CASE REPORT

Persistent Mullerian Duct Syndrome Presenting As Transverse Testicular Ectopia [TTE] Rarest of Rare: A Case Report

SreeRamulu.P.N[1], D.Srinivasan[1], Pawan Katti[1], Madhu[1]*

[1]Department of Surgery, Sri Devaraj Urs Academy of Higher Education and Research, Kolar-563101 (Karnataka) India

Abstract:

Persistent Mullerian Duct Syndrome (PMDS) is usually an accidental finding either during orchipexy or during routine inguinal hernia repair in male patients presenting with mal- descended testes. It is caused by a defect in the Mullerian Inhibiting Substance (MIS) system. Intra-operatively, Mullerian remnants consisting of an infantile uterus and fallopian tubes are usually found. Familiarity with PMDS is necessary to diagnose the condition. We are presenting a rarest of a rare a case report of a male aged 58 year presented with inguino-scrotal swelling. Subsequently the patient was taken up for surgery, when the inguinal canal was opened we found in the hernia sac well developed female reproductive system. i.e. uterus of adult size, ovaries, fallopian tubes and cervix .on the right side hence a case of transverse testicular ectopia, cut sections of uterus shows endometrial cavity measuring 2.5cm, endocervix measuring 2cm while endometrium measuring 1cm. The gonads were testes by histological examination, with features of degeneration and fibrosis with no neoplastic changes.

Keywords: Mullerian Inhibiting Substance, Mullerian Remnants, Persistent Mullerian Duct Syndrome, Transverse Testicular Ectopia.

Introduction:

Intersexual disorders are very important clinical issues with their different aspects relating to diagnosis, treatment and sex of rearing [1].They are broadly classified into disorders associated with a normal chromosome constitution and disorders associated with an abnormal chromosome constitution. Persistent Mullerian Duct Syndrome (PMDS) is a form of male intersex caused by a defect in the Mullerian Inhibiting Substance (MIS) system. Patients are phenotypically male and usually present when young with unilateral or bilateral undescended testes and an inguinal hernia into which prolapses an infantile uterus and fallopian tubes [2]. Familial cases have been reported with a probability of sex-limited autosomal recessive or X-linked recessive inheritance. An incidence of PMDS in identical twins has also been reported [3]. PMDS is often misdiagnosed due to a lack of familiarity with the condition. We hereby report our case to stress the intra-operative diagnosis of this condition with a high index of suspicion on the operating surgeon's part and because of its rarity.

Case Report:

Rare case report of a male aged 58 year is presented with right inguinoscrotal swelling and on examination he had a peculiar inguinoscrotal swelling, we could feel both the testis and swelling was not reducible into the abdomen features suggestive of obstructed inguinal hernia and apart from routine investigations patient was subjected to ultra scanning of the scrotal swelling, also confirmed as obstructed inguinal hernia and commented that the both the testis could be visualized in the scan. Subsequently under spinal anesthesia right inguinal incision was taken, when

the inguinal canal was opened and the scrotal content was retracted, out, to our surprise we found well developed female reproductive system i.e. uterus of adult size, ovaries,(mistaken for testis under our scan report) fallopian tubes and cervix.

No morphologic evidence of intraepithelial germ cell neoplasia or any evidence of testicular neoplasm was detected. No Familial history of the same kind, the patient's secondary sexual characters were well developed as evident by the photograph, and obviously the patient had no issues

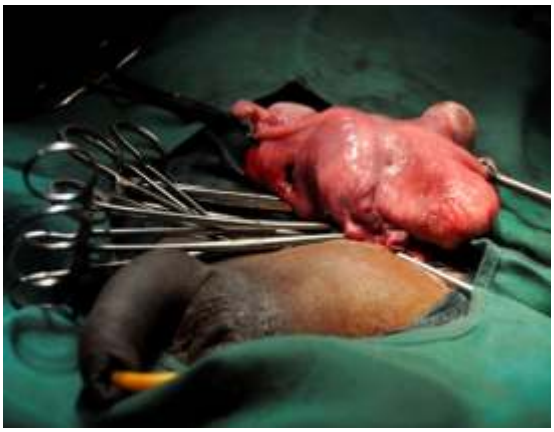

Fig. 1: Showing Well Developed Phallus an Female Reproductive System on the Same Side TTE [10%] of PMDS

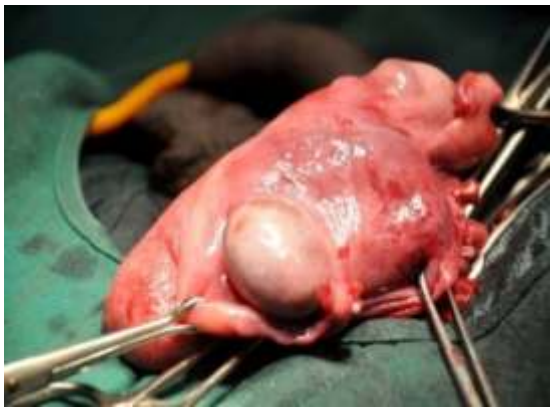

Fig. 2: Shows Well-Developed Uterus with Fundus Body, Cervix, Gonads, with Fallopian Tubes

