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Oestrogen and Oestrogen Receptors: Consequences and Implications in Human Mutations

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Background

Oestrogen action through the Oestrogen Receptor 1 (ESR1), especially ESR1A influences many physiological processes including reproduction, cardiovascular health, bone integrity, cognition and behaviour. We illustrate the consequences of an ESR1 mutation in a family with three affected members with oestrogen insensitivity syndrome.

Family Study

The index patient is a 21-year-old female, born to consanguineous parents. At 16 years she had absent breast development, primary amenorrhoea and hirsutism. Clinical assessment showed height 170cm (+1.09SDS), weight 70kg (+1.45SDS), Tanner stage B1A3P5, acne and severe hirsutism (Ferriman-Gallwey score 22). Bone age was 11 years. Laboratory studies revealed serum oestradiol 9526pmol/l (N 110-280pmol/L), elevated gonadotrophins-FSH 15 IU/L (N 2-11), LH 18 IU/L (N 2-9) with 46, XX karyotype and multicystic ovaries on pelvic ultrasound. Genetic studies demonstrated a c.1181G>A mutation in the fifth coding exon of ESR1 (Arg394His) in the ESR1 ligand-binding domain.

Both parents and one healthy sister are heterozygous for the mutation while an older sister aged 25 years and a brother aged 18 years are homozygotes. They also had pubertal delay with hypergonadotrophic hypogonadism. The brother has bilateral cryptorchidism, Tanner stage G1A1P1, with LH 13, FSH 55IU/L, serum AMH 0.6ng/ml (N 1.5-51) and Inhibin B: 5pg/ml (N >100). The older sister had hirsutism and primary amenorrhoea, Tanner stage B1A3P5, LH: 21 IU/L, FSH: 20IU/L. E2: 9476pmol/l.

Conclusion

This family study confirms the important role of oestrogen in the male as well as the female.