

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.



PLAY PICMONIC

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

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.