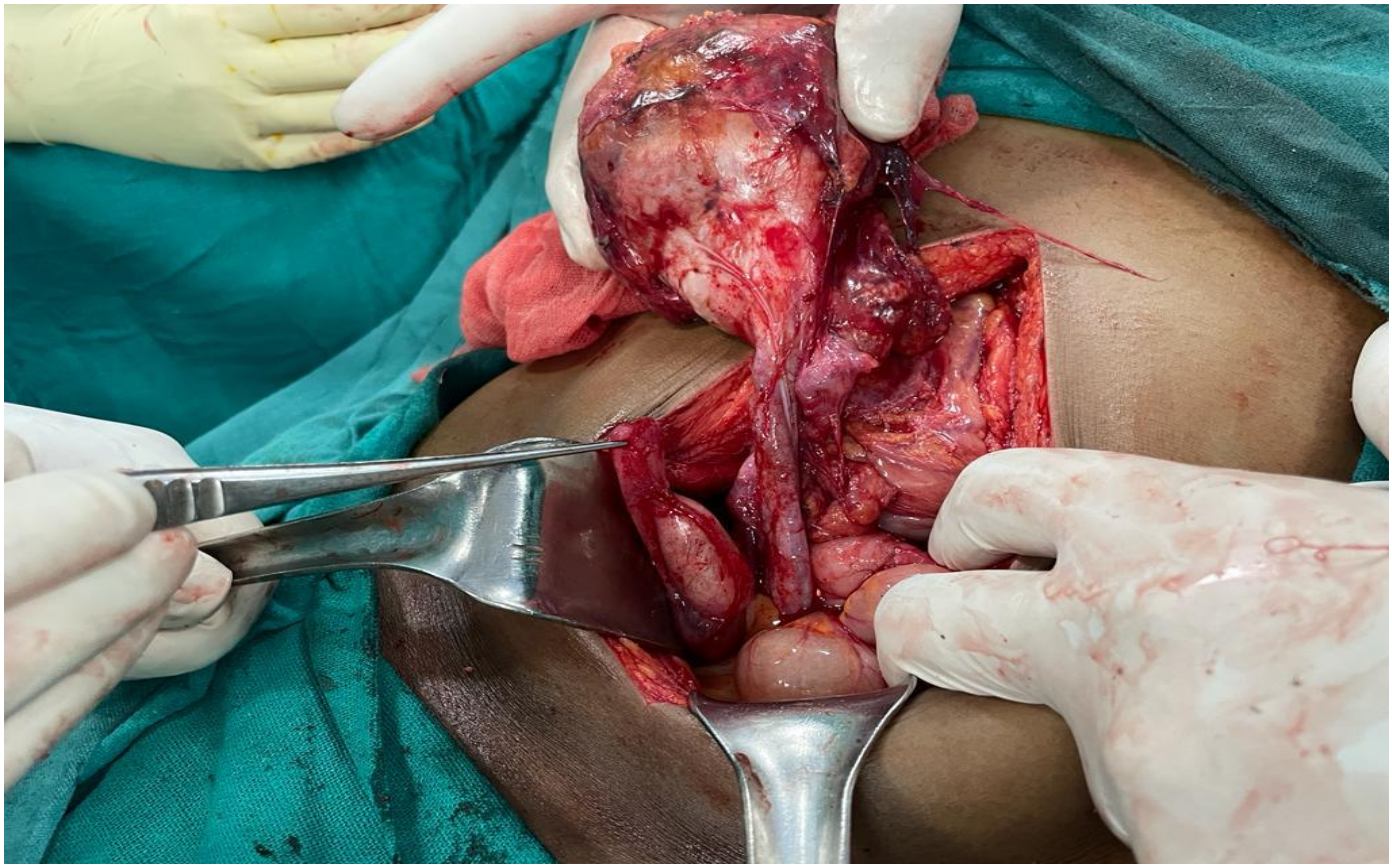
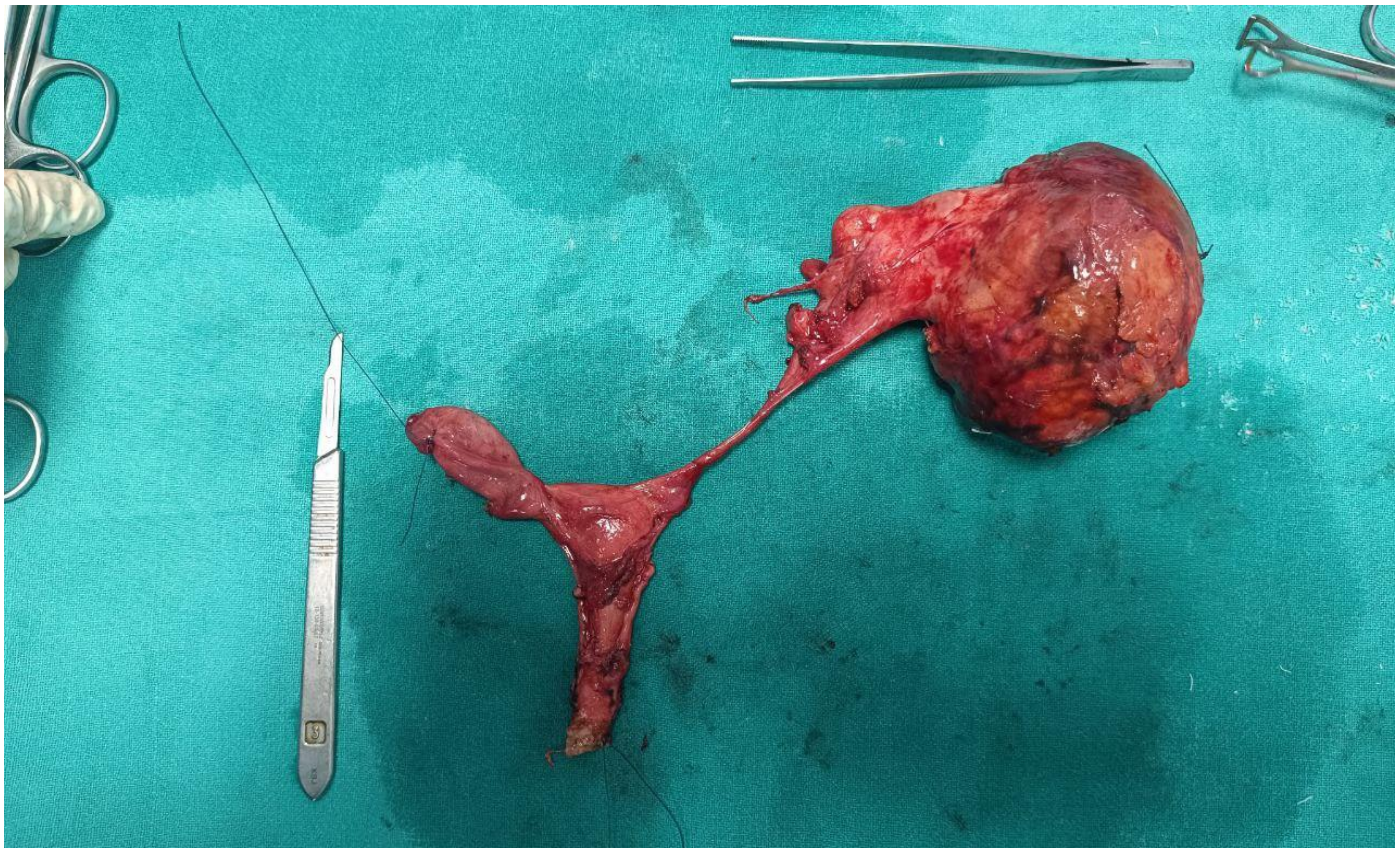
Case report:

