Abstract: A type of Male pseudohermaphroditism called persistent mullerian duct syndrome (PMDS), is a rare condition characterized by the presence of uterus and oviducts in phenotypic males (46xy). We report a case of PMDS presenting as hernia utero inguinalis with transverse testicular ectopia, normal testicular histology and primary infertility with normal male sexual function.

Keywords: Persistent mullerian duct syndrome, hernia utero inguinalis

Introduction
Persistent Mullerian duct syndrome (PMDS) is a rare form of internal male pseudohermaphroditism in which Mullerian duct derivatives are seen in a male patient. This syndrome is characterized by the persistence of Mullerian duct derivatives (i.e. uterus, cervix, fallopian tubes and upper two thirds of vagina) in a phenotypically and karyotypically male patient. The syndrome is caused either by an insufficient amount of Mullerian inhibiting factor (MIF) or due to insensitivity of the target organ to MIF.

Case Report
A 30 year old male presented to out patient department for a right inguinoscrotal swelling, which was present since 1 year and associated with pain for the past 1 week. Detailed history revealed an associated infertility and he has been married since 4 years. He reported normal sexual activity and had well developed secondary sexual characters such as facial, axillary and pubic hairs with male pattern voice. No significant family or personal history noted.

Per abdomen examination was unremarkable with no signs of intestinal obstruction. Right sided inguinal hernia was present which was partially reducible without any signs of strangulation. On genital examination, he had male genitals with normal appearing penis along with left sided cryptorchidism. With a clinical diagnosis of right sided uncomplicated inguinal hernia with left sided cryptorchidism against a background of primary infertility, patient was investigated and found to be azoospermic on semen analysis, normal testosterone level, with ultrasound showing absent left testis in left inguinal region and right testis in right inguinal hernia with some ill defined mass in the hernial sac.

Patient was operated under spinal anesthesia. Surprisingly, the right hernial sac contained the right and left well developed testicular substances, between which an underdeveloped uterus like and bilateral fallopian tube with fimbria like structures found. Other contents of omentum reduced. Since, in order to explain and to obtain consent from the patient, these structures are not removed in that first setting of open surgery.

Later in the post operative period, patient and his wife were counseled and with their consent, patient underwent second sitting procedure under general anesthesia and the structures of uterus, fallopian tubes, vaginal tube up to its entry into prostate and both testis were removed laparoscopically, followed by trans abdominal pre-peritoneal mesh repair done on right side. Post operative course was uneventful.

INGUINAL SAC CONTAINING UTERUS AND BOTH TESTIS

A CASE OF PERSISTENT MULLERIAN DUCT SYNDROME PRESENTING AS HERNIA UTERO INGUINALIS
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And there was no evidence of spermatogenesis. Patient was regularly followed and on testosterone replacement therapy.

Laparoscopic specimen extraction
With consent from the patient karyotyping was done that revealed 46XY genotype and tumor markers like beta HCG and alpha fetoprotein were within normal limits. Histopathology of the specimen revealed normal endometrial, myometrial and cervix and fallopian tube histology with normal seminiferous tubules lined by sertoli cells and foci of leydig cells. No epididymal structures found. And there was no evidence of spermatogenesis. Patient was regularly followed and on testosterone replacement therapy.

Cut section of Uterus showing normal endo and myometrium

Discussion
Müllerian (paramesonephric) ducts and wolffian (mesonephric) ducts are the anlagen of the female and male reproductive tracts, respectively. In the XY fetus, the testis differentiates by the end of the seventh gestational week. Sertoli cells begin to secrete AMH, which is responsible for the regression of the Mullerian ducts. The AMH binds to a specific Type II serine-threonine kinase transmembrane receptor (AMHR-II). Human AMH gene localized near the tip of Chromosome 19, AMHR2 gene is located on 12q13. The type of persistent Mullerian duct syndrome caused by mutation in the AMH gene will be referred to as Type I, that which forms due to mutation in the AMH receptor (AMHR) will be designated as Type II.[1] In 45%, a mutation of the anti-mullerian hormone (AMH) gene was detected; in 39% mutation of the Type II receptor of AMH was detected; in 16% the cause is unknown.

Transverse testicular ectopia (TTE) or crossed testicular ectopia is a rare form of testicular ectopia. It was first reported by Von Lenhossek in 1886 [2]. More than 100 cases have been reported in the literature [3]. Several theories have been reported to explain the genesis of TTE. Berg [4] proposed the possibility of the development of both testes from the same genital ridge. Kimura [5] concluded that if both vasa deferentia arose from one side, there had been unilateral origin but if there was bilateral origin, one testis had crossed over. Gupta and Das [6] postulated that adherence and fusion of the developing Wolffian ducts took place early, and that descent of one testis caused the second one to follow. An inguinal hernia is invariably present on the side to which the ectopic testis has migrated.

On the basis of the presence of various associated anomalies, TTE has been classified into 3 types: Type 1, accompanied only by hernia (40% to 50%); type 2, accompanied by persistent or rudimentary Mullerian duct structures (30%); and type 3, associated with disorders other than persistent Mullerian remnants (inguinal hernia, hypospadias, pseudohermaphroditism, and scrotal abnormalities) (20%). According to that classification, our case was type 1/2 TTE. TTE associated with fused vas deferens is extremely rare. This condition may hinder the testis from being placed into the scrotum during orchidopexy [7]. The clinical presentation generally includes an inguinal hernia on one side and a contralateral or sometimes a bilateral cryptorchidism [8], [9]. Usually, the correct diagnosis is not made before surgical exploration, like our case, and it is revealed during herniotomy [9]. The diagnosis of TTE can be made preoperatively by using ultrasonography [10] by an experienced sonologist. Patients with TTE are at increased risk of malignant transformation. In fact, the overall incidence of malignant transformation of gonads is 18% [11]. There have been reports of embryonal carcinoma [12], seminoma, yolk sac tumor [13], and teratoma [11]. Walsh et al. [14] in their study concluded that testicular cancer was nearly 6 times more likely to develop in cryptorchid cases whose operations were delayed until after age 10 to 11 years. Wood et al. [15] in their study showed that risk of malignancy in undescended testicles decreased if their orchidopexy performed before ages 10 to 12 years.

Orchidectomy of ectopic testis should be done, because orchidopexy offers only limited protection against future malignancy if performed after two years of age.[16] Manassero et al reported development of mixed germ cell tumor 18 years after bilateral orchidopexy.[17] Most are known to be infertile but it is preferable to remove ectopic testis, as it is prone for malignancy. If this is necessary on both sides, there is the additional problem of lifelong testosterone substitution which requires efficient patient monitoring and good patient compliance. In cases where this cannot be achieved, compromises, such as temporarily delayed orchidectomy, may be considered.[18] Testis, vas and epididymis are closely adherent running along the uterus and fallopian tubes. This gives rise to difficulty in separating the gonads and the vas without damage. Different surgical methods have been described for safe surgery. There have been at least three documented reports of adenocarcinoma in the mullerian duct remnants. So, contrary to previous suggestions, now it is recommended to remove the persistent mullerian derivatives.

The patient or his family should be completely informed of the diagnosis, the surgical options and the need for long-term follow-up. Finally, genetic counseling must be offered to the patient or his parents because of the possible chromosomal origin of the syndrome.

References

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