CONDITION

Turner syndrome

Turner syndrome is a chromosome condition affecting growth, puberty, fertility, and several body systems in females.

WHAT IT IS
A lifelong condition involving a missing or altered X chromosome

BODY SYSTEM
Growth, reproductive system, heart, hearing, kidneys, and hormones

MAIN SYMPTOMS
Short stature, delayed puberty, absent periods, hearing problems

WHAT CAUSES IT
Random missing or altered X chromosome

HOW IT IS DIAGNOSED
Chromosome analysis, sometimes prenatal diagnostic testing

MAIN TREATMENT
Growth and sex hormones, monitoring, and specialist care

CAN IT BE CURED?
No - chromosome change remains lifelong

IS IT AN EMERGENCY?
Sometimes - sudden severe chest or back pain

LONG-TERM OUTLOOK
Usually full lives with lifelong monitoring and treatment

Overview

Turner syndrome is a chromosome condition that affects females. It happens when one X chromosome is missing or has important structural changes in some or all cells.

Most people have 46 chromosomes in each cell, including two sex chromosomes. Females usually have two X chromosomes. In Turner syndrome, chromosome testing may show one complete X chromosome and a missing or altered second X. Not everyone has the same chromosome pattern.

The condition affects people differently. Short stature and reduced ovarian function are common. The ovaries usually make too little estrogen, a hormone needed for puberty, periods, bone health, and other body functions.

Turner syndrome can also affect the heart, major blood vessels, hearing, thyroid, kidneys, bones, and metabolism. Regular monitoring matters because some related conditions cause few symptoms at first but can be treated or managed when found early.

Turner syndrome may be diagnosed before birth, during childhood, around puberty, or in adulthood. Some people have clear physical features, while others have few outward signs.

Symptoms

The features of Turner syndrome differ greatly, especially in people with mosaic Turner syndrome. "Mosaic" means the chromosome change is present in some cells but not others.

Possible signs in babies include swelling of the hands and feet, a broad chest, widely spaced nipples, a low hairline, or extra folds of skin around the neck. Some babies have a heart or kidney difference found before or soon after birth.

During childhood, the most consistent feature is slower growth. A child may be shorter than expected for their age or family pattern.

When puberty would usually begin, the ovaries may not produce enough estrogen. This can lead to:

Other possible features include repeated middle-ear infections, gradual hearing loss, a high arched roof of the mouth, dental crowding, many moles, or differences in the shape of the nails.

Most people with Turner syndrome have intelligence in the usual range. Some have specific difficulties with spatial awareness, mathematics, attention, planning, or reading social cues. Verbal skills are often a strength. These patterns are not the same for everyone.

- Little or no breast development without hormone treatment
- Periods that do not start or stop unexpectedly
- Reduced fertility
- Slower development of strong bones

Causes and risk factors

Turner syndrome results from a random chromosome change during the formation of an egg or sperm, or during early development after fertilisation. It is not caused by anything a parent did before or during pregnancy.

Some people have only one X chromosome in every tested cell. This is sometimes described

as monosomy X or 45,X. "XO syndrome" is an older name, but it can be misleading because not everyone with Turner syndrome has the same chromosome finding.

Others have mosaic Turner syndrome. For example, some cells may have 45 chromosomes while others have 46. The second X chromosome can also be incomplete, rearranged, or formed into a ring.

Turner syndrome is usually not inherited. Having one child with it does not generally mean that future children will have it. Rare chromosome rearrangements may require individual genetic advice.

There are no established lifestyle or parental risk factors that reliably predict who will have Turner syndrome.

Diagnosis

Turner syndrome is confirmed with a chromosome analysis called a karyotype. This blood test examines the number and structure of chromosomes. More detailed genetic tests may be used when the result is uncertain or when mosaicism is suspected.

Testing may be offered when a child has unexplained short stature, delayed puberty, characteristic physical features, or certain heart findings. In adults, absent periods or difficulty becoming pregnant may lead to testing.

