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# Conceptual Basis of Turner Syndrome

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**Abstract**

Turner Syndrome (TS) is a relatively common chromosomal disorder affecting females. It is characterized by the partial or complete absence of one of the X chromosomes, resulting in a wide range of physical, developmental, and medical difficulties. TS has been the subject of extensive research and clinical studies, providing valuable insights into the genetic and physiological basis of female development. This article aims to provide a comprehensive overview of the conceptual basis of Turner Syndrome, its genetic origins, clinical manifestations, diagnostic criteria, and current therapeutic approaches.

**Keywords:** Turner syndrome, symptoms, X chromosome, diagnosis, treatment

## 1. Introduction

Turner Syndrome (TS) is a chromosomal condition that affects only females and is caused by the partial or complete absence of one of the two X chromosomes. Henry Turner described the syndrome in 1938, and it is named after him. The manifestation of the condition can vary, but common features include short stature, specific physical features, and dysgenesis of the genitals leading to infertility.

Turner syndrome is a hereditary disease that primarily impacts females. It is caused by the absence of one or all of the X chromosomes, the chromosomes that determine a person's sex. As a result, girls with Turner syndrome have distinct physical and intellectual characteristics. These include [1]:

- Short stature
- Neck creases
- Low hairline on the back of the neck
- Broad chest
- Narrow nose

Turner syndrome can also cause some intellectual and developmental problems, such as learning disabilities, delayed speech development, and attention and memory problems. However, with appropriate care and intervention, many people with Turner syndrome live full and productive lives. The disease was first described as hereditary in 1925 by the Soviet endocrinologist N.A. Shereshevsky. He then proved that the disease was caused by underdevelopment of the gonads and anterior pituitary gland and was associated with congenital defects. Shereshevsky-Turner syndrome was described in more detail by Dr. Henry Turner in 1938. Dr. Turner was an endocrinologist and medical geneticist working in the United States. In his seminal article, he described the clinical features of a group of girls with short stature, short necks with wing-like curls, and other physical features characteristic of this disorder. He also noted the presence of ovarian dysgenesis and infertility, which are hallmarks of Turner syndrome. Since its initial description, the syndrome has been further characterized and is now widely

recognized as a distinct genetic disorder. Today, the diagnosis of Turner syndrome is based on a combination of physical examination, genetic testing, and medical history.

### 2. X Chromosome Abnormalities in Turner Syndrome

Turner syndrome is caused by the absence of one or all of the X chromosomes (Figure 1) [3]. Generally, females possess two X chromosomes, whereas males possess one X and one Y chromosome. In Turner syndrome, females are missing either the entire X chromosome (45, X) or part of one X chromosome (45, X/46, XX mosaicism).

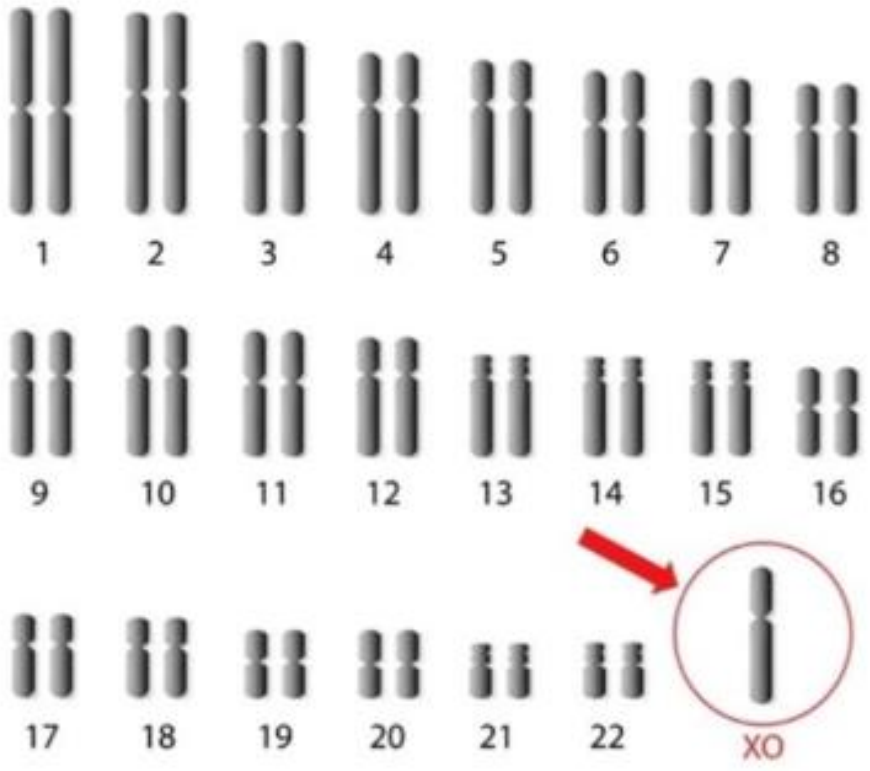


**Figure 1.** Chromosomal abnormalities in individuals with Turner syndrome [2], [3].

There are several genetic variants of Turner syndrome. These are the following:

