NATIONWIDE CHILDREN'S®
When your child needs a hospital, everything matters.

Helping Hand™ *Health Education for Patients and Families*

Turner Syndrome

Turner syndrome is a genetic condition that occurs in about 1 in every 2,500 people born female. Normally, those born female have two X chromosomes (**krow**-muh-somz) in each cell. Chromosomes are the building blocks that contain DNA and help determine how we develop.

- In "classic" Turner syndrome, cells have only one X chromosome. The second X chromosome is missing.
- In other forms of Turner syndrome, cells may have two X chromosomes, but they're formed the wrong way or have a piece missing.
- Some children also have mosaicism. This is when some cells have two X chromosomes and some do not.

Signs and Symptoms

Children with Turner syndrome may have a range of different associated conditions. They may be at greater risk of having certain heart, kidney, or endocrine problems. Some signs and symptoms may show up early, while others won't show up until they're older.

Infants	Toddlers to School-Age	Adolescents and Teens
• Puffy hands and feet • Extra skin folds around the neck • Short fingers and toes • Certain heart problems (for example, coarctation of the aorta)	• Learning difficulties • Hearing loss • Many ear infections • Short stature (including shorter than expected compared to other family members)	• Short stature • Delayed or absent puberty • Premature ovarian insufficiency (early menopause) • Loss of fertility (may not be able to get pregnant on their own)

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Diagnosis

Turner syndrome is diagnosed through a simple blood test called a karyotype. This test will count and look at the structure of the chromosomes in your child's cells. Your child's doctor or health care provider may order a karyotype if they have some of the common signs and symptoms of Turner syndrome without other explanations.

Treatment

There is no known cure for Turner syndrome. However, many conditions associated with Turner syndrome can be successfully treated. Treatment may include:

- Growth hormone therapy for short stature
- Estrogen replacement therapy for puberty, including breast growth and periods (menstruation)
- Thyroid hormone replacement
- Regular checkups for monitoring

Follow-up Appointments

- For patients with Turner syndrome, Nationwide Children's Hospital has a comprehensive care clinic with many different providers (including endocrinology, gynecology, genetics, psychology, and others) all in the same place. Call (614) 722-4425 to reach the clinic. This is also the number for the main Endocrinology Clinic.
- Schedule regular follow-up appointments for your child with any other doctors or health care providers as recommended.
- Write down all your questions as you think of them. Bring the list with you to any appointments.