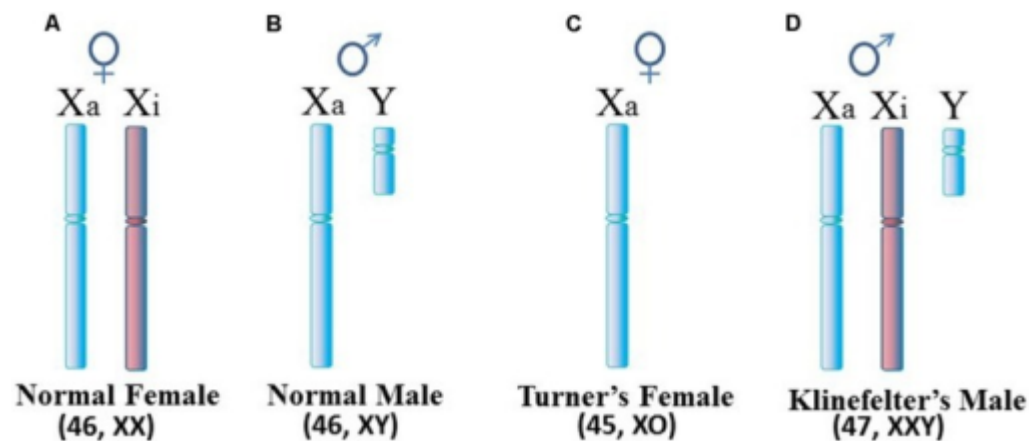


## Turner Syndrome (Gonadal Dysgenesis) — Diagnosis and Management

[See online here](#)

**The future gender of the individual coming into being is already fixed on a chromosomal level during the fertilization of the ovum. The determination and differentiation of the gender take place during the embryogenesis.**

**Disturbances of the sexual differentiation can arise due to chromosome anomalies, gene mutations and exogenous or endogenous influences, which are remarkable at last in the puberty and cause patients to consult a doctor.**



## Definition, Epidemiology, and Etiology of Turner Syndrome

Turner syndrome is a disorder of genital development **defined by a chromosomal aberration**. It is characterized by monosomy, which means that only 1 of 2 gonosomes is present (X-chromosome in this case), resulting in the karyotype 45,X0 with a female phenotype.

With a frequency of 1 in 2500–7500 (living girls), it is the most frequent gonadal chromosome anomaly in women. A high spontaneous abortion rate accompanies the syndrome. 99% of pregnancies with 45,X0 karyotype spontaneously abort.

The cause is the loss of a gonosome during the embryogenesis, which can occur at different stages of development (spermatogenesis or oogenesis, fertilization) and is **based on a nondisjunction of the chromosomes**, more specifically of the chromatids. In 70% of cases, the gonosome is the X-chromosome of the father.

A structurally modified X-chromosome can be a cause as well. Chromosomal mosaics with unobtrusive and striking cells (e.g., 46,XX/45,X0) can also occur, but the form of the pathology, in this case, is proportionally weakened.

While the occurrence of the Turner syndrome does not correlate with the age of the mother, the advanced age of the father plays a possible role in the pathogenesis.

## Symptoms and Clinical Appearance of Turner Syndrome

In the case of Turner syndrome, the phenotype, as well as the psychosexual setting, are female. The **main symptoms are primary amenorrhoea and dwarfism** (143-147 cm). The growth spurt is absent. Patients are infertile and show sexual infantilism (hypoplastic female genitals due to a lack of hormonal stimulation). While the gonads develop normally at first, they start to degenerate, and fibrosis occurs in the 3rd month of pregnancy, which finally causes the ovaries to exist only as undifferentiated tissue (streak gonads).

### The ABCs of Turner Syndrome are:

- **A**udiology
- **A**utoimmunity
- **C**ardiac defects
- **D**ermatologic manifestations
- **F**ailure to grow
- **G**astrointestinal disease
- **G**enetics
- **M**etabolic disorders
- **O**rthopedic complications
- **R**enal abnormalities -> 30-50% with horseshoe-shaped kidney being most common
- **R**eproductive and pubertal disorders
- **X**-linked disorders

