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Case Report: Persistent Mullerian Duct Syndrome

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Abstract : Persistent Mullerian duct syndrome(PMDS) is a rare, autosomal recessive intersex disorder characterized by the presence of completely developed Mullerian duct derivatives in an otherwise normally virilized male with a 46, XY karyotype. We report a case of 15year-old boy presenting with right inguinal pain, which was clinically found bilateral undescended. On ultrasonography (USG) uterus and bilateral undescended testis found, on Magnetic resonance imaging (MRI) similar findings noted. Normal male genotype found on karyotyping. Final diagnosis of PMDS was made following operation and bilateral gonadal biopsy.

Keywords : Persistent Mullerian duct syndrome, ultrasonography, MRI, karyotyping, genotype.

Introduction :

Persistent Mullerian duct syndrome (PMDS) refers to a form of internal male pseudohermaphroditism characterized by the presence of Mullerian duct derivatives (uterus, Fallopian tube, upper part of vagina) in an otherwise normal male[1]. The PMDS patients have unilateral or bilateral cryptorchidism and are assigned to the male sex at birth without hesitation. They are genetically 46 XY, with a normal male phenotype, no chromosomal abnormalities and, normal testosterone production and responsiveness.[2] PMDS is thought to result from the failure of synthesis or release of Mullerian inhibiting factor (MIF), the failure of end-organ to respond to MIF, or a defect in the timing of the release of MIF. [3]

Case report :

A 15 year-old male presented with complaint of right inguinal pain with burning urination since one month duration. A clinical examination showed bilateral empty scrotum with normal a normal phallus. USG revealed right-sided undescended testis located at the deep inguinal ring with right sided indirect inguinal hernia. Left testis near the right side of base of urinary bladder. . [Figure 1]. Uterus with endometrium also found, cystitis changes noted. [Figure 2]. MRI is done to assess the ultrasound findings showed the similar findings of USG. Karyotyping done showed the normal male genotype (44XY).

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The gross examination showed a well formed uterus with separate two testes with epididymus. [Figure 2].

Discussion :

Persistent Mullerian duct syndrome (PMDS) is a rare, autosomal recessive intersex disorder characterized by the presence of completely developed Mullerian duct derivatives in an otherwise normally virilized male with a 46, XY karyotype.[1]

The affected individual is otherwise normally virilized, with normal testosterone production, and male external genitalia.[3] PMDS was first described by Nilson in 1939, as quoted by Acikalin et al.[4] subsequently; approximately 150 cases have been reported.

Two anatomical forms of PMDS have been described. The association of unilateral cryptorchidism and contralateral hernia characterizes the most common form. One testis descends into the scrotum, and the ipsilateral uterus and fallopian tube either enter the inguinal canal, a condition known as hernia uteri inguinalis, or can be dragged into it by gentle traction, bringing the contralateral testis and fallopian tube in their wake. Often, no traction is necessary, because the contralateral testis is already in the hernia sac. Transverse testicular ectopia, as this condition is called, is extremely common in PMDS.

More rarely, PMDS takes the form of bilateral cryptorchidism—the uterus is fixed in the pelvis, and both testes are embedded in the broad ligament. These clinical variants are not determined genetically and may occur within the same sibship [1].

The exact cause of PMDS is not known; however, it is thought to result from the defects of the synthesis or release of MIF or from the MIF receptor defect. The MIF

gene has been localized to the short arm of Chromosome 19. MIF, released by the Sertoli cells of the fetal testis from seven weeks gestation onwards, is responsible for the regression of the Müllerian duct in the male fetus. A defect of the MIF gene leads to the persistence of the uterus and the fallopian tube in the male. It is likely that these remnant Müllerian structures produce cryptorchidism by hindering the normal testicular descent mechanism. [3]