*Monosomy X (45, X):* This is a condition in which a woman is missing one of her two X chromosomes. This can occur as a result of a random error during cell division in a developing egg or fetus.

*Structural anomalies:* This is a condition in which part of one of a woman's X chromosomes is missing or altered.

*Mosaicism:* This is a condition where some cells have two typical X chromosomes, while others have only one X chromosome or a structurally altered X chromosome [4].

The absence or alteration of the X chromosome can cause the characteristic physical and intellectual characteristics of Turner syndrome, as well as several other health problems. It is necessary to note that Turner syndrome is not caused by anything the mother does during pregnancy or how she raises the child. It is a spontaneous genetic event.

### 3. Symptoms of Turner Syndrome

Some of the most common physiological symptoms of Turner syndrome include (Figure 2) [3]:

**Short stature:** This is the most obvious and well-known characteristic of Turner syndrome. Girls with Turner syndrome are significantly shorter than their peers, and their growth hormone production stops at an early age.

**Ovarian Dysfunction:** Turner syndrome is associated with ovarian dysgenesis, which is the failure of the ovaries to develop normally. This leads to the absence of menstruation and infertility.

**Heart defects:** Some people with Turner syndrome may have structural heart abnormalities such as a bicuspid aortic valve, coarctation of the aorta, or patent ductus arteriosus.

**Skeletal problems:** Girls with Turner syndrome may have spinal abnormalities such as scoliosis, as well as joint problems such as dislocation.

**Kidney problems:** Some people with Turner syndrome may have structural kidney abnormalities, such as a horseshoe kidney, which is a fusion of two kidneys.

**Learning disabilities:** Some people with Turner syndrome may have difficulties with math and spatial skills, as well as mental deficits such as attention and memory problems.

**Hearing problems:** Some people with Turner syndrome may experience hearing loss, especially in the high-frequency range [5].

It is necessary to note that not all people with Turner syndrome will experience all of these symptoms, and the severity of symptoms can vary greatly. In addition, many girls with Turner syndrome can return to normal lives with proper care and treatment.

