Bilateral Seminoma Arising in Transverse Testicular Ectopia with Persistent Müllerian Duct Syndrome and its Metastasis to Rudimentary Uterus: Case Report

Persistan Müller Kanalı Sendromu ile Transvers Testiküler Ektopi Birlikteliğinde Bilateral Seminomun Rudimenter Uterusa Metastazı

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Geliş Tarihi/*Received:* 13.05.2009
Kabul Tarihi/*Accepted:* 30.01.2010

This case report was presented as poster in 18th National Pathology Congress (25-29 December 2008, Belek, Antalya, Turkey).

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ABSTRACT We report a case of a 45-year-old man with transverse testicular ectopia and persistent Müllerian duct syndrome who had bilateral germ cell tumor which showed metastasis to the rudimentary uterus. Transverse testicular ectopia in which both testes are located in one inguinal canal is a very rare condition. Persistent Müllerian duct syndrome is also a very rare form of male pseudohermaphroditism in which the Müllerian duct structures are present in genotypically normal males. Persistent Müllerian duct syndrome associated with transverse testicular ectopia is much rarer. Malignant transformation of the Müllerian ducts is even more uncommon. A variety of germ cell tumors of the testis have been reported in association with persistent Müllerian duct syndrome. However, no cases with metastasis to persistent Müllerian duct structures have been reported up to date. This represents the first reported case in which the testicular tumor metastasizes to the Müllerian duct and also the first case of bilateral testicular malignancy in transverse testicular ectopia with persistent Müllerian duct syndrome.

Key Words: Pseudohermaphroditism; seminoma; neoplasm metastasis; cryptorchidism; mullerian ducts

ÖZET Testiküler ektopi ve persistan Müller kanalı sendromu olan 45 yaşında erkek hastada saptanan rudimenter uterusa metastaz yapmış bilateral germ hücreli tümör olgusunu sunuyoruz. Her iki testisin tek inguinal kanalda yer aldığı transvers testiküler ektopi çok nadir rastlanan bir durumdur. Genetik olarak normal erkeklerde Müller kanalı elemanlarının bulunduğu persistan Müller kanalı sendromu da erkek psödohermafroditizminin çok nadir bir formudur. Transvers testiküler ektopi ile birlikte persistan Müller kanalı sendromuna ise daha da nadir olarak rastlanır. Müller kanallarının malign transformasyonu nadiren görülür. Persistan Müller kanalı sendromu ile çeşitli germ hücreli tümörler birlikte saptanmış, ancak persiste eden Müller kanalı yapılarına metastaz bugüne kadar bildirilmemiştir. Olgumuz persistan Müller kanalı sendromu ile transvers testiküler ektopi birlikteliğinde Müller kanalı yapısı olan rudimenter uterusa metastaz yapan ilk bilateral testis malignitesidir.

Anahtar Kelimeler: Psödohermafroditizm; seminom; tümör metastazı; kriptorşidizm; müllerian kanallar

Turkiye Klinikleri J Med Sci 2011;31(4):1015-8

Transverse testicular ectopia (TTE) is a very rare entity in which both testes descend through the same inguinal canal towards the same scrotal sac.[1,2] Persistent Müllerian duct syndrome (PMDS) is a rare form of male pseudohermaphroditism in which the Müllerian duct derivatives (uterus, fallopian tubes and vagina) are present in a male phenotype. PMDS is rarely associated with TTE. Malignant transformation of the Mül-

doi:10.5336/medsci.2009-13458

lerian structures is a very rare event; only three cases have been reported to date.[3-5] However no metastasis to the Müllerian duct structures has been reported yet. Furthermore, although some articles document testicular malignancy associated with PMDS and TTE, none of these cases had bilateral testicular malignancy.

This represents the first reported case in which the testicular tumor metastasizes to rudimentary uterus of the Müllerian duct and also the first case of bilateral testicular malignancy in TTE with persistent Müllerian duct syndrome.

CASE REPORT

A 45-year-old man presented with inguinal pain and right scrotal mass, which were present approximately for three and seven months respectively. His past medical history revealed right inguinal hernia repair 30 years ago. Physical examination showed a palpable mass in the right inguinal canal and scrotum which was assumed to be an ipsilateral irreductable hernia. Left testis was not palpable. The secondary sexual characteristics and external genitalia were normal. He was married and reported a normal sex life, but he had never fathered a child. Abdominal ultrasonography (USG) was unremarkable except for gas filled intestinal lumens in the hernia sac.

He underwent an operation for right inguinal hernia and bilateral cryptorchidic testis. At exploration, a leiomyomatosis uterus including blind-ending vagina-like structure and gonads on the both sides of the uterus were observed in the hernia sac. All these tissues were excised en bloc (Figure 1). Additionally, a rudimentary uterus which was initially in continuity with the mass was discovered, and excised from the hernia sac.

