A novel mutation of the AMH in an Egyptian male with persistent Mullerian duct syndrome

Inas Mazen1, Mona El Gammal1 and Mohamed Abdel Hamid2
1Clinical Genetics Department, 2Molecular Medical Genetics Department, National Research Centre, Cairo, Egypt

We have nothing to disclose

Background:

- PMDS is a relatively rare autosomal recessive disorder of sex development (DSD)
- Characterized by the presence of Müllerian duct derivatives in 46, XY phenotypic males
- PMDS is due to mutations in the AMH gene or its type II receptor gene AMHR2
- To date, more than 50 different mutations of AMH gene have been reported.

Case report:

- Here, we report a novel mutation of AMH in an Egyptian patient with PMDS.
- A 3-year-old male presented with bilateral cryptorchidism and normal male external genitalia.
- The patient’s uncle had infertility, bilateral cryptorchidism and very low serum AMH (<0.1 ng/ml).
- A laparoscopic surgery revealed a uterus and fallopian tubes.
- Serum AMH was very low (0.1 ng/ml).

Genetic analysis of AMH gene showed a homozygous novel frameshift mutation c.203delC (p.L70fs*7) in exon 1.
- This mutation is predicted to result in early truncated protein.
- Both parents were heterozygous for the mutation.

Conclusion:

PMDS should be included in differential diagnosis of cryptorchidism.

References: