Turners Syndrome

BIRTH DEFECT RESEARCH FOR CHILDREN

What is Turner's Syndrome?

Turner's Syndrome is a genetic condition that affects females only. It occurs when one of the two X chromosomes (one of the two sex chromosomes) normally found in females is incomplete or missing. The syndrome is named after Dr. Henry Turner who first described it in 1938. The most common characteristics of Turner's Syndrome include short stature, underdeveloped ovaries, and failure to mature sexually. It can also include webbed neck, skeletal problems, low posterior hairline, heart defects, kidney abnormalities, and other problems. Not all these characteristics need to be present for a diagnosis of Turner's Syndrome. There can be great variability in the severity of the abnormalities depending on how much genetic material is missing.

Turner's Syndrome

How many children have Turner's Syndrome?

Turner's Syndrome occurs in approximately one out of every 2,000-5,000 live births. In the United States, approximately 60,000 girls and women are affected and about 800 new cases appear each year. There is no reported difference in the frequency of the condition based on race or socio-economic factors.

How do you know if your child has Turner's Syndrome?

If a newborn girl has edema (swelling) of the hands and feet, excessive skin on the neck, and/or small stature, she should be tested for Turner's Syndrome. Testing may also be needed if other characteristics typical of Turner's Syndrome appear in childhood or adolescence. Turner's Syndrome is diagnosed through a blood test called a karyotype.

What causes Turner's Syndrome?

Turner's Syndrome is caused by the complete or partial absence of one of the two X chromosomes. While it is not known what causes this, it is believed to be due to abnormalities in cell division. Turner's Syndrome appears to be a random event that has not been associated with any environmental factors.

How can you help a child who has Turner's Syndrome?

Although there is no cure for Turner's Syndrome, a child can receive treatment to minimize the symptoms. Available treatment includes human growth hormone injections to maximize growth potential and estrogen replacement therapy to promote development of sexual characteristics. Additionally, surgery can be performed to correct some of the other abnormalities associated with Turner's Syndrome. Counseling may be needed to help the child face the problems and emotions associated with growing up with Turner's Syndrome. A doctor specializing in endocrinology can manage many of the most common health issues.

What's in the future for a child who has Turner's Syndrome?

Some girls with Turner's Syndrome may have differences in learning styles that make spatial learning more difficult. Thyroid problems have also been reported. Girls with Turner's Syndrome without the complications of serious cardiovascular or kidney abnormalities can be expected to have a normal life span. Modern reproductive technologies are available to help women with Turner's Syndrome become pregnant. Turner's Syndrome requires medical follow-up throughout the life.

Fact Sheet by:

Birth Defect Research Children, Inc.
www.birthdefects.org