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Case Report

An unusual case of inguinal hernia: Persistent mullerian duct syndrome with transverse testicular ectopia

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Persistent Müllerian Duct syndrome (PMDS) is a rare variety of male pseudo-hermaphroditism. It is characterized by the persistence of the uterus, fallopian tubes and upper vagina in otherwise normally virilized boys. The association of the syndrome called transverse testicular ectopia (TTE), a condition in which both testes are located in one inguinal canal is rare. Here, we report a young male patient presented with infertility, left sided inguinal hernia and right-sided undescended testis since birth, finally found to have PMDS with TTE.

Key words: Müllerian duct, testicular ectopia, undescended testis, orchidopexy.

INTRODUCTION

Persistent Müllerian Duct syndrome (PMDS) is a rare variety of male pseudo-hermaphroditism. It is characterized by the persistence of the uterus, fallopian tubes and upper vagina in otherwise normally virilized boys. Despite the normal male genotype (46 XY) and the subsequent normal development of fetal testes, müllerian structures do not regress either due to absence of Müllerian Inhibiting Substance (MIS) or lack of response to it. Since the secretion and action of testosterone is not affected, the Wolffian duct derivatives and the external genitalia of the fetus progresses in the normal male direction. This syndrome may be rarely associated with transverse testicular ectopia, a condition in which both testes are located in one inguinal canal. It was first described by Lennossek in 1886.

CASE REPORT

A 23 years old male patient is presented with infertility, left sided inguinal hernia and right sided undescended testis since birth. The patient was phenotypically male with normal secondary sexual characters. Examination of inguinoscrotal region revealed a left sided, complete, reducible inguinal hernia. The left hemiscrotum was well developed. The left testis was palpable in the inguinal canal and could be manipulated in the scrotum and also pushed into the abdomen. Two more globular, firm and partially reducible swellings were palpable in the left inguinal canal. In one of those swellings, the patient had testicular sensation and the patient had a feeling that his right testis was on the left side. On the right, the hemiscrotum was empty and the testis was not palpable along its normal course. The penis was well developed.

Ultrasound examination showed normal left testis with bulky epididymis. A non-homogenous mass (2.8 × 1.7 × 1.7 cm) was visualized in the left inguinal region. Partial herniation of small gut loops was seen through left deep ring. Right testis could not be visualized. Prostate was normally situated. Diagnostic laparoscopy showed a well developed uterus, cervix and fallopian tubes. Both the testes were visualized. They were highly mobile and were attached to round ligament on either side. As the uterus was manipulated towards the left inguinal canal, right testis was also dragged into the left inguinal canal. Biopsy was taken from the right testis.

Histopathological report of the right testis showed Leydig cell hyperplasia, seminiferous tubules containing

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sertoli cells with areas showing hyalin degeneration and fibrosis and most significantly, no germ cells were seen. Semen analysis showed no spermatozoa. Serum testosterone and anti-mullerian hormone (AMH) levels were 862.70 ng/dl and 10 ng/ml. Karyotype analysis showed 46 XY pattern.

Patient was informed about his unusual condition. Patient expressed his wish to remain as male. Thereafter, consent for hysterectomy was taken and operation was planned. On operation, hysterectomy with bilateral salpingectomy was done. Both the testis was mobilized and orchidopexy was done by placing the testis in subdartos pouch. Post operative recovery was uneventful. Histopathological report of uterus showed normal uterine tissue with proliferative endometrium. Patient was followed up after one week, one month and three months after operation. His testes and scrotum were easily palpable and there was no evidence of testicular atrophy at these visits.

DISCUSSION

Embryologically, between 7 and 8 weeks of gestation, masculinisation occurs in a male fetus. Testosterone, secreted by the Leydig cells in the testis aids the development of Wolffian duct into vas deferens, epididymis and seminal vesicle. Müllerian inhibiting substance, secreted by Sertoli cells in the testis, acts locally and unilaterally to suppress the müllerian ducts and causes their regression by 8^{th} and 10^{th} week of fetal life. PMDS is caused by a defect in either the synthesis of, or the receptor for, Müllerian - inhibiting substance (Manjunath et al., 2010; Josso et al., 2005).

