

Klinefelter's Syndrome

Klinefelter's syndrome is a relatively common genetic disorder, affecting only boys. One of the main abnormalities is lack of normal growth of the testicles during puberty and smaller than average size of the penis. This is related to problems with making the normal amount of sex-related hormones. Your son may also have difficulties with school performance and behavior, including anger problems. Treatment with testosterone (male hormone replacement therapy) can lessen the impact of Klinefelter's syndrome.

What is Klinefelter's syndrome?

Klinefelter's syndrome is a genetic (inherited) disorder affecting approximately 1 in 500 newborn boys. It is caused by inheritance of an extra X chromosome. Boys with Klinefelter's syndrome have abnormal or slow sexual development. As a result, the condition may not be recognized until your son reaches the age of puberty.

The genetic abnormality can also cause problems with intellectual ability, ranging from learning disorders to mild mental retardation. Behavior problems are common as well, including aggressive or antisocial behavior.

Boys with Klinefelter's syndrome need treatments with the male hormone testosterone to develop more normally. Special education and other services can help your child's intellectual and social development.

What kinds of problems occur with Klinefelter's syndrome?

The most typical features of Klinefelter's syndrome are:

- *Abnormal sexual development.*
 - Your son does not progress into puberty at the expected time: the testicles remain small, the penis is smaller than average, and facial hair doesn't grow normally.
 - Breast enlargement (gynecomastia) may occur.
 - Males with Klinefelter's syndrome have problems with fertility (the ability to have children).
- *Intellectual/social issues.*
 - Most boys with Klinefelter's syndrome do not have major reductions in intellectual ability. However, on average, intelligence is lower than in your child's siblings. Some boys with Klinefelter's syndrome have mental retardation, although it is usually mild.
 - Your child may have problems at school, such as poor grades or learning disorders. Speech and language development may be delayed.

- Your son may have behavior problems. Many boys with Klinefelter's syndrome are shy and immature. They may also have outbursts of angry or antisocial behavior.
- Boys with Klinefelter's syndrome tend to be tall and thin, with long legs. Some have problems with physical coordination.
- *Health problems.* Boys with Klinefelter's syndrome are at increased risk of certain medical conditions:
 - Scoliosis (abnormal curvature of the spine).
 - Undescended testicles; this occurs when the testicles don't "drop" (descend) from the abdomen into the scrotum before your child is born.
 - Certain cancers, including male breast cancer.

All of these signs vary a lot. The abnormalities are often quite mild. Klinefelter's syndrome often goes unrecognized until puberty, when sexual development doesn't occur normally. Sometimes, the problem isn't diagnosed until adulthood, when men have trouble producing children.

With modern testing techniques, the genetic abnormality causing Klinefelter's syndrome may be recognized during pregnancy.

What causes Klinefelter's syndrome?

Klinefelter's syndrome is caused by a genetic abnormality. Genetically normal boys have one X chromosome and one Y chromosome (XY).

Boys with Klinefelter's syndrome have two X chromosomes plus one Y chromosome (XXY). Some people prefer the term "XXY males" instead of Klinefelter's syndrome for the disorder.

The extra X chromosome can develop in different ways. It is unrelated to anything the parents did wrong or anything the mother did during pregnancy.

What puts your child at risk of Klinefelter's syndrome?

The risk of having a child with Klinefelter's syndrome is slightly increased for older mothers. In addition, if one child has Klinefelter's syndrome, there is a slightly increased risk for future children (1 in 100).

How is Klinefelter's syndrome diagnosed?

- Klinefelter's syndrome may be suspected because of delayed puberty, your child's appearance, and other problems.

- Genetic testing is needed to confirm the gene abnormality causing Klinefelter's syndrome. Although the XXY pattern is most common, other variants can occur. We will likely recommend a visit to a medical geneticist, a doctor specializing in genetic diseases, who can provide more complete information.

How is Klinefelter's syndrome managed?

Several types of treatment are helpful for boys with Klinefelter's syndrome:

- *Testosterone replacement.* Regular treatments with the male hormone testosterone can help to normalize your son's sexual and physical development. A doctor specializing in hormone problems (endocrinologist) can manage this part of your child's treatment.
- *Treatment for enlarged breasts.* Enlarged breasts (gynecomastia) can be a problem in boys with Klinefelter's syndrome. The problem may improve over time, and the endocrinologist can recommend treatment, if needed.
- *Help with school and behavior problems.* Your son may need extra help in school, particularly with speech/lan-

guage. Special education services are available in every state. You are entitled to expert evaluation of your child. Based on the results, an Individualized Education Program (IEP) can be developed to meet your child's educational needs.

- *Infertility treatment.* For some men with Klinefelter's syndrome, special infertility treatments may make it possible to have children.

When should I call your office?

Learning that your child has a genetic disorder is a difficult situation for any parent. Call our office if you have any questions about this disorder, especially how to access the expert medical care and other services your son needs.

Where can I get more information?

Information on resources and support for families affected by Klinefelter's syndrome/XXY conditions is available from Klinefelter Syndrome & Associates on the Internet at www.genetic.org/ks, or call 1-888-XXY-WHAT (1-888-999-9428).