Case Report

A case report of persistent Müllerian duct syndrome in a male patient with seminoma

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ABSTRACT

Persistent Müllerian duct syndrome (PMDS) is a genetic disorder characterized by the persistence of Müllerian duct derivatives, including the uterus, fallopian tubes, and upper two-thirds of the vagina in a phenotypically and genotypically male patient. We report the case of a 44-year-old male with PMDS and an abdominal lump. On examination, he had a uterus and left testis on one side and pure seminoma arising in the right testis, confirmed on histopathological examination.

Key words: Male, Persistent Müllerian duct syndrome, Seminoma, Testis, Uterus

Persistent Müllerian duct syndrome (PMDS) is a rare type of genetic disorder, where a phenotypically and genotypically male patient (46, XY) develops persistence of female reproductive organs [1]. The condition is inherited in an autosomal recessive manner and is characterized by mutations in genes linked to the hormone known as Müllerian inhibiting substance, which causes the male Müllerian ducts to regress. They have a uterus, fallopian tubes, cervix, and the upper 2/3rd of the vagina, due to the failure of regression of the Müllerian duct [2]. Persistence of these structures prevents the normal descent of the testis in the scrotum, resulting in an undescended testis on one side and an inguinal hernia on the other side. It is usually incidentally encountered during orchidopexy, laparotomy, or routine inguinal hernia repair in patients presenting with an undescended testis [3]. The risk factor for developing testicular tumors is cryptorchidism, and it has been found to be associated with PMDs [4].

CASE REPORT

A 44-year-old male presented with swelling on the right side of the scrotum for 2 months with on-and-off pain. The pain was dull, not intense, and not associated with fever, burning micturition, or vomiting. There was no history of consanguineous marriage in the family. He was married and had no children. On examination, the patient was phenotypically a normal male with a small scrotum and an empty left-sided scrotal sac since birth. He had a normal male distribution of hair. On general examination, no pallor/icterus/edema was noted. Respiratory, cardiovascular, and central nervous system examinations were within normal limits.

Computed tomography of the abdomen revealed a large heterogeneously enhancing mass lesion measuring 13 × 7 × 8 cm in size involving the right testis, epididymis, and right spermatic cord. The lesion showed right lower abdominal cavity extension. Multiple enlarged nodes were noted along the right paraaortic and periportal station, the largest measuring 1.7 × 1.6 cm in size. Blood investigation revealed a high serum lactate dehydrogenase level (783 U/L). Serum alpha-fetoprotein and beta-human chorionic gonadotropin levels were normal. After the approval of the multidisciplinary tumor board, the patient was prepared for right high inguinal orchidectomy.

Intraoperatively, a large right testicular mass was noted in the inguinal canal with rudimentary uterus and ovary-like structures identified as "Persistent Müllerian duct syndrome." The patient and his relatives were counseled about the condition. The surgical specimens were sent for histopathological examinations. On gross examination, a large testicular mass measuring 10 × 9 × 6 cm was noted attached to the right side of a small uterus, and on the left side, an atrophic testis was noted (Fig. 1a and b). However, no ovary was identified grossly.

On microscopy, the right testicular mass revealed a neoplasm arranged in sheets separated by thin fibrous

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septae composed of monomorphic cells with a moderate amount of clear cytoplasm, round vesicular nuclei, and prominent nucleoli. Large areas of necrosis were present (Fig. 2a).

Immunohistochemistry was done and revealed diffuse positive for CD117 and negative for CD30, confirming the diagnosis of seminoma right testis (Fig. 3a and b). Sections from the left testis showed atrophic seminiferous tubules with preserved Leidig cells. Sections from the spermatic cord revealed both the ductus deference and the fallopian tube's epithelium. Sections from the uterus showed endometrial glands and stroma (Fig. 2a-d). However, no ovarian tissue was seen in our case. Hence, histopathologically, the patient was diagnosed as a seminoma with PMDS.

DISCUSSION

PMDS was first described by Nilson in 1939 in a male with an inguinal hernia [5]. Normally, both Wolffian and Müllerian ducts are present in the fetus at the 7th week of gestation. After the 7th week of gestation, there is regression of the Müllerian ducts due to the secretion of anti-Müllerian hormone (AMH) by Sertoli cells. In 85% of cases, PMDS is either due to a lack of AMH secretion or end-organ resistance to AMH action as a result of AMH-II receptor gene alterations [6,7]. The remaining 15% of cases are idiopathic PMDS. The risk of malignant transformation is 18% similar to that of cryptorchidism in normal males [8]. Seminoma is the most commonly encountered tumor in PMDS patients; besides that, cases of embryonal carcinoma, yolk sac tumor, choriocarcinoma, and mixed germ cell tumor are also reported in association with PMDS [9]. The risk of malignant transformation increases after puberty; therefore, early diagnosis should be recommended. There are only a few cases (150) of PMDS described in the literature [10].

