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
- 49,XXXXY
- 48,XXXX and 49,XXXXX

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

- Toll-free helpline 888–999–9428
- Online library of publications
- Educational webinars
- Support groups
- The AXYS Clinic and Research Consortium, a national network of specialized clinics
- Professional directory
- Research recruitment
- Biennial family conference and adult retreat

Visit AXYS at www.genetic.org

P.O. Box 861, Mendenhall, PA 19357
Helpline: 1-888-999-9428
info@genetic.org

axys[sm]

47,XXX
(Trisomy X)

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. Less than 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 47,XXX (Trisomy X)

47,XXX, is the presence of an extra X chromosome in a female. It is characterized by wide variation in symptoms and severity among individual girls and women. One female in 1000 is affected by 47,XXX. An individual with Trisomy X often has some, but not all, of the symptoms that characterize the phenotype. Women with 47,XXX, are typically capable of becoming pregnant although some have lowered fertility. The chances of passing on the extra X chromosome are very small, but couples may want to have genetic counseling before attempting pregnancy.
For more information, visit **https://genetic.org/variations/about-trisomy-x/**

In infants and children:
- *Speech delay*
- *Hypotonia (low muscle tone)*
- *Motor skill development delay*
- *Learning disabilities*
- *Delayed social development; anxiety*
- *Attention deficit hyperactivity disorder (ADHD)*
- *Subtle physical signs such as epicanthic eyelid skin folds*

In teens and adults:
- *Tall stature and long limbs*
- *Ongoing learning and/or social difficulties*
- *Anxiety, mood disorder, other psychiatric difficulties*
- *Executive functioning challenges; impulsivity*
- *Delay in vocational success*
- *Occasionally, delayed puberty, lowered fertility or premature ovarian failure*

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 programs
- Vocational counseling; workplace accommodations
- Life skills training; financial literacy programs
- Consultation with gynecologist if there are concerns regarding delayed puberty, infertility, or premature ovarian failure