Persistent Mullerian Duct Syndrome in Adult : A Case Report

SOMKIAT SUNPAWERAVONG, M.D.*,
CHUSAK PRIPATNANONT, M.D.*

Abstract

A twenty-eight year old phenotypically and genotypically normal male adult was admitted with a right inguinal hernia, a right retractile testis and left cryptorchidism. During surgery, he was found to have a uterus in the right spermatic cord and a left undescended testis (intra abdominal type). The uterus was excised, left orchidectomy, right orchidoplexy and right herniorrhaphy were performed. This rare case is reported as persistent Mullerian Duct Syndrome (PMDS) which is caused by impaired action of Mullerian Inhibiting Substance (MIS) in regressing the Mullerian duct.

Key word : Persistent Mullerian Duct Syndrome, Mullerian Inhibiting Substance

SUNPAWERAVONG S & PRIPATNANONT C
J Med Assoc Thai 2000; 83: 1541-1543

The Mullerian duct regresses in males as a result of Mullerian Inhibiting Substance (MIS). The disorder of regression is known as persistent Mullerian Duct Syndrome (PMDS). The rarity of this syndrome is generally discovered by the surgeon while repairing an inguinal hernia or an undescended testis. The management of this disease is controversial.

CASE REPORT

A phenotypically normal 28-year-old male had a right inguinal hernia, a right retractile testis and left cryptorchidism. The patient had a repeated history of symptoms over a long period of time. Physical examination showed a large, but easily reducible, right indirect inguinal hernia with a retractile testis and left cryptorchidism.

* Department of Surgery, Faculty of Medicine, Prince of Songkla University, Songkhla 90110, Thailand.

Ultrasound showed the left undescended testis was on the right side of the urinary bladder. Sperm analysis showed sterility.

During surgery, the right inguinal canal was explored. A hypervascular soft tissue mass, 1.5 x 2 x 3 cm in size, was found in the mid part of the spermatic cord. In addition, a left undescended testis (intra abdominal type), 1 x 1.5 x 2.5 cm, and a right retractile testis, (1.5 x 2 x 3 cm) was found. See Fig. 1.

The soft tissue mass was excised and left orchidectomy, right orchidoplexy and herniorrhaphy were done. The pathologist reported

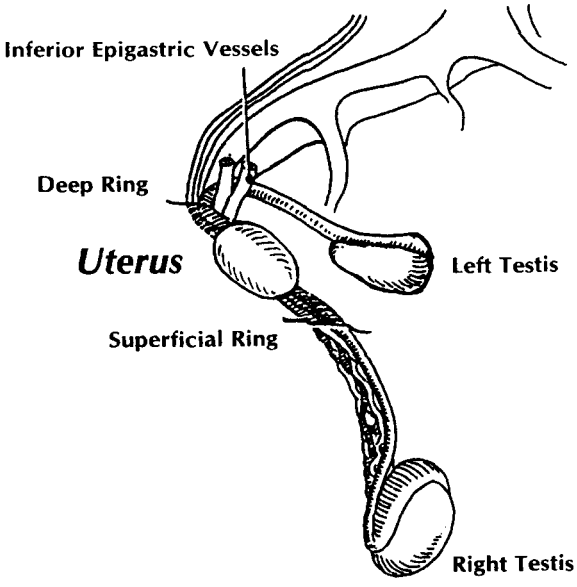

Fig. 1. Diagram demonstrates the operative finding.

the soft tissue mass as an atrophic uterus and uterine tubes, the left testis was atrophied and azoospermia. On the genetic evaluation a 46, XY karyotype and the sex hormones are shown in Table 1.

Table 1. Levels of sex hormones.

Hormones	levels	normal values
B- HCG	1 mIU/ml.	0 - 10.00
FSH	24.47 mIU/ml.	1.1 - 13.50
LH	9.58 mIU/ml.	0.4 - 5.70
Testosterone	5.11 ng/ml.	2.45 - 18.63

DISCUSSION

In normal embryogenesis; the differentiation of an indifferent gonad into the testis or ovary depends on the sex chromosomes. The testis produces two principal hormones, Mullerian Inhibiting Substance (MIS)[1] and testosterone. MIS causes regression of the Mullerian ducts, which would otherwise develop into the uterus, fallopian tubes and upper vagina. Persistent Mullerian Duct Syndrome (PMDS) is believed to be caused by a defect in the secretion or action of MIS[2,3].

