Persistent Mullerian Duct Syndrome - Case Report

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Case Report

Abstract: Persistent Mullerian duct syndrome (PMDS) refers to the presence of a uterus and sometimes other Mullerian duct derivatives in a genetically male. We report a case of male with right side cryptorchidism and left inguino scrotal mass with Mullerian duct derivatives (uterus).

Keywords: Persistent Mullerian Duct Syndrome, Mullerian inhibiting factor.

Introduction

Persistent Mullerian duct syndrome (PDMS) is a rare disorder with the presence of Mullerian duct derivatives in an otherwise phenotypical male([1]). PMDS results from the failure of synthesis or release of Mullerian inhibiting factor (MIF), the failure of end organ to respond to MIF, or a defect in timing of the release of MIF.

Case Report

A 50-year-old male, from the farming community, presented with a mass in the inguinoscrotal region since 2 years, which was gradually increasing in size but with a rapid increase in size since 1 month. The patient was married with two children. The clinical diagnosis was indirect inguinal hernia. He was operated and the specimen was sent to the department of Pathology for histopathological examination.

Gross examination showed a rudimentary uterus of size 6 × 2 × 1 cm with one side gonad of 2.5 × 1.5 cm, which was gray brown and soft to cut. Cut section of uterus shows endometrial cavity.(fig.1)

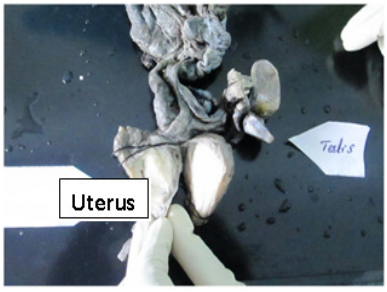

Figure 1: Hernial sac showing rudimentary uterus and one side gonad

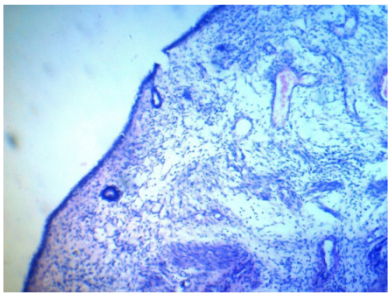

Figure 2: Endometria lining epithelium with underlying endometrial glands in primitive stroma

Microscopic examination of the uterus showed lining epithelium with underlying endometrial glands in primitive stroma (fig .2). Sections from the gonad showed semniferous tubules with intervening stroma shows leydig cell hyperplasia(fig.3) and epididymis.(fig.4)

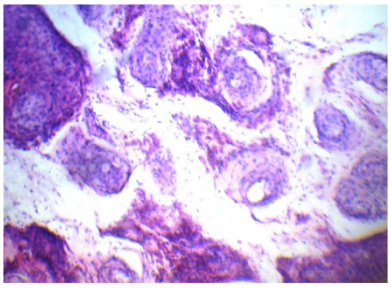

Figure 3: semniferous tubules with intervening stroma shows leydig cell hyperplasia

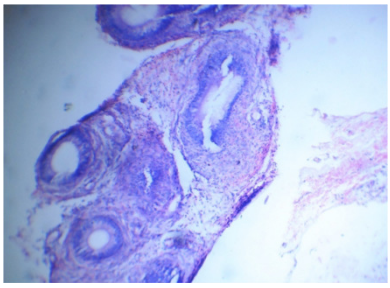

Figure 4: Showing epididymis

Discussion

PMDS is rare disorder and its prevalence is unknown. Manifests as autosomal recessive pattern due to Mutations in the AMH gene in 45% of cases, Mutations in the AMHR2 gene responsible for 40% of cases ([2]). In the remaining 15% of cases the cause is unknown. Types: Type 1 due to Mutations in the AMH gene and Type 2 due to Mutation in the AMHR gene([3]). The condition can come to attention because of a bulge in the inguinal canal of a genetically male infant due to herniation of the uterus or Occasionally the uterus is discovered during abdominal surgery for some other purpose in later childhood or adult life clinically presents either as male type (80-90%) or as female type (10-20%).Male type present as unilateral cryptorchidism with a contralateral inguinal hernia. Female type (10-20%) present as bilateral cryptorchidism([5]). Male type is divided into first type and second type. The first type also called as hernia uteri inguinalis([4]) present as descended testis and herniation of the ipsilateral corner of the uterus and the ipsilateral fallopian tube into the inguinal canal. The second type is crossed testicular ectopia, which is characterized by herniation of both testes and the entire uterus and both fallopian tubes.

Conclusion

PMDS is a rare form of male pseudo-hermaphroditism characterized by the presence of Mullerian duct structures in an otherwise phenotypically, as well as genotypically, normal man. The patient with PMDS has unilateral or bilateral cryptorchidism and is usually assigned to the male sex at birth without hesitation. Since patients are phenotypically male, the diagnosis is usually not suspected until surgery is performed for cryptorchidism or hernia repair.

Hernia uteri inguinalis is type I of the male form of PMDS, characterized by one descended testis and the herniation of the ipsilateral corner of the uterus and fallopian tube into the inguinal canal. In order to prevent further complications such as infertility and malignant change, the surgeon should be aware of PMDS while dealing with patients who present with unilateral or bilateral cryptorchidism.

In summary, in cases of unilateral or bilateral cryptorchidism associated with hernia, as in our patient's case, the possibility of PMDS should be kept in mind.

Consent

Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

References

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