Turner syndrome

Description

Turner syndrome is a chromosomal condition that affects development in people who are assigned female at birth. Females typically have two X chromosomes, but in individuals with Turner syndrome, one copy of the X chromosome is missing or altered.

The most common feature of Turner syndrome is short stature, which becomes evident by about age 5. Reduced functioning of the ovaries, the female reproductive organs that produce egg cells (oocytes) and female sex hormones, is also very common. The ovaries develop normally at first, but egg cells usually die prematurely and most ovarian tissue breaks down before birth.

Many affected individuals do not undergo puberty unless they receive hormone therapy, and most are unable to become pregnant naturally. A small percentage of people with Turner syndrome retain normal ovarian function through young adulthood.

About 30 percent of individuals with Turner syndrome have extra folds of skin on the neck (webbed neck), a low hairline at the back of the neck, puffiness or swelling (lymphedema) of the hands and feet, skeletal abnormalities, or kidney problems. One-third to one-half of individuals with Turner syndrome are born with a heart defect, such as a narrowing of the large artery that leaves the heart (coarctation of the aorta) or abnormalities of the valve that connects the aorta to the heart (the aortic valve). Complications associated with these heart defects can be life-threatening.

Most people with Turner syndrome have normal intelligence. Developmental delays, nonverbal learning disabilities, and behavioral problems are possible, although these characteristics vary among affected individuals.

Frequency

This condition occurs in about 1 in 2,000 newborns worldwide who were assigned female at birth. However, this condition is much more common among pregnancies that do not survive to term (miscarriages and stillbirths).

Causes

Turner syndrome is related to the X chromosome, which is one of the two sex chromosomes. People typically have two sex chromosomes in each cell: generally,

females have two X chromosomes, while males have one X chromosome and one Y chromosome. Turner syndrome results when one normal X chromosome is present in cells and the other sex chromosome is missing or structurally altered. The missing genetic material affects development before and after birth.

About half of individuals with Turner syndrome have monosomy X, which means each cell in the individual's body has only one copy of the X chromosome instead of the usual two.

Turner syndrome can also occur if one of the X chromosomes is partially missing or rearranged rather than completely absent.

Some people with Turner syndrome have a chromosomal change in only some of their cells, which is known as mosaicism. People with Turner syndrome caused by X chromosome mosaicism are said to have mosaic Turner syndrome.

Researchers have not determined which genes on the X chromosome are associated with most of the features of Turner syndrome. They have, however, found that the loss of one copy of a gene called *SHOX* likely causes short stature and skeletal abnormalities in people with Turner syndrome.

Learn more about the gene and chromosome associated with Turner syndrome

- SHOX
- x chromosome

Inheritance

Most cases of Turner syndrome are not inherited. When this condition is caused by monosomy X, the chromosomal abnormality occurs as a random event during the formation of reproductive cells (eggs and sperm) in the affected person's parent. 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 lose a sex 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 a single X chromosome in each cell and will be missing the other sex chromosome.

Mosaic Turner syndrome is also not inherited. In an affected individual, it occurs as a random event during cell division in early fetal development. As a result, some of an affected person's cells have the usual two sex chromosomes, and other cells have only one copy of the X chromosome. Other sex chromosome abnormalities are also possible in people with X chromosome mosaicism.

Rarely, Turner syndrome caused by a partial deletion of the X chromosome can be passed from one generation to the next.

Other Names for This Condition

- 45,X
- Monosomy X
- TS
- Turner's syndrome
- Ullrich-Turner syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Turner syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0041408/)

Genetic and Rare Diseases Information Center

- Turner syndrome (https://rarediseases.info.nih.gov/diseases/7831/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Turner syndrome%22)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Turner+Syndrome%5BMAJR%5D%29+AND+%28Turner+syndrome%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D)

References

- Bondy CA. New issues in the diagnosis and management of Turner syndrome. RevEndocr Metab Disord. 2005 Dec;6(4):269-80. doi: 10.1007/s11154-005-6185-z. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16311945)
- Bondy CA. Turner syndrome 2008. Horm Res. 2009 Jan;71 Suppl 1:52-6. doi:10.1159/000178039. Epub 2009 Jan 21. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19153507)
- Bondy CA; Turner Syndrome Study Group. Care of girls and women with

Turnersyndrome: a guideline of the Turner Syndrome Study Group. J Clin EndocrinolMetab. 2007 Jan;92(1):10-25. doi: 10.1210/jc.2006-1374. Epub 2006 Oct 17. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17047017)

- Doswell BH, Visootsak J, Brady AN, Graham JM Jr. Turner syndrome: an updateand review for the primary pediatrician. Clin Pediatr (Phila). 2006May;45(4): 301-13. doi: 10.1177/000992280604500402. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16703153)
- Gravholt CH, Andersen NH, Conway GS, Dekkers OM, Geffner ME, Klein KO, Lin AE,Mauras N, Quigley CA, Rubin K, Sandberg DE, Sas TCJ, Silberbach M, Soderstrom-Anttila V, Stochholm K, van Alfen-van derVelden JA, Woelfle J, Backeljauw PF; International Turner Syndrome Consensus Group. Clinical practiceguidelines for the care of girls and women with Turner syndrome: proceedings fromthe 2016 Cincinnati International Turner Syndrome Meeting. Eur J Endocrinol. 2017Sep;177(3):G1-G70. doi: 10.1530/EJE-17-0430. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/28705803)
- Ho VB, Bakalov VK, Cooley M, Van PL, Hood MN, Burklow TR, Bondy CA. Majorvascular anomalies in Turner syndrome: prevalence and magnetic resonanceangiographic features. Circulation. 2004 Sep 21;110(12):1694-700. doi:10.1161/01.CIR.0000142290.35842.B0. Epub 2004 Sep 7. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15353492)
- Hong D, Scaletta Kent J, Kesler S. Cognitive profile of Turner syndrome. DevDisabil Res Rev. 2009;15(4):270-8. doi: 10.1002/ddrr.79. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20014362) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3114458/)
- Matura LA, Ho VB, Rosing DR, Bondy CA. Aortic dilatation and dissection inTurner syndrome. Circulation. 2007 Oct 9;116(15):1663-70. doi:10.1161/CIRCULATIONAHA.106.685487. Epub 2007 Sep 17. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17875973)
- Morgan T. Turner syndrome: diagnosis and management. Am Fam Physician. 2007Aug 1;76(3):405-10. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17708142)
- Ostberg JE, Conway GS. Adulthood in women with Turner syndrome. Horm Res. 2003;59(5):211-21. doi: 10.1159/000070220. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12714784)
- Sybert VP, McCauley E. Turner's syndrome. N Engl J Med. 2004 Sep16;351(12):1227-38. doi: 10.1056/NEJMra030360. No abstract available. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15371580)

Last updated August 1, 2023