PERSISTENT MULLERIAN DUCT SYNDROME - CASE REPORT -

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Introduction

The Persistent Mullerian Duct Syndrome (PMDS) is a rare disorder of sex development characterized by the persistence of Mullerian derivatives in otherwise normally virilized man. All affected subjects are genotypically and phenotypically male.

It results from mutations in the gene encoding the anti-Mullerian hormone (45%) or its receptor (40%), and has an autosomal recessive inheritance.

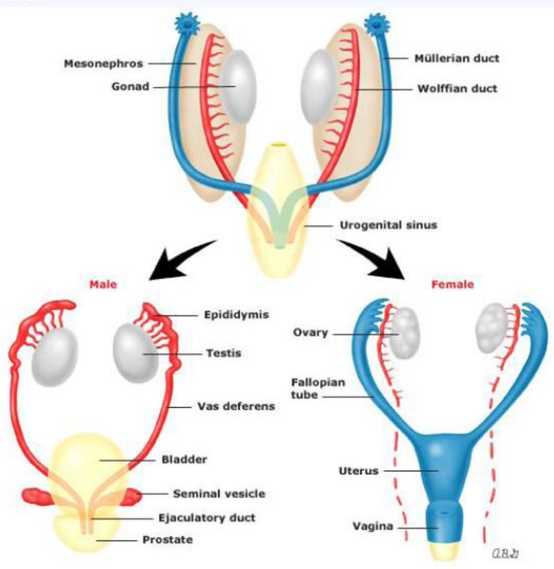

Fig 1 – Development of the internal genitalia[1]

Case report

♂, 62 years old, referred to our Endocrinology unit for evaluation after the incidental discovery of uterus and bilateral Fallopian tubes during emergency laparotomy for acute peritonitis.

Medical history:

→ Bipolar disorder diagnosed at age 33

→ Crohn's disease diagnosed after the episode of peritonitis

Medications:

→Risperidone, biperiden, lorazepam, alprazolam

Birth and developmental history:

→Eutocic birth at home without complications; appropriate pubertal development; no history of gynecomastia; without erectile dysfunction or changes in libido, but childless although there was childbearing intention.

Family history:

→3 brothers, all with children

Physical examination:

→ Weight: 99Kg; Height:165cm; BMI: 36.7kg/m², arm span 170 cm, upper-to-lower segment ratio 0.94; male phenotype with normal penis but missing testicles in the scrotum; no dysmorphisms; no gynecomastia; normal hair distribution and male pattern voice.

Case report

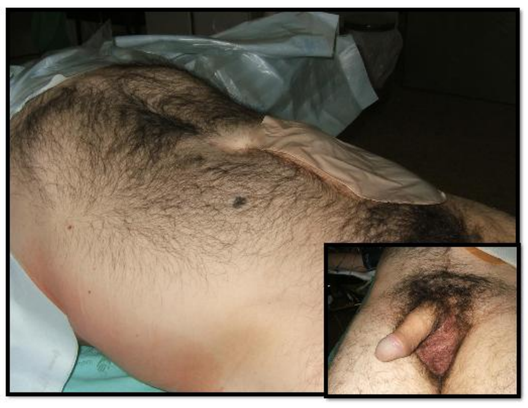

Fig. 2 – Normal male habit and detail showing empty scrotum

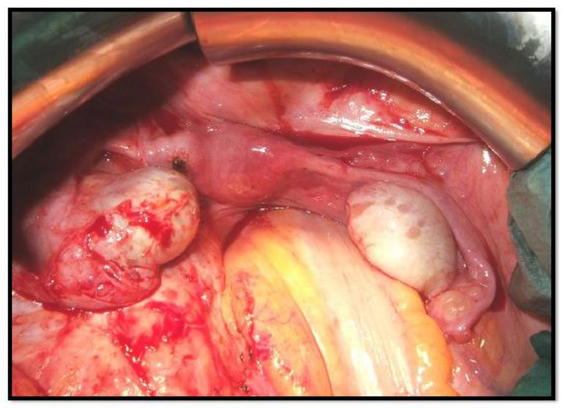

Fig. 3 – Intraoperative photograph showing gonads, uterus and fallopian tubes

Investigation

→ MRI: "uterus in median location with normal anatomical pattern, inserted in the region of the seminal vesicles; prostate and seminal vesicles with normal characteristics; gonads in ovarian topography with uncharacteristic signal behavior for ovarian tissue, suggesting ovotestes"

→ Karyotype 46, XY

→ Normal bone densitometry

Table I. Analytical results

FSH	42.5 mUI/mL	(1.5-12.4)
LH	16.2 mUI/mL	(1.7-8.6)
Free testostosterone	5.7 pg/mL	(13-40)
Dihydrotestosterone	14.8 pg/mL	(300-850)
Δ4-androsthenedione	1.7 ng/mL	(0.8-2.1)
17-hydroxyprogesterone	1.1 ng/mL	(0.2-2.5)
Estradiol	40 pg/mL	(7.6-42.6)
Alpha-fetoprotein	3.3 ng/mL	(<10.9)
B -HCG	<1 IU/L	(<1)
Cortisol	15 µg/dL	(5-25)
TSH	0.9 µUI/mL	(0.4-4.4)
Free T4	1.2 ng/dL	(0.9-1.8)
Prolactin	4.8 ng/mL	(2.1-17.7)
PSA	0.88 ng/mL	(<4.5)

The patient allowed the removal of only one gonad.

Histology → atrophic testicular tissue, scattered microcalcifications, without signs of malignancy.

Discussion

Early diagnosis and treatment of PMDS can reduce the risk of degeneration and testicular malignancy associated with prolonged cryptorchidism. Surgical replacement of the gonads in the scrotum can not ensure fertility if there is abnormal connection of the testicles to the excretory ducts.

References: 1) Normal sexual development in www.uptodate.com 2) Persistent Mullerian duct syndrome in www.orpha.net 3) Current concepts in disorders of sexual development, Gonul Olcal, J Clin Ped Endo, 2011, 3 105-114 4) Persistent mullerian duct syndrome: a case report and review of the literature, Vaibhab J Nayak et al, International Journal of Applied and Basic Medical Research, 2014, vol 4, issue 2 5) Disorders of sex Development: An update, Harry Ostrer, J Clin Endocrinol Metab, 2014 99:1503-1509

17 ECE
173--EP
Reproduction, endocrine disruptors and signalling
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DOI: 10.3252/pso.eu.17ece.2015
Poster presented at: ECE2015 16–20 May 2015 DUBLIN, IRELAND 17th EUROPEAN CONGRESS OF ENDOCRINOLOGY
PosterSessionOnline.com