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Persistent mullerian duct syndrome in a child: case report and review of literature

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Abstract Herein we report of a case of persistent mullerian duct syndrome diagnosed on laparoscopy. Current knowledges and management are discussed.

Key Words: Persistent mullerian duct syndrome; laparoscopy; orchidopexy; undescended testis

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Introduction

Persistent mullerian duct syndrome (PMDS) is a rare form of male pseudohermaphroditism characterized by the retention of mullerian derivatives in an otherwise normally virilized male [1]. Various procedures have been described for the treatment of PMDS. At present, experience in the surgical treatment of this abnormality is limited to sporadic case reports describing open surgical exploration. In recent decades, laparoscopy has been used for patients with PMDS [2]. Our aim was to present a case of laparoscopically diagnosed PMDS, and review the current evidence for

the use of diagnostic and therapeutic modalities.

Case report

A healthy, 17-month-old male infant was initially evaluated for bilateral undescended testes. He had undergone a previous exploration for right-sided at same hospital, where a viable testis along with the hernia sac with abnormal thick and short spermatic cord and with fimbria-like epididymis was found. Right inguinal herniotomy and orchidopexy was performed after difficult mobilization, and no gonadal biopsy obtained. The patient was transferred our department for further investigation of right nonpalbable testis in the same center. On examination, he had a well-developed phallus and hypoplastic scrotum. A gonad was palpable in the proximal right inguinal canal, and left testis was nonpalpable. Neither ultrasonography reports commented about the presence or absence of right gonad and internal genital structures. Six months later, he was scheduled for laparoscopy.

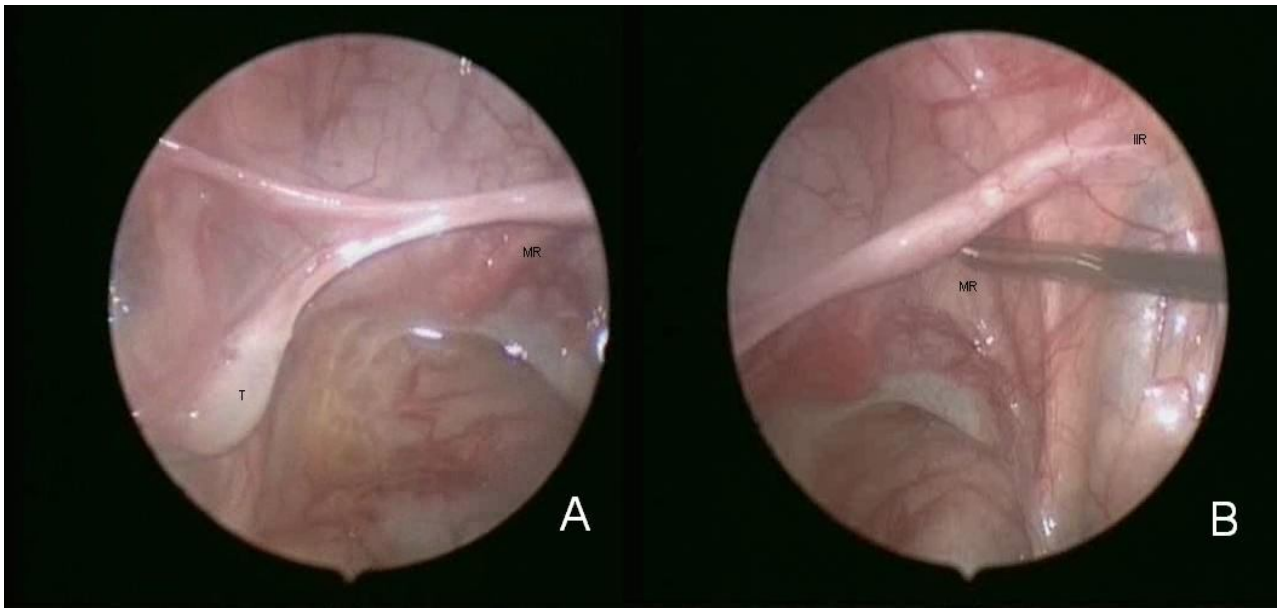

Figure 1. Laparoscopic photographs show the left intra-abdominal testis (T) in a position analogous to ovary, mullerian remnants (MR) in the midline (A), and the remnants and gonadal vessels on the right side are passing through a closed internal inguinal ring (IIR) with traction (B).

