Introduction

Humans are usually born with 46 chromosomes, which are arranged in 23 pairs. One of each pair of chromosomes comes from our mother in the egg and the other of the pair comes from our father in the sperm.

The chromosomes are numbered from 1 to 22 and the last pair, known as X and Y, are the sex chromosomes as they determine whether the person is a boy or a girl. Boys usually, but not always, have the sex chromosomes XY and girls usually, but not always, have the sex chromosomes XX.

Occasionally a girl is born with only one X chromosome in each cell; this is known as Turner Syndrome. The picture below shows the chromosome pattern of someone with Turner syndrome.

![Chromosome Pattern of Turner Syndrome](image)

Some females have abnormalities in one of their two X chromosomes. This can also lead to Turner Syndrome.

About 1 in 1,800 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.

Mosaic Turner Syndrome

In about half of the cases of Turner Syndrome one X chromosome is missing from or is changed in some, but not all, of the cells of the
body. This is referred to as “Mosaic Turner Syndrome”. Girls with Turner Syndrome in a mosaic form often have fewer features of Turner Syndrome and are more likely to enter puberty naturally.

**What causes Turner syndrome?**

Turner syndrome occurs when there is loss or alteration of an X chromosome during the making of sperm or eggs, or in the early developing embryo. It is not known why this happens. 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.

**When is the diagnosis made?**

Turner Syndrome can be suspected because of features seen on an ultrasound scan performed during pregnancy. A CVS (*chorionic villus sample*) or amniocentesis test can confirm a diagnosis. Turner Syndrome 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 Turner Syndrome is not diagnosed until early childhood when tests may be carried out because a girl is not growing 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 an effect on many parts of the body. It is variable. Most girls with Turner Syndrome would have some, but not all of the following:

- Short stature (*not as tall as other girls*).
- Widening of the neck (“webbing”).
- Ovaries that do not function and infertility.
• 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*).
• Kidney problems - horseshoe kidneys
• Thyroid problems - under activity of the thyroid
• Hearing problems

**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 (endocrinologist) 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, their progress at school is generally good and many go on to university. A small proportion of affected girls may have specific learning difficulties, particularly with mathematics and geometry. Some girls with Turner Syndrome have difficulty with activities involving dexterity, e.g. fine finger movements and co-ordination.

**Puberty**

In the majority of girls with Turner Syndrome, the eggs in the ovaries degenerate and disappear in early childhood and the ovaries stop functioning properly before the age that puberty would normally begin. The ovaries normally produce the sex hormones oestrogen and progesterone which control puberty and the menstrual cycle.
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 rarely able to become pregnant naturally because their ovaries are no longer producing eggs. A very small proportion of young women with Turner Syndrome, who enter puberty naturally without needing hormone treatment, may then be fertile for a short time.

Girls with Turner Syndrome 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). Adult women need monitoring on a regular basis.

**Other sources of information:**

Turner Syndrome Support Society (UK)
13 Simpson Court
11 South Avenue
Clydebank Business Park
Clydebank
Scotland
G81 2NR
www.tss.org.uk

*Factual information presented in this communication is based on accurate contemporaneous peer reviewed literature. Evidence of sources can be provided on request.*