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OLGU SUNUMU/CASE REPORT

Dev Abdominal Seminom ve Persistan Müllerian Kanal Sendromlu Olgunun Sunumu

A Case Report of the Presenting with Huge Abdominal Seminoma and Persistent Mullerian Duct Syndrome

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ÖZ

Persistan Müllerian Kanal Sendromu çok nadir görülen bir erkek psödohermofroditizm nedenidir. Bu hastalık genotipik ve fenotipik olarak normal olan erkek bireylerde görülen Müllerian kanal kalıntılarının (uterus, fallopian tüpleri ve vajenin üst 2/3'ü) varlığı ile karakterizedir. Bu hastalarda en sık görülen başvuru semptomları kasık fıtığı, inmemiş testis, transvers testiküler ektopi ve infertilitedir. Persistan Müllerian Kanal Sendromu hastalarında testis malignite oranı %15'e kadar yükselmektedir. 38 yaşında erkek hasta karın ağrısı ve karında şişlik şikayeti ile hastanemize başvurdu. Özgeçmişinde inmemiş testis nedeniyle on yaşında yapılan bilateral orşiektomi ve infertilite öyküsü vardı. Ameliyat sırasında kitleye yapışık uterus benzeri yapılar olduğu görüldü. Bu olguda geç tanı almış bir Persistan Müllerian Kanal Sendromu hastası ve ona eşlik eden seminom vakası anlatılmıştır.

ABSTRACT

Persistent Mullerian Duct Syndrome is a very rare form of male pseudo hermaphroditism. It is characterized by the presence of Mullerian duct derivatives (uterus, fallopian tubes and upper two-third of vagina) in genotypically and phenotypically normal males. The most common presenting symptoms are inguinal hernia, undescended testis and transverse testicular ectopia and infertility. The risk of malignancy in testes associated with Persistent Mullerian Duct Syndrome is up to 15%. We report a 38 year-old, phenothpically male patient, presenting with huge abdominal mass. He had underwent bilateral orchiectomy due to cryptoorchidism at 10 years old. At exploration, uterus like structure was observed adherent to mass. The case explained in this report is an example of delayed recognition of Persistent Mullerian Duct Syndrome and subsequent seminoma.

Introduction

Persistent Mullerian Duct Syndrome (PMDS) is characterized by the presence of uterus, fallopian tubes and upper two-third of vagina in genotypically and phenothpically normal males. Approximately 200 PMDS case reports are present in the literature (1). Most of the patients were diagnosed in childhood and adulthood. The most common presenting symptoms were inguinal hernia, undescended testis, transverse testicular ectopia and infertility (1-2). The case explained in this report is an example of delayed recognition of PMDS and subsequent seminoma.

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Case Report

A 38 year-old male patient presented to our clinic with complains of abdominal pain, constipation, urinary urgency for a few months. The patient was married for about ten years and was infertile. The patient had underwent orchiectomy bilaterally in childhood for cryptoorchidism.

Physical examination revealed a phenothpically male with masculine pattern of external genitalia. He had truncal obesity and low muscle proportion. A firm, tender mass, measuring 20 x 15 cm in diameter was palpable in the both lower quadrants of abdomen. There were bilateral orchiectomy insicion scars on the inguinal regions. No more remarkable findings were present at the physical examination. Complete blood count test revealed anemia, hemoglobin level was 10,3 g/dL. Blood biochemical tests, serum glucose, serum urea, creatinine, electrolytes, liver function tests, coagulation analysis were normal. Abdominal ultrasonography revealed an irregular shaped, heterogenous composition intraabdominal mass. The Abdominal Computed Tomography (CT) showed a huge mass, measuring 15 x 12 x 18 cm in diameter, begining from the pelvis, in the lower quadrants of the abdomen, at the midline localization. The mass was heterogenous in composition, lobulated, rich in vascular supply. It was displacing the intestine and compressing the urinary bladder. It was close relation to internal and external iliac vessels, and there were multiple enlarged lymph nodes in the aortocaval region. Radiologically it was similar to Gastrointestinal Stromal Tumor (Figure 1, 2).

