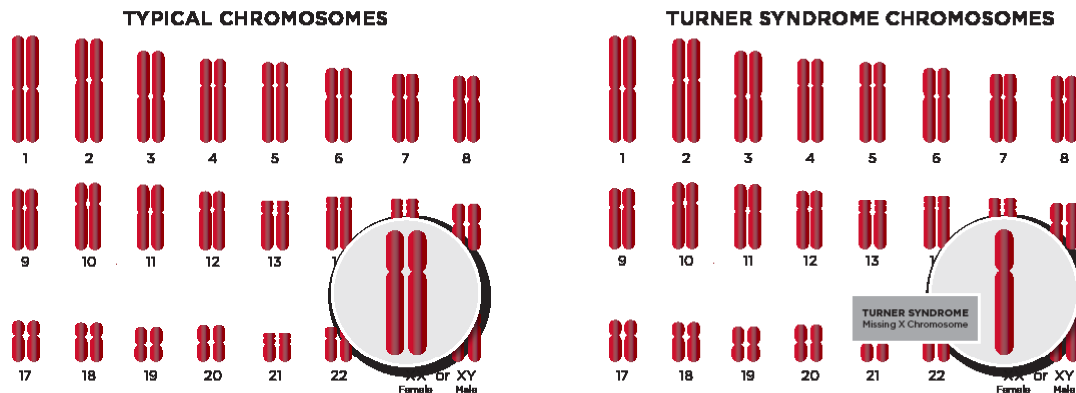


Turner Syndrome

What is Turner syndrome?

Turner syndrome, also known as monosomy X, affects around 1 in every 2,000 to 2,500 live female births. Estimates indicate more than 70,000 women and girls living in the U.S. have Turner syndrome, a chromosome disorder involving the X chromosome.

Chromosomes are the packages of genetic information, made of DNA, that contain the instructions the body uses to build a person. Chromosomes come in 23 pairs, with most people having 46 total chromosomes, which includes a pair of "sex chromosomes." In most females, those sex chromosomes are two "X" chromosomes; however, in most cases of Turner syndrome, the affected individual has only one X chromosome. In other cases of Turner syndrome, there may be only part of the second X chromosome missing, or there may be a structural abnormality involving the second X chromosome. Turner syndrome almost always occurs as a chance event, and rarely ever runs in families.



Turner syndrome has a wide array of symptoms that can affect different organ systems.

The most common features of Turner syndrome include:

- » Short stature
- » Abnormal ovarian development leading to premature ovarian failure
- » Birth defects of the heart, kidneys, and skeleton
- » Wide, webbed neck
- » Swelling associated with blood pressure problems
- » Hormonal irregularities, including thyroid dysfunction

It is important to note that girls with Turner syndrome typically have normal intelligence, although some may experience mild learning disabilities.

A Cardinal Glennon St. Louis Fetal Care Institute nurse is available 24 hours a day, seven days a week to discuss referrals with physicians and potential families by calling 314-268-4037, option 2.

Phone 314-268-4037, option 2
Toll-free 1-877-SSM-FETL (776-3385)
Web stlouisfetalcare.com
Email fetalcare@ssmhealth.com
Facebook facebook.com/fetaldocs

Turner Syndrome

How is Turner syndrome diagnosed?

Turner syndrome is often diagnosed before birth through tests called amniocentesis and chorionic villus sampling (CVS). Both advanced diagnostic tests can be arranged through the Cardinal Glennon St. Louis Fetal Care Institute. Diagnosis can also be made after birth, typically through blood testing.

Genetic screening such as noninvasive prenatal testing (NIPT) identifies pregnancies at increased probability of having Turner syndrome. Additionally, prenatal ultrasound may reveal physical findings associated with Turner syndrome, such as fluid accumulation and swelling as well as heart or kidney abnormalities.

What can I expect if I have been told my unborn baby may have Turner syndrome?

At the Cardinal Glennon St. Louis Fetal Care Institute, you will meet with a team of experts who can provide information about your baby's diagnosis.

This will involve:

- » Maternal-fetal medicine specialists
- » Genetic counselors
- » Clinical geneticists
- » Social workers
- » Footprints coordinators/palliative care nurses
- » Neonatologists

Additionally, based on your unique pregnancy, you may have follow-up visits with other pediatric specialists.

In most cases, a detailed fetal ultrasound will be performed during your first visit to the Cardinal Glennon St. Louis Fetal Care Institute. Amniocentesis is also available if this test has not been performed yet.

A Cardinal Glennon St. Louis Fetal Care Institute nurse coordinator will serve as your primary contact and will coordinate the care of you and your baby throughout the pregnancy.

The goal of the Cardinal Glennon St. Louis Fetal Care Institute team is to provide families with the support, information, knowledge and options to make the decisions that are best for their baby. We recognize that the diagnosis of Turner syndrome affects a family greatly and our hope is to be able to provide families with the resources they need during a difficult and overwhelming time.

How will Turner syndrome affect my baby after delivery?

Because of the complex nature of Turner syndrome, babies often face medical challenges during delivery and in the newborn period, which is why it is recommended that they be delivered at a hospital with the team, experience, and facilities to handle their unique needs. Every baby's situation is different, and the Cardinal

Glennon St. Louis Fetal Care Institute team works with each family to prepare for their baby's delivery and to create a care plan to follow after birth.

How will Turner syndrome affect my baby into childhood?

Turner syndrome doesn't have a cure, but females with the condition can lead full, productive lives with the help of therapies that improve their health and well-being. Those affected by Turner syndrome benefit from regular medical checkups and addressing associated chronic illnesses, such as high blood pressure and hormonal issues.

As your child matures, growth hormone therapy and estrogen therapy may be appropriate. Talk to an endocrinologist before choosing a course of treatment.

What are the treatment options for babies with Turner syndrome?

Treatment of Turner syndrome is customized for each female and involves a comprehensive medical plan that addresses each patient's specific symptoms. Early intervention is important in ensuring children with Turner syndrome reach their potential.

Babies with Turner syndrome will likely need the care of a multidisciplinary medical team, which may include:

- » Pediatricians
- » Medical geneticists
- » Cardiologists
- » Surgeons
- » Endocrinologists

Preparing for the birth of a baby with Turner syndrome can be overwhelming and scary. That's why we're available to help 24 hours a day, 7 days a week. For more information or to schedule an appointment, call us at 314-268-4037, opt. 2 or toll free at 877-SSM-FETL (877-776-3385).
