CASE REPORT

Persistent Mullerian Duct Syndrome in a Post Orchideopexy Patient with Gyanaecomastia and Hypospadias: A Case Report

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Abstract:

Persistent Mullerian Duct Syndrome (PMDS), a rare form of male pseudohermaphroditism. It is characterized by the persistence of Mullerian duct structures (uterus, fallopian tubes and upper two-thirds of vagina) in otherwise normally virilized males (Karyotype 46XY). The exact cause of PMDS is not known, however it is thought to result from the defect of the synthesis or release of Mullerian inhibiting factor (MIF) or from a MIF receptor defect. Herein we report a case of PMDS with gynaecomastia and hypospadias in a post orchidopexy patient.

Keywords: Pseudohermaphroditism, Mullerian duct, Mullerian inhibiting factor

Introduction:

Persistent Mullerian Duct Syndrome (PMDS) is a rare form of internal male pseudohermaphroditism, in which mullerian duct derivatives are present in a genotypic (46XY) and phenotypic male [1]. Over 150 cases have been reported in the literature, mostly in adults. Typically, these phenotypic males have unilateral or bilateral undescended testes, bilateral fallopian tubes, a uterus, and an upper vaginal draining into a prostatic utricle. PMDS results from either a deficiency of Anti-Mullerian Hormone (AMH) activity or by an abnormality in its receptor anti-mullerian hormone, produced by fetal testicular sertoli cells, is responsible for the involution of embryonic mullein structures in normal males [2].

Case Report:

A 16 year male patient came with history of swelling of left testis since one week. No history of trauma. No history of fever, vomiting, patient had undergone left laparoscopic orchidopexy five years back. General physical examination revealed bilateral gynaecomastia (Fig.1).

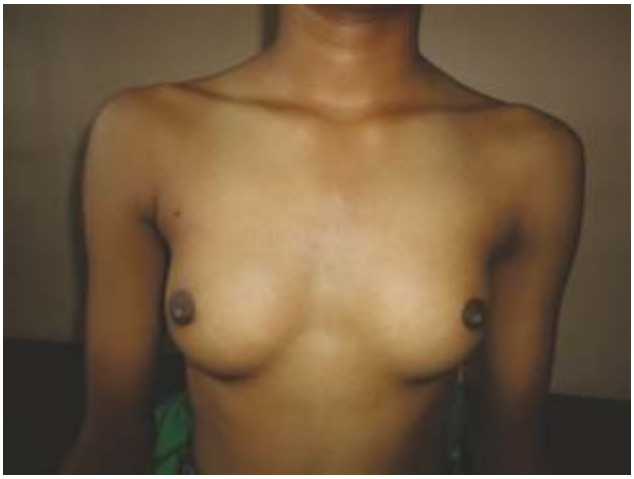

Fig. 1: Clinical Photograph Showing Significant Bilateral Gynaecomastia

Local examination showed a solitary swelling in left scrotum oval in shape, bosselated surface, non tender, spermatic cord was not palpable. Swelling was measuring 9x7x5 cm in size. Right testis was normal. Genital examination revealed penoscrotal hypospadias with chordee. Urinalysis, complete blood count and serum electrolyte were normal. Hormonal profile details are tabularized as below Table 1.

Table 1: Shows Hormone Profile

Tests	Test Values	Reference Value
Luteinizing hormone	11.8 mIu/ml	Men 1.1-7.0 Female 2.0-25
Testosterone	4.47 ng/ml	Male 3.0-10.6
Follicle-stimulating hormone	32I U/L	Male 4-10 Female 10-20
Prolactin	24.2 ng/ml	Men 3-25 Female 5-35

Ultrasound of scrotum showed left testis was completely liquefied and replaced with hypoechoic collection features suggestive of left pyocoele/testicular abscess. right funicultis with epidymoorchitis. But this ultrasound finding was not clinically correlating; hence we planned to get a Magnetic Resonance Imaging (MRI) of inguinoscrotal region.

MRI revealed large multi loculated cystic collection noted in between layers of tunica vaginalis which showed features suggestive of high cholesterol/fat content-with features of chylocoele, with non visualization of testis and epididymis–liquefation. Patient underwent left inguinal exploration and the swelling was removed in Toto and sent for histopathology examination (Fig.2 and Fig, 3). The postoperative course was uncomplicated.

