CASE REPORT – OPEN ACCESS

International Journal of Surgery Case Reports 37 (2017) 72–75

Contents lists available at ScienceDirect

International Journal of Surgery Case Reports

journal homepage: www.casereports.com

# A rare case of male pseudohermaphroditism-persistent mullerian duct syndrome with transverse testicular ectopia – Case report and review of literature

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ARTICLE INFO

*Article history:*
Received 15 March 2017
Received in revised form 1 June 2017
Accepted 5 June 2017
Available online 15 June 2017

*Key words:*
Persistent mullerian duct syndrome
Transverse testicular ectopia
Testis
Cryptorchidism
Orchidopexy
Case report

ABSTRACT

*INTRODUCTION:* Persistent Mullerian duct syndrome (PMDS) is a rare type of male pseudohermaphroditism. Transverse testicular ectopia (TTE) is characterized by one testis moving to the opposite side and both testes traversing the same inguinal canal.
*CASE PRESENTATION:* An 11-month-old boy presented with bilateral cryptorchidism. The left testis was not palpable; the right testis was canalicular with a right inguinal hernia. Ultrasound showed both testes located in the right inguinal canal. Right inguinal exploration revealed two testes with intact spermatic cords. A primitive uterus with fallopian tubes was also identified on opening the processus vaginalis. After herniotomy, bilateral orchidopexy was carried out (left orchidopexy through a *trans*-septal approach). Karyotyping confirmed a male gender (46XY). One year after the operation, ultrasound showed both testes to be in good condition.
*DISCUSSION:* PMDS is caused by defects in the gene that encodes Antimullerian hormone(AMH). Treatment aims to correct cryptorchidism and ensure appropriate scrotal placement of the testes. Malignant transformation is as likely as the presence of abdominal testes in an otherwise normal man. Failing early surgical correction, gonadectomy must be offered to prevent malignancy. Division of the persistent mullerian duct structures is indicated only in patients where persistence interferes with orchidopexy.
*CONCLUSION:* TTE should be suspected in patients presenting with inguinal hernia on one side and cryptorchidism on the other side. Herniotomy and bilateral orchidopexy is optimal. Removal of mullerian structures may injure the artery to vas deferens and is hence not recommended. Follow-up for fertility assessment in the latter years should be counselled.



## 1. Introduction

Persistent Mullerian duct syndrome (PMDS) occurs in a phenotypic and genotypic male who has a concurrent uterus, fallopian tubes, and an upper vagina as the mullerian ducts fail to regress. In transverse testicular ectopia (TTE), one of the testes moves to the opposite side and both testes pass the same inguinal canal. The concurrence of TTE and PMDS is extremely rare.

We report a patient with PMDS and TTE who presented with a right-sided inguinal hernia and left undescended testis. This child was managed in our tertiary care institution. This report highlights the pathogenesis and management of PMD and TTE, the possible complications of dividing these structures and the necessity for fertility follow-up and gonadectomy in certain situations.

## 2. Patient information

An 11-month-old boy was brought by his mother to our hospital for evaluation of bilateral cryptorchidism.

## 3. Clinical findings

The left testis was impalpable, the right testis was canalicular and a right inguinal hernia was present.

## 4. Timeline

Cryptorchidism was detected by the caregivers only at 11 months and the patient was brought in.

## 5. Diagnostic assessment

Ultrasound revealed a small left and a larger right testis, both located in the right inguinal canal. Karyotyping was 46XY.

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http://dx.doi.org/10.1016/j.ijscr.2017.06.016

