Triple X syndrome

Triple X syndrome, also called trisomy X or 47,XXX, is characterized by the presence of an additional X chromosome in each of a female's cells. Although females with this condition may be taller than average, this chromosomal change typically causes no unusual physical features. Most females with triple X syndrome have normal sexual development and are able to conceive children.

Triple X syndrome is associated with an increased risk of learning disabilities and delayed development of speech and language skills. Delayed development of motor skills (such as sitting and walking), weak muscle tone (hypotonia), and behavioral and emotional difficulties are also possible, but these characteristics vary widely among affected girls and women. Seizures or kidney abnormalities occur in about 10 percent of affected females.

Frequency

This condition occurs in about 1 in 1,000 newborn girls. Five to 10 girls with triple X syndrome are born in the United States each day.

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).

Triple X syndrome results from an extra copy of the X chromosome in each of a female's cells. As a result of the extra X chromosome, each cell has a total of 47 chromosomes (47,XXX) instead of the usual 46. An extra copy of the X chromosome is associated with tall stature, learning problems, and other features in some girls and women.

Some females with triple X syndrome have an extra X chromosome in only some of their cells. This phenomenon is called 46,XX/47,XXX mosaicism.

Inheritance Pattern

Most cases of triple X syndrome are not inherited. The chromosomal change usually occurs as a random event during the formation of reproductive cells (eggs and sperm). An error in cell division called nondisjunction can result in reproductive cells with an abnormal number of chromosomes. For example, an egg or sperm cell may gain an extra copy of the X chromosome as a result of nondisjunction. If one of these atypical

reproductive cells contributes to the genetic makeup of a child, the child will have an extra X chromosome in each of the body's cells.

46,XX/47,XXX 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 two X chromosomes (46,XX), and other cells have three X chromosomes (47,XXX).

Other Names for This Condition

- 47,XXX
- 47,XXX syndrome
- triplo X syndrome
- trisomy X
- XXX syndrome

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
  /primer/testing/genetictesting
- Genetic Testing Registry: Trisomy X syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0221033/

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22triple+X+syndrome%22

Other Diagnosis and Management Resources

- Association for X and Y Chromosome Variations (AXYS): Trisomy X Syndrome
  https://genetic.org/variations/about-trisomy-x/
- MedlinePlus Medical Tests: Karyotype Genetic Test
  https://medlineplus.gov/lab-tests/karyotype-genetic-test/

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
- Medical Tests: Karyotype Genetic Test
  https://medlineplus.gov/lab-tests/karyotype-genetic-test/

Genetic and Rare Diseases Information Center

- 47 XXX syndrome
  https://rarediseases.info.nih.gov/diseases/5672/47-xxx-syndrome

Educational Resources

- MalaCards: triple x syndrome
  https://www.malacards.org/card/triple_x_syndrome
- 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/triple-x-syndrome
- Orphanet: Trisomy X
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=3375
- University of Michigan Health System
  http://www.med.umich.edu/yourchild/topics/xxxsyn.htm

Patient Support and Advocacy Resources

- Association for X and Y Chromosome Variations (AXYS)
  https://genetic.org/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/trisomy-x/
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/chromoso.html#xxx
- Unique: The Rare Chromosome Disorder Support Group (UK)
  https://www.rarechromo.org/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28triple+x%5BTIAB%5D%29+OR+%2847+AND+XXX%5BTI%5D%29+OR+%28trisomy+x%5BTIAB%5D%29+OR+%28triplo+x%5BTIAB%5D%29+OR+%28xxx+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

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