

# Turner Syndrome

## Information Guide

### 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 boys because they are born with the sex chromosomes XY and girls are girls because they 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 have only one normal X chromosome, they are 100 percent female, although fertility problems in later life are usual.

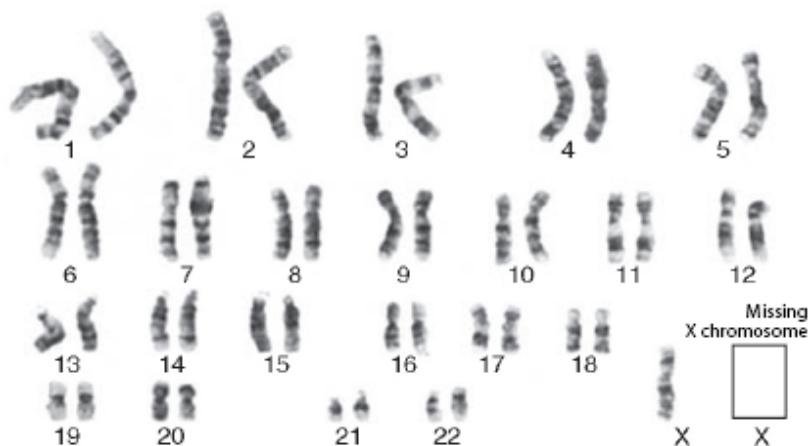
Turner Syndrome is named after Dr Henry Turner who first described it in 1938.

### 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) have been identified as being associated with an increased risk of having a baby with Turner Syndrome.

### Diagnosis and chromosomal findings

The diagnosis of Turner Syndrome is confirmed by chromosomal analysis.



In Turner Syndrome there will usually only be 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 chromosome 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) only some 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 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 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 ('neck webbing')
- 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)
- A difference about the way the kidney is formed

## **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.

### **Developmental progress**

Girls with Turner Syndrome usually have normal intelligence and their progress at school is usually 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 coordination 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 additional information or support**

If you have any further questions about Turner Syndrome, please contact:

**Genetic Medicine**

6<sup>th</sup> Floor, St Mary's Hospital, Oxford Road, Manchester M13 9WL

Telephone 0161 276 6506

Fax 0161 276 6145

Department staffed Monday–Friday, 9.00am to 5.00pm.

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 or 020 8995 0257

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

**The Turner Syndrome Support Society (UK)**

1/8 Irving Court, Hardgate, Clydebank G81 6BA

Telephone 01389 380385 or 01389 872511

Email [turner.syndrome@tss.org.uk](mailto:turner.syndrome@tss.org.uk)

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

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