During pregnancy, ultrasound findings or a prenatal screening test may suggest Turner syndrome. Screening does not confirm the diagnosis. Cell-free DNA testing, sometimes called non-invasive prenatal testing, examines DNA fragments in the pregnant person's blood and can produce false-positive results.

Diagnostic prenatal testing uses cells collected by chorionic villus sampling or amniocentesis. Chromosome testing after birth may still be recommended because placental cells do not always have the same chromosome pattern as the baby.

After diagnosis, an assessment usually checks growth, blood pressure, heart and aorta structure, hearing, kidney anatomy, thyroid function, liver health, blood sugar, cholesterol, and bone health. The aorta is the large artery carrying blood away from the heart.

Treatment and management

Treatment does not change the chromosome pattern. It supports growth and development and reduces the effects of related health conditions.

Growth hormone may be offered during childhood to improve adult height. The expected benefit is influenced by when treatment starts, the child's growth pattern, and individual response. A paediatric hormone specialist usually monitors treatment and growth.

Estrogen replacement is commonly used when the ovaries do not make enough estrogen. It helps start and maintain puberty, supports the uterus, and protects bone health. A progesterone-type hormone is usually added later to produce regular shedding of the womb lining when appropriate. Hormone treatment is adjusted over time rather than given as one fixed plan.

Management may also involve:

Some people with mosaic Turner syndrome have spontaneous periods and may become pregnant without fertility treatment. For many others, pregnancy may be possible using a donated egg or embryo. Adoption and choosing not to become a parent are also options.

Pregnancy can place added strain on the aorta. Anyone with Turner syndrome considering pregnancy needs a detailed heart and aorta assessment beforehand. Pregnancy may be unsafe when certain cardiovascular problems are present.

- Regular blood pressure, heart, and aorta checks
- Hearing tests and treatment for ear problems
- Thyroid, liver, cholesterol, and blood sugar tests
- Screening for coeliac disease, an immune reaction to gluten
- Bone health assessment and support
- Educational or psychological support for specific learning needs
- Fertility counselling and reproductive care

Complications

Congenital heart differences are important complications. These include a bicuspid aortic valve, which has two flaps instead of three, and narrowing of the aorta, called coarctation. High blood pressure is also common and may not cause symptoms.

The aorta can gradually widen. Rarely, its wall can tear, known as aortic dissection. This is a medical emergency. Lifelong cardiovascular imaging is important even after a normal childhood assessment.

Other possible complications include hearing loss, an underactive thyroid, coeliac disease, liver test abnormalities, diabetes, high cholesterol, and reduced bone strength. Kidney differences

may increase the chance of urinary infections or high blood pressure.

Reduced ovarian function usually limits fertility. If some Y-chromosome material is found during genetic testing, there may be an increased risk of a tumour in underdeveloped gonadal tissue. A specialist will explain whether preventive surgery should be considered.

Anxiety, low mood, social difficulties, or reduced self-confidence can occur, particularly around body image, fertility, or feeling different from peers. These concerns deserve the same attention as physical health.

Prevention

Turner syndrome cannot be prevented. It results from a chromosome event that is usually random and is not caused by diet, medication, exercise, stress, or something a parent did.

Prenatal screening or testing can identify the condition before birth, but it does not prevent it. Genetic counselling can help families understand test results, the chance of recurrence, and available pregnancy options without directing them toward a particular decision.

Complications cannot always be prevented, but planned monitoring can find many of them early. Blood pressure control, heart surveillance, hearing care, hormone treatment, and attention to bone and metabolic health can reduce some long-term risks.

Living with it and what to expect

Turner syndrome is lifelong, but its effects change with age. During childhood, care often focuses on growth, hearing, learning, and congenital heart or kidney findings. During adolescence, puberty, hormone treatment, periods, body image, and increasing independence become more important.

In adulthood, monitoring continues even when you feel well. Heart and aorta imaging, blood pressure checks, hearing tests, and hormone and metabolic assessments may be needed at intervals based on your findings. Moving from children's services to coordinated adult care helps avoid gaps in follow-up.

