

An Unusual Persistent Mullerian Duct Syndrome in a child in Abha city: A Case Report

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Received: January 2019; Accepted: February 2019; Published: March 1, 2019

Citation: Youssef Ali Mohamad Alqahtani et al. An Unusual Persistent Mullerian Duct Syndrome in a child in Abha city: A Case Report. World Family Medicine. 2019; 17(3): 17-20. DOI: 10.5742/MEWFM.2019.93628

Abstract

Background: Persistent Mullerian duct syndrome (PMDS) is a rare condition that is characterized by the presence of the Mullerian duct structures and is phenotypically and genotypically male. It could result from insufficiency of Mullerian inhibiting factor (MIF) or its receptors.

Case presentation: A 9 month-old Syrian boy was admitted to Abha Maternity and Children Hospital with a previous history of a huge left inguinal swelling since 8 hours, vomiting 4 times, and with yellowish discharge. Routine examinations and investigations were done and the boy was diagnosed with left unilateral inguinal hernia with obstruction and during surgery left ovotestis with fallopian tubes and rudimentary uterus were detected. The histopathology showed no signs of malignancy. After two weeks from left inguinal hernia repair, the boy presented with right incarcerated hernia. The boy underwent right inguinal herniotomy and right

gonadopexy. During the operation, right ovotestis, with vas and fallopian tube were detected. The tube was resected and the sac was dissected; vas and vessels were secured. The boy had no sexual dysfunction and chromosomal investigation showed normal male karyotype. The testosterone level was less than the normal range (0.087 nmol/l).

Conclusion: The PMDS is a rare condition and during early stages cannot be detected; the only diagnostic procedure is when the children are tested for other diseases such as hernia or cryptorchidism. The correct and early diagnosis depends on genetic investigation and endocrinology. Surgery is the treatment of choice.

Key words: Persistent Mullerian duct syndrome (PMDS), Obstructed inguinal hernia, male, Mullerian inhibiting factor, Mullerian duct derivatives.

Background

Persistent Mullerian duct syndrome (PMDS) is a rare condition that has presented in only 150 cases in the literature (1). It is characterized by the presence of the Mullerian duct structures and is phenotypically and genotypically male but the exact etiology is still a debate. It could result from insufficiency of Mullerian inhibiting factor (MIF) or its receptors(2).

The PMD patients are supposed to have normal genitalia and sexual characteristics. Among males, inguinal hernia is characterized by descending of the testis and presence of inguinal hernia. The second type of PMD is ectopia and hernia of both testis (3, 4).

Most of the inguinal hernias present in the groin (75%). Hernias have many complications including obstruction of the bowel and strangling among older subjects(5). Also, ectopic testis diagnosed among PMD patient could result in cryptorchidism among males. The tumors of the testicular germ cell tumors have been reported but are very rare among the Mullerian duct derivatives. If the patients was diagnosed to be phenotypically male, tumor and PMD are not suspected until the time of surgery for hernia repair or treatment of cryptorchidism(6, 7).

Case presentation

A 9 months-old Syrian boy was admitted to Abha Maternity and Children Hospital with a previous history of huge left inguinal swelling since 8 hours, vomiting 4 times, yellowish discharge and these symptoms are the most common symptoms of persistent inguinal hernia according to the literature review(6, 7). Routine examinations and investigations were normal. No history of medical chronic condition, surgical operation, allergy to medications was present and he was diagnosed with left unilateral inguinal hernia with obstruction and the patient was prepared for surgery. The child had urgent left inguinal herniotomy, gonadopexy and diagnostic laparoscopy after 3 days of admission. During operation there was left and right ovotestis with fallopian tubes and rudimentary uterus; thus the boy was referred for consultation in the OPD after 2 weeks (Figure 1).

The histopathology of the tissue from the left fibroid and vas side of the testis showed normal testis composed of capsule, lobule and convoluted seminiferous tubules. The tubules were enclosed by thick basal lamina surrounded by muscles cells. The tubules contained spermatogenic cells and Sertoli cells. No malignancy was seen. At discharge, the HB was 9.7 mg/dl, WBCs were 6.87×10^3 and normal UE.

After two weeks from left inguinal hernia repair, the boy presented with right incarcerated hernia. The boy underwent right inguinal herniotomy and right gonadopexy. During the operation, right ovotestis, with vas and fallopian

tube were found. The tube was resected and the sac was dissected; vas and vessels were secured. 2 biopsies were taken from the vas site and the fimbrial site.

The boy had no sexual dysfunction and chromosomal investigation showed normal male karyotype 46 XY. The testosterone level was less than the normal range (0.087 nmol/l). The ALT level was within normal range, Fe, BUN and creatinine were lower than the normal range and complete blood picture was done. The pathological report showed immature testicular tissue with no signs of malignancy.

Discussion

Pseudo-hermaphroditism is a condition that occurs in males where the testis presents but the internal genital organs are not fully virilized(8, 9).

PMDS is a rare condition that can presented Mullerian duct derivatives and is seen in males. There is still debate about the exact etiology but the deficiency of MIF hormone or its receptors could result in PMDs(10-12).

The MIF is responsible for relapse of the Mullerian duct among male embryos thus any defect on its release from the Sertoli cells in fetal growth since the seventh week of gestation may lead to the persistence of a uterus and fallopian tube in males(13, 14).

This could result in cryptorchidism or inguinal hernia(15, 16)as in the case of our patients who developed left and right inguinal hernia.

The testicles were kept due to absence of malignancy after pathological examination. Also, the tube was resected and the sac was dissected,; vas and vessels were secured. However, other studies showed increased risk of embryonal carcinoma, yolk sac tumor and seminoma among PMDS patients but this difference could be attributed to the fact that our patient was still 9 months old as the risk of tumor is increased by age (1, 2, 4, 7).

Our patient had a low level of testosterone and the chromosomal investigation showed normal male karyotype with 46XY chromosome pattern. In the same respect, normal male development of the epididymis, seminal vesicles and sperm duct needs testosterone from the Leydig cells (17). Also, the AMH deficiency or deficiency of its receptors in the male fetus could result in PMDs development. The testosterone level is not influenced by PMDs thus the chromosomal pattern is presented as 46XY and normal external genital organs as well as development of Wolffian ducts derivatives. However, the patient is phenotypically male, and cryptorchidism could occur (6, 18).

Figure 1:



Fallopian tube



Ovotestis



Left ovotestis



Uterus

Conclusion

The PMDS is a rare condition and during early stages cannot be detected but the only diagnostic procedure is when the children are tested for other diseases such as hernia or cryptorchidism. The correct and early diagnosis depends on genetic investigation and endocrinology. Surgery and removal of the PMDs derivatives is the treatment of choice and further investigations are needed.

Conflict of Interest:

The authors declare that there is no conflict of interest regarding the publication of this paper.

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