Two clinical variants of PMDS are encountered. The more common variant of PMDS is characterized by unilateral cryptorchidism and contralateral inguinal hernia. When in such patients the uterus is present in the hernial sac, the condition is referred to as hernia uteri inguinalis. In transverse testicular ectopia, both the testes are located on one inguinal side and the opposite inguinal canal and scrotum are empty. In the rarer variant, patients may present with bilateral cryptorchidism where the uterus is in the pelvis and both the testes are embedded in the broad ligament. In the PMDS-affected individuals, the testis is usually histologically normal, apart from the spectrum of lesions due to longstanding cryptorchidism. [2,3,5]

Like other undescended testes, the gonads of these patients are at increased risk of malignant transformation. There have been case reports of embryonal carcinoma, seminoma, yolk sac tumor and teratoma in patients of PMDS. [4] The overall incidence of malignant change has been found to be 15%. [2] In our case, testicular tissue showed no malignancy.

Infertility is common with absence of spermatozoa seen at semen analysis. There have been a few reported cases of fertility, although absolute proof of paternity was not established[5].

The main therapeutic considerations are the potential for fertility and prevention of malignant change. Surgical management is geared towards preserving fertility and orchipexy is done to retrieve the testis and position it in the scrotum. During the surgery, the uterus is usually removed and attempts are made to dissect away Müllerian tissue from the vas deferens in order to preserve fertility. Orchidectomy is only indicated for testis that cannot be mobilized to a palpable position.

Testosterone replacement is required by those undergoing orchidectomy or by those with low levels of testosterone. The patient and his family must be informed about the diagnosis, the associated complications and the surgical options. Genetic counseling must be offered to them because of the possible chromosomal origin of the syndrome[6]. In conclusion, while dealing with the cryptorchid patients, radiologists should be well aware of the entity of PMDS. These patients are prone for complications like infertility and neoplastic transformation.

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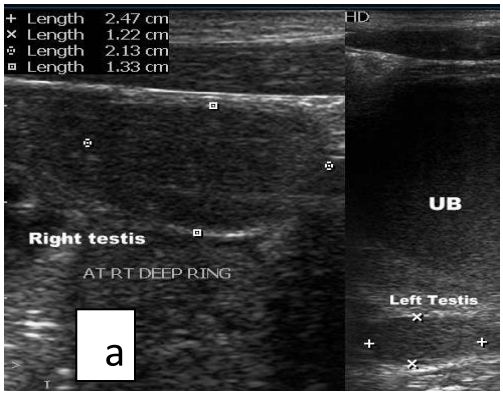

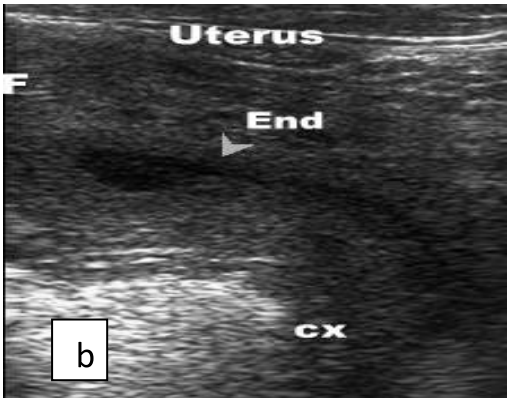

Fig.1 :a) On USG revealed right-sided undescended testis located at the deep inguinal ring with right sided indirect inguinal hernia. Left testis near the right side of base of urinary bladder.b) USG image shows Uterus with endometrium also found.

