PERSISTENT MULLERIAN DUCT SYNDROME PRESENTING AS AN INGUINAL HERNIA: A CASE REPORT

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ABSTRACT

A brief report of persistent mullerian duct syndrome (PMDS) with 46XY karyotype that is one of the rarest variety of disorders of sexual differentiation (DSD) accounting only 5% cases of all is being presented. A 21 years old male with a left inguinal hernia and absent right testis presented in surgical outdoor and was operated. On exploration, female genital organs like uterus and fallopian tubes along with contralateral testis were present in the left inguinal canal as a content of sliding left inguinal hernia.

KEYWORDS Mullerian duct syndrome, karyotype, disorders of sexual differentiation (DSD), cryptorchidism, inguinal hernia

Introduction

The term disorder of sexual differentiation refers to a child born without clear demarcation being a male or female phenotype. True bisexuality is ability to function both as male and female to the point of self-reproduction has never been reported. However, cases of intersexuality, there being no complete male, no complete female or both sexes having certain features and organs belonging to each other, have been recognized. Such cases usually present in early childhood with ambiguous genitalia, although presentation in late adulthood is not rare. Late diagnosis may have a severe psychological impact on the patient. Hence, early detection and treatment are essential. Our case is a 21-year-old gentleman who presented with absent right testis and left an inguinal hernia with well-developed secondary sexual characteristics and male external genitalia.

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Case Report

A 21-year-old phenotypically well-developed male patient presented in the outpatient department of our institute with a left inguinal hernia. On examination, the patient had normal external male genitalia. He was found to have left an inguinal hernia and normal left testis in left hemiscrotum. However right testis could not be palpated in right hemiscrotum or inguinal canal, with right hemiscrotum being underdeveloped. As per patient he had only one testis since childhood and had never consulted any doctor regarding it. Besides this, his medical history was insignificant. There was no hypospadias, gynecomastia or any other physical anomaly. He presented normal morning erection and ejaculation and gave no history of urethral bleeding. A diagnosis of left inguinal hernia and undescended right testis was made. Ultrasonography was done. However right testis could not be located in right inguinal canal or abdomen. All routine investigations were within normal range. MRI was planned to locate right testicle but the patient refused, and he asked to operate for a symptomatic hernia only at this time. He was explained about complications that can occur in the undescended testicle. He was taken up for left inguinal hernia surgery. Upon contralateral exploration testis along with uterus and fallopian tubes were present as contents of left sliding inguinal hernia. Since the patient was phenotypically male with well developed male external genitalia, uterus, and fallopian tubes were excised preserving both gonads and vasa defferens. The right gonad was mobilized and brought to right hemiscrotum

