

in this group occur after puberty, long term followup is required.

PMDS needs to be differentiated from mixed gonadal dysgenesis. In addition of having Mullerian structures, mixed gonadal dysgenesis is characterized by ambiguous genitalia, a testis is present unilaterally and gonadal streak on contralateral side¹³.

Pre-operative diagnosis is not possible, the diagnosis is made incidently during herniorrhaphy, during exploration for cryptorchidism or for tumour arising from undescended testis as in our case. The treatment for persistent mullerian structures is controversial, some clinician recommended that they should be removed because leaving behind uterus and vagina which is often communicated with the urethra can lead to recurrent urinary tract infection¹⁴. Edward, Tark and David have reported two cases of bladder outlet obstruction caused by massive dilatation of persistent mullerian duct remnants²⁰. While most of the surgeons recommended that the mullerian structures should not be removed because their close proximity to the vas might lead to their destruction during removal. By this, means any possible fertility should be preserved⁸.

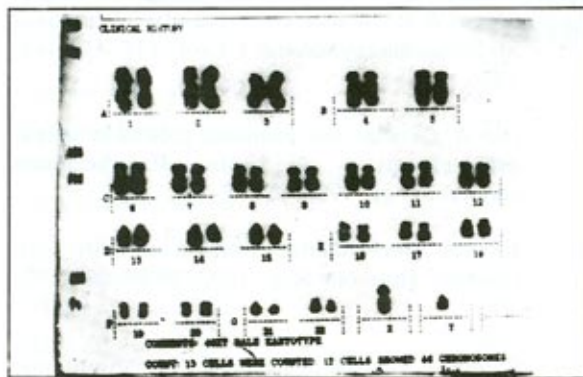
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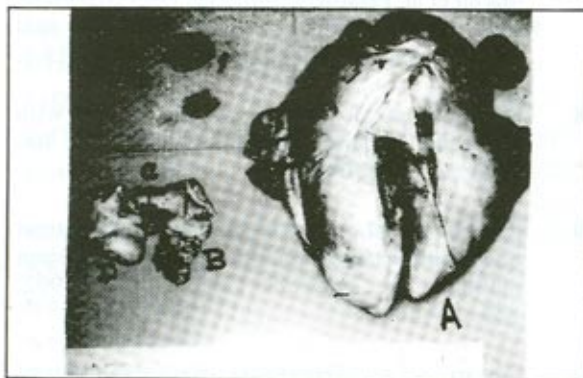
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Chromosomal analysis showing 46XY male karyotype

The patient subsequently underwent an explorative laparotomy. The findings were a large lobulated well encapsulated suprapubic mass and normal looking right testis which was attached to midline structures between bladder and rectum resembling uterus with fallopian tube. The large mass was not connected with the rudimentary uterus (Fig.4) The large suprapubic mass and right testis with attached uterus were surgically removed and the histopathology showed.

1. Mixed germ cell tumour arising from left undescended testis.
2. Bilateral vas and fallopian tube like structures present
3. Right undescended testis with sertoli cells only
4. Uterus and cervix were identified. The uterus measures 6 x 4 x 2.5 cms. The uterine cavity was small measuring 3.5 cms. The uterine and cervical cavity reveal endometrial and endocervical cells lining respectively.



Surgical specimen of a large tumor arising from left testis (A) and a rudimentary uterus, vas & fallopian tube (B) and testis (D) on the opposite side.

Discussion

Persistent Mullerian duct syndrome (PMDS) also known as hernia uteri inguinale and internal male Pseudohermaphroditism³ is a rare disorder of sexual differentiation characterized by the retention of Mullerian derivatives in patients otherwise normally virilized.

Since the original report of the syndrome by Nelson 1939⁴ about 160 cases have been reported including 5 from Pakistan^{5,6}. Most refer to isolated cases while few involved siblings^{7,8}. The view that PMDS is hereditary is supported by the familial occurrence in a few of the reported cases.

The regression of the Mullerian ducts is by the action of Mullerian inhibiting substance first described by Jost in 1953⁹, a glycoprotein with molecular weight of 140,000 - 200,000 D.

The MIF is secreted by fetal sertoli cells acts locally and ipsilaterally to cause regression of mullerian ducts. Its action is independent of testosterone which causes the Wolffian ducts to differentiate. Recently the MIS gene has been localized to the short arm of chromosome 19¹², but the location of the gene for the MIS receptors remains unknown.

