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ORIGIN, TREATMENT AND PREVENTION OF KLINEFELTER SYNDROME

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Annotation: Klinefelter syndrome (KS), also known as 47,XXY, is an aneuploid genetic condition where a male has an additional copy of the X chromosome. The primary features are infertility and small, poorly functioning testicles. Usually, symptoms are subtle and subjects do not realize they are affected. Sometimes, symptoms are more evident and may include weaker muscles, greater height, poor motor coordination, less body hair, breast growth, and less interest in sex. Often, these symptoms are noticed only at puberty. Intelligence is usually average, but reading difficulties and problems with speech are more common.

Key words: Klinefelter syndrome , treatment , drug , XXY, XY, chromosome.

Klinefelter syndrome occurs randomly. The extra X chromosome comes from the father and mother nearly equally. An older mother may have a slightly increased risk of a child with KS. The syndrome is defined by the presence of at least one extra X chromosome in addition to a Y chromosome yielding a total of 47 or more chromosomes rather than the usual 46. KS is diagnosed by the genetic test known as a karyotype.

While no cure is known, a number of treatments may help. Physical therapy, occupational therapy, speech and language therapy, counselling, and adjustments of teaching methods may be useful. Testosterone replacement may be used in those who have significantly lower levels. Enlarged breasts may be removed by surgery. Approximately half of affected males have a chance of fathering children with the help of assisted reproductive technology, but this is expensive and not risk free. XXY males have a ~15-fold higher risk of developing breast cancer than typical males, but still lower than that of females. People with the condition have a nearly normal life expectancy.

Klinefelter syndrome is one of the most common chromosomal disorders, occurring in one to two per 1,000 live male births. It is named after American endocrinologist Harry Klinefelter, who identified the condition in the 1940s. In 1956, the extra X chromosome was identified as the cause. Mice can also have the XXY syndrome, making them a useful research model.

Klinefelter syndrome is a genetic condition that results when a boy is born with an extra copy of the X chromosome. Klinefelter syndrome is a genetic condition affecting males, and it often isn't diagnosed until adulthood.

Klinefelter syndrome may adversely affect testicular growth, resulting in smaller than normal testicles, which can lead to lower production of testosterone. The syndrome may also cause reduced muscle mass, reduced body and facial hair, and enlarged breast tissue. The effects of Klinefelter syndrome vary, and not everyone has the same signs and symptoms.

Most men with Klinefelter syndrome produce little or no sperm, but assisted reproductive procedures may make it possible for some men with Klinefelter syndrome to father children.

Signs and symptoms of Klinefelter syndrome vary widely among males with the disorder. Many boys with Klinefelter syndrome show few or only mild signs. The condition may go undiagnosed until

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adulthood or it may never be diagnosed. For others, the condition has a noticeable effect on growth or appearance.

Signs and symptoms of Klinefelter syndrome also vary by age.

Signs and symptoms may include:

- Weak muscles
- Slow motor development — taking longer than average to sit up, crawl and walk
- Delay in speaking
- Problems at birth, such as testicles that haven't descended into the scrotum

Klinefelter syndrome occurs as a result of a random error that causes a male to be born with an extra sex chromosome. It isn't an inherited condition.

Humans have 46 chromosomes, including two sex chromosomes that determine a person's sex. Females have two X sex chromosomes (XX). Males have an X and a Y sex chromosome (XY).

Klinefelter syndrome can be caused by:

- One extra copy of the X chromosome in each cell (XXY), the most common cause
- An extra X chromosome in some of the cells (mosaic Klinefelter syndrome), with fewer symptoms
- More than one extra copy of the X chromosome, which is rare and results in a severe form

Extra copies of genes on the X chromosome can interfere with male sexual development and fertility.

Your doctor will likely do a thorough physical exam and ask detailed questions about symptoms and health. This may include examining the genital area and chest, performing tests to check reflexes, and assessing development and functioning.

The main tests used to diagnose Klinefelter syndrome are:

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- Hormone testing. Blood or urine samples can reveal abnormal hormone levels that are a sign of Klinefelter syndrome.
- Chromosome analysis. Also called karyotype analysis, this test is used to confirm a diagnosis of Klinefelter syndrome. A blood sample is sent to the lab to check the shape and number of chromosomes.

A small percentage of males with Klinefelter syndrome are diagnosed before birth. The syndrome might be identified in pregnancy during a procedure to examine fetal cells drawn from the amniotic fluid (amniocentesis) or placenta for another reason — such as being older than age 35 or having a family history of genetic conditions.

Klinefelter syndrome may be suspected during a noninvasive prenatal screening blood test. To confirm the diagnosis, further invasive prenatal testing such as amniocentesis is required.

If you or your son is diagnosed with Klinefelter syndrome, your health care team may include a doctor who specializes in diagnosing and treating disorders involving the body's glands and hormones (endocrinologist), a speech therapist, a pediatrician, a physical therapist, a genetic counselor, a reproductive medicine or infertility specialist, and a counselor or psychologist.

Although there's no way to repair the sex chromosome changes due to Klinefelter syndrome, treatments can help minimize its effects. The earlier a diagnosis is made and treatment is started, the greater the benefits. But it's never too late to get help.

Treatment for Klinefelter syndrome is based on signs and symptoms and may include:

- Testosterone replacement therapy. Starting at the time of the usual onset of puberty, testosterone replacement therapy can be given to help stimulate changes that normally occur at puberty, such as developing a deeper voice, growing facial and body hair, and increasing muscle mass and sexual desire (libido). Testosterone replacement therapy can also improve bone density and reduce the risk of fractures, and it may improve mood and behavior. It will not improve infertility.
- Breast tissue removal. In males who develop enlarged breasts, excess breast tissue can be removed by a plastic surgeon, leaving a more typical-looking chest.
- Speech and physical therapy. These treatments can help boys with Klinefelter syndrome who have problems with speech, language and muscle weakness.
- Educational evaluation and support. Some boys with Klinefelter syndrome have trouble learning and socializing and can benefit from extra assistance. Talk to your child's teacher, school counselor or school nurse about what kind of support might help.
- Fertility treatment. Most men with Klinefelter syndrome are typically unable to father children because few or no sperm are produced in the testicles. For some men with minimal sperm production, a procedure called intracytoplasmic sperm injection (ICSI) may help. During ICSI, sperm is removed from the testicle with a biopsy needle and injected directly into the egg.
- Psychological counseling. Having Klinefelter syndrome can be a challenge, especially during puberty and young adulthood. For men with the condition, coping with infertility can be

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difficult. A family therapist, counselor or psychologist can help work through the emotional issues.

If you notice symptoms of Klinefelter syndrome in yourself or your son, talk to your health care professional. You may be referred to a specialist for testing and diagnosis.

Here's some information to help you get ready for your appointment. If possible, bring a family member or friend with you. A trusted companion can help you remember information and provide emotional support.

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LITERATURE:

1. Fogel F., Matulskiy A., Genetika cheloveka, v 3-x tomax, per. s angl., M., 1990.

Muxsin Zokirov.[1]

2. www.google.com
3. www.wikipedia.com
4. https://www.webmd.com/men/klinefelter-syndrome