Androgen Insensitivity (Complete)

Androgen insensitivity syndrome occurs when a person who is genetically male with 46 chromosomes and XY sex chromosomes has a defect in the androgen receptor so that the body cannot respond to androgens. The unresponsiveness of the body to the presence of androgenic hormones prevents masculinization of the male genitalia as well as the development of male secondary sexual characteristics. Therefore, individuals with complete androgen insensitivity appear female. These individuals have female external genitalia. However, they typically have a rudimentary vagina with an absent uterus and fallopian tubes due to the presence of anti-mullerian hormone secreted from the Sertoli cells of the testes. They also often have scant sexual hair. Because the testes form in an androgen-independent process during the embryonic stage of development, individuals with this syndrome still have testes that may be located intraabdominally or in the labia majora. Individuals with this syndrome have a higher risk for germ cell malignancies, and, therefore, the testes are often surgically removed. Additionally, there is a lack of feedback inhibition of the hypothalamic-pituitary-gonadal axis. Therefore, there are increased levels of luteinizing hormone and testosterone, which are aromatized into estrogen.

PLAY PICMONIC

Defect in Androgen Receptor
Broken Receptor
This syndrome is caused by a mutation in the androgen receptor gene. Therefore, the body cannot respond to androgens.

Phenotypically Female
Feminine Woman
The unresponsiveness of the body to the presence of androgenic hormones prevents masculinization of the male genitalia as well as the development of male secondary sexual characteristics. Therefore, individuals with complete androgen insensitivity appear female.

(46, XY)
46 XY Top
People with androgen insensitivity syndrome have a genetic composition of 46 chromosomes with XY sex chromosomes. Therefore, these individuals are genetically male.

Female External Genitalia
Bikini covering Female External Genitalia
The unresponsiveness of the body to the presence of androgenic hormones prevents masculinization of the male genitalia as well as the development of male secondary sexual characteristics. Therefore, individuals with complete androgen insensitivity are phenotypically female with female external genitalia.

Rudimentary Vagina
Rooted Vagina-violet
The Mullerian system, which includes the fallopian tubes, uterus, and upper portion of the vagina, typically regresses due to the presence of anti-mullerian hormone secreted from the Sertoli cells of the testes. Therefore, individuals with androgen insensitivity syndrome are born without fallopian tubes, a uterus, and a rudimentary vagina that ends as a blind pouch.

Scant Sexual Hair
Shaving Sexual Hair
Individuals with androgen insensitivity syndrome have diminished or absent sexual hair due to a lack of androgen response.

Testes Often in Labia Majora
Testes in Labia-leis
During the embryonic stage of development, testes form in an androgen-independent process that occurs due to the influence of the SRY gene on the Y chromosome. Therefore, individuals with this syndrome still have testes that may be located intraabdominally or may herniate into the labia majora.

Testes Surgically Removed to Prevent Malignancy
Testes Removed From Labia-leis and Malignant-man
Immature sperm cells in the testes are arrested at an early stage and do not mature since sensitivity to androgens is required for spermatogenesis. Individuals with this syndrome have a higher risk for germ cell malignancies, and the testes are often removed.

Increased Estrogen, Testosterone, and LH

Up-arrow Easter-Egg, Testes-Stereo, and Luge

Due to the androgen receptor defect, there is a lack of feedback inhibition of the hypothalamic-pituitary-gonadal axis. Therefore, there are increased levels of luteinizing hormone and testosterone, which are aromatized into estrogen.

Absent Uterus and Fallopian Tubes

Uterus with Fallopian Tubes Leaving

The Mullerian system, which includes the fallopian tubes, uterus, and upper portion of the vagina, typically regresses due to the presence of anti-mullerian hormone secreted from the Sertoli cells of the testes. Therefore, individuals with androgen insensitivity syndrome are born without fallopian tubes and a uterus and with a rudimentary vagina.