Persistent Müllerian Ducts Syndrome: A Case Report

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Abstract

Persistent mullerian duct syndrome (PMDS) is usually an accidental finding during routine inguinal hernia repair in male patients. Intraoperatively, mullerian remnants consisting of an infantile uterus and fallopian tubes are usually found. Herein we report a case of PMDS in a 3-year-old boy presenting with unilateral undescended testes.The right testis was not palpable and the diagnostic ultrasound shows a non-visualized right testis neither in scrotum nor in the right inguinal region.

Keywords - *Persistent mullerian duct syndrome, Hernia uteri inguinale, anti-mullerian hormone, cryptorchidism.*

I. INTRODUCTION

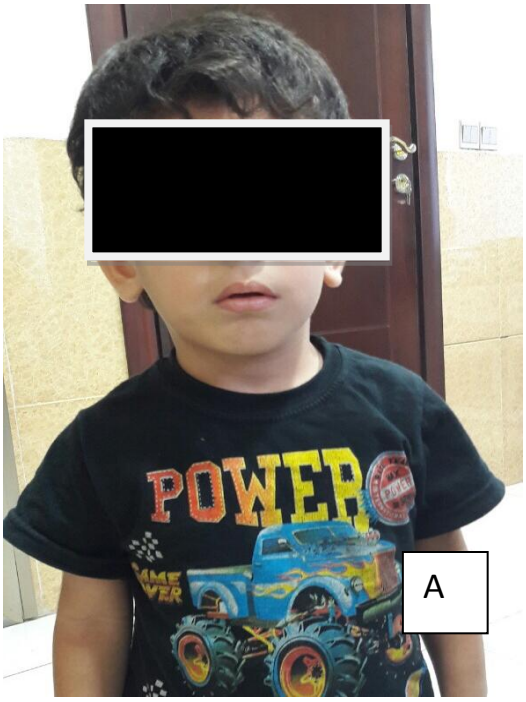
Persistent Mullerian duct syndrome (PMDS) is a rare disorder of sexual reproductive system that affects males; however, the prevalence of the condition is yet unknown. Males with PMDS have normal phenotyping consisting of normal male external genitalia as well as a uterus and fallopian tubes.

The uterus and fallopian tubes during emberyonal life develop from the Müllerian duct, which is present in both male and female and normally in males it becomes artretic but it remains in those with PMDS. The main signs of PMDS may include (cryptorchidism) or inguinal hernia.

Here we report a case of PMDS who presented with inguinal hernia and the hernia content were sent to histopathology and was incidentally found to be a case of PMDS

II. CASE REPORT

A three year-old toddler was brought to our Shar hospital in Sulaimani city in Iraq for evaluation of unilateral cryptorchidism that was phenotypically male(Figure-1). Informed consent was obtained from the parents of the child to publish the case. Clinical findings: The right testis was impalpable and a right inguinal hernia was present.



Diagnostic Ultrasound shows a non-visualized right testis neither in scrotum nor in the right inguinal region with normal size and texture of the left testis and epididymis with no obvious hydrocele nor varicocele.
During herniotomy and right sided orchidopexy a thick cord like structure was found in the right inguinal region and was sent for histopathologicalexamination.
Pathological examination:Grossly, A three centimeters tubular structure was received, sliced and all was submitted for histopathological evaluation.
Microscopical finding: An infantile uterus comprising of normal-looking endometrial lining with few endometrial glands surrounded by myometrium (Figure 2) with two patent normal looking fallopian tubes(Figure 3).