He underwent an operation for the abdominal mass. At exploration, a huge mass deplacing the normal anatomical organs was present in the pelvis and lower

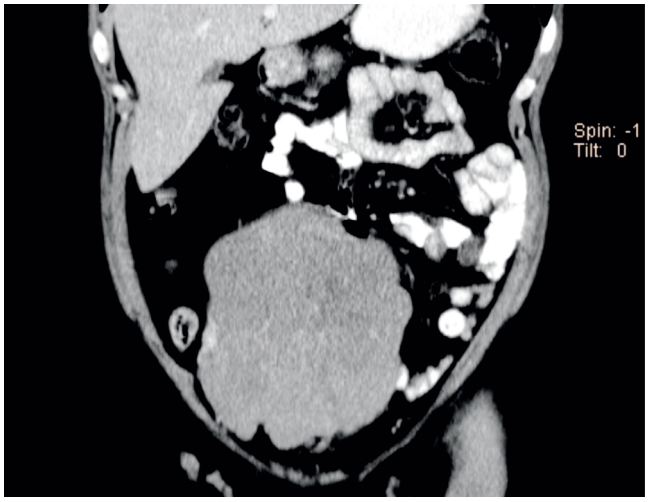

Figure 1. Computed Tomography findings of the abdominal mass, displacing the intestine and compressing the urinary bladder.

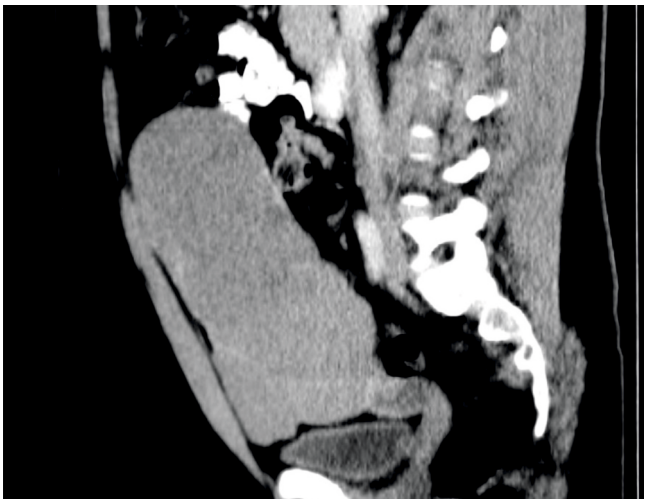

Figure 2. Computed Tomography findings of the abdominal mass, displacing the intestine and compressing the urinary bladder.

quadrants of the abdomen. Adherent to mass, uterus and fallopian tubes like structures were observed. The tumor was also adherent to seminal vesicule and prostate inferiorly. Adherent tissues were excised with the tumoral mass.

Macroscopic examination demonstrated a necrotic tumoral mass 22 x 17 x 13 cm in diameter. Adherent to the mass, the rudimentary uterus was 10 x 3 x 2 cm in size (Figure 3). Histological examination confirmed seminal vesicule, endoservix and endometrium at the same patient (Figure 4, 5). The tumor cells were composed of hyperchromatic nucleus, eosinophilic cytoplasm and has a fibrous stroma infiltrated by lymphocytes. The center of the mass was necrotic. Immunohystochemically it was positive for CD117 and D2 40, negative for CD30, PLAP, EMA, AFP and LCA. The pathological result was seminoma (Figure 6).

The patient was genotypically male, karyotype analysis was 46 XY.

Further ivestigation for seminoma metastasis, thorax CT revealed no metastases. The patient recieved Bleomycin, Etoposide and Cisplatin (BEP) chemotheraphy. It has beeen two years after the surgery, there is no local recurrence and distant metastases. Consent form was taken from the patient.

