



## Klinefelter syndrome

Other Names: 47,XXY

*Klinefelter syndrome is a genetic condition that affects males and is caused by the presence of at least one extra X chromosome.*

### Characteristics of Klinefelter syndrome

Klinefelter syndrome is the most common disorder of sex chromosomes in humans occurring in about 1 in every 500 males. Usually, humans have 46 chromosomes. Chromosomes come in pairs with 1-22 the same in males and females. The 23rd pair is known as the sex chromosomes: males have one X and one Y while females have two X's. In Klinefelter syndrome, there are at least two X chromosomes and one Y chromosome present. Klinefelter syndrome can affect physical and cognitive male development, but symptoms and signs can vary among affected males.

Most males with Klinefelter syndrome have tall stature, narrow shoulders, broad hips, and sparse body hair. Intelligence can range from normal to delayed speech and language development. The testes are typically decreased in size. Most males are infertile due to changes in the testes that causes reduced to absent production of sperm. Males with Klinefelter syndrome typically have lower testosterone levels than normal males. A small number of these males will develop gynecomastia (breast development), which increases risk for developing male breast cancer. Additionally, males with Klinefelter syndrome have an increased chance of developing osteoporosis, obesity, metabolism problems, and diabetes.

The features of Klinefelter syndrome are very variable. Some of the features can be very mild so that the affected men often receive the diagnosis only at the adulthood by their infertility. The effects on physical and cognitive development are more severe with a greater number of extra X chromosomes.

### Diagnosis/Testing

Klinefelter syndrome is a chromosomal condition and is most often diagnosed by karyotype (chromosome analysis).

### Management/Surveillance

Since males with Klinefelter syndrome tend to have testosterone levels that are below normal, testosterone replacement therapy is sometimes prescribed by an endocrinologist (a doctor specializing in hormones). Testosterone replacement therapy can help improve concentration, promote the growth of body hair, increase the appearance of muscles, improve self-esteem and mood, increase sex drive, energy, and strength. Testosterone treatments do not prevent or fix the infertility in this condition; however, an infertility specialist may be able to help some couples get pregnant. Males with Klinefelter syndrome might need therapy (speech, physical, occupational) if they have developmental delays or they might need extra help in school for learning disabilities. Males with Klinefelter syndrome are also at increased risk to develop attention deficit hyperactivity disorder, autoimmune disorders, depression, osteoporosis, varicose veins, and rare tumors called extragonadal germ cell tumor.

### Mode of inheritance

In most cases, Klinefelter syndrome is not inherited and is considered a sporadic event with the chance of having a baby with Klinefelter syndrome influenced by the mother's age.

## Risk to family members

The greatest known risk factor for having a child with Klinefelter syndrome is based on maternal age. However, having had a child with Klinefelter syndrome also increases the chance of having another child with Klinefelter syndrome.

## Special considerations

None

## Resources

The American Association for Klinefelter Syndrome Information and Support (AAKSIS)

<http://www.aaksis.org>

Association for X and Y Chromosome Variations

<http://www.genetic.org/>

Genetics Home Reference: Klinefelter syndrome

<http://ghr.nlm.nih.gov/condition/klinefelter-syndrome>

## References

[Groth, K. et al. \(2013\).](#) "Klinefelter Syndrome-A Clinical Update." *Journal of Clinical Endocrinology and Metabolism* 98(1): 20-30.

[Hong, D. et al. \(2014\).](#) "Cognitive and neurological aspects of sex chromosome aneuploidies." *Lancet Neurology* 13(3): 306-318.

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