Macroscopic examination demonstrated that the mass was composed of a huge right testicular tumor and left ectopic testicular tumor, which were of 16 x 9 x 6 cm and 7 x 7 x 3 cm diameters respectively. These structures were connected to each other via a spermatic cord-like structure (Figure 1). The rudimentary uterus was 8 x 4 x 2 cm in size and a white, well circumscribed nodular area of 1.5 cm in size was seen on the myometrium.

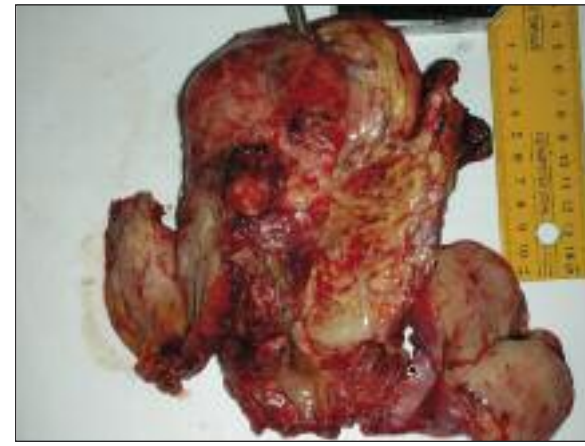


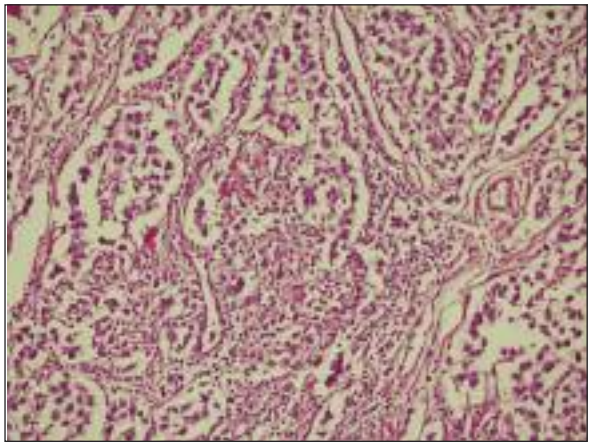
FIGURE 1: A huge right testicular tumor resembling a leiomyomatous uterus has a blind ending vagina-like structure on one side (arrow) and the left ectopic testicular tumor is bound to the huge mass with a spermatic cord like structure on the other side (arrow head).

Histological examination confirmed a testicular seminoma confined to both testes (Figure 2). Vagina-like pouch was lined by epithelium, which mostly represents Müllerian duct remnants (Figure 3). The rudimentary uterus contained only endocervical glands and the aforementioned nodular lesion on the myometrium was diagnosed as seminoma metastasis (Figure 4). The vasa deferentia and uterine tubes were also observed on one side of the uterus.

Further investigation including alpha fetoprotein (AFP) and beta human chorionic gonadotropin (βHCG) were within normal limits. Thorax computed tomography (CT) imaging was normal findings but abdominal CT showed enlarged paraaortic lymph nodes. The patient received three cycles of chemotherapy with bleomycin, etoposide and cisplatin. The patient has been tumor-free for 16 months after the initial treatment.

DISCUSSION

TTE is a very rare condition in which both testes are located in one inguinal canal.[2] PMDS is also a rare condition in which Müllerian derivatives are present in a male with a normal male phenotype.[1,3] The syndrome is an autosomal or X-linked recessive disorder caused by insufficient Müllerian duct inhibiting factor or unresponsiveness of the end organ.[1,6]



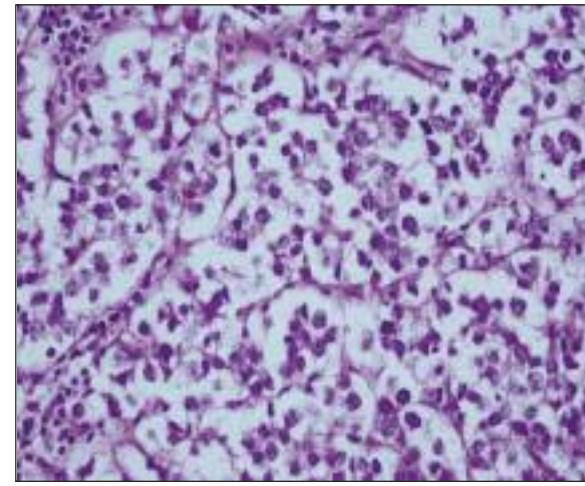


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FIGURE 2a, b: Classical seminoma with uniform tumoral cells and fibrovascular septa infiltrated by lymphocytes (a- H&E stain, x110; b- H&E stain, x440).

