TURNER SYNDROME

Ullrich–Turner Syndrome, Bonnevie-Ullrich-Turner Syndrome, Gonadal Dysgenesis, 45,x, Monosomy X, Ts

Turner syndrome is a genetic disorder in which a female is partly or completely missing one X chromosome that results in ovarian dysgenesis.

診断 | 女性

Related Diagnoses:
- Obesity
- Anovulation
- Autoimmune disorders
- Premature ovarian failure
- Hypogonadism
- Amenorrhoea
- Gonadal dysgenesis

ℹ️ About Turner syndrome

Turner’s syndrome is a genetic disorder that affects only females. Turner syndrome is caused by a chromosomal abnormality in which all or part of one of the X chromosomes is missing or altered. While most people have 46 chromosomes, people with TS usually only have 45. This condition occurs in about 1 out in 2,500 female live births worldwide (but it is much more common among miscarriages and stillbirths).

There are different types of Turner syndrome depending on how much of the X chromosome is missing. Roughly half of the females with classic Turner syndrome have monosomy X, which means each cell in the body has only one copy of the X chromosome than the typical two. This may be due to a nondisjunction in the father. Meiotic errors that lead to the production of X with p arm deletions or abnormal Y chromosomes are also mostly found in the father. Isochromosome X or ring chromosome X on the other hand are formed equally often by both parents. Overall, the functional X chromosome mostly comes from the mother. In mosaic Turner syndrome, a complete X chromosome is missing in just some cells. In most cases, Turner syndrome is a sporadic event, and for the parents of an individual with Turner syndrome the risk of recurrence is not increased for subsequent pregnancies. Rare exceptions may include the presence of a balanced translocation of the X chromosome in a parent, or where the mother has X0 mosaicism restricted to her germ cells.

Signs and symptoms vary among those affected. Heart defects, diabetes, and low thyroid hormone occur more frequently. Most people with TS have normal intelligence. Many, however, have troubles with spatial visualization such as that needed for mathematics. Vision and hearing problems occur more often.

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Diagnosis is based on physical signs and genetic testing. A diagnosis of Turner syndrome can be made during preimplantation genetic screening, prenatal tests, at birth, or at any other time. Pre-implantation genetic screening (PGS) is a comprehensive chromosome screening in which an embryo biopsy is taken either at day 3 or day 5 and all 24 chromosomes are examined—the 22 non-sex chromosomes plus the two sex chromosomes (X & Y). As a result of PGS, embryo with proper genetic constitution may be transferred into woman’s uterus. Procedures during prenatal diagnosis include sampling of a small piece of tissue from the placenta or sampling of the amniotic fluid taken from the uterus to detect for any abnormal amniocentesis. Also, a doctor can suspect Turner syndrome based on certain physical features of an individual such as poor physical development and heart or kidney problems. Other tests include blood hormone levels, echocardiogram, MRI of the chest, pelvic exam, or ultrasound of the reproductive organs. Nonetheless, a doctor usually confirms Turner syndrome with a special blood test called a karyotype. This blood test produces an image of the composition of an individual’s chromosomes. From the image, the doctor can identify whether one of the sex chromosomes is missing or partially deleted. The common karyotype that causes Turner syndrome is called “45X” which means an individual has 44 autosomes and a single X chromosome. A karyotype can also reveal mosaic Turner syndrome, which is when an individual’s body is composed of cells with two X chromosomes and some having one X chromosome.
Turner syndrome occurs in between one in 2000 and one in 5000 females at birth. All regions of the world and cultures are affected about equally. People with TS have a shorter life expectancy, mostly due to heart problems and diabetes. As a chromosomal condition, there is no cure for Turner syndrome. However, much can be done to minimize the symptoms. Scientists have developed several treatments to correct some of the problems associated with Turner syndrome. Treatment becomes easier and effective if the disease is diagnosed at an early stage. Non-medical treatments such as psychological therapy and sex education can also help to reduce the mental stress and impact of complications associated with Turner syndrome. Overall, regular checkups and appropriate care can give most girls and women the potential live relatively healthy and independent lives.

