Transverse testicular ectopia associated with persistent Müllerian duct syndrome treated by transseptal orchiopexy

A case report

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Abstract

Rationale: Persistent Müllerian duct syndrome (PMDS) is rare form of male pseudohermaphroditism characterized by the presence of uterus and fallopian tubes with normal external genitalia and secondary sexual characteristics. Transverse testicular ectopia (TTE) is also a rare form of testicular ectopia that may be associated with PMDS.

Patient concerns: We present a 2-year-old boy who presented with bilateral non-palpable testes with left inguinal mass.

Diagnosis: TTE with PMDS.

Interventions: On exploration, both testes were present in the left inguinal region. Uterus and fallopian tubes were located between the testes. A hysterectomy was performed with resection of the underdeveloped fallopian tubes. Bilateral orchiopexy was performed by placing both gonads into subdartos pouches in each scrotum with transseptal approach.

Outcomes: Both testes were palpable in both the scrotum at 1-year postoperative follow-up and we are planning a regular follow-up.

Lessons: In case of TTE with PMDS, optimal surgical approach with orchiopexy and excision of Müllerian duct should be needed. A long-term postoperative follow-up is necessary for assessment of malignant transformation and infertility.

Abbreviations: MIF = Müllerian duct inhibitory factor, PMDS = persistent Müllerian duct syndrome, TTE = transverse testicular ectopia.

Keywords: child, persistent Müllerian duct syndrome, transverse testicular

1. Introduction

Persistent Müllerian duct syndrome (PMDS) may result from the failure of synthesis of Müllerian duct inhibitory factor (MIF) or the failure of activation of MIF in end organs.[1,2] In most cases external genitalia shows normal male appearance, so the diagnosis is often made incidentally during herniorrhaphy or exploration for undescended testis. Subsequently, more than 200 cases have appeared in the literature.[3] Transverse testicular ectopia (TTE) is a rare form of ectopic testis. In this condition, both testes are located in one inguinal side. The association between PMDS and TTE is even more uncommon. We report a case of an infant in which PMDS and TTE were diagnosed at operation for bilateral undescended testes.

2. Case report

A 2-year-old boy was admitted with bilateral undescended testes. On physical examination, both testes were not palpable in the scrotum and a firm mass was palpated in the left inguinal area. Other physical findings were normal. Ultrasonography showed 2 undescended testes in left inguinal canal and herniation of omental fat through the left inguinal canal (Fig. 1). At the time of operation, the left inguinal region was explored initially. Around 2 gonads were found in the left inguinal area, both gonads were similar sized and located vertically. There were 2 vas deferenses and vascular structures accompanying each gonad, between them tubular structures resembling an immature uterus and fallopian tubes were located (Fig. 2). Both gonads were placed into subdartos pouches in each scrotum by the transseptal approach. The tubular structures were carefully removed and the histopathologic examination showed PMD (Figs. 2 and 3). After operation, all hormonal studies showed normal and the karyotype based on peripheral blood was 46 XY. This study was approved by the institutional review board of the Chonnam National University Hospital. Informed consent for publication of the case details was given by the patient’s family.
3. Discussion

TTE is a rare form of testicular ectopia of uncertain embryological etiology, which is commonly associated with other anomalies like PMDS, inguinal hernia, hypospadias, true hermaphroditism, pseudohermaphroditism, or scrotal anomaly. Several theories explain the genesis of TTE. The possibility of the development of both testes form the same genital ridge was reported by Berg.\(^4\) Kimura concluded that if both vas deferenses arose from one side, there had been unilateral origin but if there was bilateral origin, one testis had crossed over.\(^5\) Gupta and Das\(^6\) hypothesize that the adherence and fusion of the developing Wolffian ducts took place early, and that descent of one testis caused the second one to follow.

TTE can be classified to 3 types by the presence of associated disorders: Type 1, accompanied only by hernia (40% to 50%); type 2, accompanied by persistent or rudimentary Müllerian duct structures (30%); and type 3, associated with disorders other than persistent Müllerian remnants (inguinal hernia, hypospadias, pseudohermaphroditism, and scrotal abnormalities) (20%).\(^7\)

The overall incidence of malignant transformation of gonads in TTE is 18%.\(^8\) For the preservation of fertility, a surgical approach in the form of orchiopexy is recommended after the diagnosis of TTE is made. Either trans-septal or extra-peritoneal transposition orchiopexy is possible. A search for Müllerian remnants and other anomalies and long term postoperative follow-up should be necessary.

PMDS shows karyotype of 46 XY and normal male external genitalia, with internal Müllerian duct structures. In PMDS patients undescended testes, fallopian tubes, uterus or upper vagina may be seen and in most cases diagnosis are made by the presence of those structures while exploration is performed.

![Figure 1. Ultrasonography of left inguinal area showing both testes.](image)

![Figure 2. Macroscopic and microscopic features. Macroscopically, the rudimentary uterus (arrowhead) and fallopian tube (arrow) were observed (A). Microscopically, uterus was composed of endometrial tissue, myometrium and perimetrium (B, D). Fallopian tube was composed of mucosa with branching folds and muscular wall (C).](image)
Transseptal orchiopexy is the operative procedure of choice.\textsuperscript{[9]} A testis with longer spermatic cord and vessels can be placed in the empty scrotum after crossing the scrotal midline septum.\textsuperscript{[10]} It is controversial whether the Müllerian duct remnants should be removed or not. However, most clinicians recommend the removal and undergoing biopsy of the persistent Müllerian structures. The vas deferens and vessels should not be dissected extensively because of the possibility of trauma. In this case, both testes were well palpated in both sides of scrotum at 1-year postoperative follow-up and we are planning a regular follow-up.

TTE associated with PMDS is a rare case which is incidentally discovered during surgery of undescended testis. In these patients, optimal surgical approach with orchiopexy and excision of Müllerian duct is needed. A long term postoperative follow-up is necessary for assessment of malignant transformation and infertility.

**Author contributions**

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