

Turner syndrome: What you need to know

Turner syndrome is a genetic condition that affects the body's chromosomes and how girls and women develop. In this handout, you will learn about Turner syndrome, including its causes, symptoms, diagnosis and treatment.

WHAT IS TURNER SYNDROME?

Turner syndrome (TS) is a genetic condition in girls in which one X chromosome or part of one of the X chromosomes from some or all cells of the body is missing. This can lead to problems with growth and physical development.

WHAT CAUSES TS?

No one knows why girls are born with TS. TS almost always happens at random. TS is not passed down through families.

What are chromosomes and how do they work?

Most girls have two X chromosomes and 46 chromosomes in total. Most boys have one X and one Y chromosome with 46 chromosomes in total. The X and Y chromosomes are called **sex chromosomes**. The other 46 chromosomes are called **autosomes**. These chromosomes contain all the genes that control the information for development and how our bodies work throughout life. People with differences in chromosomes often have differences in how their bodies develop.

Girls with TS may have only 45 chromosomes in some or all the cells in their bodies. This is because they have lost an X chromosome or a piece of an X chromosome in some or all their cells. This is fairly common, occurring in about 1 in every 2,500 girls.

WHAT ARE THE SYMPTOMS OF TS?

Many girls with TS have no health problems, but they can be shorter than average. There are some other health issues that may come with TS. These can be evaluated and treated by your child's care team. Some health issues can include:

- **Underdeveloped ovaries (reproductive organs).** This can lead to delayed or absent puberty. Girls with TS might need medications to help start the puberty process.
- **Puffy neck or swollen hands and feet at birth**
- **Problems with the heart, hearing, how the thyroid works and certain gastrointestinal (stomach and intestine) conditions.**
- **Kidneys that have developed differently.** Sometimes there is only one kidney or the 2 kidneys are joined together. This is called **horseshoe kidney** because of how the joined kidneys are shaped.
- **Learning differences,** especially with math or learning directions. These differences do not typically affect overall intelligence.
- **In adults, trouble with fertility.** Girls with TS who do not go through puberty on their own may have trouble getting pregnant as adults. This is because their bodies do not produce eggs to be fertilized. These girls are otherwise entirely normal and can have normal, happy lives and sex lives when they are of an appropriate age. Choices for having children are changing with the years. Although many young women with TS choose to adopt children, other ways to have children may be available. When your child is of an appropriate age, have them ask the care team about what choices are available.

Information on diagnosis, treatment and additional resources on the back >>>

HOW DO DOCTORS DIAGNOSE TS?

Diagnosis of TS can happen at any age, depending on your child's symptoms and development. Doctors usually diagnose TS through a blood test, a cheek swab (a cotton swab rubbed against the inner cheek) or both. The cells from the sample are then grown and analyzed for chromosomes in the laboratory. It can be hard to diagnose TS just through checking your child's medical symptoms.

Before birth

Turner syndrome may be diagnosed before birth if a pregnant woman has prenatal genetic testing to check her baby's developing chromosomes.

After birth

After birth, chromosome tests to check for TS might be done if a female baby has a puffy neck, hands or feet, or is born with heart disease.

During childhood

Other girls are diagnosed because they are short and growing slowly or because they do not start puberty at a normal age. In general, it is hard to tell what other medical conditions a person with TS may have based on the chromosome test.

HOW DO DOCTORS TREAT TS?

Treatment usually involves medication to help your daughter grow taller and go through puberty. There is no cure for TS, but treatment can help with growth and development. Other treatments help treat medical conditions that can occur with TS, such as kidney or heart issues.

Height

Girls with TS can grow taller if they and their parents wish. This is done through daily injections of a medication called **growth hormone**. Your daughter will receive growth hormone during childhood while she is growing. These injections are given at home with a very small needle. Most children adjust to the small injections over time. These injections are not usually painful.

Puberty

If your daughter with TS does not start puberty on their own, they can receive medications that help bring on physical signs of puberty. These medications are female hormones known as **estrogen** and **progesterone**.

Other medical conditions

Treatment for other conditions depends on your daughter's condition and symptoms. They are treated the same way in girls with TS as they are in people without TS. This can include heart disease, kidney issues, thyroid problems, hearing difficulties, extra weight gain, diabetes, learning problems in school, anxiety and trouble dealing with stress.

WHERE CAN I LEARN MORE ABOUT TS?

- **Your daughter's care team at MGHfC**

- **The Turner Syndrome Society of the United States**
www.turnersyndrome.org

This organization is comprised of people with TS, their families and medical professionals. There are many local chapters and an annual meeting for both TS women and their families and professionals. This organization has been instrumental in developing and updating international guidelines for management.

- **The Turner Syndrome Foundation**
www.turnersyndrome.org

This organization is composed of people with TS and their families and organizes many worthwhile activities and supports.

- **Boston International Turner Syndrome Summit**
www.bitssummit.org

This is an 8-day residential learning experience for girls with TS ages 12-18 offered each year at the end of July and beginning of August. It is organized by people with TS and parents of girls with TS. It is a wonderful bonding and learning experience. Doctors from MGHfC participate in this program.

- **The Magic Foundation**
www.magicfoundation.org

Help and information for many different disorders of growth including Turner Syndrome

- ***Standing Tall with Turner Syndrome* by Claudette Beit-Aharon**

This book is edited by a mother local to Boston and has wonderful stories of growth and resilience written by girls who have Turner syndrome.

Pediatric Endocrine and Diabetes Center

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www.massgeneralforchildren.org/endocrine

For more information please call

617-726-2909

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