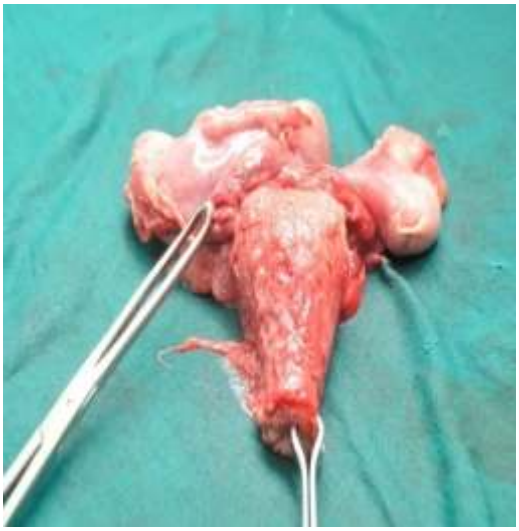

Fig. 3: Extracted Specimen with Full Developed Female Reproductive System

Fig.4. Gross Section Consists of the Uterus and Cervix with Two Testes

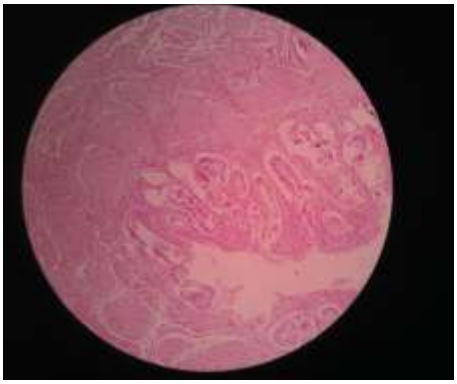

Fig. 5: Section shows Atrophic Endometrium with Normal Myometrium and Perimetrium

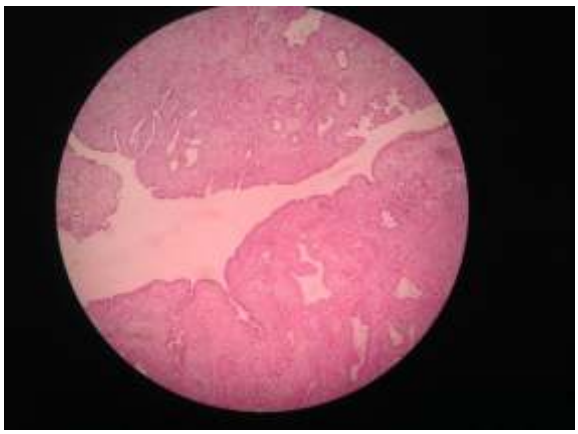

Fig. 6: Cervix shows Normal Endocervix Histology No Ectocervix

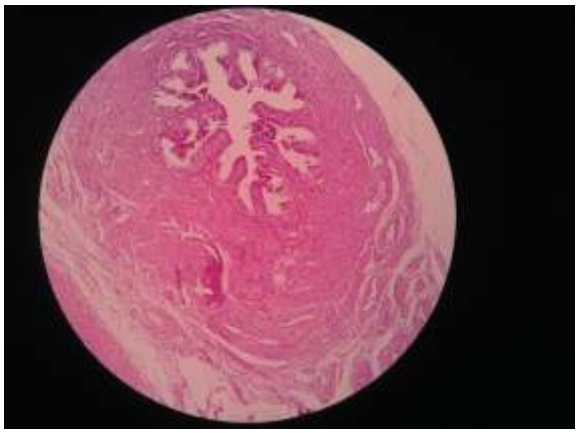

Fig.7: Section shows Normal Histology of Fallopian Tubes

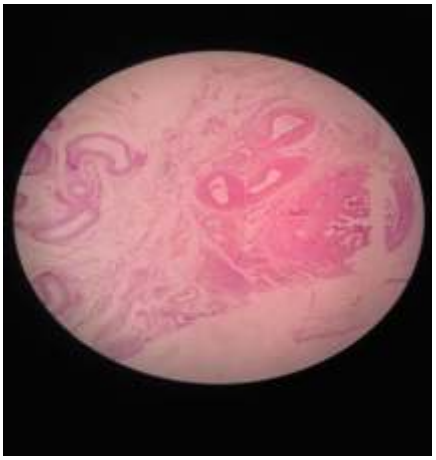

Fig 8: Section Shows Tube Like Structure on The Side of the Large Testis Shows Normal Epididymis

