MAYO CLINIC

Diseases and Conditions

Turner syndrome

By Mayo Clinic Staff

Turner syndrome, a condition that affects only girls and women, results when a sex chromosome (the X chromosome) is missing or partially missing. Turner syndrome can cause a variety of medical and developmental problems, including short height, failure to start puberty, infertility, heart defects, certain learning disabilities and social adjustment problems.

Turner syndrome may be diagnosed before birth (prenatal), during infancy or in early childhood. Occasionally the diagnosis is delayed until the teen or young adult years in those who have mild signs and symptoms of Turner syndrome.

Nearly all girls and women with Turner syndrome need ongoing medical care from a variety of specialists. Regular checkups and appropriate care can help most girls and women lead relatively healthy, independent lives.

Signs and symptoms of Turner syndrome may vary significantly.

Before birth

Prenatal ultrasound of a baby with Turner syndrome may show:

- Large fluid collection on the back of the neck or other abnormal fluid collections
- Heart abnormalities
- Abnormal kidneys

At birth or during infancy

In some girls, a number of physical features and poor growth are apparent early. Signs of Turner syndrome at birth or during infancy may include:

- Wide or weblike neck
- Receding or small lower jaw
- High, narrow roof of the mouth (palate)

- Low-set ears
- Low hairline at the back of the head
- Broad chest with widely spaced nipples
- Short fingers and toes
- Arms that turn outward at the elbows
- Fingernails and toenails that are narrow and turned upward
- Swelling of the hands and feet, especially at birth
- Slightly smaller than average height at birth
- Delayed growth

In older girls, teens and young women

For some girls, the presence of Turner syndrome may not be readily apparent. Signs and symptoms in older girls, teenagers and young women that may indicate Turner syndrome include:

- No growth spurts at expected times in childhood
- Short stature, with an adult height of about 8 inches (20 centimeters) less than might be expected for a female member of her family
- Learning disabilities, particularly with learning that involves spatial concepts or math, though intelligence is usually normal
- Difficulty in social situations, such as problems understanding other people's emotions or reactions
- Failure to begin sexual changes expected during puberty — due to ovarian failure that may have occurred by birth or gradually during childhood, adolescence or young adulthood
- Sexual development that "stalls" during teenage years
- Early end to menstrual cycles not due to pregnancy
- For most women with Turner syndrome, inability to conceive a child without fertility treatment

When to see a doctor

Sometimes it's difficult to distinguish the signs and symptoms of Turner syndrome from other disorders. So it's important to get a prompt, accurate diagnosis and appropriate care. See your doctor if you believe your daughter shows signs of Turner syndrome or if you have concerns about her physical, sexual or behavioral development.

Most people are born with two sex chromosomes. A boy inherits the X chromosome from his mother and the Y chromosome from his father. A girl inherits one X chromosome from each parent. If a girl has Turner syndrome, one copy of the X chromosome is missing or significantly changed.

The genetic alterations of Turner syndrome may be one of the following:

- **Monosomy.** The complete absence of an X chromosome generally occurs because of an error in the father's sperm or in the mother's egg. This results in every cell in the body having only one X chromosome.
- **Mosaicism.** In some cases, an error occurs in cell division during early stages of fetal development. This results in some cells in the body having two complete copies of the X chromosome. Other cells have only one copy of the X chromosome, or they have one complete and one altered copy.
- **Y chromosome material.** In a small percentage of Turner syndrome cases, some cells have one copy of the X chromosome and other cells have one copy of the X chromosome and some Y chromosome material. These individuals develop biologically as girls, but the presence of Y chromosome material increases the risk of developing a type of cancer called gonadoblastoma.

Effect of the chromosomal errors

The missing or altered X chromosome of Turner syndrome causes errors during fetal development and other developmental problems after birth — short stature, ovarian failure and learning disabilities. Physical characteristics and health complications that arise from the chromosomal error vary greatly.

The loss or alteration of the X chromosome occurs randomly. Sometimes, it's because of a problem with the sperm or the egg, and other times, the loss or alteration of the X chromosome happens early in fetal development.

Family history doesn't seem to be a risk factor, so it's unlikely that parents of one child with Turner syndrome will have another child with the disorder.

Turner syndrome can affect the proper development of several body systems. A number of complications may occur, including:

- **Heart problems.** Many girls and women with Turner syndrome were born with heart defects or even slight abnormalities in heart structure that increase their risk of serious complications. Defects in the main blood vessel leading out of the heart (aorta) increase the risk of a tear in the inner layer of the aorta (aortic dissection). A defect in the valve between the heart and the aorta may also increase the risk of a narrowing of the valve (aortic valve stenosis).
- **Risk factors for cardiovascular disease.** Women with Turner syndrome have an increased risk of diabetes and high blood pressure — conditions that increase the risk of developing diseases of the heart and blood vessels.
- **Hearing loss.** Hearing loss is common among girls or women with Turner syndrome. In some cases, this is 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.

