Turner Syndrome

Turner syndrome is a medical condition that affects approximately one in every 3,000 females. Dr. Henry Turner, an endocrinologist, first described the condition in 1938 when he observed a set of common physical features in some of his female patients, but it wasn't until 1960 that a chromosomal abnormality associated with the condition was actually described.

What Is Turner Syndrome?

"Turner syndrome is [the result of] a chromosomal abnormality in which a female infant is born with only one X chromosome, instead of the usual two [or is missing part of one X chromosome]," explains Kevin R. Johnson, MD, a pediatrician in Gainesville, Georgia. In most cases, untreated females with this disorder are typically short in stature (average final adult height is 4 feet and 7 inches) and may have a variety of associated physical features and medical problems.

Because females with Turner syndrome (TS) do not have proper ovarian development, they usually fail to develop secondary sexual characteristics during adolescence and are infertile as adults. However, advances in medical technology, including hormonal therapy and in vitro fertilization, can help women with this condition.

Other complications of TS may include kidney and heart abnormalities, high blood pressure, obesity, diabetes mellitus, cataracts, thyroid problems, and arthritis.

Girls with TS usually have normal intelligence, but some may experience learning difficulties, particularly in mathematics. Many also have a problem with tasks requiring spatial skills, such as map reading, or visual organization.

Although they are not at increased risk for psychological problems, some girls do have problems with body image or self-esteem. Children with this disorder may also be hyperactive.

How Is Turner Syndrome Diagnosed?

Upon physical examination, your child's doctor may note certain features, such as a lack of breast development, a webbed neck, short stature, and abnormal development of the arms.

"A special blood test called a **karotype** is used to identify patients with this problem," Dr. Johnson says. Several physical characteristics may be noted at birth, which can alert your child's doctor to the possibility of TS and cause him or her to order the karotype. A karotype test that indicates TS reveals 45 chromosomes (only one X chromosome), instead of the normal 44 autosomes and 2 X chromosomes (XX) typically found in females. In some girls with TS, two X chromosomes may be present, but one is misshapen or missing a piece.

The identifying physical features of TS include:

- short stature
- webbed neck
- low hairline at the back of the head
- abnormal eye features (drooping of eyelids)
- abnormal bone development
- lack of breast development at the expected age (usually by age 13)
- absence of menstruation (amenorrhea)

People who have TS may vary widely in their characteristics or symptoms of the disorder. Some may have many features or symptoms, whereas others experience only a few. With appropriate medical care, early intervention, and ongoing support, a person with TS can lead a normal, healthy, and productive life.

Treating Turner Syndrome

Because TS is a chromosomal disorder, there is no cure for the condition. However, there are a number of treatments that can help:

- Growth hormone, either alone or with other hormone treatment, may improve growth and will usually increase final adult height. The U.S. Food and Drug Administration has approved growth hormone for the treatment of TS, and many insurance plans now cover this special treatment.
- Estrogen replacement therapy is often initiated when the child reaches 12 or 13 years of age to stimulate the development of secondary sexual characteristics (breast development and menstrual periods). This therapy will not reverse infertility.
- Cardiac surgery may be necessary to correct specific heart defects.
- Recently developed reproductive technologies can help women with TS become pregnant. Fertilized donor eggs can be used to create embryos, which can be inserted into the uterus of women with TS. With the help of hormone treatment, these women carry developing fetuses to term.