Discussion

The normal gonadal determination stage begins in the 7th embryonic week. In normal fetus, both Wolffian and Mullerian ducts are present at 7th week. In a normal female, genital structures are derived from Mullerian

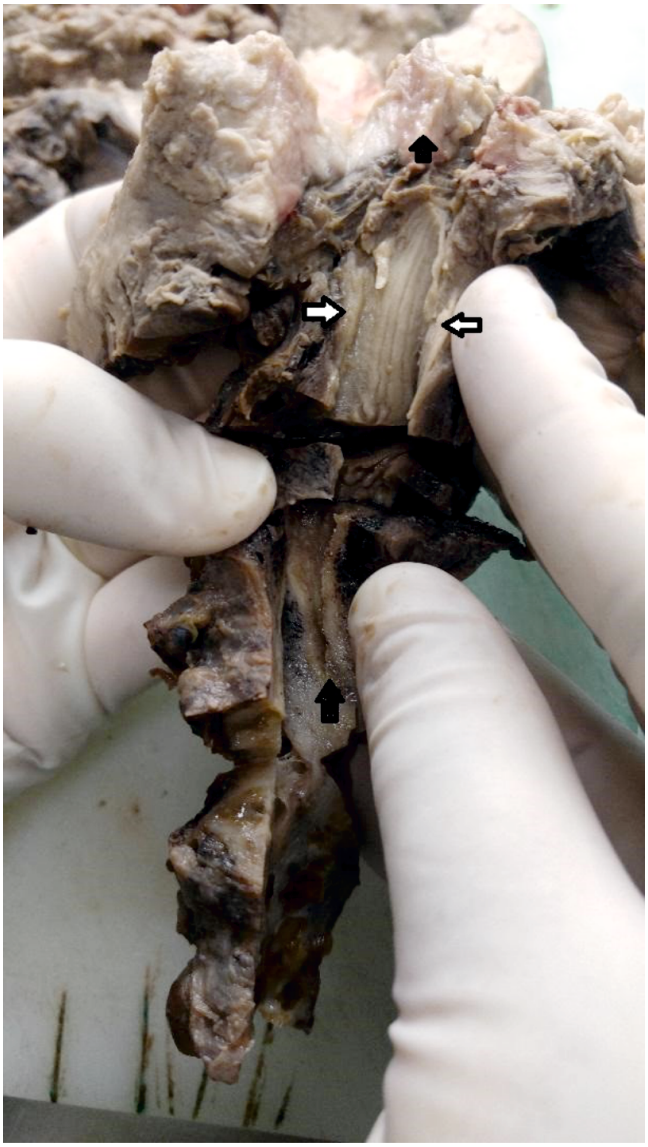

Figure 3. Macroscopic examination demonstrated a tumoral mass, adherent to the mass, the rudimentary uterus and cervix.

duct, on the other hand in male, Wolffian duct is the precursor. Antimullerian hormone (AMH) or Mullerian inhibiting substance (MIS) is produced by Sertoli cells, leads to the regression of Mullerian ducts (uterus, fallopian tubes and upper two-third of vagina) in the male fetus during the 8th and 9th embryonic weeks (3).

Persistent Mullerian Duct Syndrome was first described by Nilson in 1939 (4, 5). PMDS is a very rare form of male pseudohermaphroditism that is characterized by the presence of Mullerian duct derivatives in genotypically and phenothpically normal males (6-7). Deficiency of AMH secretion or end organ resistance to action of AMH due to mutations of gene of AMH-II receptor leads to PMDS. This syndrome is an autosomal or X linked recessive inherited disorder (6).

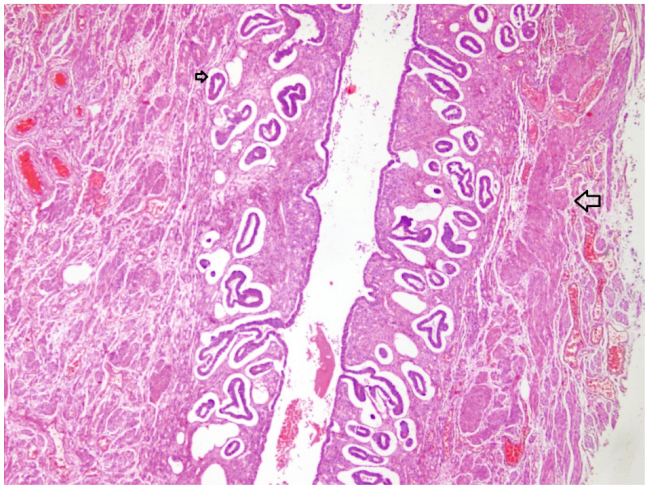

Figure 4. Histopathological findings showing Mullerian Duct residues, Endometrium.

