



With Knowledge, Support and Action

About KS&A

KS&A's mission is to help individuals with one or more extra X and/or Y chromosomes and their families lead fuller and more productive lives.

KS&A serves individuals and families affected by X and Y chromosome aneuploidies including Trisomy X, 47,XXY (Klinefelter syndrome), 47,XYY and associated conditions including 48,XXXY; 48,XXYY; 48,XXXX; 49,XXXXY; 49,XXXXX

We fulfill our mission through:

Support—providing service and support geared to the needs of our members, their families, and the clinical and educational professionals who assist them

Education—assuring that up-to-date, scientifically accurate information is in the hands of the appropriate people at the times they need it, as well as fairly and respectfully reported in the media

Advocacy—helping individuals to get the help and support they need whenever and wherever they need it; raising awareness about these conditions among medical and service professionals

Research—encouraging the advancement of research and other scholarly activities that support the generation and dissemination of knowledge about these conditions

Treatment—helping make available new, improved and more affordable treatments and interventions that will address the health and educational needs of adults, adolescents and children who are affected by extra X and y chromosome variations

Visit KS&A online at www.genetic.org
to learn more about the organization's programs

Services available to all online visitors:

- Toll-free information hotline staffed by professional social worker and telephone support volunteers
- An online library of resources and current articles and other publications
- Access to conferences and webinars covering a variety of treatment and education topics
- Regional and online support groups
- A newsletter
- Information about and opportunities to participate in research and clinical studies
- Directory of services and clinicians
- Personalize crisis support

Become a member of KS&A and support our organization's efforts to assist individuals and families. Membership prices begin at \$25 for individuals and just \$10 for adults with the condition.

Membership Benefits:

- Access to KS&A's online forums
- Free or discounted access to webinars, videos, conferences and other educational materials
- Fellowship with a supportive group of individuals, families and professionals dedicated to improving the lives of those with X and Y chromosome variations

Donate to KS&A:

KS&A relies on your donations to support its important outreach, advocacy and education work. We welcome your donations online. www.genetic.org



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X and Y CHROMOSOME VARIATIONS

(SEX CHROMOSOME ANEUPLOIDY)



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About X and Y Chromosome Variations

X and Y chromosome variations are the most common chromosomal aneuploid conditions that affect humans. Ordinarily, all human cells have 46 chromosomes, 22 pairs of autosomes, and 1 pair of sex chromosomes, either one X and one Y chromosome in a male, or two X chromosomes in a female. Any variation in *number* of sex chromosomes results in sex chromosome aneuploidy (SCA).

X and Y chromosome variations affect 1 in 500 persons. Females can be affected by 47,XXX (Trisomy X). Males can be affected by 47,XXY (Klinefelter syndrome) or 47,XYY. There are also rare SCA conditions involving 48 and 49 chromosomes, as well as Turner syndrome, in which a female has only one X chromosome.

Sex chromosome variations are syndromes characterized by an enormous range of functioning and symptom presentation. This is characteristic of many syndromes or medical conditions described by a collection of symptoms. Persons with extra X and Y chromosomes have one or more of these symptoms, but rarely have all of them. Some of the symptoms common to these trisomy conditions may include speech delay, low muscle tone and coordination difficulties, learning disabilities, or tall stature. There may also be anxiety, depression, attention deficits, and social immaturity. Intellectual disability is not common but may affect a small percentage.

Most symptoms can be addressed successfully by early diagnosis and detection, appropriate treatments such as speech and occupational therapy, and special education services, if learning disabilities are involved. While persons with X and Y chromosome variations may be somewhat slower than typical to achieve maturity as adults, they usually live independently, have careers and form families and typical adult relationships.

For all trisomy conditions, there is marked **under-diagnosis**. Children and adults with 47,XXY, 47,XXX and 47,XYY do not look different from others. Their differences and symptoms are often subtle. The “phenotype” (physical and cognitive characteristics) is highly variable from one person to the next. For this reason, unless a child is diagnosed prenatally because of a mother’s increased risk for genetic problems, pediatricians often neglect to test for SCA. Early diagnosis and support can be critical to prevent unnecessary complications.

Diagnosis requires a specialized blood test, called a karyotype or a buccal swab test called XCAT. In some cases, FISH (fluorescence in situ hybridization), or microarray analysis is performed. Prenatal diagnosis requires amniocentesis or chorionic villus sampling, or a noninvasive prenatal test called NIPT. Diagnostic testing is appropriate when a child or an adult displays some of the constellation of symptoms listed below or when there is a delay in reaching puberty, incomplete puberty, infertility or premature ovarian failure.

In **children**, the most common symptoms of SCA are:

- Delay in developing speech
- Low muscle tone (hypotonia) and delayed motor development
- Delayed social development
- Learning disabilities
- Subtle physical signs such as clinodactyly (a curved little finger)
- Anxiety or social withdrawal

In **older teens and adults**, SCA may be characterized by:

- Tall stature and long limbs
- Continued learning and/or social difficulties
- Delay in vocational success
- Anxiety, depression or other mood or psychiatric disorder
- Dental problems

Klinefelter syndrome, 47,XXY, occurs in one in 600 males. It has some additional signs and symptoms, including undescended testicles or occasionally, hypospadias, in an infant. Adolescents may begin puberty normally, but the testes nearly always eventually fail, making most men with XXY hypogonadal (lacking in adequate testosterone levels) and infertile. Recent advances in hormone replacement and assisted reproduction, can reduce the impact of failed testes, and can allow some men with XXY to become biological parents.

XYY occurs in 1 in 1000 males. They rarely develop hypogonadism, and most are able to father children.

Trisomy X occurs in 1 in 1000 females. Although a small percentage of women with 47,XXX have lowered fertility, most can become pregnant normally. A certain percentage of women with Trisomy X, however, will develop premature ovarian failure (early menopause). The risk of either XYY or Trisomy X parents passing on the extra chromosome to offspring is small.

Effective therapies for those with SCA are:

- Speech, occupational and physical therapy when indicated. Very young children are often eligible for early intervention services
- Special education services or educational accommodations when necessary
- Social skills training programs
- Family or individual counseling
- Behavioral consultation
- Regular exercise, a healthy diet, and upper body strengthening programs
- Vocational counseling and workplace accommodations when necessary
- Hormone treatments when indicated, generally for adolescents and adults who have 47,XXY (Klinefelter Syndrome)