

Oncology

Cancer on cryptorchid testis revealing a Persistent Müllerian Duct Syndrome: A rare case

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ABSTRACT

The persistence of Müllerian derivatives syndrome or PDMS is a rare form of male pseudohermaphroditism. Its association to ectopic testicular cancer is even rarer. Because of its rarity it is difficult to diagnose preoperatively. We present a case of a cancer on cryptorchid testis revealing a persistent Müllerian duct syndrome.

Introduction

The persistence of Müllerian derivatives syndrome or PDMS is a rare form of male pseudohermaphroditism due to the lack of secretion or action of the antimüllerian hormone or AMH. It is characterized by the presence in an individual of a uterus, the fallopian tubes and the upper part of the vagina with, moreover, male-type external genitals. The karyotype is usually normal of the XY type. Its association with testicular cancer often developed on cryptorchid testis is an exceptional syndromic association described for the first time by NILSON in 1939.

Case presentation

A 46-year-old patient, presenting with primary infertility for 20 years, has been consulting for abdominal and pelvic pain that has been evolving for 2 days. He had a male morphotype, the scrotum was empty and the penis was well developed. The palpation of the abdomen showed an abdominopelvic mass of 13 cm. No other abnormalities were found. Abdominal CT showed a large, 13 cm long, well-defined right abdominal-pelvic mass with an endopelvic extension and right retro-vesical extension (Fig. 1). Thoracic CT was normal. There was a very high elevation of AFP at 95880 ng/ml, the rest of the biological examinations were normal. The patient was operated on by a laparotomy. Intraoperative exploration found a hemoperitonium of about 1 L, an abdominal tumor developing on the right gonad of 13 cm ruptured on the left side (Fig. 2) blocked by the large epiploon with presence of 2 fallopian tubes, a normal left gonad, a 5 cm uterine cavity and a 2.5 cm prostatic utricle (vaginal cavity). It was a tumor developing on a cryptorchid right testicle with persistence of Müllerian derivatives. Complete excision of the tumor and müllerian duct derivatives were

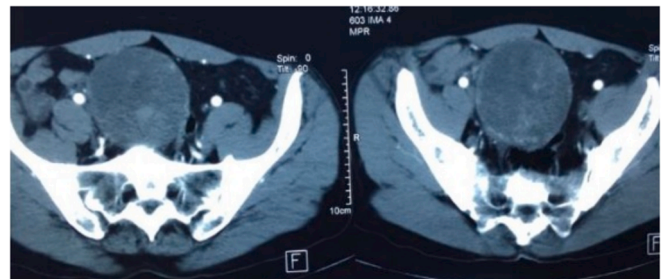


Fig. 1. Abdominal CT showing a large, 13 cm long, well-defined right abdominal-pelvic mass with an endopelvic extension and right retro-vesical extension.

performed. The post-operative follow-ups were simple and the patient was put out after 3 days of hospitalisation. The examination of the 13 cm specimen showed two fallopian tubes, two epididymides, the uterus and a left atrophic gonad, a left seminal vesicle 2.5 × 1x1 cm, a prostatic utricle. Histopathological findings showed a germ tumor with several histological components: predominantly vitelline (≥75%), seminoma (≤20%) and embryonic carcinoma (≤5%) (Fig. 3). The immunohistochemical study confirmed the mixed nature of this germ tumor. chemotherapy with BEP 4 cycles was started on the 15th post-operative day. The patient did not show for follow-up, Then was hospitalised after 9 months in a general condition impairment and died.

Discussion

Persistent Müllerian Duct Syndrome (PMDS) is a rare form of internal male pseudo-hermaphroditism. This condition defined by the

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Fig. 2. 1: intraoperative photo of the ruptured tumor and specimen after excision: a-right testicular tumor b- Fallopian tube + left testis c- Uterus d-vaginal cavity e-right seminal Vesicle.

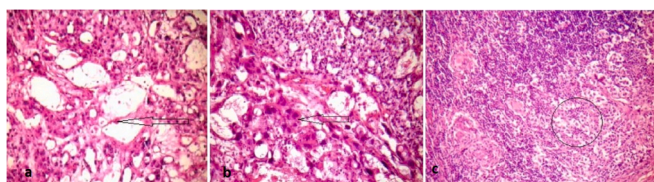


Fig. 3. Histopathological findings: a-vitelline component b-embryonic carcinoma c-seminoma.

presence of structures derived from the Mullerian duct (i.e uterus, cervix, fallopian tubes and upper part of vagina) in a normal genotypically and phenotypically male. This disorder caused by deficiency in the production of Müllerian inhibiting factor (MIF) from the immature Sertoli cells in the newly formed testes.¹ it may be detected in infancy or adulthood. In a male fetus, Leydig cells and Sertoli cells of the newly formed testes start producing testosterone and MIF, respectively at the 8th week of gestation. Any defect in this normal pathway leads to persistence of mullerian duct structures. This remnant Mullerian structures lead to cryptorchidism by hindering the normal testicular descent.² The secretion and response of target organs to testosterone are not affected, so the wolffian duct structures and external genitalia progress to a normal male. PMDS is categorized into male form and female form according to the anatomical structure. The most common form is the male form and constitutes more than 80% of the case. Type I (60–70%): Have intra-abdominal mullerian structures and testes in a position analogous to that of ovaries, type II (20–30%) also known as hernia uteri inguinal it has one testis in hernial sac or scrotum together with mullerian structures and type III (10%): have both testes located in the same hernia sac along with fallopian tubes and uterus (transverse testicular ectopia).³ Due to its rarity PMDS is rarely diagnosed before surgery. the reason for consultation is generally non-specific, with vague abdominal and pelvic pain. The fortuitous discovery of the

disease during an exploration of infertility has been reported. When associated with cryptorchid testes tumor, like the case of our patient, patients may present anorexia, weight loss, general condition impairment or symptoms related to the tumor extension. Surgical treatment consists of exeresis of the tumor and Mullerian derivatives. IT can be done by classic surgery or laparoscopic surgery.⁴ In our case, classic surgery was performed. The risk of malignancy in an ectopic testis in a case of PMDS is similar to that in a cryptorchid testis in a healthy male.⁵ the prognosis of the disease depends on the characteristics of the tumor and chemotherapy reduces the risk of recurrence.

Conclusion

PMDS with mixed germ tumor is a rare entity. Because of its rarity it is difficult to diagnose preoperatively and hence it is difficult to predict intraoperatively. Surgeons and pathologists should be aware of this situation when dealing with cryptorchidism and malignancy in it. The risk of developing malignancy and other symptoms is higher in an intra-abdominal testis. When attempting for surgery for germ tumor in such a scenario the mullerian remnants should also be removed.

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