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.

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