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Persistent Mullerian Duct Syndrome: A case report

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Abstract
The Müllerian duct is the structure that later develops into the female reproductive tract during embryology. It usually obliterates in males. Persistent Müllerian duct syndrome is a disorder characterized by the presence of female reproductive organs in a male individual. The common presentations in males are undescended testes or inguinal hernias. The uterus and fallopian tubes are noted during surgery. This syndrome is due to deficiency of fetal anti-Müllerian hormone which is caused by mutations of the gene for anti-Müllerian hormone or anti-Müllerian hormone receptor. The testosterone levels are normal that's why the development of external genitalia is normal. Imaging investigations are the key to establishing the diagnosis. The treatment modality is surgical for replacing the gonads into their normal position and probably a hysterectomy. We reported a case of a 30-year-old male presented with painful, tender, firm and mobile supra pubic swelling. On urogenital examination he had a well-developed penis with no urethral meatus, the scrotum divides into two parts forming labia majora-like structure with an opening in between. The testicles were not palpable. Imaging investigations reveal the cystic pelvic mass and bilateral testes were not visualized. On laparotomy, a distended uterus with bilateral fallopian tubes was found. The uterus consisted foul smelling pus and the testicles were found. Drainage of pus and then a hysterectomy was done. The patient did well and was discharged home. Globally a few cases of Persistent Müllerian duct syndrome were reported but with a variation of manifestation. Persistent Müllerian duct syndrome is a rare condition.

Keywords: Persistent mullerian duct syndrome, A case at Dodoma Tanzania, Benjamin Mkapa Hospital

Introduction

The Müllerian duct is the embryonic structure that later on develops into the female reproductive tract which is the oviduct, uterus, cervix and upper vagina. It usually obliterates during early development in males, but it is retained in those with persistent Müllerian duct syndrome.
The presentations in males are undescended testes or inguinal hernias. Occasionally, both testes are undescended (bilateral cryptorchidism). Often one testis has descended into the scrotum normally, and one has not. The uterus and fallopian tubes are noted during surgery. The testes and female reproductive organs can be located in unusual positions.

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Persistent Müllerian duct syndrome is due to an autosomal recessive congenital disorder and is a form of pseudohermaphroditism due to the presence of Müllerian derivatives (Imbeaud S et al 1996).

The condition is due to a deficiency of fetal anti-Müllerian hormone effect caused by mutations of the gene for anti-Müllerian hormone or the anti-Müllerian hormone receptor, also may be due to insensitivity to anti-Müllerian hormone (Renu D, et al 2010).

Genetically, and - Müllerian hormone or Müllerian inhibiting substance is secreted by Sertoli cells during an individual's whole life. Its role is to obliterate the Müllerian ducts. The Sertoli cells in males secrete Anti – Mullerian Hormone, through the presence of a Y chromosome (Josso N et al, 2005)

Persistent Müllerian duct syndrome is very rare globally because up to the year 2017, only 150 cases were documented (Nerune SM, et al; 2010).

Mullerian duct derivatives are present in a male foetus up to the 8th week of gestation, and their regression is mediated by the müllerian inhibitory factor produced by Sertoli cells. Failure of synthesis or release of müllerian inhibitory factor causes the persistence of müllerian structures (Manjunath B G et al: 2010). The Wolffian duct development progresses in a normal direction because the testosterone levels are normal. This leads to normally developed external genitalia.

Investigations are mainly imaging (CT, US and MRI). These can distinguish the Müllerian duct derivatives such as the uterus, fallopian tubes, and the upper part of the vagina. A specific ELISA test can be used to determine Anti Mullerian Hormone levels in the serum and is a useful screening method to guide the molecular diagnosis. In most cases, Persistent Müllerian duct syndrome is usually discovered incidentally during surgery for undescended testes or inguinal hernia in boys with normal external genitalia.

Treatment is surgical and consists of replacement of the gonads within the scrotum, requiring careful dissection of the Müllerian derivatives. Total hysterectomy is not recommended because of the risk to the vas deferens. But otherwise, a hysterectomy can be offered to improve the chances of fertility and to prevent the occurrence of neoplastic tissue formation (Colacurci N, et al: 1997).

Case report

We report a case of a 30-year-old male presented at our surgical clinic with the complaint of progressive mild tender supra pubic swelling (figure 1). He reported the swelling to be of eight months duration, there were neither gastrointestinal nor urinary manifestations associated with the swelling. No history of fever was reported. Neither history of marriage nor childbirth. On examination he was an average-built man with an obvious symmetrical supra pubic swelling, extending above the umbilicus, which was mild- tender and mobile.

On urogenital examination he had a well-developed penis with no urethral meatus, the scrotum divides into two parts forming labia majora-like structure with an opening in between (Figures 2 and 3). Upon catheterization, greenish-yellowish pus was drained. The testicles were not palpable. Ultrasound examination and then a CT scan showed features suggestive of cystic pelvic mass and the impression was a mesenteric cyst.

Bilateral testes were not visualized in the scrotum/inguinal canal/abdomen. On explorative laparotomy under general anaesthesia, a huge, distended uterus was visualized with bilateral fallopian tubes (Figures 4, 5, and 6). When opened, the greenish-yellowish pus with a foul smell of approximately 6 litres was extracted (figure 7). On culture and sensitivity of the pus, the Staphylococcus aureus isolated which was sensitive to vancomycin and cefazolin. No testicles were found. A hysterectomy was done. The patient was kept on intravenous antibiotics and analgesics and was discharged on the fifth day postoperatively after fully recovery.

