What Is Turner Syndrome?

Turner Syndrome (TS) is a condition that affects only girls and women. It is identified by a difference in the genetic make-up of those who are affected. 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 X 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. A synthetic form of human growth hormone (GH) is available for medical use. GH is sometimes given to girls with TS to help them grow. GH therapy may be used as treatment in cases of growth hormone deficiency, or short stature. At the typical age of puberty, girls with Turner Syndrome are usually prescribed hormone replacement therapy to induce normal female developmental changes. This therapy is often continued through most of the adult years.

What Causes Turner Syndrome?

Turner Syndrome occurs when a piece of genetic information (part or all of the X chromosome) gets "dropped" during a process called meiosis, when sex cells divide to form sperm in males, and eggs in females. To date, TS is not thought to be associated with environmental factors or any other factors generally associated with genetic problems, such as advanced maternal age.

Diagnosing Turner Syndrome

Girls and women are diagnosed at various stages of life, from the prenatal stage to adulthood. The age of diagnosis has been decreasing with better awareness of TS in the medical community. Usually, there is something ‘different’ about the girl or woman that makes the parent or the doctor investigate the possibility of TS. The diagnosis of is made by a blood test which looks at the complete set of chromosomes of the individual. This is called a karyotype. Karyotype analysis shows if one of the X chromosome pair is missing from the full chromosome set, or if there are any structural differences in the X chromosomes. Before birth, diagnosis of TS can be made by taking a sample of amniotic fluid or other fetal tissue to look at the fetal karyotype. Ultrasound is also used to screen for patterns often seen in TS, such as fluid buildup around the neck, and kidney or heart abnormalities. After birth, diagnosis of TS is confirmed by taking a sample of blood or other tissue to obtain a karyotype. Because there is a range of health, developmental, social, and learning challenges which might affect girls and women with TS to different degrees, 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.

Recognizing Turner Syndrome

These are the most common reasons that prompt an investigation into whether a girl or woman has TS, but they are not the only reasons:

Infants: Small size, puffy hands and feet, extra skin folds at the side and back of neck, and heart abnormalities.

Young children: Small size in relation to peers (below “normal” growth chart for both height and weight), as well as other signs such as recurrent ear infections or hearing problems.

Teens: Small stature and absence of development of breast tissue or menstruation at expected age.

Adults: Irregular menstruation, problems with fertility, small stature, as well as issues with hearing, heart, or blood pressure.

DID YOU KNOW?

The earlier that learning and health problems are identified, the better the chance that the person with TS can be given the help she needs in order to succeed.
Physical Characteristics and Health Issues

Each individual with TS is unique. However, there are certain characteristics, either potential issues or physical attributes, which are linked to TS. Please note that, except for details under "Consistent findings with TS", all other characteristics are variable, and are not seen in every individual with TS. Different karyotypes will result in a different likelihood of these findings.

**Consistent findings with TS**
- Short stature (average adult height is 4’8” or 142cm)
- Infertility (a small percentage of women with TS do conceive naturally)

**Possible Physical characteristics (other than stature)**
- Broad chest
- Wide and short neck with excess skin that joins the neck with collar bone ("neck webbing")
- Lymphedema, or fluid built-up in hands and feet

**Social and Behavioral Issues**
- Feeding problems in infancy and childhood
- Visual and spatial learning challenges
- ADHD / Nonverbal Learning Disability
- Difficulty with social skills

**Potential Health Issues**
- Hearing problems, including chronic or recurrent middle ear infections hearing loss
- Kidney and urinary tract abnormalities
- Heart abnormalities
- Hypothyroidism
- High blood pressure
- Obesity

**Less Likely Health Issues (less than 20%)**
- Eye problems such as ptosis,(eyelid drooping) or strabismus("lazy eye")
- Curvatures of the spine, scoliosis/kyphosis
- Diabetes, types 1 and 2
- Osteoporosis and increased likelihood of fractures
- Cataracts
- Autoimmune disease conditions such as celiac disease and osteoarthritis

**Treatment for Turner Syndrome**

TS is not a disease, but a genetic condition that is associated with a range of health concerns. 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 the appropriate specialist. For example, girls and women with the heart conditions common in TS should be followed by a cardiologist. New medical developments allow women with TS who are interested in starting a family to seek the help of an obstetrician who specializes in in-vitro fertilization. A girl with TS may get growth hormone under a pediatric endocrinologist. Some areas have coordinated TS clinics where patients see various specialists in the same day. These may be found at www.turnersyndrome.org.

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**Are there different types of Turner syndrome?**

Girls who are missing one complete X chromosome have "classic" TS, or 45,X karyotype.

A karyotype is the complete set of chromosomes in a person’s cells. Over half (about 54%) of women with Turner Syndrome have the 45,X karyotype.

Others are missing only part of the second X chromosome, or have some structural rearrangements of the chromosome or have a mosaic karyotype, which refers to a missing or rearranged X chromosome in some but not all cells in the body.

It is important for someone with TS to know her karyotype since different karyotypes are associated with different potential health issues.

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**References**


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**For More Information**

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**Thank you to the Turner Syndrome Society of Canada.**