

Fact Sheet

Spotlight on Turner Syndrome

Information Compiled By Tanni Anthony

What is Turner Syndrome?

Turner syndrome has been recognized since the 1930s when Otto Ullrich and Henry Turner described females with the association of short stature and other physical features. It was not until 1959 that the chromosomal nature of the condition was described.

Turner syndrome affects approximately one out of every 2,500 female live-births worldwide. It encompasses a spectrum of features, from major heart defects to minor cosmetic issues. Some individuals with Turner syndrome may have only a few features, while others may have many. Almost all people with Turner syndrome have short stature and loss of ovarian function, but the severity of these problems varies considerably amongst individuals.

Features may include:

General Appearance: Some individuals with Turner syndrome have a short neck with a webbed appearance, a low hairline at the back of the neck, and low-set ears. Hands and feet of may be swollen or puffy at birth and often have soft nails that turn upward at the ends when the individual is older. All these features appear to be due to obstruction of the lymphatic system during fetal development. Another characteristic cosmetic feature is the presence of multiple pigmented nevi, which are colored spots on the skin, or moles.

Short Stature: Almost all individuals with Turner syndrome have short stature. This is partially due to the loss of one copy of the SHOX gene on the X-chromosome. This particular gene is important for long bone growth. The loss of SHOX may also explain some of the skeletal features found in Turner syndrome, such as short fingers and toes, and irregular rotations of the wrist and elbow joints. Linear growth is attenuated in utero, and statural growth lags during childhood and adolescence, resulting in adult heights of 143-145 cm (approximately 4 feet 8 inches). Final adult height in Turner syndrome can be increased by a several inches if growth hormone (GH) treatment is given relatively early in childhood.

Puberty/Reproduction: Unknown genes on the X-chromosome regulate the development and functions of the ovary. Most individuals experience loss of ovarian function early in childhood, and do not enter puberty at the normal age.

Some teenagers may undergo some breast development and begin menstruating, but cease further development and menses during the later teen years. A few women with Turner syndrome have apparently normal ovarian function with regular menses until the mid-20s before ovarian failure occurs. A few spontaneous pregnancies have been reported.

Cardiovascular: From 5-10% of children with this condition are found to have a severe constriction of the major blood vessel coming out from the heart, a condition known as "coarctation of the aorta." This can be surgically corrected as soon as it is diagnosed.

Other major defects in the heart and its major vessels are reported to a much lesser degree. Approximately 30% of individuals with Turner syndrome are reported to have "bicuspid aortic valves," meaning that the major blood vessel from the heart has only two rather than three components to the valve regulating blood flow. Comprehensive screening and evaluation by a cardiologist with expertise in congenital heart conditions is essential for individuals with Turner syndrome at the time of diagnosis. The evaluation should include cardiac magnetic resonance as well as echocardiography and ECG.

Many women with Turner syndrome have high blood pressure, which may be apparent even in childhood. In some cases this high blood pressure may be due to aortic constriction, or to kidney problems. In a majority of women, however, no specific cause for the high blood pressure has been found.

Kidney: Kidney malformations affect approximately 1/3 of individuals with Turner syndrome. While the kidneys usually function normally, there may be a tendency towards urinary infections.

Osteoporosis: There is a high incidence of osteoporosis (thin or weak bones) in adult women.. Osteoporosis leads to loss of height, curvature of the spine and increased bone fractures. The primary cause of osteoporosis in individuals with Turner syndrome appears to be inadequate circulating estrogen in the body. Women who have low levels of estrogen due to ovarian failure can take estrogen treatments, which will help prevent osteoporosis.

Diabetes: Type II diabetes, also known as adult onset diabetes, has a high occurrence rate in individuals with Turner syndrome. There is twice the risk of the general population for developing this disease. The reason for the high risk of diabetes amongst individuals with Turner syndrome is unknown, but there appears to be insufficient insulin release in response to glucose. Diabetes type II can be controlled through careful monitoring of blood-sugar levels, diet, exercise, regular doctor visits and sometimes medication.

Thyroid: Approximately one third of individuals with Turner syndrome have a thyroid disorder, usually hypothyroidism. Symptoms of this condition include decreased energy, dry skin, cold-intolerance and poor growth. It is often caused by an immune system attack on the thyroid gland (also known as Hashimoto's thyroiditis). The condition can be treated with thyroid hormone supplements.