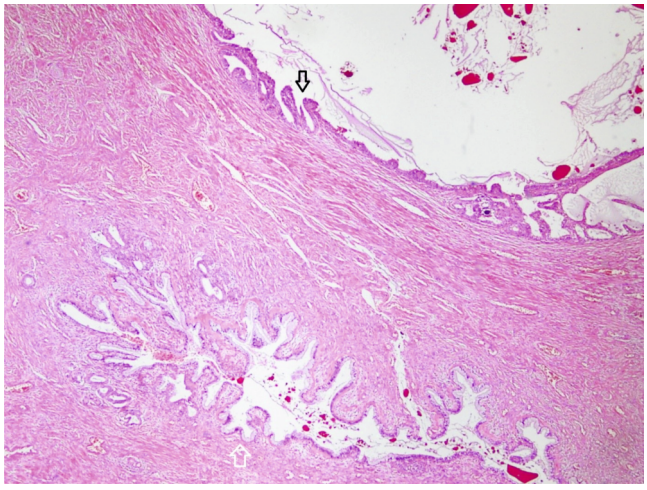

Figure 5. Arrows showing seminal vesicule, endocervix and endometrium.

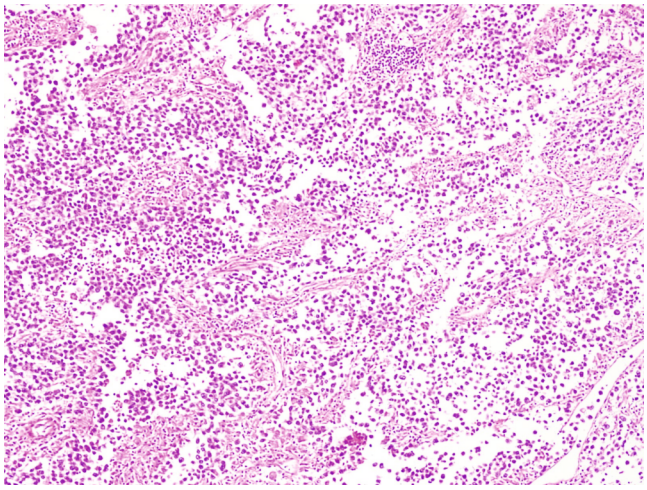

Figure 6. The histopathological findings of Seminoma (tumor cells with polygonal shaped large nuclei and prominent nucleoli, rich in fibrovascular stroma and lymphocytes).

PMDS has the malignancy risk associated with undescended testes. The risk of malignant transformation of the gonadal tissue for PMDS is 15%, similar to the rate of malignancy in abdominal testes in a man without PMDS (8). A variety of germ cell tumors can develop. Seminoma is the most common testicular germ cell tumor, others are embryonal carcinoma, yolk sac tumor, gonadoblastoma and teratoma (9).

The surgical treatment of PMDS is still controversial (6). Arising from the Mullerian remnants, the literature uncovered 11 patients developing malignancy (1). Adenocarcinoma, squamous cell carcinoma, papillary cystadenocarcinoma and adenosarcoma are presented in literature. One group of surgeons consider excision of Mullerian remnant structures when possible. However, it has the risk of damaging blood supply of vas deferens during surgical removal of Mullerian structures. Other group of surgeons choose more conservative approach and recommend only orchidopexy without Mullerian remnants structure removal (10).

In our case, it is important to point out the history of the patient. He had bilateral orchiectomy in childhood for cryptoorchidism. He had neglected PMDS diagnosis in childhood. Meanwhile, he had delayed recognition of PMDS and subsequent seminoma. We recommend to remember the rare cases of PMDS for infertile patients with the inguinal hernia, undescended testis and transverse testicular ectopia.

Yazarlık katkısı: Fikir/Hipotez: NT, Tasarım: NT Veri toplama/Veri işleme: NT, AU, AC, EK Makalenin hazırlanması: NT, AU, AC, EK.

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