PMDS can occur sporadically or inherited either as X-linked or autosomal dominant sex-limited trait. PMDS has also been reported in association with Klinefelter's syndrome, Turner's syndrome and Mayer-Rokitansky-Kuster-Hauser syndrome (Hoshiya et al., 2003). PMDS cases are divided into three categories (Manjunath et al., 2010):

1. Majority (60 to 70%) with bilateral intra-abdominal testes, in a position analogous to ovaries.
2. Smaller group (20 to 30%), with one testis in the scrotum, associated with contralateral inguinal hernia whose contents are testis, uterus and tubes (classical presentation of hernia uteri inguinale).
3. Smallest group (10%) where both the testes are located in the same hernial sac along with the müllerian structures (transverse testicular ectopia - TTE). PMDS accounts for 30 to 50% of all cases of TTE. Jordan first described the syndrome of TTE with PMD in 1895.

Affected individuals have normal male phenotype, normal virilisation, undergoes normal male puberty, and may even be fertile if the gonads are placed in the scrotum (Josso et al., 2005). Pre operative diagnosis is not made in most cases and it is usually an operative surprise during orchidopexy or inguinal hernia repair.

In patients with bilateral abdominal testes, the rudimentary uterus lies in the centre with both the gonads located in a position analogous to the ovaries. This may lead to confusion during surgery regarding the real genetic and gonadal sex of the patient. A two staged procedure as described by Loeff et al. (1994) including a gonadal biopsy in stage one followed by re-exploration of the patient several months later in stage two is not needed. The presence of epididymis and the vas deferens would unequivocally reveal that the gonad is a testis and a decision to perform an orchidopexy can be taken during initial exploration itself. However, testicular biopsy may be needed to rule out mixed gonadal dysgenesis or carcinoma *in situ*. Testicular biopsy may transgress the blood-testes barrier, which may result in the production of antisperm antibodies. Thus, Berkman opined that testicular biopsy is not needed as long as the testis is placed at a location where it can be palpated. Impalpable testicular malignancies can be detected through ultrasound (Berkman, 1997). Orchidectomy is indicated in patients when the testes cannot be mobilised to palpable location outside the abdomen as the incidence of malignancy intraabdominal is high (Vandersteen et al., 1997). Seminoma, embryonal cell tumours and teratomas have been reported in such testes, even after orchidopexy.

In some cases, müllerian remnants prevent the mobilisation of the testes. In such a situation, the müllerian structures can be split in the midline, thus, mobilising the testes to reach the scrotum on either sides. Microvascular autotransplantation of the testis is another alternative (Brandli et al., 2005). Complete excision of the müllerian duct remnants may compromise the vascularity and integrity of vas deferens. At times, the vas deferens may be incorporated into the wall of the vagina or it may lie in close proximity to the fallopian tubes and the vagina (Lima et al., 2000). Vandersteen et al. (1997) evaluated the risk of injury to the vas deferens during excision of the müllerian remnants versus the risk of retaining müllerian structure and concluded that the müllerian remnants should not be excised, as malignancy had not been reported till then. Berkman (1997) too opined, since no malignancy was reported, removal of remnants was not necessary. However, since 1997, several cases of malignancies arising from the retained müllerian structures have been reported. Theil et al. (2005) reported the death of a 14-year-old boy with PMDS, due to metastasis of adenosarcoma arising from the müllerian remnant (Thiel and Erhard, 2005). Romero et al. (2005) reported a 39-year old man who developed adenocarcinoma of endocervical origin, arising from müllerian remnant (Romero et al., 2005). Shinmura et al. (2002) reported clear cell adenocarcinoma arising from uterus in a 67-year-old man with PMD. Such recent

