What is Turner Syndrome?

Turner syndrome (TS) occurs in approximately one in 2,000 live female births. TS is a chromosomal condition that describes girls and women with common features, physical traits and medical conditions that are caused by the complete or partial absence of the second sex chromosome.

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.

Causes

Turner syndrome occurs when all or part of one of the X chromosomes is absent or altered before or soon after the time of conception. It is not connected to or passed on from either parent, and there is nothing a person can do to increase or decrease the likelihood of their child having Turner syndrome.

Diagnosis

Turner syndrome can be diagnosed before birth with an amniocentesis, or any time during their life (prior to age 50) with a specific blood test called a karyotype. A karyotype shows the number and visual appearance of the chromosomes found in the cells of a person. Turner syndrome is usually diagnosed before or during teen years because of short stature and a lack of pubertal changes.

Key Traits & Conditions

TS conditions may include, but are not limited to:

• Delayed puberty • Heart defects such as aortic coarctation and bicuspid aortic valve • Edema (swelling) of the hands and feet, especially at birth • Infertility due to non-functioning ovaries • Kidney, thyroid and liver concerns • Hearing loss • Frequent ear infections • Normal intelligence with specific learning disorders • Psychosocial challenges • Scoliosis • Celiac disease

TS physical traits may include, but are not limited to:

• Short stature (under 5 ft.) • Broad, short neck • Receding lower jaw • Multiple moles • Low-set ears

Treatment

The treatment for those with Turner syndrome should be individualized. Physicians, families and patients should decide on treatment options together.

Major advances in the treatment of Turner syndrome have made a significant difference in the lives of girls and women with TS. Many young girls and teenagers diagnosed with TS start on growth hormone therapy to achieve greater height. Puberty is initiated at the same time as peers with the use of estrogen replacement therapy. Estrogen therapy is continued for a multitude of reasons, including the prevention of osteoporosis. Continuous heart, liver and thyroid evaluations are needed from birth throughout adult life.

For the complete treatment guidelines, see the "Clinical Practice Guidelines for Turner Syndrome" found at www.turnersyndrome.org.

Turner
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Advances in heart surgery have improved the lives of hundreds of girls and women with TS. Because hearing loss often occurs early in Turner syndrome, hearing evaluations are critical. The social and psychological impact of Turner syndrome cannot be underestimated and should be a priority in the care of any girl or woman with TS. 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.

Research

TSSUS is advancing research through the Turner Syndrome Research Registry (TSRR). TSSUS advocates for research that will support the needs of the TS community, shared information among researchers and collaboration of research within the Turner Resource Network. The Turner Research Registry was created to support research, but more importantly, to sustain research for years to come. The TSRR is solely funded and managed by TSSUS, and is endorsed by our partners in the Turner Resource Network.
More information can be found about the Registry at www.turnersyndrome.org

The Turner Syndrome Society of the United States (TSSUS) is a national 501(c)(3) nonprofit organization dedicated to providing support and health-related information to those with Turner syndrome (TS), their families and the medical community. TSSUS aids in the diagnosis and treatment of TS.

TSSUS is the largest grass-roots Turner syndrome organization in the world, with more than 30 years of dedicated service to the TS community. TSSUS is the leading authority on the condition.

Key Programs Include:

- Annual TS Patient & Family Conference
- TS Research Registry
- TS Days Regional Events
- Local Resource Groups across the Country
- Turner Syndrome Professional Symposiums
- TSSUS Chasing Butterflies Walks for Awareness
- Providing comprehensive resource materials
- Personally assisting the TS community

Our Mission:

The Turner Syndrome Society of the United States advances knowledge, facilitates research and provides support for all touched by Turner syndrome.

Our Vision:

Every woman and girl with Turner syndrome will have cutting edge health care ensuring the best quality of life.

Turner Syndrome Society of the United States®

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https://www.facebook.com/turnersyndromesociety/

TSRR Turner Syndrome Research Registry

Turner Resource Network

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