

The mission of AXYS

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
- 47,XXX (Trisomy X)
- 48,XXYY (XXYY Syndrome)
- 48,XXXY
- 48,XXXX
- 49,XXXXY
- 49,XXXXX



Donate to the XXYY Project

The XXYY Project operates as a project of AXYS, a 501(c)(3) organization. We rely on donations to fund our important support, advocacy and education work. Please consider making a tax deductible, online donation to the XXYY Project by visiting the AXYS website, and clicking on the XXYY Project in the drop down box.

www.genetic.org/donate/

About the XXYY Project

The XXYY Project was organized by members of the XXYY Parent Network, an informal association of parents, founded in 1998. It currently operates under the umbrella of AXYS, the association of X and Y chromosome variations.

The XXYY Project offers information and support for affected individuals and family members world-wide. The XXYY Project has been instrumental in expanding the AXYS Clinic and Research Consortium and in promoting research projects into XXYY Syndrome.



Contact the XXYY Project

Website



Brochure



<https://genetic.org/variations/about-xyyy/>

1-267-338-4262

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48, XXYY (XXYY Syndrome)



What is XYY Syndrome?

Typical males are born with only one X and one Y Chromosome. 48, XYY is a rare sex chromosome aneuploidy (SCA) condition affecting approximately 1:18,000 - 1:40,000 male births. XYY Syndrome is characterized as a developmental disability and neurodevelopmental disorder.



XYY is often described as a variant of Klinefelter Syndrome, 47, XYY, due to sharing similar physicality.



Research has established that XYY has its own distinct characteristics, with more complex neurodevelopmental features. Symptoms can be numerous and severe, and may require special treatment and management.



A New Look at XYY Syndrome:
Medical and Psychological Features

How is XYY diagnosed?

48, XYY is most commonly identified by a standard karyotype or chromosomal microarray (CMA) performed on blood, amniotic fluid or buccal swab. Prenatal diagnosis is becoming more regular, but XYY is most commonly diagnosed during childhood when physical and/or developmental concerns warrant genetic testing.

What are the symptoms?

XYY characteristics recognized by parents and medical professionals:

- Tall (taller than expected for family)
- Curved pinky finger (Clinodactyly)
- Developmental delays / Delayed Milestones
- Speech and language impairment
- Behavior outbursts & mood swings
- Learning disabilities & possible low IQ
- ADHD & Autism Spectrum Disorders
- Allergies, asthma & respiratory illnesses
- Tremors
- Flat feet
- High arches (pes cavus)
- Testosterone deficiency & delayed or lack of sexual development
- Infertility
- Undescended testes
- Significant dental problems
- Seizures
- Breast development (gynecomastia)
- Low muscle tone (hypotonia)
- Heart problems
- Deep vein thrombosis
- Eosinophilic Esophagitis (EOE)

Not all individuals with XYY have all these traits.



What are possible treatments?

Boys with XYY benefit from evaluations and early intervention therapies to address developmental, cognitive, and physical delays. Developing a treatment plan with a medical team that includes therapies, behavioral interventions, education supports, and possible psychotropic medications is advised. Hormone therapy can be initiated for boys who cannot make adequate testosterone on their own.

Common myths

An XYY diagnosis can be distressing to learn, and getting good, reliable information on how to help your son thrive is essential.

- Cognitive abilities vary among boys with XYY.
- Many men with XYY can live independently with community supports, while some may require supervision in a residential setting.
- XYY is often mistaken for other syndromes, and is often paired with additional diagnosis.

