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Pathology

Persistent Mullerian Duct Syndrome – Rare form of Intersex Disorder – A Case Report

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Abstract **Case Report**

Persistent Mullerian duct syndrome is a rare form of intersex disorder, characterized by presence of the uterus and fallopian tubes in a otherwise 46 XY male.[1] We report a 21 years old boy with bilateral cryptorchidism presented with bilateral inguinal hernia having presence of uterus and bilateral fallopian tubes diagnosed as PMDS.

Keywords: Persistent Mullerian duct syndrome, intersex disorders, and male pseudohermaphroditism.

INTRODUCTION

Persistent Mullerian duct syndrome (PMDS) is a rare form of intersex disorder which is characterized by failure of regression of Mullerian duct structures like the uterus and fallopian tubes in a otherwise normal male. Individuals with PMDS are genetically 46 XY, without any chromosomal abnormalities [1]. Here we report a 21 years old boy with bilateral cryptorchidism presented with bilateral inguinal hernia having presence of uterus and bilateral fallopian tubes.

CASE REPORT

A 21 years old male patient presented with complaints of bilateral inguinal hernia of which right sided was scrotal type with evidence of hydrocele and left sided was bubuocele type. He had absent testes since birth with fully developed secondary sexual characters. MRI of whole abdomen showed bilateral empty scrotal sacs, and presence of uterus with elongated cervix. Right testis seen placed right posterior to the fundus of urinary bladder and left testis adherent to the fundus of uterus. Cervix seen opening in the prostate. Semen analysis showed no presence of sperm after 4 days of abstinence. Serum testosterone level was 5.39 ng/ml (biological reference value – 3.0-10.6 ng/dl) and LH and FSH was 16.15 mIU/ml (biological reference value – 1.7-8.6 mIU/ml) and 43.89 mIU/ml (biological reference value – 1.5-12.4 mIU /ml) respectively. Anti Mullerian hormone level in serum was 0.56 ng/ml (biological reference value – 0.6-13.7 ng/ml). Genetically he was 46 XY. Open laparotomy was done and uterus and bilateral fallopian tube was removed and right orchidopexy was done. Uterus and both fallopian tube were received for HPE. Uterus with cervix measured 5x3x2 cm and cervix at external os measured 1 cm. Both fallopian tubes were 1 cm in length. Sections from right fallopian tube showed structure of fallopian tube along with focal areas of epididymis like structure in the wall (Fig 1). Left fallopian tube showed fibroconnective tissue lined by pseudostratified columnar epithelium which was like Vas deference (Fig 2).

Fig-1: Microphotograph shows focal areas of epididymis like structure in the wall of right fallopian tube. (HE, x100)

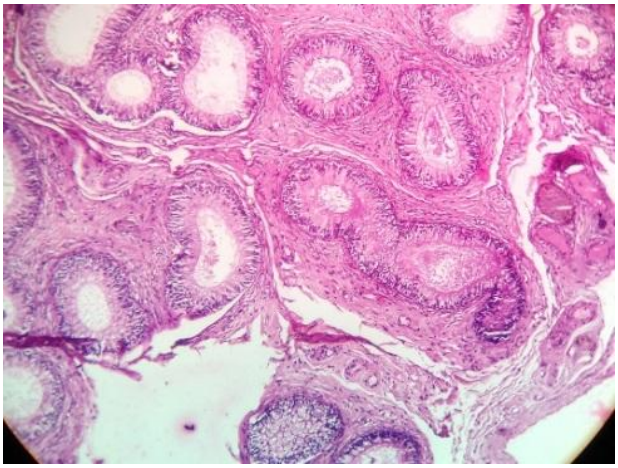

Fig-2: Microphotograph shows Vas deference like structure in the wall of right fallopian tube. (HE, x400)

Sections from uterine body showed lining of non-secretory endometrial glands (Fig 3) and sections from cervix showed endocervical lining and focal seminal vesicle like structure in the wall (Fig 4).

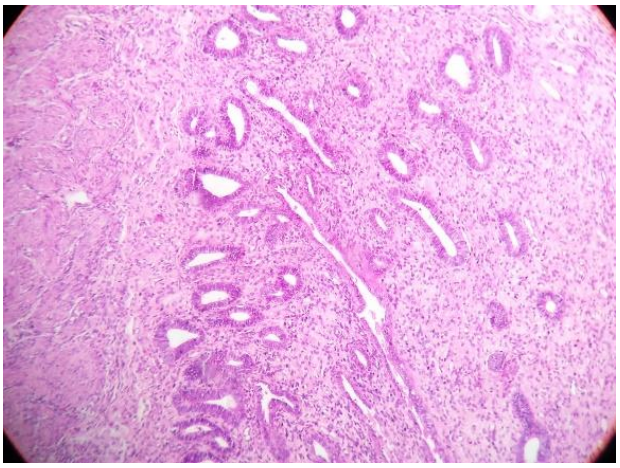

Fig-3: Microphotograph shows non secretory endometrial glands (HE, x100).

