Persistent Mullerian Duct Syndrome: Report of Two Boys with Associated Transverse Testicular Ectopia

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Persistent mullerian duct syndrome (PMDS) is an unusual form of male pseudohermaphroditism characterized by the presence of a uterus and fallopian tubes in an otherwise differentiated male with a 46XY karyotype[1]. Since the first report in 1939, approximately 150 new cases have been reported. Most refer to isolated cases, while a few involve siblings[2].

Transverse testicular ectopia (TTE) is a rare condition in which one-testis crosses the midline and both the two testes are found on one inguinal sid[3]. The ectopic testis may lie at the internal ring, in the inguinal canal or in the scrotum. Lenhossek in 1886 first described this entity and since then about 100 cases have been reported[4].

The association of PMDS and TTE is recorded in adult literature[5], but to date only 9 pediatric cases have been reported in the English literature (Table)[6,7].

Table . Clinical data of the cases of PMDS associated with TTE.

Study	Year	Age	Side	Hernia	Mullerian structure
Pellegrini[4]	1913	2 years	Right	Present	uterus and tubes
Stonham[4]	1828	9 months	Left	Present	uterus
Link[4]	1930	1.5 year	Right	Present	uterus and tubes
Stauber[4]	1965	10 month*	Right	Present	uterus and tubes
Stauber [4]	1965	3 month*	Left	Present	uterus
Fourcroy[5]	1982	4 years	Right	Present	mullerian duct
Mahfouz[6]	1990	3 month	Right	Present	uterus and tubes
Mahfouz[6]	1990	27 days	Right	Present	uterus and tubes
Karnak[7]	1996	9 month	Right	Present	uterus
Mandhan et al (two cases)					
Case 1	1997	2 year	Left	Present	uterus and tubes
Case 2	1998	7 month	Right	Present	uterus

*Siblings

Two new cases are reported.

Case Report

Case 1

A 2 year old boy was admitted with a left inguinal hernia. His left gonad was palpable in scrotum, right half of scrotum was empty and penis was normal in appearance. He also had an umbilical hernia. Parents were not related and there was no history of inguinal hernia or sexual ambiguity in siblings. A diagnosis of left-sided inguinal hernia, right undescended testis and umbilical hernia was made. During surgery for the left inguinal hernia when traction was applied on the sac, another gonad and a tubular structure were unexpectedly encountered coming from abdomen through internal ring. Hernia was repaired and all structures were placed in their original position after taking biopsies from two gonads and tubular structure. Further work up revealed 46XY karyotype, presence of a uterus in the pelvis on abdominal ultrasound and a normal cystourethrogram. Histologically the two gonads were testis and tubular structure as infantile uterus (Figure).

