

Diversity in sex development, including Androgen Insensitivity Syndrome, Klinefelter Syndrome and Turner Syndrome.

Inheriting too many or too little copies of sex chromosomes (23rd pair) leads to atypical chromosome patterns, which can cause serious developmental complications. These complications can change hormone levels and lead to lifelong conditions affecting gender development.

Differences in sex development (DSD) is recognised as a group of rare conditions involving genes, hormones and reproductive organs, including genitals.

Some people have a chromosome pattern other than the usual XY or XX. They may have one X chromosome (XO), or they may have an extra chromosome (XXY).

Their internal and external sex organs can be either male or female, but they may not go through a full physical development at puberty. For example, a child with female sex organs may not start having periods.

Androgen Insensitivity Syndrome

Androgen Insensitivity Syndrome (AIS) is a condition that affects sexual development before birth and during puberty. People with this condition have one X chromosome and one Y chromosome in each cell. In people with androgen insensitivity syndrome, the body's cells and tissues are unable to respond to certain male sex hormones (called androgens) that are important for normal male sexual development before birth and during puberty. As a result, affected individuals may have external sex characteristics that are typical for females or have features of both male and female sexual development.

AIS is a condition where individuals with a typical male (XY) chromosome pattern have a reduced or absent response to androgens, the male sex hormones, due to androgen receptor dysfunction. There are three forms of AIS; complete, partial, and mild.

Complete androgen insensitivity syndrome occurs when the body does not respond to androgens at all. People with this form of the condition have external sex characteristics that are typical of females. Affected individuals do not have a uterus. They have male internal sex organs (testes) that are undescended, which means they are located in the pelvis or abdomen instead of outside the body. As such, affected individuals do not menstruate and are unable to conceive a child (infertile). People with complete androgen insensitivity syndrome also have sparse or absent hair in the pubic area and under the arms.

The partial and mild forms of androgen insensitivity syndrome occur when the body's tissues are partially sensitive to the effects of androgens. People with partial androgen insensitivity can have genitalia that look typical for females, genitalia that have both male and female characteristics, or genitalia that look typical for males. People with mild androgen insensitivity are born with male-typical sex characteristics, but they are often infertile and tend to experience breast enlargement at puberty.

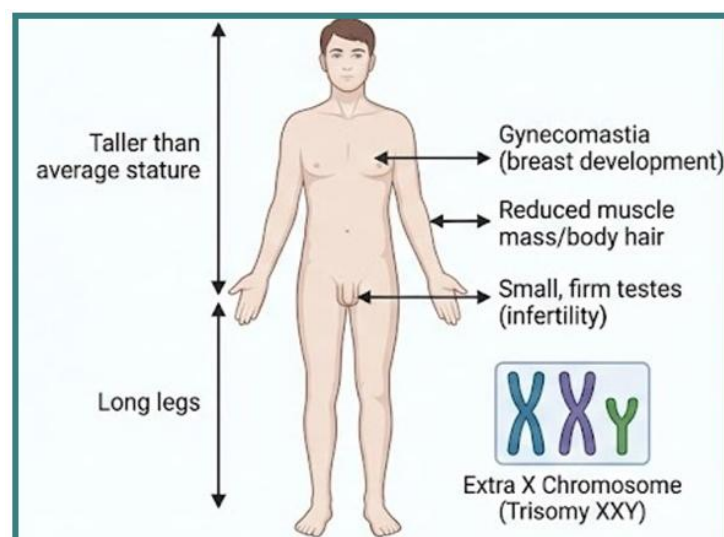
AIS is inherited with an X-linked recessive pattern. The gene associated with this condition is located on the X chromosome, which is one of the two sex chromosomes. In males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females (who have two X chromosomes), a variant would have to occur in both copies of the gene to cause the disorder. Because it is unlikely that both copies of a gene would be altered, males are affected by X-linked recessive disorders much more frequently than females.

Klinefelter Syndrome

Klinefelter syndrome is a genetic condition in which **males** inherit an additional X chromosome (XXY). Symptoms may include breast growth, infertility, osteoporosis and learning difficulties. Treatments usually involve physical and emotional therapy, as well as hormone replacement.

Boys and men with Klinefelter syndrome are still genetically male and often will not realise they have this extra chromosome, until later in life. Research has shown that the average intelligence (e.g. IQ) of boys with Klinefelter Syndrome is lower than that of boys with the usual number of chromosomes. Approximately 75% of boys with Klinefelter's have difficulty learning to read and will require special help on a one-to-one basis at school. This is possibly related to the particular difficulties that many of them have with short attention span, poor memory and difficulty in keeping things in the right order.

Boys with Klinefelter Syndrome often grow faster than usual, especially in the length of their legs. On average, a man with Klinefelter Syndrome will be about 186cm (6ft 2in) tall. Puberty usually occurs at the normal time and usually progresses normally. However, in some cases the growth of the penis may slow down towards the end of puberty, and the facial, pubic and underarm hair may be quite sparse. A mild degree of breast development (gynaecomastia) can occur in about two-thirds of boys with Klinefelter Syndrome. Men with Klinefelter Syndrome do not usually produce sperm, and so they have always been considered to be infertile, except in very rare cases.



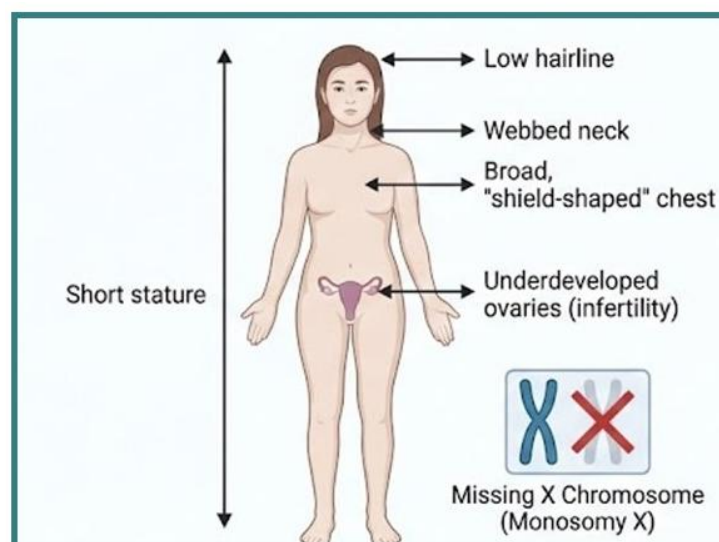
Turner Syndrome

Turner syndrome is a rare genetic condition that affects **females** when one of the X chromosomes is completely or partially missing (XO). Therefore, a girl born with only one X chromosome is known to have Turner Syndrome.

This chromosomal difference can lead to a range of physical, developmental, and medical challenges, including short stature, delayed puberty, heart conditions, and infertility. It is often diagnosed during pregnancy or shortly after birth, however, symptoms become more obvious at puberty, when a lack of expected puberty changes, such as menstruation, can lead to a diagnosis.

Most girls with Turner Syndrome would have some, but not all, of these things:

- Short stature (not so tall as other girls)
- Widening of the neck ('webbed neck')
- Ovaries that do not function and infertility
- An underactive thyroid gland
- A broad chest with widely spaced nipples
- A heart murmur, sometimes associated with narrowing of the aorta (the main blood vessel that comes out of the heart)
- A difference about the way the kidney is formed



This disorder happens by a random error in cell division during the formation of reproductive cells or early foetal development. There is no cure for the condition, but treatments help manage symptoms. Doctors often use growth hormone therapy to improve adult height and hormone replacement to trigger puberty.

