Persistent Mullerian Duct Syndrome - A Rare Anomaly

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Abstract

Persistent mullerian duct syndrome is a disorder of male pseudo-hermophraditism characterized by persistence of uterus, fallopian tubes and upper two third of vagina in otherwise normally virilized phenotypically and genotypically male (46XY). Patients may present with hernia, hydrocele, or impalpable udescended testis at any age group and most of them are diagnosed intraoperatively. Awareness among the surgeons about this rare association helps in appropriate management.

Keywords: Hernia uteri inguinale, Persistent mullerian duct syndrome, Undescended testis

INTRODUCTION

Von Lenhossek first reported the rare entity of Transverse testicular ectopic (TTE) in 1886. Jordan in 1895 described transverse testicular ectopia associated with persistent mullerian duct syndrome (PMDS).[1] Nelson in 1939 first descrbed this association in a man with inguinal hernia as hernia uteri inguinale. About 150 cases of PMDS have been reported in literatue, whereas TTE is still scarer.[2] Presence of both testes on one side of scrotum is known as TTE. It is rare to find combination of PMDS & TTE in a single patient. Patients present with absent testis, hernia, or infertility during infancy, childhood or adulthood. Diagnosis is made incidentally during groin hernia or orchidopexy operations or imaging.[3] Pre operative diagnosis is practically difficult.[4] There are 2 morphological types of PMDS: Female type (10-20%) having bilateral (BL) undescended testes (UDT) and no hernia. Uterus and fallopian tubes are fixed to pelvis and testes embedded in broad ligament. Male type (80-90%) having unilateral UDT and contralateral inguinal hernia containing mullerian duct (MD) structures and testis. Male type has 2 sub types. Type I - hernia uteri inguinale with TTE, hernia sac containing MD structures and both testis. Type II - classic hernia uteri inguinale, hernia sac containing ipsilateral fallopian tube and ipsilateral testis.

CASE REPORTS

We report 5 cases of PMDS which were incidentally detected during groin operations.

Case 1

2 year old boy was brought BL impalpable UDT, empty scrotum and a normal penis Diagnostic Laparoscopy (DL) revealed uterus and fallopian tubes fixed to pelvis and both testes were embedded in broad ligament. Suprapubic exploration done. Both the testes and adherent uterus with fallopian tubes mobilised in toto. We had to split the Uterus meticulously in midline without damaging the vascularity of testes in order to bring down both the testes into the scrotum. Orchidopexies were done. It was female type of PMDS (Figure 1).

Case 2

3 year old boy presented left sided impalpable UDT, normal penis and empty left hemiscrotum. DL revealed inguinal hernia on right side with left testis on right side. We also found a rudimentary uterus and fallopian in close relation testis. It was male -sub type I form of PMDS. Groin exploration done on right side (Figure 2). Herniotomy and sub-dartous pouch orchidopexies done. Mullerian structures were biopsied.

Case 3

2 year old boy was brought with right sided hydrocele. During herniotomy fallopian tube was seen attached to the hernial sac, which was placed back into the abdomen. It was males subtype II form of PMDS. Post-operatively the Karyotype was 46XY and gonadal biopsy confirmed to be testis (Figure 3).

Case 4

5 year old boy presented with right sided obstructed inguino-scrotal hernia. On emergent groin exploration was done. The contents were viable intestinal loops which were reduced. During herniotomy we could find there were two spermatic cords with two testes. In between the two testes we found PMD structures. Herniotomy and subdartous orchidopexy done (Figure 4)

Case 5

1 year old boy (sibling of case-2) was presented with right sided inguinal hernia. Examination revealed a normal penis and an impalpable testis on left side. During herniotomy, we found that some mass attached to the right spermatic cord which was very difficult to deliver. Hence the skin and fascial incisions were extended. Applying traction to the cord revealed fallopian tubes, uterus and left testis. Herniotomy and orchidopexy was done (Figure 5).

