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Robotic/laparoscopic hysterectomy & orchiopexy for familial persistent mullerian duct syndrome

Abstract

Background: Persistent Mullerian duct syndrome (PMDS) in which Mullerian duct derivatives are seen in a male patient is a rare form of pseudohermaphroditism. PMDS is characterized with undescended testes and underdeveloped uterus in genetically and phenotypically male infants or adults.

Case series summary: We present three siblings in which they were referred to our institute with primary infertility and were investigated furtherly. Diagnosed with PMDS and underwent robotic/laparoscopic hysterectomy and orchiopexy.

Discussion: Robotic/laparoscopic approach in the management of PMDS has been published in the pediatric and adult age groups. In the literature, the approaches concluded to be achievable.

Conclusion: As familial PMDS has been presented, minimally invasive approach is feasible with good results.

Keywords: PMDS, orchiopexy, hysterectomy, robotic, laparoscopic

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Saud A Almousa, Abdulrahman I Aljasser, Ahmad M Alhazmi, Naif A Aldhaam, Abdullah S Alfakhri, Maher S Moazin

Department of Urology, King Fahad Medical City, Saudi Arabia

Correspondence: Saud A Almousa, Department of Urology, King Fahad Medical City, Saudi Arabia, Email S_almous@hotmail.com

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Abbreviations: PMDS, persistent mullerian duct syndrome; MIF, mullerian inhibiting factor; MIF, müllerian inhibitory factor; MISR-II, II MIS receptor

Introduction

Mullerian duct derivatives in a male patient are a rare form of internal male pseudohermaphroditism which is known as persistent Mullerian duct syndrome.[1] Insufficient amount of Mullerian inhibiting factor (MIF) and insensitivity of the target organ to MIF are causes of this rare syndrome.[2] In 1939, this rare condition (<1 in 200,000) has been described by Nilson.[3] PMDS is typically characterized by the presence of small underdeveloped uterus in association with undescended testes in genetically male infant or adult with normal external genitals and virilization.[4] Familial PMDS involving two siblings has been described radiographically for the first time in 1976, in which recommendations for surgical management have been made.[5] We report three siblings of familial PMDS underwent robotic/laparoscopic hysterectomy and orchiopexy.

Case series

Three brothers were referred to our institute with primary infertility. The first brother was a 41 years old male. On physical examination, revealed to have normal general appearance, male phenotype, and secondary sexual characteristics. The left testicle was palpable, but right scrotum was empty. Laboratory investigations revealed high FSH & LH, normal prolactin and testosterone. Semen analysis showed azoospermia. Karyotyping showed 46, XY. The patient was admitted for further investigation and diagnostic laparoscopy. MRI pelvis, showed a tortious structure in between bladder and rectum, Resembling as uterus (Figure 1). After counseling the patient, he was consented for diagnostic laparoscopy plus hysterectomy. Uterus was removed and right testis was not found in abdomen. Operative time took 130 minutes, blood loss was 100 ml. Discharged on day 3 postoperatively.

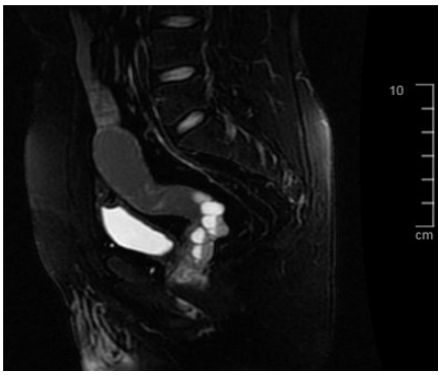

Figure 1 Uterus detected by MRI pelvis.