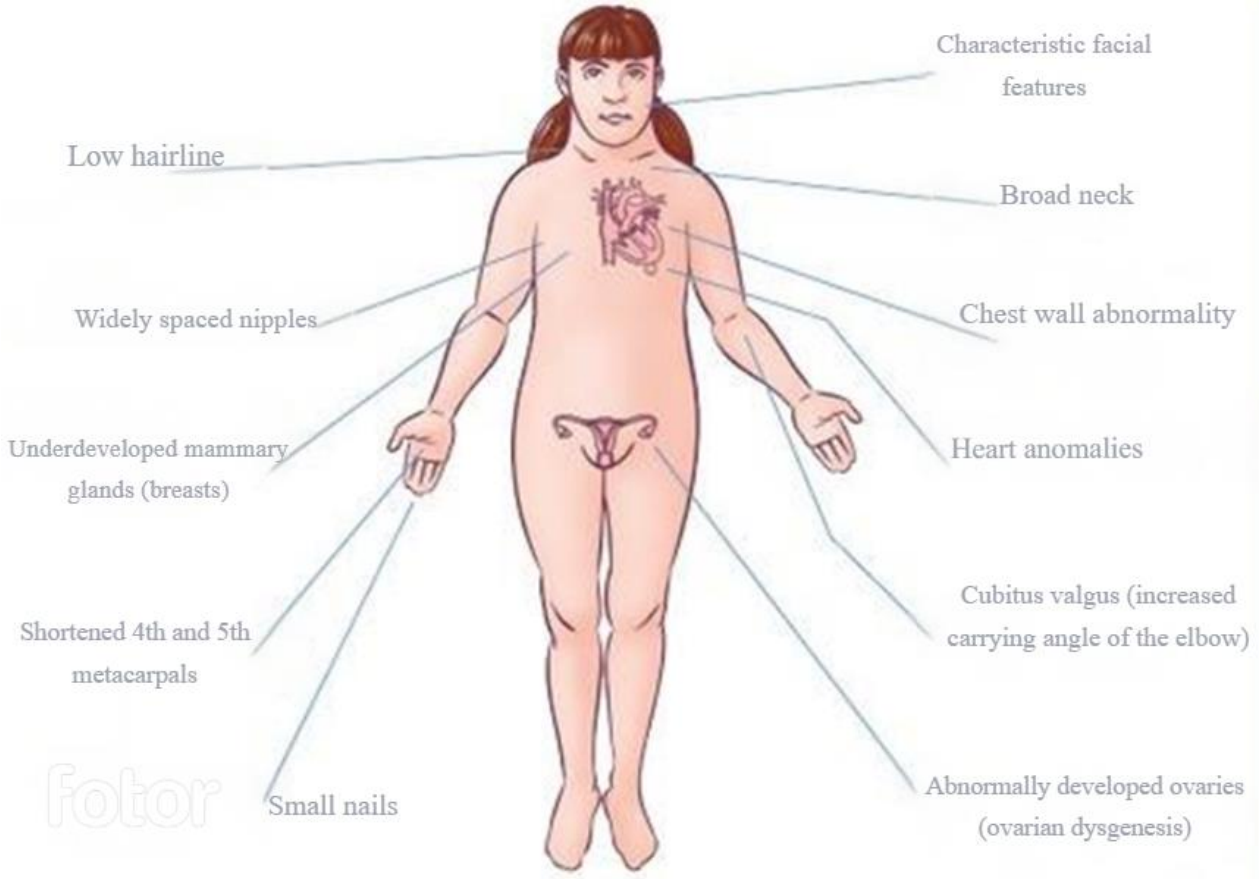


**Figure 2.** Appearance of a person with Turner syndrome [3].

**4. Diagnosis of Turner Syndrome**

Turner syndrome is usually diagnosed through a combination of physical examination, genetic testing, and medical history. Some diagnostic tools that may be used to diagnose Turner syndrome include:

**Physical examination:** During the examination, the doctor detects the characteristic physical signs of Turner syndrome, such as short stature, a broad chest, a short neck with wing-like folds, a low hairline at the back of the neck, narrow hips, and a high palate.

**Blood tests:** Blood tests can be used to analyze levels of hormones such as follicle-stimulating hormones (FSH), which are essential for ovarian function. Elevated FSH levels may indicate that the ovaries are not functioning properly, which is a characteristic feature of Turner syndrome.

**Chromosome analysis:** Chromosome analysis, also known as karyotyping or cytogenetic testing, is a laboratory test used to determine the number and structure of chromosomes. In Turner syndrome, a woman may have only one X chromosome (45, X) or she may have two X chromosomes, but one of them has a structural change (45, X/46, XX mosaicism).

**Ultrasound:** Ultrasound can be used to assess the condition of internal organs such as the heart, kidneys, and ovaries. In some cases, ultrasounds can detect structural abnormalities characteristic of Turner syndrome.

It should be noted that the diagnosis of Turner syndrome is not always clear; several tests may be required to clarify the diagnosis, but the final diagnosis is made based on the results of cytogenetic research (karyotype analysis).
Prenatal tests performed during pregnancy may reveal the presence of Turner syndrome (Figure 3) [6].

**Ultrasound:** A first-trimester screening test that assesses the fluid accumulation behind the fetus's neck. An aberrant reading signifies a chromosomal abnormality. This diagnostic test is conducted between 10 and 12 weeks of gestation for women with an abnormal first-trimester screening result. A minor specimen of the placenta, referred to as the chorionic villus, is examined for chromosomal anomalies [7].

Amniocentesis is conducted between 15 and 18 weeks of gestation. A sample of amniotic fluid, which surrounds the fetus, is collected and examined for aberrant protein levels that may signify particular diseases.

