# CASE PRESENTATION

## Persistance of Remnants of the Mullerian Duct

By Mazin A. Ismail*

**ABSTRACT**

**We report four cases where persistant structures of the Mullerian duct were found in male patients, and discuss the aetiology, clinical presentation, complications and management of this condition.**

Persistance of remnants of the Mullerian duct is a rare congenital entity in the male. The patient is essentially a phenotypically and karyotypically normal male with normally developed secondary male sexual characteristics.

The condition was first described by Nilson as "Hernia uteria inguinalis," which presented as a unilateral cryptorchidism with a contralateral inguinal hernia that contained a rudimentary uterus and Fallopian tubes.[1]

We report four cases which have been diagnosed at St. Vincent's Hospital, Dublin, and the Children (Temple Street) Hospital, Dublin during the period 1973 – 1983.

**CASE No. 1**

A 3-months old boy presented with bilateral cryptorchidism and hypospadius. He was found to have 45XY chromosomal configuration in most of the cells examined while a buccal mucosal smear showed the absence of "Barr bodies" in the cells, suggestive of a male configuration.

At the age of 4 years, the child's groin was explored, the findings were a uterus-like structure with two Fallopian tubes and a single right-sided gonad which was proved histologically to be a testis. The testis was fixed in the right inguinal canal while the uterus and tubes were excised.

An endocrine follow up of the patient was intended to commence at the age of eight years as the child was too young for endocrine assessment at the time of the surgery.

**CASE No. 2**

A 9-months old boy presented with bilateral cryptorchidism, a rudimentary scrotum and a small penis lying between two "Pseudolabial" folds. A Karyotype study showed 45XY configuration while a buccal smear confirmed the absence of "Baar bodies" in the cells, indicating a male chromosomal configuration.

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Sydney Parade Avenue
Sandymount
Dublin 4, Eire

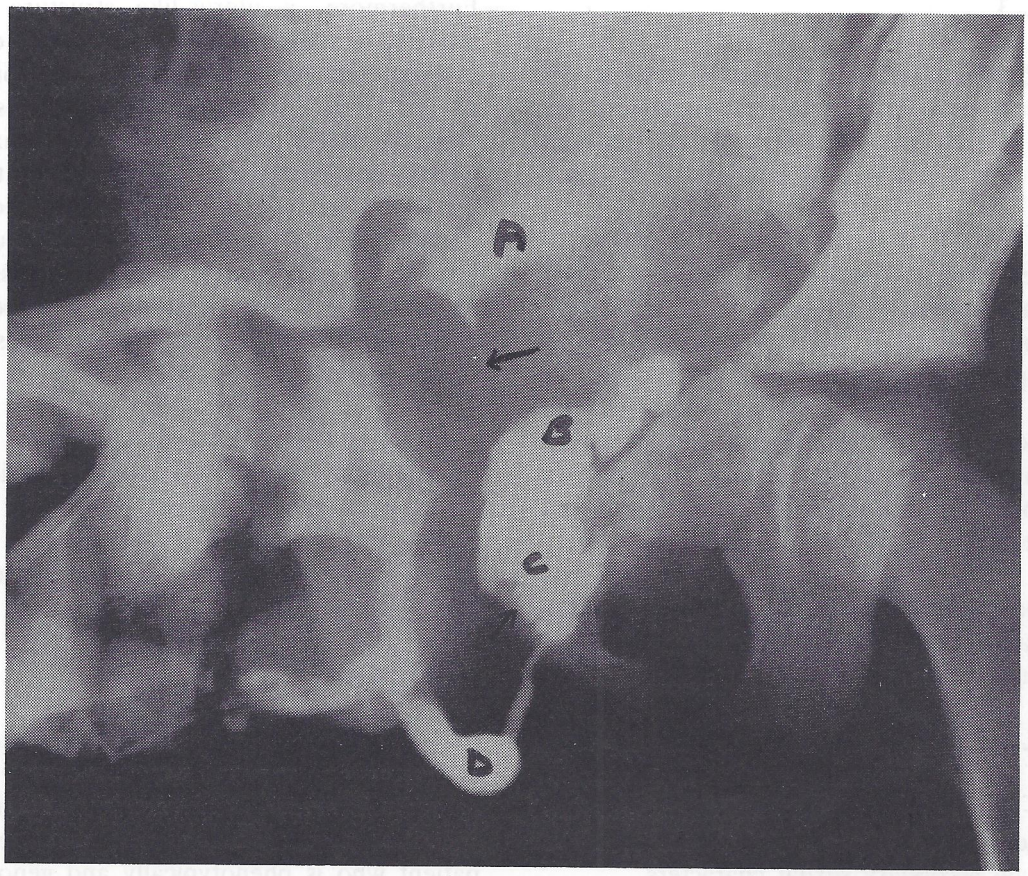


**Figure 1, (Case 2)**

Ascending Cystourethrogram in case 2, showing the fistula between the urethra and the "vagina" and the fistula between the bladder and the "uterus":

A– Bladder.
B– Uterus.
C– Blind-ending vagina.
D– Urethra.

An ascending cystourethrogram showed what appeared to be a "Vagina" communicating with the urethra via a fistula. This vagina also opened into a uterus-like structure that in turn communicated via another fistula with the bladder (Fig. 1).

Surgical exploration was postponed to the age of 8 years and at laparotomy a uterus full of urine was seen communicating with the bladder via a vagina. Also, a pair of gonads were seen on either side of the uterus. One of the gonads was attached to the end of a Fallopian tube while the other was attached to the bladder by a "vas deferens" like structure (Fig. 2). The two gonads were preserved while the vagina, uterus and tubes were excised.

An endocrine assessment was started post-operatively and repeated at yearly intervals. This showed no evidence of hypogonadism.

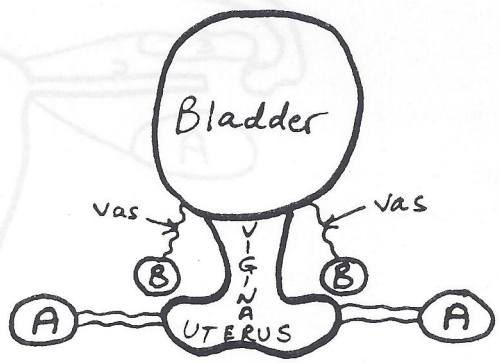


**Figure 2, (Case 2)**

Findings at laparotomy in case 2:
A– Gonad.
B– Gonad.
C– Fallopian tube.

**CASE No. 3**

A 3½-weeks old boy presented with a left undescended testis lying in the left inguinal canal and could not be brought down into the scrotum while the right testis was normally located in the scrotum.

The left groin was explored at the age of 4 years when it was found that there was only a "peritoneal tube" lying in the left inguinal canal, which on further exploration intraperitoneally was found to extend to the base of the bladder. There was no trace of the missing left testis anywhere. On histological examination, this peritoneal tube was found to be composed of a rudimentary uterus with no fallopian tube or ovary.

Subsequently, a Karyotype study revealed a normal male configuration (45XY), while a buccal mucosal smear showed no "Barr bodies."

**CASE No. 4**

A 21-years old male presented with bilateral cryptorchidism. He had a normal-sized penis and normal male secondary sexual characters.

On laparotomy, two gonads were found intraperitoneally, attached to the posterior abdominal wall and could not be mobilised to the scrotum. Furthermore, a uterus-like structure was seen attached to a blind-ending vagina, a pair of fallopian tubes extending from the uterus to the gonads, and a pair of "Round ligament" like structures extending from the uterus and entering the inguinal canal on either side. All these structures and the two gonads were exicsed. Histological examination of the gonads showed atrophic testicular tissue in both gonads with no trace of ovarian tissue, while examination of what was believed to be a pair of Fallopian tubes revealed a "vas deferens" attached to an epididymis (Fig. 3).

Subsequently, a Karyotype study revealed a normal male (46XY) configuration, while a buccal smear showed no Barr bodies.

The patient received Testosterone supplements and was followed up periodically.

**DISCUSSION**

Persistance of Mullerian Duct Structures is a congenital anomaly in which remnants of the Mullerian duct (uterus, fallopian tubes, vagina, round ligaments, broad ligaments) are preserved in a patient who is phenotypically and genotypically a male with normal male secondary sexual characters.

Typically, the patient presents with unilateral, or more commonly bilateral, cryptorchidism with or

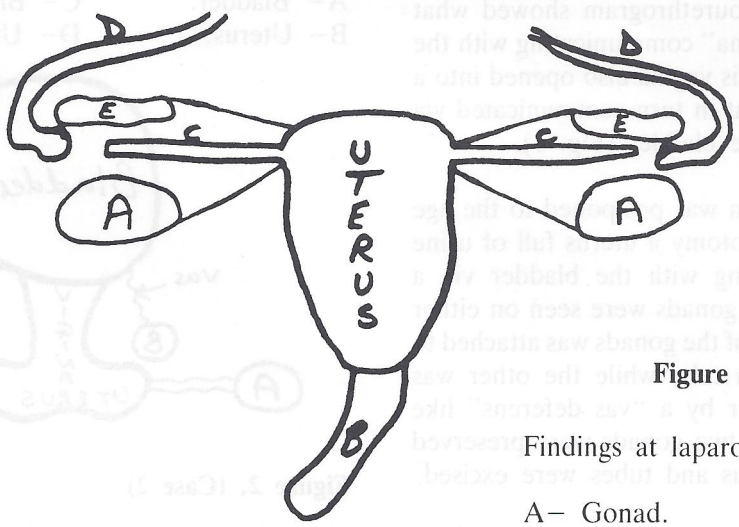


**Figure 3, (Case 4)**

Findings at laparotomy in case 4:

A– Gonad.
B– Blind-ending vagina.
C– Fallopian tube.
D– "Broad ligament" – like structure.
E– Epididymis.

without an indirect inguinal hernia which may contain some Mullerian duct remnants. More likely, such structures remain intra-abdominally in which case the uterus opens into a blind-ended vagina, and the undescended testis-or testes-lie at the fimbrial end of the fallopian tubes, the usual site of the ovary.

The aetiology of this condition is related to an abnormality of the "Mullerian Regression Factor" secreted by the Sertoli cells of the testis. This abnormality could either be in the form of deficient synthesis of the factor by the cells of the undescended testis, or a defective function of that factor – although it is sufficiently produced – resulting in failure to accomplish complete regression of the Mullerian structures. Failure of regression could also be due to the resistance of the Mullerian structures to the action of the Mullerian Regression Factor or to delayed release of the factor after the critical period of differentiation.[2,3]

Complications associated with persistance of Mullerian duct structures include:

a) Infertility with azoospermia, but there have been a few reports of fertility among these patients although paternity has not been established in any of these cases.[2]

b) Testicular neoplasia including seminoma, teratoma, embryonal carcinoma and yalk sac carcinoma.[4,5]

c) Development of retention cysts within the Mullerian remnants. These may become infected[6] or exert pressure on adjacent organs as in the case of the bladder causing outflow obstruction.[7]

d) Urinary incontinence due to pooling of urine in the Mullerian remnants which communicate with the bladder, thus acting as a bladder diverticulum.

Persistant Mullerian duct syndrome is difficult to diagnose pre-operatively because of the normally developed external genitalia but is mostly diagnosed after surgical exploration for cryptorchidism or inguinal hernia repair.[2] However, if a suspicion of "Intersex" is entertained, on the basis of cryptorchidism, chromosomal studies (in the form of a karyotype study showing a normal male 46XY configuration, and a buccal smear confirming the absence of Barr bodies in the cells) are highly suggestive.

Lower abdominal ultrasonography is recommended to detect the rudimentary Mullerian structures, localising the missing intra-abdominal gonad(s), and demonstrating retention cysts within these structures.

Intravenous urography has proved inaccurate in demonstrating the Mullerian remnants particularly when they do not communicate with the urinary system or are not large enough and in a position to cause bladder compression or outflow obstruction. On the other hand, voiding cysto-urethrography has proved as the most accurate diagnostic procedure in demonstrating any communication between the Mullerian structures and the lower urinary tract.[6]

Laparascopy is a technique most useful in detecting Mullerian remnants where ultrasonography has failed and CT-scanning is not available, as well as to obtain a biopsy from the intra-abdominal gonad(s) to prove their histological nature, and lastly to assess the level of descent of the gonad(s) and decide on the appropriate procedure required to mobilise the testis down to the scrotum (i.e. single or two-stage orchidopexy or microvascular anastomotic technique).[8] The use of laparoscopy is, however, not recommended below the age of 3 years due to the discrepancy between the size of the abdominal cavity and the instrument.[8]

Finally, assessment of sex hormones in the serum invariably shows elivated FSH and LH levels as a response to the markedly repressed or completely absent spermatogenesis while serum testosterone levels remain normal since the endocrine function of the cryptorchid testis(es) is not altered.

The surgical management of this condition is controversial. Many authorities advocate retaining the Mullerian remnants, if asymptomatic, to avoid irreparable damage to the vasa-differentia which run close to the ectopic uterus and vagina[3,6] thus sacrificing any potential fertility. Others insist that all Mullerian remnants must be removed because of the associated complications, particularly recurrent urinary infection when these remnants communicate with the urinary tract.[2]

Furthermore, surgeons are still divided as to

what constitutes the optimal surgical treatment for the intra-abdominal gonads. Some advocate their removal even if they have been proven to be histologically normal so as to avoid the risk of malignant change.[2] Others, however, maintain that as long as there is no histological evidence of noeplasia, every effort should be made to mobilise them to the scrotum.[8] Such cases should then be monitored periodically for neoplastic changes using annual estimation of serum markers for testicular tumours such as B-HCG and Alfafoetoprotein.[9]

In the rare event of a unilateral maldescended testis co-existing with Mullerian remnants, while the contralateral testis is normally descended, the descent of the maldescended testis should be facilitated by surgical means, but no attempt should be made at excising it unless it shows clear histological evidence of neoplasia.[8] If neoplasia develops later in either testis, they will be at a more accessable position for management.[2]

After surgery, the patient should be followed up with endocrine assessment to determine the need for androgen supplements. This is necessary for all cases where both cryptorchid testes have been removed, so as to enhance the growth of the penis and maintain the normal developement of the male secondary sexual characters.

## CONCLUSION

**Persistance of remnants of the Mullerian system in the phenotypicly and karyotypicly normal male is related to failure of synthesis or function of the Mullerian Regression Factor which may also account for the variable degree of testicular maldescent commonly associated with this syndrome. The condition must be excluded whenever unilateral or bilateral cryptorchidism is encountered. Prompt steps must be taken to ensure early descent or removal of the maldescended gonad(s), with or without excision of the Mullerian structures and are necessary to avoid the complications of testicular maldescent.**

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