

Management of a Rare Case: Transverse Testicular Ectopia Associated With Persistent Mullerian Duct Syndrome

Nadir Bir Olgunun Yönetimi: Kalıcı Mülleryen Kanal Sendromu ile İlişkili Transvers Testiküler Ektopi

© Mehmet Semih Demirtaş¹, © Mustafa Tuşat²

¹Aksaray Training and Research Hospital, Clinic of Pediatrics, Aksaray, Turkey

²Aksaray University Faculty of Medicine, Department of Pediatric Surgery, Aksaray, Turkey

Abstract

Transvers testiküler ektopi (TTE), her iki testisin de aynı hemiskrotuma veya inguinal bölgeye migrasyonu ile karakterize erkek çocuklarda nadir görülen bir konjenital anomalidir. TTE olgularına, mülleryen kanal yapılarının varlığı ile birlikte fenotipik olarak erkek olan, karakterize kalıcı mülleryen kanal sendromu eşlik eder. Persistan Mülleryen kanal sendromu, anti-mülleryen hormonun veya reseptörünün genetik kusurundan kaynaklanır. Bu olguda, sağ kasıkta şişlik ve palpe edilemeyen sol testis ile başvuran 12 yaşında bir erkek çocukta persistent mülleryen duktus sendromu ile ilişkili bir transvers testis ektopisi bildirdik. Hasta fıtık kesesinin yüksek ligasyonu, mülleryen yapıların eksizyonu ve transeptal orşiopeksi ile tedavi edildi. Olguların cerrahi yaklaşımında, testislere ve diğer komşu yapılara zarar vermeden, mülleryen yapıların mümkün olduğunca çıkarılması ve testislerin palpe edilebilmesi gerektiğini düşünüyoruz.

Anahtar Kelimeler: Transvers Testiküler Ektopi, Persistent Mülleryen Kanal Sendromu, İnguinal Herni, İnmemiş Testis, Transeptal Orşiopeksi

Öz

Transverse testicular ectopia (TTE) is a rare congenital anomaly in boys, which is characterized by the migrate of both testicles towards the same hemiscrotum or inguinal region. TTE cases are accompanied by the characterized persistent mullerian duct syndrome, which is phenotypically male, with the presence of mullerian duct structures. Persistent Mülleryen duct syndrome is caused either by the genetic defect of the anti-mullerian hormone or its receptor. In this case, we reported a TTE associated with persistent mullerian duct syndrome in a 12-year-old boy who presented with swelling in the right groin and non-palpable left testis. The patient was treated with high ligation of the hernia sac, excision of mullerian structures and transeptal orchiopexy. In the surgical approach of the cases, we think that removing the mullerian structures as much as possible and making the testicles palpable without damaging the testicles and other adjacent structures are important.

Key Words: Transverse Testicular Ectopia, Persistent Mullerian Duct Syndrome, Inguinal Hernia, Undescended Testis, Transeptal Orchiopexy

Introduction

Transverse testicular ectopia is a rare congenital anomaly in which both the testis migrates ipsilaterally towards the same hemiscrotum. As a result, both testicles can be found in the deep inguinal canal or hemiscrotum on the same side (1).

Persistent mullerian duct syndrome is a rare type of hermaphroditism characterized by the presence of mullerian structures such as the uterus, tuba uterina and the upper third of the vagina in a boy with a completely normal appearance. Transverse testicular ectopia (TTE) and persistent mülleryen duct syndrome (PMDS) coexist very rarely and 57 cases have been

Address for Correspondence/Yazışma Adresi: Mehmet Semih Demirtaş
Aksaray Training and Research Hospital, Clinic of Pediatrics, Aksaray, Turkey
Phone: +90 382 520 10 20 E-mail: md.semihdemirtas@gmail.com ORCID ID: orcid.org/0000-0003-2965-1811
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reported so far (2). We report a case of TTE associated with PMDS applied to clinic with swelling in the right groin and left non-palpable testis.

