Aromatase deficiency due to a novel CYP19A1 mutation in an Egyptian patient with ambiguous genitalia

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There is nothing to disclose…..

Back ground

- Mutations in *CYP19A1* gene have been described in both females and males.
- to date only 20 cases with aromatase deficiency have been reported.
- In newborns, aromatase deficiency should be considered in the etiology of 46,XX DSD, after ruling out congenital adrenal hyperplasia.

Case report

- Here we report a patient who was presenting at the age of 20 years old reared as boy, he was a product of consanguineous parent.
- He had no palpable testis, hypoplastic scrotum, phallus was penis like =3 cm, there was a penoscrotal hypospadias.
- His karyotype was 46,XX and SRY was negative. Pelvic sonar showed a small hypoplastic uterus and no testis could be identified

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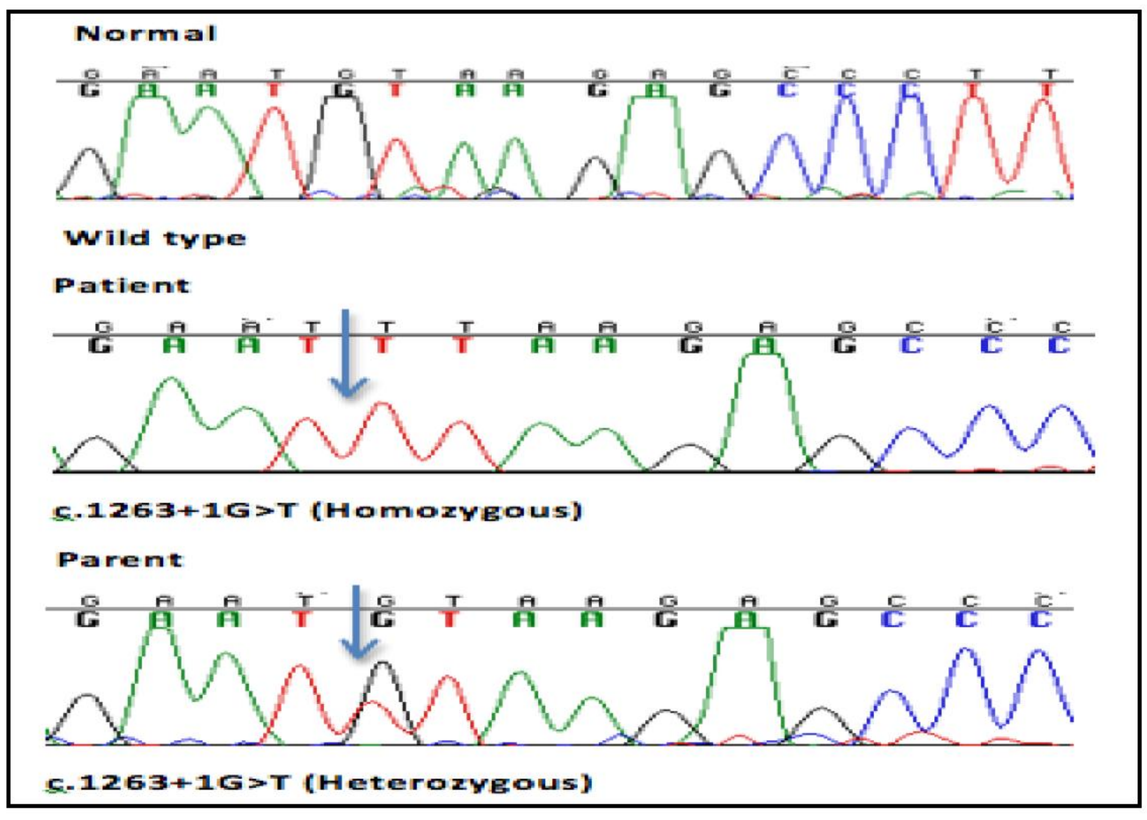
- Testosterone and its precursors were normal excluding congenital adrenal hyperplasia, pathology of gonadal biopsy showed ovarian stroma negative for oocytic follicle.

Molecular study

- The entire coding region of the *CYP19A1* was amplified and directly sequenced using Sanger Sequencer.
- A novel splice site mutation in the donor splice site of exon 9 was identified in our patient, c.1263+1G>.
- The mutation was found in the homozygous form in the patient and both parents were heterozygous for the mutation.
- The mutation was not found in 200 normal chromosomes of Egyptian origin and was predicted to be disease causing by Mutation Taster, PolyPhen2 and SIFT.



Conclusion

- This is the first report of such a rare disorder in an Egyptian patient with 46,XX DSD emphasizing the importance of mutational analysis of *CYP19A1* gene for appropriate management and better choice of sex

References

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