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

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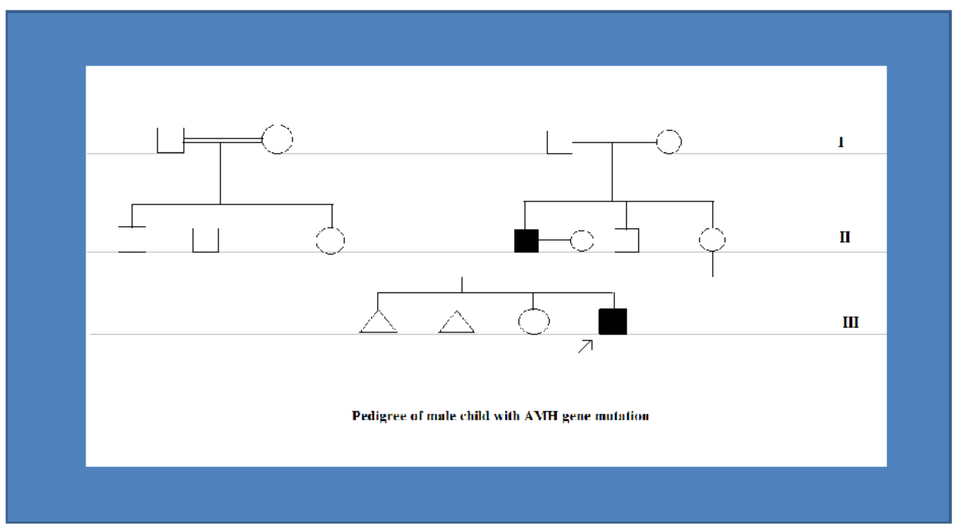
We have nothing to disclose

## 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.



Pedigree of male child with AMH gene mutation

Genetic analysis of AMH gene showed a homozygous novel frameshift mutation c.203delC (p.L70Cfs*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.**

## 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 (<0.1ng/ml).
- A laparoscopic surgery revealed a uterus and fallopian tubes.
- Serum AMH was very low (0.1ng/ml).

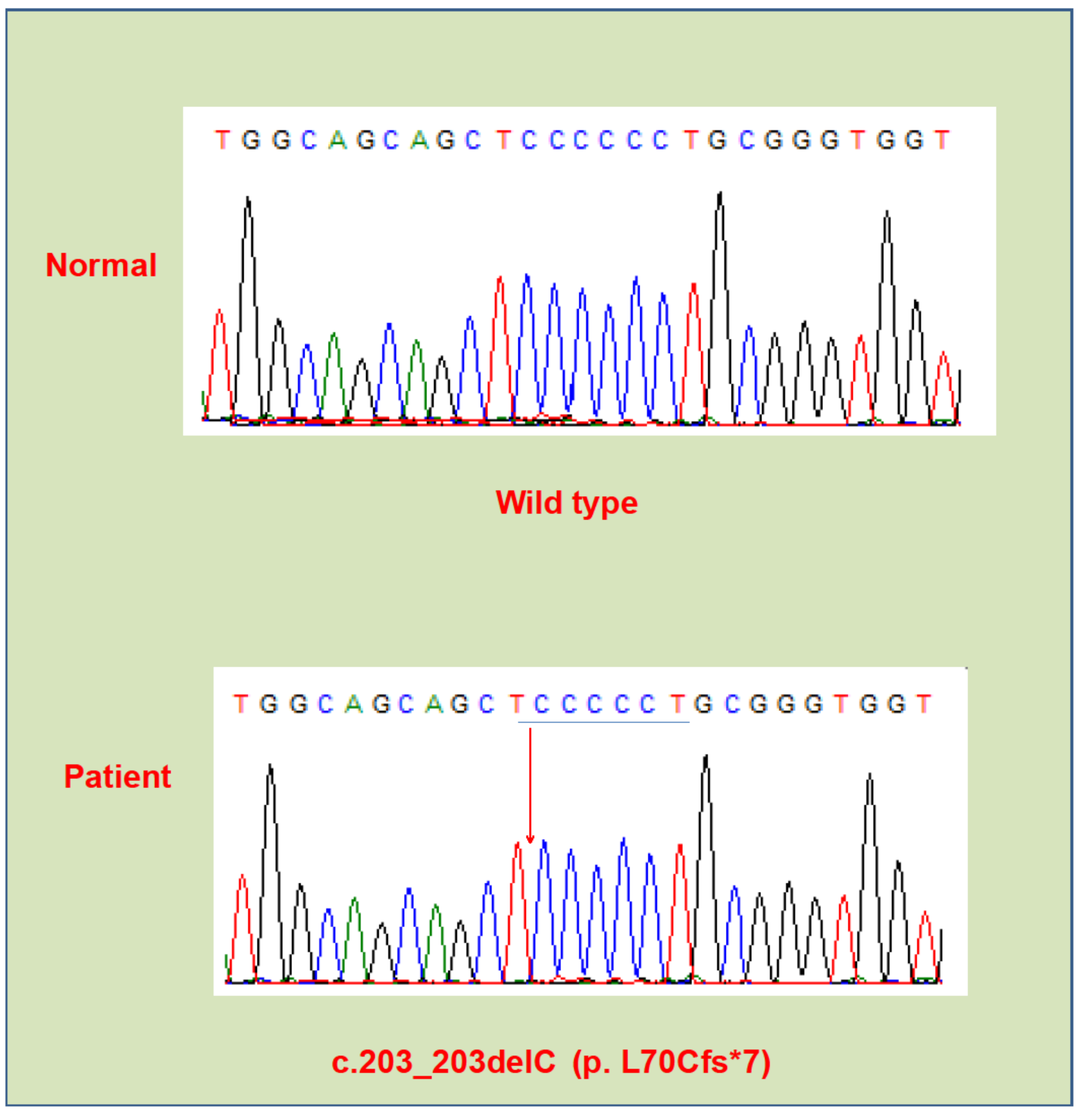



Figure 1: Portion of the sequencing electrophoregram showing the novel frameshift mutation identified in exon 1 of the AMH gene in our patient. The arrow indicates the mutation site.

## Refrences:

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