About Turner Syndrome

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Turner syndrome is variable, and each girl and woman will have unique health needs and characteristics.

Turner syndrome (TS) occurs in approximately one out of every 2,000-4,000 female live births. It is a chromosomal condition describing girls and women with common features, physical traits and medical conditions caused by the complete or partial absence of the second sex chromosome. TS was first described in the United States in 1938 by Dr. Henry Turner.

At the basic level, the missing genetic material prevents the female body from growing and maturing naturally. Turner syndrome is variable, and each girl and woman will have unique health needs and characteristics. Most girls and women have two complete X chromosomes (the sex chromosomes for females). Turner syndrome is caused by the absence of all or part of the second sex chromosome in some or all of the cells of the body. The most consistent features of TS are short stature and lack of ovarian development, however, there are other symptoms and characteristics that can appear in varying degrees, depending on each person’s unique genetic makeup.

Symptoms and Characteristics of TS

Medical concerns:

- chronic middle ear infections
- hearing loss
- distinctive heart, liver, and kidney abnormalities
- autoimmune disorders, such as underactive thyroid and celiac disease
- difficulty with nonverbal communication skills, spatial relationships, such as driving or riding a bike, and executive functions

Facial appearance, such as:

- down-turning eyelids
- low-set and prominent ears
- small jaw
- high, narrow roof of mouth

Other characteristics:

- neck webbing (short, thick neck)
- short stature
- delayed puberty
- reduced fertility
- lymphedema (puffy hands and feet)
- turned up nails
- short roots of teeth

The specific karyotype (genetic makeup of the chromosomes) does not always predict the appearance or medical symptoms of a person with TS.

What Causes Turner Syndrome?

Turner syndrome occurs when all or part of one of the X chromosome is lost before or soon after the time of conception. To read a more detailed description of how this happens, please refer to the Guide for Families, pages 3-6.

There is nothing that either parent did before or at the start of the pregnancy that caused or increased the risk of TS for their daughter. The age of a parent, ethnicity, diet, or other factors are unrelated to the conception of a child with Turner syndrome.
The Clinical Practice Guidelines for TS recommends:

- All females with suspected TS should have a genetic blood test, called a karyotype.
- If mosaicism (a form of TS) is strongly suspected, but not confirmed with a standard karyotype, additional genetic tests should be done to confirm or rule out TS.
- A blood karyotype is usually adequate, but skin cells or cheek (via swab) cells may be examined if there is an ongoing suspicion of TS or low-level mosaicism.

A final diagnosis is made by a blood test, called a karyotype, which looks at the complete set of chromosomes of an individual. A karyotype analysis determines if one of the X chromosome pairs is missing from the full chromosome set, or if there are any structural differences in the X chromosomes.

Before birth, a diagnosis of TS may be made by a maternal serum screening, by taking a sample of amniotic fluid or other fetal tissue to look at the fetal karyotype or an ultrasound. Because ultrasound and maternal serum screening (blood tests) are not 100% reliable, genetic karyotype testing is essential after the baby is born so that the diagnosis of TS can be confirmed.

There are a range of health, developmental, social, and learning challenges which might affect girls and women with TS to different degrees, so it is important to diagnose TS as early as possible.

Early diagnosis helps doctors determine whether the girl or woman has health issues that need treatment or need to be followed.

NOTE: Males may have a 45,X/46,XY male mosaic karyotype, but this is not Turner syndrome. A TS diagnosis only applies to phenotypical females (those born with female sexual organs internally and externally). TSSUS professional advisors disagree with the practice of diagnosing males with TS and encourage physicians to contact TSSUS to consult with a TS genetic specialist for questions related to TS karyotypes.

Treating Turner Syndrome

TS is a chronic rare condition (clinically it is considered a disease, but of course, not infectious), associated with a range of health concerns. The Clinical Practice Guidelines for the care of girls and women with Turner syndrome is a 70-page document describing health care recommendations for physicians and providers of care. The Patient and Family version of the Clinical Practice Guidelines is a 16-page document to support the partnership between those with TS, parents of girls with TS and medical providers.

TS is not directly treated by a doctor, but the individual health concerns that arise for each girl or woman with TS should be monitored and treated by an appropriate specialist.

Treatment should be individualized and physicians, families, and patients should decide on treatment options together.

Major advances in treatment include growth hormone therapy to achieve greater height, and estrogen replacement therapy to mimic natural estrogen.

Other treatments specific to TS include advances in heart surgery, hearing evaluations, in-vitro fertilization, and social and psychological testing. Connecting with others who share this unique bond and who have similar experiences and concerns is of great benefit. With the help of medical specialists and a good social support system, a woman with TS can expect to live a satisfying, healthy life.