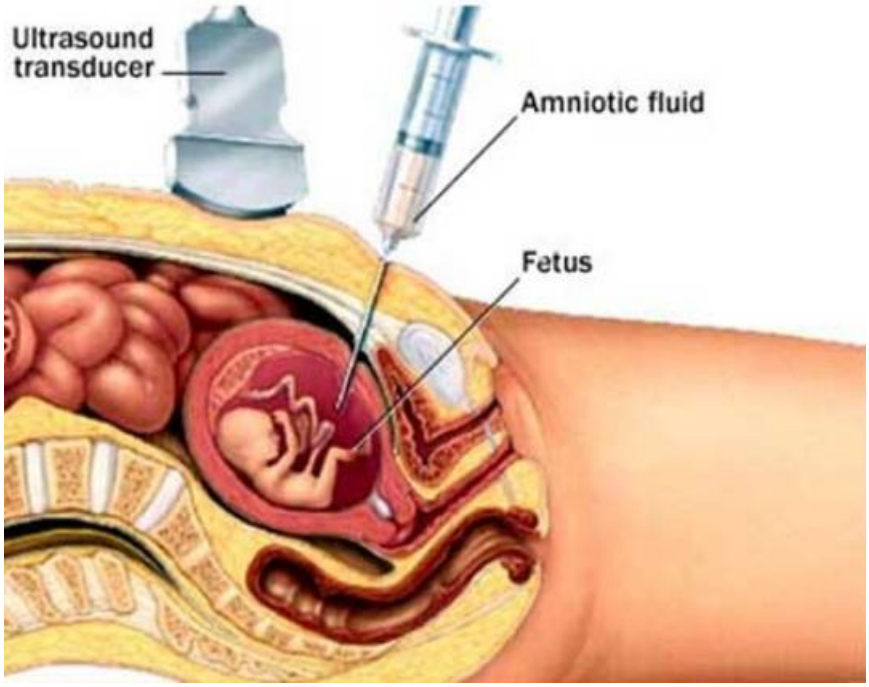


**Figure 3.** Tests to detect disease during pregnancy [6].

## 5. Treatment Strategies in Turner Syndrome

Turner syndrome is a chronic disorder without a cure; nevertheless, recent advancements in genetics and current treatment choices have significantly improved the lives of affected girls and women. Females with Turner syndrome may experience additional medical issues that require ongoing treatment and management throughout their lives. Growth hormone supplementation represents conventional therapy for height enhancement. The increase in height at the end of treatment is dependent upon various circumstances, including the age at which therapy begins, the length of treatment, and the administered hormone dosage [8]. Supplementing with growth hormones should begin at age 4 or as soon as the disease is recognized, whichever happens first. Research indicates that after 3-7 years of treatment, patients may achieve an increase in height of roughly 8-10 cm. No adverse consequences have been recorded. The majority of individuals necessitate ovarian hormone therapy, specifically estrogen, a female sex hormone. This can begin between the ages of 12 and 15 to initiate puberty and sustain normal female endocrine function. This is also crucial to the formation of additional sexual behaviors. Hormone replacement treatment (HRT) is crucial as it mitigates the risks linked to ovarian failure, particularly osteoporosis and cardiovascular disease. Hormone levels should be regularly monitored [8].

New efforts in assisted reproduction have created the possibility of pregnancy in these patients. Pregnancy rates of over 50% have been reported in patients with spontaneous menstruation. However, these patients exhibit a higher susceptibility to miscarriage and chromosomal anomalies. Twins are more typical. In a separate team, where patients possess completely striated ovaries, fertilization can be achieved via donated eggs. Heart valve problems appear in 30% of females with the condition. Surgical intervention or regular consultations with a cardiologist, accompanied by routine ultrasonography or echocardiography. Cardiac imaging assessments should be performed [9]. Hypertension is induced by the constriction of the aorta. Narrowing can be repaired surgically, or hypertension can be managed with pharmacological interventions. Hearing aids can be used to treat hearing issues brought on by childhood middle ear infections. Estrogen therapy can be used to treat osteoporosis, or bone thinning. Thyroid function tests should be used to monitor thyroid diseases. It is necessary to control metabolic diseases like diabetes and obesity. Ultrasound is necessary on a regular basis for kidney abnormalities. Regular exercise and a healthy lifestyle can be beneficial. Although the condition has no known treatment, females who suffer can live normal lives, but they typically require the care of multiple doctors.

## 6. Conclusion

When TS is diagnosed, the family should be provided with professional support from a medical geneticist, pediatric endocrinologist, or physician specializing in TS. TS can be diagnosed prenatally by ultrasound (USG) demonstrating fetal edema or a cystic hygroma of the neck. Ultrasonographically, left heart defect, renal anomalies, growth retardation, or relatively short limbs may indicate TS.

## Author Contributions

Laman A. Huseynova conducted the literature search and drafted the manuscript. Saltanat A. Aghayeva supervised the study, provided critical guidance on content and structure, and reviewed the manuscript.

## Conflict of Interest

The authors declare no conflicts of interest.

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**Abbreviations**

Turner Syndrome (TS), Follicle-Stimulating Hormone (FSH), Hormone Replacement Treatment (HRT), Ultrasonography (USG).

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