**Associated diseases**

Among autoimmune disorders associated with TS, Hashimoto’s thyroiditis has been estimated to affect around 50% of TS patients. Other most commonly associated autoimmune disorders are: celiac disease (CD), ulcerative colitis, Crohn’s disease, psoriasis, idiopathic thrombocytopenic purpura, vitiligo and juvenile rheumatoid arthritis.

**Complications**

There are many complications and adverse effects associated with those who have Turner syndrome. These can include arthritis, cataracts, diabetes, heart defects, high blood pressure, kidney problems, middle ear infections, obesity and scoliosis in adolescence.

**Risk factors**

No environmental risks are known and the mother's age does not play a role. Even family history doesn’t seem to be a risk factor.

### Impact on fertility

Women with Turner syndrome are almost universally infertile since TS is characterized by primary amenorrhea, premature ovarian failure and streak gonads. Usually estrogen replacement therapy is used to spur growth of secondary sexual characteristics at the time when puberty should onset. While very few women with Turner syndrome menstruate spontaneously, estrogen therapy requires a regular shedding of the uterine lining (“withdrawal bleeding”) to prevent its overgrowth. Withdrawal bleeding can be induced monthly, like menstruation, or less often, usually every three months, if the patient desires. Estrogen therapy does not make a woman with nonfunctional ovaries fertile, but it plays an important role in assisted reproduction; the health of the uterus must be maintained with estrogen if an eligible woman with Turner syndrome wishes to use IVF (using donated oocytes). While some women with Turner syndrome have successfully become pregnant and carried their pregnancies to term, this is very rare and is generally limited to those women whose karyotypes are not 45,X. Even when such pregnancies do occur, there is a higher than average risk of miscarriage or birth defects, including Turner syndrome or Down syndrome. As more women with Turner syndrome complete pregnancy thanks to modern techniques to treat infertility (donor egg can be used to create an embryo, which is carried by the Turner syndrome woman), it has to be noted that pregnancy may be a risk of cardiovascular complications for the mother. Indeed, several studies had suggested an increased risk for aortic dissection in pregnancy. Three deaths have even been reported. The influence of estrogen has been examined but remains unclear. It seems that the high risk of aortic dissection during pregnancy in women with Turner syndrome may be due to the increased hemodynamic load rather than the high estrogen rate. Of course these findings are important and need to be remembered while following a pregnant patient with Turner syndrome.

### Prevention

Turner syndrome cannot be prevented. This genetic disorder occurs randomly, but can be diagnosed very soon i.e. during preimplantation genetic screening of early embryo.
Symptoms

Signs and symptoms vary among those affected. The following is a list of common symptoms of Turner syndrome. It is important to note that an individual may have any combination of symptoms and would unlikely have all symptoms.

- short stature
- lymphedema (swelling) of the hands and feet
- broad chest (shield chest) and widely spaced nipples
- low hairline
- low-set ears
- reproductive sterility
- rudimentary ovaries gonadal streak (underdeveloped gonadal structures that later become fibrosed)
- amenorrhea, or the absence of a menstrual period
- increased weight, obesity
- shield shaped thorax of heart
- shortened metacarpal IV
- small fingernails
- characteristic facial features
- webbed neck from cystic hygroma in infancy
- coarctation of the aorta
- bicuspid aortic valve
- horseshoe kidney
- visual impairments sclera, cornea, glaucoma, etc.
- ear infections and hearing loss
- high waist-to-hip ratio (the hips are not much bigger than the waist)
- attention Deficit/Hyperactivity Disorder or ADHD (problems with concentration, memory, attention with hyperactivity seen mostly in childhood and adolescence)
- nonverbal learning disability (problems with math, social skills and spatial relations)

Other features may include a small lower jaw (micrognathia), cubitus valgus, soft upturned nails, palmar crease, and drooping eyelids. Less common are pigmented moles and a high-arch palate (narrow maxilla).

Therapies

Self therapy

Non-existing.

Conventional medicine

Pharmacotherapy

Growth hormone

Doctors might use a shot of a growth hormone known as Genotropin (Pfizer). Growth hormone, either alone or with a low dose of androgen, will increase growth and probably final adult height. During childhood and adolescence, growth hormones may help a child grow taller. According to the National Human Genome Research Institute, “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”. The growth hormone works by stimulating the growth and reproduction of cells. Unfortunately, growth hormones cannot stimulate bone growth after puberty.

