Genetics Home Reference
Your Guide to Understanding Genetic Conditions
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Turner syndrome

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What is Turner syndrome?

Turner syndrome is a chromosomal condition that affects development in females. The most common feature of Turner syndrome is short stature, which becomes evident by about age 5. An early loss of ovarian function (ovarian hypofunction or premature ovarian failure) is also very common. The ovaries develop normally at first, but egg cells (oocytes) usually die prematurely and most ovarian tissue degenerates before birth. Many affected girls do not undergo puberty unless they receive hormone therapy, and most are unable to conceive (infertile). A small percentage of females with Turner syndrome retain normal ovarian function through young adulthood.

About 30 percent of females 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 leaving the heart (coarctation of the aorta) or abnormalities of the valve that connects the aorta with the heart (the aortic valve). Complications associated with these heart defects can be life-threatening.

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

How common is Turner syndrome?

This condition occurs in about 1 in 2,500 newborn girls worldwide, but it is much more common among pregnancies that do not survive to term (miscarriages and stillbirths).

What are the genetic changes related to Turner syndrome?

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: 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 a female's 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 sex chromosomes. Turner syndrome can also occur if one of the sex chromosomes is partially missing or rearranged rather than completely absent. Some women with Turner syndrome have a chromosomal change in only some of their cells, which is known as mosaicism. Women 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, identified one gene called *SHOX* that is important for bone development and growth. The loss of one copy of this gene likely causes short stature and skeletal abnormalities in women with Turner syndrome.

Read more about the *SHOX* gene and the X chromosome.

Can Turner syndrome be inherited?

Most cases of Turner syndrome are not inherited. When this condition results from 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 females 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.

Where can I find information about diagnosis or management of Turner syndrome?

These resources address the diagnosis or management of Turner syndrome and may include treatment providers.

- Genetic Testing Registry: Turner syndrome
- MedlinePlus Encyclopedia: Ovarian Hypofunction
- MedlinePlus Encyclopedia: Turner Syndrome

You might also find information on the diagnosis or management of Turner syndrome in Educational resources and Patient support.

General information about the diagnosis and management of genetic conditions is available in the Handbook. Read more about genetic testing, particularly the difference between clinical tests and research tests.

To locate a healthcare provider, see How can I find a genetics professional in my area? in the Handbook.

Where can I find additional information about Turner syndrome?

You may find the following resources about Turner syndrome helpful. These materials are written for the general public.

- MedlinePlus - Health information (3 links)
- Genetic and Rare Diseases Information Center - Information about genetic conditions and rare diseases
- Additional NIH Resources - National Institutes of Health (2 links)
- Educational resources - Information pages (12 links)
- Patient support - For patients and families (3 links)

You may also be interested in these resources, which are designed for healthcare professionals and researchers.

- Genetic Testing Registry - Repository of genetic test information (1 link)
- ClinicalTrials.gov - Linking patients to medical research
- PubMed - Recent literature

What other names do people use for Turner syndrome?

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

For more information about naming genetic conditions, see the Genetics Home Reference Condition Naming Guidelines and How are genetic conditions and genes named? in the Handbook.

What if I still have specific questions about Turner syndrome?

Ask the Genetic and Rare Diseases Information Center.

Where can I find general information about genetic conditions?

The Handbook provides basic information about genetics in clear language.

- What does it mean if a disorder seems to run in my family?
- What is a chromosome?

- Can changes in the number of chromosomes affect health and development?
- Are chromosomal disorders inherited?

These links provide additional genetics resources that may be useful.

- Genetics and Health
- Resources for Patients and Families
- Resources for Health Professionals

What glossary definitions help with understanding Turner syndrome?

aneuploidy ; aorta ; artery ; atypical ; cell ; cell division ; chromosome ; deletion ; disabilities ; dysgenesis ; egg ; gene ; hormone ; hormone therapy ; infertile ; inherited ; kidney ; lymphedema ; monosomy ; mosaic ; mosaicism ; nondisjunction ; ovarian ; puberty ; reproductive cells ; sex chromosomes ; short stature ; sperm ; stature ; syndrome ; tissue

You may find definitions for these and many other terms in the Genetics Home Reference Glossary.

See also Understanding Medical Terminology.

References (10 links)

The resources on this site should not be used as a substitute for professional medical care or advice. Users seeking information about a personal genetic disease, syndrome, or condition should consult with a qualified healthcare professional. See How can I find a genetics professional in my area? in the Handbook.

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