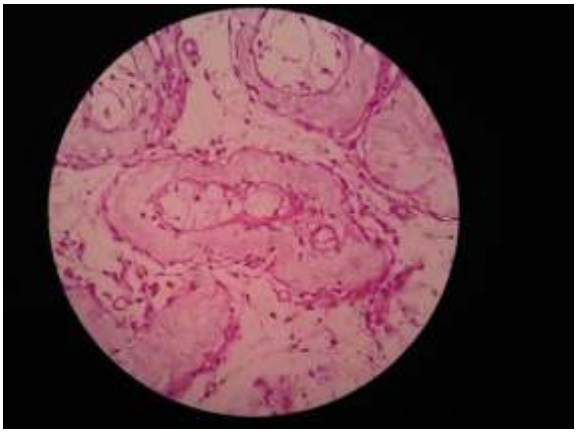

Fig 9: Section of the Smaller Testis Shows Tubules Showing Only Serotoli Cells with Few Hyalinized, Seminiferous

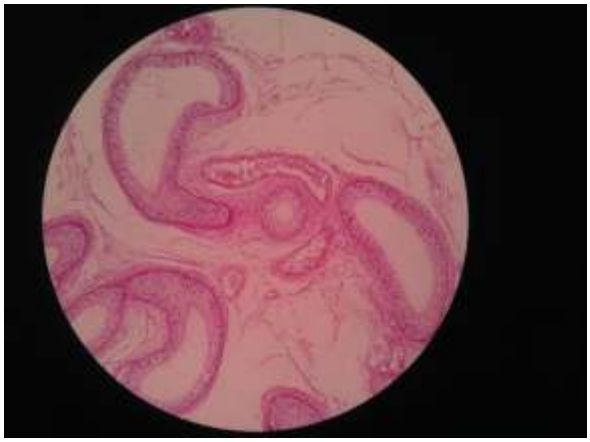

Fig.10 Section Shows Normal Histology of Vas Deferens

Results:

Peripheral blood for karyotype and 46XY genotype and hormonal assay was performed as shown in table 1. This shows the patients genotypes similarity with the female

Table 1: Shows Hormonal Assay Distinguishing between Observed Values and Normal Values

Hormonal assay	Observed values	Normal values
Total testosterone	14.7 ng/dl	Male-260-1000 ng/dl Female: 15-70ng/dl
Testosterone bioavailable	4.25 ng/dl	Male Value: 66-417ng Female-6-5ng/dl
Free testosterone	0.25ng/dl	Male: 5-21 Female: 0.01-0.85
Sex hormone binding globulin	42.69 nmol/l	14-48 nmol/l
Estrogen	25.91 pg/ml	Male-10-50 pg/ml Female :20-350 pg/ml
Lutinizing hormone	17.10miu/ml	Male-1.7-8.7, Female: 1-95.6miu/ml
Mullerian Inhibiting substance	Zero	

Histopathology Findings:

The hernia sac was measuring 9cm in length 2.5cm in breath, 3.5 cm in width, uterus and cervix measuring in 5cm in length, 2.5cm in breath 3.5cm in width, one testis measuring in 3cm length 2cm in breath 2cm in width. Another 2cm in length, 1.7 in breath, 5cm width, spermatic cord measuring in 1.5.cm in length and 0.7.cm in breath, cut sections of uterus shows endometrial cavity measuring 2.5 cm. Endocervix measuring 2cms. Endometrium m.1cm in thickness, also identified two tube like structures extended from uterus to the testis one measuring 6cms in length another 5 cms. Seminiferous tubules show interstitial fibroses, and marked with Leydig cell hyperplasia. Features are the smaller testis is consistent with atrophic testis with complete absence of spermatozoa. Section of the larger testis shows majority of seminiferous tubules showing maturation arrest to the spermatocyte stage. Few tubules showing mature spermatozoa few tubules are completely hylainized also seen in leydig cell hyperplasia. No morphologic evidence of intraepithelial germ cell neoplasia or any evidence of testicular neoplasm was detected. Intra operative blood was drawn for hormonal assay.

Discussion:

A PMDS or hernia uterus inguinal is a rare form of male pseudohermaphroditism characterized by the presence of mullerian duct structures in 46 XY phenotypic males. It is caused by a defect in the MIS system [4]. The MIS is a large glycoprotein that sertoli cells produce early in fetal life as early as seven weeks of intrauterine life. The gene responsible for the substance is on chromosome 19. MIS is also known as anti-mullerian hormone. The primary function of MIS is to cause regression of the mullerian (paramesonephric) ducts in the male fetus. MIS is first secreted in effective amounts 56-62 days after fertilization, and the process of mullerian regression is

normally completed by about Day 77, after which the mullerian tissue is no longer sensitive to MIS. Another important function of MIS is to initiate testicular descent, principally by its postulated regulatory control over the gubernaculum testis [2-4]. These two functions of the MIS explain the clinical findings of PMDS.