- **Kidney problems.** About one-third of girls with Turner syndrome have some malformation of the kidneys. Although these abnormalities generally don't cause medical problems, they may increase the risk of high blood pressure and urinary tract infections.
- **Immune disorders.** Girls and women with Turner syndrome have an increased risk of certain immune system disorders, including a condition that causes an underactive thyroid (hypothyroidism). This disorder results in the low production of hormones important for controlling heart rate, growth and metabolism. There's also an increased risk of diabetes, inflammatory bowel disease, and wheat intolerance (celiac disease), a condition that affects how your body processes carbohydrates in food.
- **Dental problems.** Poor or abnormal tooth development may lead to a greater risk of tooth loss. The shape of the roof of the mouth and lower jaw often results in crowded teeth and a poorly aligned bite.
- **Vision problems.** Girls with Turner syndrome have an increased risk of weak muscle control of eye movements (strabismus) and farsightedness (hyperopia).
- **Skeletal problems.** Problems with the growth and development of bones increase the risk of abnormal curvature of the spine (scoliosis) and forward rounding of the upper back (kyphosis). Women with Turner syndrome are also at increased risk of developing weak, brittle bones (osteoporosis).
- **Pregnancy complications.** Most women with Turner syndrome are infertile. However, a very small number of women may become pregnant spontaneously, and some can become pregnant with fertility treatment. Because women with Turner syndrome are at increased risk of aortic dissection during pregnancy, they should be evaluated by a cardiologist before pregnancy. They're also at risk of developing high blood pressure and gestational diabetes during pregnancy.
- **Psychological issues.** Girls and women with Turner syndrome may have learning disabilities, particularly math and spatial concepts, difficulties functioning well in social situations, and an increased risk of attention-deficit/hyperactivity disorder (ADHD).

How you learn if your daughter has Turner syndrome will depend on the degree to which it has affected her development.

- **Before birth.** Certain features may be detected on prenatal ultrasound screening and confirmed with prenatal diagnostic testing.
- **At birth.** If certain conditions — such as a webbed neck or other distinct physical features — are readily apparent at birth, diagnostic tests will likely begin before your child leaves the hospital.
- **During childhood or teen years.** Your family doctor or pediatrician may suspect the disorder later in your daughter's childhood if she isn't growing at an expected rate or isn't beginning puberty at an expected time. Diagnostic testing can confirm the diagnosis.

Well-baby visits and annual checkups

It's important to take your daughter to all regularly scheduled well-baby visits and annual

appointments throughout childhood. These visits are an opportunity for your doctor to take height measurements, note delays in expected growth and identify other problems in physical development.

Your daughter's doctor may ask questions such as:

- What concerns do you have about your daughter's growth or development?
- How well does she eat?
- Has your daughter begun to show signs of puberty?
- Is she experiencing any learning difficulties at school?
- How does she do in peer-to-peer interactions or social situations?

Talking to your doctor about Turner syndrome

If your family doctor or pediatrician believes that your daughter shows signs or symptoms of Turner syndrome and suggests diagnostic tests, you may want to ask these questions:

- What diagnostic tests will we need?
- When will we know the results of the tests?
- What specialists will we need to see?
- How will you screen for disorders or complications that are commonly associated with Turner syndrome?
- How can I help monitor my daughter's health and development?
- Can you suggest educational materials and local support services regarding Turner syndrome?

If your family doctor or pediatrician suspects that your daughter has Turner syndrome, a lab test will likely be done to analyze your daughter's chromosomes using cells from a blood sample. The test results are a specialized image (karyotype) that enables a specialist to count and judge the condition of each chromosome in a sample.

Prenatal diagnosis

A diagnosis is sometimes made during fetal development. Certain features on an ultrasound image may raise suspicion that your baby has Turner syndrome or another genetic condition affecting development in the womb. Prenatal screening tests that evaluate the baby's DNA in the mother's blood (cell-free fetal DNA testing or noninvasive prenatal screening) may also indicate an increased risk of Turner syndrome.

Your pregnancy and childbirth specialist (obstetrician) may ask if you're interested in additional tests to make a diagnosis before your baby's birth. One of two procedures can be performed to test for Turner syndrome:

- **Chorionic villus sampling.** This involves removal of a small piece of tissue from the placenta.

- **Amniocentesis.** In this test, a sample of the amniotic fluid is taken from the uterus.

Discuss the benefits and risks of prenatal testing with your doctor.

Health care team

Because Turner syndrome can result in several developmental problems and medical complications, several specialists may be involved in screening for specific conditions, making diagnoses, recommending treatments and providing care.