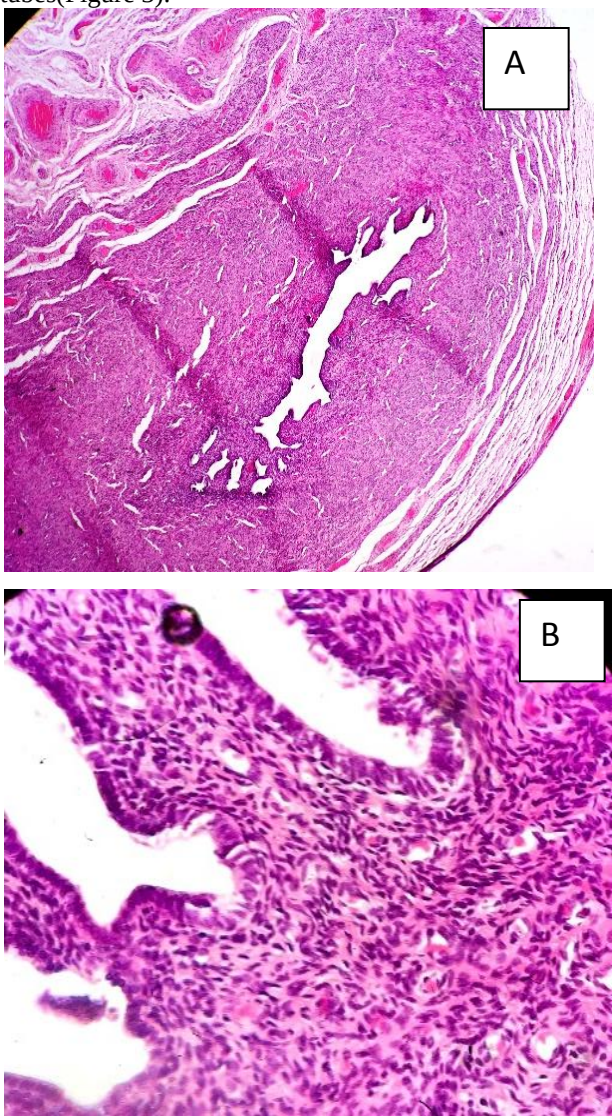


Figure 2. (A) Infantile uterus (x100), (B) Endometrial glands and stoma (x 400)

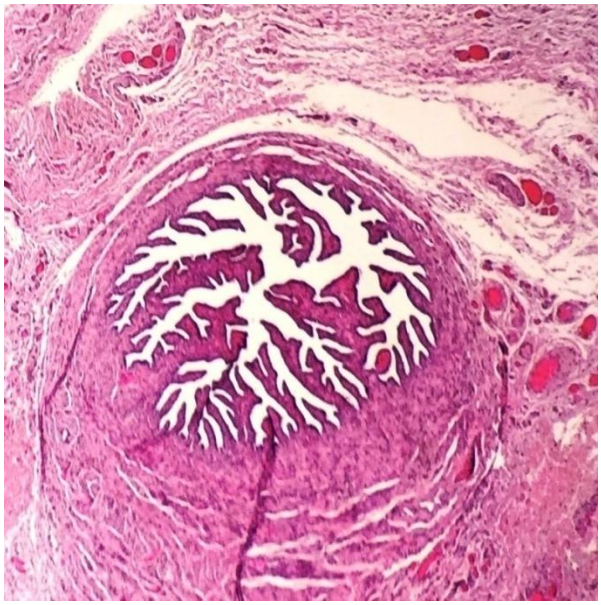


Figure 3 A well developed fallopian tube.

III. DISCUSSION

Persistency of the Müllerian ducts syndrome (PMDS) is a rare condition of male pseudo hermaphroditism, and only sporadic descriptions were made in either men or animals.[1]During normal male embryological development, under the influence of antimullerian hormone (AMH), Mullerian ducts have totally disappeared at 10 weeks of fetal development, releasing the testes from their initial position in the pelvis and allowing them to reach the scrotum, provided the cord is of sufficient length. If the Mullerian ducts fail to regress, the testes remain suspended in the broad ligament. In females, the broad ligament is solidly attached to the pelvis, but in PMDS the connection is usually flimsy or nonexistent, allowing the testes, still attached to the Fallopian tubes, to descend towards the inguinal canal and scrotum, dragging the uterus in their wake.[2]Patients are phenotypically male and usually present when young with unilateral or bilateral cryptorchid testes and an inguinal hernia into which prolapsed infantile uterus and fallopian tubes are found.[3]

PMDS is defined as the presence of mullerian derivatives, uterus, and Fallopian tubes in otherwise normally masculinized 46,XY subjects. The PMDS patient is outwardly completely male, the urethra opens at the tip of the penis (there is no hypospadias).[4]
Persistency of Müllerian Ducts derivates in genetic males is associated with impaired secretion of Anti-Müllerian hormone (AMH) or to AMH receptor deficiency [5-7]. Genetic anomalies in AMH secretion or functioning will interfere with the process of Müllerian Ducts regression.Between 1990 and 2016, molecular

studies in 157 families with PMDS were done. Mutations of either the *AMH* or *AMHRII* gene have been detected in 88% of cases. A few individuals with *AMH* or *AMHRII* mutations may lack mullerian remnants, whereas in others with clinical features of the condition, no mutations can be detected.[4]
Sometimes, Müllerian derivatives are not found in subjects bearing an *AMH* or *AMHRII* mutation. They may be totally asymptomatic.[8]

Familial cases have been reported with a probability of sex-limited autosomal recessive or X-linked recessive inheritance. An incidence of PMDS in identical twins has also been reported.[9] Infertility is the most frequent complication of PMDS but normal spermatogenesis has also been reported. A proportion of 11% of fertile patients in Kuwait and neighboring populations was regestered.[10] All these patients except for one presented with either transverse testicular ectopia or hernia uteri inguinalis, i.e., at least one testis was in a normal scrotal position.[11]unfortunately no such registration found in Iraq.So fertility is rare but possible in PMDS provided two conditions are met: at least one testis should be normally descended and the excretory ducts should be intact.[4]

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