KLINEFELTER SYNDROME (47, XXY SYNDROME)

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ABSTRACT

Klinefelter Syndrome (47, XXY) is a chromosomal variation in males in males in which one extra X chromosome is present, resulting in a XXY sex chromosome karyotype. At birth, most neonates with 47, XXY (KS) have no dysmorphic or unusual features. Most individuals with 47, XXY (KS) are identified though prenatal diagnosis or when the child does not progress through puberty completely or adequately. The genetic variation is irreversible, so symptomatic treatment is recommended.

***Keywords**: Klinefelter syndrome, XXY, Karyotype.*

INTRODUCTION

In 1942, Dr.Harry Klinefelter, and his co-workers found nine men with small testes, aspermia, elevated urinary gonadotrophins and gynecomastics. These persons were later designated as Klinefelter's syndromic. Chromosomes are packages of genes found in every cell in the body. There are 2 types of chromosome, called the sex chromosomes, which determine the genetic sex of a baby. These are named either X or Y. Usually, a female baby has 2 X chromosomes (XX) and a male has 1 X and 1 Y (XY). But in Klinefelter syndrome, a boy is born with an extra copy of the X chromosome (XXY).Klinefelter syndrome is a chromosomal condition in boys and men that can affect physical and intellectual development.

DEFINITION

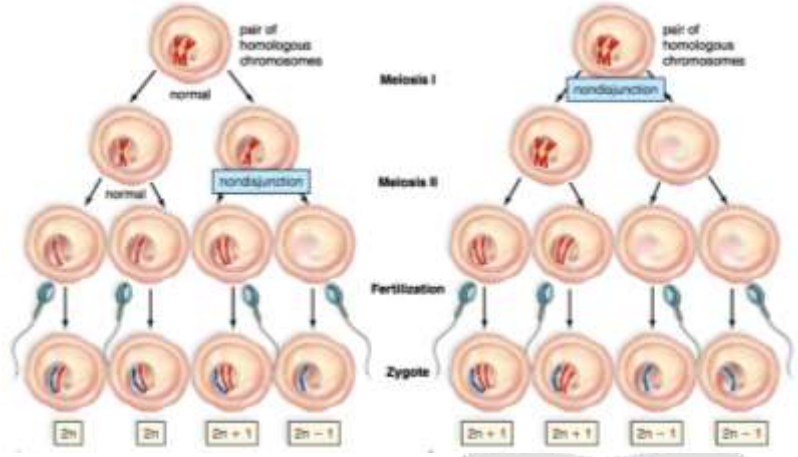
Klinefelter Syndrome (47, XXY) is a chromosomal variation in males in which one extra X chromosome is present, resulting in a XXY sex chromosome karyotype.

PREVALENCE

- This syndrome, evenly spread in all ethnic groups, has a prevalence of 1-2 in every 1000 males in the general population
- 3.1 % of infertile males have Klinefelter syndrome

CAUSES AND RISK FACTORS

- Klinefelter syndrome is caused by an additional X chromosome.
- Klinefelter syndrome is not directly inherited – the additional X chromosome occurs as a result of either the mother's egg or the father's sperm having the extra X chromosome (an equal chance of this happening in either), so after conception the chromosome pattern is XXY rather than XY.
- But the risk of a woman having a son with Klinefelter syndrome may be slightly higher if the mother is over 35 years of age.
- The extra X chromosome is retained because of a nondisjunction event during Meiosis I (gametogenesis) and Meiosis II in females.



- Meiosis I (gametogenesis): Nondisjunction occurs when homologous chromosomes (X and Y sex chromosomes) fail to separate, producing a sperm with an X and a Y chromosome. Fertilizing a normal (X) egg produces an XXY offspring. The XXY chromosome arrangement is one of the most common genetic variations from the XY karyotype, occurring in about 1 in 500 live male births.
- Meiosis II in females: Another mechanism for retaining the extra X chromosome is through a nondisjunction event during meiosis II in the female. Nondisjunction will occur when sister chromatids on the sex chromosome, in this case an X and an X, fail to separate. An XX egg is produced which, when fertilized with a Y sperm, yields XXY offspring.

SIGNS AND SYMPTOMS

Babies

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

Boys and teenagers

Signs and symptoms may include:

- Taller than average stature
- Longer legs, shorter torso and broader hips compared with other boys
- Absent, delayed or incomplete puberty
- After puberty, less muscle and less facial and body hair compared with other teens
- Small, firm testicles
- Small penis
- Enlarged breast tissue (gynecomastia)
- Weak bones
- Low energy levels
- Tendency to be shy and sensitive

- Difficulty expressing thoughts and feelings or socializing
- Problems with reading, writing, spelling or math

Men

Signs and symptoms may include:

- Low sperm count or no sperm
- Small testicles and penis
- Low sex drive
- Taller than average height
- Weak bones
- Decreased facial and body hair
- Less muscular compared with other men
- Enlarged breast tissue
- Increased belly fat

DIAGNOSIS

- Hormone Testing Blood or urine samples can reveal abnormal hormone levels that are a sign of Klinefelter syndrome.
- A spermiogram can also be part of the further investigation. Often there is an azoospermia present, rarely an oligospermia.
- Chromosome Analysis Also called karyotype analysis. This test is used to confirm a diagnosis of Klinefelter syndrome. Blood test called a karyotype (on lymphocyte) and is the standard diagnostic. A blood sample is sent to the lab to check the shape and number of chromosome.
- Prenatal Testing: many cases have been diagnosed through amniocentesis or chorionic villus sampling (CVS). The procedure is done in the first trimester (during the first three month of pregnancy, it's important to establish a foundation of good health) and the fetal cells needed for examination are taken from the placenta.

TREATMENT

- **Testosterone Replacement Therapy:**

It helps to stimulate changes in the time of puberty. Changes such as developing a deeper voice, growing facial and body hair, and increasing muscle mass and penis size. It will not result in testicle enlargement or improve infertility.

- **Breast Tissue Removal**

In males who develop enlarged breasts, excess breast tissue can be removed by a plastic surgeon, leaving a more normal looking chest.

- **Speech and Physical Therapy**

These treatments can help boys with Klinefelter syndrome overcome 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. Child's teacher, school counsellor or school nurse about some additional support.

- **Fertility Treatment**

Intracytoplasmic sperm injection (ICSI) for minimal sperm production patient. During ICSI, sperm is removed from the testicle with a biopsy needle and injected directly into the egg.

- **Psychological counselling**

A family therapist, counsellor or psychologist can help work through the emotional issues

COMPLICATIONS

Klinefelter syndrome may increase the risk of:

- Autism spectrum disorder, Anxiety and depression, Social, emotional and behavioural problems, such as low self-esteem, emotional immaturity and impulsiveness
- Infertility and problems with sexual function
- Weak bones (osteoporosis)
- Heart and blood vessel disease
- Breast cancer and certain other cancers
- Lung disease
- Autoimmune disorders such as lupus and rheumatoid arthritis

CONCLUSION

Klinefelter syndrome is fairly common. Klinefelter syndrome person are more likely to have low self-confidence or shyness, which can make things harder. Counsellors and therapists can give guys practical skills to help them feel more confident in social settings. Most boys who have it go on to have a good and healthy life, especially if they get the help they need from doctors, speech therapists, and other experts.

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