Second brother is a 48 years old male. He underwent bilateral inguinal hernia repair during childhood. He has normal male general appearance, normal phallus, but underdeveloped scrotum and non-palpable testes bilaterally. Semen analysis showed azoospermia. Diagnostic laparoscopy revealed bilateral gonads and uterus. Pathology reported gonadal biopsies as fibrotic seminiferous tubules with normal uterine wall. The patient underwent robotic hysterectomy Figure 2(A) plus bilateral orchiopexy Figure 2(B). Pathology report concluded the specimen negative for malignancy, only myometrial wall and endometrial tissue consistent with uterine tissue. Third sibling, a 31 years old male, referred as case of bilateral undescended testes with left inguinal hernia. Further investigations were carried out, revealed to have high FSH & LH, semen analysis showed azoospermia. Karyotyping resulted in 46, XY. Ultrasonographic images viewed left inguinal testis. Diagnostic laparoscopy revealed right intra-abdominal testis and uterus. The patient underwent laparoscopic hysterectomy plus bilateral orchiopexy. Surgical pathology was reviewed and demonstrated underdeveloped uterine tissue. Surgical pathology slides showing uterine tissue, vas deferens, epididymeal tissue, and seminiferous tubules for the three siblings (Figure 3).

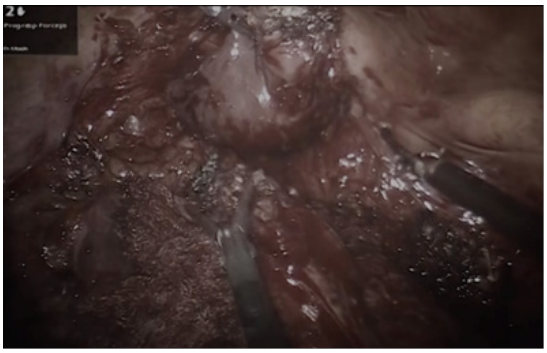

Figure 2A Surgical removal of the uterus.

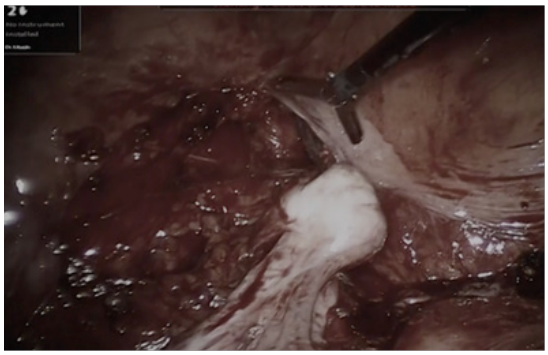

Figure 2B Robotic orchiopexy.

