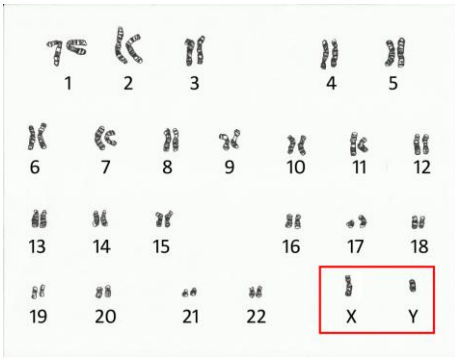
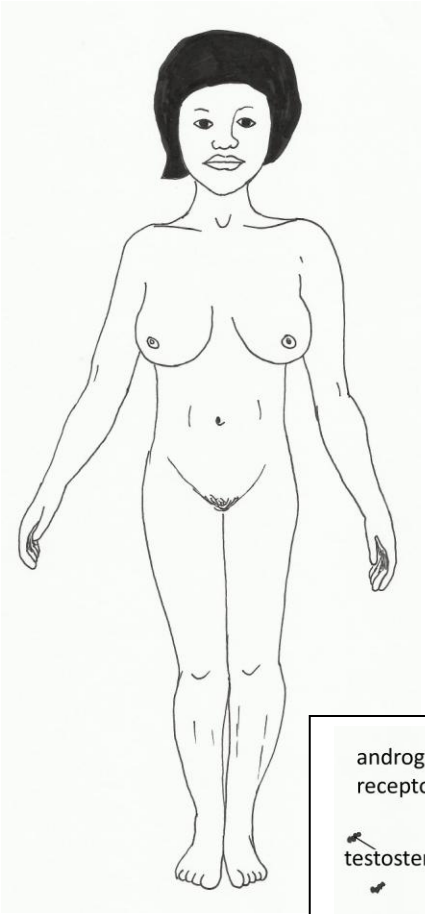
Androgen Insensitivity Syndrome (AIS) 46XY

occurs in approximately 1/20,400 baby "boys"





Living with Androgen Insensitivity Syndrome

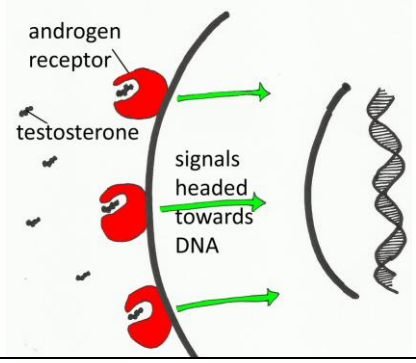
Individuals living with AIS have a Y chromosome containing the SRY gene. In response to the product of the SRY gene, the gonads develop into testicles.

The testicles are able to produce a normal amount of testosterone. However the body cannot detect and respond to the testosterone, and so the body develops as though testosterone is not present. The body is missing the receptor molecule that could bind the testosterone and pass along a signal to the DNA of the cell thereby activating the genes necessary for masculinizing the body. The genital tubercle cannot grow into a penis. The Wolffian duct cannot develop into the epididymis, the vas deferens, and the seminal vesicles. The typical masculine body shape and muscle development do not occur, male facial and body hair do not develop, and the voice does not change at puberty.

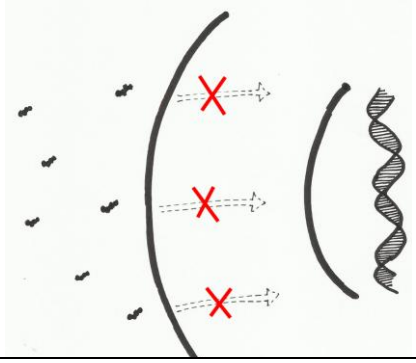
The person living with AIS develops a female appearance externally with a normal appearing clitoris and a shallow vagina and typical female sized breasts. The testicles may be in the abdomen, in the inguinal canal, or in the labia majora.

Many AIS individuals do not know that there is anything unusual about their body until they reach the age of puberty and fail to start menstruation. During medical evaluation of amenorrhea or infertility the presence of AIS may be discovered.

Most individuals with AIS have a female identity, and they might get married as women, and adopt children if they desire to build a family.



Testosterone binds to androgen receptors and signals are sent to genes in the DNA.



Testosterone with nowhere to bind is unable to send signals to genes in the DNA.