PERSISTENT MULLERIAN DUCT SYNDROME - A CASE REPORT

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ABSTRACT

An eighteen month old phenotypically and genotypically normal male child was admitted with a left inguinal hernia and a right undescended testis. At operation, he was found to have a uterus, bilateral fallopian tubes, and a vagina in the left hernial sac. Bilateral orchidopexies and excision of the persistent Mullerian duct structures were carried out. This rare case of persistent Mullerian duct syndrome is due to a defect in Mullerian regression, which is in turn controlled by the Mullerian inhibiting substance (MIS). Orchidopexy with excision of the persistent Mullerian duct structures is usually not possible without damage to the vas deferens which is closely adherent to the wall of the uterus. The alternative of leaving the persistent Mullerian duct structures alone and performing a staged or primary orchidopexy has been suggested.

Keywords: Persistent Mullerian duct syndrome, male pseudo-hermaphroditism, Mullerian inhibiting substance

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INTRODUCTION

The paramesonephric or Mullerian duct first appears in the human embryo at the 10 mm stage of development, and at the 30 - 32 mm stage, begins to regress in the male as a result of a glycoprotein secreted by the developing testes known as Mullerian inhibiting substance (MIS)[1]. Incomplete obliteration of the caudal part of this ductal system can give rise to cysts lying in the midline between the bladder and rectum, and these may communicate with the posterior urethra[2]. Apart from these disorders of regression, there is also a well-documented syndrome known as the persistent Mullerian duct syndrome which is due to the failure of regression of the Mullerian ductal system. This was first described by Nilson in 1940 as "hernia uteri inguinales" or "internal male pseudohermaphroditism"[3]. The classical presentation is a patient who is phenotypically and genotypically a male, with unilateral cryptorchidism and a contralateral hernia which contains Mullerian duct structures such as the uterus, fallopian tubes and vagina. This anomaly is attributed to lack of or impaired action of Mullerian inhibiting substance.

The rarity of this syndrome in surgical practice gives rise to problems regarding the recognition of this syndrome, and the course of action to be taken.

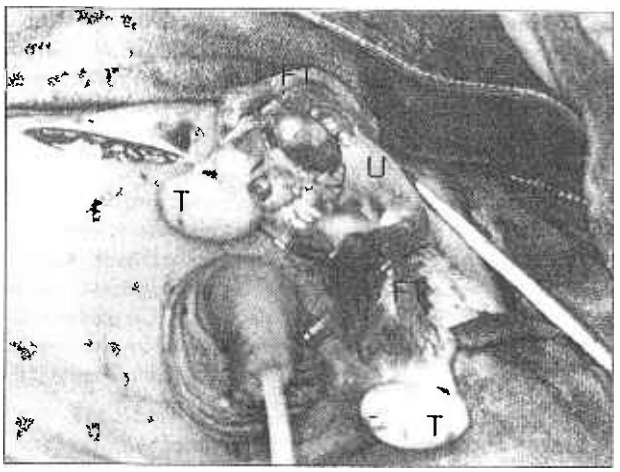
CASE REPORT

AK was first seen as a new-born with bilateral undescended testes. He had a normal penis and scrotum. Chromosome studies showed a normally banded 46,XY karyotype.

On follow-up over the next few months, the left testis was felt in the scrotum, but this was associated with a left inguinal hernia. The right testis remained undescended. At the age of 18 months, he was admitted for surgery. The pre-operative diagnosis was a left inguinal hernia with a right undescended testis.

At operation, the right inguinal canal was explored first, but the testis could not be found. The left inguinal canal was then explored. The hernial sac was opened, and in it was found a uterus with bilateral fallopian tubes arising from it (Fig 1). At the end of each fallopian tube was a gonad, one of which was in the left scrotum, and the other one in the inguinal canal. The epididymis were rudimentary, and the vas deferens could not be identified in the spermatic cord. At the other end of the uterus was a vagina. There were no communications with the urethra or bladder. Bilateral gonadal biopsies were done, and the gonads were replaced in both scrotal sacs. The uterus, vagina and tubes were removed.

Fig 1 – Photograph Taken During Operation Showing T = Testis, U = Uterus, FT = Fallopian Tube



Histopathological examination confirmed the presence of an immature but fully formed uterus, and a single fallopian tube. The other tubular structure thought to be a fallopian tube was actually a vas deferens and epididymis. The testicular biopsies showed immature testes with no ovarian elements.

Post-operative recovery was uneventful. The intravenous urogram and micturating cystourogram did not show any abnormalities. Serum luteinising hormone, follicle stimulating hormone, testosterone and oestrogen studies were normal.

This child is now 4 years old, and both testes are in the scrotum, and of normal size.

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DISCUSSION

An understanding of normal embryogenesis helps to explain the aetiology of this disorder[4]. Differentiation of an indifferent gonad into a testis or ovary depends on the sex chromosomes. The testis produces two principal hormones, Mullerian inhibiting substance (MIS) and testosterone. MIS causes regression of the Mullerian ducts, which would otherwise develop into the uterus, fallopian tubes and upper vagina. Testosterone induces the development of the internal male genital structures, that is, the vas deferens, seminal vesicles, and epididymis. When metabolised to 5-alpha-dihydrotestosterone, it stimulates the development of the prostate, glans, penis and scrotum. By contrast, female development is an autonomous process, and does not require the presence of either gonad or hormone. Male pseudohermaphroditism results from defective formation or action of androgen or MIS.

The male pseudohermaphrodite has male gonads and karyotype but varying degrees of virilisation of the internal and external genital tracts. Persistent Mullerian duct syndrome (PMDS) represents a small fraction of this broad spectrum of male pseudohermaphroditism.

It is believed that PMDS is caused by a defect in the secretion or action of MIS[5]. There is also a possibility that the MIS is normally produced, but the Mullerian ducts exhibit varying resistance to it. The fact that testicular descent is often impaired in this condition gives rise to a postulation that MIS may also be involved in the descent of the testes. Measurement of the production of MIS in infants with undescended testes suggests that it is lower than normal, although this could be the result rather than the cause of the maldescent[6].

Pre-operative diagnosis of PMDS is often impossible because of the normally developed penis and scrotum[7]. Diagnosis is usually made during operation for inguinal hernia or cryptochidism, as in our case.

Of the more than 80 previous reports of PMDS, the majority have been isolated cases, but a few cases of siblings have been described[8,9]. Infertility is usual in these cases. However, there are 6 reported cases of fertility in males[10,11] but paternity was not satisfactorily documented. The fact that fertility may be present causes a dilemma in the surgical management[7]. Orchidopexy, with the removal of the Mullerian duct structures is usually not possible without damage to the vas deferens. Leaving the uterus and tubes behind is not a problem because malignancy in these structures have not been reported. However, leaving the testis in the abdominal cavity in order to avoid damage to the vas seems pointless from the point of view of fertility, and leaving a testis in the abdominal cavity predisposes to malignancy.

Based on these considerations, the logical management would be to do an orchidopexy and perform a total hysterectomy, even though this requires severing the vas and sacrificing potential fertility; the development of malignancy in an intrascrotal testis is more easily detected and managed[7]. However, other groups[8] have advocated performing a staged or primary orchidopexy and leaving the uterus, tubes, or vagina alone to preserve any possible fertility.

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