Case Report

A 12-year-old foreign male patient was referred from the pediatric outpatient clinic to the pediatric surgery department due to the absence of left testicle and swelling in the right groin for the last 6 months. Swelling increased when coughing or carrying heavy objects. On examination, the patient had normal secondary sexual characteristics with a male pattern of external genitalia. There was the silk glove sign in right inguinal region. The left side of the scrotum was undeveloped. Left testis was not palpable and two 2x2.5 cm masses suspected of having testicles in the right scrotum of the patient were palpated. Afterwards, it was confirmed by ultrasonography (USG) that both testicles were in the right scrotum. One of them was in high scrotal location and our pre-diagnosis was TTE. In USG imagination revealed a right inguinal hernia and the testis at the base of the right hemiscrotum was measured 29x22 mm in size while the high-placed testis was measured as 29x26 mm in the right hemiscrotum. The vas deferens and vascular structures belonging to each testis were observed. The parenchyma of both testicles was homogeneous. No structure was seen in the left scrotum. There was a suspicious cystic structure behind the bladder. No other urologic anomalies were identified. Magnetic resonance imaging also revealed same findings. Moreover, the uterus-like cystic structure was observed on the right side of the bladder with a size of 30x15 mm and its extension was observed in the right inguinal canal (Figure 1). Serum follicle stimulating hormone, luteinizing hormone, prolactin testosterone and dihydrotestosterone (DHT) levels were normal while anti-Müllerian hormone (AMH)>24.20 ng/mL (0.73-16.05 ng/mL) was found. Karyotype analysis of the patient was reported as 46, XY. Detailed information about the current diagnosis and treatment was given to the family of the patient. Signature was obtained for the informed consent form from the family. The patient was

operated with the pre-diagnosis of the right inguinal hernia and TTE associated with PMDS. Both gonads, their cord and vascular structures, and rudimentary structures of the müllerian ducts delivered from the incisional wound after right inguinal exploration was done. Both gonads macroscopically were testes and their dimensions (3x2.5 cm) were similar. Each testicle had a normal appearance with the epididymis, vas deferens and testicular vessels separated. Both spermatic cords in close relation with a rudimentary uterus and bilateral fallopian tube were seen to enter the abdomen through the right deep inguinal ring. An rudimentary uterus and bilateral fallopian tube with fimbria were seen between both testicles (Figure 2). Uterus and fallopian tubes were carefully dissected from the testicles, vas deferens and vascular structures (Figure 2). Later, the hernial sac was separated carefully and a inguinal hernia repair was performed. All müllerian structures was removed completely and sent for histopathology (Figure 2). Per-operative biopsies were taken from each testis. Testicular biopsies were reported as testicular tissue, while rudimentary structure was reported as uterus with inactive endometrium with bilateral fallopian tubes and cervix. Left testicle was brought down to the left hemiscrotum transeptally via the right inguinoscrotal canal. Then, the right testis was placed and fixed on the pouch of the right hemiscrotum scrotum, and transeptal orchiopexy was completed (Figure 3). The patient, who had no complications in the postoperative period, was called to the outpatient clinic and discharged. In the examination of the patient who came to the outpatient clinic, no pathological examination finding was found, and abdominal and pelvic USG imaging was also found to be normal.

Discussion

TTE with unknown etiology has been classified into three types: Type 1 associated with only inguinal hernia (40-50%), type 2 accompanied by PMDS (20-30%), and type 3 associated with other anomalies such as hypospadias, pseudohermaphroditism and scrotal abnormality (1-3). The ectopic testis may be located

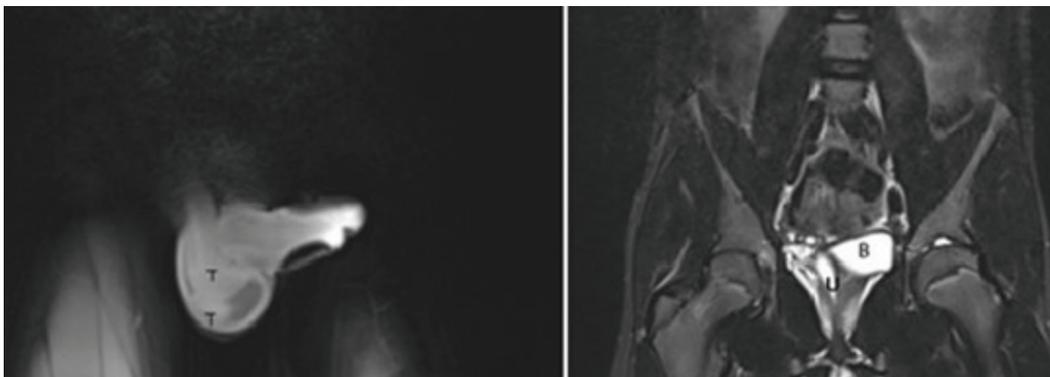


Figure 1: MRI of the pelvis and abdomen of patient
MRI: Magnetic resonance imaging