Vision and Hearing Issues: Refractive error (nearsightedness, farsightedness, and/or astigmatism) is very common (around 40%). There is high rate of amblyopia (almost 30%) and strabismus or poor eye alignment (33%). There is a danger that parents and community doctors may concentrate exclusively on other medical features of Turner's syndrome, resulting in delayed recognition of strabismus and developing amblyopia. Other potential, though less frequent, vision concerns with Turner syndrome include color vision challenges, X-linked congenital stationary night blindness, glaucoma and abnormal irides. The high prevalence for some type of visual conditions would argue for early systematic screening of children with Turner's syndrome. Early and regular eye exams are important to ensure optimal visual performance. If a visual impairment is present that affects school performance, the child will require the services of a certified teacher of students with visual impairments.

Hearing loss is common among girls or women with Turner syndrome. This may be due to the gradual loss of nerve function. Also, slight abnormalities in the shape of the skull increase the risk of frequent middle ear infections and hearing loss related to these infections. Otitis media (ear infection) is extremely common in girls particularly in infancy and early childhood. Changes in the structure of the middle ear and eustachian tube (tube connecting the middle ear with the back of the throat) probably account for poor drainage and ventilation in the middle ear and easier movement of germs from the throat into the ear - all of which play a role in causing frequent middle ear infections and effusions (fluid in the middle ear), especially in early childhood. Individuals may develop early sensorineural (nerve) hearing loss. This hearing loss may begin in childhood and appears to worsen over time, so that more than 90% of women with Turner syndrome in their forties have some degree of hearing loss, with more than 25% of them needing hearing aids. It is recommended that individuals with Turner syndrome should have their hearing and middle ear function checked regularly throughout life. If hearing loss is present that affects school performance, the child will require the services of a certified teacher of students who are deaf/hard of hearing.

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Cognitive Function/Educational Issues: In general, individuals with Turner syndrome have normal intelligence. However, some girls and women with Turner syndrome may have difficulty with specific visual-spatial coordination tasks (e.g. mentally rotating objects in space) and learning math (geometry, arithmetic). This very specific learning problem has been termed the "Turner neurocognitive phenotype" and appears due to loss of X-chromosome genes important for selected aspects of nervous system development. Verbal skills are usually normal or superior.

Education and Medical Supports

Individuals with the diagnosis of Turner syndrome will often be eligible for the services of a state project serving students who are deafblind. Education specialists trained in sensory disabilities should be members of the student's Individualized Education Program team, if vision

and/or hearing loss are diagnosed. Medical specialists will play an important role in ensuring that the individual with Turner syndrome has appropriate treatments and clinical care.

References:

Information retrieved from: <http://turners.nichd.nih.gov/publications.html>

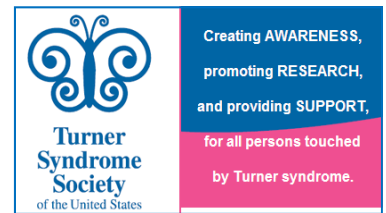
Information retrieved from: <http://www.bing.com/health/article/mayo-126690/Turner-syndrome?q=turner+syndrome>

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Parkin, M. W. P. (2009). *Hearing loss in Turner syndrome*. *International Journal of Pediatric Otorhinolaryngology*, 73 (2):243-7. Epub 2008 Dec 9.

Resources

Turner Syndrome Society of the United States: The Turner Syndrome Society of the United States (TSSUS) is a national non-profit 501 (c) (3) organization that provides health-related resources to patients, families and physicians for the diagnosis and treatment of Turner syndrome. There are chapters and support groups located throughout the country. TSSUS holds an annual conference with speakers. For more information, go to: www.turnersyndrome.org



For more information about the CO Services for Children and Youth with Combined Vision and Hearing Loss Project contact:

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Fact Sheets from the Colorado Services to Children and Youth with Combined Vision and Hearing Loss Project are to be used by both families and professionals serving individuals with vision and hearing loss. The information applies to children, birth through 21 years of age. The purpose of the Fact Sheet is to give general information on a specific topic. More specific information for an individual student can be provided through personalized technical assistance available from the project. For more information call (303) 866-6681 or (303) 866-6605. Updated: 2/11