

# Turner Syndrome

## What is Turner syndrome?

Turner syndrome is a genetic condition that affects only girls and women. It occurs when 1 of the 2 X chromosomes normally found in females is missing or incomplete.

## How common is it?

- Turner syndrome is very common, occurring at about 1 out of 2,500 births.
- Approximately 800 infants are diagnosed each year as having it, and 60,000 girls and women in the United States are affected.

## What are some common characteristics of children who have Turner syndrome and of Turner syndrome as children present with it?

- The most common characteristics of Turner syndrome include
  - Short stature
  - Arms that turn out slightly at the elbows
  - Webbed neck
  - Low hairline at the back of the head
- Possible complications of Turner syndrome include
  - Congenital heart disease, especially coarctation (narrowing) of the aorta.
  - Gastroesophageal reflux (GER) (increased spitting up) and difficulty growing and gaining weight.
  - Thyroid problems.
  - Kidney problems.
  - Frequent ear infections.
  - Normal intelligence but often with learning disabilities (eg, visual-spatial weaknesses; problems with copying designs, right/left directions, or math).
  - Younger children may have increased anxiety.
  - Orthopedic problems, including dislocated hips and scoliosis (curvature of the spine).

## Who might be on the treatment team?

- The treatment team for a child with Turner syndrome includes the primary care provider in the medical home.
- Pediatric specialists in genetics, endocrinology, and orthopedics are often involved, and a pediatric specialist in cardiology is sometimes involved.
- Physical therapy may be necessary for children with orthopedic problems.

- Special education targeted at visual-spatial weaknesses and counseling for anxiety may be needed.

## What adaptations may be needed?

### Medications

No special medications are needed for children with Turner syndrome, but the Care Plan may include medications or special exercises for these children, if they have other conditions (eg, heart conditions). See Heart Conditions, Nonstructural, Quick Reference Sheet (page 139) for more details.

### Physical Environment and Other Considerations

- Avoid placing infants in backpack carriers, umbrella strollers, walkers, and jumpers because these can increase GER.
- Many children will appear younger than their same-age peers because of their short statures. Be sure to take their ages into account as you interact with them.
- Infants may need *GER precautions*, which are measures to keep them from spitting up or vomiting their food. These precautions can include keeping the infant upright after feedings, frequent burping, and having the infant sleep with the head of the bed angled up (see Gastroesophageal Reflux Disease [GERD] Quick Reference Sheet [page 129]).
- Many children are at risk for teasing because of their physical appearances. Work to foster self-confidence with the child as well as understanding among the child's classmates.

## What should be considered an emergency?

Children with Turner syndrome can experience sudden shearing of the wall of their aortas (the aorta is the main blood vessel from the heart), which is an emergency. This condition is uncommon, but any severe chest pain or sudden pallor associated with decreased activity should prompt a call to emergency medical services (911).

## What are some resources?

- Medline Plus: “Turner Syndrome” (Web page), <https://medlineplus.gov/ency/article/000379.htm>
- National Institutes of Health, US National Library of Medicine, Genetics Home Reference: “Turner Syndrome” (Web page), <https://ghr.nlm.nih.gov/condition/turner-syndrome>
- Turner Syndrome Society of the United States: [www.turnersyndrome.org](http://www.turnersyndrome.org), 1-800-365-9944

