

Turner Syndrome



Knowledge
to grow by



Turner syndrome

What is Turner syndrome? Turner syndrome is a genetic disorder in girls, present in approximately 1 in 2,500 births.

Turner **syndrome** is named after Henry Turner, a doctor who was the first to identify features of girls with the syndrome in 1938. Turner syndrome is caused by a missing X chromosome or a missing part or structural abnormality of one of the X chromosomes that affect physical sex development, height, and other characteristics.

Turner syndrome

Children with Turner syndrome usually have multiple medical issues. This means that more than one doctor may be involved in managing your child's condition.

What causes Turner syndrome?

The entire body is made of tiny units called cells. Inside each cell are 23 pairs of **chromosomes**. Chromosomes contain a person's **genes**. Genes control traits like hair color and eye color and are passed down from generation to generation.

One of these pairs of chromosomes controls a person's sex. These are the sex chromosomes. In males, this pair contains an X and a Y chromosome: XY. In females, this pair contains two X chromosomes: XX.

In a girl with Turner syndrome, one of the following may be the case:

- There is only one X chromosome, and the other one is missing
- There are two X chromosomes, and part of one of them is missing or it has a structural abnormality

It is also possible for a Y chromosome to be involved in Turner syndrome. A geneticist or genetic counselor can further explain the specifics of your child's chromosome report.



Features of Turner syndrome

Some of the common physical traits and conditions of Turner syndrome are:

- Heart defects
- Short neck with a webbed appearance
- Low hairline at the back of the neck
- Differently shaped, low-set ears
- High-arched palate
- Small jaw
- Drooping eyelids
- Short stature
- Ovarian failure, resulting in failure to progress through **puberty** at a normal age
- Broad chest
- Larger number of moles on the skin
- Lymphedema (puffy hands and feet)

Some girls with Turner syndrome have only a few of these traits. Most have ovarian failure, in which the ovaries do not work properly. Because of this, puberty will generally not occur on its own. Nearly all have short stature.

A girl with Turner syndrome may have legs that are a little shorter than her trunk. On average, the adult height of a woman with Turner syndrome is 4 feet 8 inches. A few women may reach a height of 5 feet.

Like many people with short stature, girls and women with Turner syndrome often find it difficult to keep their weight in a healthy range for their height. Therefore, monitoring weight closely and encouraging exercise and healthy food choices from an early age are important.

Diagnosing Turner syndrome

If your daughter is shorter than average, she may be seen by an **endocrinologist**. This doctor is a specialist who is trained to diagnose and treat children with growth problems. An endocrinologist will perform an evaluation to find the cause of short stature. If there is a medical condition, the doctor can suggest medicines that may help the child grow.

What does it mean if the doctor says your daughter has short **stature** caused by Turner syndrome? It means that your child is shorter than average for her age group.

To diagnose Turner syndrome, a doctor does a blood test to examine a child's genetic makeup. This test is called a **karyotype**. This test counts the number of chromosomes and can identify any that are abnormally shaped or have missing pieces.

Girls may be diagnosed at birth or in childhood, although some may not be diagnosed until much later.

Growth charts

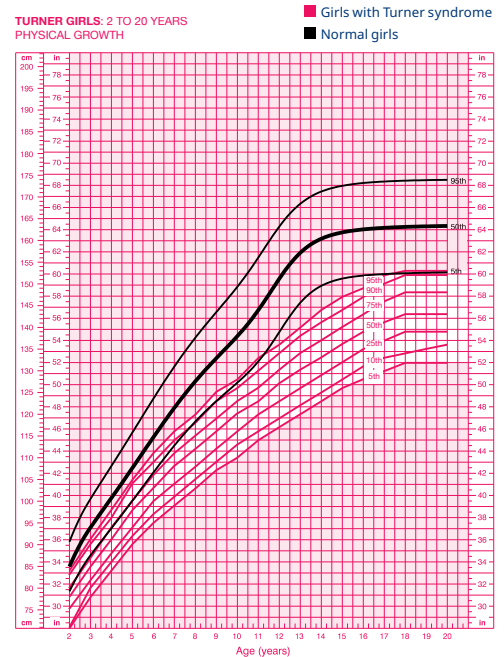
Growth charts are used to track a child's height and weight so that he or she can be compared with the statistical norm. This "norm" is the average height and weight of other children who are the same sex and age. Separate growth charts are used for boys and girls. However, special growth charts are used for girls with Turner syndrome. These are the appropriate tools for your endocrinologist to use in monitoring your daughter's growth compared with other girls with Turner syndrome.

In general, most children grow at least 2 to 2 1/2 inches every year between age 3 and puberty, which, for girls, generally starts between ages 9 and 14.