A 29 years old sexually active male came with the complaints of abdominal pain which was sudden in onset and progressive in nature over right hypochondrium and right lumbar region and he felt the lump in abdomen not associated with vomiting, diarrhoea, icterus, melena and hematemesis. Secondary male sexual characters are well appreciated. On examination per abdomen, it is globular with central inverted umbilicus, no colour change, no dilated veins no scar mark present afebrile, no tenderness, guarding rigidity not present. Mass of 15×12 cms felt over right hypochondrium extending till umbilicus region and right iliac region. Scrotal sac formed with bilateral testes not palpable in the scrotum. Triphasic CECT abdomen shows large ill-defined solid heterogeneously enhancing mass lesion noted in abdomen and pelvis of size 13.9× 13.5 × 22 cms large retroperitoneal deposit in left para-aortic region left renal hilum of size 9.2 × 4.6 cms mild hydronephrosis mild ureteric obstruction with multiple peritoneal nodular deposits peri rectal deposits suggestive of malignant mass. True cut biopsy shows round cell tumour and oncopathology report suggestive of germ cell tumour especially germinoma. MRI pelvis shows hypoplastic uterus. Patient was started on chemotherapy. After 6 months patient was planned for surgery for the removal of tumour. Patient came for pre anaesthetic check-up. Patient was completely assessed and examined. Modified Mallampati scoring 2 with three finger mouth opening. All routine investigations were within normal limits. Patient was shifted inside the OT and all the standard parameters were attached and monitored. Under all aseptic precautions, epidural catheter insertion was performed in sitting position at L2-L3 level and the placement is confirmed with loss of resistance technique. we attained the epidural space at 3.5cms and the catheter is fixed at 10 cms since we have to achieve sensory block up to T6 level. He was premedicated with 4mg ondansetron,0.2 mg glycopyrollate,1mg of midazolam and 100 mcg of fentanyl. Preoxygenation done with 100% O2 for 3 minutes via ventilatory circuit and induction was done with 100mg propofol and 100 mg succinyl choline. Conventional laryngoscopy was performed using a Macintosh blade. A Cormack-Lehane grade 1 view was obtained on the first attempt, and a 7.5 endotracheal tube was placed without difficulty. Anaesthesia is maintained with oxygen and nitrous oxide mixture, 0.2 -2% isoflurane and IV atracurium was given as maintenance throughout the surgery. Volume of 20 ml Bupivacaine 0.125% and 5 mcg/cc of fentanyl was taken in a syringe and started it as infusion at a rate of 4ml per hour via epidural for intra operative analgesia. Patient was reversed and extubated with glycopyrrolate 0.5mg and neostigmine 2.5 mg. Intraoperatively MAP was maintained between 65 to 70 mm/hg throughout the 3 hours of surgery. Blood loss was assessed and transfused accordingly. Patient was shifted out of the OT with epidural catheter in-situ for postoperative pain management. Biopsy report came after three days which reveals mature endometrial tissue, fallopian tube, gonadal ridges, immature testis without spermatogenesis and vas deferens suggestive of mixed gonadal dysgenesis or persistent mullerian duct syndrome. No residual tumour foci and architectural features are suggestive of pre therapy seminoma residual cancer burden score is 0. Hormonal assays came as elevated FSH and LH with decreased testosterone levels. Karyotyping was done and the report comes as XY which conclude the diagnosis of mixed gonadal dysgenesis or persistent mullerian duct syndrome.