### Further, facultative symptoms are:

- Short neck
- Pterygium colli (webbed neck)
- Low hairline
- Lymphedema on the back of the hand and the foot (in the 1st weeks of life) giving a sausage-like appearance to fingers
- Infants have a higher incidence of congenital hip joint dislocation
- Cubitus valgus (x-formed arms)
- Shield thorax with widely diverged nipples and funnel chest
- Multiple (benign) pigment nevi
- Nail and ear dysplasia
- Ptosis, strabismus, cataracts, red-green color blindness may be present
- Incidence of gastrointestinal bleeding, Crohn's disease, and ulcerative colitis are increased
- Malformations of the kidneys and the urinary system
- Malformations of the skeleton (e.g., deformities of the spine) and anomalies of the ligamentous apparatus
- Heart defect: e.g., aortic stenosis, aortic coarctation, anomalies of the pulmonary veins
- Hypertension may be present

These symptoms do not necessarily occur together and can be pronounced to a different

extent, according to the person. The intelligence of the affected persons can be normal or reduced. There are often partial inefficiencies (e.g., mathematics, spatial orientation).

As already mentioned above, some corporal anomalies can occur with the presence of a chromosomal mosaic. Depending on the ovarian function, puberty development and even spontaneous pregnancy are possible. But, spontaneous pregnancy may be accompanied by elevated abortion rates and congenital malformations of the children.

## Diagnosis and Differential Diagnosis of Turner Syndrome

Turner syndrome is often diagnosed when patients consult a doctor due to amenorrhoea and the absence of puberty.

Due to the insufficiency of the ovaries, very little estrogen is produced that triggers superior centers to produce more hormones. Correspondingly, raised levels of follicle-stimulating hormone (FSH) and luteinizing hormone (LH), as well as reduced levels of estrogen, are found during an endocrinological examination. As the disorder lies at the level of the gonads, it is also called hypergonadotropic hypogonadism or primary hypogonadism, as these terms are synonymous.

Chromosomal analysis of peripheral lymphocytes is appropriate to diagnose the condition. The karyotype of the patient can be examined with the karyogram.

The differential diagnosis must exclude tumors (e.g., gonadoblastoma, dysgerminoma).

Patients with virilizing symptoms should be examined for the presence of Y chromosome material; these patients may have malignant gonadoblastoma or testicular tissue.

Prenatal	Neonatal	Adolescence
Ultrasound findings include fetuses who present with: <ul style="list-style-type: none"> <li>• Intrauterine growth retardation (IUGR)</li> <li>• Large septate cystic hygromas</li> <li>• Nuchal thickening               <ul style="list-style-type: none"> <li>• Short femur</li> <li>• Total body lymphangiectasia and cardiac defects</li> </ul> </li> </ul>	Physical findings include: <ul style="list-style-type: none"> <li>• Low birth weight</li> <li>• 30% present with lymphedema of upper and lower extremities</li> <li>• Webbed neck (pterygium colli)</li> <li>• High arched palate</li> <li>• Low-set prominent ears</li> <li>• Low posterior hairline</li> <li>• Micrognathia</li> </ul>	The most common presentation is: <ul style="list-style-type: none"> <li>• Short stature</li> <li>• Amenorrhea</li> <li>• Lack of secondary sexual characteristics</li> </ul> 30% of patients undergo some degree of spontaneous puberty.

## Therapy of Turner Syndrome

Turner syndrome is **not curable as the disorder is at the molecular level** (missing chromosome). However, treatment is aimed at symptomatic therapy, which improves the physical and psychic characteristics of the patient.

Hormone substitution is induced at the beginning of puberty (approx. at the age of 12 or 13). Estrogens enhance the growth of primary and secondary sexual characteristics (vagina, uterus, breast development, pubic hair) and prevent osteoporosis and arteriosclerosis. For example, a preparation of estradiol valerate (1-2 mg) is used. Gestagens should be given at least 10 days a month as they trigger menstruation; hence, the endometrium is transformed and not stimulated continuously, which prevents the development of an endometrium carcinoma.

The final height of the patients can be increased by up to 10 cm through a dose of synthetic growth hormone (hGH) in early childhood.

Infertility is common in women affected with Turner's syndrome. But, pregnancy via egg donation can be considered.

Note: The sterility of patients with the karyotype 45,X0 is irreversible.

Surgical therapy for the removal of disturbing stigmata (e.g., pterygium colli) and psychosocial care for the patients is important.

## Complications of Turner Syndrome

Women who have Turner syndrome come down more frequently than the standard population with cardiovascular diseases, diabetes mellitus, thyroid diseases, and inflammable bowel diseases.

The life span of patients can be reduced by an average of 13 years.

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