5 Alpha Reductase Deficiency

5 alpha reductase deficiency is an autosomal recessive disorder caused by a mutation of the 5 alpha reductase gene. This enzyme normally converts testosterone to 5 alpha dihydrotestosterone (DHT) in peripheral tissues. Therefore, a defect in this enzyme leads to the inability to convert testosterone to DHT. DHT is a potent androgen that is necessary for the development of male external genitalia in utero. Individuals with a 5 alpha reductase deficiency are born with normal internal genitalia including testicles and Wolffian structures but usually have ambiguous external genitalia until puberty. During puberty around age twelve, it is hypothesized that rising testosterone levels are able to generate sufficient levels of DHT to undergo some masculinization including descending of the testes, growth of facial/body hair, deepening of the voice, and enlargement of penis. Individuals with this disorder typically have normal or slightly increased leutinizing hormone levels and often have normal testosterone/estrogen levels as compared to androgen insensitivity syndrome which has elevated LH, testosterone, and estrogen levels.



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Autosomal Recessive
Recessive-chocolate
This disorder is inherited in an autosomal recessive fashion.

Inability to Convert Testosterone to DHT
Testes-stereo blocked from Dyed-hydra-testes-stereo
This enzyme normally converts testosterone to 5 alpha dihydrotestosterone (DHT) in peripheral tissues. Therefore, a defect in this enzyme leads to the inability to convert testosterone to DHT.

Normal Internal Genitalia
Internal Testes
Individuals with a 5 alpha reductase deficiency are born with normal internal genitalia including testicles and Wolffian structures but usually have ambiguous external genitalia until puberty.

Ambiguous External Genitalia until Puberty
Question-mark-dress on Testes
DHT is a potent androgen that is necessary for the development of male external genitalia in utero. Therefore, individuals with 5 alpha reductase deficiency typically have ambiguous external genitalia until puberty.

Masculinization During Puberty with Growth of External Genitalia
Testes exposed with Pubic-hair
During puberty around age twelve, it is hypothesized that rising testosterone levels are able to generate sufficient levels of DHT to undergo some masculinization including descending of the testes, growth of facial/body hair, deepening of the voice, and enlargement of penis.

LH can be Increased
Up-arrow Luge
Individuals with this disorder typically have normal or slightly increased leutinizing hormone levels and often have normal testosterone/estrogen levels as compared to androgen insensitivity syndrome which has elevated LH, testosterone, and estrogen levels.

Normal Estrogen, LH, and Testosterone levels
Normal-bar Easter-egg, Luge, and Testes-stereo
Individuals with this disorder typically have normal or slightly increased leutinizing hormone levels and often have normal testosterone/estrogen levels as compared to androgen insensitivity syndrome which has elevated LH, testosterone, and estrogen levels.

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