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Transvers Testiküler Ektopia ve Aort Koarktasyonu Birlikteliği: Yeni Bir Antite mi?

Coexistence of Transverse Testicular Ectopia and Coarctation of Aorta: A New Entity or Just a Co-incidence?

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ÖZ

Giriş: Transvers testiküler ektopi (TTE), her iki testisin aynı skrotal kompartmana taşınmasından kaynaklanan nadir bir patolojidir. Nadir de olsa TTE ile bazı kardiyak anomaliler bildirilmiş ancak daha önce TTE ile birlikte aort koarktasyonu (CoA) bildirilmemiştir.

Olgu Sunumu: Burada sol inguinal bölgede şişlik, solda inmemiş testis ve sağ impalpabl testis nedeniyle başvuran 6 aylık TTE hastası sunulmaktadır. Olgunun prenatal tanı ve klinik bulgularla kardiyak genişlemesi olduğu tespit edlip, hemen tedavi edilen ciddi CoA mevcuttu. Ultrasonda, sol inguinal bölgede iki testis varlığı gösterildi. Olguya laparoskopik Trans-septal orşiopeksi prosedürü uygulandı. Hastamızda sağ testis ektopik yerleşime sahipken inmemiş testis, fıtık ve ektopik testis sol taraftaydı.

Sonuç: Nadir olmasına rağmen, TTE kriptorşidizmin ayırıcı tanısında düşünülmeli ve diğer konjenital anormallikler ile birlikteliği akla gelmelidir.

Anahtar Kelimeler: transvers testiküler ektopi, inguinal herni, aort koarktasyonu, transseptal orşiopeksi

ABSTRACT

Objective: Transverse testicular ectopia (TTE) is a rare pathology resulting from the migration of both testicles into the same scrotal compartment. Although rare some cardiac anomalies had been reported with TTE but coexistence of TTE and coarctation of aorta (CoA) has not been reported before.

Case Report: Here we describe a 6-month old patient with TTE who presented with swelling in the left inguinal region, left undescended testicle, and impalpable testicle on the right side. The prenatal diagnosis of cardiac enlargement and clinical findings let us diagnose severe CoA that had to be managed immediately. The ultrasound examination showed the presence of two testicles in the left inguinal area and a laparoscopic procedure, trans-septal orchiopexy, was applied. In our patient, the right testicle had an ectopic location, while undescended testis, hernia and ectopic testis were on the left side.

Conclusion: Although rare, TTE should be considered in differential diagnosis of cryptorchidism and it might be present with other congenital abnormalities.

Keywords: transverse testicular ectopia, inguinal hernia, coarctation of aorta, transeptal orchiopexy

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INTRODUCTION

An ectopic testicle is defined as 'a testicle' that localizes at any other site than the scrotum after changing its direction following its passage through the inguinal canal. The least frequent type of ectopic testicle is the transverse testicular ectopia (TTE), in which both testicles are localized in a single inguinal canal (1). The diagnosis is often made incidentally during surgery.

Persistent Mullerian Duct Syndrome (PMDS) describes preserved Müllerian structures (uterus, fallopian tubes, the upper two-thirds of vagina) in a karyotypically normal male. PMDS is an inherited disorder caused by genetic mutations of either anti-Mullerian hormone (AMH) or its receptor. Herein, we report an infant with preoperative diagnosis of TTE without PMDS and severe CoA. The patient had 46, XY karyotype and no renal abnormalities. Genetic analysis revealed a recessive mutation in the AMH-receptor gene. To the best of our knowledge, this combination has not been reported before.

CASE REPORT

The patient was a 6-month-old infant admitted for non-palpable testicle on the right side and inguinal herniation on the left. He was the third child of non-consanguineous parents. The first child was a six years old healthy girl, the second child was a boy who died on the 11th day of life due to neonatal sepsis. In the last trimester of this pregnancy, unilateral cardiac enlargement and bradycardia were detected. After birth, the patient had shortness of breath during feedings. Due to this prenatal history and weak femoral pulses on physical examination, a consultation from the department of pediatric cardiology was requested. Upon detection of aortic coarctation by echocardiography, simultaneous angiography and dilatation were performed (Figure 1).

