Learning About Turner Syndrome

What is Turner syndrome?

Turner syndrome is a chromosomal condition that alters development in females. Women with this condition tend to be shorter than average and are usually unable to conceive a child (infertile) because of an absence of ovarian function. Other features of this condition that can vary among women who have Turner syndrome include: extra skin on the neck (webbed neck), puffiness or swelling (lymphedema) of the hands and feet, skeletal abnormalities, heart defects and kidney problems.

This condition occurs in about 1 in 2,500 female births worldwide, but is much more common among pregnancies that do not survive to term (miscarriages and stillbirths).

Turner syndrome is a chromosomal condition related to the X chromosome. [ghr.nlm.nih.gov]

Researchers have not yet determined which genes on the X chromosome are responsible for most signs and symptoms of Turner syndrome. They have, however, identified one gene called SHOX that is important for bone development and growth. Missing one copy of this gene likely causes short stature and skeletal abnormalities in women with Turner syndrome.

What are the symptoms of Turner syndrome?

Girls who have Turner syndrome are shorter than average. They often have normal height for the first three years of life, but then have a slow growth rate. At puberty they do not have the usual growth spurt.

Non-functioning ovaries are another symptom of Turner syndrome. Normally a girl's ovaries begin to produce sex hormones (estrogen and progesterone) at puberty. This does not happen in most girls who have Turner syndrome. They do not start their periods or develop breasts without hormone treatment at the age of puberty.

Even though many women who have Turner have non-functioning ovaries and are infertile, their vagina and womb are totally normal.

In early childhood, girls who have Turner syndrome may have frequent middle ear infections. Recurrent infections can lead to hearing loss in some cases.

Girls with Turner Syndrome are usually of normal intelligence with good verbal skills and reading skills. Some girls, however, have problems with math, memory skills and fine-finger movements.

Additional symptoms of Turner syndrome include the following:

- An especially wide neck (webbed neck) and a low or indistinct hairline.
- A broad chest and widely spaced nipples.
- Arms that turn out slightly at the elbow.
- A heart murmur, sometimes associated with narrowing of the aorta (blood vessel exiting the heart).
- A tendency to develop high blood pressure (so this should be checked regularly).
- Minor eye problems that are corrected by glasses.
- Scoliosis (deformity of the spine) occurs in 10 percent of adolescent girls who have Turner syndrome.
- The thyroid gland becomes under-active in about 10 percent of women who have Turner syndrome. Regular blood tests are necessary to detect it early and if necessary treat with thyroid replacement
- Older or over-weight women with Turner syndrome are slightly more at risk of developing diabetes.
- Osteoporosis can develop because of a lack of estrogen, but this can largely be prevented by taking hormone replacement therapy.

How is Turner syndrome diagnosed?

A diagnosis of Turner syndrome may be suspected when there are a number of typical physical features observed such as webbed neck, a broad chest and widely spaced nipples. Sometimes diagnosis is made at birth because of heart problems, an unusually wide neck or swelling of the hands and feet.

The two main clinical features of Turner syndrome are short stature and the lack of the development of the ovaries.

Many girls are diagnosed in early childhood when a slow growth rate and other features are identified. Diagnosis sometimes takes place later when puberty does not occur.

Turner syndrome may be suspected in pregnancy during an ultrasound test. This can be confirmed by prenatal testing - chorionic villous sampling or amniocentesis - to obtain cells from the unborn baby for chromosomal analysis. If a diagnosis is confirmed prenatally, the baby may be under the care of a specialist pediatrician immediately after birth.

Diagnosis is confirmed by a blood test, called a karyotype. This is used to analyze the chromosomal composition of the female. More information about this will be discussed in the section "Is Turner syndrome inherited?"

What is the treatment for Turner syndrome?

During childhood and adolescence, girls may be under the care of a pediatric endocrinologist, who is a specialist in childhood conditions of the hormones and metabolism.

Growth hormone injections are beneficial in some individuals with Turner syndrome. Injections often begin in early childhood and may increase final adult height by a few inches.

Estrogen replacement therapy is usually started at the time of normal puberty, around 12 years to start breast development. Estrogen and progesterone are given a little later to begin a monthly 'period,' which is necessary to keep the womb healthy. Estrogen is also given to prevent osteoporosis.