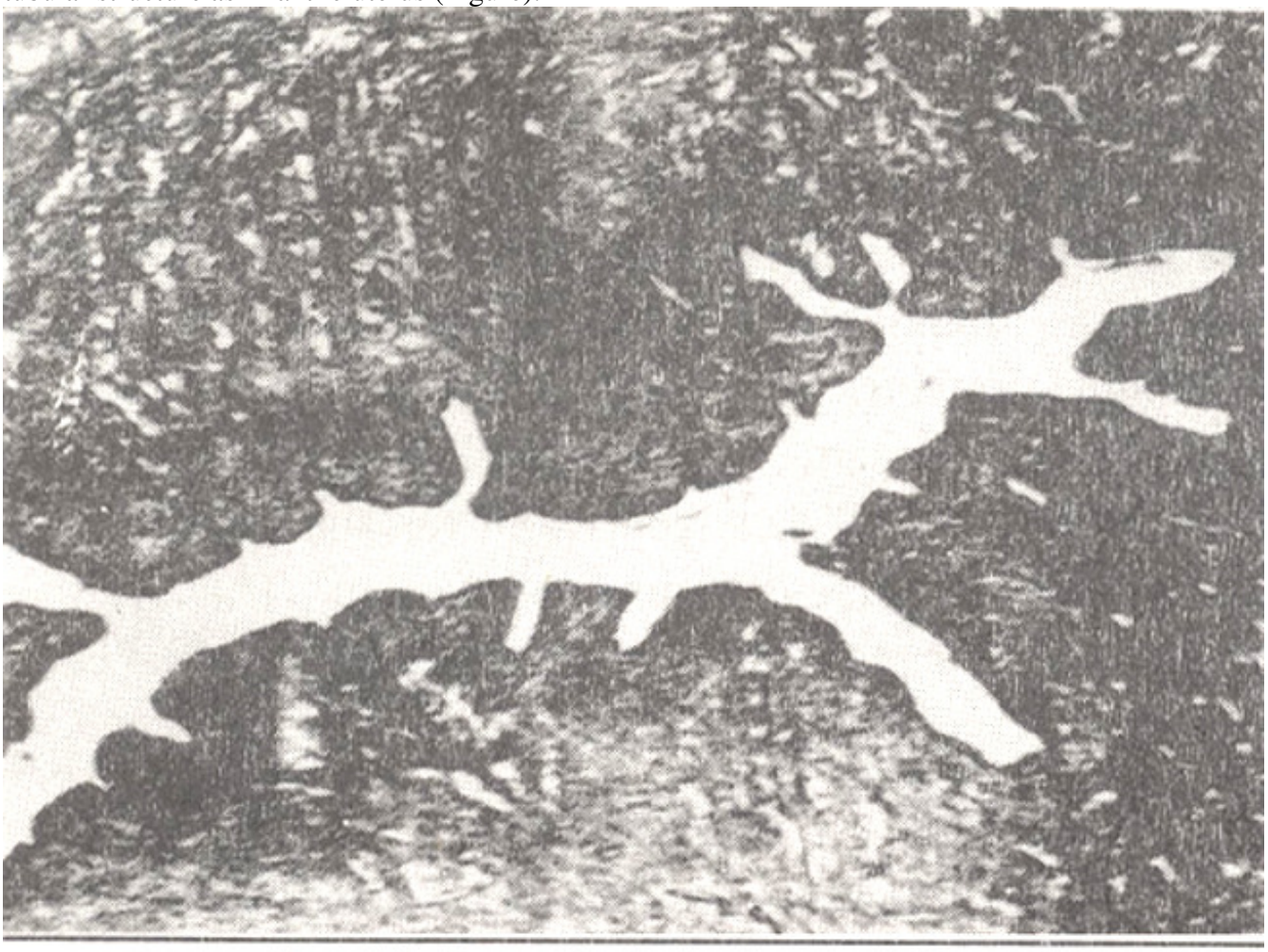

Figure. Infantile uterus lined with columnar epithelium.

Exploration through a transverse subumbilical incision showed the uterus and two fallopian tubes to be present in the midline behind the urinary bladder, the right testis was in the left pelvis with its separate blood supply and vasa deferentia were in close relation with the fallopian tubes and uterus. The vas vessels of right testis were mobilized carefully from the mullerian remnants and the relation with the fallopian tubes and uterus. The vas vessels testis was fixed in right subdartos pouch through the same inguinal canal. Both fallopian tubes and uterus were excised leaving a pedicle of myometrium intact. The umbilical hernia was also repaired. Postoperative course was uneventful.

Case 2

A 7-month-old boy was admitted for right inguinal hernia with empty and underdeveloped left hemiscrotum. His penis was normal in appearance. During surgery for the right inguinal hernia when sac was pulled, an additional gonad came out through the internal ring from the abdominal cavity. Biopsies of the two gonads were and the right inguinal hernia was repaired after replacing the two gonads in their respective position. Further work up showed the presence of a small uterus in the pelvis on abdominal ultrasound, normal cystourethrograrn and 46XY karyotype. The two gonads were. testes on histological examination. Laparotomy carried out by a transverse subumbilical incision revealed a small uterus present posterior to the urinary bladder and the left testis in the right pelvis with its separate vessels and vas. No fallopian tubes were identified. The vas and vessels of left testis were mobilized and it was placed in left subdartos pouch through the same inguinal canal. The uterus was excised leaving intact a pedicle of myometrium. Postoperative course was smooth and histopathological report confirmed the findings of a uterus.

Discussion

Mullerian duct structures (MDS) are present in the male fetus until the eighth week of gestation and their regression is mediated by the mullerian-inhibiting factor (MIF), a glycoprotein produced by fetal Sertoli's cells. Failure of synthesis or release of MIF, the defect of end organs to respond to MIF or error in the timing of release of MIF causes persistence of MDS in male fetuses'. Transverse testicular ectopic (TTE) is one of the rarest forms of testicular ectopia with uncertain embryological etiology. An aberrant gubernaculum, tes.ticular adhesion, fusion and adhesion of developing wolffian ducts and the traction on a testis by PMS are the suggested embryological explanation[1,8]. It seems possible that the mechanical effect of the PMS prevents testicular descent and also drags both testicles towards the same inguinal side or hemiscrotum and thus produces TTE.

Preoperative diagnosis of TTE is suspected when the child presents with an empty hemiscrotum with an additional mass in the contralateral hemiscrotum. In presence of TTE the possibility of PMDS should be suspected as it is a common cause of this abnormality. However, the diagnosis is often made incidentally during the operation of inguinal hernia or during exploration for undescended testis. When two gonads of approximate size and shape alongwith mullerian duct remnants are encountered during inguinal operation, further work up for associated malformations and sex determination should be considered before abdominal exploration. A complete radiological evaluation in TTE is necessary because a variety of associated genitourinary anomalies have been reported[9]. Although no additional genitourinary malformations were encountered in our cases, the association of an umbilical hernia has not been recorded in literature previously.

The standard approach to the patients of PMDS with associated TTE includes gonadal biopsy to confirm the sex and exclude the possibility of mix gonadal dysgenesis, removal of mullerian structures, fixation of two testes in their respective hemiscrotum and inguinal hernia fixation. Any tissue between the vasa deferentia should be sent for histopathology to identify the mullerian remnants. Excision of MDS is considered essential to prevent sexual maturation hypertrophy of uterus and accumulation of blood causing abdominal discomfort and mass. Current surgical recommendation for MDS include the proximal salpingectomies and a hysterectomy leaving intact a pedicle of myometrium as this facilities preservation of fertility and hormonal functions, especially when the child is young10. For TTE, transseptal fixation or modified Ombredanne technique, in which both cords and vessels of two testicles pass through the same inguinal canal, is recommended, but this carries a risk of damage to both vas and vessels of two testis whenever there is trauma or infection7. Thus we fixed both the ectopic testes through the ipsilateral inguinal canals into their respective hem i scrotum.

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