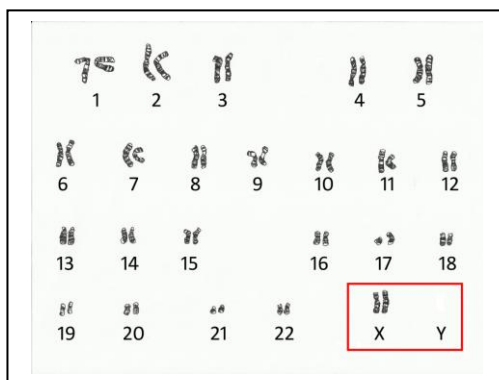


# Congenital Adrenal Hyperplasia (CAH) 46XX

occurs in approximately 1/10,000 to 1/18,000 babies



## Living with Congenital Adrenal Hyperplasia

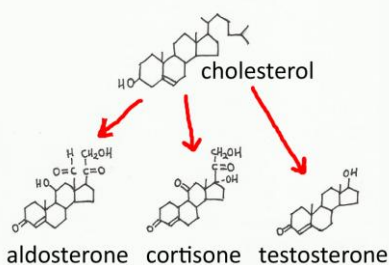
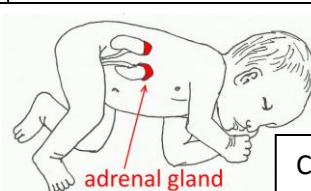
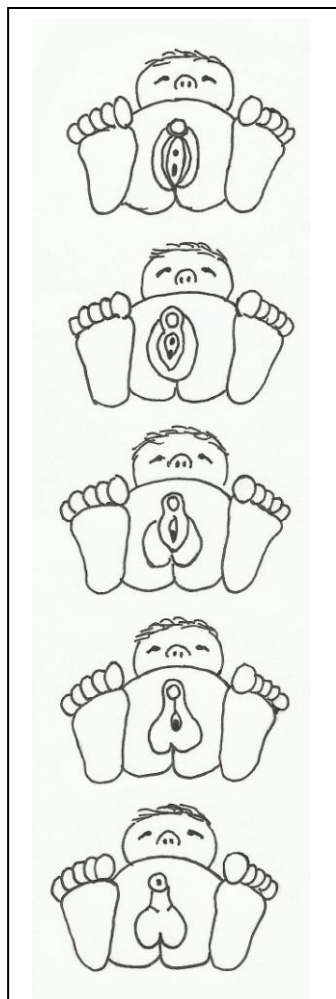
Individuals living with CAH have no Y chromosomes or testicles but they do have an adrenal gland that produces higher than normal levels of testosterone.

The molecule cholesterol is supposed to be used by the adrenal glands to make testosterone as well as other hormones. When enzymes are missing for making certain other hormones, more cholesterol is converted into testosterone than usual. It also means that the individual will need hormone treatment to provide the missing hormones that the adrenal gland cannot make.

The person with CAH is exposed to high levels of testosterone during the time when the internal reproductive structures and external genitalia are being formed. Under the influence of these high levels of testosterone, the differentiation process is therefore pushed away from the usual female developmental pathway expected in the absence of a Y chromosome, and instead pushed towards the male developmental pathway. The genital tubercle enlarges and becomes more like a penis and less like a clitoris. The labia become fused and become more like a scrotum. However, because the individual living with CAH does not have a Y chromosome or testicles, their internal reproductive organs consist of a uterus and a vagina.

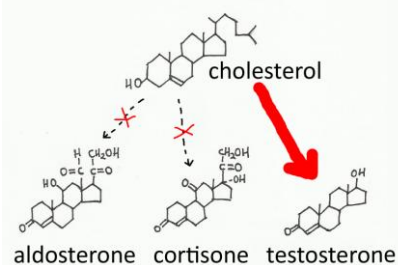
At birth, babies with CAH have ambiguous external genitalia that are not clearly male or female. Hopefully, their condition is correctly recognized right away so that doctors and parents know that the child is an XX individual with CAH and is not an XY male with a small penis and undescended testicles. In some cases, recognizing CAH means that life saving treatment with missing hormones can begin before it is too late.

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Inside the adrenal gland, cholesterol is converted into multiple hormones.

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When there are missing enzymes that make it impossible to make certain other hormones, more of the cholesterol gets converted to testosterone.