International Journal of Science and Research (IJSR)
ISSN (Online): 2319-7064
Index Copernicus Value (2013): 6.14 | Impact Factor (2013): 4.438

Persistent Mullerian Duct Syndrome (Type 1) – A Rare Anomaly

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Abstract: ***Background: Persistent Mullerian duct syndrome (PMDS) is a rare form of internal male Pseudo-Hermaphroditism in which Mullerian Duct derivatives persists in a phenotypically & karyotypically male patient. It was associated with Asymmetric Gonadal dysgenesis(AGD). The exact incidence is not known but till date around 400 cases have been reported. AGD refers to individuals who usually have a differentiated gonad on one side and a streak gonad or testis on the other side. Case summary: A 27 year old male patient presented with cryptorchidism. On exploratory laparotomy uterus with bilateral adnexa were noted & Histopathology revealed features of AGD. The closest DD for AGD is True hermaphroditism. Conclusion: Persistent Mullerian duct syndrome (PMDS) is a rare disorder. It is important to diagnosis this entity early because 30% of cases progress to Gonadoblastomas. The very rare nature of this entity & grave prognosis merits its reporting.***

Keywords: Gonadal dysgenesis , True hermaphroditism, Gonadoblastomas, MIF, cryptorchidism

1. Introduction

Persistent Mullerian duct syndrome (PMDS) is a rare form of internal male Pseudo-Hermaphroditism in which Mullerian Duct derivatives (Uterus, Cervix, Fallopian tubes and Upper 2/3rd of vagina) are present in a phenotypically and karyotypically male patient[1]. It is usually associated with AGD a very rare entity. The exact incidence is not known but till date around 400 cases have been reported[2].

The PMDS Syndrome is caused either by insufficient amount of Mullerian inhibiting Factor (MIF) or due to insensitivity of the target organs to MIF. AGD refers to individuals who usually have a differentiated gonad on one side and a streak gonad or streak testis on other side. Few authors apply the term to patient who show testicular differentiation on either side , bilateral streak testis or bilateral dysgenetic testis[1] .

The diagnosis of PMDS & AGD is very important as it has important clinical implications for gender assignments and the decision for early gonadectomy can be taken as 30% of cases progress to gonadoblastomas [3].

The closest DD for PMDS with AGD is True hermaphroditism which can only be diagnosed by following the strict histopathological criteria necessary for diagnosis

2. Case Report

A 21 Year old married male patient presented with bilateral cryptorchidism and left inguinal Hernia. He had a male phenotype with well develop secondary sexual characters.

On Examination External genitalia were normal with empty scrotal sac & swelling in left scrotal region. Past history included primary infertility. USG revealed bilateral cryptorchidism and inguinal hernia. Semen analysis and Karyotyping were not done. On exploratory laparatomy uterus with bilateral fallopian tubes and ovary like structures were noted and was sent for histopathological examination.

Gross: Uterocervix measured 8x5x4cms,both tubes measured 4cms & each ovary 2.5x2x1cms

Microscopy: Endometrium was in proliferative phase and both side fallopian tubes showed normal histology. No ovarian stroma or primordial follicles were identified. Section studied through globular tissue attached to fallopian tubes show seminiferous tubules with immature sertoli cell and few primitive germ cell. Section studied through separately sent testicular biopsy shows incompletely spermatocytic maturation arrest. So based on above findings a diagnosis of PMDS with AGD was made.