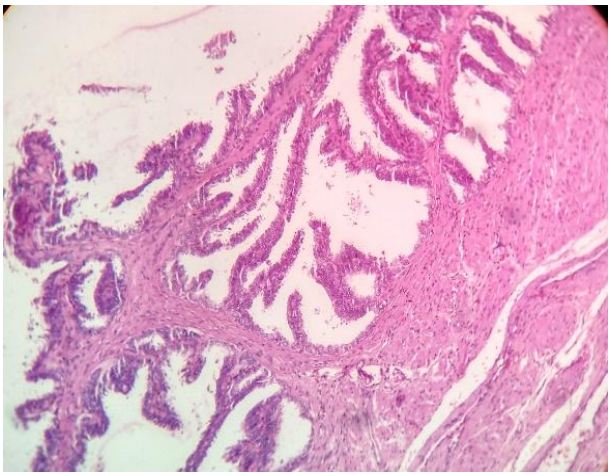

Fig-4: Microphotograph shows seminal vesicle like structure in cervix wall. (HE, x100)

DISCUSSION

Embryologically between 7th and 8th week of gestation, masculinization occurs in a male foetus. Testosterone secreted by the Leydig cells in the testis helps in development of Wolffian duct into Vas deferens, epididymis and seminal vesicle. Anti Mullerian hormone secreted by Sertoli cells in the testis acts locally to suppress the Mullerian ducts and causes their regression by 8th to 10th week of gestation [2]. (Fig 5).

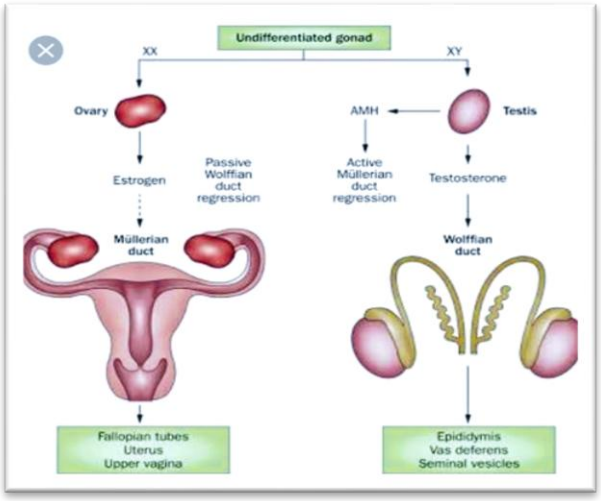

Fig-5: Differentiation of gonads in human. (Hutson, JM & Beasly, the Surgical Examination of Children 2013)

Persistent Mullerian duct syndrome is a rare form of internal male pseudo-hermaphroditism characterised by presence of Mullerian duct derivative in the genotype and phenotype of males with 46 XY karyotype. Due to deficiency of Anti Mullerian Hormone or defect in AMH type II receptor PMDS occurs [3].

PMDS can occur sporadically but most commonly, it occurs as autosomal recessive trait due to mutation in AMH and AMH II gene. Patient with type 1 PMDS have defect in AMH gene located at 19p13 which was seen in 45% cases. Patient with type II PMDS have AMH receptor located 12q13, seen in 40% cases and in other 16% cases causes are unknown [4].

PMDS patients present with male external genitalia with normal development of penis and scrotum but with cryptorchidism. There are three clinical presentation of PMDS- 1. Majority (60-70%) with bilateral intraabdominal testis in apposition analogous to ovaries. 2. Smaller group (20-30%) with one testis in the scrotum associated contralateral inguinal hernia whose contents are testis, uterus and tubes (classical presentation of hernia uteri inguinale). 3. Smallest group (10%) where both the testes are located in the same hernia sac along with the mullerian structures (transverse testicular ectopia-TTE)[5].

Treatment of PMDS is exclusively surgical and aims to correct cryptorchidism. The testes are usually histologically normal, though the germinal cells may be affected due to long standing cryptorchidism. The overall incidence of malignant transformation in these testes is 18%, similar to rate in abdominal testes in otherwise normal male. Early surgical intervention is necessary for testicular preservation, failing which gonadectomy must be offered to prevent malignancy. Adequate surgery involves bilateral orchidopexy[6].

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