



# Turner Syndrome

## WHAT IS TURNER SYNDROME?

Turner syndrome (TS) is a genetic condition that occurs when a female infant is born with a missing or changed X chromosome. Chromosomes contain the genes that instruct your body how to develop and function. Typically, a person has 46 chromosomes, including the two "sex chromosomes" that determine gender. Females usually have two X chromosomes.

TS occurs in about 1 in 2,000 female births worldwide. The reason for the missing or changed X chromosome appears to be a random event, so any girl could be born with TS.

## DID YOU KNOW?

**Early diagnosis and treatment of Turner syndrome in girls can help promote normal growth and development.**

## WHAT ARE THE PHYSICAL FEATURES OF TURNER SYNDROME?

The features of TS can vary widely from patient to patient, so the condition may not be diagnosed until a girl reaches the age of puberty or even later. The most common features of TS are short stature (with an average adult height of about 4'8", if untreated) and ovaries that do not produce female hormones or eggs. As adults, most women with TS cannot become pregnant without medical help.

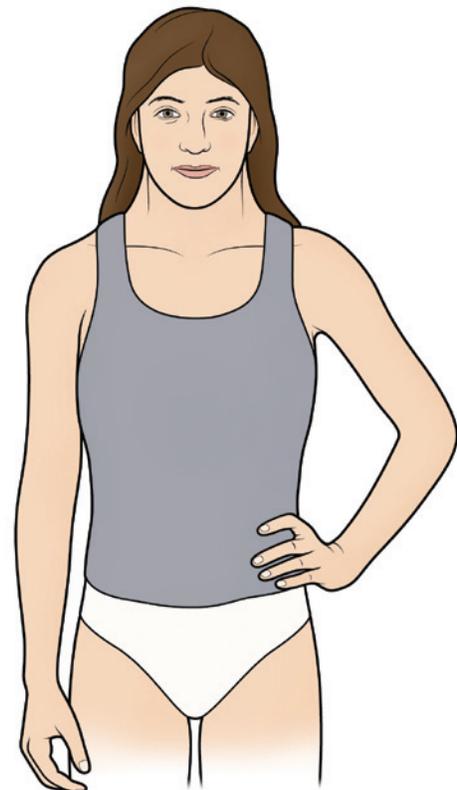
Apart from short stature, there may be other physical features:

### Head and neck

- Eye problems (drooping eyelids, "lazy" eye)
- Low-set ears
- Small lower jaw
- Low hairline at the back of the head
- Wide and short neck, sometimes with excess skin that joins the neck with the collarbone (called "neck webbing")

### Torso

- Broad chest
- Curvature of the spine (scoliosis)



## Hands, arms, and feet

- Arms that turn out more than usual at the elbows
- Missing 4th or 5th knuckle
- Puffiness of the hands and/or feet
- Narrow fingernails and toenails

Girls with TS may also have many moles on their skin.

## WHAT HEALTH PROBLEMS CAN OCCUR WITH TURNER SYNDROME?

Girls and women with TS are at risk for congenital (present at birth) abnormalities of the heart and kidneys, high blood pressure, chronic or repeated middle ear infections, hearing loss, diabetes, underactive thyroid gland, bowel disorders, and non-verbal learning disabilities (like difficulty with math). Other possible medical problems include being overweight, dental crowding, and osteoporosis (brittle bones).

Because of their physical conditions, health concerns, and infertility, some girls and women with TS may have low self-esteem, anxiety, or depression.

## HOW IS TURNER SYNDROME DIAGNOSED?

Physical features may suggest TS, but the only reliable way to diagnose the condition is by a karyotype analysis. In this test, a blood sample is examined under a microscope to detect missing or altered chromosomes.

## HOW IS TURNER SYNDROME TREATED?

Because TS is a genetic disorder, there is no cure, but some of its symptoms can be treated individually. Although TS is a lifelong condition, it can be successfully managed. Two options are:

- **Growth hormone therapy.** Girls with TS may get injections of a synthetic form of human growth hormone to increase their height. Treatment may start in the pre-school or early school years, if the diagnosis of TS is made early.
- **Hormone replacement therapy.** Most girls with TS need estrogen (hormone) therapy to develop the physical changes that normally occur at puberty—breast growth and menstruation. Estrogen is available as a patch, gel, spray, pill, or injection. After menstruation begins, a second hormone, progesterone, is added to ensure regular menstrual cycles. Hormone therapy usually continues until the typical age for menopause (about age 50).

## EDITORS

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The Hormone Health Network offers free, online resources based on the most advanced clinical and scientific knowledge from The Endocrine Society ([www.endo-society.org](http://www.endo-society.org)). The Network's goal is to move patients from educated to engaged, from informed to active partners in their health care. This fact sheet is also available in Spanish at [www.hormone.org/Spanish](http://www.hormone.org/Spanish).

Turner Syndrome Fact Sheet

## If your daughter has TS: Questions to ask her doctor

- What are her treatment options?
- What are the advantages and disadvantages of each?
- What is her likely adult height without growth hormone treatment?
- What conditions should I be watching for?
- Which specialists should be involved in her care?
- Should she see a pediatric endocrinologist?

## RESOURCES

- Find-an-Endocrinologist: [www.hormone.org](http://www.hormone.org) or call 1-800-HORMONE (1-800-467-6663)
- MedlinePlus (National Institutes of Health-NIH): [www.nlm.nih.gov/medlineplus/turnersyndrome.html](http://www.nlm.nih.gov/medlineplus/turnersyndrome.html)
- Eunice Kennedy Shriver National Institutes of Child Health & Human Development (NIH): [www.nichd.nih.gov/health/topics/Turner\\_Syndrome.cfm](http://www.nichd.nih.gov/health/topics/Turner_Syndrome.cfm)
- Mayo Clinic: [www.mayoclinic.com/health/turner-syndrome/DS01017](http://www.mayoclinic.com/health/turner-syndrome/DS01017)
- Turner Syndrome Society: [www.turnersyndrome.org](http://www.turnersyndrome.org)