ISSN: 0975-3583, 0976-2833 VOL 15, ISSUE 01, 2024





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2. DISCUSSION

Persistent Mullerian duct syndrome is a disorder of sexual development that affects males. Males with this disorder have normal male reproductive organs, though they also have a uterus and fallopian tubes, which are female reproductive organs. The uterus and fallopian tubes are derived from a structure called the Mullerian duct during development of the foetus. The Mullerian duct usually breaks down during early development in males, but it is retained in those with persistent Mullerian duct syndrome. Affected individuals have the normal chromosomes of a male (46, XY) and normal external male genitalia.

The first noted signs and symptoms in males with persistent Mullerian duct syndrome are usually undescended testes (cryptorchidism) or soft out-pouching's in the lower abdomen (inguinal hernias). The uterus and fallopian tubes are typically discovered when surgery is performed to treat these conditions. The testes and female reproductive organs can be located in unusual positions in persistent mullerian duct syndrome.[1]

MDS or persistent mullerian duct syndrome is a rare phenomenon, and a high degree of suspicion is essential in diagnosing it. Clinically, PMDS cases are divided into three categories:

1. Majority (60–70 %) with bilateral intra-abdominal testis, in a position analogous to ovaries (female variant) .
2. Smaller group (20–30 %) with one testis in the scrotum, associated with contralateral inguinal hernia whose contents are the testis, uterus and tubes (classical presentation of hernia uteri inguinale, male variant)[2] .
3. Smallest group (10 %) where both the testes are located in the same hernial sac along with the Mullerian structures (transverse testicular ectopia (TTE), male variant)[3,4] .

