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Persistent Mullerian Duct Syndrome -A Rare Case Report

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Abstract **Case Report**

Persistent mullerian duct syndrome is a disorder of sexual development that commonly affects male, also known as Hernia uteri inguinale. They have male external genitalia with admixture of male and female reproductive organs. Common clinical presentation is with infertility or inguinal hernia. The main modality of therapy is surgery. Early diagnosis and therapy maintains fertility and prevents malignant change. We are reporting a case of persistent mullerian duct syndrome in a 40 year old male who presented with inguinal hernia with preserved fertility.

Keywords: Persistent mullerian duct syndrome, male, inguinal hernia, fertile.

INTRODUCTION

PMDS is first described by Nilson in 1939 as hernia uteri inguinalis. It is a rare disorder of sexual development, a type of male pseudohermaphroditism. It is familial and autosomal recessive disorder [1]. This is caused by defect in mullerian inhibiting substance resulting in defective regression of uterus and fallopian tube [2]. So far in English literature, around 158 cases of PMDS have been reported [3].

CASE REPORT

A 40 years old male presented with acute pain and swelling in the right inguinal region since 2 days. Detailed clinical history revealed presence of swelling in right inguinal region and absence of right testis in the scrotal sac since birth. Family history: He was married and had two children, with no such cases in the family. Clinical examination showed normally developed penis with descended testis in left scrotal sac and absence of testis in right scrotal sac. In the right inguinal region, a 7x5cm pyriform shaped tender, warm swelling was present with absence of reducibility and cough impulse. A diagnosis of right sided obstructed inguinal hernia with right cryptorchidism was made. On ultrasonography of right inguinal region, hernial sac was seen with omentum, part of bowel, undescended testis as contents. Patient was posted for emergency hernioplasty.

Intraoperatively, exploration of right inguinal canal revealed an indirect inguinal hernia sac with well-formed uterus along with a tube like structure, testis, part of ileum and omentum as contents. Total excision of uterus and testis was done and operation was completed by hernioplasty. We received uterus with tube like structure and testis for histopathological examination. Gross examination of specimens excised showed a well formed uterus measuring 5x3x2cm with attached tube like structure of length 1cm with no ovaries. Cut section of uterus showed endometrial cavity with poorly formed cervix. Testis was atrophic measuring 4x3x1cm.On histopathological examination, sections from uterus showed endometrium with glands, stroma and myometrium (Figure 2).Sections from tube like structure showed structure of fallopian tube (Figure 3).Sections from testis showed atrophied and hyalinised seminiferous tubules with complete arrest of spermatogenesis and prominence of Leydig cells (Figure 4&5).There was no evidence of malignancy in the sections studied.

A diagnosis of persistent mullerian duct syndrome was made based on history, clinical examination and histopathological findings. Patient recovered well after the surgery.

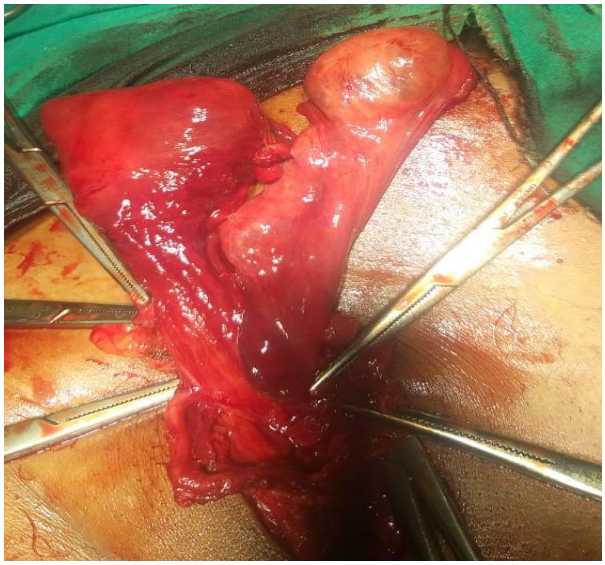

Fig-1: Intraoperative photograph showing uterus with fallopian tube and testis during hernioplasty

