Klinefelter Syndrome: What You Need to Know

Klinefelter syndrome (KS or 47, XXY) is a genetic condition in which males are born with an extra X chromosome. In this handout, you will learn about Klinefelter syndrome and its symptoms. You will also learn how doctors diagnose and treat Klinefelter syndrome.

WHAT IS KLINEFELTER SYNDROME?

Klinefelter syndrome (KS) is a genetic condition in which boys are born with an extra copy of the X chromosome (piece of genetic material). A person with KS has 47 chromosomes inside their cells instead of the usual 46. KS is also called 47, XXY.

Males with KS typically have small testicles that sometimes make less testosterone (male sex hormone) than usual. Many men also have low sperm counts that can lead to infertility.

Other common challenges in males with KS can include learning disabilities and trouble with social interactions.

WHAT CAUSES KS?

KS happens at random. It is not anyone’s fault. KS is not passed down from parent to child.

WHAT ARE THE SYMPTOMS OF KS?

KS symptoms are different for every person. In most cases, symptoms do not show up until the teen or young adult years. In other cases, symptoms show up during infancy or childhood.

Infants

- Delayed motor skills, such as sitting up or walking
- Speech delays
- Undescended testicles (when one or both testicles have not dropped into their usual position outside of the body)

Children

Physical symptom and signs

- Taller than average height
- Longer legs, shorter torso, and wider hips than other boys
- Increased breast tissue (gynecomastia)
- Low muscle tone (hypotonia)
- Low energy levels

Social development symptom and signs

- Challenges with social interactions
- Challenges with learning in school, such as with reading, writing or math
- Attention deficits (ADD)

Symptoms continued on back page >>>
Teens and young adults

- Low or no sperm production
- Infertility
- Low sex drive
- Incomplete or delayed puberty (change from childhood to adulthood)
- Less facial or body hair
- Small, firm testicles
- More belly fat and less muscle than usual
- Anxiety and depression

HOW DO DOCTORS DIAGNOSE KS?
Symptoms of KS are often subtle. They can also look similar to other medical conditions. This can make KS more challenging for doctors to diagnose.

In many cases, doctors diagnose KS during or after puberty. Tests can include:

- A physical examination
- Genetic testing
- Hormone testing (done through a blood sample)

KS can also be diagnosed **prenatally** (before birth). Tests can include:

- Cell-free DNA testing
- Chorionic villus sampling
- Amniocentesis

HOW DO DOCTORS TREAT KS?
There is no cure for KS. Treatment focuses on you or your child’s individual symptoms and medical needs. Your care team will talk with you to create an individualized treatment plan. This will often include being seen by different types of doctors including a clinical geneticist, an endocrinologist or a fertility specialist, and a developmental medicine specialist or a psychologist.

Medication and therapy options for KS can include the following:

**Medication**

- **Testosterone replacement therapy** to help replace the testosterone the body does not make. You can meet with an endocrinologist (hormone doctor) to decide if this option is appropriate. Testosterone replacement therapy comes as a gel, patch or injection.
- **Surgery** to remove extra breast tissue, if present
- If you are an adult or your child is a teen who wants children in the future, **fertility treatments** can help improve the chances of having children.

**Therapies**

- **Speech and language therapy** to help children cope with speech delays or challenges
- **Physical therapy** to build muscle strength
- **Occupational therapy** to gain everyday skills for living and work
- **Behavioral therapy or counseling** to cope with depression, anxiety or low self-esteem
- **Help in school** to cope with learning challenges or disabilities