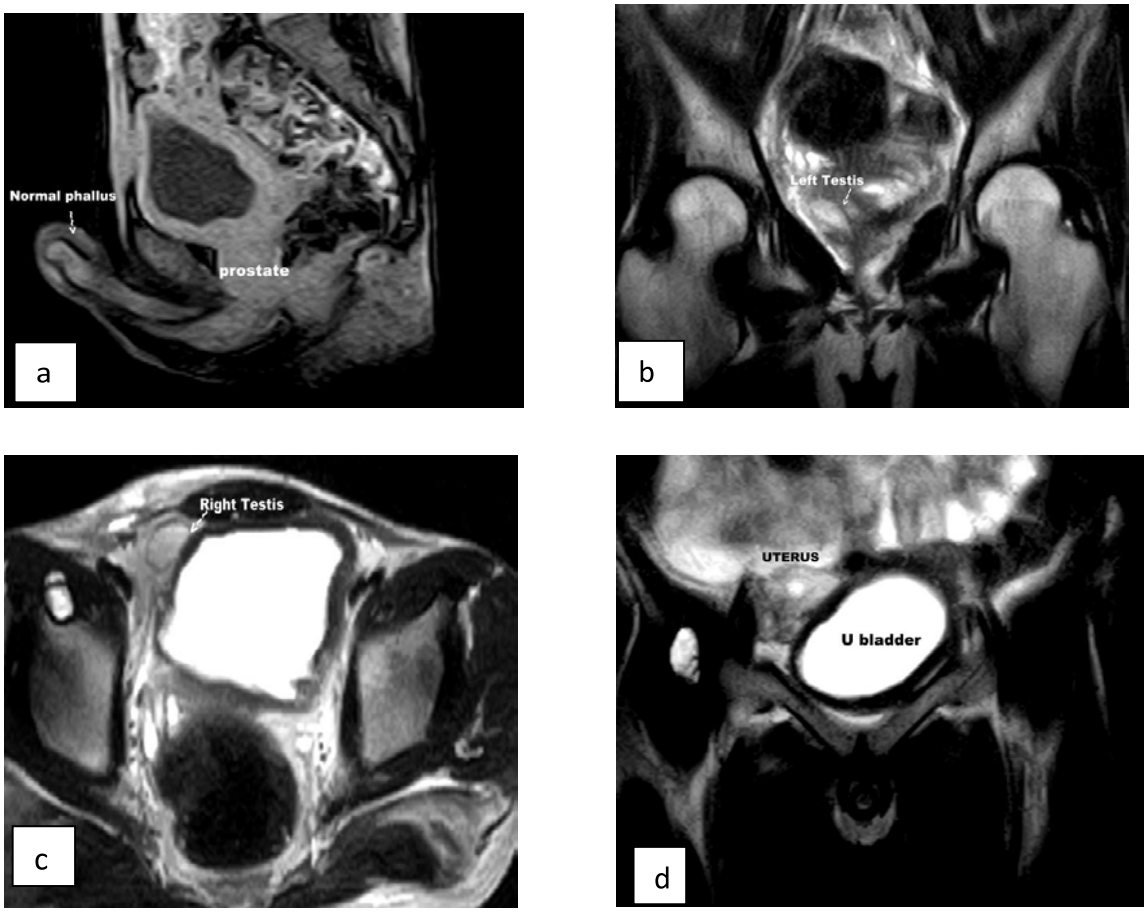

Fig 2 a) MRI Sag image showing prostate and phallus. b)Left testis near the right side of base of urinary bladder. c) MRI T2 WI axial image showing right-sided undescended testis located at the deep inguinal ring d) MRI T2 WI coronal image showing uterus above the urinary bladder.

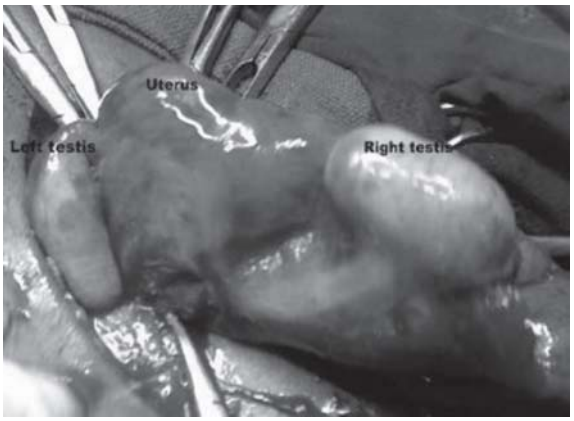

Fig 3 : Post op showing uterus and bilateral testis

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