

Trisomy X

Description

Trisomy X, also called triple X syndrome 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 trisomy X have normal sexual development and are able to conceive children.

Trisomy X 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. Seizures or kidney abnormalities occur in about 10 percent of affected females.

Frequency

This condition occurs in about 1 in 1,000 female newborns; however, many of these affected individuals are never diagnosed. Five to 10 people with trisomy X 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).

Trisomy X 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 affected individuals.

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

[Learn more about the chromosome associated with Trisomy X](#)

- x chromosome

Inheritance

Most cases of trisomy X 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
- Triple X syndrome
- Triplo X syndrome
- Trisomy X
- XXX syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Trisomy X syndrome (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0221033/>)

Genetic and Rare Diseases Information Center

- Trisomy X (<https://rarediseases.info.nih.gov/diseases/5672/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Clinical Trials

- ClinicalTrials.gov (<https://clinicaltrials.gov/search?cond=%22Trisomy X%22>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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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