## The mission of AXYS is to help individuals with one or more extra X or Y chromosomes and their families to live fuller and more productive lives.

**AXYS serves individuals and families affected by Sex Chromosome Aneuploidy including:**

- 47,XXY (Klinefelter syndrome)
- 47,XYY
- 47,XXX (Trisomy X)
- 48,XXYY and 48,XXXY

## Donate to AXYS

AXYS is a 501c3 organization. It relies on donations to fund our important support, advocacy and education work. Please consider making a tax deductible, online donation to AXYS at

**www.genetic.org/donate/**



## Services Available to the X and Y Variations Community

- Helpline helpline@genetic.org or 1-267-338-4262
- Online library of publications
- Educational webinars
- Support groups
- The AXYS Clinic and Research Consortium, a network of specialized clinics in the US
- Professional directory
- Research recruitment
- AXYS Family Conference

## Visit AXYS at www.genetic.org

**PO Box 253**
**Church Hill, MD 21623**
**info@genetic.org**

axys[sm]

# 47,XYY
# (Jacobs Syndrome)

## Diagnosing X and Y Chromosome Variations

X and Y chromosome variations affect 1 in 500 persons. Because children and adults with X/Y variations do not look “different”, and symptoms vary so much from one person to another, doctors frequently neglect to test for this genetic variation.

Children can be identified prenatally through non-invasive prenatal screening of a blood sample, also known as cell-free DNA. This is only a screening test that may show a high risk for sex chromosome variation. Diagnosis prenatally can take place through amniocentesis or chorionic villus sampling (CVS).

Children and adults can be diagnosed with specialized blood tests including karyotyping, microarray, or FISH (fluorescence in situ hybridization). Testing for sex chromosome aneuploidy can be obtained through a health care provider or a genetic counselor.

## About 47,XYY

47,XYY syndrome, sometimes called Jacobs syndrome, is the presence of an extra Y chromosome in males. It is characterized by wide variation in symptoms and severity among individuals. One male in 1000 is affected by 47,XYY. An individual with 47,XYY, usually has some, but not all, of the symptoms characteristic of the condition. In many, no symptoms are apparent; others are more significantly affected.

For more information, visit **https://genetic.org/variations/about-xyy/**

In infants and children:

- *Speech delay*
- *Hypotonia (low muscle tone)*
- *Motor skill development delay*
- *Learning disabilities*
- *Delayed social development; immaturity*
- *Mild autism spectrum disorder*
- *Attention deficit hyperactivity disorder (ADHD)*
- *Subtle physical signs such as clinodactyly (curved little finger)*

In teens and adults:

- *Tall stature and long limbs*
- *Learning and social difficulties*
- *Anxiety, mood disorder, or other psychiatric difficulties*
- *Executive functioning challenges; impulsivity*
- *Delay in vocational success*
- *Dental problems*
- *Very occasionally, hypogonadism or testicular failure*

Men with 47,XYY, are typically capable of fathering children and do not have lowered fertility. The chances of passing on the extra Y chromosome are very small, but couples may want to have genetic counseling before attempting pregnancy.

## Effective Treatment and Interventions

- Speech, occupational and physical therapy; very young children benefit from early intervention services
- Educational accommodations and special education services
- Family and individual therapy; behavioral consultation
- Social skills training
- Vocational counseling; workplace accommodations
- Life skills training; financial literacy programs
- Consultation with an endocrinologist if hypogonadism is suspected