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/

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

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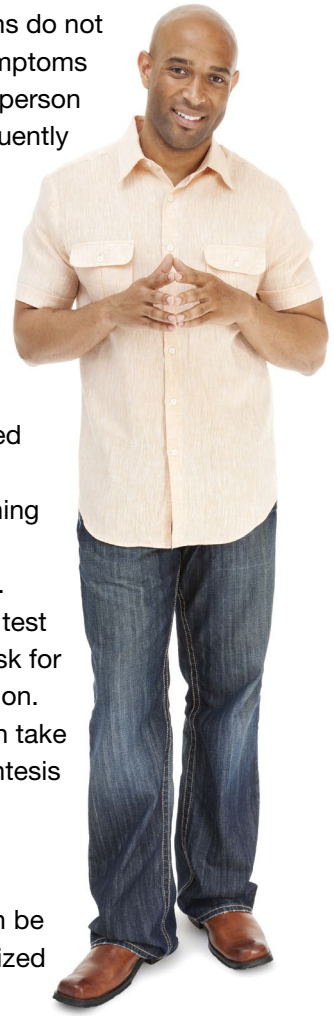
47,XXY
(Klinefelter Syndrome)

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. 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,XXY (Klinefelter Syndrome)

47,XXY, is the presence of an extra X chromosome in a male. One male in 500 is affected by 47,XXY. An individual with 47,XXY, usually has some, but not all, of the symptoms that characterize the phenotype. Nearly all adults have very low fertility, but in some, this is the only symptom. Others may be more significantly affected with psychosocial and medical complications. There is wide variation in how individuals are affected.
For more information, visit **https://genetic.org/variations/about-xxy/**

In infants and children:

- *Speech delay*
- *Hypotonia (low muscle tone) and motor skill development delay*
- *Cryptorchidism (undescended testicle)*
- *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*
- *Incomplete puberty; small testes; hypogonadism and infertility*
- *Ongoing learning and/or social difficulties*
- *Anxiety, mood disorder, other psychiatric difficulties*
- *Executive functioning challenges; impulsivity*
- *Delay in maturity, vocational success*
- *Dental problems*
- *Risk of developing Type II diabetes, osteoporosis, gynecomastia*

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
- Supplemental testosterone and other hormone treatments, if indicated
- Vocational counseling; workplace accommodations
- Life skills training; financial literacy programs
- Dental health preservation including sealants and frequent cleanings