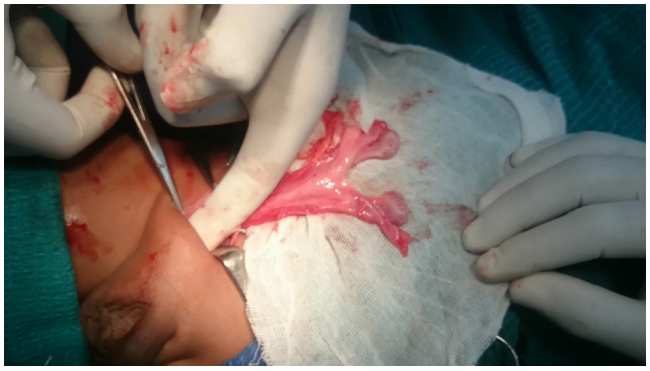

**Fig. 1.** Both testes in right inguinal hernia.

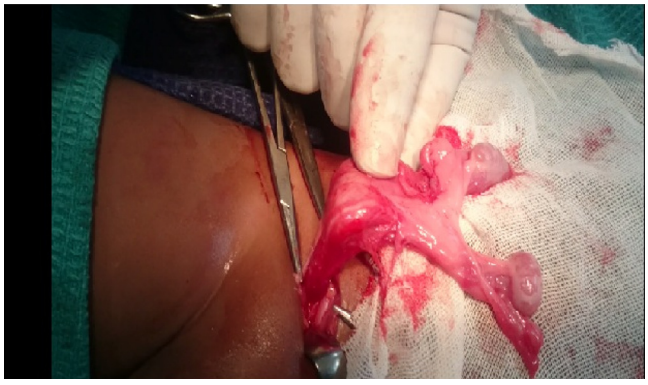

**Fig. 2.** Tubes and Uterus (Primitive) clearly seen.

## 6. Therapeutic intervention

An inguinal exploration was planned to correct the cryptorchidism. Pre-operative assessment and anesthetic clearance was obtained. The surgery was performed under general anesthesia with caudal block by a senior pediatric surgeon. Right inguinal exploration revealed bilateral testes with intact spermatic cords and a sac in the right inguinal region suggesting transverse testicular ectopia(Fig. 1). When the processus vaginalis was opened, a primitive uterus together with fallopian tubes was identified confirming the presence of persistent mullerian duct syndrome (Figs. 2 and 3). After performing herniotomy, bilateral orchidopexy was carried out (left orchidopexy through a *trans*-septal approach). The mullerian structures were not divided to prevent injury to vessels supplying the testes and vas deferens. The patient was discharged the following day. Sutures were removed on the 10th postoperative day and no apparent complications were detected.

## 7. Follow-up and outcomes

Postoperative follow-up was uneventful. One year after the operation, ultrasound showed both testes to be in good condition.

## 8. Discussion

An ectopic testis is one which is found in a location not along the standard path of testicular descent. This differentiates this anomaly from an undescended testis. Crossed or transverse testicular ectopia was first described by Von Lenhossek in 1886 [1]. TTE is characterized by both testes descending through a single inguinal canal and being on the same side with contralateral cryptorchidism.

The exact number of documented cases of TTE till date has not been reported. Fourcroy et al. summarized that about 100 cases of

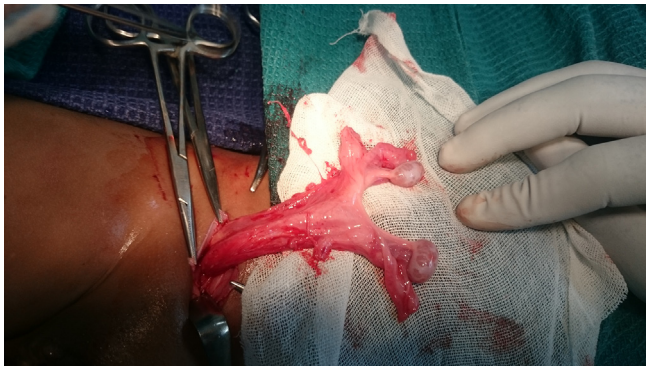

**Fig. 3.** TTE with PMDS.

TTE had been reported till 1982 [2]. Our PubMed search identified 152 cases published after 1982 bringing the number of reported cases to about 260 till date.

TTE associated with PMDS is extremely rare and Ferri et al. state that 10 cases had been reported till 1999 [3]. Review of the PubMed database using 'persistent mullerian duct syndrome with transverse testicular ectopia' identified 47 more cases since 1999. Thus, to our knowledge there are only 57 reported cases of TTE with PMDS till date.

Various anatomic factors (defective implantation, rupture or tearing of the gubernaculum, obstruction of the internal inguinal ring, adhesions between the testis and adjacent structures, late closure of the umbilical ring, etc.) are suggested as causative factors in the failure of testicular descent.

TTE has been classified as three types: Type I accompanied by hernia (40–50%),Type 2 by persistent or rudimentary mullerian duct structures (30%) and Type 3 associated with disorders such as hypospadias, pseudohermaphroditism and scrotal abnormalities.

TTE patients sometimes have associated PMDS. Mullerian structures include fallopian tubes, uterus and the upper part of the vaginal canal. PMDS is characterized by the persistence of these structures in their primitive form in male children.

PMDS is caused by defects in the gene that encodes the synthesis or action of Mullerian inhibiting Factor or Antimullerian hormone [4–8]. AMH is secreted by the sertoli cells of the developing testes by 7 weeks of gestation. The other cause of persistence of Mullerian ducts, testicular dysgenesis, usually affects both Sertoli and Leydig cells. Persistence of Mullerian derivatives is then associated with external genital ambiguity.

Soon after testicular differentiation, AMH gene expression is induced by SOX9 in sertoli cells which results in an ipsilateral regression of the mullerian ducts by 8 weeks of gestation, before the emergence of testosterone secretion or the stimulation of the wolffian ducts [9].

The gene encoding AMH I is located in the short arm of chromosome 19(19p13.3). AMH signaling is mediated via a heterodimeric receptor consisting of type 1 and type 2 serine/threonine kinase receptor, the type 2 part of the receptor mediates ligand specificity and the type 1 receptor activates a downstream signaling cascade. ALK 2, ALK 3 and ALK 6 have all been linked to type 1 signaling and decreased expression or deletion of the former two disrupts mullerian duct regression [10]. The type 2 receptor is called as AMHR2 and is located on chromosome 12(12q13). AMH mutations are often autosomal recessive. Affected patients usually have an AMH gene mutation in 45%, and in the AMRH2 receptor gene in another 39%. In the remaining 15%, no mutation was detected, implicating genes coding for other factors in the AMH transduction cascade [11]. AMH and AMH2 receptor mutations are transmitted through an autosomal recessive pattern and are symptomatic only in males.