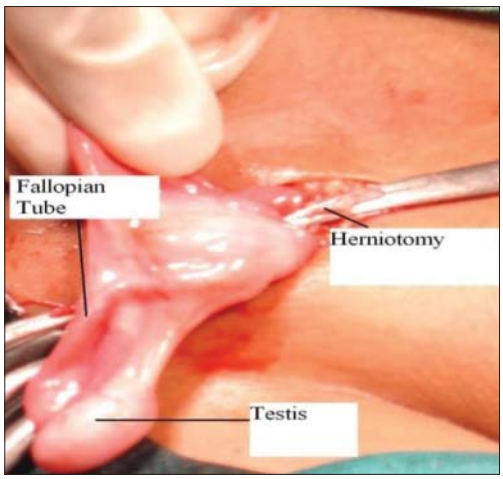

Figure 3: Male type of PMDS-subtype II

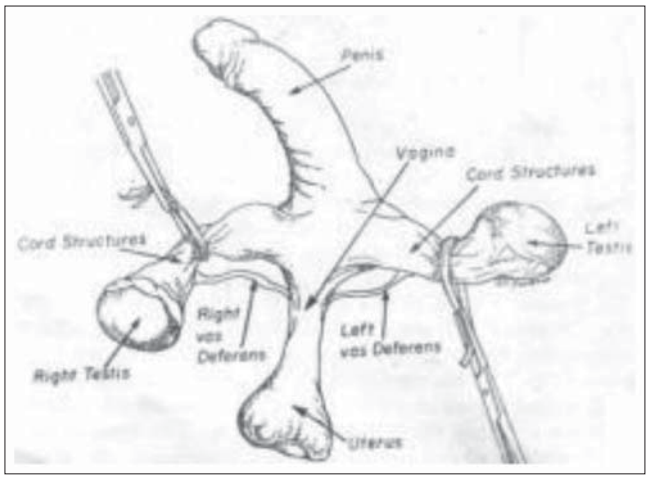

Figure 1: Female type of PMDS

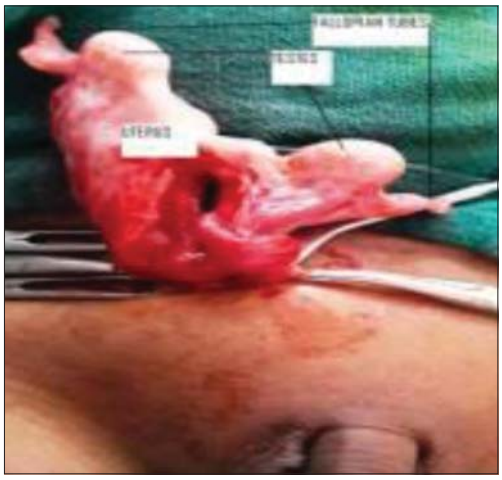

Figure 4: Male type of PMDS-subtype I

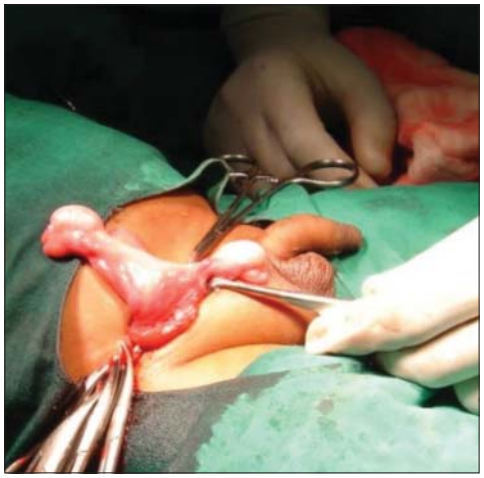

Figure 2: Male type of PMDS-subtype I

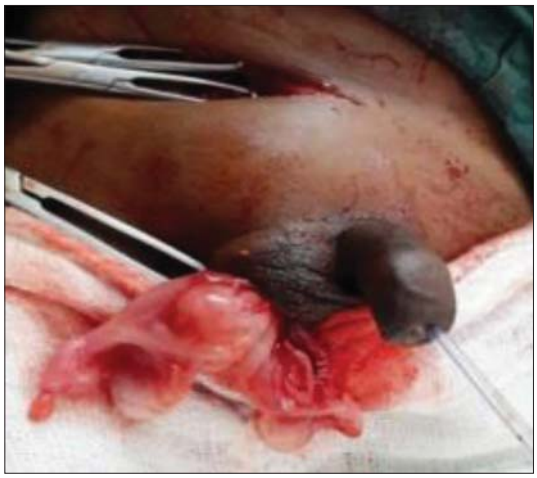

Figure 5: Male type of PMDS-subtype I

Follow Up

Karyotyping was 46XY, testis and MD were confirmed by biopsy in all. All the boys were followed up at 1 weak, 6 months and 1 year interval. Fairly good sized testes in the scrotal sacs were seen all of them except one atrophied testis of case no. 1.