reports of malignant degeneration of the müllerian remnants warrant a rethink into the management options of the müllerian remnants. In our case, we excised the uterine remnants which were located away from the vas deferens. Manjunath et al. (2010) recommended stripping/destroying the mucosa of the retained müllerian remnants to reduce the risk of malignancy and simultaneously preventing damage to the vas deferens and disruption of collateral blood supply to the testes. Recurrent urinary tract infection (UTI), stones and voiding disturbances are also known to occur in cases of retained müllerian remnants which are in connection with the prostatic utricle. The risk of UTI and stone formation can be reduced by destruction of the mucosa of the vaginal remnant as it will obliterate the cavity due to adhesions. If a two stages Stephen-Fowler procedure is contemplated for abdominal undescended testes, then, the excision of the müllerian remnant may be hazardous for the collateral circulation of the testes. Excision of the uterus and the fallopian tubes may disrupt the vasal collateral blood supply, which should be retained for the viability of the testis. In such patients, midline splitting of the müllerian remnants and excision of the mucosa would allow the complete mobilisation of both the testes preserving the collateral blood supply from the mesometrium.

In PMDS patients with TTE, mobilisation of the testis is usually easier. Trans-septal orchidopexy is the treatment of choice, along with excision of the müllerian remnants, as mentioned earlier. Lima et al. recommended that all cases of TTE should undergo abdominal exploration to rule out PMDS. Martin et al. (1992) documented spermatogenesis in a patient with TTE. Fertility is rare in patients with PMDS. Imbeaud et al. reported three cases of PMDS with testicular degeneration and opined that anatomical abnormality may favour testicular torsion and early loss of testis (Imbeaud et al., 1995). Laparoscopy has huge benefits in the diagnosis as well as the treatment of PMDS [8]. Shirasaki et al. described laparoscopic excision of the uterus followed by orchidopexy in 1-year-old child with PMDS (Shirasaki et al., 2003). Turaga et al. (2006) described an algorithm-based approach for hernia uteri inguinale, depending on laparoscopic findings. Ng and Koh, (1997) described one stage orchidopexy for intraabdominal testes in PMDS with division of the testicular vessels, with careful reservation of the collateral blood supply around the müllerian remnants. El-Gohary et al. (2003) described laparoscopic orchidopexy in PMDS by splitting the müllerian remnants in the midline and thus achieving adequate length for the testes to reach the scrotum [18]. The technique of laparoscopically stripping the mucosa of the müllerian remnant can be used during such a procedure, so as to reduce the risk of malignancy. Laparoscopy has the advantage of improved access to proximal vagina which is located deep in the pelvis where it opens into the prostatic utricle. Screening of the siblings and second degree relatives is necessary, as PMDS has autosomal recessive inheritance. Ultrasonography and MRI have been reported to be of value in locating the müllerian remnants (Otsuka et al., 2003; Di Cesare et al., (1998). But we feel that all the siblings with undescended testis should be subjected to diagnostic laparoscopy.

CONCLUSION

Correct diagnosis of this condition warrants familiarisation of the surgeon with PMDS entity while dealing with patients of cryptorchid or undescended testis of all age groups. When clinically the diagnosis is in doubt, diagnostic laparoscopy and biopsy is confirmatory. Recent reports of malignancy in the müllerian remnants being high; we recommend excision of müllerian remnants carefully without compromising the vascular supply of the vas deferens and testes. During the process of dissection if the surgeon feels that the vascularity to vas deferens and testis may be at undue risk, stripping off of the mucosa of müllerian remnants is recommended as the malignancy often arises from the mucosa.

REFERENCES

Berkman F (1997). Persistent müllerian duct syndrome with or without transverse testicular ectopia and testis tumours. Br. J. Urol., 79(1): 122–126.

Brandli DW, Akbal C, Eugsster E, Hadad N, Havlik RJ, Kaefer M (2005). Persistent müllerian duct syndrome with bilateral abdominal testis: Surgical approach and review of the literature. J. Pediatr. Urol., 1(6): 423–427.

