

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 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)
- 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 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 eg, 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.

Translation and interpretation service

Do you have difficulty speaking or understanding English ?

আপনি কি বঙ্গভাষীতে বুঝতে কিংবা বুঝাতে পারছেন ? (BENGALI)

क्या आपको अंग्रेजी बोलने या समझने में कठिनाई है ? (HINDI)

તમે ભાષા કારણે વાતચીત કરવામાં મુશ્કેલી આવે છે ? (GUJARATI)

ਕਿ ਤੁਹਾਨੂੰ ਅੰਗ੍ਰੇਜ਼ੀ ਬੋਲਣ ਜਾਂ ਸਮਝਣ ਵਿਚ ਦਿੱਕਤ ਹੈ ? (PUNJABI)

Miyey ku adagtahay inaad ku hadasho Ingriisida aad sahamto (SOMALI)

هل لديك مشاكل في فهم اوالتكلم باللغة الانجليزية ؟ (ARABIC)

你有困難講英語或明白英語嗎？ (CANTONESE)

کیا آپ کو انگریزی سمجھنے اور سمجھانے میں دقت پیش آتی ہے؟ (URDU)

☎ Telephone Number 0161 276 6202/6342

This information sheet was compiled from a number of sources including the booklet 'Turner Syndrome: A guide for patients and parents', produced by the Child Growth Foundation. Thanks also to the Kennedy-Galton Centre.

We would like to acknowledge our Clinical Genetics colleagues at Guy's and St Thomas' Hospital NHS Trust who originally designed and wrote this leaflet.

Please let us know if you would like this leaflet in another format (eg. large print, Braille, audio).

Seen in clinic by(doctor)

and(genetic counsellor)

Telephone number

Family reference number

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Turner Syndrome

Information for parents
and families



INVESTOR IN PEOPLE



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 per cent female, however, 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.

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.cgf.org.uk

The Turner Syndrome Support Society (UK)

1/8, Irving Court
Hardgate
Clydebank G81 6BA
Telephone: 01389 380385 or 01389 872511
E-mail: turner.syndrome@tss.org.uk
Website: www.tss.org.uk

Suggestions, concerns and complaints

If you wish to make a comment, have a concern or want to complain, it is best in the first instance to speak to the manager of the ward or department involved.

The Trust has a Patient Advice and Liaison Service (PALS) who can be contacted on (0161) 276 8686 and via e-mail: pals@cmmc.nhs.uk. They will help you if you have a concern, want advice or wish to make a comment or complaint.

Information leaflets about the service are readily available throughout the Trust. Please ask any member of staff for a copy.

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

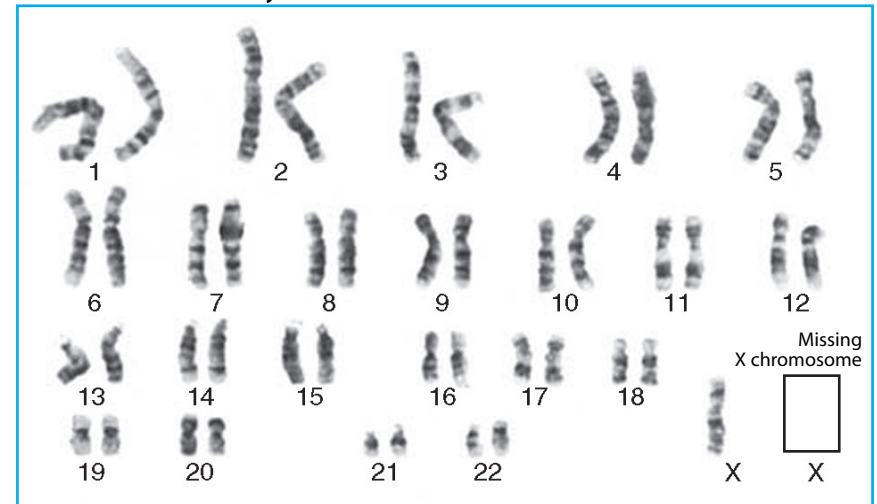
If you need more advice about any aspect of Turner Syndrome, you are welcome to contact:

Genetic Medicine
Sixth Floor
Saint Mary's Hospital
Oxford Road
Manchester, M13 9WL
Telephone: 0161 276 6506 Reception
Facsimile: 0161 276 6145

Department staffed Monday - Friday 9.00 am to 5.00 pm

Diagnosis and chromosomal findings

The diagnosis of Turner Syndrome is confirmed by chromosomal 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) 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.