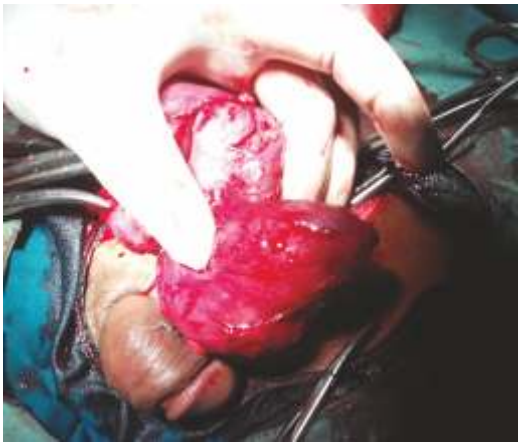

Fig. 2: Intraoperative Findings of Uterus, Fallopian Tubes and Ovaries.

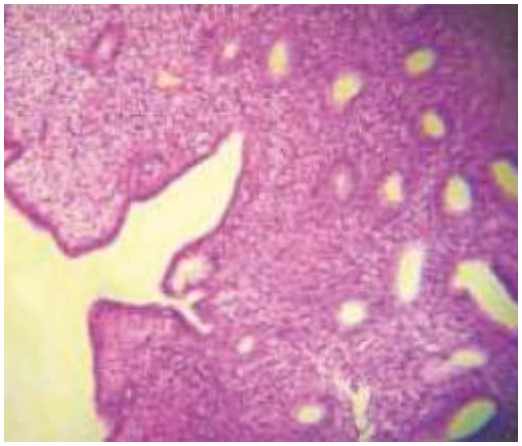

Fig. 3: Photomicrograph Showing Histopathology of Fallopian Tube (H and E, X20)

Discussion:

Persistent Mullerian Duct Syndrome (PMDS) is rare, characterized by the presence of well developed or rudimentary uterus, cervix, vagina and fallopian tubes in normal 46XY male. It is a familial syndrome associated with autosomal recessive mode of inheritance [3]. Typically these phenotypes were unilateral or bilateral undescended testis or bilateral fallopian tubes, uterus or upper vagina draining into prostatic utricle. The Mullerian Inhibiting Substance (MIS) is a large glycoprotein that sertoli cells produce early in fetal life. The gene responsible for the substance is on chromosome 19 [4]. The primary function of MIS is to cause regression of the Mullerian (paramesonephric) ducts in the male fetus. MIS is first secreted in effective amounts 56-62 days after fertilization, and the process of Mullerian regression is normally completed by about Day 77, after which the Mullerian tissue is no longer sensitive of MIS [5].

Two anatomic variants of PMDS have been described, the male type and the female type [6]. The most common variant is the male form, encountered in 80-90% of cases and characterized by unilateral cryptorchidism with a contralateral inguinal hernia. The male form of PMDS can be of two types. The first type is hernia uteri inguinalis, which is usually characterized by a descended testis and herniation of the ipsilateral corner of the uterus and the ipsilateral fallopian tube into the inguinal canal. The second type is crossed testicular ectopia, which is characterized by herniation of both testes and the entire uterus and both fallopian tubes.

The diagnosis of PMDS is made incidentally during surgical exploration for cryptorchidism or herniorraphy as the mullerian remnants are not palpable on abdominal, rectal or scrotal examination. Intraoperative methods of diagnosis, especially the gonadal biopsy, can be performed to rule out mixed gonadal dysgenesis and developing malignancy [7]. Diagnosing PMDS is based on a combination of anatomic and clinical findings. Imaging features, although classic are often missed. Ultrasound scan and MRI failed to identify the structures in our patient too. Management of PMDS is exclusively surgical. The main therapeutic objectives are preservation of testes, spermatogenesis and fertility and protection against testicular malignancies. Open or laparoscopic orchidopexy is a preferred surgical option in prepubertal patients [8].

Fertility has been reported rarely in a few cases. Martin *et al* [9] had reported a 32 year old man with transverse testicular ectopia and a persistent mullerian duct that had a normal sperm count, but the motility index was zero, implying an intrinsic defect in spermatogenesis. There have been reports of different types of malignancy, such as seminoma, teratoma, yolk sac carcinoma, embryonal carcinoma and choriocarcinoma in postpubertal patients. Infertility is frequent in PMDS because most are axoospermic [10].

Conclusion:

The treatment of PMDS requires correct diagnosis of the condition and distinguishing it from other intersex disorders. In all such cased karyotyping, and testicular biopsies are helpful. This helps to ascertain the genetic sex and present of functional testicular tissue. Serum AMH levels also help in diagnosis before puberty.

The clinicians should be aware of the entity of PMDS while dealing with the cryptorchid patients. This helps to reduce and foresee complications like infertility and neoplastic transformation.

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