Case Report

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Surgery Section

A Rare Case Report of Inguinal Hernia with Persistent Mullerian Duct and Klinefelter Syndrome

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ABSTRACT

Inguinal hernia in male is a common problem but having female reproductive organs in hernial sac is rare. It occur because of failure of mullerian duct to regress in a male fetus during embryonic development, result in a syndrome known as Persistent Mullerian Duct Syndrome (PMDS), which is a rare entity of male pseudohermaphroditism. We hereby present a case of 21-year-old male patient reported with complains of cryptorchidism and inguinal hernia. Generally diagnosis of PMDS was established during investigation like ultrasonography, MRI for localization of undescended testis and during surgical exploration for inguinal hernia or cryptorchidism. Our patient was operated by bilateral inguinal incision; hernial sac contained adult size uterus fallopian tube and upper 2/3rd of vagina. On karyotyping it was found that he was a case of klinefelter syndrome also. Association of PMDS with klinefelter syndrome is very rare.

Keywords: Male pseudohermaphroditism, Persistent mullerian duct syndrome, Undescended testis

CASE REPORT

A 21-year-old, tall muscular gentleman, farmer by occupation, presented with right sided absent testis and left inguinal region swelling since childhood. Swelling on left side was initially small and reducible, but it had increased in size, reached upto base of scrotum, became irreducible and painful from last 10 days. There were no cough impulse over left inguinal swelling, getting above the swelling was not appreciable.

Per abdomen examination was normal, there were no abdominal distension, bowel sound was present, bowel and bladder habit was normal, per rectal examination was normal. Rest of the systemic examination was normal.

USG suggested absence of testis in right scrotum and inguinal canal, left testis was found in left scrotal sac. MRI whole abdomen showed that right testis was not seen in scrotum, inguinal canal and pelvis, left testis was seen in scrotum [Table/Fig-1].

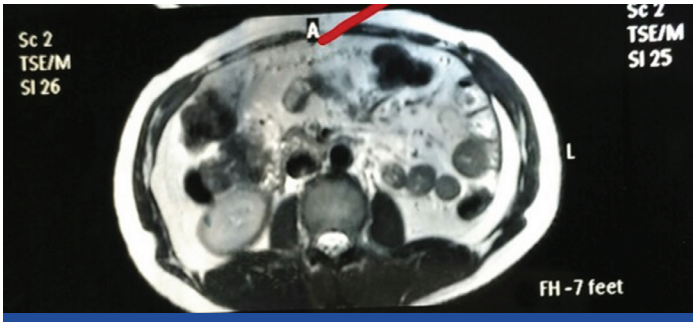
He was operated upon through bilateral inguinal incision under spinal anaesthesia. On right side no testis was found and on left side hernia sac contained an adult –sized uterus, fallopian tubes, broad ligament, vas deferentia and both testis, which were atrophied. Total inguinal hysterectomy with bilateral salpingectomies, bilateral orchidectomy and mesh hernioplasty [Table/Fig-2,3] were performed through the same incision.

Histopathological examination of the resected specimen revealed uterus, cervix and bilateral adnexa. Both gonads identified on either side. Microscopic examination from cervix and endomyometrium showed focal presence of mullerian epithelium with remnants of the mesonephric duct [Table/Fig-4]. Fallopian tube on both side were lined by mullerian epithelium. The adjacent parametrial tissue showed ductal differentia with epididymis.

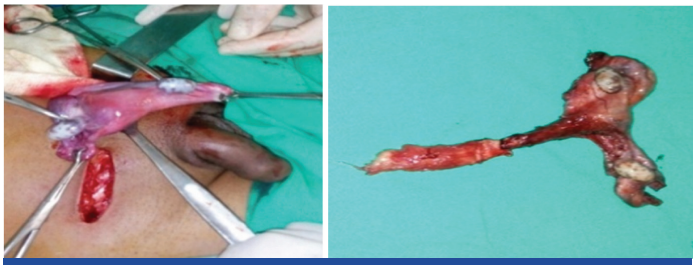
Section from both testis revealed many sclerosed and hyalinized 'ghost' seminiferous tubules with reduced spermatogonia. Spermatogenesis is absent [Table/Fig-5]. Overall histomorphology favours mullerian duct syndrome with marked testicular atrophy.

Post operative hormonal assay showed testosterone 31.41ng/dl (reference value 241-827ng/dl), estradiol 12pg/ml (11- 44pg/ml), dehydroepiandrosterone sulphate 119ug/dl (238-539 ug/dl), progesterone 0.60ng/ml (0.28-1.22ng/ml).

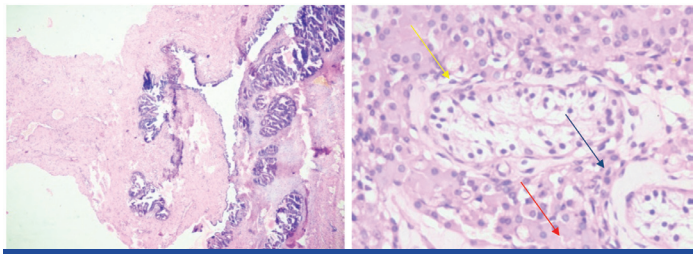
Chromosomal analysis (GTG-banding with 550 band resolution) revealed 47, XXY pattern in all the cells analysed. This chromosomal complement confirmed klinefelter syndrome.