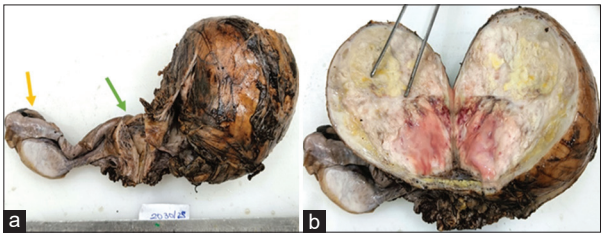

Figure 1: (a) Right large testicular mass, uterus (green arrow), left testis with spermatic cord (orange arrow), (b) cut surface of right testicular mass showing solid grayish white to yellowish area

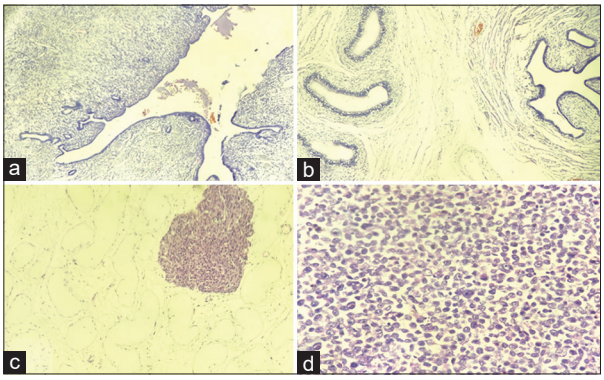

Figure 2: (a) Uterine stroma lined by endometrial glands, (b) ductus deference (left side) and fallopian tube (right side), (c) Atrophic left testis with preserved Leidig cells, (d) right testicular seminoma (hematoxylin and eosin, ×400)

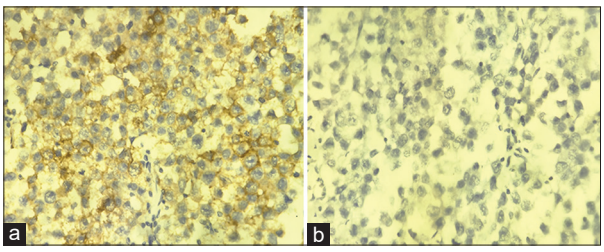

Figure 3: (a) Membranous positivity of CD117, (b) no immunoreactivity with CD30 (immunohistochemistry, ×400)

Since patients with PMDS are phenotypically male, the diagnosis is almost always incidental during surgical exploration. PMDS should be distinguished from mixed gonadal dysgenesis, where the Müllerian structures are generally present, unilaterally a testis and contralaterally a streak gonad is present. Most of the PMDS patients usually suffer from infertility and an inguinal hernia [11]. In our case, the patient was married but had no children. Infertility occurs due to testicular hypoplasia and ejaculatory duct obstruction due to compression by Müllerian duct structures.

The importance of identifying PMDS remains the potential for fertility and prevention of malignant transformation. Surgical management, such as orchiopexy, is available for preserving fertility and repositioning of the testis into the scrotum, herniorrhaphy with hysterectomy, and bilateral salpingectomy. Orchidectomy is indicated in cases where the testes cannot be mobilized or have undergone malignant transformation. The management of seminoma depends on the clinical stage and lymph nodes involvement: (a) Surgery for tumor removal, with or without radiation therapy to lymph nodes in the abdomen after the surgery and lifelong follow-up, or (b) tumor removal surgery, followed by chemotherapy and lifelong follow-up. In our case, the patient underwent surgical removal of testes with tumor and uterus, followed by platinum-based chemotherapy (bleomycin, etoposide, and platinum regimen – bleomycin, etoposide, and cisplatin) and follow-up. Currently, the patient is doing well and receiving the fourth cycle of chemotherapy.

CONCLUSION

The patients with PMDS are phenotypically and genotypically male without any clinical signs or symptoms. Hence, diagnosis is almost always incidental. Since the risk of malignant transformation is similar to that of cryptorchid testis, early detection and treatment are recommended. Müllerian structures should be removed, if possible, to avoid the risk of malignant transformation and long-term follow-up, if necessary.

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