Estrogens

Another treatment is estrogen replacement therapy. Estrogen replacement therapy such as the birth control pill has been used since the condition was described in 1938 to promote development of secondary sexual characteristics. Estrogens are crucial for maintaining good bone integrity, cardiovascular health and tissue health. It is usually started at the time of normal puberty to induce breast development and other sexual characteristics. Women with Turner Syndrome who do not have
spontaneous puberty and who are not treated with estrogen are at high risk for osteoporosis and heart conditions.

Surgical therapy

Surgeries can correct other medical conditions such as heart defects and kidney problems.

Assisted reproduction

Most adult women with Turner syndrome already have established ovarian failure with high serum follicle-stimulating hormone (FSH) levels at the time they wish to start a family. Although this doesn’t indicate complete absence of viable follicles, ovarian stimulation for homologous in vitro fertilization is often not realistic and patients are usually guided towards egg donation.

In vitro fertilization with donor oocytes and subsequent embryo transfer has been the predominant fertility option for such patients. Women with Turner mosaicism (46XX/45XO) and normal FSH levels may have an adequate ovarian reserve and undergo attempts at traditional assisted reproduction (IVF-ICSI). At the time of retrieval, an ovarian biopsy may be performed in order to directly evaluate the ovarian karyotype. A successful pregnancy may result from the embryos demonstrating a normal karyotype after preimplantation genetic screening (PGS).

The combination of ovarian tissue cryobanking and immature oocyte collection from the tissue, followed by in vitro maturation and vitrification of matured oocytes represent promising approaches to fertility preservation for young women with mosaic Turner syndrome.

Find more about related issues

Diagnoses

Obesity
A disease of excess body fat that can have a negative effect on health, leading to reduced life expectancy and other health problems.
Learn more at: www.fertiltypedia.org/therapy/diag/obesity

Anovulation
Failure of the ovaries to release an oocyte over a period of time generally exceeding 3 months.
Learn more at: www.fertiltypedia.org/therapy/diag/anovulation

Autoimmune disorders
A condition arising from an abnormal immune response to a normal body part.
Learn more at: www.fertiltypedia.org/therapy/diag/autoimmune-disorders-1

Premature ovarian failure
The loss of function of the ovaries before age 40.
Learn more at: www.fertiltypedia.org/therapy/diag/premature-ovarian-failure

Hyponadism
A medical term which describes a diminished functional activity of the gonads – the testes and ovaries.
Learn more at: www.fertiltypedia.org/therapy/diag/hyponadism

Amenorrhoea
The absence of a menstrual period in women of reproductive age.
Learn more at: www.fertiltypedia.org/therapy/diag/amenorrhoea
Gonadal dysgenesis
Any congenital developmental disorder of the reproductive system characterized by a progressive loss of germ cells on the developing gonads.
Learn more at: www.fertilypedia.org/therapy/diag/gonadal-dysgenesis

Organs

Ovary
The ovum-producing organs of the internal female reproductive system
Learn more at: www.fertilypedia.org/edu/organs/ovary

Reproductive cells

Oocyte
A female germ cell involved in reproduction.
Learn more at: www.fertilypedia.org/edu/reproductive-cells/oocyte

Biological control

Follicle-stimulating hormone
FSH is a hormone secreted by the anterior pituitary gland. It regulates the development, growth, pubertal matur and reproductive functions of the body
Learn more at: www.fertilypedia.org/edu/biological-control/follicle-stimulating-hormone

Risk factors

High level of FSH
FSH levels above what an expected levels for one's age and is indicator of proper ovarian function.
Learn more at: www.fertilypedia.org/therapy/rf/high-level-of-fsh

High level of LH
A condition with high blood luteinizing hormone (LH) leading to irregular periods and reduced fertility in both females and males.
Learn more at: www.fertilypedia.org/therapy/rf/high-level-of-lh

Symptoms

Absence of menstrual periods
The absence of a menstrual period in a woman of reproductive age.
Learn more at: www.fertilypedia.org/edu/symptoms/absence-of-menstrual-periods

ADHD
A developmental disorder in which a person has a persistent pattern of impulsiveness and inattention, with or without a component of hyperactivity.
Learn more at: www.fertilypedia.org/edu/symptoms/adhd

