

## Turner's Syndrome

Described by Dr. Henry Turner in 1938 as manifested with short stature, webbed neck, cubitus Valgus and sexual infantilism. Grumbach used the term "gonadal dysgenesis" to describe the syndrome. Many girls may have distinctive characteristics, while some girls may show few.

Turner's Syndrome occurs in 1 in 3,000 live female births. Approximately 98% of pregnancies with Turner's Syndrome abort spontaneously and approximately 10% of fetuses from pregnancies that have spontaneously aborted have Turner's Syndrome.

The syndrome represents a wide spectrum of clinical presentation, the most common of which is the classic Turner's Syndrome with 45 XO karyotype, less common the Mosaic Turner's Syndrome with Mosaic sex chromosome Karyotypes 45, X/46, XX, 45, X/46, XY.

Short stature is almost a consistent finding in Turner's Syndrome, the cause of which is multifactorial, including intrauterine growth retardation, gradual decline in height velocity in childhood, absence of pubertal growth spurt and to end organ resistance resulting from skeletal dysplasia. Patients with Turner's Syndrome may have abnormal body proportions characterized by markedly shortened lower extremities. The ultimate height range is between 55 to 58 inches. Familial height may play a role in determining the ultimate height in girls with Turner's Syndrome

Children with Turner's Syndrome may have the following physical findings; congenital lymphedema, low posterior hair line, webbed neck, prominent ears, high arched palate, micrognathia, broad chest, cubitus valgus, multiple pigmented nevus, abnormal finger nails, intestinal telangiectasia and hypoplastic nipples. Cardiovascular anomalies are common and the most clinically frequent is coarctation of the aorta. Echocardiographic studies however, showed non stenotic bicuspid aorta valve might be the most common cardiovascular lesion in Turner's Syndrome

Renal anomalies occur in 1/3 to 1/2 of girls with Turner's Syndrome with monosomic patients at great risk. The most common anomaly is a horse shoe kidney. There is an increased frequency for chronic lymphocytic thyroiditis and diabetes mellitus or carbohydrate intolerance. Patients with Turner's Syndrome are prone to keloid formation. The prevalence of mental retardation appears to be no greater than that in general population. However, many patients have a specific deficit in special ability and frequently exhibit gross and fine motor dysfunction. The bone age is retarded along with shortening of the fourth metacarpal bone. Osteoporosis may also be seen.

Normal pubertal development and spontaneous menstrual periods do not occur in the majority of children with Turner's Syndrome. Mosaic forms of gonadal dysgenesis are seen in female adolescents with primary amenorrhea and in young women with premature ovarian failure. It is estimated however, that 3 to 8% of 45X Karyotype patients and 12 to 21% of females with sex chromosome mosaicism may have normal

pubertal development and spontaneous menstrual periods. Pregnancies have occurred in patients with 45X and 45, X/46XX Karyotypes.

Gonadal dysgenesis should be entertained in all short girls, girls with unexpected primary or secondary amenorrhea and girls with lymphedema. Chromosome Karyotyping and serum gonadotropin determination are indicated in the workup of suspected patients. Cardiac and renal evaluation is indicated if the diagnosis of Turner's Syndrome is confirmed.

Different modalities of therapy are available, including low dose estrogen therapy, anabolic steroids, growth hormone alone or in combination with androgens or estrogens.

Most patients with Turner's Syndrome 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 and it is recommended that therapy begin when the patient expresses concern about the onset of puberty. Various estrogenic and progestational agents and schedules have been used. The response to growth hormone therapy varies from patient to patient. A suitable course of therapy consists of oral estrogen for 6 to 12 months or until menstruation occurs, therefore, the estrogen can be given on the 1st 25 days of each calendar month. Withdrawal bleeding usually occurs 3 to 5 days after the estrogen therapy is stopped. Oral progestational agents should be given from the 15th to 25th day of each month to improve breast development, cause coiling of the glands of the endometrium and increase the vascularity of the stroma. All patients on long term exogenous female hormone therapy require periodic gynecological examinations, as patients with Turner's Syndrome have an increased risk of developing neoplasms arising from the rudimentary streak gonads, including gonadoblastoma and dysgerminoma.

*For further information concerning Turner's Syndrome, please contact your endocrinologist.*

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The MAGIC Foundation is a national nonprofit organization created to provide support services for the families of children afflicted with a wide variety of chronic and/or critical disorders, syndromes and diseases that affect a child's growth. Some of the diagnoses are quite common while others are very rare.

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