Many people with Turner syndrome attend mainstream school, work, have relationships, and live independently. Some benefit from support with mathematics, navigation, organisation, or social situations. A learning assessment can identify practical accommodations rather than assuming broad intellectual disability.

Fertility can be an emotionally difficult subject. Early, sensitive discussions allow time to

understand fertility preservation, donor conception, adoption, or a life without children. No one option is right for everyone.

Long-term health is strongly influenced by heart and blood vessel findings, blood pressure, and access to ongoing care. Many people live active, fulfilling lives, but Turner syndrome is linked with increased health risks and may shorten life expectancy, especially when cardiovascular complications are present.

When to get medical help

Call emergency services now for sudden, severe chest, upper back, neck, or abdominal pain, especially if it feels tearing or is accompanied by breathlessness, fainting, sweating, weakness, or trouble speaking. These symptoms can have several causes, but in Turner syndrome they require urgent assessment for an aortic emergency.

Seek urgent medical assessment for new chest pressure, unexplained severe shortness of breath, fainting during activity, a very rapid or irregular heartbeat with weakness, or new neurological symptoms.

Arrange routine medical follow-up if you notice:

Children should be assessed if their growth slows, puberty has not started as expected, or swelling of the hands and feet is present in infancy. These findings do not prove Turner syndrome, but they can justify further evaluation.

- Reduced hearing, ringing in the ears, or repeated ear infections
- New headaches or dizziness that could relate to blood pressure
- Unusual tiredness, feeling cold, constipation, or weight changes
- Periods stopping unexpectedly
- Persistent abdominal symptoms or unexplained changes in liver tests
- Increasing anxiety, low mood, or difficulty coping

Common questions

Is Turner syndrome inherited from my parents?

Usually not. It most often results from a random chromosome event in an egg, sperm, or early embryo. It is not caused by a parent's health, age, or behaviour. Rare chromosome rearrangements may need more detailed genetic counselling.

Does Turner syndrome affect intelligence?

Most people have intelligence in the usual range. Some have specific difficulties with spatial tasks, mathematics, attention, organisation, or interpreting social signals. Speech and reading skills may be relative strengths. Support can be tailored to the person's actual learning profile.

Can I get pregnant if I have Turner syndrome?

Some people, particularly those with mosaic Turner syndrome, become pregnant without fertility treatment. Many need a donated egg or embryo because the ovaries have stopped working early. A heart and aorta assessment is essential before fertility treatment or pregnancy because pregnancy is not safe for everyone with Turner syndrome.

What is mosaic Turner syndrome?

Mosaic Turner syndrome means the chromosome change is present in only some cells. It may cause milder or different features, but the chromosome result alone cannot predict exactly how someone will be affected. Heart and other health checks remain important.

Will I need estrogen for life?

Estrogen replacement is often continued through the age when natural menopause would usually occur, and sometimes longer after an individual review. The form and timing may change with age, health, personal preference, and whether the uterus is present.

What is the life expectancy with Turner syndrome?

Many people live into older adulthood and have full, independent lives. Average life expectancy is reduced, mainly because of heart, aorta, and blood pressure complications. Regular cardiovascular monitoring and treatment of related conditions are important parts of long-term care.

Sources

- Gravholt CH, Andersen NH, Conway GS, et al. Clinical practice guidelines for the care of girls and women with Turner syndrome: proceedings from the 2023 Aarhus International Turner Syndrome Meeting . European Journal of Endocrinology . 2024;190(6):G53-G151.
- Gravholt CH, Viuff MH, Just J, Sandahl K, Brun S, van der Velden J. Turner Syndrome . GeneReviews , University of Washington, Seattle.
- Silberbach M, Roos-Hesselink JW, Andersen NH, et al. Cardiovascular Health in Turner Syndrome: A Scientific Statement From the American Heart Association . Circulation: Genomic and Precision Medicine . 2018;11(10):e000048.

- NHS. Turner syndrome . Overview, symptoms, diagnosis, and treatment.
- MedlinePlus Genetics, U.S. National Library of Medicine. Turner syndrome . Genetic causes, features, and inheritance.