Persistent Mullerian duct syndrome: Rare but important aetiology of an inguinal hernia and cryptorchidism in boys

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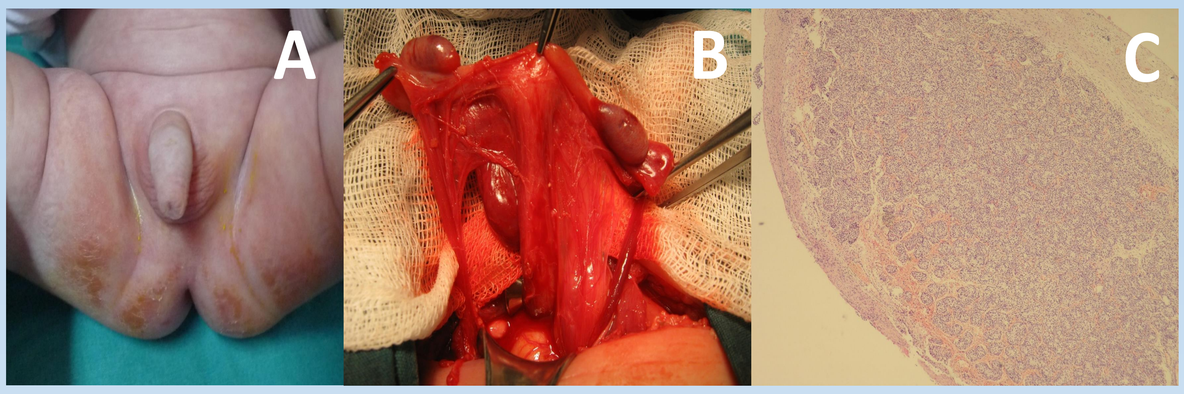
Background

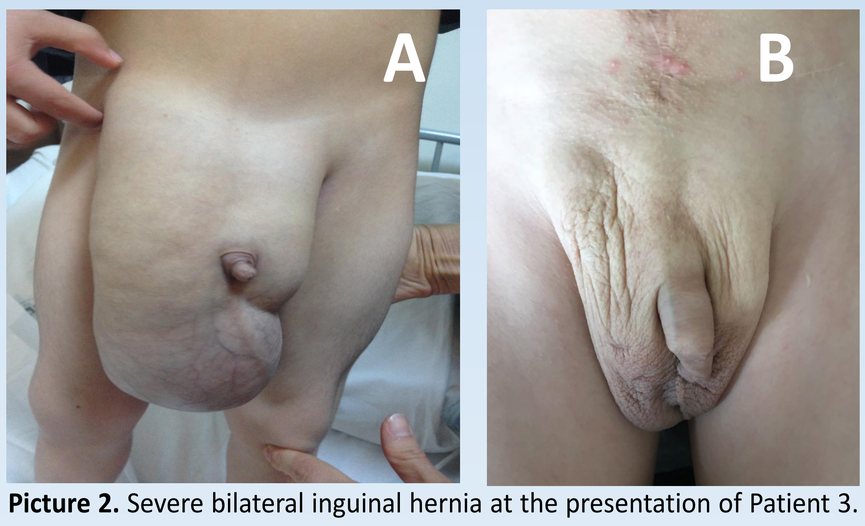
Anti-Mullerian hormone (AMH), secreted by immature Sertoli cells, provokes the regression of male fetal Mullerian ducts. Homozygous loss of function mutations in genes encoding AMH (*AMH*) or its receptor (*AMHRII*) lead to the persistent Mullerian duct syndrome (PMDS) which is characterized by the presence of uterus, fallopian tubes, cervix and vagina in otherwise normally virilized 46,XY males. Typical clinical features along with plasma AMH levels and genotyping establish the diagnosis of PMDS. However, surgical management and long-term follow up of these patients is challenging.

Case Reports

We present 4 cases with PMDS presented with cryptorchidism and inguinal hernia **(Table)**. Inguinal exploration for cryptorchidism or inguinal hernia by laparoscopy revealed incidental findings of Mullerian remnants **(Picture 1, 2, 3)**. Biopsies taken from gonads in each patient revealed testicular tissue with variable degree of immaturity. The biopsy of Mullerian remnants did not reveal any malignancy. All patients were genotypically male. Clinical and genetic characteristics of the patients are presented in Table. We opted a single stage laparotomy to split the fundus of the uterus in the midline to release testes and to avoid damaging vas deferens or the deferential artery during orchidopexy. The postoperative course was uneventful.



Picture 1. Normal male external genital appearance of patient 1 (A). Mullerian remnants (uterus, fallopian tubes) with normal appearing testes observed during operation (B), normal semineferous tubules and microscopic structure of testis biopsy of patient 1 (C).



Picture 2. Severe bilateral inguinal hernia at the presentation of Patient 3. Pre-op (A) and post-op (B) appearances of external genitalia.

Picture 3. Surgical approach to spare neurovascular bundle of testicular structures in patient 4. Uterus-like remnant (A) is dissected at the middle and mobilized *en-block* to repair cryptorchidism and inguinal hernia (B).

Table. Clinical and genetic characteristics of the patients with PMDS

Patient	Age of presentation	Signs/symptoms at presentation	Other anomalies	FSH (mIU/mL)	AMH concentration	Gene/mutation	Reference
I	26 days	Bilateral inguinal hernia, bilateral cryptorchidism	-	2.47	<0.1 ng/ml (45-266)	*AMH* c.G301A, p.(Gly101Arg) (homozygous)	*Three families from West Turkey, Pakistan and India
2	8 yr 9 mos	Unilateral inguinal hernia, bilateral cryptorchidism	-	0.8	138.5 ng/ml (33-60.2)	*AMHRII* c.1510C>T, p.(Arg504Cys) (homozygous)	*Two PMDS patients from Germany and Italy
3	1 yr 6 mos	Bilateral inguinal hernia, bilateral cryptorchidism	-	0.46	0.02 ng/ ml (45-266)	*AMH* c.1577C>T, p.(Cys526Phe) (homozygous)	**novel**
4	3 yrs 8 mos	Bilateral inguinal hernia, bilateral cryptorchidism	-	2.88	0,02 ng/ml (45-266)	*AMH* c.1673G>A, p.(Gly558Asp) (homozygous)	**novel**

*Picard JY, et al. Sex Dev 2017;11:109–125

Conclusion

PMDS serves as a remarkable management dilemma due to 2 main complications namely infertility and cancer. The surgeon should bear in mind that a cryptorchidism and an inguinal hernia in presence of Mullerian duct structures in male phenotypes should suggest PMDS. The management of PMDS cases is far more complicated than the ones with isolated cryptorchidism and/or inguinal hernia. Long-term reproductive and endocrinological surveillance is warranted.