Other effects of persistent Mullerian duct syndrome may include the inability to father children (infertility) or blood in the semen (hematospermia). Also, the undescended testes may break down (degenerate) or develop cancer if left untreated.

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition. However, persistent Mullerian duct syndrome affects only males. Females with two mutated copies of the gene do not show signs and symptoms of the condition.

Preoperative diagnosis of PMDS is difficult, but all patients (of any age group) who present with bilateral cryptorchidism or one-sided inguinal hernia with contralateral undescended testis or one-sided inguinal hernia with a palpable mass above the normally descended testis should ideally be investigated for chromosomal studies and ultrasonography or MRI to assess the presence of uterus with fallopian tubes and testis. However, most studies have found ultrasonography to be of limited value in picking up Mullerian structures [5]. Diagnosis of PMDS is based on a combination of high index of suspicion and anatomic, clinical and imaging findings. Simple per rectal examination may reveal the presence of uterus with fallopian tubes. MRI has also been done to detect the mass when PMDS presents as an intra-abdominal mass.

Differential diagnosis of PMDS includes disorders of sexual differentiation with defective regression of the mullerian derivatives including mixed gonadal dysgenesis (MGD). Biopsy

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is required to confirm the sex and to differentiate from MGD. A workup to rule out associated genitourinary malformation should be done. In our case, both the patients had no associated genitourinary malformation

Distinguishing intersex disorders from PMDS is important as the management is different. In the literature, there are case reports of PMDS patients with infertility as well as patients with three to five children, so there is no definitive documentation of PMDS with infertility[6].

Josso et al. reported that if the gonads are placed in the scrotum, fertility is retained . Martin et al. documented spermatogenesis in a patient with TTE . Fertility is rare in patients with PMDS[6]. Imbeaud et al. reported three cases of PMDS with testicular degeneration and opined that anatomical abnormality may favour testicular torsion and early loss of testis[7] .

Management of PMDS is controversial. Earlier studies opined that while removing the mullerian remnants, there is a high probability of damage to vas deferens and its blood supply with no disadvantage of conversion into malignancy, so they should not be removed[8,9]. Thiel and Erhard reported the death of a 14-year-old boy with PMDS, due to metastasis of adenosarcoma arising from the mullerian remnant [10].Romero et al. reported a 39-year-old man who developed adenocarcinoma of endocervical origin, arising from the mullerian remnant[11]. The overall incidence of malignant transformation in these testes is 18 % similar to the rate in abdominal testes[12]. There have been reports of embryonal cell carcinoma, seminoma, choriocarcinoma and teratoma in patients with cryptorchidism and persistent Mullerian duct. Apart from malignancy, retained mullerian remnants which are in connection with the prostatic utricle are known to cause recurrent UTI, stones and voiding disturbances. Surgical management is aimed at preserving fertility by performing orchiopexy for repositioning of the testis into the scrotum if feasible or at least getting the testis to a palpable suprascrotal location. Herniorrhaphy with hysterectomy and bilateral salpingectomy with preservation of the vas deferens retained to preserve fertility should also be done[13,14] . Orchidectomy is indicated for testes that cannot mobilised or have undergone a malignant change or if streak/dysgenetic gonads have been detected [15]. Routine orchidectomy should not be done as the capacity of the virilisation must be preserved.

In this case since there is biopsy proven malignant mass[16] with undescended testis with some peritoneal metastasis which has to be removed surgically. Extensive exploration is needed for the surgeons. Case discussed with surgeons and understanding about the duration of surgery and necessity of post-operative analgesia we preferred general anaesthesia along with epidural catheter insertion over spinal anaesthesia.

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