KS&A

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**

KS&A

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TRISOMY X
47,XXX
SYNDROME

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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.

Trisomy X
47,XXX Syndrome

47,XXX is a trisomy condition affecting **females** in which there is an extra Y chromosome. It affects 1 in 1,000 females.

In **children**, Trisomy X may produce some of the following symptoms:

- Delay in developing speech
- Low muscle tone (hypotonia) and delayed motor development
- Delayed social development
- Learning disabilities
- Lowered IQ
- Anxiety and other emotional difficulties
- Epicanthal eyelid skin folds
- Occasionally, genito-urinary malformtions or delayed puberty

In **older teens and adults**, 47,XXX may also be characterized by:

- Tall stature and long limbs
- Continued learning and/or social difficulties
- Delay in vocational success
- Anxiety, depression or other mood disorder
- Dental problems
- Occasionally, lowered fertility or premature ovarian failure (early menopause)

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 above or when there is infertility or premature ovarian failure.

Effective Therapies and Treatment for Trisomy X

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 when indicated
- Special education services or educational accommodations when necessary
- Social skills training programs
- Family or individual counseling
- Behavioral consultation
- Regular exercise and upper body strengthening programs

Adolescents and adult women with Trisomy X can ecan benefit from the following support services:

- Psychological counseling when indicated
- Vocational counseling and workplace accommodations when necessary
- A healthy diet and regular exercise to maintain cardiovascular health

Most adult women with Trisomy X can expect to become pregnant normally, although there may be lowered fertility. The risk of transmitting the additional X chromosome to offspring is low, but couples may wish to obtain genetic counseling if pregnancy is being contemplated.

Most women with Trisomy X go through menopause normally. There are instances of premature ovarian failure (early menopause), which may require hormone treatment.