Our mission is to help individuals with one or more extra X and/or Y chromosomes and their families lead fuller and more productive lives.

Visit AXYS at genetic.org to learn more about the organization’s programs.

Services available to all online visitors

- Toll-free information and support helpline staffed by experienced, trained volunteers
- An online library of resources and current articles and additional publications
- Access to conferences and webinars covering a variety of treatment and education topics
- Regional and online support groups
- Information about and opportunities to participate in research and clinical studies
- Assistance finding services and clinicians
- Personalized crisis support

Donate to AXYS and support our organization’s efforts to assist individuals and families.

Donations to AXYS are tax-deductible.

P.O. Box 861, Mendenhall, PA 19357
Helpline: 888-999-9428
info@genetic.org
About X and Y Chromosome Variations

X and Y chromosome variations affect 1 in 500 persons. Approximately 75% are never diagnosed.

Typical human cells have 46 chromosomes: 22 pairs of autosomes and 1 pair of sex chromosomes. Females have two X chromosomes (46,XX). Males have one X and one Y (46,XY). Variations are called sex chromosome aneuploidy (SCA). These are the most common chromosomal aneuploid conditions that affect humans.

The most common SCA in females is 47,XXX (Trisomy X). The most common in males are 47,XXY (Klinefelter syndrome) or 47,YYY. There are numerous other SCA conditions involving 48 and 49 chromosomes, as well as Turner syndrome (45,X).

SCA is associated with symptoms that vary along a continuum from mild to severe. The associated symptoms may include physical, hormonal, neurocognitive, psychiatric and social issues that can present functional challenges in daily life. Symptoms may include speech delay, low muscle tone and poor coordination, learning disabilities, and tall stature. There may also be anxiety, depression, attention deficits, and social immaturity. Intellectual disability is not common but may affect a few. It is rare to have all symptoms.

Most symptoms can be addressed successfully by early diagnosis and detection, and with appropriate treatments such as speech and occupational therapy. Special education services are indicated if there are learning disabilities. Persons with SCA may be somewhat slower achieving maturity as adults, but many live independently, have careers, and form families and typical adult relationships.

In children, 47,XXX may produce some of the following symptoms:
- Speech delay
- Hypotonia (low muscle tone)
- Delayed social development
- Learning disabilities
- Pervasive developmental disorder (PDD-NOS) or mild autism spectrum disorder
- Attention deficit and/or hyperactivity

In older teens and adults, 47,XXX may also be characterized by:
- Very tall stature and long limbs
- Severe acne
- Continued learning and/or social difficulties
- Delay in vocational success
- Anxiety, depression or other mood disorder
- Impulsivity
- Dental problems
- Occasionally, hypogonadism (failure of testes to produce adequate male hormone)

Men with 47,XXX are typically capable of fathering children and do not have lowered fertility. They may wish to have genetic counseling before becoming pregnant.

Effective Therapies & Treatment for 47,XXX Syndrome (if indicated)

**Infants and very young children** may be eligible for early intervention. Children can benefit from special education services including:
- Speech therapy
- Occupational and physical therapy for motor problems
- Special education services or accommodations
- Social skills training programs
- Family or individual counseling
- Behavioral consultation
- Regular exercise and upper body strengthening programs

**Adolescents** may develop severe acne. A dermatologist should treat appropriately to prevent scarring.

**Adults** may take several years longer to develop the emotional maturity to become self-supporting and capable of living independently. Interventions that may be helpful include:
- Psychological counseling
- Vocational counseling and workplace accommodations
- Dental health preservation/restoration
- A healthy diet and regular exercise to maintain cardiovascular health

Occasionally, a **boy with 47,XXX** fails to complete puberty or an adult experiences testicular failure, resulting in hypogonadism. This can be treated with hormone replacement by an experienced endocrinologist.

**Diagnostic tools:** Karyotype blood test; XCAT buccal swab test; FISH (fluorescence in situ hybridization); or microarray.

**Prenatal diagnostic tools:** Amniocentesis; chorionic villus sampling; or, NIPT (noninvasive prenatal test).

**Diagnostic testing** is indicated when a child or an adult displays some of the constellation of symptoms listed below.

47,XXX is a trisomy condition in which there is an extra Y chromosome. It affects 1 in 1,000 males. Many have no symptoms, but some are more significantly affected.

**Diagnostic testing** is indicated when a child or an adult displays some of the constellation of symptoms listed below.

**Diagnostic tools:** Karyotype blood test; XCAT buccal swab test; FISH (fluorescence in situ hybridization); or microarray.

**Prenatal diagnostic tools:** Amniocentesis; chorionic villus sampling; or, NIPT (noninvasive prenatal test).

**Diagnostic testing** is indicated when a child or an adult displays some of the constellation of symptoms listed below.