

## A Patient Guide to Turner Syndrome

### What Is Turner Syndrome?

First described by pioneer endocrinologist, Dr. Henry Turner in 1938, Turner syndrome (TS) is a genetic condition that affects only girls and women. Females typically have two complete X chromosomes (the sex chromosomes). 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.

### 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 known to be associated with environmental factors or any other factors generally associated with genetic problems, such as advanced maternal age.

### How is Turner Syndrome Diagnosed?

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 TS 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, other fetal tissue, or maternal blood 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.



**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.

### Recognizing Turner Syndrome

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

- Prenatal** Can be discovered through prenatal blood tests. Excess fluid, thickening of skin at back of the neck, shorter limbs, or heart defect may be detected during routine ultrasound.
- Infants** Small size, puffy hands and feet, extra skin folds at the side and back of neck, or heart abnormalities.
- Children** Small size in relation to peers (below “normal” on 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.

## Physical Characteristics and Related Health Conditions

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 in TS

Short stature (average adult height is 4'8" or 142cm)  
Infertility (a very small percentage of women with mosaic TS do conceive naturally)

### Possible Physical Characteristics

Broad chest  
Wide and short neck with excess skin joining the neck and collar bone, also known as "neck webbing."  
Lymphedema, or fluid build-up in hands and feet

### Common Social and Behavioral Differences

Visual and spatial learning challenges, such as math, riding a bike, or driving  
ADHD  
Difficulty understanding sarcasm, jokes, social cues, and nonverbal communication, such as facial expressions and body language  
Difficulty with social skills, such as personal space, making conversation, and maintaining eye contact  
Anxiety and obsessive tendencies, such as rumination and repetitive questions.

### Are There Different Types of Turner Syndrome?

Girls who are missing one complete X chromosome have what is called monosomy X, often referred to as "classic TS."

The karyotype is 45X, meaning they have 45 of the possible 46 chromosomes, and only one of the two X chromosomes.

Others are missing only a part of the second X chromosome, or have some structural rearrangements of the chromosome.

Girls and women who have a missing or rearranged chromosome in some, but not all cells of the body have a mosaic karyotype.

An individual's karyotype can determine which of the potential features or health concerns she might have.

### Potential Health Concerns

Feeding problems in infancy and childhood  
Chronic or recurrent middle ear infections  
Conductive and/or sensorineural hearing loss  
Heart abnormalities  
Hypothyroidism  
Hypertension (high blood pressure)  
Obesity

### Less Likely Health Concerns

Diabetes, Types 1 and 2  
Vision/eye problems, such as ptosis (eyelid drooping) or strabismus ("lazy eye")  
Osteoporosis and increased risk of fractures  
Cataracts  
Autoimmune disease such as celiac, osteoarthritis, or Crohn's

## Treatment for Turner Syndrome

TS is a chronic rare 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.

### For more information contact us:

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