

Turner Syndrome: A Guide for Families

What is Turner syndrome?

Turner syndrome is a genetic condition that affects 1 in every 2,000 to 2,500 live-born girls. Usually, a girl is born with 2 X chromosomes (46,XX) in each cell. A girl with Turner syndrome is missing one whole X chromosome (45,XO) or part of an X chromosome. Sometimes, some cells will have 2 X chromosomes, but other cells have only one (*mosaicism*). This is not an inherited condition, so it is unlikely to happen again in the same family.

What are the common signs and symptoms of Turner syndrome?

Turner syndrome can cause a variety of medical and developmental problems. The signs and symptoms depend on how the cells are affected by changes to the X chromosome.

Typically, only one-third of patients are diagnosed in infancy. Sometimes, a prenatal ultrasound may identify certain abnormalities of the heart, kidneys, or lymph collection of the neck (a *cystic hygroma*). Findings in infancy that may suggest Turner syndrome include

- Puffy hands and feet (lymphedema)
- Extra folds of skin over neck or swelling of lymph vessels in the neck
- Heart abnormalities, particularly coarctation of the aorta

Signs of Turner syndrome leading to diagnosis in childhood may include

- Short stature (seen in all patients)
- Webbing of neck
- Low hairline at the back of the head
- A high-arched roof of the mouth
- An angle at the elbows when the arms are stretched out
- Frequent ear infections and hearing problems
- Failure of puberty to start by age 13 years

Most girls with Turner syndrome have normal intelligence and good reading skills, but some girls with Turner syndrome have problems with math, especially geometry. Other girls with Turner syndrome have social uneasiness.

How is Turner syndrome diagnosed?

Turner syndrome is diagnosed by a special blood test called a *karyotype*. This test examines the chromosomes in a sample of cells. Screening for Turner syndrome is often recommended for girls with heights below the third percentile on the growth chart, especially when combined with some of the physical features listed previously. In girls who are 10 years or older, blood levels of a pituitary hormone called *follicle-stimulating hormone* (FSH) are usually very elevated due to failure of the ovaries to develop normally.

How is Turner syndrome treated?

There is no genetic treatment. However, appropriate medical management can help with the associated medical problems. This requires involvement and coordination of different specialists to address the medical and psychosocial aspects of Turner syndrome.

Without treatment, most patients with Turner syndrome are short (average height of about 4'8"–4'9"). Growth hormone treatment can improve these patients' final adult height. Growth hormone treatment is recommended for girls with Turner syndrome whose height is below the fifth percentile on a growth curve.

Because Turner syndrome can affect the normal development of ovaries, most patients will have little or no breast development, absence of menstrual cycles, and infertility. Estrogen replacement with the help of a pediatric endocrinologist is recommended at an age when girls normally go through pubertal changes. The estrogen replacement treatment will help with breast development, promote menstrual cycles, and improve bone health. For fertility issues, reproductive technologies, such as *in vitro fertilization* (IVF), may be used under the guidance of a reproductive endocrinologist. Importantly, evaluation by a cardiologist is essential before undergoing IVF.

Patients will need to be monitored for heart defects and high blood pressure by a cardiologist at certain intervals throughout life. They also need to be evaluated by a nephrologist for any potential kidney problems or high blood pressure. Audiological evaluations to assess for the development of hearing loss are beneficial. Educational evaluation is helpful to evaluate learning and psychological issues and develop appropriate strategies to maximize the patient's future educational and vocational opportunities.

*Pediatric Endocrine Society/American Academy of Pediatrics
Section on Endocrinology Patient Education Committee*

American Academy of Pediatrics

DEDICATED TO THE HEALTH OF ALL CHILDREN®



PES PEDIATRIC ENDOCRINE SOCIETY