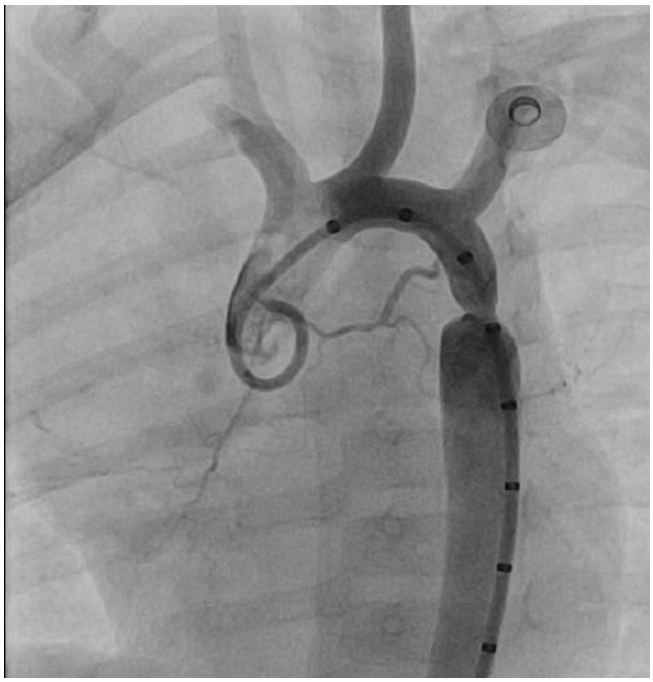

Figure 1: Angio image of aortic coartation

Scrotal ultrasound (US) performed after physical examination revealed that both testicles were in the middle and proximal segment of the left inguinal canal, with homogenous parenchyma. The sizes of the testicles were 7x12 mm and 6x12 mm. No testicular tissue was present in the right inguinal canal and right scrotum. Laboratory and hematological findings were normal. Karyotype analysis determined a 46, XY structure. Anti-Müllerian hormone (AMH) level was 70 ng/ml (range: 39.1-91.1). In addition, blood samples were obtained for AMH receptor mutation analysis. An elective surgery was scheduled. External genital examination revealed apalpable testicle on the left inguinal area and herniation, while right hemiscrotum was unoccupied by any testicular tissue (Figure 2).

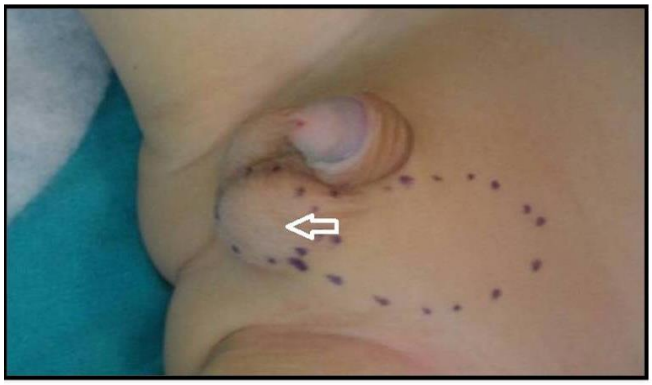

Figure 2. Right scrotum empty, both testes in the left hemiscrotum.

During laparoscopy, two testicles entering the left internal inguinal orifice were identified. Both testicles were clearly visible protruding from the left inguinal incision, establishing a diagnosis of TTE. (Figure 3a and 3b).

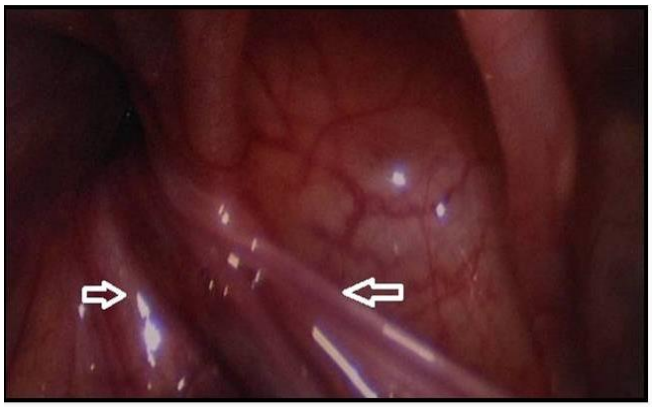

Figure 3a. Intraoperative appearance; transverse testicular ectopia and open left side inguinal hernia

