

Persistent Müllerian Duct Syndrome: A Challenge in Diagnosis and Treatment

Persistent Müllerian duct syndrome (PMDS) is a very important issue in clinical practice, mainly because of its rarity and difficult diagnosis and management. PMDS was first described by Nilson^[1] in 1939 and is caused by a defect in the Müllerian inhibiting factor (MIF) system. In the current issue of the Journal, Elabd and Almalki described a case of PMDS that warrants some highlighting of this condition.^[2]

Patients are genotypically and phenotypically male, but they have a uterus and fallopian tubes due to failure of Müllerian duct regression. These structures do not regress due to a failure in the production of MIF or insensitivity of the target organ to MIF.^[3] However, affected males can be sterile because testes cannot be properly connected with the excretory ducts, due to aplasia of the epididymis and the upper part of the deferens. Fertility is rare but possible if at least one testis is scrotal, and its excretory ducts are intact. Testosterone levels and the sexual function are usually normal except if testicular degeneration is found. PMDS is an autosomal recessive disorder (the gene responsible of disease is on chromosome 19),^[4] so the diagnosis is challenging. Only 300 cases have been reported in literature up to 2016.^[5]

PMDS is usually discovered accidentally during surgery for cryptorchidism or inguinal hernia repair in males with normal external genitalia. Although ultrasonography and magnetic resonance imaging are reported to play a role in locating the Müllerian remnants, laparoscopy has a distinctive advantage in diagnosing PMDS.^[6] There are three clinical variants of PMDS. The 60%–70% of the patients show bilateral intra-abdominal testes in a position analogous to ovaries. The second group (20%–30%) is characterized by one testis in the scrotum with a contralateral inguinal hernia, which contains testis, uterus, and tubes. In the least common form (10%) both the testes are located in the same hernia sac

along with the Müllerian structures.^[7] Sometimes, in patients with intraabdominal testes, a rudimentary uterus is found in pelvis, and the Müllerian remnants prevent the mobilization of the testes.^[8]

The surgical approach is justified by the malignant tumors identified in Müllerian remnants. Moreover, the Müllerian duct can be connected with the seminal vesicle causing urinary tract infections, periodic hematuria, stones, and urination disorders. In some cases, is possible to remove the mucosa of the retained Müllerian structures to reduce the risk of malignancy, without compromising the integrity and vascularity of the vas deferens.^[9] Despite PMDS is well-described in the scientific literature, the contributions made by Almalki and Elabd^[2] is an important addition, because the syndrome was usually diagnosed during in childhood and adolescence while in this case PMDS was discovered in a 47-year-old patient. It should be advisable to perform an early diagnosis of this condition, to reduce the likelihood of unrecognized malignancy in middle age. Moreover, the surgery at a younger age reduces the risks of damaging the vas deferens. The increasingly widespread use of genetic tests can identify people who hold the mutated gene and recommend about the risk in who is trying to have a child. A multidisciplinary team (including urologists, gynecologists, geneticists, and psychiatrists) and an holistic approach to these cases are recommended. In particular, it is important to involve a psychiatrist or a psychologist for the evaluation of the psychological impact of the disease on the patient.

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Conflicts of interest

There are no conflicts of interest.

Compliance with ethical principles

This article does not contain any studies involving human participants performed by any of the authors.

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