



47,XYY syndrome

47,XYY syndrome is characterized by an extra copy of the Y chromosome in each of a male's cells. Although many males with this condition are taller than average, the chromosomal change sometimes causes no unusual physical features. Most males with 47,XYY syndrome have normal production of the male sex hormone testosterone and normal sexual development, and they are usually able to father children.

47,XYY syndrome is associated with an increased risk of learning disabilities and delayed development of speech and language skills. Affected boys can have delayed development of motor skills (such as sitting and walking) or weak muscle tone (hypotonia). Other signs and symptoms of this condition include hand tremors or other involuntary movements (motor tics), seizures, and asthma. Males with 47,XYY syndrome have an increased risk of behavioral, social, and emotional difficulties compared with their unaffected peers. These problems include attention deficit hyperactivity disorder (ADHD); depression; anxiety; and autism spectrum disorder, which is a group of developmental conditions that affect communication and social interaction.

Physical features related to 47,XYY syndrome can include increased belly fat, a large head (macrocephaly), unusually large teeth (macrodontia), flat feet (pes planus), fifth fingers that curve inward (clinodactyly), widely spaced eyes (ocular hypertelorism), and abnormal side-to-side curvature of the spine (scoliosis). These characteristics vary widely among affected boys and men.

Frequency

This condition occurs in about 1 in 1,000 newborn boys. Five to 10 boys with 47,XYY syndrome are born in the United States each day. Many affected males are never diagnosed or not diagnosed until later in life.

Causes

People normally have 46 chromosomes in each cell. Two of the 46 chromosomes, known as X and Y, are called sex chromosomes because they help determine whether a person will develop male or female sex characteristics. Females typically have two X chromosomes (46,XX), and males have one X chromosome and one Y chromosome (46,XY) in each cell.

47,XYY syndrome is caused by the presence of an extra copy of the Y chromosome in each of a male's cells. As a result of the extra Y chromosome, each cell has a total of 47 chromosomes instead of the usual 46. It is unclear why an extra copy of the Y chromosome is associated with tall stature, learning problems, and other features in some boys and men.

Some males with 47,XYY syndrome have an extra Y chromosome in only some of their cells. This phenomenon is called 46,XY/47,XYY mosaicism.

Inheritance Pattern

Most cases of 47,XYY syndrome are not inherited. The chromosomal change usually occurs as a random event during the formation of sperm cells. An error in cell division called nondisjunction can result in sperm cells with an extra copy of the Y chromosome. If one of these atypical reproductive cells contributes to the genetic makeup of a child, the child will have an extra Y chromosome in each of the body's cells.

46,XY/47,XYY mosaicism is also not inherited. It occurs as a random event during cell division in early embryonic development. As a result, some of an affected person's cells have one X chromosome and one Y chromosome (46,XY), and other cells have one X chromosome and two Y chromosomes (47,XYY).

Other Names for This Condition

- Jacob's syndrome
- XYY karyotype
- XYY syndrome
- YY syndrome

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
[/primer/testing/genetictesting](#)
- Genetic Testing Registry: Double Y syndrome
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3266843/>

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%2247%2CXYY+syndrome%22+OR+%22XYY+Karyotype%22+OR+%22XYY+syndrome%22>

Other Diagnosis and Management Resources

- Association for X and Y Chromosome Variations: Tell Me About 47,XYY
<https://genetic.org/variations/about-xyy/>

Additional Information & Resources

Health Information from MedlinePlus

- Health Topic: Learning Disabilities
<https://medlineplus.gov/learningdisabilities.html>
- Health Topic: Speech and Communication Disorders
<https://medlineplus.gov/speechandcommunicationdisorders.html>

Genetic and Rare Diseases Information Center

- 47, XYY syndrome
<https://rarediseases.info.nih.gov/diseases/5674/47-xyy-syndrome>

Educational Resources

- MalaCards: 47,xyy
<https://www.malacards.org/card/47xyy>
- March of Dimes: Chromosomal Conditions
<https://www.marchofdimes.org/baby/chromosomal-conditions.aspx>
- Merck Manual Consumer Version
<https://www.merckmanuals.com/home/children-s-health-issues/chromosome-and-gene-abnormalities/xyy-syndrome>
- Orphanet: 47,XYY syndrome
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=8

Patient Support and Advocacy Resources

- Association for X and Y Chromosome Variations
<https://genetic.org/>
- National Organization for Rare Disorders (NORD)
<https://rarediseases.org/rare-diseases/xyy-syndrome/>
- Resource list from the University of Kansas Medical Center
<http://www.kumc.edu/gec/support/chromoso.html#xyy>
- The Focus Foundation
<https://thefocusfoundation.org/x-y-chromosomal-variations/xyy/>
- Unique: XYY
https://www.rarechromo.org/media/information/Chromosome_Y/XYY%20FTNW.pdf

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28xyy%5BTI%5D%29+OR+%2847,xyy%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

Medical Genetics Database from MedGen

- 47,XYY syndrome
<https://www.ncbi.nlm.nih.gov/medgen/1432905>

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