

## What is 47,XXY / Klinefelter syndrome?

Klinefelter syndrome, 47,XXY, is a chromosomal condition that affects males. It is the leading cause of infertility and hypogonadism. Every cell in the body contains 23 pairs of chromosomes for a total of 46 chromosomes. The twenty-third pair, the sex chromosomes, are different for males and females. Females have two copies of the X chromosome. Males have one X and one Y, or 46XY. In Klinefelter syndrome, males have an extra copy of the X chromosome. The result is 47,XXY in each cell. Some males are 46,XY/47,XXY or mosaic, which means that some of their cells have one X chromosome and one Y chromosome (XY), while other cells have the extra X (XXY). Less common variations result when there is more than one copy of the X or Y in each cell. These variations include 48,XXXY; 48,XXYY; 49,XXXXY; and 46,XX males, which is termed sex-reversal syndrome.

## How common is this condition?

47,XXY is estimated to occur in one out of 500 males, making it the most common chromosomal disorder. However, the variations are far less common. The syndrome was named after physician, Dr. Harry Klinefelter, who published a report in 1942 about nine men with enlarged breast development, sparse facial and body hair, small testes, and an inability to produce sperm. In the late 1950's, other researchers found that the cause was due to an extra X chromosome. Because all of the manifestations described by Dr. Klinefelter are not found in the majority of 47,XXY males, the term 47,XXY or XXY is preferred when referring to this condition.

## Is 47,XXY / Klinefelter syndrome inherited?

No. It is a random event that happens when paired chromosomes fail to separate in the first or second stage of meiosis.

## What causes 47,XXY?

The exact cause of 47,XXY is unknown, and the extra chromosome can come from either parent.

## How is 47,XXY diagnosed?

The diagnosis is made by taking a blood sample which will provide a "karyotype" or photographic illustration of the chromosomes in a single cell.

## When is one diagnosed?

Diagnosis can occur at any time in a person's life. The characteristics of 47,XXY are often subtle, so many XXY's go undiagnosed for years. Diagnosis is most likely to be made:

- as the result of prenatal testing
- during childhood when developmental delays in areas like speech and language are present
- at puberty when a boy's secondary sexual characteristics are not progressing in the typical way
- during fertility studies, when a couple has been unsuccessful in conceiving a child

*and in some instances*

- if continuing physical or psychological difficulties are present

## Does 47,XXY affect one's health?

XXY's have an increased risk for:

- autoimmune disorders, including diabetes and thyroiditis
- hypothyroidism
- breast cancer
- osteoporosis
- leg ulcers
- depression
- dental problems

## Does 47,XXY affect fertility?

Generally, XXY's are infertile, but a semen analysis is suggested to be sure. Some 46,XY/47,XXY's have preserved testicular function. Recent advances in male infertility treatment, such as testicular sperm extraction coupled with in vitro fertilization and a procedure called ICSI (intracytoplasmic sperm insertion), can result in a full term pregnancy without any chromosomal abnormalities.

Other options for creating a family are donor insemination and adoption.

## 47,XXY—THE EARLIER YEARS

### What does a 47,XXY newborn look like?

47,XXY newborns look like any other newborn. Unless the infant was tested prenatally, it is unlikely that anyone would suspect this condition.

## Does 47,XXY affect children and their development?

No one can predict for certain what effect the extra chromosome will have on an individual. Personalities in this group are as varied as in the general population. In addition, the spectrum of manifestations is wide.

However, the following characteristics are said to occur more often in children with 47,XXY. Keep in mind that a child may have only one or two features, while others may have more:

- delayed speech
- gross and fine motor delays
- sensory integration difficulties, which may include sensitivity to things like sounds, touch, or movement
- mild hypotonia or low muscle tone
- ADHD
- auditory processing problems
- language-based learning disabilities, including reading and written language difficulties
- social skill deficits
- anxiety
- depression
- gynecomastia or swelling of breast tissue during puberty

## Why is early diagnosis important?

47,XXY is a leading cause of male infertility and testosterone deficiency, yet the vast majority of KS individuals will reach adulthood without diagnosis.

Early diagnosis means:

- developmental delays can be addressed sooner
- appropriate educational interventions can be determined
- hormone replacement can be timed appropriately
- having greater knowledge and understanding of one's diagnosis
- gaining better health management throughout life

## An additional thought

Current studies have shown that the diagnosis of Klinefelter syndrome does NOT indicate mental retardation, deviant behavior, or other broad generalizations. Most individuals diagnosed with this condition have average to superior intelligence with only about 20% scoring below average on standardized intelligence tests.