A punch biopsy was taken from left gonad, and procedure terminated. Histology Laparoscopy via 5-mm port revealed an average-sized left testis attached to mullerian structures (uterus and fallopian tubes), and the remnants and gonadal vessels on the right side were seen passing through a closed internal ring (orchidopexy and repaired hernia) (Fig. 1 A and B) of the testicular biopsy revealed mature testis consistent with cryptorchid testes. His karyotype was 46 XY. Hormone levels were normal. At the age of 29 months, the patient underwent laparotomy via suprapubic transverse incision. A bulldog clamp was applied to left testicular vessels, and the left testis mobilized after dividing of spermatic vessels (Fowler- Stephens) when no ischemia was seen. The cervicouterine stricture was split longitudinally in the midline to achieve successful orchidopexy after mobilizing the left testis. A subdartos pouch was created in the scrotum, left testis passed through the open inguinal canal to the scrotum, and the hernia was repaired intrabdominally. Postoperative period was uneventful. Follow-up after one year showed right testis in the inguinal canal and left one in the scrotum to be normal.

Discussion

PMDS as a distinct entity can be explained by inadequate mullerian suppression from hormonal influences. The hypotheses for PMDS causation include failure of synthesis or release of mullerian inhibiting substance (MIS) by testicular Sertoli cells, the failure of end organs to respond to MIS, or a defect in the timing of the release of MIS despite the normal male genotype (46 XY) [2,4].

Nilson first described PMDS in a man with an inguinal hernia in 1939 as hernia uteri inguinal [5]. The current incidence of PMDS is felt to be higher than historically reported in literature, secondary to improved diagnostic imaging, better pathologic diagnosis and earlier correction of cryptorchidism [6]. Clinically, the affected patient presents with bilateral cryptorchidism and an inguinal hernia with a palpable testis within the hernia sac. Although imaging techniques may help to investigate the intersex abnormalities, preoperative diagnosis of PMDS is practically impossible because of

the normally developed penis and scrotum. The diagnosis is usually made during an operation for inguinal hernia or bilateral undescended testes [7]. The present case was recognized during a laparoscopic evaluation of left nonpalpable testis.

Three groups of PMDS have been described [3,6,8,9]. Group 1 (female type): Bilateral intra-abdominal testes in a position analogous to ovaries. Group 2 (male type): One testis is found in a hernia sac or scrotum along with the uterus and tubes (hernia uterus inguinal). Group 3 (male type): Both the testes are located in the same hernial sac along with the müllerian structures (transverse or crossed testicular ectopia). Our case was considered as female type, but the patient had been referred to our department as left nonpalpable testis.

When the mullerian structures are encountered during exploration, to exclude the possibility of mixed gonadal dysgenesis, verification of the karyotype and gonadal biopsy should be done [10]. We approached the patient with PMDS in two stages. In first stage, a testicular biopsy was obtained and gender determination was done, and definitive operation performed six months later as a second stage. Loeff et al. [11] also performed the two-stage procedure: testicular biopsies were obtained during the initial operations in each patient, and orchidopexies and the removing of müllerian remnants were done as second stage procedure at several months later. Nevertheless, the staged approach and testicular biopsy in these patients is still under discussion.

The surgical approach of orchiopexy and hernia repair with/without removing mullerian structures in series is consistent with the optimal surgical management recommended in the literature [2,4,6]. While some authors recommend the removal of residues are due to the risk of malignancy, others proposed that surgical excision of persistent mullerian duct structures may result in ischemic and/or traumatic damage to the spermatic cords and testes [2,6,10,12]. In some cases, short and thick müllerian remnants may prevent the orchidopexy, in such a condition, it has been reported that splitting the müllerian remnants in the midline

can achieve an adequate length for the testes to reach the scrotum [2,3,10]. We split the uterine remnant in the midline, and not removed mullerian remnants because of the risk of testicular blood supply damage.

The use of laparoscopy in the management of PMDS has been sporadically reported since 1997. In the last two decades, the laparoscopic approach to the disease has been increasingly popular [3,6]. Laparoscopic approach is a simple, effective and less invasive method of dealing with PMDS. As in our case, mullerian remnants can be diagnosed during diagnostic laparoscopy for impalpable testes. Although laparoscopy is simple and diagnostic, there is reported that the entire procedure can perform laparoscopically such as excision or splitting of mullarian remnants, orchidopexy [3,6,9,10]. Turaga et al. [6] also described an algorithm-based approach for hernia uteri inguinale, depending on laparoscopic findings.

This case report demonstrates that the patients with nonpalpable testes or undescended testes which have abnormal appearance should be evaluated carefully. Optimal surgical management in the patients with PMDS is orchiopexy or staged Fowler-Stephen procedure leaving the uterus and fallopian tubes in their natural place. Laparoscopy has important benefits in the diagnosis as well as the treatment of PMDS. The patients without removal of mullerian remnants should be followed-up closely because of the risk of malignancy.

CONFLICT OF INTEREST

None declared

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