Di Cesare E, Di Bartolo De Vincentiis V, Maffey MV, Splendiani A, Masciocchi C (1998). US and MRI in a case of persistent müllerian duct syndrome. Pediatr. Radiol., 28(11): 865-867.

El-Gohary MA (2003). Laparoscopic management of persistent müllerian duct syndrome. Pediatr. Surg. Int., 19(7): 533–536.

Hoshiya M, Christian BP, Cromie WJ, Kim H, Zhan Y, MacLaughlin DT, Donahoe PK (2003). Persistent müllerian duct syndrome caused by both a 27-bp deletion and a novel splice mutation in the MIS type II receptor gene. Birth Defects Res. A Clin. Mol. Teratol., 67(10): 868–874.

Imbeaud S, Rey R, Berta P, Chaussain JL, Wit JM, Lustig RH, De Vroede MA, Picard JY, Josso N (1995). Testicular degeneration in three patients with the persistent müllerian duct syndrome. Eur. J. Pediatr., 154(3): 187–190.

Josso N, Belville C, di Clemente N, Picard JY (2005). AMH and AMH receptor defects in persistent müllerian duct syndrome. Hum. Reprod. Update, 11(4): 351–356.

Lima M, Domini M, Libri M (1997). Persistent müllerian duct syndrome associated with transverse testicular ectopia: A case report. Eur. J. Pedtr. Surg., 7(1):60–62.

Lima M, Morabito A, Libri M, Bertozzi M, Dòmini M, Lauro V, Strano C, Messina P, Tani G (2000). Laparoscopic removal of persistent müllerian duct in a male: Case report. Eur. J. Pedtr. Surg., 10(4): 265–269.

Loeff DS, Imbeaud S, Reyes HM, Meller JL, Rosenthal IM (1994). Surgical and genetic aspects of persistent müllerian duct syndrome. J. Pediatr. Surg., 29(1): 61–65.

Manjunath B G, Vasanth G Shenoy, Preetham Raj (2010). Persistent müllerian duct syndrome: How to deal with the müllerian duct remnants - a review. Indian J. Surg., 72: 16–19.

Martin EL, Benette AH, Cromie WJ (1992). Persistent müllerian duct syndrome with transverse testicular ectopia and spermatogenesis. J.

Urol. 147(6):1615–1617.

Ng JW, Koh GH (1997). Laparoscopic orchidopexy for persistent müllerian duct syndrome. Pediatr. Surg. Int., 12(7): 522–525.

Otsuka T, Nakazono T, Matsuo Y, Kanou T, Masaki Z, Kudo S (2003). Persistent müllerian duct syndrome suggested by magnetic resonance imaging. Radiat. Med., 21(6): 277–299.

Romero FR, Fucs M, Castro MG, Garcia CR, Fernandes Rde C, Perez MD (2005). Adenocarcinoma of persistent müllerian duct remnants, case report and differential diagnosis. Urology, 66(1): 194–195.

Shinmura Y, Yokoi T, Tsutsui Y (2002). A case of clear cell adenocarcinoma of the müllerian duct in persistent müllerian duct syndrome: The fi rst reported case. Am. J. Surg. Pathol., 26(9): 1231–1234.

Shirasaki Y, Nagai A, Nasu Y, Iguchi H, Kumon H (2003). Laparoscopic removal of müllerian structures and orchiopexy for persistent müllerian duct syndrome. Urology, 62(6): 1121.

Thiel DD, Erhard MJ (2005). Uterine adenosarcoma in a boy with persistent müllerian duct syndrome: fi rst reported case. J. Pedtr. Surg., 40(9): e29–31

Turaga KK, St Peter SD, Calkins CM, Holcomb GW 3rd, Ostlie DJ, Snyder CL (2006). Hernia uterus inguinale: a proposed algorithm using laparoscopic approach. Surg. Laparosc. Endosc. Percutan. Tech., 16(5): 366–367.

Vandersteen DR, Chaumeton AK, Ireland K, Tank ES (1997). Surgical management of persistent müllerian duct syndrome. Urology, 49(6): 941–945.