This team may evolve as your child's needs change, and your family doctor or pediatrician can coordinate the care. Specialists on your care team may include some or all of these professionals:

- Hormone disorder specialist (endocrinologist)
- Heart specialist (cardiologist)
- Specialist in women's health (gynecologist)
- Specialist in skeletal disorders (orthopedist)
- Ear, nose and throat (ENT) specialist
- Dental specialist in correcting problems with the alignment of teeth (orthodontist)
- Specialist in vision problems and other eye disorders (ophthalmologist)
- Mental health provider, such as a psychologist or psychiatrist
- Developmental therapist, who specializes in therapy to help your child develop age-appropriate behaviors, social skills and interpersonal skills
- Special education instructors
- Medical geneticist

The primary treatments for nearly all girls and women include hormone therapies:

- **Growth hormone.** Growth hormone therapy is recommended for most girls with Turner syndrome. The goal is to increase height as much as possible at appropriate times during your daughter's childhood and teen years. Growth hormone treatment is usually given several times a week as injections of somatropin (Humatrope, Genotropin, Saizen, others). If height is very short, some doctors may recommend an androgen called oxandrolone (Oxandrin) in addition to the growth hormone.
- **Estrogen therapy.** Most girls with Turner syndrome need to start estrogen and related hormone therapy in order to begin puberty and achieve adult sexual development. Estrogen may also help your daughter grow when used along with growth hormone. Estrogen replacement therapy usually continues throughout life, until a woman reaches the average age of menopause.

Because of the range of symptoms and complications, other treatments are tailored to address your daughter's particular problems.

Transition to adult care

It's important to help your daughter prepare for the transition from care with your family doctor or pediatrician to her own adult medical care. Her doctor can continue to coordinate care among a number of specialists throughout her life.

- Regular checkups have shown substantial improvements in the quality and length of life for women with Turner syndrome.
- Problems that often arise during adulthood include hearing loss, eye problems, high blood pressure, diabetes and osteoporosis.
- Periodic follow-up with a heart specialist (cardiologist) is important, as are regular ultrasounds of the heart (echocardiography), a test that uses sound waves to produce an image of the heart.
- Healthy lifestyle habits, such as maintaining proper weight and exercising regularly, are important throughout life.

Pregnancy and fertility treatment

Few women with Turner syndrome can become pregnant without fertility treatment. Those who can are still likely to experience failure of the ovaries and subsequent infertility very early in adulthood. So it's important to discuss timing of pregnancy with your doctor.

Some women with Turner syndrome can become pregnant with the donation of an egg or embryo. This requires a specially designed hormone therapy to prepare the uterus for pregnancy.

In most cases, women with Turner syndrome have relatively high-risk pregnancies. It's important to discuss those risks with your doctor.

The Turner Syndrome Society of the United States and other organizations provide educational materials, resources for families and information about support groups. Groups for parents provide an opportunity to exchange ideas, develop coping strategies and locate resources.

Peer groups for girls with Turner syndrome can help reinforce your daughter's self-esteem and provide her with a social network of people who understand her experience with Turner syndrome.

References

1. Turner syndrome. Genetics Home Reference. http://ghr.nlm.nih.gov/condition/turner-syndrome. Accessed July 22, 2014.
2. Learning about Turner syndrome. National Human Genome Research Institute. http://www.genome.gov/19519119. Accessed July 22, 2014.
3. Turner syndrome. National Organization for Rare Disorders (NORD). https://www.rarediseases.org/rare-disease-information/rare-diseases/byID/112/viewFullReport. Accessed July 22, 2014.

4. Turner syndrome. Eunice Kennedy Shriver National Institute of Child Health and Human Development. http://turners.nichd.nih.gov/index.html. Accessed July 22, 2014.
5. Saenger P. Clinical manifestations and diagnosis of Turner syndrome (gonadal dysgenesis). http://www.uptodate.com/home. Accessed July 22, 2014.
6. Milbrandt T, et al. Turner syndrome. Pediatrics in Review. 2013;34:420.
7. Saenger P. Management of Turner syndrome (gonadal dysgenesis). http://www.uptodate.com/home. Accessed July 22, 2014.
8. Turner Syndrome Society of the United States. http://www.turnersyndrome.org/. Accessed July 26, 2014.
9. Wick MJ (expert opinion). Mayo Clinic, Rochester, Minn. Aug. 1, 2014.
10. Hoecker JL (expert opinion). Mayo Clinic, Rochester, Minn. July 27, 2014.
11. Freriks K, et al. Long-term effects of previous oxandrolone treatment in adult women with Turner syndrome. European Journal of Endocrinology. 2013;168:91.

Aug. 23, 2014

Original article: http://www.mayoclinic.org/diseases-conditions/turner-syndrome/basics/definition/CON-20032572