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Case Report

Transverse testicular ectopia with persistent Mullerian duct syndrome: a rare anomaly

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ABSTRACT

Persistent Mullerian duct syndrome is a disorder of male pseudo-hermaphroditism characterized by persistence of uterus, fallopian tubes and upper two third of vagina in otherwise normally virilized phenotypically and genotypically male (46XY). Aim of study was to create an awareness among the surgeons about the pathophysiological, morphological and clinical presentations of this rare entity along with its management. Between 2009 to 2013 we came across 5 boys with Persistent Mullerian Duct Syndrome (PMDS) with varied clinical spectrum in each, viz hernia, hydrocele, right sided Un-Descended Testis (UDT), left sided UDT and bilateral UDT. All were diagnosed intra-operatively and managed accordingly, 4 by Open Orchidopexy (OP) with 1 requiring midline splitting of the uterus and 1 laparoscopic orchidopexy. Awareness of this rare anomaly associated with inguinal hernia or undescended testis and also diagnostic laparoscopy for impalpable testis helps in appropriate management.

Keywords: Persistent Mullerian duct syndrome, Hernia uteri inguinale, Undescended testes

INTRODUCTION

Von Lenhossek first reported the rare entity of Transverse Testicular Ectopia (TTE) in 1886.[1] Jordan in 1895 described transverse testicular ectopia associated with Persistent Mullerian Duct Syndrome (PMDS).[1] Nelson in 1939 first described this association in a man with inguinal hernia as hernia uteri inguinale.[2] About 150 cases of PMDS have been reported in literature, 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.[3] Patients present with absent testis, hernia, or infertility during infancy, childhood or adulthood.[3] 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) Un-Descended 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.[3]

CASE REPORT

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

Case 1

2 year old boy was brought BL impalpable UDT. Diagnostic Laparoscopy (DL) revealed uterus and fallopian tubes fixed to pelvis and both testes embedded

in broad ligament. Suprapubic exploration and mobilisation were done. Uterus split in midline without damaging the vascularity of testes. Both testes brought separately into scrotum and orchidopexy done. It was female type of PMDS.

Case 2

3 year old boy presented left sided impalpable UDT. DL revealed hernia on right side with both testes on right side. In between the 2 testes uterus and fallopian were present. It was male - sub type I form of PMDS. Herniotomy and orchidopexies were done.

Case 3

2 year old boy was brought with right sided hydrocele. Groin exploration showed fallopian tube which was placed back into the abdomen and herniotomy done. It was males subtype II form of PMDS.

Case 4

5 year old boy presented with right sided obstructed inguino-scrotal hernia. On exploration, the contents were intestinal loop with both testis having PMD structures in between. Bowel reduced. Herniotomy was done and subdartous orchidopexy was done.

Case 5

1 year old boy, sibling of case 2 was brought with right sided hernia. Examination revealed impalpable testis on left side. During herniotomy, we found that some mass attached to the right spermatic cord which on applying traction revealed fallopian tube, uterus and left testis. Herniotomy and orchidopexy were done.

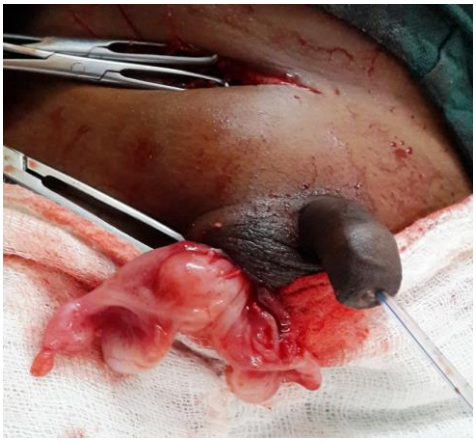

Figure 1: Herniotomy and orchidopexy shows persistent Mullerian duct syndrome.

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.

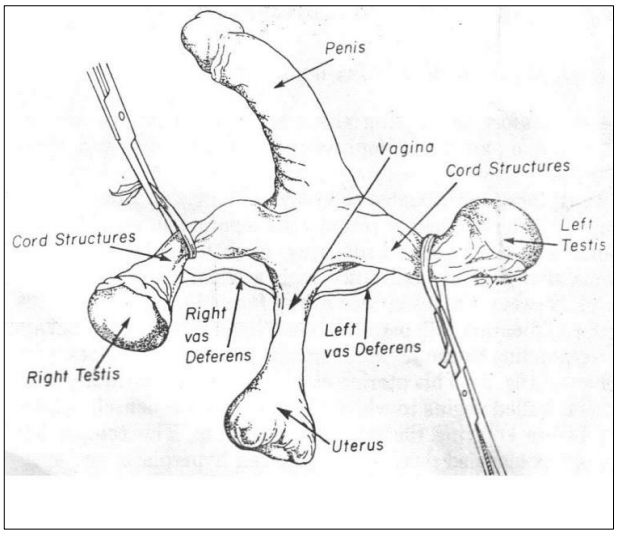

Figure 2: Cord structures of undescended testes.

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.[1] 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.[1] 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 labelling these boys as vanishing testis syndrome.[1]

TTE should be suspected in all patients with unilateral hernia with contralateral non-palpable 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.[7]

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 tumorigenesis 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 gonal 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 densely 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 an 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 make aware of risk of testicular malignancy and infertility, including genetic councelling.[8]

CONCLUSION

Awareness among the surgeons the possible forms of PMDS and TTE helps to plan the proper line of management of this which is encountered incidentally during operation. 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 counselled.

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