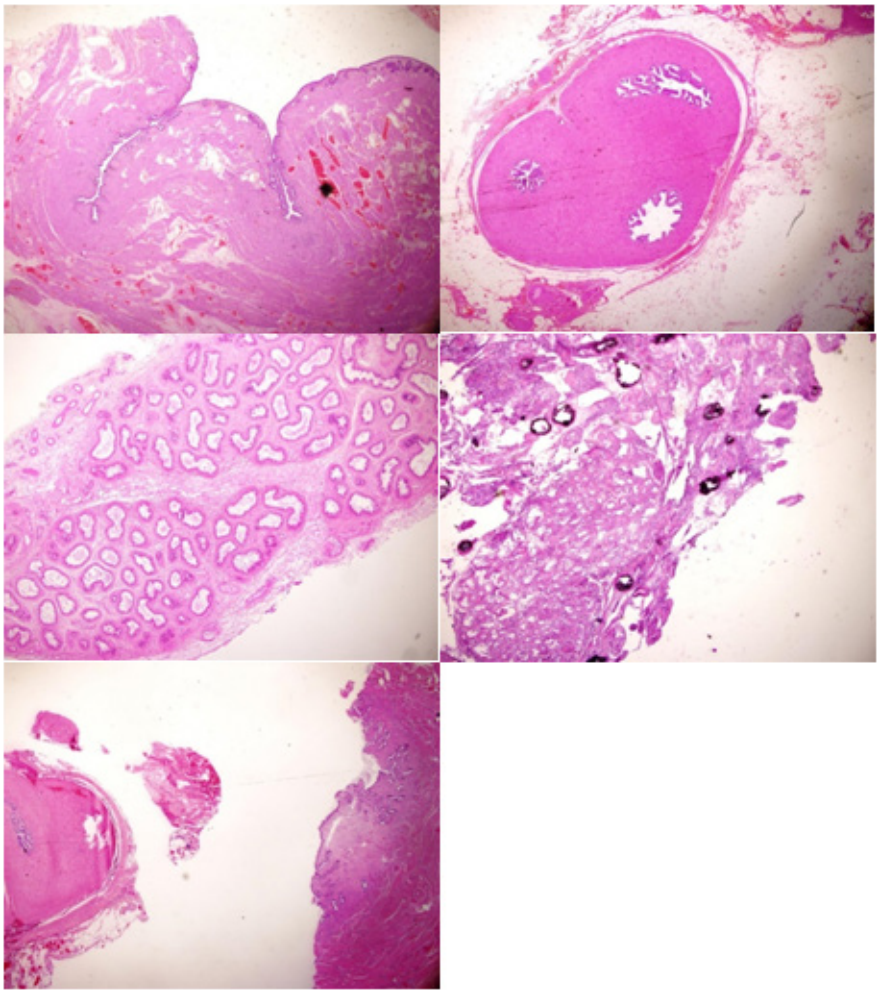

Figure 3 Pathology slides.

Discussion

While there have been reports of consanguinity, the mechanism of PMDS is likely to be multi-factorial. The majority (85%) of cases seems to be affected by autosomal recessive inheritance.[6] Though, the origin of the remaining 15% of cases remains unknown.[7] Jonathan A, W and his colleagues reported a robot assisted laparoscopic hysterectomy, gonadal biopsy, and orchiopexies in an infant with persistent mullerian duct syndrome.[8] Another reported case, published in 2015 by W. L. I. Smith-Harrison. A 27-year-old male

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with a history of bilateral cryptorchidism status post orchiopexy at 18 months presented with intermittent, painless, gross hematuria, and hematospermia. He underwent robotic surgical removal of the remnant Mullerian structures. Surgical pathology demonstrated uterine-like smooth muscle with a central cavity.[9] The risk of malignancy in an ectopic testis in a case of PMDS is similar to that in a cryptorchid testis in a healthy male.[10] Germ cell tumors have been reported in the testis in cases of PMDS, however tumors of the Mullerian duct derivatives are very rare.[11] A laparoscopic hysterectomy and bilateral orchiectomy done by Palanisamy for a patient with PMDS, the histopathological analysis showed the presence of rudimentary uterus with cervix. Both testis showed presence of atrophic seminiferous tubules without any evidence of spermatogenesis. Distinctly, section from right testes had showed presence of seminoma, stage I, which was confirmed by immunohistochemistry.[12]

Müllerian duct derivatives are present in the 8th week of gestation in a male fetus, and their regression is mediated by müllerian inhibitory factor (MIF) produced by Sertoli cells.[13] Some of the PMDS patients have defect in MIS gene located at 19p13, and others have defective gene for type II MIS receptor (MISR-II), located at 12q13. Approximately 85 % of PMDS cases are due to mutation of either MIS or MISR-II genes, in similar proportions. In 15 % of cases, the cause for PMDS is unknown.[14] PMDS can be inherited either as X-linked or autosomal recessive sex-limited trait. PMDS can occur also sporadically. Mutation in MIS and MISR-II gene has autosomal recessive transmission.[15]

Conclusion

We hereby report a family of three siblings with primary infertility all underwent hysterectomy plus orchiopexy. To our knowledge, only few cases of familial PMDS reported in the literature. Minimally invasive approach is feasible with good outcome.

Acknowledgments

None.

Conflicts of interest

The author declares there is no conflict of interest.

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