Each chart has lines called percentile curves, or percentiles. These lines represent the percentage of children at the same height or weight for that age group.

A percentile is a way to show ranking. For example, if a 10-year-old girl is in the 50th percentile for height, that means 50% of 10-year-old girls are taller and 50% are shorter. On the other hand, if a 2-year-old girl is in the 5th percentile for height, that means 95% of 2-year-old girls are taller and 5% are shorter.

To record your child's growth, the doctor will draw a line connecting height measurements for your child at several points. This is called a growth curve. This growth curve is made by comparing your child to children without Turner syndrome. The growth curve for most children usually falls along one of the percentiles on the growth chart. The doctor will also draw similar lines for weight.



Growth chart for girls with Turner syndrome aged 2 to 20 years.

Reproduced from *Archives of Disease in Childhood*, Lyon AJ, 60, 932-935, 1985, with permission from BMJ Publishing Group Ltd.



If it is left untreated, slow growth can continue through childhood and into adulthood. Most girls with Turner syndrome show a decline in growth rate by 3 years of age, and growth continues to be slower than average throughout childhood.

Turner syndrome and puberty

Puberty is when a child's body matures into an adult body. As part of this, the child has a time of very rapid growth. This is called a growth spurt. A girl with Turner syndrome usually will not have a growth spurt because puberty does not generally occur on its own. Your endocrinologist will help you decide the best time to help your daughter go through puberty.

Treatment of short stature due to Turner syndrome

There are treatments available to help girls with short stature due to Turner syndrome reach an adult height that is close to average. Treatment typically lasts for a specific period of time while the potential for growth exists. Duration of treatment also depends on how well the child responds to therapy.

There are also treatment options available to address other symptoms of Turner syndrome, such as late-onset puberty.

Girls with Turner syndrome should be treated as soon as they are diagnosed. To learn more about treatment options for your child, speak with your pediatrician or pediatric endocrinologist.

Follow-up visits

After diagnosing your child with Turner syndrome, the endocrinologist will likely ask you for regular follow-up visits.

Support your child

The self-esteem of children with short stature has much to do with how they see their bodies. Children who feel good about themselves and who feel loved by their families may feel fine about their short stature.

Girls with Turner syndrome may face social challenges because of their height. Remind your child that a person's worth has nothing to do with height. But it has everything to do with who that person is.

Talk to your doctors or see page 13 to learn more about Turner syndrome support groups.



Insurance

Questions about coverage

You may have questions about insurance coverage for treatment of Turner syndrome. There are programs that may help you get the assistance you need. In many cases, assistance is just a phone call away. The manufacturer of your child's treatment may be able to answer your questions about insurance coverage and reimbursement, and may be able to help you find additional coverage to ensure that your child's treatment continues smoothly.

Getting help

With your permission, your doctor may contact the pharmaceutical manufacturer so that you can get help with completing insurance paperwork. The doctor may submit a Statement of Medical Necessity, a recent growth chart (if applicable), and any available insurance information.

Manufacturers' assistance programs

If your doctor has prescribed a treatment option for your child but you are unable to resolve insurance reimbursement issues, the pharmaceutical manufacturer may be able to provide therapy free of charge on a short-term basis. There may also be assistance available from other patient and not-for-profit support organizations.



Use these lines for questions you may have for the doctor, and notes from your conversation.

ADDITIONAL RESOURCES

You can find information and support from these organizations:

Hormone Health Network

The public education affiliate of The Endocrine Society
8401 Connecticut Avenue, Suite 900
Chevy Chase, MD 20815-5817
Tel: 1-800-HORMONE (467-6663)

hormone.org

Turner Syndrome Society

11250 West Road, Suite #G
Houston, TX 77065
Tel: 1-800-365-9944 or 1-832-912-6006

www.turnersyndrome.org

Turner Syndrome Foundation

P.O. Box 726
Holmdel, NJ 07733
Tel: 1-800-594-4585
Fax: 1-800-594-3862
Email: info@tsfusa.org

www.turnersyndromefoundation.org

GLOSSARY

Here are definitions for some words in this brochure that may be new to you. If there are other words that you need to understand better, your child's doctor or nurse can help you.

Chromosomes

Thread-like strands that contain a person's genes.

Endocrinologist

A specially trained doctor who diagnoses and treats diseases of the glands and hormone imbalances.

Genes

The functional units on a chromosome that transmit characteristics from parents to their children.

Karyotype

A test to examine chromosomes in a sample of cells.

Puberty

The growth period when a child becomes a young adult reaching sexual maturity.

Stature

The standing height of a person.

Syndrome

A group of signs and symptoms that form a recognizable pattern of an abnormality.

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