Turner Syndrome

What is it?

Turner syndrome is a chromosomal condition that affects girls. Girls with Turner syndrome most likely will have short stature and lack ovaries. Many girls also have extra folds of skin on the neck, a low hairline at the back of the neck, swelling of the hands and feet, skeletal anomalies, or kidney problems. Some girls may have a heart defect called coarctation of aorta or aortic valve defects. If left untreated, these heart defects can be life-threatening. Few girls will have development delays, nonverbal learning disabilities, or behavioral problems.

How common is it?

About 1 in 2,500 girls born each year will have Turner syndrome. It is more common in pregnancies that result in miscarriage or stillbirth.

What causes it?

Turner syndrome is caused when only one normal X chromosome is present and the other is missing or there has been a change in the structure of it. Normally, girls have two X chromosomes and boys have one X and one Y chromosome.

How is it diagnosed?

Turner syndrome is confirmed by chromosomal analysis. A **chromosomal analysis** is a test that looks at and determines the number of chromosomes in a person. Turner syndrome can be diagnosed during pregnancy or after birth.

How is it treated?

Treatment will vary from person to person. Girls with Turner syndrome may need different therapies to grow and develop normally. Your child's doctor will discuss appropriate treatment options with you.

Indiana State Department of Health

BDPR
Indiana Birth Defects and Problems Registry

For more information:

U.S. National Library of Medicine, Genetics Home Reference
https://ghr.nlm.nih.gov/condition/turner-syndrome#

Mayo Clinic
https://www.mayoclinic.org/diseases-conditions/turner-syndrome/symptoms-causes/syc-20360782