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Dysmorphology

Persistent Müllerian Ducts Syndrome: A Case Report

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Abstract **Case Report**

We report the case of a 6 years old child, admitted for management of testicular ectopia, the clinical examination found an empty scrotum, a normal looking penis and a urethral meatus in place. The scrotal ultrasound showed an intra-abdominal testicle (left illiac fossa) of homogeneous echostructure on the left side, while the right testicle was not visualized. The gonadotropins LH and FSH levels were low, as well as the level of testosterone, the secretion of which is stimulated after administration of Pregnyl. Laparoscopy revealed 2 gonadal formations on the same side, 2 tubes and a median formation reminiscent of the uterus. The karyotype of the patient was male 46XY, the dosage of anti Müllerian hormone (AMH) was high, confirming an abnormality of its receptors, which explains the persistence of the Müllerian ducts in this child. The child underwent a left gonadectomy with bilateral hysterectomy and adnexectomy, associated with a lowering of the right testicle. At 9 years, pubertal development was classified as T3P4 according to Tanner's classification, scrotal ultrasound showed a free left testicular compartment with testicular calcifications classified as grade 3, and tumor marker levels were normal.

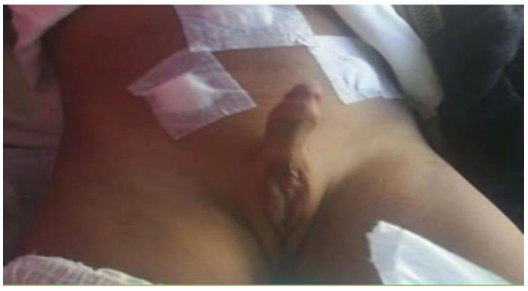
Keywords: Müllerian ducts; Testis tumor; Pseudohermaphroditism; Cryptorchidism.

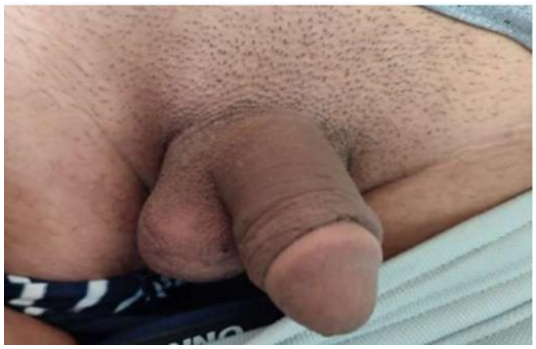
INTRODUCTION

Müller duct persistence syndrome (MDPS) represents a rare form of internal male pseudohermaphroditism and is characterized by the presence of the uterus, fallopian tubes and upper vagina in a normally virilized boy with a 46 XY karyotype.

OBSERVATION

We report the case of a 6 years old child, admitted for management of testicular ectopia, the clinical examination found an empty scrotum, a normal looking penis and a urethral meatus in place. The scrotal ultrasound showed an intra-abdominal testicle on the left side (left illiac fossa) with a homogeneous echostructure, while the right testicle was not visualized. The gonadotropins LH and FSH levels were low, as well as the level of testosterone, the secretion of which is stimulated after administration of Pregnyl. Laparoscopy revealed 2 gonadal formations on the same side, 2 tubes and a median formation reminiscent of the uterus. Histopathological examination revealed dystrophic germ cells with microcalcifications in the testicular parenchyma, without evidence of ovarian or tumour structures, the patient's karyotype was male 46XY. The anti-Müllerian hormone (AMH) assay showed a high level, confirming an abnormality of its receptors, which explains the persistence of Müllerian ducts in this child. The child benefited initially from a hysterectomy, bilateral adnexectomy, associated with a lowering of the 2 testicles. Then a left orchiectomy (fibrotic testicle) in a second step. At 9 years, pubertal development was classified as T3P4 according to Tanner's classification, scrotal ultrasound showed a free left testicular compartment with testicular calcifications classified as grade 3, and tumor marker levels were normal.





DISCUSSION

The embryo is undifferentiated until the 8th week of embryonic development, there is coexistence of the 2 outlines of the male and female genital tracts: the Wolff and Müller ducts. In men, the secretion of testosterone and antimüllerian hormone (AMH) by the fetal testes controls sexual differentiation into the male morphotype. AMH is secreted by Sertoli cells from the 7th week of amenorrhea (SA), and leads to the regression of müllerian structures. In the absence of AMH, the Müllerian ducts lead to the formation of 2 fallopian tubes, the uterus and the upper 2/3 of the vagina. Testosterone, secreted by the Leydig cells from the 9th SA onwards, has a direct effect on the wolffian structures that lead to the formation of the vas deferens, the epididymis and the seminal vesicles. It also has an indirect effect on the virilization of the urogenital sinus and external genitalia through dihydrotestosterone. In the urogenital sinus, the prostate develops while the vagina regresses. At the level of the external genitalia, there is a lengthening of the anogenital distance, fusion of the genital buds and closure of the urethra. In PMDS patients, the serum testosterone level is therefore normal. The lack of AMH synthesis or the insensitivity to AMH before the critical age of 8 SA would explain the persistence of the Müllerian ducts responsible for the three clinical forms of PMDS:

- The female form, with non-palpable testes bilaterally in the pelvic position;
- The male form (90%) associating inguinal hernia and contralateral testicular migration defect;
- Transverse testicular ectopia where both testicles and tubes are located in the same hemi-scrotum. PMDS is an autosomal recessive disorder. The karyotype is always type 46 XY, two karyotypic abnormalities are possible, firstly the mutation of the AMH gene (chromosome 19), as well as the mutation of the AMH type II receptor (chromosome 12). The diagnosis is often made in early childhood during surgery for cryptorchidism or strangulated hernia repair or rarely in adults during a hypofertility assessment or a testicular tumor.

The objective of the management is the preservation of fertility and hormonal function, which implies early surgical management in childhood, especially for testicular lowering.

There is no formal recommendation for the systematic removal of müllerian structures, however long-term monitoring is essential through clinical examination, imaging such as pelvic ultrasound or pelvic MRI, given the risk of malignant degeneration.

CONCLUSION

PMDS should be considered in any bilateral cryptorchidism or inguinal hernia associated with a suspicious mass. The objective of the management is the preservation of fertility and hormonal function with regular monitoring because of the risk of malignant degeneration.

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