In PMDS, the MIS is (1) not produced also known as AMH-negative forms, a mutation of the AMH gene is thought to have been responsible for the condition.^{10,11} (2) Produced in insufficient quantity (3) Defective (4) Produced after the critical period for differentiation (8th week of gestation^{13,14,15}) (5) Produced normally but the mullerian ducts exhibit varying degrees of resistance to it. This syndrome may be associated with transverse testicular ectopia¹⁶, hirschsprung's disease¹⁷ and splenic gonadal fusion¹⁸.

PMDS is characteristically associated with cryptorchidism and often with inguinal hernia. Like other undescended testis, these gonads are at high risk of malignant transformation. The overall incidence of malignant transformation in these gonads is 15% (Seminoma 46%, embryonal Ca 15%, teratoma 15%, Yolk Sac 8%, Choriocarcinoma 8%, Mixed germ cell 8%)¹⁹. which is similar to the rate of abdominal testes in normal men, so the absence of mullerian inhibiting substance does not appear to increase the relative risk of testicular malignancy and so far malignancy arising from Mullerian structures of PMDS patients have not been reported.

To our best of knowledge, we report the second case of a mixed germ cell tumour in a patient with PMDS. The first was reported by James A. Eastham in 1992¹⁹. The incidence of malignancy is same as other cryptorchid testes. Preservation of virilization can be accomplished by orchidopexy. Even if it is performed early in life, does not decrease the risk of malignancy. Since most tumours

PERSISTENT MULLERIAN DUCT SYNDROME WITH MIXED GERM CELL TUMOUR

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Abstract

Persistent Mullerian Duct syndrome (PMDS) is a rare disorder of sexual development, it is characteristically associated with undescended testes. Testicular tumours are not uncommon in this group of patients. We report a case of PMDS with mixed germ cell tumour in the left undescended testis.

Key Words : Persistent Mullerian Duct Syndrome, Mullerian Duct, Cryptorchidism.

Introduction

PMDS is a rare disorder of sexual development characterized by normal virilization of the external genitalia but persistence of mullerian duct structures (Fallopian tubes, uterus and upper vagina). Mullerian inhibiting substance (M.I.S.) or anti-mullerian hormone is responsible for the regression of fetal mullerian structures. These patients are genotypic (46 XY) and phenotypic males with undescended testes. Like other undescended testes, these gonads are at an increased risk of malignant transformation.

Case Report

A 25 year old man presented to our consulting clinic with complaints of vague lower abdominal pain



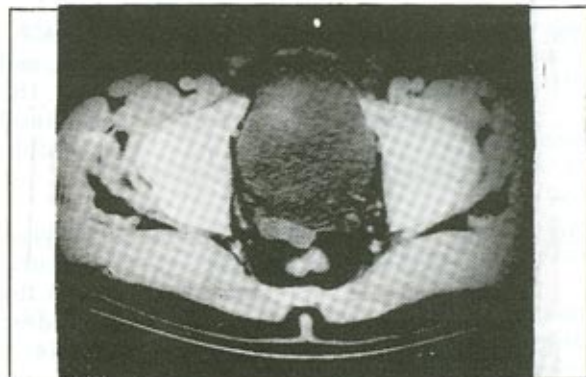
The hypoplastic scrotum and extent of the abdominal mass is marked on skin

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and primary infertility. He was married for 11/2 years and had no problem with his sexual relationship. On clinical examination the patient was a well built man with normal secondary male sexual characters. On abdominal examination he had a large mass palpable in the suprapubic region extending upto umbilicus. The mass was hard, non-tender with well demarcated upper and lateral margins and mobile in a transverse plane. His scrotum was hypoplastic and testes were not palpable, penis was of normal size and appearance. (Fig.1).

Clinical diagnosis was a tumour arising from an undescended testes. Investigations revealed serum testosterone 826 mcg% (270-1070). Serum B-HCG 256 mlu/ml (0-5). Serum a - fetoprotein 349 mcg/ml (0-10). These were suggestive of testicular tumour other than pure seminoma or pure choriocarcinoma.

Ultrasound of the lower abdomen showed a large 13 x 9 cms solid mass sitting at the roof of the bladder and extending upto the umbilicus. The echogenicity was very heterogeneous. It was well circumscribed and not invading the bladder. CT scan showed a round large 10 cms well defined heterogeneous mass in the lower abdomen. (Fig.2) The contrast scan showed irregular areas of contrast enhancement. Fascial planes in the lower abdomen and pelvis were intact. The urinary bladder was separated from the mass. Pelvic and para aortic lymph nodes were not enlarged. A normal sized right testis was noted in abdominal cavity, measuring 4x3 cm. (Fig.2). Genotyping showed normal male pattern (46 XY). (Fig.3).



CT Scan showing a well defined heterogenous mass in the lower abdomen.