Turner Syndrome

Amid Habib, M.D., F.A.A.P.
Pediatric Endocrinology
Diplomat, American Board of
Pediatrics and
Pediatric Endocrinology

Altamonte Springs, Florida

A Member of
The MAGIC Foundation's
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Turner Syndrome

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6645 W. North Avenue
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Described by Dr. Henry Turner in 1938, and manifested with short stature, webbed neck, cubitus vulgus (arms turned out slightly at elbow), and sexual infantilism. Grumback used the term "Gonadal Dysgenesis" (abnormal development of the ovaries) to describe the syndrome. Many girls may have distinctive characteristics, while some girls show few.

Turner Syndrome (TS) occurs in approximately 1 in 2500 live female births. Approximately 98% of pregnancies with TS abort spontaneously. Approximately 10% of fetuses from pregnancies that have spontaneously aborted have TS.

The syndrome represents a wide spectrum of clinical presentation. The most common of which is the classic TS with 45XO (one X chromosome is missing completely). Less common, the mosaic TS with 45X/46XX,45X/46XY. Isochromosome happens when one chromosome is partially missing.

Short stature is almost a consistent finding in TS. Most children with TS show gradual decline in growth rate between 3 and 11 years of age, drifting considerably below the lower normal percentiles for age. Without treatment, the ultimate height range is between 4'7" to 4'10". Family height may play a role in determining the ultimate height in girls with TS.

Children with TS may have the following physical findings: congenital lymphedema (puffy hands and feet at birth), low posterior hairline, webbed neck, prominent ears, high arched palate, micrognathia (small jaw), broad chest, hypoplastic nipples, cubitus valgus (arms turned out slightly at the elbow), multiple pigmented nevus (moles), abnormal fingernails (turned up at the end), intestinal telangiectasia (malformation of the intestinal blood vessels). Cardiovascular anomalies are common and the most clinically frequent coarctation (narrowing) of the aorta (main artery coming out of the heart), echocardiographic studies, however, show non stenotic bicuspid aorta (malformed heart valve) might be the most common cardiovascular lesion in TS.

Kidney anomalies occur in 1/3 to 1/2 of girls with TS, with monosomic (one chromosome is missing). The most common anomaly is a horseshoe kidney. There is an increased frequency of chronic lymphocytic thyroiditis (under active thyroid) and diabetes mellitus. Patients with TS are prone to keloid (excessive growth of scar tissue).

The prevalence of mental retardation appears to be no greater than that of the general population. However, many patients have a specific deficit in spatial ability and frequently exhibit gross and fine motor dysfunction. The bone age is delayed. Osteoporosis may also be seen.

Normal pubertal development, growth spurts and spontaneous periods do not occur in the majority of children with TS. Mosaic forms are seen in female adolescents with primary amenorrhea (failure to start menstrual cycles) and in young women with premature ovarian failure. It is estimated, however, that 3 - 8% of 45XO karyotype patients and 12 - 21% of females with sex chromosomes mosaicism may have normal pubertal development and spontaneous menstrual cycles. Pregnancies have occurred in patients with TS.

Gonadal dysgenesis should be entertained in all short girls, girls with unexpected primary or secondary amenorrhea (absence of menstrual cycles) and girls with lymphedema. Chromosome studies are indicated in the work-up of suspected patients. Heart, kidney and hearing evaluation is indicated if the diagnosis of TS is confirmed.

Different modalities of therapy are available, including low dose estrogen therapy, anabolic steroids, growth hormone alone or in combination with the above. The response to growth hormone therapy varies from patient to patient. The most gains are seen when growth hormone is started early (before 9 years of age) and estrogen therapy is started late (after 14 years of age). Patients receiving growth hormone must be monitored closely for increased intracranial pressure, hypothyroidism, glucose intolerance, edema, and any changes in the nevi. Potential benefit and risk associated with growth hormone treatment must be considered.

Most patients with TS will require substitution of female hormone therapy for development of secondary sexual characteristics and menstruation. The time of initiation of therapy varies with each patient. Some recommend the estrogen therapy begin when the patient expresses concern about the onset of puberty.

For further information concerning TS please contact your Pediatric Endocrinologist