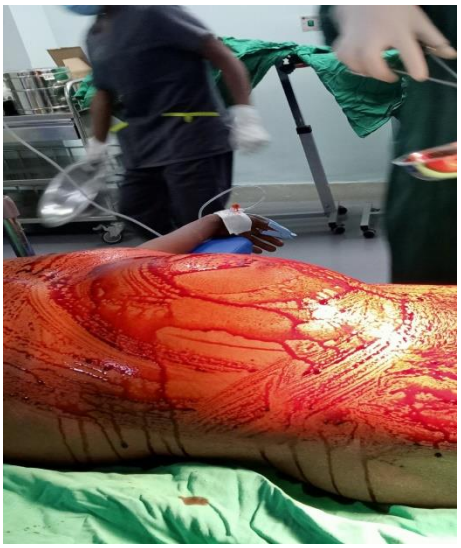

Figure 1: Displays a patient with abdominal distension prepared and ready for exploratory laparotomy

Figure 2

Figure 3

Both figures 2 and 3 show a penis which has no urethral opening and an opening is shown in between the divided scrotum.

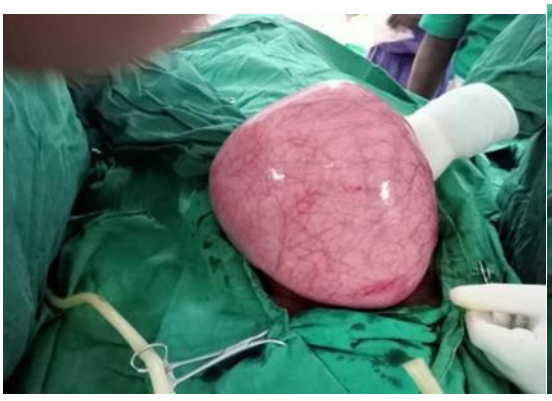

Figure 4

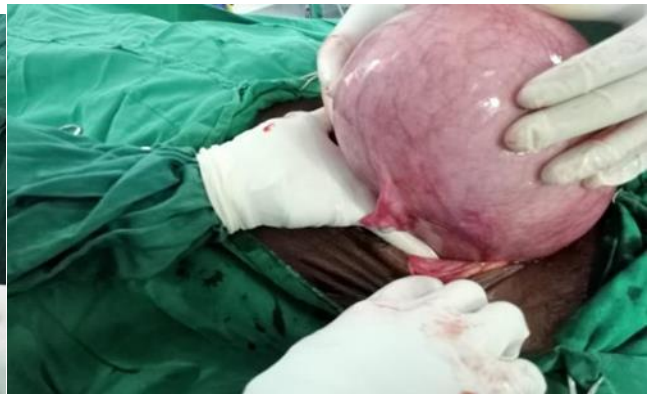

Figure 5

Figure 3, 4 and 5 displays a distended uterus with fallopian tubes on its sides

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Figure 6

Figure 7: Shows a pus which was drained from the distended uterus

Discussion

Persistent Müllerian duct syndrome is a sexual development disorder characterized by the presence of female reproductive organs in individuals with both normal chromosomes (46, XY) and a normal phenotype of a male. Male sex differentiation is driven by 2 hormones, testosterone and anti-müllerian hormone, responsible for the regression of müllerian ducts in male fetuses.

According to the literature, Persistent Mullerian Duct Syndrome is a rare condition and even in our hospital this condition is not common. A few cases were reported in different articles. Diagnosis is often made incidentally during surgery for an inguinal hernia or during exploration for cryptorchidism. Different cases of Persistent Mullerian Duct Syndrome reported were found to have variations of how they present. In the year 2013, Vijaya Patil et al reported a case of Persistent Mullerian Duct Syndrome in which a patient presented with reduceable painful right-sided groin swelling who had no children and had no sexual dysfunction. Secondary sexual characteristics were well developed and the patient had undescended testes. The scrotum was well developed but semen analysis showed azoospermia (Indian J Surg et al; 2013). In our case, the secondary sexual characteristics were well-developed but the hernia was not present.

In some cases, the reports of histopathological studies of gonads show different variations. For example, in a case reported in 2014 by Vaibhav Nayak et al, a microscopic study of the right gonad showed normal-looking seminiferous tubules with spermatogonia at its various maturation levels for the age of the patient and a normal-looking epididymis. The left gonad showed the structure of the

uterus comprising normal-looking endometrium and myometrium. [8] In our case, the male gonads were not seen radiologically and intraoperatively probably there was testicular agenesis or ectopic testicles.

In patients with intraabdominal testes, both the gonads may be located in a position analogous to the ovaries, with a rudimentary uterus in the centre and the mullerian remnants preventing the mobilization of the testes (Int J Appl Basic Med Res. 2014).

In one case reported in 2021 by Marah Mansour et al, a patient with Persistent Mullerian Duct Syndrome was found to present with abdominal pain and a large cystic mass in the right iliac fossa which after investigations revealed a dermoid cyst in an undescended right-sided testis. Also, a large internal iliac lymph node was found. However, ovaries were not found (Manjunath BG et al; 2010, Mansour, M et al 2021). Our case showed cystic swelling located on the suprapubic region and it was a pus filling the uterus and causing the distension.

Surgical options also depend on the presentation of the condition i.e. no common or single treatment modality. Generally, there should be a removal of mullerian remnants to correct the pathology noted.

Conclusion

Persistent Mullerian Duct Syndrome is a rare condition. Mostly is detected incidentally during surgery for hernia repair, undescended testes or laparotomies for other conditions. Fertility is always unaffected. The pt is phenotypically and genotypically normal.

Acknowledgement

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Conflict of interest

The Authors have no conflict of interest to declare

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