Unilateral cryptorchidism with a contralateral hernia (male form) is the most common encountered variant with a percentage of 80-90% among the two anatomic variants of PMDS.[2,3,7] Commonly, one testis is palpated in the scrotum with herniation of the ipsilateral uterus and Fallopian tubes into the inguinal canal; a condition known as hernia uteri inguinale. Although very rare, in some instances both testes and the Müllerian structures herniate towards the same hemiscrotum producing TTE.[2,7]

In the second anatomic variant (female form) bilateral testes are non-palpable and embedded in the round ligaments with the uterus fixed in the pelvis.[2,7] PMDS commonly presents during child-

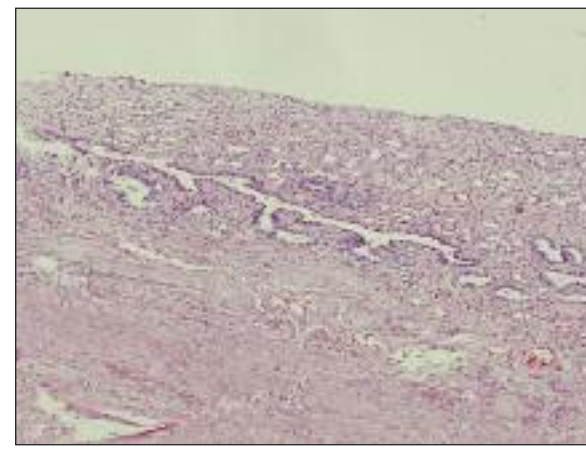
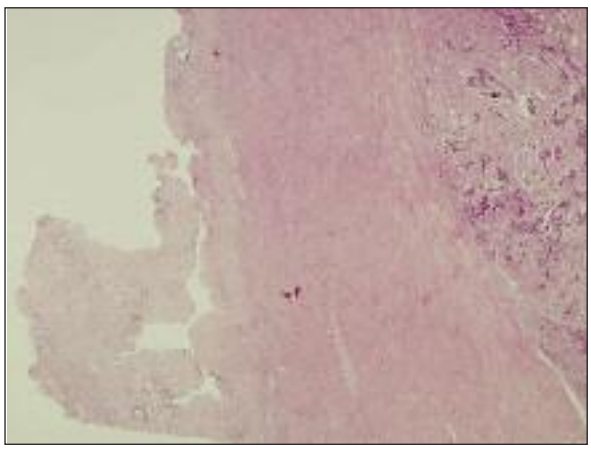


FIGURE 3: Vagina-like pouch is composed of muscle layer that is lined by epithelium which mostly represents "Müllerian" duct remnants (H&E stain, x40).



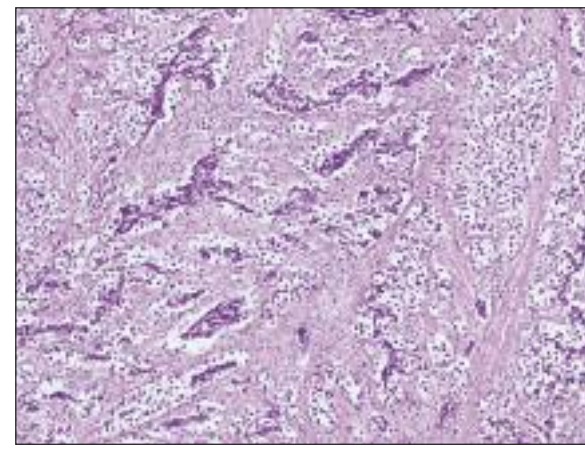


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FIGURE 4a, b: Seminoma metastasis in the myometrial wall of the rudimentary uterus containing only endocervical glands (a- H&E stain, x20; b- H&E stain, x220).

hood with inguinal hernias containing Müllerian structures (hernia uteri inguinale), and associated cryptorchidism.[6,8] Since secondary sex characteristics are normal, the syndrome is usually discovered in adults during hernioplasty or assessment of infertility.[8] Most of these patients are infertile.[6] Presentation with gonadal tumors is rare.[8] The risk of malignant transformation in these gonads is reported to be 15% which is similar to the rate of malignant transformation in other cryptorchid testes.[1,3,8] A variety of germ cell tumors have been reported in patients who have PMDS, seminoma being the most common one.[1,8,9] Bilateral tumors are rare; only six bilateral germ cell tumors have been documented.[9] However, we were not able to find any published case of bilateral testicular malignancy in TTE with PMDS. Thus, this is the first case with metastasis of a testicular neoplasm to one of the Müllerian duct structures.

Diagnosis of PMDS is based on combination of all anatomical, pathological and clinical findings. Ideally it is complemented by obtaining the karyotype.[10] Chromosomal analysis could not be performed in our case due to the technical limitations in our instutition.

The surgical treatment of TTE with PMDS is still controversial.[2] It is recommended to carry out orchidopexy in early stage of life if possible, or removal through standard methods if there is high risk of malignancy.[2,6,7] Main considerations in the management of Müllerian structures are the possibility of fertility and the risk of malignant change.[7] Since the blood supplies of vas deferens show close relation with the Müllerian structures, there is a high risk of damage during surgical removal of Müllerian structures.[7,8] Before2002, there had been a tendency towards leaving these structures in situ because no malignant change was reported before.[3,8] However, by demonstrating metastasis of the testicular tumor to the Müllerian duct structure, we recommend to reconsider treatment policy regarding leaving these structures in situ.

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