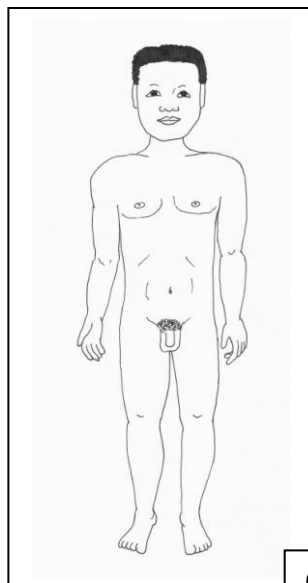
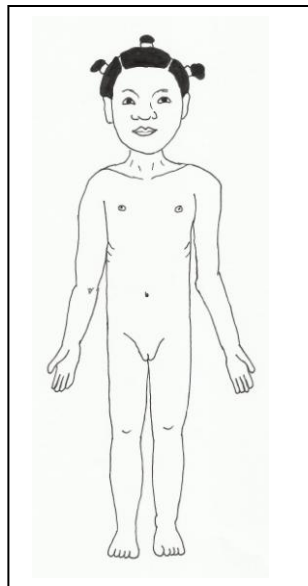
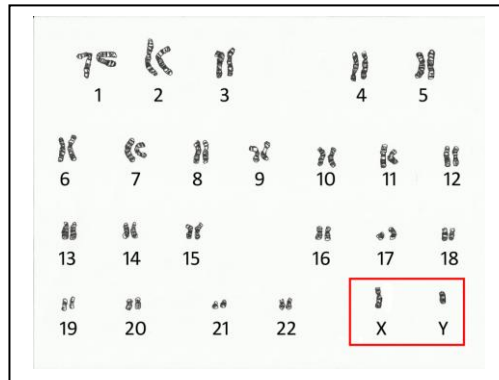


5-alpha-reductase deficiency 46XY

very rare except in a few particular villages scattered around the world



Living with 5-alpha-reductase deficiency

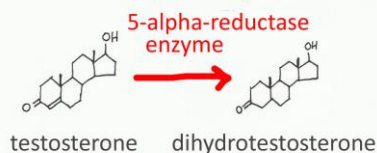
Individuals living with 5-alpha-reductase deficiency are missing an enzyme that is necessary in order for the body to convert testosterone to dihydrotestosterone. Without dihydrotestosterone the genital tubercle cannot turn into a penis during fetal development.

Amazingly - although the first growth stage of a penis does not occur in the womb - the second growth stage of the penis at puberty is able to occur. A child who has looked like a female at birth dramatically develops a male body at puberty.

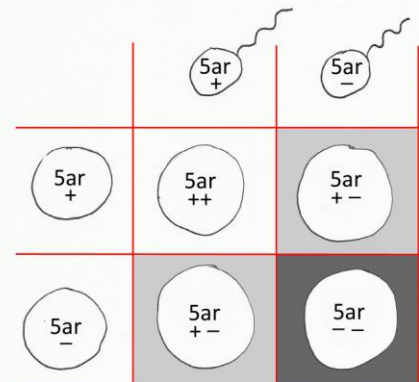
Since the testicles were inside the abdomen or pelvis all through early childhood, the testicles are likely to have been damaged by the high body temperature, and the individual is usually infertile.

Because this condition requires the presence of a rare mutation on both of the number 2 chromosomes, 5-alpha-reductase deficiency typically only occurs in families where people with the rare mutation have for many generations married distant relatives (as occurs in some small isolated villages).

In the Dominican Republic, in the village of Las Salinas, one out of every 90 males is born looking like a girl. Because this is relatively common in their village, the people of Las Salinas have learned to expect that some girls will become boys. The affected individuals usually adopt a male identity in adulthood, and are able to do so with the support and acceptance of family and friends.



Only if both egg and sperm carry a defective gene for the 5-alpha-reductase enzyme will a baby be produced with a deficiency syndrome. If one gamete has a good gene and one gamete has a defective gene the baby will be a carrier.



5ar⁺ = presence of normal 5-alpha-reductase gene
5ar⁻ = absence of normal 5-alpha-reductase gene