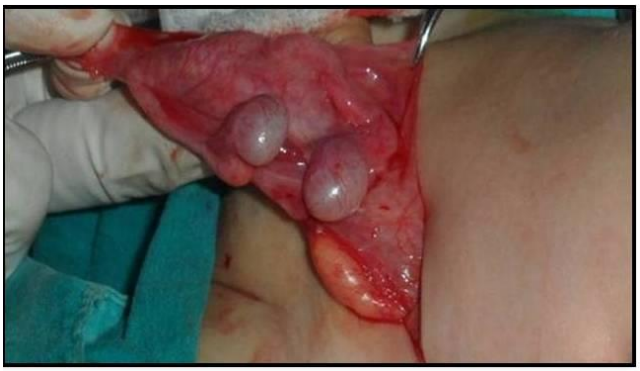

Figure 3b. Intraoperative appearance; left and right vas deferences in the left internal inguinal orifice.

A structure reminiscent of a rudimentary uterus observed between the testicles was excised. Two small scrotal incisions were made and a Kelly clamp was passed to the abdominal cavity through the right hemiscrotal incision in order to bring down the right testicle. This was guided by laparoscopy. On the left side, a herniorrhaphy was carried out. Both testicles were fixed to the scrotum. Histopathological examination of the cross-sections prepared from the surgical specimen did not reveal any uterine epithelium or myometrium. The mutation analysis was executed in INSERM, France and it detected a mutation/polymorphism in the AMH receptor (AMHRII) gene, a G to A transition in the 11th last exon, at position 7381, changing codon CGG to CAG, and Arg 494 to Gln .

The Doppler US examination one month after the operation showed normal testicular size within the scrotum, homogenous parenchymal echogenicity, symmetric and normal perfusion. The patient was healthy and free of any complications at 1 year of follow-up.

DISCUSSION

Transverse testicular ectopia, a rare form of testicular ectopia, is also referred to as the “crossed testicular ectopia” or “unilateral double testis” results from the migration of testicles through/into a single inguinal canal or hemiscrotum (1).

Transverse ectopic testicle is usually associated with an indirect inguinal herniation (50%) or PMDS (30%), or infrequently with a mixed variety including herniation, hypospadias, disorders of sexual differentiation, and scrotal abnormalities (20%) (2).

The type of the possible genetic defect in these patients can be predicted from the level of serum AMH, which is normally low or undetectable in patients with AMH gene mutations and at the upper limit of normal in receptor mutations (3). Our patient had a normal level of serum AMH, and the mutation analysis confirmed a defect in the AMH-receptor gene. To the best of our knowledge, no cases with AMH receptor gene defect and CoA have been reported in the literature so far. Josso et al. stated that other severe congenital abnormalities were present in half of their patients with AMH receptor gene defect (4).

The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is aplasia of Müllerian duct derivatives in a phenotypically female and this syndrome might be considered as a mirror image of PMDS (5). The MRKH-syndrome is frequently associated with other malformations including renal, vertebral and cardiac abnormalities (6). Recently, severe aortic stenosis were reported in a patient with Müllerian agenesis and renal abnormalities (7). CoA seen in our patient might be a different presentation of TTE.

Previously, most cases were diagnosed during hernia repair, while more recently, preoperative US allows detection of these patients before surgery (8). Similarly, our patient was diagnosed preoperatively by US performed after physical examination.

Currently, laparoscopy is considered both diagnostic and therapeutic in these patients. Accordingly, the importance of performing a laparoscopic intervention has been underscored by several authors including Dean and Shak (9). Here we report a case, of a 6-month-old boy preoperatively diagnosed with TTE by ultrasound examination and treated with laparoscopy-assisted surgery.

Once a diagnosis of TTE is made, a conservative surgical approach in the form of orchiopexy is recommended for the preservation of fertility. The treatment for testicular ectopia involves either a transeptal or extraperitoneal transposition orchiopexy (10). In TTE cases, a transeptal orchiopexy is a recommended surgical option. Testicular biopsy is not needed in the case of TTE with normal male karyotype.

The patient characteristics that have made us report this case include the preoperative detection of TTE ultrasound, its concurrent occurrence with aortic coarctation, the role of AMH receptor mutation in its development, and requirement for a multi-disciplinary approach for its management.

Informed Consent: Informed consent was obtained from the patient's parents for publication of this case report and accompanying images.

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