KLINEFELTER SYNDROME

What is Klinefelter syndrome?

Klinefelter syndrome is a disorder that affects only males. Males normally have an X chromosome and a Y chromosome (XY). But males who have Klinefelter syndrome have an extra X chromosome (XXY), giving them a total of 47 instead of the normal 46 chromosomes.

People with this disorder develop as males with subtle characteristics that become apparent during puberty. They are often tall and usually don't develop secondary sex characteristics, such as facial hair or underarm and pubic hair. The extra X chromosome primarily affects the testes, which produce sperm and the male hormone testosterone.

How do people get Klinefelter syndrome?

Normally, males inherit one X chromosome from their mother and one Y chromosome from their father. But those who have Klinefelter syndrome inherit an extra X chromosome.

Klinefelter syndrome is typically caused by what is called nondisjunction. If a pair of sex chromosomes fails to separate during the formation of an egg (or sperm), this is referred to as nondisjunction. When that egg unites with a normal sperm to form an embryo, that embryo may end up with three copies of the sex chromosomes (XXY) instead of the normal two (XY). The extra chromosome is then copied in every cell of the baby's body.

What are the symptoms of Klinefelter syndrome?

Many people with this disorder have no idea they have it until they hit puberty or try to have children. At puberty, men with this syndrome often develop more breast tissue than normal, have a less muscular body, and grow very little facial or body hair. When men with Klinefelter syndrome try to have children, most discover that they are sterile because they cannot produce sperm. Learning disabilities (not categorized as mental retardation) are also a common problem for them.
How do doctors diagnose Klinefelter syndrome?

Klinefelter syndrome is most often diagnosed in adulthood using a karyotype, an analysis of the patient's chromosomes taken from a blood sample.

Klinefelter syndrome may also be diagnosed during a woman's pregnancy. Doctors can look for the chromosome abnormality in cells taken from the amniotic fluid that surrounds the fetus (amniocentesis), or from the placenta (chorionic villus sampling (CVS)).

How is Klinefelter syndrome treated?

Hormone replacement therapy is the best way to treat this disorder. Teenagers are typically given testosterone injections to replace the hormone that would normally be produced by the testes. Synthetic testosterone works like natural testosterone - it builds muscle and increases hair growth.

Interesting facts about Klinefelter syndrome

Klinefelter syndrome is one of the most common genetic abnormalities. It affects between 1 in 500 and 1 in 1,000 males.

The disorder is named for Dr. Harry Klinefelter, who first reported its symptoms in 1942.