



Turner syndrome (sometimes called 45,X or monosomy X) occurs when a girl has complete or partial absence of her second X chromosome in some or all of her cells. This missing chromosome causes them to be shorter than average in height and can affect how their ovaries work. Each girl and woman with Turner syndrome are unique, but are more likely than average to have other health issues including heart and kidney problems, autoimmune disorders, hearing and vision loss, and other health issues.

Individuals with Turner syndrome typically have normal intelligence, although some may have specific learning disabilities and speech or expressive language delays. Turner syndrome is variable, meaning some individuals have more significant health and developmental concerns while others have less.

### **What Causes Turner syndrome?**

The majority of Turner syndrome occurs when there are differences with formation of a parent's egg or sperm. Some instances of Turner syndrome can be caused by errors that happen when the cells are dividing very early in a baby's development. The exact reason for these occurrences is not known. While many girls and women with Turner syndrome will be missing the second X chromosome in all of the cells in their body, some will only be missing the second X chromosome in some of their cells. Girls and women with this mixture of cells have *mosaic* Turner syndrome, and may have fewer health or developmental issues than those who have non-mosaic Turner syndrome.

Less commonly, Turner syndrome occurs when there is loss of only a portion of a second X chromosome that can occur from a variety of mechanisms.

### **What are the Health and Developmental Concerns Associated with Turner syndrome?**

During pregnancy, it may be diagnosed through genetic testing such as a [chorionic villus sampling \(CVS\)](#) or [amniocentesis](#) or suspected through a blood test such as [cell-free DNA \(cfDNA\)](#) analysis. Keep in mind that [cfDNA](#) analysis cannot tell for certain if a baby has a variation in their number of X and Y chromosomes. It is important to remember that the signs and symptoms of Turner syndrome can vary quite a bit from person to person. While [prenatal diagnostic testing](#) can provide a 'yes' or 'no' answer about whether a pregnancy has Turner syndrome, it can not predict the specific health concerns that a baby with Turner syndrome might have due to the variability of the condition.

While many pregnancies with Turner syndrome will not appear different in any way, while



some will show differences on prenatal [ultrasound](#). These signs can include large collections of fluid at the back of a baby's neck (an increased nuchal translucency or a cystic hygroma) or around the baby's body (hydrops) as well as heart defects. For reasons that are unclear, the majority of pregnancies with Turner syndrome result in miscarriage. The miscarriage can occur in any of the three trimesters, but most often occurs in the first trimester.

Despite the high rate of miscarriage, the outlook for girls with Turner syndrome at birth is quite good. During the newborn period, some girls with Turner syndrome will no obvious signs or health issues, while others may show puffy hands and feet (edema), a short thick neck (webbed neck), and/or heart problems. Short stature is the most common physical difference for girls with Turner syndrome, and is usually apparent by school age. Early loss of ovarian function is also very common, causing many individuals with Turner syndrome to need hormone treatments to transition through puberty. Many women with Turner syndrome experience infertility as adults, but a small number do retain their ovarian function a bit longer and can conceive a child during young adulthood.

Other differences in development that can commonly be seen in girls and women with Turner syndrome include a low hairline at the base of the neck, skeletal differences, kidney problems, and hearing and vision loss. Autoimmune issues such as thyroid disorders and diabetes may also be more common in women with Turner syndrome.

Intelligence is typically in the normal range but nonverbal learning disabilities, developmental delays, and differences in behavior can be a little more common in girls with Turner syndrome.

### **If I have a baby with Turner syndrome, what is the chance I will have another baby with Turner syndrome?**

In most cases, the chance that a woman will have another baby with an X and Y chromosome variation is thought to be less than 1%, as the majority of cases of X and Y chromosome variation are not inherited. However, some rare cases of Turner syndrome that involve the loss of a portion of a second X chromosome can potentially be passed from one generation to the next.

### **Treatment for Turner syndrome**

There is no "cure" for Turner syndrome. Many babies with Turner syndrome do not need any special treatment or intervention as infants. However, infants and children known to



have Turner syndrome should have medical and developmental evaluations that look specifically for health issues that are associated with Turner syndrome, which can allow for early detection and changes to the baby's medical management. These evaluations should include a heart evaluation and an ultrasound of the kidneys. Congenital (from birth) heart defects are identified in half of girls with Turner syndrome, and can be life-threatening.

School age girls with Turner syndrome should be seen by an endocrinologist (who specializes in hormones) to determine the optimal timing of hormone therapy to assist them through puberty and to reach a more typical final adult height. Regular hearing and vision screens as well as screens for diabetes, thyroid problems, and celiac disease are other common evaluations for girls with Turner syndrome. If any symptoms of learning disabilities or social/emotional difficulties arise during childhood, early intervention and support in school can be helpful.

Throughout adulthood individuals with Turner syndrome continue to receive hormone therapy, and should work with their providers to manage their overall health. This should include particular attention to blood pressure, heart, bone, and liver function.

Women with Turner syndrome have a decreased likelihood to conceive a pregnancy naturally. This chance also decreases quickly with age. Therefore, women with Turner syndrome who desire a family may not want to delay their family planning, or may wish to consider oocyte cryopreservation (freezing of their eggs) when possible. They may also wish to consider other options for motherhood such as egg donation, adoption, and gestational carriers (surrogates). Women with Turner syndrome that do carry a pregnancy have an increased risk for cardiovascular complications during the pregnancy.

### **How common is Turner syndrome?**

Turner syndrome (45,X) is estimated to occur in one of every 2500 female births. It is unknown exactly how many pregnancies that result in miscarriage are affected with Turner syndrome.

### **Outlook for individuals with Turner syndrome:**

Many girls and women with Turner syndrome will be shorter than average in height and most will experience infertility. About half will also have heart problems that can in some cases be life-threatening. Some may experience difficulty in learning in some areas. However, many individuals with Turner syndrome have success in school, including higher education, and work in a variety of career settings. The majority of women with Turner



syndrome will need coordinated medical care throughout their life, but can expect to live full and productive lives.

Click [here](#) to learn more about scheduling a genetic counseling appointment for pregnancy-related questions.

Click [here](#) to learn more about scheduling a genetic counseling appointment for infertility or preconception questions.

Click [here](#) to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.

## **OTHER RESOURCES**

[Turner Syndrome Foundation](#)

[Turner Syndrome International](#)

[Turner Syndrome Society of Canada](#)

[Turner Syndrome Society of the United States](#)

[Turner Syndrome Support Society \(UK\)](#)