

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 (Jacobs syndrome)
- 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/

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Services Available to the X and Y Variations Community

- Helpline helpline@genetic.org or 888-999-9428
- 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



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**X and Y
Chromosome Variations**
(Sex chromosome aneuploidy)

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. Only 25 to 30 percent of individuals are properly diagnosed in their lifetimes.

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 X and Y Chromosome Variations

X and Y chromosome variations (sex chromosome aneuploidy) are characterized by wide variation in symptoms and severity among individuals. An individual with one of the X/Y chromosome variations usually has some, but not all, of the symptoms. Most individuals with X and Y chromosome variations finish school, settle into careers, and live independently as adults. X and Y chromosome variations are not inherited.

In infants and children:

- *Speech delay*
- *Hypotonia (low muscle tone)*
- *Motor skill development delay*
- *Learning disabilities*
- *Delayed social development*

In teens and adults:

- *Tall stature and long limbs*
- *Learning and social difficulties*
- *Anxiety or other psychiatric difficulties*
- *Executive functioning challenges*
- *Delay in vocational success*

47,XXY (Klinefelter syndrome) occurs in 1 in 600 males www.genetic.org/variations/about-47xy

Infants and children may have undescended testicles. Puberty may be delayed, and most adults will have hypogonadism (inadequate testosterone levels) and lowered fertility.

47,XYY (Jacobs syndrome) occurs in 1 in 1000 males www.genetic.org/variations/about-xyy

Jacobs syndrome does not cause lowered fertility.

47,XXX (Trisomy X) occurs in 1 in 1000 females www.genetic.org/variations/about-xxx

Occasionally females with Trisomy X may experience lowered fertility.

48,XXYY is rare, occurring in 1 in 17,000 males www.genetic.org/variations/about-xyyy

Males with 48 chromosomes may share many of the characteristics of Klinefelter syndrome, but symptoms are often more pronounced.

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

