

Growth

Girls with Turner syndrome tend to be short. Their growth rate may be normal for the first 2 or 3 years but then slow down. Girls with Turner Syndrome may need to be referred to a growth specialist and the treatment options discussed. Growth hormone is often used to increase the final height even though they are not growth hormone deficient.

Intelligence

Girls with Turner Syndrome usually have normal intelligence and their progress at school is generally good. The eggs in the ovaries degenerate and disappear in early childhood and the ovaries stop functioning properly well before the normal age of puberty. The ovaries normally produce the sex hormones oestrogen and progesterone.

It is oestrogen that is needed to start puberty. Oestrogen therapy can be given to start puberty. Oestrogen is used to produce breast development and oestrogen and progesterone together help produce regular periods.

Ongoing oestrogen replacement is important for prevention of osteoporosis in later life.

Infertility

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

Although the ovaries of girls with Turner Syndrome do not function they do have a normal womb and vagina. This means they are able to have an entirely normal sex life. Some women with Turner Syndrome have had a successful pregnancy using donated eggs and in vitro fertilisation (IVF).

For more information

You may wish to contact a support group specifically for girls with Turner Syndrome and their families.

The Child Growth Foundation

2 Mayfield Avenue
London, W4 1PW
Telephone: 020 8994 7625/ 020 89950257
Website: www.cgf.org.uk

The Turner Syndrome Support Society (UK)

13 Simpson Court, 11 South Avenue
Clydebank Business Park, Clydebank,
Scotland, G81 2NR
Telephone: 01419528006
Website: www.tss.org.uk

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Birmingham Women's 
NHS Foundation Trust

Turner Syndrome

An information leaflet for
parents and families

If you need more advice about any aspect
of Turner Syndrome please contact:

Clinical Genetics Unit
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NHS Foundation Trust
Mindelsohn Way
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Birmingham
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Turner Syndrome

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

Introduction

Humans are usually born with 46 chromosomes which are arranged in 23 pairs. Our chromosomes are numbered according to size from 1 to 22 and the last pair are the sex chromosomes. It is the sex chromosomes that determine whether a baby is male or female. Boys are born with the sex chromosomes XY and girls are born with two X chromosomes XX.

Occasionally a girl is born with only one X chromosome and this is known as Turner syndrome. The second X chromosome is usually missing but occasionally it may be present but unusual in some way. The X chromosome can be unusual in several ways; there can be some chromosome material missing; it may have formed itself into a ring; or in very rare cases there may be only one chromosome and part of a Y chromosome.

It is estimated that about 1 in 2000 to 1 in 2,500 girls has Turner Syndrome so in Britain there are about 10,000 girls and women who have Turner Syndrome. Even though these girls and women only have one X chromosome they are female but may have fertility problems.

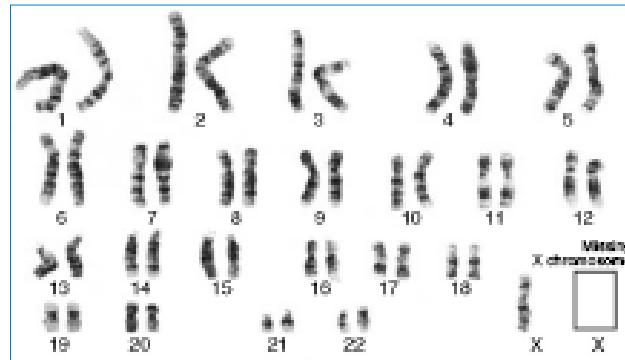
What causes Turner Syndrome?

Sometimes when the egg or sperm are forming there is an error in cell division

which results in an individual with 45 chromosomes instead of 46. In Turner syndrome it is an X chromosome that is missing. This is a chance event and no risk factors such as maternal age or diet during the pregnancy have been associated with an increased risk of having a baby with Turner Syndrome.

Diagnosis

Girls with Turner Syndrome often have a typical appearance and this is often how the diagnosis is first suspected. The diagnosis can be confirmed by looking at the chromosomes. The picture below is an enlarged photograph of chromosomes from a person with Turner syndrome.



When one X chromosome is missing this is called classical Turner Syndrome. Sometimes one X chromosome is missing from some of the cells of the body or is unusual in some of the cells of the body. This is called Mosaic Turner Syndrome. Girls with mosaic Turner Syndrome 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. If Turner Syndrome is suspected it can be confirmed by checking the baby's chromosomes. This can be done during the pregnancy by either an amniocentesis test or a chorionic villus sampling (CVS) test.

Usually the diagnosis is made later. It may be suspected shortly after birth because the baby has a wide neck (neck webbing), puffy hands and feet and occasionally a problem with the heart.

Often the diagnosis is not made until early childhood because a girl is not growing as well as expected. Some girls are not diagnosed until puberty because their periods or puberty seem late in starting.

Features of Turner Syndrome

Turner Syndrome can have effects on many parts of the body. Some common features are:

- Short stature (not so tall as other girls)
- Widening of the neck (webbed neck)
- Ovaries that do not function and infertility.
- A broad chest with widely spaced nipples.
- A heart murmur sometimes associated with a narrowing of the main blood vessel that comes out of the heart called the aorta.
- Sometimes kidney malformations.