Persistent Mullerian Duct Syndrome with Testicular Seminoma in Transverse Testicular Ectopia

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Persistent Mullerian Duct Syndrome (PMDS) is a disorder of male pseudohermaphroditism characterized by the persistence of Mullerian duct derivatives (uterus, fallopian tubes, and upper two-third of vagina) in a phenotypically and genotypically male. Transverse testicular ectopia (TTE) is a rare congenital anomaly in which both gonads migrate toward same hemiscrotum. About 150 cases of PMDS and 100 cases of TTE have been reported in previous studies. Testicular tumor in patients with PMDS with TTE is very rare. We report a case of testicular seminoma in a 35-year-old male patient with PMDS and TTE. Preoperative diagnosis was not possible in most of the reported cases.

Keywords: Persistent Mullerian Duct syndrome, seminoma, transverse testicular ectopia

INTRODUCTION

Persistent Mullerian Duct Syndrome (PMDS) is a rare type of pseudohermaphroditism in phenotypically and genotypically male. They have uterus, fallopian tubes, and cervix due to the failure of regression of Mullerian duct.[1] In a normal male and female structures derived from Wolffian duct and Mullerian duct, respectively, are present. Patients with PMDS have defect in secretion of anti-Mullerian hormone (AMH) or the AMH receptors resulting in persistence of Mullerian duct.[2] Transverse testicular ectopia (TTE) is a rare congenital anomaly in which both testes descend through same inguinal canal. About 20% of TTE cases are associated with PMDS.[3] We report a case of seminoma in a 35-year-old male patient in TTE with PMDS.

CASE REPORT

A 35-year-old male patient noticed lump in abdomen and complained of pain in abdomen for 1 year. The patient was married for 4 years with primary infertility. No significant past history was present. On clinical examination, he was averagely built and well-nourished with well-developed secondary sexual characters. Per abdominal examination revealed lump in the right inguinal region. On genitourinary examination, bilateral testis was absent in the hemiscrotum with well-developed penis. No hypospadias or epispadias was present. Karyotyping was not done in our case.

Inguinoscrotal ultrasonography revealed absence of bilateral testis in the scrotal sac. On computed tomography abdomen, there was presence of large mass in the right inguinal region with the presence of another small ovoid mass attached to its one end. The clinical and radiological diagnosis was right cryptorchidism with testicular tumor with the absence of another testis. Tumor markers were not done in our case.

We received a right orchidectomy specimen. On gross examination, testis was enlarged in size measuring 17 cm × 5.5 cm × 6 cm. Cut section of the mass showed large areas of necrosis with the presence of viable yellowish areas. On one side of spermatocord, there was seen a triangular structure with tube-like structure. Proximal end of spermatocord showed a small nodule measuring 1.5 cm × 0.8 cm [Figures 1 and 2]. Histopathological examination from the testicular tumor showed the features of seminoma.

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with large areas of coagulative necrosis with atrophic testis at the periphery [Figure 3]. Sections from triangular structure on one side of spermatic cord showed endomyometrium and cervix [Figures 4 and 5]. Sections from tube-like structure confirmed the presence of fallopian tube [Figure 6]. Small ovoid mass at proximal end of spermatic cord showed other atrophic testis [Figure 7]. To conclude, histopathological examination confirmed presence of both the testis on the right side with the presence of uterus and cervix (persistent Mullerian duct structures). Hence, the final diagnosis was testicular seminoma in TTE with PMDS.

**DISCUSSION**

PMDS was first described by Nilson.[4] In human fetus, both Wolffian and Mullerian ducts are present at 7th week of gestation. After 7th week of gestation, Sertoli cells secrete AMH which causes regression of Mullerian duct. In 85%
of cases, PMDS is either caused by deficiency of AMH secretion or by end-organ resistance to action of AMH due to mutations of gene of AMH-II receptor. In the remaining, 15% of cases cause is unknown (idiopathic PMDS).[5] In PMDS, the incidence of malignant transformation is 18% similar to incidence rate of malignant transformation in cryptorchidism in normal males.[6] Seminoma is the most frequently encountered tumor in patients with PMDS; there are also reports of embryonal carcinoma, yolk sac tumor, choriocarcinoma, and mixed germ cell tumor.[7] About 150 cases of PMDS described in the literature. Since patient of PMDS is phenotypically male, diagnosis of PMDS is usually not suspected before surgical exploration. Coexistence of PMDS with TTE is even rare.[8] PMDS must be distinguished from mixed gonadal dysgenesis in which the Mullerian structures are generally present, a testis is present unilaterally, and there is contralateral streak gonad.

TTE also called as crossed testicular ectopia is a congenital anomaly in which both the testis migrates through single inguinal canal. The first case was reported in the literature by Lenhossek in 1886.[8] Different theories have been proposed to explain the genesis of TTE. Berg proposed the possibility of development of both the testis from the same genital ridge.[9] Kimura stated that if both testis differentia arose from one side, there had been unilateral origin but if there was bilateral origin one testis had crossed over.[10]

TTE is classified into three clinical types according to the presence of additional abnormalities. Type 1 is the most common, accompanied by inguinal hernia. Type 2 is associated with persistent or rudimentary Mullerian duct structures. Type 3 is rare associated with disorders other than persistent Mullerian duct remnants (inguinal hernia, hypospadias, pseudohermaphrodite, and scrotal abnormality).[11] According to above-mentioned classification, our case was Type 2 TTE.

**CONCLUSION**

In a case of bilateral cryptorchidism associated with inguinal mass, possibility of TTE should be kept in mind. One should also look for persistent Mullerian duct structures as it may be associated with TTE.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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**Conflicts of interest**

There are no conflicts of interest.

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