Klinefelter's Syndrome

Klinefelter's syndrome is a genetic disorder, in which patients are born with at least one extra X chromosome, giving them a total of 47 chromosomes rather than 46. Patients are born male, with the genetic composition of XXY. They often display a eunuchoid body shape, gynecomastia, a female hair distribution pattern, and testicular atrophy. cognitive and developmental delays are seen. Patients often have trouble with language learning and reading at a young age, and also display motor delays. This includes achieving developmental milestones much later than normal (sitting, crawling, walking). Later in life, this can translate as inability to participate in sports, and trouble with academic success. Secondary to abnormal Leydig cell function, lab investigations will show decreased testosterone, increased LH and increased estrogen. Additionally, in these patients there is impaired gamete production and/or regulation.



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Pathophysiology

Male (XXY)
XXY-sign
Klinefelter's syndrome is a genetic disorder, in which patients are born with at least one extra X chromosome, giving them a total of 47 chromosomes rather than the 46. 47, XXY is the most common sex chromosome aneuploidy in males born with this syndrome.

Signs and Symptoms

Eunuchoid Body Shape
Tall-with-Long-extremities-and-Wide-hips
Individuals with Klinefelter's typically have a eunuchoid body habitus, with long extremities and wide hips. These patients are typically taller than average males of the same age. Patients with this condition display underdeveloped sexual organs, and specifically, because of testicular atrophy, male hormone production is deficient.

Gynecomastia and Female Hair Distribution
Man with Breast-tassels
Gynecomastia, or increased breast tissue, is present in about a third of affected individuals. Additionally, men with Klinefelter’s display female hair distribution, showing decreased facial and body hair growth.

Testicular Atrophy
Testicle @-trophy
Patients born with Klinefelter’s syndrome will develop microorchidism, or testicular atrophy. This testicular atrophy, where there is diminished size and function of the testicles, leads to a primary hypogonadism, characterized by low serum testosterone.

Dysgenesis of Seminiferous Tubules
Disc-genie Semi-tubes
Those with Klinefelter’s syndrome develop seminiferous tubule dysgenesis. This is a dysfunction by which the seminiferous tubules exhibit an abnormal cytoarchitecture and extensive hyalinization; the testes are small, with few spermatozoa formed.

Possible Developmental Delay
Developmental-bus
In Klinefelter’s syndrome cognitive and developmental delays are seen. Patients often have trouble with language learning and reading at a young age, and also display motor delays. This includes achieving developmental milestones much later than normal (sitting, crawling, walking). Later in life this can translate as inability to participate in sports, and trouble with academic success.

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Hypogonadism

Hippo-gonads

In Klinefelter’s syndrome there is diminished size and function of the testicles, leading to a primary hypogonadism. Hypogonadism described as diminished functional activity of the gonads, characterized by low serum testosterone in this syndrome. Additionally, in patients there is impaired gamete production and/or regulation.

Decreased Inhibin B

Down-arrow Inhibiting-chains on (B) Bee

Inhibin B is produced by the gonads and downregulates FSH synthesis and inhibits FSH secretion. Due to hypogonadism in these patients, lab workup will reveal decreased inhibin B.

Increased FSH

Up-arrow Fish

In Klinefelter's Syndrome, primary hypogonadism leads to low serum testosterone and decreased inhibin. A consequence of this decreased inhibin is that FSH synthesis is not downregulated, resulting in increased FSH levels.

Abnormal Leydig Cell Function

Abnormal Lady-bug

Leydig cells are found adjacent to the seminiferous tubules in the testes, and produce testosterone in the presence of LH. In Klinefelter's Syndrome, Leydig cells are dysfunctional, and this results in decreased testosterone, increased LH and increased estrogen.

Decreased Testosterone

Down-arrow Testes-stereo

In Klinefelter's Syndrome, primary hypogonadism leads to low serum testosterone, and consequently high serum FSH, LH, and estrogen.

Increased LH

Up-arrow Luge

In Klinefelter's Syndrome, primary hypogonadism leads to low serum testosterone, and consequently high serum FSH, LH, and estrogen.

Increased Estrogen

Up-arrow Easter-egg

In Klinefelter's Syndrome, primary hypogonadism leads to low serum testosterone, and consequently high serum FSH, LH. The increase in serum FSH up-regulates aromatase, which leads to increased conversion of testosterone to estrogen.

Barr body (inactivated X chromosome)

Bar Body

A Barr body is the inactive X chromosome in a female somatic cell, rendered inactive in a process called lyonization. The Lyon hypothesis states that in cells with multiple X chromosomes, all but one are inactivated during embryogenesis. Consequently in Klinefelter's Syndrome, or XXY, there is a single Barr body, with one normally functioning X chromosome.

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