DISCUSSION

PMDS are otherwise normally differentiated 46XY male. Embryologically, upto 6th week all fetuses have both male (Wolfian) and female (Mullerian) genital ducts. After 7th week, in male fetuses (46XY), the Mullerian ducts regress[3] mediated by Mullerian inhibiting substance (MIS) or anti-mullerian harmone (AMH) produced in immature fetal sertoli cells.[2] While the Wolfian ducts continue to differentiate into epidydimis, vas and seminal vesicle.[3] PMDS is attributed to AMH deficiency or AMH receptor defectivity[2] or AMH may not expressed in the critical period of before 8 weeks of gestation. It is inherited as an autosomal recessive or X-linked recessive mutation of short arm of chromosome 19.[1] Exact pathogenesis is known in about 85% of cases. Type I PMDS (45%) is due to AMH deficiency and type II PMDS (40%) is due to receptor defects and in the remaining 15% the exact cause is unknown.[3] TTE is rare form of ectopic testis which is rarely associated with PMDS. Normal testicular descent is impeded by the close association of the testis and vasa to broad ligament. This mechanical effect of PMD structures prevents testicular descent or leads both testes to descend towards the same hemiscrotum.[1] As the androgen levels are normal, penile development is not affected and testicular histology is not affected apart from lesions due to UDT.[5] Awareness of this phenomenon is essential to avoid labeling these boys as vanishing testis syndrome.[1]

TTE should be suspected in all patients with unilateral hernia with contralateral nonpalpable testis and ultrasound should be done. If TTE is present it is itself an indirect indicator of PMDS.[5] Preoperative imaging can be done using ultrasound, computed tomography and magnetic resonance imaging[6] and diagnostic Laparoscopy. Serum AMH levels remain fairly high till 2 years age, measurable till puberty and later remains undetedtable.[7] Hence Serum AMH levels are useful only in prepubertals.

Overall incidence of testicular tumors in PMDS is about 18%, which is comparable to that of individuals with UDT.[3] There are no reports of malignancy arising from retained MD structures.[1]

Mixed gonadal dysgenesis (MGD) is the differential diagnosis. In MGD there is presence of ambiguous genitalia, unilateral testis, and contralateral streak gonad. In addition mullerian structures are normally present and gender assignment is female, with XO/XY mosaic karyotyping.[1] In contrast PMDS show normal virilisation of male external genitalia, 46XY karyotype and gonadal biopsy is suggestive of testis.

Management is exclusively surgical. The main objectives are preservation of testis with its vascularity and protecting the testis against malignancy with preserving its hormonal functions by open or laparoscopy.[3] The vasa is densly adherent to vagina and can be dissected free only with great difficulty, placing the vas at risk of injury. In fact some surgeons advocate leaving the uterus and other derivatives in situ to avoid possible injury to vasa.[1] It should be done with extreme care and dexterity to avoid ischemic gonadal damage. Removal of MD is not advised, rather it should be pushed back into pelvis.[3] It is a conservative surgical approach[4] by simple orchidopexy, so that the testis is in a easily palpable and accessible position if malignancy occurs.[5] Every effort should be made to preserve the testis and vas for possible future fertility, though fertility has been reported in a very few cases.[4] Hysterectomy is recommended only if PMD structures limit scrotal orchidopexy.[5] Parents should made aware of risk of testicular malignancy and infertlity, including genetic councelling.[8]

CONCLUSION

Awareness amoung the surgeons the possible forms of PMDS and TTE helps to plan the proper line of mangement of this which is encounterd incidentally during opertion. Use of laparoscopy in impalpable testis prevents from wrong labelling of some boys as vanishing testis. Management is by simple scrotal orchidopexy with preservation of vascularity of testis. Risk of testicular tumour and infertility has to be addressed. Parents should be genetically councelled.

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