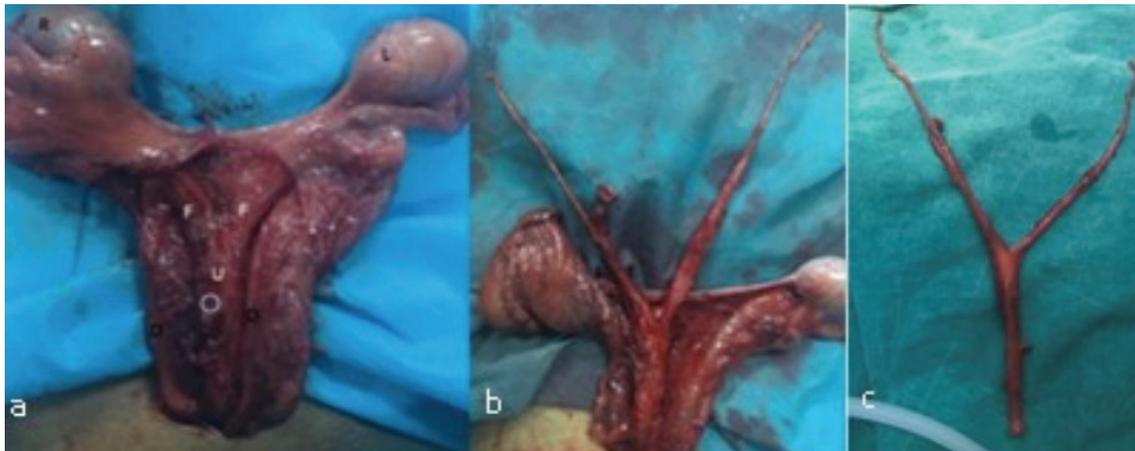


Figure 2: Intraoperative view of the mullerian remnants and testes, dissection of rudimentary duct structures, excised specimen



Figure 3: Stages of transseptal orchiopexy

in the abdomen, at the level of the internal inguinal ring or in the same hemiscrotum. TTE is often accompanied by ipsilateral inguinal hernia and contralateral non-palpable testicle (1,3,4).

Sexual differentiation is controlled by testosterone and AMH. PMDS mainly due to two causes: Failure of AMH secretion, end organ resistance to AMH. Since serum testosterone and DHT remain within normal range, patients with PMDS are phenotypically and genotypically males with normal secondary sexual characteristics (5). In TTE cases accompanied by PMDS, the diagnosis is usually made by seeing the rudimentary uterus and fallopian tubes during the operation of undescended testis or inguinal hernia (6). Although ultrasonography is helpful for diagnosis, it is not a good imaging method in intraabdominally located testicles. MR is the best non-invasive method for detecting intraabdominal testes and mullerian residues (3,6,7).

The mixed gonadal dysgenesis (MGD) is the most important disease to consider in the differential diagnosis of PMDS. Since AMH and testosterone secretion are also impaired in MGD, there are both mullerian canal residues and disorders in

external genitalia. In addition, patients with MGD generally have karyotype of 46 XO or 46 XY (8). In our case, the result of karyotype analysis was reported as 46 XY. Testosterone, follicle stimulating hormone, luteinising hormone and prolactin levels measured to support differential diagnosis were normal.

The best surgical approach in TTE cases with PMDS is excision of mullerian rudiments, orchiopexy with inguinal hernia repair, or orchidectomy when necessary and the operation should include the biopsy of both testicles (1,6). Transseptal orchiopexy technique is generally used in TTE surgical treatment. In this technique, the testis with a longer spermatic cord is placed in the subdartos pouch on the opposite side, while the other testis is placed on the same side. In bilateral orchiopexy, each testis is passed through a separate inguinal incision through a separate inguinal canal. In order to perform this technique, testicular vessels and vas deferens should be of sufficient length (2,6).

Some authors do not recommend removing the mullerian ducts because of the risk of damage to the vas deferens and vascular structures and the low rate of malignancy from

mullerian residues. However, recent studies have revealed that the left mullerian remains have a malignant potential (2,6,9). Therefore, we removed all the mullerian structures in our case. In addition, many testicular tumors such as seminoma, choriocarcinoma, embryonal carcinoma and teratoma have been reported in PMDS patients with undescended testicles (2,10). The postoperative follow-up period of the patient was specified and It was decided to follow the patient at regular intervals of 6 months. Although virilization is usually normal in patients with PMDS, almost all patients are infertile (10).

As a conclusion, in cases of TTE with PMDS, The case management requires that the condition is recognized by the surgeon and verified by biochemical testing, radiological imagination, testicular biopsies and chromosomal studies. In the surgical approach of the cases, we think that removing the mullerian structures and their attachments as much as possible and making the testicles palpable without damaging the testicles and other adjacent structures.

Ethics

Informed Consent: Signature was obtained for the informed consent form from the family.

Peer-reviewed: Externally peer-reviewed.

Authorship Contributions

Surgical and Medical Practices: M.T., Concept: M.S.D., Design: M.S.D., Data Collection or Processing: M.S.D., Analysis or Interpretation: M.S.D., M.T., Literature Search: M.T., Writing: M.S.D., M.T.

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