

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.

Are there different types of Turner Syndrome?

Girls who are missing one complete X chromosome have "classic" TS, often referred to as an "XO" or 45,X karyotype. Others are missing only part of the second X chromosome, or have some structural rearrangements of the chromosome. A mosaic karyotype 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 their karyotype since different karyotypes are associated with different potential health issues.

How common is Turner Syndrome?

TS affects approximately 1 in every 2,000 - 2,500 females born in the population. Thus we would expect that there are over 6,000 individuals with TS in Canada.

What causes Turner Syndrome?

Turner Syndrome occurs when a piece of genetic information gets "dropped" during division of the sex cells - or during the process of meiosis. To date, TS is not thought to be associated with environmental factors or any other factors generally associated with genetic problems.

Can Turner Syndrome be 'passed down' in a family?

TS is not an inherited condition and, therefore, is not passed down from one generation to another. In rare cases, a family may have more than one child with TS, but in those cases the loss of a sex chromosome during cell division happened twice to the same family by chance. In fraternal twins, it is possible for one twin to have TS and the other not to have TS.

What signs indicate TS?

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 child - small size in relation to peers (often off the "normal" growth chart for both height and weight) Other signs such as recurrent ear infections or hearing problems

Teen - small stature, does not develop breast tissue or menstruate at expected age

Adult - does not menstruate regularly, problems with fertility, small stature in conjunction with hearing, heart or blood pressure problems

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How is Turner Syndrome diagnosed?

Diagnosis of TS is confirmed by taking a sample of blood, amniotic fluid, or other tissue, and examining the genetic material. The resulting genetic 'map' is called a karyotype. Girls and women are diagnosed at various stages of their life, from the prenatal stage to adulthood. The age of diagnosis has been decreasing with better awareness of TS in the medical community. Most girls with TS now are diagnosed at birth or when they are very young. Usually, there is something 'different' about the girl or woman that makes the parent or the doctor investigate the possibility of TS

Why is it important to know if someone has Turner Syndrome?

The absence of all or part of one X chromosome is associated with a range of health, developmental, social and learning challenges which affect girls and women with TS to different extents. It is important to diagnose TS as early as possible so that the doctor can determine whether the girl or woman has health issues that need treatment or need to be watched. Also, 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.

Can Turner Syndrome be treated?

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 the supervision of a pediatric endocrinologist.

(see the TS Health Care Checklist, or the InfoClip on "What health care should a person with TS receive?)

What health & social issues could be associated with TS?

Each individual with TS is unique. This list presents some of the potential issues which may or may not be present

- Short stature; average height is 4'8"
- Infertility
- Heart abnormalities
- Hearing problems including chronic or recurrent middle ear infections and hearing loss
- Feeding problems in infancy & childhood
- Kidney and urinary tract differences
- Diabetes
- Puffy hands and feet, edema may persist into adulthood
- Pigmented moles on skin
- Eye problems (drooping eyelids, "lazy" eye)
- Cataracts
- Wide and short neck with excess skin that joins the neck with collar bone ("neck webbing")
- High arched palate with associated dental problems
- Broad chest
- Curvature of the spine (scoliosis)
- Overweight
- Brittle bones (osteoporosis)
- High blood pressure
- Visual spatial learning challenges
- Difficulty with social skills

For more detailed information, see our information book "Turner Syndrome: Across the Lifespan" which can be ordered at www.turnersyndrome.ca