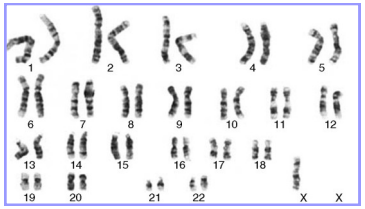
Girls with TS have a normal womb and vagina and will be able to have an entirely normal sex life. Some women with TS have been able to have a child using fertility treatments using donated eggs and in vitro fertilisation (IVF).

What causes Turner Syndrome?

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

In TS there will usually be only one X chromosome, so making 45 chromosomes in total (see below). When one X is missing in all the cells of the body, this is known as 'Classical Turner Syndrome.' The missing X has been lost some time during cell division in the course of sperm or egg production.



Some TS is caused by a rearrangement in one of the two X chromosomes. Where this is identified, a referral to the Clinical Genetics is recommended for specific advice for the pregnancy.

Mosaicism

Sometimes one X chromosome is missing from, or rearranged in, some but not all, of the cells of the body. This is referred to as 'mosaic Turner Syndrome.'

Girls with one missing X chromosome in a mosaic form may have fewer features of Turner Syndrome and are more likely to enter puberty naturally.

If an X chromosome rearrangement is found in some but not all cells, a referral to Clinical Genetics is recommended for specific advice.

For more information:

Clinical Genetics Departments in Scotland

Aberdeen Royal Infirmary. Tel: 01224 552120
Fax: 01224 559390 (Aberdeenshire, Moray, Highland, Western & Northern Isles)

Ninewells Hospital, Dundee. Tel: 01382 632035
Fax: 01382 645731(Tayside, Perth & Kinross, Angus, North East Fife)

Western General Hospital, Edinburgh. Tel: 0131 651 1012 Fax: 0131 651 1013 (Borders, Lothian, South West Fife)

Yorkhill Hospital, Glasgow. Tel: 0141 201 0808
Fax: 0141 201 0361 (Glasgow, Argyll & Bute, Ayrshire, Dumfries & Galloway, Forth Valley)

The Turner Syndrome Support Society (UK)

An organisation ran by and for individuals with TS and their families.
12 Simpson Court, 11 South Avenue
Clydebank Business Park, Glasgow, G81 2NR
Helpline: 0845 2307520 or 0141 952 8006
E-mail: Turner.syndrome@tss.org.uk
Website: www.tss.org.uk

Antenatal Results & Choices (ARC)

An organisation providing non-directive support throughout the antenatal testing process.
73 Charlotte Street
London, W1T 4PN
Helpline: 0207 631 0285
Email: info@arc-uk.org
Website: www.arc-uk.org.uk

NHS Greater Glasgow and Clyde

Scottish Clinical Genetics Forum

Turner Syndrome

Following antenatal diagnosis

This is Emie diagnosed with Turner Syndrome in utero. And Kacey who was diagnosed at birth. Picture reproduced by the kind permission of the Turner Syndrome Support Society UK.

Acknowledgements

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Introduction

Turner Syndrome (TS) is caused by a missing or partially missing X chromosome (see overleaf).

The cause of TS is currently unknown, however, it is not a result of anything that the parents did or did not do during the pregnancy.

Features of Turner Syndrome

Most girls and women born with TS are able to lead **relatively normal, healthy lives**. They will need regular health checks and treatment throughout their lives. There is considerable variation but the commoner characteristics of TS are:

- Not as tall as other girls (4'10'' average adult height without treatment). However, height is often increased by Growth Hormone treatment in childhood.
- About 15-50% have a heart malformation (e.g. a heart murmur sometimes associated with narrowing of the aorta, the main blood vessel that comes out of the heart). Surgical intervention is occasionally required.
- The majority have infertility (due to the ovaries not developing properly).

The Diagnosis of TS in pregnancy

Turner Syndrome is sometimes suspected when an ultrasound scan is performed during pregnancy. This may because lymphoedema (fluid accumulation in the tissues) is identified. The diagnosis of TS can be confirmed by an amniocentesis test or a chorionic villus sampling (CVS) test.

Lymphoedema (fluid accumulation) may be mild or serious and may impact on whether the pregnancy survives in the womb. Overall, most pregnancies with TS will be lost naturally. However, lymphoedema may reduce should the pregnancy become more advanced and more active in the womb. The Obstetrician can advise about the chances in each specific pregnancy and about options.

Decisions in pregnancy are individual and people should not be pressured into a decision. Both Obstetricians and the Clinical Genetics service can offer non-directive information and support, specific for each pregnancy.

A referral to Clinical Genetics can help to explain the implications and options for the pregnancy, as well as whether it could happen in future pregnancies.

The organisations 'Antenatal Results and Choices' and the 'Turner Syndrome Support Society' can also offer information and support to parents facing pregnancy decisions. Contact details for these organisations are overleaf.

Living with Turner Syndrome

About 1 in 2500 girls are born has TS. In Britain, it is estimated that there are about 10,000 girls and women who have TS. TS was first described by Dr Henry Turner in 1938.

Every person with TS is individual and most will have some and NOT ALL of the main features.

Growth

The growth rate of girls with TS may be normal for the first 3 or 4 years before slowing down. 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.

Developmental progress

Girls with TS usually have normal intelligence and their progress at school is generally good. Some 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

The ovaries normally produce the sex hormones, oestrogen and progesterone. 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 TS 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.

Although treatment normally allows girls with TS to enter puberty alongside girls of their own age, they may benefit from support for issues of low-self esteem and shyness.

Infertility

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