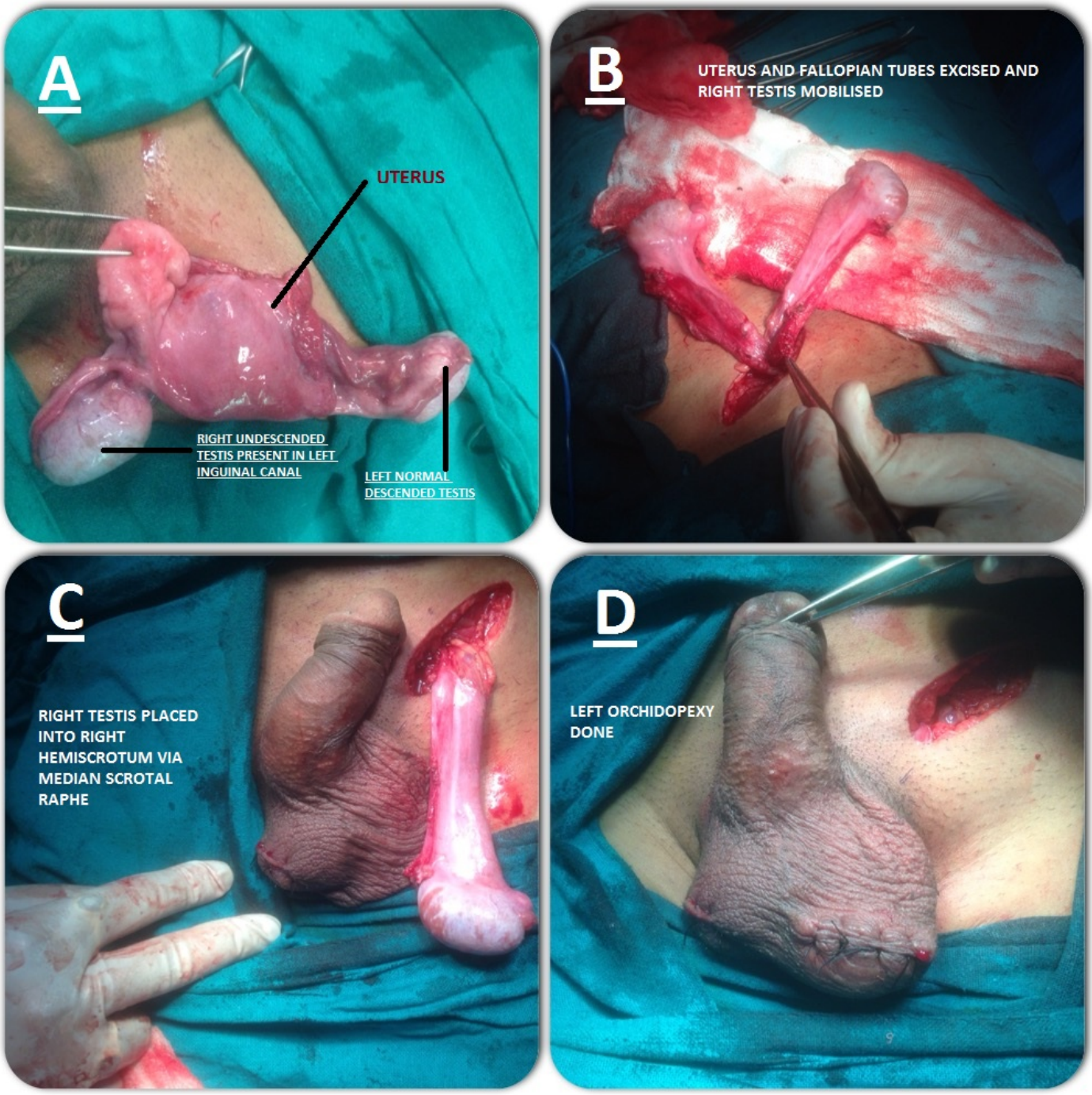

Fig.1: A. Picture is showing uterus, fallopian tubes, bilateral gonads including left normally descended testis and right contralateral testis. Ovaries were not present separately. B. Mullerian structures i.e. uterus and fallopian tubes excised. Testes mobilized. C. Right testes placed into right hemiscrotum into a dartos pouch. D. After bilateral orchidopexy.

through midline scrotal raphe and was fixed in dartos pouch. Similarly left orchidopexy was done (Fig. 1A, 1B, 1C, 1D). A gonadal biopsy was not taken. Histopathology report of excised tissue confirmed uterus and fallopian tubes. Testosterone level measured postoperatively was 2.42 ng/ml that is within normal limits. Karyotyping done postoperatively was 46XY.

Discussion

150 cases of PMDS have been reported in the literature, but its combination with transverse testicular ectopia (TTE) as in our case where there is the presence of both testes on one side of the scrotum or inguinal canal is extremely rare. Diagnosis is usually made incidentally during a groin hernia or orchidopexy operations or imaging[1,2]. PMDS is a rare form of internal male pseudohermaphroditism that constitutes a group of patients with 46XY karyotype with normal male external genitalia and internal müllerian duct structures, bilateral fallopian tubes, uterus, and upper vagina draining into prostatic utricle associated with uni/bilateral undescended testes. This condition is commonly diagnosed when müllerian tissue is encountered during inguinal hernia surgery or orchidopexy.

Patients with PMDS are divided into three categories: (1) majority (60-70%) with bilateral intra-abdominal testes in a position analogous to ovaries; (2) smaller group (20-30%) where one testis is found in a hernia sac or scrotum in association with a contralateral inguinal hernia (3) third group (10%), in which both testes are located in a same hernia so along with fallopian tubes and uterus[2]. Our patient belongs to the rarest group, transverse testicular ectopia associated with PMDS, presenting with right undescended testes and left an inguinal hernia Upon contralateral exploration testis along with uterus and fallopian tubes were present as contents of left sliding inguinal hernia. Mullerian and Wolffian ducts are both present at seven weeks of gestation however testis differentiates by the end of the 7th gestational week in a male child. Factors that control normal male differentiation: testosterone and Mullerian inhibiting factor (MIF) are secreted by Sertoli cells.

Testosterone promotes differentiation of wolfian ducts into the epididymis, vas deferens, and seminal vesicles. PMDS patients have a deficiency of MIF or MIF receptor. Tissues are often intertwined, resulting in non-patency of vas deferens or other parts of the reproductive, excretory ducts resulting in infertility and azoospermia[3-4]. The testes are usually histologically normal in patients with PMDS with an overall incidence of malignant transformation around 18%, similar to the rate in abdominal testes in otherwise healthy men[5]. Since patients are phenotypically male, diagnosis is not usually suspected until surgery for cryptorchidism or hernia repair[6].

The treatment of PMDS is relatively straightforward all patients are phenotypic males who require orchidopexy by open or laparoscopy. Early orchidopexy with the removal of Mullerian structures is the preferred approach. Vasa deferentia lies near the uterus and proximal vagina. Therefore, preservation of necessary müllerian structures to avoid injury to the vasa is recommended to preserve fertility. Parents should make aware of the risk of testicular malignancy and infertility, including genetic counseling.

Conclusion

Surgeons should be aware of rare entities like TTE and PMDS in cases of unilateral or bilateral undescended testis associated with an inguinal hernia so that patients can be diagnosed preoperatively especially in prepubertal age as fertility can be preserved and future risk of malignancy can be addressed. In the case of incidental discovery of Mullerian structures during inguinal hernia repair, we should keep PMDS in mind and manage the patient appropriately.

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None

Disclosure Statement

Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

Competing Interests

The authors declare no conflict of interest.

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