Persistent Mullerian Duct Syndrome, Types I and II

Alternative Names
PMDS
Pseudohermaphroditism, Male Internal
Hernia Uteri Inguinale
Persistent Oviduct Syndrome
Female Genital Ducts in otherwise Normal Male
Mullerian Derivatives, Persistent

WHO International Classification of Diseases
Congenital malformations, deformations and chromosomal abnormalities

OMIM Number
261550

Mode of Inheritance
Autosomal recessive with male sex limitation

Gene Map Locus
19p13.3-p13.2, 12q13

Description
The persistent Mullerian (paramesonephric) duct, or the uterine hernia, syndrome is a rare disorder of male sexual development. It is characterized by the persistence of the uterus, fallopian tubes and upper vagina in otherwise normally virilized boys. Despite the normal male genotype and the subsequent normal development of fetal testis, Mullerian structures do not regress due to failure in production or in response to the anti-Mullerian hormone. Since the secretion and action of testosterone is not affected, the Wolffian (mesonephric) duct derivatives and the external genitalia of the fetus progress in the normal male direction. An intersex condition is, therefore, not usually suspected but the malformation is incidentally detected during operative treatment of associated abnormalities such as inguinal hernia and descended testis, generally in the first year of life.

The diagnosis of persistent Mullerian duct syndrome is often established when a uterus and/or fallopian tube is found along with undescended testes in a genotypically and phenotypically normal male child during repair of inguinal hernia. The relative risk of testicular tumor is not increased in patients with persistent Mullerian duct syndrome if orchiopexy is performed before two years of age. The aetiology of persistent Mullerian duct syndrome is poorly understood. Some studies suggest that inheritance which may be either X-linked or autosomal recessive with male sex limitation is a possible cause of the disorder. The observation that most cases of persistent Mullerian duct syndrome are bilateral is noteworthy. Anti-Mullerian hormone is secreted by the Sertoli cells of the fetal testis and acts ipsilaterally. The sensitivity of Mullerian duct to this hormone is present only during the ambisexual stage of the embryonic period.

Molecular Genetics
A mutation in the genes encoding anti-Mullerian hormone or the AMH receptor leads to the two forms of persistent Mullerian duct syndrome, referred to as type I and type II, respectively.

Epidemiology in the Arab World

Kuwait
Naguib et al. (1989) reported an Arab Bedouin family including four males (2 brothers and 2 of their maternal uncles) with uterine hernia syndrome. All had a male chromosome constitution and phenotype, inguinal hernia, cryptorchidism, and persistence of Mullerian derivatives. Histopathological studies confirmed the presence of both testicular tissue and Mullerian derivatives. The presence of two affected brothers and two affected maternal uncles suggested X-linked inheritance, but the consanguinity of the parents of the maternal
uncles suggested autosomal recessive inheritance.

In 1990, Mahfouz et al. described two phenotypically normal unrelated males with persistent Mullerian duct structures and crossed testicular ectopia during routine herniorrhaphy. Both had a normal 46 XY karyotype. In each case, the vascular supply to the ectopic testis originated from the appropriate ipsilateral side.

**Oman**

Rizk et al. (1998) reported an 18-month old Omani boy with right-sided inguinal hernia and bilateral cryptorchidism. The parents were not consanguineous and family history was unremarkable. At operation, there was an unusual hernial sac with a very wide neck connecting to the pelvic peritoneum and containing a structure that resembled an infantile uterus with two fallopian tubes interposed between two gonads lying above each other on the right side of the pelvic peritoneum. Postoperative recovery was uneventful. Karyotyping was 46 XY.

**United Arab Emirates**

Between 1993-2002, El-Gohary (2003) used both diagnostic and operative laparoscopy in the management of five cases of persistent Mullerian duct syndrome. Two siblings from two different families accounted for four of the cases. Their age at presentation ranged from 7 months to 5 years of age. They presented with cryptorchidism and inguinal hernias. The impalpable testes were on the left in three, on the right in one and bilateral in one. The latter case had been managed previously in another hospital by an open technique, and the diagnosis was missed. Transverse testicular ectopia was present in two unrelated boys. All the cases were managed by splitting the uterus in the midline and then bringing the testis with the vas and attached uterine tissue into the scrotum. Three of the five cases were managed laparoscopically. Follow-up of 6 months to 10 years showed satisfactory results in four of the five cases.

[See also: Oman > Rizk et al., 1998].

**References**


**Contributors**

Ghazi O. Tadmouri: 11.5.2005