Clinically, PMDS cases are divided into three categories. Majority (60-70%) with bilateral intra-abdominal testis in apposition analogous to ovaries , Smaller group (20-30%) with one testis in the scrotum associated contra lateral inguinal hernia whose contents are testis, uterus and tubes (classical presentation of hernia uteri inguinale), Smallest group (10%) where both the testes are located in the same hernial sac along with the mullerian structures (transverse testicular ectopia-TTE). **This is type we encountered, thus rarest of rare group**.

A review of the literature shows that in patients with intra-abdominal testes both the gonads may be located in a position analogous to the ovaries, with a rudimentary uterus in the center and the mullerian remnants preventing the mobilization of the testes [3].

Manjunath *et al*[3] have reported two cases of PMDS (familial), one with bilateral intra-abdominal testes and the other having hernia uteri inguinale with TTE **[our case]**.Our case did not show any dysplastic changes or malignancy. However, Berkmen [4] has reported three cases with PMDS associated with testicular malignancy: Two cases were of testicular seminoma and the remaining one case showed both testicular seminoma and teratoma. Compared with a normally descended testis, the bilateral undescended testis has a 7-35% increased risk of developing a malignant tumor, especially seminoma. Moreover, the risk of developing a malignancy is greater in an abdominal than in an inguinal testis. Seminoma is the most common histological type in bilateral undescended patients or in PMDS. The diagnosis of PMDS is made incidentally during surgical exploration for cryptorchidism or herniorraphy as the mullerian remnants are not palpable on abdominal, rectal or scrotal examination. Intra-operative methods of diagnosis, especially the gonadal biopsy, can be performed to rule out mixed gonadal dysgenesis and developing malignancy [3, 4].Mixed gonadal dysgenesis is a disorder associated with an abnormal sex chromosome constitution, characterized by ambiguous genitalia with unilateral testis and a streak gonad contralaterally, with persistence of mullerian duct structures on the side of the streak [2].

Another clinical presentation is that of crossed testicular ectopia or transverse testicular ectopia. It is usually seen to be associated with PMDS. It is a rare congenital anomaly in which both the gonads migrate toward the same hemi-scrotum [5] [**Our case]**.

In cases of abdominal undescended testis, where a two-stage Stephen Fowler procedure is contemplated, excision of Mullerian remnants may be hazardous for the collateral circulation of the testes. Midline splitting of the Mullerian remnants and excision of the mucosa are advocated to allow orchidopexy. However, as no malignancy occurs in the retained Mullerian ducts, hysterectomy should not be performed [3, 4]. A conflict exists whether orchidectomy should be performed as orchidopexy offers only limited protection against future malignancy. **Thus in our case, with due consent of patient, we contemplated orchidectomy since the patient already was 58 year old.**

Conclusion:

PMDS has an autosomal recessive inheritance. Screening of siblings and second-degree relatives is necessary followed with genetic counseling. Although ultrasonography and magnetic resonance imaging are reported to play a role in locating the Mullerian remnants, laparoscopy has a distinctive advantage in diagnosing PMDS. Familiarity of the operating surgeon with this disease condition would increase the chances of correctly diagnosing and appropriately dealing with the Mullerian remnants.

Acknowledgement:

We thank the contribution from Dr. Kusma Assistant Professor, Department of Pathology and also Dr. Somashekarum, Department of Anesthesia in this study and helping us to operate respectively.

References

1. Ocal G. Current concepts in disorders of sexual development. *J Clin Res Pediatr Endocrinol* 2011:3:105-14
2. Robboy SJ, Bentley RC, Russell P, Anderson P. Pathology of abnormal sexual development. In: Fox H, Wells M. Haines and Taylor-Obstetrical and Gynecological Pathology. 5th ed. United Kingdom: Churchill Livingstone, Elsevier; 2003: 1209-32.
3. Manjunath BG, Shenoy VG, Raj P. Persistent müllerian duct syndrome: How to deal with the müllerian duct remnants- A review. *Indian J Surg* 2010; 72:16-9.
4. Berkmen F. Persistent müllerian duct syndrome with or without transverse testicular ectopia and testis tumours. Br J Urol 1997; 79:122-6.
5. Moslemi MK, Ebadzadeh MR, Al-Mousawi S. Transverse testicular ectopia, case report and review of literature. *Ger Med Sci* 2011; 9:Doc15.

[*] ***Author for Correspondence:*** *Dr. D. Srinivasan, Department of Surgery, Sri Devaraj Urs Academy of Higher Education and Research, Kolar-563101 (Karnataka) India Email: drsrinivasandorai@gmail.com Cell : 08105576075*