TURNER SYNDROME

What is Turner syndrome?

Turner syndrome is caused by a missing or incomplete X chromosome. People who have Turner syndrome develop as females. The genes affected are involved in growth and sexual development, which is why girls with the disorder are shorter than normal and have abnormal sexual characteristics.

How do people get Turner syndrome?

Normally, females inherit one X chromosome from their mother and one X chromosome from their father. But females who have Turner syndrome are missing one of their X chromosomes.

Turner syndrome is typically caused by what is called nondisjunction. If a pair of sex chromosomes fails to separate during the formation of an egg (or sperm), this is referred to as nondisjunction. When an abnormal egg unites with a normal sperm to form an embryo, that embryo may end up missing one of the sex chromosomes (X rather than XX). As the embryo grows and the cells divide, every cell of the baby's body will be missing one of the X chromosomes.

The abnormality is not inherited from an affected parent (not passed down from parent to child) because women with Turner syndrome are usually sterile and cannot have children.

In about 20 percent of Turner syndrome cases, one X chromosome is abnormal. It may be shaped like a ring, or missing some genetic material.

About 30 percent of girls with the disorder are only missing the X chromosome in some of their cells. This mixed chromosome pattern is known as mosaicism. Girls with this pattern may have fewer symptoms because they still have some normal (XX) cells.

One of the missing genes on the X chromosome is the SHOX gene, which is responsible for long bone growth. The missing SHOX gene is the reason girls who have the disorder are unusually short. Other missing genes regulate ovarian development, which influences sexual characteristics.

What are the symptoms of Turner syndrome?

Turner syndrome affects growth and sexual development. Girls with this disorder are shorter than normal, and may fail to start puberty when they should. This is because the ovaries (which produce eggs, as well as the sex hormones estrogen and progesterone) fail to develop properly.

Women with Turner syndrome appear to have a stocky appearance, arms that turn out slightly at the elbow, a receding lower jaw, a short webbed neck, and low hairline at the back of the neck.

Other medical symptoms include: lymphedema (swelling of hands and feet), heart and/or kidney defects, high blood pressure, and infertility (inability to have children).
How do doctors diagnose Turner syndrome?

About half of the cases are diagnosed within the first few months of a girl's life by the characteristic physical symptoms (swelling of the hands and feet, or a heart defect). Other patients are diagnosed in adolescence because they fail to grow normally or go through puberty.

When the doctor suspects Turner syndrome, a blood sample can be used to make a karyotype (a chromosome analysis) and the diagnosis can be confirmed.

Turner syndrome may be diagnosed during pregnancy with a chorionic villus sampling (CVS) or amniocentesis. Alternatively, an ultrasound (a machine that uses sound waves to look inside a mother's uterus) can identify the disorder by its physical symptoms before the baby is born.

How is Turner syndrome treated?

Hormone replacement therapy is the best way to treat this disorder. Teenagers are treated with growth hormone to help them reach a normal height. They may also be given low doses of androgens (male hormones which females also produce in small quantities) to increase height and encourage normal hair and muscle growth. Some patients may take the female hormone estrogen to promote normal sexual development.

Interesting facts about Turner syndrome

Turner Syndrome affects 60,000 females in the United States. This disorder is seen in 1 of every 2000 to 2500 babies born, with about 800 new cases diagnosed each year.

In 75-80% of cases, the single X chromosome comes from the mother's egg; the father's sperm that fertilizes the egg is missing its sex chromosome.

Turner syndrome is named for Dr. Henry Turner, who in 1938 published a report describing the disorder.

The average height of an untreated woman with Turner syndrome is 4 feet 8 inches.

A female fetus (normally XX) can survive with only one X chromosome, but a male fetus (normally XY) could not survive with only one Y chromosome. This is because not having an X chromosome is much worse than not having a Y chromosome. The Y chromosome carries very few genes essential for life. In contrast, the X chromosome is a much longer DNA molecule and contains many, many genes that are needed for cells to function.