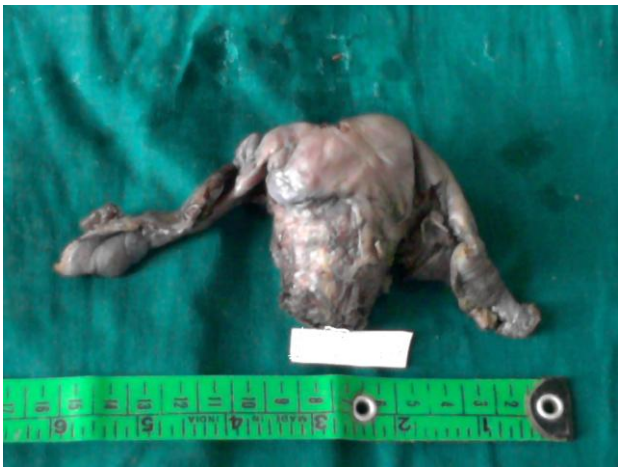

Figure 1: Utero-Cervix with bilateral Fallopian tubes & Ovaries

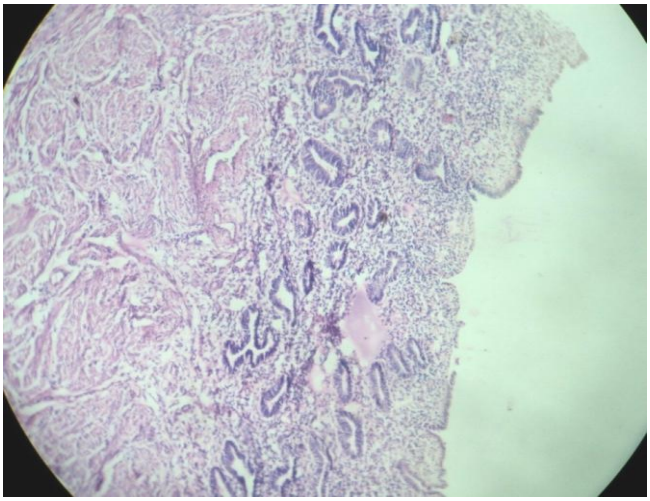

Figure 2: H & E; shows Proliferative Endometrium

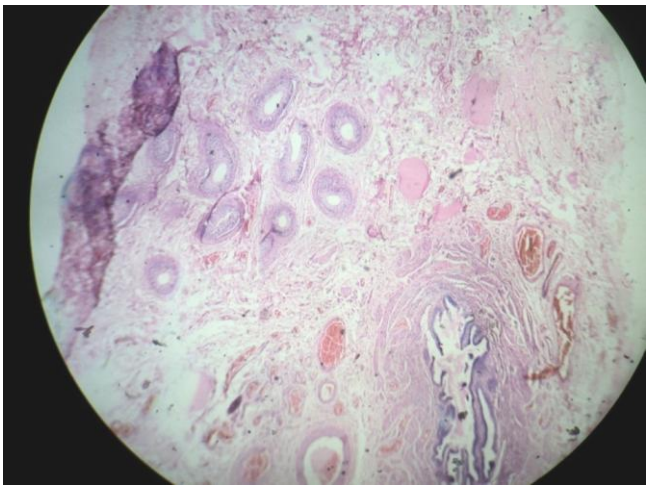

Figure 3: L P; H & E Fallopian tube & epididymis & vasdeferens

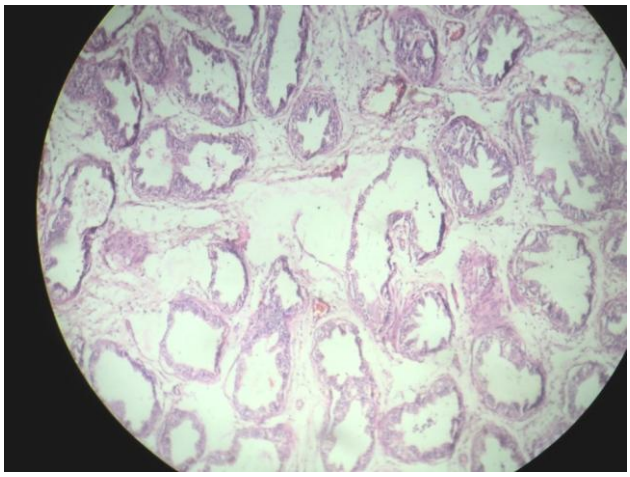

Figure 4: L P; H & E Seminiferous Tubule with Spermatocytic Arrest & immature Sertoli cells

3. Discussion

Children born with intersex disorder comprise about 1.7% of all live birth[4]. Ovo-testicular disorder of sexual differentiation or True hermaphroditism is a rare intersex disorder characterized by the presence of both ovarian tissue and testicular element regardless of their karyotype. The rarity is because most of the fetuses do not survive[5]. Persistent Mullerian duct syndrome (PMDS) is a rare form of internal male Pseudo-Hermaphroditism in which Mullerian duct Derivatives (Uterus, Cervix, Fallopian tubes and Upper 2/3rd of vagina) are present in a phenotypically and karyotypically male patient with normal secondary sexual characters[1]. It is due to deficiency or lack of MIF.

In human fetus the Mullerian & wolffian duct both are present at 7 week of gestation & in males by this time the testis differentiates. Normal sex differentiation is controlled by Testosterone, Dihydrotestosterone and MIF. Sertoli cells secretes MIF which leads to regression of mullerian ducts. Testosterone has a direct effect on Mullerian Duct and promotes differentiation of epididymis, vasdeferens, seminal vesicles. Dihydrotestosterone induces male development of external genitalia**.**

PMDS = WOLFFIAN + MULLERIAN ducts due to deficiency of MIFs[1].

PMDS :- has 2 anatomical Variants
Type 1(Males) & Type 2 (Females)
Type 1 is more common accounting for 80 – 90% of all cases. It again has two Subtypes, Type 1a & Type 1b

Type 1a = Hernia uteri inguinalis ie C/b descended testis and herniation of ipsilateral uterus & fallopian tube in the inguinal canal.

Type 1b = Crossed testicular ectopia C/b herniation of both the testis and the entire Uterus & both the fallopian tubes

Type 2 = C/b bilateral Cryptorchidism, with the testis fixed in the round ligament in an ovarian position.[1]
Our case presented with bilateral cryptorchidism and left inguinal Hernia. He had a Male Phenotype with empty scrotal sac & swelling in left inguinal region. This probably falls into Type 1 PMDS.

AGD is the term used if one gonad shows more complete development and can be identified as an ovary or testis (usually testis) and other gonad is streak[6]. This case was finally reported as PMDS with AGD. True hermaphroditism was the closest differential diagnosis and was excluded by following the strict histological criteria necessary for diagnosis.

The strict histological criteria necessary for diagnosis of true hermaphroditism is OVO-TESTIS i.e well define testicular elements like solid seminiferous tubules with immature sertoli cells and few primitive germs cell and ovarian stroma comprised of numerous primordial or mature follicles containing primary oocytes [5,6]

It is important to differentiate the two condition because it is necessary for gender assignment and in PMDS with AGD 30% patients develop germ cell tumors like dysgerminoma, yolk sac tumor, embryonal carcinoma and hence gonadectomy is necessary[7,8]

The histological examination in this case reveals testicular element with seminiferous tubules and immature sertoli cells

and endometrium in proliferative phase with structure of both fallopian tubes. The diagnosis of PMDS , AGD , Pseudohermaphroditism & true hermaphroditism requires detailed Clinical, Radiological & Histopathological Examination.

4. Conclusion

PMDS with AGD is rare phenomenon and it is important to diagnose this entity because 30% of cases progress to Gonadoblastomas.

The very rare nature of this entity & its Dismal Prognosis merits its reporting.

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