PERSISTENT MÜLLERIAN DUCT SYNDROME IN TWIN BROTHERS CAUSED BY A NOVEL MUTATION IN THE AMHR2 GENE

KidZ Health Castle

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Sex differentiation, gonads and gynaecology or sex endocrinology

- ✓Persistent Müllerian Duct Syndrome (PMDS) is a rare form of recessive autosomal inherited sex development disorder.
- ✓Normally masculinized XY males can present with bilateral cryptorchidism or unilateral cryptorchidism associated with an inguinal hernia.
- ✓Most frequent genetic causes are mutations in Anti-Müllerian Hormone (AMH) & AMHR 2 genes, situated respectively on chromosome 19 (13.3p19) & chromosome 12 (13.q12).

Our aim is to report the different clinical (genital) presentation of a novel mutation in the *AMHR 2* gene in monochorionic diamniotic twin boys

Healthy Turkish, consanguineous parents
Two older female siblings with unexplained retinitis pigmentosa
Eventless MCDA twin pregnancy
Delivery at 36 weeks of pregnancy with normal birth weight and length
Bilateral cryptorchidism without penile abnormalities
Ultrasound: intra-abdominal testes suspected

Examination at referral (14 months old)

Normal scrotum
Normal penile length
No inguinal hernia
Synophris and slight hypertrichosis

Body weight (kg)	7.93 (-0.86 SDS)
Body length (cm)	79.5 (+0.18 SDS)
Serum testosterone* (µg/L)	5.98
Serum dihydrotestosterone* (µg/L)	0.50

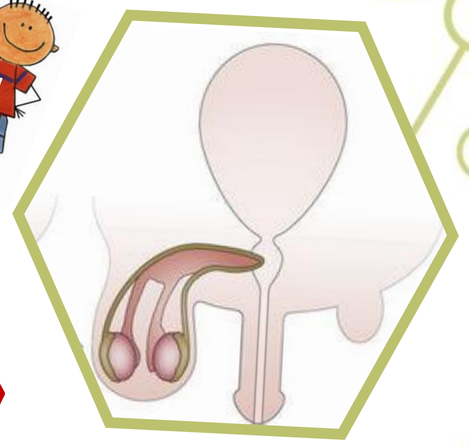
Underdeveloped scrotum
Normal penile length
No inguinal hernia
Synophris and slight hypertrichosis

Body weight (kg)	7.93 (-0.86 SDS)
Body length (cm)	79.5 (+0.18 SDS)
Serum testosterone* (µg/L)	6.21
Serum dihydrotestosterone* (µg/L)	0.56

* After hCG administration

46, XY karyotype
Likely pathogenic homozygous variant in AMRH 2 gene: c.1473>G p.(Asp491Glu)

A novel, similar mutation in the Anti-Müllerian Hormone Receptor 2 gene showed a crossed testicular ectopia and female type of Persistent Müllerian Duct Syndrome in non-identical twin boys.



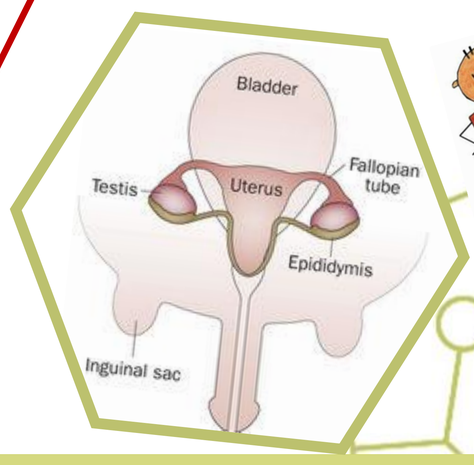


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Figure: Hutson J, Grover S, O'Connell M and Pennell S. Malformation syndromes associated with disorders of sex development. *Nature Reviews Endocrinology* volume10, pages 476–487(2014)

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