Persistent Mullerian Duct Syndrome

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Abstracts: Persistent mullerian duct syndrome is a rare form of internal male pseudohermaphroditism in which mullerian duct derivatives are present in a genotypic (46XY) and phenotypic male. We report a rare case of persistent mullerian duct syndrome in an adult fertile male in whom mullerian duct derivatives (uterus and fallopian tubes) were found in the right hernia sac at herniotomy. [Kaore A et al NJIRM 2012; 3(1) : 153-154]

Key Words: Persistent mullerian duct syndrome, male pseudohermaphroditism, male.

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Introduction: Persistent mullerian duct syndrome (PMDS) is a rare form of internal male pseudohermaphroditism in which mullerian duct derivatives (uterus and fallopian tubes) are present in a genotypic (46XY) and phenotypic mal[1]. It occurs due to failure of regression of mullerian ductal system in genotypically normal males[2,3]. This was first described by Nilson in 1940 as "hernia uteri inguinales" or " internal male pseudohermaphroditism"[4]. Only about 150 cases have been reported in literature mostly in adults and mainly from Western, European and Middle Eastern settings. PMDS patients are usually infertile and fertility is rare in then. This article presents rare case of PMDS in a fertile adult male.

Case report : A 35 years old married fertile male having children presented with right inguinal hernia. Both the testes were in normal position and were of normal size. At herniotomy rudimentary uterus and fallopian tubes were found in the hernia sac on right side. Uterus was attached by bands of fibers tissue to both the testes. These structures were removed along with both the testes and were sent for histopathological examination.

Gross Examination of the Specimen: Specimen received showed rudimentary uterus with two fallopian tubes. One fallopian tube was shorter than the other. The longer tube had a thin pedicle at the end with pea size cystic structure attached to it representing ovary. The uterus was attached by bands of fibrous tissue to both the testes (photograph 1).

Cut section of uterus showed a cavity extending from upper pole to lower pole. The inner surface of the cavity was smooth. No distinct cervix and vagina were seen .

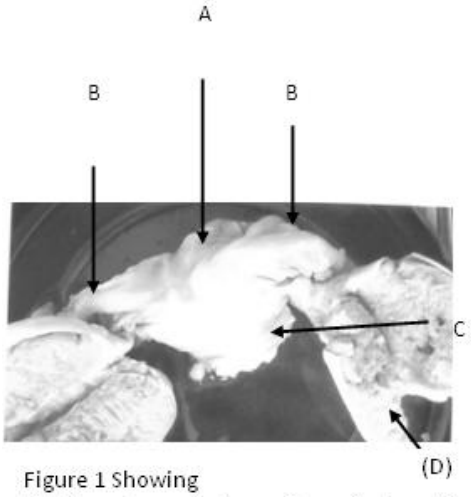

Figure 1 Showing
(A) Showing posterior surface of uterus like structure
(B) With 2 cords distinctly arising from upper pole (fundal region)
(D) Testes attached with uterus by
(C) Fibrous band.

Histopathological Study:

Uterus : Sections from upper pole (fundus), middle part and lower pole (cervical end): Similar histopathological picture is seen in all the three sections. The sections show outer tissue composed of smooth muscle and fibrocollagenous tissue with blood vessels (myometrium). The inner lining tissue shows glands of varying size lined with epithelial cells. These glands are inactive and are surrounded by stromal tissue (Endometrium) .

Fallopian tubes: Sections from both the fallopian tubes show irregular small lumen lined with columner epithelium and surrounded by thick fibro collagenous tissue and blood vessels .

Pea size tissue with pedicle (ovary):The section shows cystic areas surrounding cellular tissue. The tissue shows distinct nuclei with abundant cytoplasm (granulose cells) while adjacent tissue shows theca cell like structure. It is surrounded by fibrocollagen tissue and blood vessels .

Testes:Sections from both the testes show normal testicular tissue.

Discussion : PMDS is a rare form of internal male pseudohermaphroditism caused by deficiency of mullerian inhibiting factor (MIF)[5]. PMDS patients are both genotypically (46XY) and phenotypically male.

In a human fetus both mullerian ducts and wolffian ducts are present at seven weeks of gestation. Mullerian ducts and wolffian ducts are the anlagen of the female and male reproductive tracts respectively.

In the XY fetus the testes differentiates by the end of the seventh week of gestation. Sertoli cells begin to secrete MIF which is responsible for regression of the mullerian ducts in male fetus. In the absence of MIF the mullerian duct differentiates into fallopian tubes, uterus and upper vagina.

PMDS is caused by absence are deficiency of MIF before the eighth week of fetal life or abnormality in its receptor[6]. Since secretion and action of testosterone is not affected the Wolffian duct derivatives and external genitalia of the fetus develop in a normal male direction. An intersex condition is therefore usually not suspected but is incidentally detected during operative treatment such as for inguinal hernia as is the case in present patient.

Infertility is usual in these patients. However there are few reported cases of fertility .The patient is married and is having two children but absolute proof of paternity could not be established in our patient although histopathological examination of both the testes is normal.

In PMDS patients the prevalence of gonadal malignant transformation is very high[7]. Hence bilateral orchidectomy was done in the present case even though testes were intrascrotal.

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