Arthritis
A term often used to mean any disorder that affects joints.
Learn more at: www.fertilypedia.org/edu/symptoms/arthritis

Cataracts
A clouding of the lens in the eye that affects vision.
Learn more at: www.fertilypedia.org/edu/symptoms/cataracts
Cognitive impairment
Problems with thought processes.
Learn more at: www.fertilitypedia.org/edu/symptoms/cognitive-impairment

Hearing loss
A partial or total inability to hear.
Learn more at: www.fertilitypedia.org/edu/symptoms/hearing-loss

Heart defects
A defect in the structure of the heart and great vessels which is present at birth.
Learn more at: www.fertilitypedia.org/edu/symptoms/heart-defects

Increased level of FSH
A condition with high serum follicle-stimulating hormone (FSH) concentration.
Learn more at: www.fertilitypedia.org/edu/symptoms/increased-level-of-fsh

Infertility
The failure to achieve a clinical pregnancy after 12 months or more of regular unprotected sexual intercourse.
Learn more at: www.fertilitypedia.org/edu/symptoms/infertility

Lymphedema (swelling) of the hands and feet
An impaired lymphatic return and swelling of the hands and feet.
Learn more at: www.fertilitypedia.org/edu/symptoms/lymphedema-swelling-of-the-hands-and-feet

Micrognathia
A condition characterized by an undersized lower jaw.
Learn more at: www.fertilitypedia.org/edu/symptoms/micrognathia

Middle ear infections
The inflammation of the middle ear often due to bacterial infections.
Learn more at: www.fertilitypedia.org/edu/symptoms/middle-ear-infections

Overweight
Body weight that’s greater than what is considered healthy for a certain height.
Learn more at: www.fertilitypedia.org/edu/symptoms/overweight

Reduced height
A height of a human being which is below typical.
Learn more at: www.fertilitypedia.org/edu/symptoms/reduced-height

Scoliosis
Learn more at: www.fertilitypedia.org/edu/symptoms/scoliosis

Short fingers and small fingernails
Shortness of the fingers and toes or shortness of the fingernails.
Learn more at: www.fertilitypedia.org/edu/symptoms/short-fingers-and-small-fingernails

Vision and hearing problems
A decreased ability to see and hear to a degree that causes problems.
Learn more at: www.fertilitypedia.org/edu/symptoms/vision-and-hearing-problems

Therapies
**Egg donation**
Process by which a woman donates eggs for purposes of assisted reproduction or biomedical research.
Learn more at: [www.fertilitypedia.org/edu/therapies/egg-donation](http://www.fertilitypedia.org/edu/therapies/egg-donation)

**Embryo donation**
The giving of embryo to another person or couple for conception or to research.
Learn more at: [www.fertilitypedia.org/edu/therapies/embryo-donation](http://www.fertilitypedia.org/edu/therapies/embryo-donation)

**ICSI**
A micromanipulative fertilization technique in which a single sperm is injected directly into an egg.
Learn more at: [www.fertilitypedia.org/edu/therapies/icsi](http://www.fertilitypedia.org/edu/therapies/icsi)

**Preimplantation genetic screening**
The term PGS is used to denote procedures that do not look for a specific disease but to identify embryos at risk of de-novo occurring aneuploidies
Learn more at: [www.fertilitypedia.org/edu/therapies/preimplantation-genetic-screening-1](http://www.fertilitypedia.org/edu/therapies/preimplantation-genetic-screening-1)

**Sperm donation**
The procedure in which a man (sperm donor) provides his sperm for fertility treatment.
Learn more at: [www.fertilitypedia.org/edu/therapies/sperm-donation](http://www.fertilitypedia.org/edu/therapies/sperm-donation)

**Standard IVF**
A process in which an egg is fertilised by sperm outside the body: in vitro. Own or donated gametes may be used.
Learn more at: [www.fertilitypedia.org/edu/therapies/standard-ivf](http://www.fertilitypedia.org/edu/therapies/standard-ivf)

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**Gallery**

**Neck Turner**
Girl with Turner syndrome before and immediately after her operation for neck-webbing.

**Puffy feet - Turner syndrome**
Lymphedema, puffy legs of a newborn with Turner syndrome.
45,X - Turner syndrome

45,X karyotype, showing an unpaired X at the lower right.

Sources

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