Babies born with a heart murmur or narrowing of the aorta may need surgery to correct the problem. A heart expert (cardiologist) will assess and follow up any treatment necessary.

Girls who have Turner syndrome are more likely to get middle ear infections. Repeated infections may lead to hearing loss and should be evaluated by the pediatrician. An ear, nose and throat specialist (ENT) may be involved in caring for this health issue.

High blood pressure is quite common in women who have Turner syndrome. In some cases, the elevated blood pressure is due to narrowing of the aorta or a kidney abnormality. However, most of the time, no specific cause for the elevation is identified. Blood pressure should be checked routinely and, if necessary, treated with medication. Women who have Turner syndrome have a slightly higher risk of having an under active thyroid or developing diabetes. This should also be monitored during routine health maintenance visits and treated if necessary.

Regular health checks are very important. Special clinics for the care of girls and women who have Turner syndrome are available in some areas, with access to a variety of specialists. Early preventive care and treatment is very important.

Almost all women are infertile, but pregnancy with donor embryos may be possible.

Having appropriate medical treatment and support allows a woman with Turner syndrome to lead a normal, healthy and happy life.

Is Turner syndrome inherited?

Turner syndrome is not usually inherited in families. Turner syndrome occurs when one of the two X chromosomes normally found in women is missing or incomplete. Although the exact cause of Turner syndrome is not known, it appears to occur as a result of a random error during the formation of either the eggs or sperm.

Humans have 46 chromosomes, which contain all of a person's genes and DNA. Two of these chromosomes, the sex chromosomes, determine a person's gender. Both of the sex chromosomes in females are called X chromosomes. (This is written as XX.) Males have an X and a Y chromosome (written as XY). The two sex chromosomes help a person develop fertility and the sexual characteristics of their gender.

In Turner syndrome, the girl does not have the usual pair of two complete X chromosomes. The most common scenario is that the girl has only one X chromosome in her cells. Some girls with Turner syndrome do have two X chromosomes, but one of the X chromosomes is incomplete. In another scenario, the girl has some cells in her body with two X chromosomes, but other cells have only one. This is called mosaicism.

NHGRI Clinical Research on Turner Syndrome

NHGRI is not currently conducting clinical research on Turner syndrome.

- *ClinicalTrials.gov* provides information on clinical research studies. Currently, there are clinical trials at other institutes and organizations who are enrolling individuals with Turner syndrome. They include:
    - Long-Term Growth and Skeletal Effects of Early Growth Hormone Treatment in Turner Syndrome [clinicaltrials.gov]

- **Current NHGRI Clinical Studies**
- **Search ClinicalTrials.gov** [clinicaltrials.gov]
- **Clinical Research FAQ**

Additional Resources for Turner Syndrome

- **Turner Syndrome** [nichd.nih.gov]
  From the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development

- **Turner Syndrome** [turners.nichd.nih.gov]
  More from the *Eunice Kennedy Shriver* National Institute of Child Health and Development.

- **Turner Syndrome** [ghr.nlm.nih.gov]
  From Genetics Home Reference

- **Turner's Syndrome** [nlm.nih.gov]
  From Medline Plus

- **Turner Syndrome** [nlm.nih.gov]
  More from Medline Plus

- **Turner Syndrome Society of the United States** [turnersyndrome.org]
  Encourages medical research, the dissemination of state-of-the-art TS information, and social support services to individuals, families, physicians and the general public.

- **The Magic Foundation: Turner Syndrome** [magicfoundation.org]
  A national non-profit organization created to provide support services for the families of children afflicted with a wide variety of disorders, syndromes or diseases that affect a child's growth.

- **Turner Syndrome** [rarediseases.org]
  From the National Organization for Rare Disorders

- ***Health Supervision for Children With Turner Syndrome*** [pediatrics.aappublications.org]
  An article from *Pediatrics*, the official journal of the American Academy of Pediatrics

- **Turner Syndrome** [emedicine.medscape.com]
  From eMedicine

- **Turner Syndrome** [rarediseases.info.nih.gov]
  Information from the Genetics and Rare Diseases Information Center.

- **Finding Reliable Health Information Online**
  A listing of information and links for finding comprehensive genetics health information online.

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