

Introduction

Turner syndrome is a genetic disorder that affects a girl's development. Girls who have it are short, and most are infertile. Girls and women with Turner syndrome are at risk for many health problems, including high blood pressure, kidney problems and diabetes. There is no cure. But treatment can help reduce symptoms.

This reference summary explains Turner syndrome. It covers the symptoms and causes of Turner syndrome, as well as diagnosis and treatment options.



What is Turner Syndrome?

Turner syndrome is a genetic disorder. This means that it is caused by a problem with a person's genes. But it is generally not inherited. This means that it is not passed on from parents to daughter. Turner syndrome affects how a girl grows and develops. It does not affect males. About 1 of every 2,500 females born worldwide each year has Turner syndrome. This disorder affects all races and regions of the world equally. There are no known environmental risks for Turner syndrome.

In most cases, women with Turner syndrome are infertile. This means that they cannot get pregnant naturally. Turner syndrome cannot be prevented. It is a genetic problem that is caused by a random error. There is nothing the father or mother can do to prevent the error from happening. But there are many options for treatment.



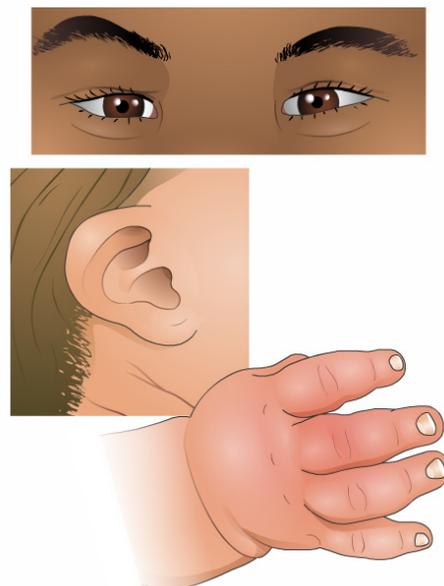
This document is for informational purposes and is not intended to be a substitute for the advice of a doctor or healthcare professional or a recommendation for any particular treatment plan. Like any printed material, it may become out of date over time. It is important that you rely on the advice of a doctor or a healthcare professional for your specific condition.

Symptoms

Turner syndrome causes a variety of symptoms in girls and women. For some people, symptoms are mild. For others, Turner syndrome can cause serious health problems. At birth, a girl with Turner syndrome may be slightly shorter than average. She may also have swelling of the hands and feet.

Signs and symptoms of Turner syndrome in babies may include:

- Arms that turn outward at the elbows.
- Drooping eyelids.
- Fingernails turned upward.
- Low-set ears.
- Sensitivity to noise.
- Short fingers and toes.
- Broad chest with widely spaced nipples.
- Low hairline in the back.
- Narrow roof of the mouth.
- Small lower jaw.
- Wide or web-like neck with folds of skin from tops of shoulders to sides of neck.



Delayed growth and sexual development is also common in girls with Turner syndrome. They may not have growth spurts that are common in childhood. They may also not start their periods or develop breasts during puberty.

Signs and symptoms of Turner syndrome in older girls and teens can include:

- Difficulty in social situations.
- Learning disabilities, often accompanied by normal intelligence.
- Shorter height, with an adult height of about 8 inches or 20 centimeters less than normal for her family.
- Shorter menstrual cycles.



This document is for informational purposes and is not intended to be a substitute for the advice of a doctor or healthcare professional or a recommendation for any particular treatment plan. Like any printed material, it may become out of date over time. It is important that you rely on the advice of a doctor or a healthcare professional for your specific condition.

Most women with Turner syndrome are also unable to get pregnant without fertility treatment.

Causes

Turner syndrome happens when there is a problem with the genes. Genes tell the body to make certain substances. Every person has thousands of genes. Our genes make us look the way we do. They also have a lot to do with our health. Most people have 46 chromosomes in each cell. Half of these are from their mother. The other half are from their father. This means 23 chromosomes come from each parent.



The 23rd pair of chromosomes are called the sex chromosomes. They determine whether a person is male or female. Females have two X chromosomes (XX) in most of their cells. Males have one X chromosome and one Y chromosome (XY) in most of their cells.

Turner syndrome most often happens when a female has one normal X chromosome, but the other X chromosome is missing. Other forms of Turner syndrome happen when a part of one of the two chromosomes is missing or changed in some way. The error that leads to the missing chromosome usually happens during the formation of the egg or sperm. Because a girl with Turner syndrome is missing part or all of a chromosome, certain genes are missing. The loss of these genes leads to the symptoms of Turner syndrome.



Sometimes, girls with Turner syndrome have some cells that are missing one X chromosome and some cells that are normal. This is because not every cell in the body is exactly the same. This condition is called mosaicism. If the second sex chromosome is lost from most of a girl's cells, then it is likely that she will have symptoms of Turner syndrome. If the chromosome is missing from only some of her cells, she may have no symptoms or only mild symptoms.

