Turner syndrome is a condition that effects development in females. It occurs when all or part of an X chromosome is missing in all or most of the cells in a female's body.

Humans have 46 chromosomes. These contain all of a person's genes and DNA. Two of the chromosomes are the sex chromosomes, which determine a person's gender. Both of the sex chromosomes in females are X. This is written as XX. Males have an X and a Y chromosome, which is written as XY. The two sex chromosomes help a person develop fertility and the sexual characteristics of their gender.

In Turner syndrome, a girl does not have the usual pair of two complete X chromosomes. This is thought to be a random error that occurs during the formation of the sperm or egg.

- Most often, the girl has only one X chromosome in her cells.
- In other cases, the girl has some cells in her body with two chromosomes, but other cells have only one. This is called mosaicism.
- There can also be an addition of Y material.

Only about 1% of pregnancies in which the fetus has Turner syndrome results in live births. The others end in miscarriage or stillbirth.

### Symptoms

Females who have Turner syndrome are shorter than average. Girls often have normal height for the first three years of life, but then have a slow growth rate. They do not have the usual growth spurt at puberty.

Those with Turner syndrome usually have normal intelligence and good verbal and reading skills. Some may have problems with memory skills, math and fine finger movements.

Some physical features of Turner syndrome include:
- Wide neck with extra skin.
- A broad chest and widely spaced nipples.
- Puffiness or swelling (lymphedema) of the hands and feet.
- Heart murmur that is sometimes related to narrowing of the major vessel (aorta) leaving the heart.

Health concerns may include:
- Frequent middle ear infections especially in early childhood, which may sometimes lead to hearing loss.
- A tendency for high blood pressure.
- Thyroid problems – most often an underactive thyroid gland.
Because of a lack of developed ovaries, women with Turner Syndrome are usually unable to conceive a child. The vagina and womb are normal so they may carry a pregnancy conceived using donor eggs.

**Diagnosis**

Turner syndrome may be diagnosed during pregnancy, at birth or later.

**During pregnancy** – It may be suspected during an ultrasound exam and can be confirmed by prenatal testing. This can be through an amniocentesis or chorionic villous sampling. Cells are obtained for chromosomal analysis.

**At birth** – Turner syndrome may be suspected at birth when physical features are observed. It is confirmed with a blood test.

**Later** – It may be diagnosed in early childhood when a slow growth rate and other features are identified. Sometimes it is diagnosed later when puberty does not occur.

**Treatment**

Babies born with a heart murmur or narrowing of the aorta may need surgery to correct the problem.

Girls and adolescents may be under the care of a pediatric endocrinologist who is a specialist in childhood conditions of the hormones and metabolism.

Some girls with Turner syndrome may benefit from growth hormone injections.

Estrogen replacement therapy is usually begun at the time of normal puberty at around age 12. This is given to start breast development. Later estrogen and progesterone are given to begin monthly periods. This is needed to keep the womb healthy.

Regular health check-ups are important to watch for and treat some of the conditions that may occur with Turner syndrome.

With appropriate medical care and support, women with Turner syndrome can lead normal lives.

**Research**

Many research studies are conducted on health conditions and treatments related to Turner syndrome.

Clinicaltrials.gov is a service of the U.S. National Institutes of Health. The website provides information on current research studies and a database of research results.

**Information and Support**

These organizations offer more information on Turner Syndrome.

The Eunice Kennedy Shriver National Institute of Child Health and Human Development - nichd.nih.gov

Turner Syndrome Society of the U.S. 800-365-9944, www.turnersociety.org,

U.S. Centers for Disease Control and Prevention - 800-232-4636 www.cdc.gov/ncbddd/birthdefects