Congenital Anomalies

Case Report of a Man With Sertoli Cell Only, Transverse Testicular Ectopia, and External Auditory Canal Atresia

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Abstract

Transverse testicular ectopia (TTE) is an uncommon congenital anomaly, reported mostly as pediatric case reports. Hereewith we report a 43-year-old man presenting with sertoli cell only, Transverse testicular ectopia and external auditory canal atresia.

Introduction

Transverse testicular ectopia (TTE) is a rare congenital anomaly with uncertain etiology. Around 150 cases have been reported in the available literature, mostly as pediatric case reports. Herewith, we present, for the first time, a 43-year-old man with sertoli cell only syndrome (SCOS), TTE and external auditory canal (EAC) atresia. The patient had normal male type secondary sex characteristics.

Case presentation

A 43-year-old patient had applied to our outpatient department with the chief complaints of infertility and left inguinal bulging. On physical examination, along with EAC atresia (microti) (Fig. 1), the right testis was unpalpable. The left testicle was in place with normal dimensions. There was a second mass on the posterosuperior position with a 2.5 cm longitudinal axis and very similar feeling of normal testis, and an evident hernia on the left side. Hematologic examination and biochemistry lab data were within normal limits. LH and total testosterone values were also within normal range, together with slightly elevated FSH value (14.29 mIU/mL; 1.5–12.4). There was azoospermia in the first semen analysis. Repeated semen analysis, 3 months later, revealed severe oligoasthenozoospermia. The karyotype of the patient is 46, XY with no Y chromosome micro deletions. Scrotal Color Doppler ultrasound and MRI examinations revealed that both testicles were in the left hemiscrotum. The lower testis had 50 × 27 × 43 mm dimensions and the posterosuperior located testis had 26 × 14 × 23 mm dimensions. Total abdominal ultrasonic examination revealed no developmental anomaly of urinary system.

The patients had a history of mediastinal cyst operation a year ago. We performed bilateral inguinal exploration. Both of the testicles were located in the left scrotum. After completion of biopsies, right orchiopexy of the posterosuperiorly located ectopic testis with crossed over cord structures and left inguinal herniorrhaphy operations were performed. There was no Müllerian duct remnant. Histologic examination revealed SCOS (Fig. 2) with few focal spermatogenesis areas. Family history of the patient revealed that mother and father were cousins, and one of his uncles had the diagnosis of EAC atresia, without infertility problem.

Discussion

TTE is a rare form of testicular ectopia. It was first described by Von Lengerke in 1886. Several etiologic factors have been suggested for the development of TTE. First of all, mechanical inhibition of testicular descent by Müllerian ducts remnants. However,
no Müllerian duct remnants have been observed in our case. Persistent Müllerian duct remnants (PMDR) are considered to be due to lack of Müllerian-inhibiting substance (MIS). The presence of appendices testis, in our case, indicates that MIS functioned normally during embryogenic development. Second, fusion of Wolffian ducts is also considered a possible mechanism. There was no common ductus deferens in our case. Third, familial cases of TTE have been reported and mostly associated with PMDR. There was no TTE diagnosis among the relatives of our patient.

Three types of TTE have been described; Type 1, associated with hernia (40–50%), type 2, accompanied by PMDR (30%), type 3, associated with hypospadias, pseudo hermaphroditism, and scrotal anomalies (20%). Our case carries similarities with that of type 1.

Association of genital abnormalities with ear anomalies are presented in CHARGE syndrome, but genital anomalies such as micropenis and cryptorchidism are defined as minor signs. To our knowledge, the association of EAC atresia with TTE as in our case has not yet been reported. EAC atresia in our case was in accordance

Figure 1. Congenital microtia (small, malformed auricle/pinna) and external auditory canal atresia.

Figure 2. Expansion of interstitial space along with increased connective tissue. Atrophic appearance in the seminiferous tubules. Basement membrane thickening (A, HE, ×100/HPF). Tubules with thickened basal membrane are lined by sertoli cells some of which are altered in shape and detached from the basal membrane, and devoid of germ cells (B, HE, ×400/HPF).

Figure 3. Bone window images of axial cranial computed tomography (CT) scan at the level of auricles and external auditory canals. There is a normal left external auditory canal (EAC). The right auricula has a non-uniform structure with a 2-mm AP axis diameter from external auditory canal meatus, and presents morphologic characteristics in accordance with EAC atresia. The ossified auricles bilaterally are normal with right microtia.
with Tanzer classification type 2a, as the external ear canal atresia together with complete hypoplasia (microtia: small, malformed auricle/pinna) (Fig. 3). Conductive type hearing loss is the most frequent in those patients, was also present in our case. Our patient underwent reconstructive surgery with unsatisfactory results for this deformity.

Preoperative diagnosis of TTE may be established with ultrasound examination, magnetic resonance imaging and venography, but still up to 65% is diagnosed at inguinal hernia operations. Contrary to a few authors suggesting that testicular biopsy is unnecessary for the TTE with normal male karyotype, definitive diagnosis is established first with gonadal biopsy and followed by orchiopexy.1

Primary testicular failure affects less than 1% of all men and 10% of those with fertility problem.1 Hypospermatogenesis, maturation arrest and Sertoli cell only syndrome (SCOS) with or without focal spermatogenesis islands are the most frequent histologic patterns. SCOS may often be associated with Y chromosomal micro deletions. Our case had no micro deletions of Y chromosome, and had focal spermatogenetic islands. Even in severe spermatogenesis defects, acceptable pregnancy and implantation rates were reported with the use of ICSI.4 Although sperm retrieval rates is lower in infertile patients with SCOS, successful sperm retrieval rates up to 86% were reported when focal spermatogenesis is present, and even 19% when pure SCOS pattern is present.5 Along with these suggestions, we had strongly recommended ICSI to our patient. Nowadays, our patients’ wife is about the completion of first trimester of pregnancy and hospitalized for abortion risk.

TTE patients bear an increased risk of malignant transformation. Malignant transformation incidence rates up to 18% have been reported.1 It is reported that risk of malignancy in undescended testicles decreased if their orchiopexy operation performed before the age of 10.1 In our patient, we preferred conservative surgery in form of orchiopexy to preserve fertility potential of the patient, despite decreased chance.

Conclusion

Our case suggests that detailed urogenital examination should be performed for the patients with external ear channel atresia.

Conflicts of interest

The authors declare that they have no relevant conflicts of interest.

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