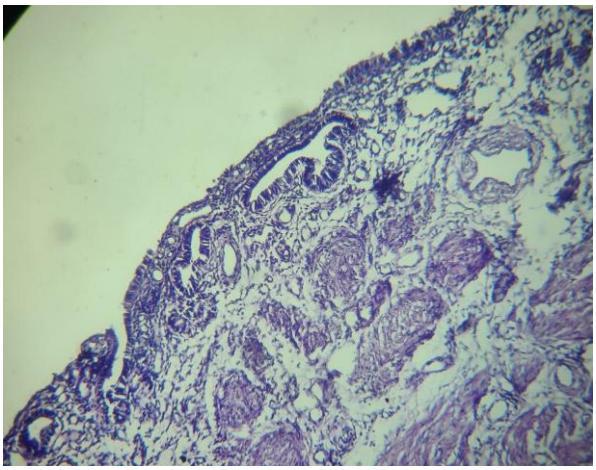

Fig-2: Sections from uterus showed endometrial glands and endometrial stroma (H&E, 100X).

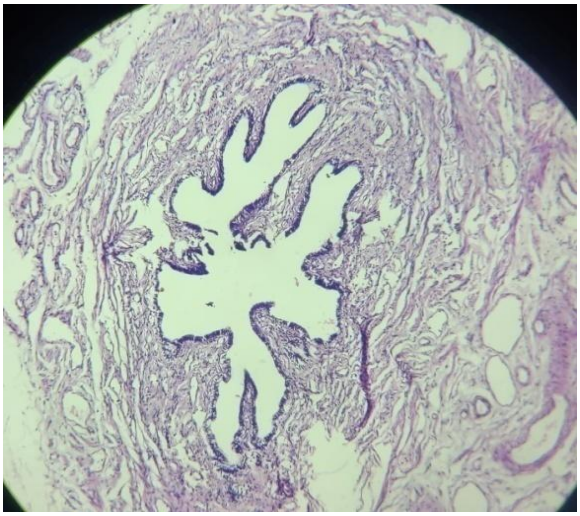

Fig-3: Sections from fallopian tube showed mucosal folds lined by pseudotratified squamous epithelium with underlying muscular wall (H&E, 100X)

DISCUSSION

Persistent mullerian duct syndrome (PMDS) is a male pseudohermaphroditism caused by defect in mullerian inhibiting system [5-9]. Genetically PMDS is of two types. PMDS type 1 is due to mutation in anti mullerian gene on chromosome 19 resulting in decreased secretion of anti-mullerian hormone [6]. PMDS type 2 is due to mutation in anti-mullerian receptor gene on chromosome 12, leading to resistance for the action of anti-mullerian hormone during first trimester in antenatal period .Under the influence of SRY gene on Y chromosome in a male fetus, anti-mullerian hormone secreted from sertoli cells of testis causes regression of mullerian duct thereby facilitating the testicular descent [7]. Development of external genitalia is normal under the influence of testosterone hormone secreted by Leydig cells of testes [8].

Persistent mullerian duct syndrome may be in two forms: 1.Female form with uterus and bilateral intra-abdominal testes occupying position of ovaries [7]. As per Natarajan S [1], female form is the most common presentation. 2. Male form is two types: type 1 with one descended testis and contralateral inguinal hernia with uterus, fallopian tube and undescended testis as contents, type 2 with inguinal hernia containing both testes and mullerian structures as contents, known as transverse testicular ectopia [7]. As per Prakash N [6] male form type 1 with one descended testis and contralateral inguinal hernia is the most common presentation. According to Gujar [7], a case of male form of PMDS presented as an obstructed inguinal hernia in the left side with right cryptorchidism

Extensive survey of English literature showed majority of cases reported were female form of persistent mullerian duct syndrome and male form of transverse testicular ectopia. The present case presented with obstructed inguinal hernia on the right side with cryptorchidism and presence of uterus, fallopian tube and opposite side descended testis (male form type 1).As per literature, most common malignancy reported in undescended testis of persistent mullerian duct syndrome was seminoma. In the present case there was no evidence of malignancy.

Karyotyping, genetic testing for mullerian inhibiting system, mullerian inhibiting hormonal assay adds to the diagnosis which is not in the present case due cost issues. Preoperative diagnosis helps in careful planning of surgery. The effectiveness of treatment lies in careful excision of mullerian structures without injuring vas deferens and orchidopexy.

CONCLUSION

Whenever a clinician sees a child with unilateral/bilateral cryptorchidism, there should be high degree of suspicion for the presence of persistent mullerian duct syndrome. Recognition of PMDS at an early age is important for preventing complications. A multidisciplinary approach is needed to diagnose and treat this condition successfully

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