

### Developmental progress

Girls with Turner Syndrome usually have normal intelligence and their progress at school is generally good. However a small proportion of girls with Turner Syndrome may have specific learning difficulties. Sometimes they have particular difficulties with mathematics and geometry, but their reading age may be advanced. Activities involving dexterity, e.g. fine finger movements and co-ordination, can occasionally be a problem.

### Puberty

In girls with Turner Syndrome the eggs in the ovaries degenerate and disappear in early childhood and the ovaries stop functioning properly well before the age that puberty would normally begin. The ovaries normally produce the sex hormones oestrogen and progesterone and it is oestrogen that is needed to start puberty. When the ovaries do not function, puberty will only occur if replacement oestrogen therapy is given. The great majority of girls with Turner Syndrome do not start their periods or develop the adult female body shape without the help of some hormone treatment. Oestrogen is used to start off breast development, and progesterone and oestrogen together help produce regular periods.

### Infertility

Girls with Turner Syndrome are almost always infertile, because their ovaries are unable to produce eggs. A very small proportion of young women with Turner Syndrome may have a short time during their life when they are fertile.

Although girls with Turner Syndrome have non-functioning ovaries, they do have a normal womb and vagina, and will be able to have an entirely normal sex life. Some women with Turner Syndrome have had successful pregnancies using donated eggs and in vitro fertilisation (IVF).

### For more information about Turner Syndrome:

Clinical Genetics Departments

Northern Scotland (main base Aberdeen)

Tel: 01224 552120 Fax: 01224 559390

(Aberdeenshire, Moray, Highland, Western & Northern Isles)

Tayside (main base Dundee)

Tel: 01382 632035 Fax: 01382 645731

(Perth & Kinross, Angus, North East Fife)

South East Scotland (main base Edinburgh)

Tel: 0131 651 1012 Fax: 0131 651 1013

(Borders, Lothian, South West Fife)

West of Scotland (main base Glasgow)

Tel: 0141 201 0808 Fax: 0141 201 0361

(Glasgow, Argyll & Bute, Ayrshire, Dumfries & Galloway, Stirling,

Lanarkshire, Falkirk)

You may wish to contact the [Child Growth Foundation](#) or the [Turner Syndrome Support Society \(UK\)](#), which are support groups specifically for girls with Turner Syndrome and their families.

### The Child Growth Foundation

2 Mayfield Avenue, Chiswick

London W4 1PW

Telephone: 020 8994 7625/ 020 8995 0257

Website: [www.cgf.org.uk](http://www.cgf.org.uk)

### The Turner Syndrome Support Society (UK)

1/8 Irving Court, Hardgate

Clydebank G81 6BA

Telephone: 01389 380385/872511

E-mail address: [Turner.syndrome@tss.org.uk](mailto:Turner.syndrome@tss.org.uk)

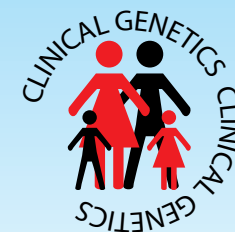
Website: [www.tss.org.uk](http://www.tss.org.uk)



### Seen in clinic by.....

This information sheet was compiled from the booklet 'Turner Syndrome: A guide for Patients and Parents', by the Child Growth Foundation. Thanks also to the Kennedy-Galton Centre. The leaflet was written by Clinical Genetics, Guys Hospital & updated by Genetic Interest Group Scotland. Updated Nov 2001

# Turner Syndrome



An information sheet  
for professionals

## Introduction

Humans are usually born with 46 chromosomes, which are arranged in 23 pairs. One of these pairs determines whether a baby is male or female, and these are known as the sex chromosomes. Boys are born with the sex chromosomes XY, and girls are born with two X chromosomes (XX).

Occasionally, however, a girl is born with only one X chromosome and this is known as **Turner Syndrome**.

The second X chromosome is usually missing in Turner Syndrome. Occasionally it may be present, but abnormal in some way. About 1 in 2,500 girls has Turner Syndrome. In Britain, it is estimated that there are about 10,000 girls and women who have Turner Syndrome. Even though these girls only have one normal X chromosome, they are 100 percent female. They may, however, have fertility problems in later life.

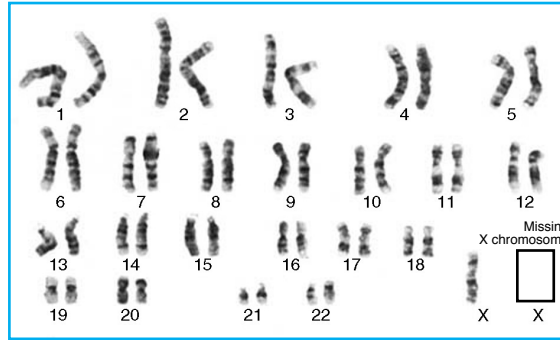
Turner Syndrome is named after Dr. Henry Turner, who first described it in 1938.

## What causes Turner Syndrome?

The cause of the missing or abnormal X chromosome in Turner Syndrome is not known. No risk factors, such as raised maternal age, diet during pregnancy etc have been identified as being associated with an increased risk of having a baby with Turner Syndrome.

## Diagnosis and chromosomal findings

Girls with Turner Syndrome often have a typical appearance, and this is how the diagnosis is often first suspected. The diagnosis can be confirmed by chromosome analysis.



In Turner Syndrome there will usually be only one X chromosome instead of two, so making 45 chromosomes in total. The picture above is an enlarged photograph of chromosomes from a person with Turner Syndrome.

When one X is missing in all the cells of the body this is often described as "classical Turner Syndrome". The missing X has been lost some time during cell division in the course of egg or sperm production. Some females have abnormalities in one of their two X chromosomes. This can also lead to Turner Syndrome.

Sometimes one X chromosome is missing from, or is abnormal in, some, but not all, of the cells of the body. This is referred to as "mosaic Turner Syndrome". In this case, some of the cells have a normal chromosome pattern but others do not. Girls with Turner Syndrome in a mosaic form often have less obvious physical characteristics and may not have fertility problems.

## When is the diagnosis made?

Turner Syndrome is sometimes suspected when an ultrasound scan is performed during pregnancy. When a scan suggests Turner Syndrome, it can be confirmed by checking the baby's chromosome pattern. This can be done during pregnancy by either an amniocentesis test or a chorionic villus sampling (CVS) test.

Usually, however, the diagnosis is made later. It may be suspected shortly after birth because a baby has an unusually wide neck ("neck webbing"), puffy hands and feet, or occasionally a problem with the heart. Often a girl with Turner Syndrome is not diagnosed until early childhood, because she is not growing as well as expected. Some girls are diagnosed as teenagers when they are taken to the doctor because their periods or puberty seem to be late in starting.

## Features of Turner Syndrome

Turner Syndrome can have effects on many parts of the body. Some of the most common features are listed below. Most girls with Turner Syndrome would have some, but not all, of these things.

- Short stature (not so tall as other girls).
- Widening of the neck ("webbed neck").
- Ovaries that do not function and infertility.
- An underactive thyroid gland.
- A broad chest with widely spaced nipples.
- A heart murmur, sometimes associated with narrowing of the aorta (the main blood vessel that comes out of the heart).

## Aspects of Turner Syndrome

### Growth

Girls with Turner Syndrome tend to be short. Their growth rate may be normal for the first 2 or 3 years before slowing down. There are several ways to try and improve the growth of girls with Turner Syndrome, and a girl will usually need to be referred to a child growth specialist so that her individual needs can be assessed and the treatment options discussed. Although girls with Turner Syndrome do not have growth hormone deficiency, growth hormone is often used to increase their final height.