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Placental Aromatase Deficiency

Placental Aromatase Deficiency is characterized by decreased estrogen levels and increased androgens. It presents with ambiguous genitalia, primary amenorrhea, masculinization, tall stature, and/or osteoporosis. Management strategies include hormone replacement therapy, calcium and vitamin D supplementation, and surgery.



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Characteristics

Decreased Estrogen

Down-arrow Easter-egg

Aromatase deficiency involves a mutation of the CYP19A1 gene, which encodes for the enzyme aromatase. The non-functional enzyme will lead to the inability to synthesize estrogen from androgens, which will lead to decreased estrogen levels.

Increased Androgens

Up-arrow Android-genie

As a result of deficient aromatase, androgens (e.g. testosterone and androstenedione) will accumulate.

Presentation

Ambiguous Genitalia

Question-mark-dress on Testes

In neonatal females, the genitalia is ambiguous despite normal development of internal genital organs.

Primary Amenorrhea

(1) wand with Amen-tampon

During pubescence, there is impaired maturation of secondary sexual characteristics. This presents as primary amenorrhea and virilization (e.g. hirsutism, severe acne) in females.

Masculinization

Mask with Large Beard

The mothers of affected fetuses can experience masculinization (i.e. maternal virilization) during pregnancy as the fetal androgens can cross the placenta. This may start at 12 weeks gestation and typically disappears after delivery.

Tall Stature

Tall Statue

During childhood, patients may present as tall-for-height and with osteoporosis (e.g. fractures following minimal trauma).

Osteoporosis

Ostrich-with-porous bones

Osteoporosis is generally a disease of postmenopausal women when estrogen levels decline. However, the low estrogen levels in this disease will result in poor bone density i.e. osteoporosis.

Management

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Hormone Replacement Therapy

Harmonica and Med-bottles

Treatment of aromatase deficiency includes both estrogen and progesterone HRT.

Calcium and Vitamin D

Calcium-cow and Viking-Daisy

In order to address the osteoporosis, which can present in both males and females, both calcium and vitamin D supplementation can be provided.

Surgery

Surgeon

Surgical correction of the ambiguous genitalia in young females may also be an option.