



Klinefelter Syndrome

WHAT IS KLINEFELTER SYNDROME?

Klinefelter syndrome is a group of conditions that affects the health of males who are born with at least one extra X chromosome. Chromosomes, found in all body cells, contain genes. Genes provide specific instructions for body characteristics and functions. For example, some genes determine height and hair color. Other genes influence language skills and reproductive functions. Each person typically has 23 pairs of chromosomes. One of these pairs (sex chromosomes) determines a person's sex. A baby with two X chromosomes (XX) is female. A baby with one X chromosome and one Y chromosome (XY) is male.

Most males with Klinefelter syndrome, also called XXY males, have two X chromosomes instead of one. The extra X usually occurs in all body cells. Sometimes the extra X only occurs in some cells, resulting in a less severe form of the syndrome (called mosaic Klinefelter syndrome). Rarely, a more severe form occurs when there are two or more extra X chromosomes.

DID YOU KNOW?

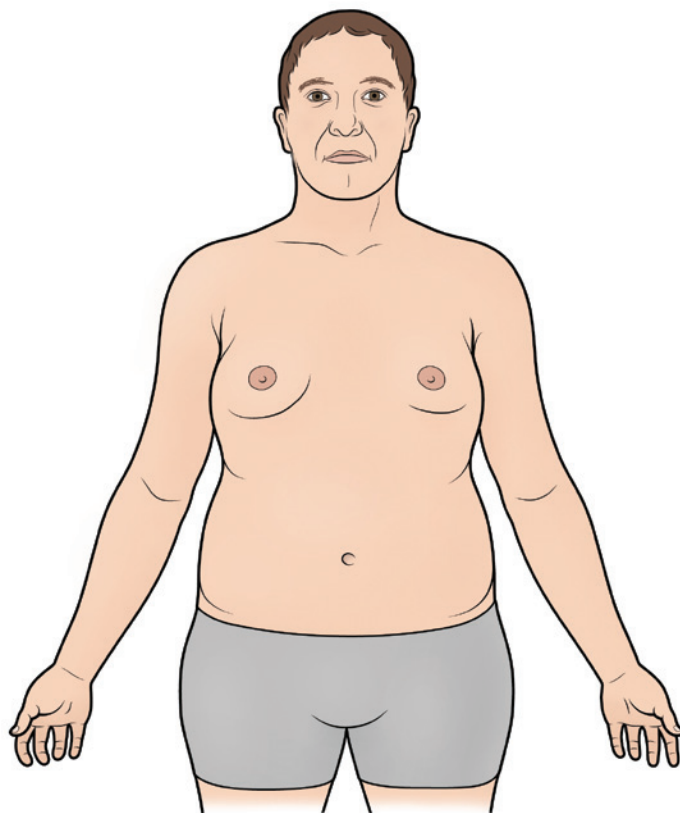
Klinefelter syndrome is the most common sex-chromosome abnormality, affecting about one in every 500 to 700 men.

WHAT CAUSES KLINEFELTER SYNDROME?

The addition of extra chromosomes seems to occur by chance. The syndrome is not inherited from the parents. The addition occurs in the sperm, the egg, or after conception.

WHAT ARE THE SIGNS AND SYMPTOMS OF KLINEFELTER SYNDROME?

Signs and symptoms can vary. Some males have no symptoms but a doctor will be able to see subtle physical signs of the syndrome. Many males are not diagnosed until puberty or adulthood. As many as two-thirds of men with the syndrome may never be diagnosed. Many men with mosaic Klinefelter syndrome have few obvious signs except very small testicles.



SIGNS AND SYMPTOMS BY AGE GROUP

Infants and young boys may have:	Adolescents may ALSO have:	Adults may ALSO have:
Problems at birth, such as testicles that haven't dropped into the scrotum or a hernia*	Very small, firm testicles	Low testosterone (male hormone) levels
A small penis	Enlarged breasts, called gynecomastia	Infertility from a lack of sperm
Weak muscles	Long legs but a short trunk	Decreased sex drive
Speech and language problems, such as delayed speech	Above-average height	Problems getting or keeping an erection
Problems with learning and reading	Reduced muscle bulk	Other difficulties, such as being unable to make plans or solve problems
Problems fitting in socially	Sparse facial and body hair	
Mood and behavioral problems	Delayed puberty	
	Low energy levels	

* when an internal organ bulges through a body cavity wall into the scrotum

HEALTH PROBLEMS LINKED TO KLINEFELTER SYNDROME

Klinefelter syndrome can lead to weak bones (osteoporosis), varicose veins, and autoimmune diseases (when the immune system acts against the body), such as lupus or rheumatoid arthritis. XXY males have an increased risk for breast cancer and cancers that affect blood, bone marrow, or lymph nodes, such as leukemia. They also tend to have excess fat around the abdomen (which raises the risk of health problems), heart and blood vessel disease, and type 2 diabetes.

HOW IS KLINEFELTER SYNDROME DIAGNOSED?

Diagnosis is based on a physical examination, hormone testing, and chromosome analysis. The syndrome can also be diagnosed before birth but testing is not routinely done at that time.

EDITORS

Adrian Dobs, MD, MHS
Alvin M. Matsumoto, MD

The Hormone Health Network offers free, online resources based on the most advanced clinical and scientific knowledge from The Endocrine Society (www.endo-society.org). The Network's goal is to move patients from educated to engaged, from informed to active partners in their health care. This fact sheet is also available in Spanish at www.hormone.org/Spanish.

March 2012, 2nd Edition

Klinefelter Syndrome Fact Sheet

WHAT IS THE TREATMENT FOR KLINEFELTER SYNDROME?

Treatment can help males overcome many of the physical, social, and learning problems that are part of the syndrome. Males with Klinefelter syndrome should be seen by a team of health care providers. The team may include endocrinologists, general practitioners, pediatricians, urologists, speech therapists, genetic counselors, and psychologists. Surgery may be needed to reduce breast size. With treatment, men can lead very normal lives.

Experts recommend testosterone replacement, starting during puberty, for proper development of muscles, bones, male sex characteristics such as facial hair, and sexual function. Continued treatment throughout life helps prevent long-term health problems. Testosterone replacement does not cure infertility, however. Infertility treatments require specialized—and costly—techniques, but some men with Klinefelter syndrome have been able to father children.

Questions to ask your doctor

- Will diagnostic tests and treatment be covered by my insurance?
- What are my (or my child's) options for testosterone therapy? What are the benefits and risks?
- What can I do manage other health problems linked to Klinefelter syndrome?
- Will I (or my child) be able to have children?
- Will I (or my child) have normal sexual function?
- Should I see an endocrinologist for my (or my child's) care?

RESOURCES

- Find-an-Endocrinologist: www.hormone.org or call 1-800-HORMONE (1-800-467-6663)
- National Institute of Child Health and Human Development Information Resource Center, National Institutes of Health (NIH): www.nichd.nih.gov/health/topics/klinefelter_syndrome.cfm or call 1-800-370-2943
- Genetic and Rare Diseases Information Center, NIH: www.rarediseases.info.nih.gov/GARD/Condition/8705/Klinefelter_syndrome.aspx or call 1-888-205-2311
- Genetics Home Reference website, National Library of Medicine, NIH: www.ghr.nlm.nih.gov/condition=klinefeltersyndrome



www.hormone.org