This rare condition was first described by Nilson in 1940 as "hernia uteri inguinales"[4]. The classical presentation is a patient who is phenotypically and genotypically a male, with unilateral cryptorchidism and a contralateral hernia which contains Mullerian duct structures such as the uterus, fallopian tubes and vagina. An etiology of this syndrome may be genetically inherited either by autosomal recessive genes with sex limitation or X-linked recessive mutation[5].

Preoperative diagnosis of PMDS is difficult. It is usually made during an operation for inguinal hernia or cryptochidism. Treatment is controversial[6-10] with most clinicians recommending excising the persistent Mullerian structures. But due to the close relationship between the vas deferens and the Mullerian structures, it is easy to injure the vas deferens. Some clinicians suggest leaving the persistent Mullerian structures because malignancy has not been reported and no problems exist.

Recommendation in children, especially between one and two years old, is that an orchidoplexy should be performed to correct the undescended testis and careful follow-up conducted due to reports of testicular tumor. But in contrast, such as in our case, an orchidectomy should be performed in adults on the undescended testis to avoid malignancy.

(Received for publication on March 2, 1999)

REFERENCES

1. Josso N, Picard JY, Tran D. The anti-mullerian hormone. Recent Prog Horm Res 1977; 33: 117-67.
2. Donahoe PK, Ito Y, Morikawa Y, Hendren WH. Mullerian inhibiting substance in human testes after birth. J Pediatr Surg 1977; 12: 323-30.
3. Hutson JM, Donahoe PK, Budzik GP. Mullerian inhibiting substance : a fetal hormone with surgical implications. Aust NZ J Surg 1985; 55: 599-605.
4. Nilson O. Hernia uteri inguinales beim manne. Acta Chir Scand 1940; 83: 231-48.
5. Beheshti M. Familial persistent mullerian duct syndrome. J Urol 1984; 131: 968.
6. Peppis C, Constantinides C, Chiotis D, Dacou VC. Persistent mullerian duct structures in cryptorchid male infants:surgical dilemmas. J Pediatr Surg 1979; 14: 128.
7. Donahoe PK. Neoseminal vesicle created from retained mullerian duct to preserve the vas in male infants. J Pediatr Surg 1988; 23: 272.
8. Fernandes ET, Hollabaugh RS, Young JA, Wilroy SR, Schriock EA. Persistent mullerian duct syndrome. Urology 1990; 36: 516-8.
9. Yip CH, Chang KW. Persistent mullerian duct syndrome-a case report. Singapore Med J 1991; 32: 363-4.
10. Loeff DS, Imbeaud S, Reyes HM, Meller JL, Rosenthal IM. Surgical and genetic aspects of persistent mullerian duct syndrome. J Pediatr Surg 1994; 29: 61-5.

กลุ่มอาการเพอร์ซิสเต็นท์ มูลเลอเรียน ดัคท์ ในผู้ใหญ่

สมเกียรติ สรรพวีรวงศ์, พ.บ., ชูศักดิ์ ปริพัฒนานนท์, พ.บ.*

ผู้ป่วยชายไทย อายุ 28 ปี มาโรงพยาบาลด้วยอาการมีไส้เลื่อนลงถุงอัณฑะ ด้านขวาร่วมกับภาวะไม่มีลูกอัณฑะด้านซ้าย ได้รับการผ่าตัดที่ขาหนีบขวาเพื่อรักษาอาการไส้เลื่อน พบก้อนมดลูก และลูกอัณฑะซ้าย ผู้ป่วยได้รับการตัดก้อนมดลูก และลูกอัณฑะซ้ายร่วมกับการเย็บซ่อมรักษาอาการไส้เลื่อนผู้ป่วยรายนี้ได้รับการวินิจฉัยเป็น Persistent Mullerian Duct Syndrome (PMDS) ซึ่งเกิดจากความผิดปกติในการทำงานของ Mullerian Inhibiting Substance (MIS) ลักษณะความผิดปกตินี้พบน้อยมาก โดยเฉพาะอย่างยิ่งในผู้ใหญ่

คำสำคัญ : Persistent Mullerian Duct Syndrome, Mullerian Inhibiting Substance

สมเกียรติ สรรพวีรวงศ์, ชูศักดิ์ ปริพัฒนานนท์
จดหมายเหตุทางแพทย์ ฯ 2543; 83: 1541-1543

* ภาควิชาศัลยศาสตร์, คณะแพทย์ศาสตร์ มหาวิทยาลัยสงขลานครินทร์, สงขลา 90110