This document is for informational purposes and is not intended to be a substitute for the advice of a doctor or healthcare professional or a recommendation for any particular treatment plan. Like any printed material, it may become out of date over time. It is important that you rely on the advice of a doctor or a healthcare professional for your specific condition.

Complications

Turner syndrome can cause complications that may affect many different parts of the body. Hearing problems and ear deformities are common in people with Turner syndrome. They may need hearing aids as children or adults. Girls with Turner syndrome may get ear infections easily.

The heart can also be affected. About five to ten percent of girls with Turner syndrome have a narrowed aorta. The aorta is the main blood vessel that moves blood out of the heart to the rest of the body. The narrowed aorta can be corrected with surgery. About 30% of girls with Turner syndrome have an abnormal valve between the heart and the aorta. The abnormal valve usually does not cause symptoms. But in some cases, it can lead to infection of the valve. It may also damage the aorta.



Kidney problems happen in about one third of people with Turner syndrome. This can lead to an increased risk for urinary tract infections. People with Turner syndrome are at high risk for type 2 diabetes. For this reason, women with Turner syndrome should be checked regularly for diabetes.



Many women with Turner syndrome have osteoporosis. This is a condition that causes weak bones and bone fractures. Women with Turner syndrome are at a higher risk for osteoporosis because their bodies do not make enough estrogen.

Thyroid disorders happen in about one third of people with Turner syndrome. Medicines can treat this condition. Gluten intolerance and celiac disease can also happen in people with Turner syndrome. These conditions can be managed by eating a diet that is free of gluten.

Diagnosis

Your health care provider will first ask about symptoms and medical history. He or she will also perform a physical exam.

This document is for informational purposes and is not intended to be a substitute for the advice of a doctor or healthcare professional or a recommendation for any particular treatment plan. Like any printed material, it may become out of date over time. It is important that you rely on the advice of a doctor or a healthcare professional for your specific condition.

Health care providers base a diagnosis of Turner syndrome on:

- Physical symptoms.
- The results of a genetic blood test.

The genetic blood test used to diagnose Turner syndrome is called a karyotype test. It shows if one of the X chromosomes is partially or completely missing. Turner syndrome can be diagnosed during pregnancy.



Newborns may be diagnosed after heart problems are detected or after certain physical features are noticed. Some girls may be diagnosed as teenagers because of a slow growth rate or because puberty does not start.

Treatment

There is no cure for Turner syndrome. But treatments can help control and lessen symptoms. If Turner syndrome is diagnosed in early childhood, human growth hormone may be given. These hormone injections can often increase adult height by a few inches. Estrogen replacement therapy can help start sexual development that normally begins at puberty. This includes breast development and the development of wider hips.

Health care providers may prescribe a combination of estrogen and progesterone to girls who have not started their periods by age 15. Estrogen also provides protection against bone loss. Regular health checks are important to care for the various health problems that can result from Turner syndrome.



Summary

Turner syndrome is a genetic disorder that affects a girl's development. Girls who have it are short, and most are infertile. In some people, Turner syndrome can cause serious health problems, such as a narrowed aorta or kidney problems.

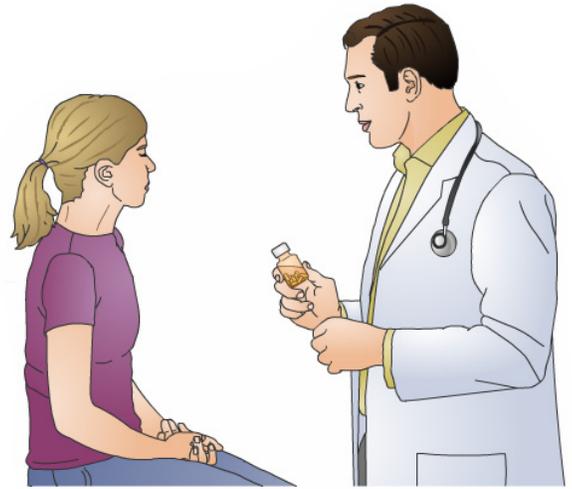
This document is for informational purposes and is not intended to be a substitute for the advice of a doctor or healthcare professional or a recommendation for any particular treatment plan. Like any printed material, it may become out of date over time. It is important that you rely on the advice of a doctor or a healthcare professional for your specific condition.

Turner syndrome happens when there is a problem with the genes. Turner syndrome most often happens when a female has one normal X chromosome, but the other X chromosome is missing. Other forms of Turner syndrome happen when a part of one of the two chromosomes is missing or changed in some way.

Health care providers base a diagnosis of Turner syndrome on:

- Physical symptoms.
- The results of a genetic blood test.

Some girls are diagnosed as newborns. Others may not be diagnosed until their teenage years. Girls and women with Turner syndrome are at risk for many health problems, including high blood pressure, kidney problems and diabetes. There is no cure. But treatment can help reduce symptoms. Treatment often includes hormone replacement.



This document is for informational purposes and is not intended to be a substitute for the advice of a doctor or healthcare professional or a recommendation for any particular treatment plan. Like any printed material, it may become out of date over time. It is important that you rely on the advice of a doctor or a healthcare professional for your specific condition.