[Table/Fig-1]: MRI showing Right testis not seen in scrotum, inguinal canal or in abdomen. RED Arrow - Iso to hypointense mass abutting the gut loop.



[Table/Fig-2]: Pre-operative view showing uterus fallopian tube, testis, and penis. **[Table/Fig-3]:** Resected specimen.



[Table/Fig-4]: Endometrium 4x – endomyometrium shows focal presence of mullerian epithelium with remnants of the mesonephric duct aggregated in the mid portion of the wall. **[Table/Fig-5]:** Tetis with 40x Stained with haematoxylin and eosin; Yellow arrow leyding cell hyperplasia, Blue arrow no mature sperm seen, Red arrow sertoli cells.

DISCUSSION

During development, all embryos have both Wolffian ducts and Müllerian ducts. In this stage, the internal organs are bipotential, meaning they have the potential to develop into both male and female sex organ [1-4].

Male differentiation is determined by SRY gene located on Y chromosome [5]. Sertoli cells of testis begin to secrete AMH (Anti

Mullerian Hormone), which is responsible for the regression of the Mullerian ducts. Remaining duct i.e., Wolffian ducts develop into the vas deferens and the seminal vesicle [2].

When mullerian duct fails to regress in a male fetus because of some pathology results in a syndrome known as persistent mullerian duct syndrome (PMDS), In it male fetus, have wolffian derivatives male structures along with female structure of mullerian derivatives (uterus, fallopian tube upper two third of vagina) also. PMDS is inherited as an autosomal recessive [1] or X-linked recessive mutation of short arm of chromosome 19 [4,6]. Exact pathogenesis is known in about 85% of cases [7]. Two main types are known. 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 [4,6,8,9].

AMH is a glycoprotein secreted by Sertoli's cells of the fetal testes. AMH, also known as Mullerian Inhibitory Substance (MIS), has two transmembrane receptors in the mesenchyme of fetal Mullerian ducts. Type-I receptors are non-specific while type-II AMH receptors are specific for its actions [1,6,8,9].

In PMDS normal testicular descent is impeded by the close association of the testis and vasa to broad ligament, it prevents testicular descent or leads both testes to descend towards the same hemiscrotum [4,6]. As the androgen levels are normal, penile development is not affected and testicular histology is not affected apart from lesions due to UDT [1].

Morphologically PMDS occur in male and female form. Female form (10-20%) is characterized by the presence of bilateral cryptorchidism with no herniation of Mullerian duct structures and testes. Uterus and fallopian tubes are fixed in the pelvis and testes are embedded in the broad ligament. Male form (80-90%) is characterized by the presence of unilateral cryptorchidism with contralateral inguinal hernia containing the Mullerian structures and the testes [1,6,8,9]. Male form is subdivided into two types. In type-I, hernia sac contains uterus, both fallopian tubes and both testes (hernii uteri inguinalis with TTE). Our patient belonged to type-I. In type-II, hernia sac contains uterus, ipsilateral fallopian tube and ipsilateral testis (classic hernii uteri inguinalis) [8-10].

One testis is usually in the scrotum, the uterus and fallopian tube being pulled into the canal by traction on the undescended testis. The contralateral testis and the fallopian tube may also appear in the hernia sac, as in our patient.

In present case, Mullerian duct derivatives were discovered unexpectedly during herniorrhaphy or surgical exploration for cryptorchidism. Overall incidence of testicular tumourigenesis in patients with PMDS is about 18% [4,10,11], which is comparable to that of normal individuals with undescended testes. Various testicular tumours that have been reported in such patients include seminoma, teratoma, embryonal carcinoma, choriocarcinoma, mixed germ cell tumour, leiomyoma and adenocarcinoma of uterus. Diagnosis of PMDS is established when Mullerian duct structures are discovered incidentally either during routine imaging for localization of undescended testes or surgical exploration for cryptorchidism or inguinal herniorrhaphy.

Karyotyping should also be done to find out any chromosomal abnormalities associated with PMDS, As seen in our patient, he had klinefelter syndrome along with PMDS.

If patient presents in pediatric age the initial procedure may need to include replacement of the gonads and Mullerian structures within the pelvis and repair of the inguinal hernia [4,7]. After confirmation of the diagnosis of PMDS, definitive surgery should be performed to remove the corpus of the uterus and fallopian tubes to enable fixation of the testes in the scrotum.

If patient present during adult life total hysterectomy, bilateral salpingectomies and orchidectomies become imperative if testis could not be brought to a palpable position, testes are atrophic or there is strong suspicion of malignant transformation [10,11]. However, before proceeding to such radical surgery, it is highly desirable that the patient and his family should be thoroughly counseled [5] about the diagnosis, the different surgical options and the need for long-term follow-up and androgen replacement therapy after orchiectomy. It is also recommended that an informed written consent should be obtained before surgery.

CONCLUSION

By this case report we want to emphasise that PMDS is a rare entity and proper evaluation should be done, when patient present to us with unilateral undescended testis and contralateral inguinal hernia. Semen analysis and investigation to locate undescended testis like USG or MRI abdomen should be done. Diagnostic laproscopy is helpful to locate intra abdominal testis as well as to find out this kind of rare entity. When diagnosis of PMDS is confirmed then karyotyping should be performed to identify other syndrome associated with it as in our case klinfelter syndrome was there. Disease and its treatment should be explained to patient and his attendant and after proper consent surgery should be performed. If bilateral orchidectomy is done then lifelong supplementation of testosterone should be started.

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