

Klinefelter Syndrome: A Guide for Families

What is Klinefelter syndrome?

It is a condition in boys caused by the presence of an extra X chromosome. Boys normally have one X and one Y chromosome, but most boys with Klinefelter syndrome have two X and one Y chromosome. It is relatively common, occurring in about 1 of every 500 baby boys. There is a wide range of findings in this condition, and many cases are not diagnosed until adulthood.

What are the common signs and symptoms?

The number and severity of symptoms vary widely. The most common reason for suspecting that a boy may have Klinefelter syndrome is when a doctor notices that in late puberty, the testicles are much smaller than what is normal. Boys may have undescended testicles, and the penis may be smaller than average. Most boys with Klinefelter syndrome are tall and have relatively long arms and legs. Although early in puberty, the testicles may produce normal amounts of the male hormone testosterone, this often lags as puberty progresses. This may be accompanied by somewhat less secondary sex characteristics, including decreased body hair or decreased muscle development. Sperm formation may be decreased or absent, and sometimes breast development occurs during puberty.

Some people are only very mildly affected and may not have Klinefelter syndrome diagnosed until they are adults, when they may have difficulty fathering children. Others have problems early in life with delayed developmental milestones or with learning problems during their school years.

How is Klinefelter syndrome diagnosed?

The syndrome is diagnosed by a special genetic blood test called a karyotype or chromosome analysis that shows the presence of an extra X chromosome. At the time of puberty, blood levels of

the pituitary hormones follicle-stimulating hormone (called FSH) and luteinizing hormone (LH) increase as a result of decrease in testicular function, and measurement of these hormones aids in making the correct diagnosis.

How is Klinefelter syndrome treated?

If testosterone levels are abnormally low, treatment with testosterone during and after adolescence may help many of the psychological symptoms and may increase secondary sex characteristics. If breast enlargement is a long-term problem, it may be treated surgically. Achieving fertility is more difficult. Special urologic techniques may help in identifying and retrieving sperm cells for fertility treatments. Support groups are available for individuals with Klinefelter syndrome and their families.

What is the long-term outlook?

Despite the range of possible problems seen in boys with this condition, the vast majority do very well. Specific educational interventions may be helpful during school years to overcome learning problems. Although there is no real cure for the condition, many of the symptoms can be treated. Overall, the predictions for long-term health and life expectancy are excellent.

*Pediatric Endocrine Society/American Academy of Pediatrics
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