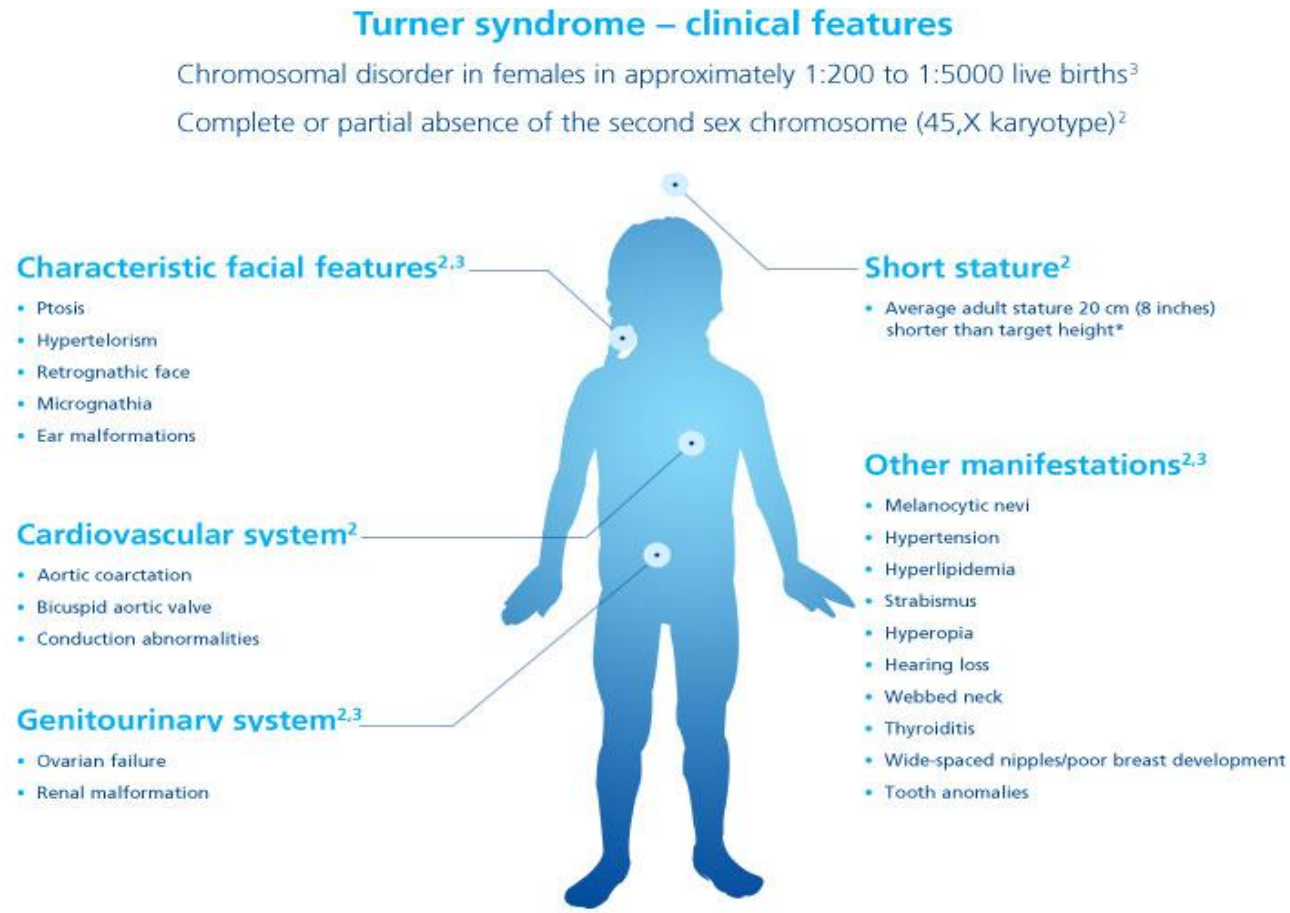
NHS
Southport and Ormskirk Hospital
NHS Trust

PATIENT INFORMATION

Turner Syndrome

What is Turner Syndrome?

Turner Syndrome is a chromosomal condition where there is a lack of, or abnormality of, the second X chromosome (45 XO). It affects only females. Turner Syndrome is usually characterised by short stature and non-functioning ovaries which cause the absence of sexual development and infertility. Despite the poor or absent ovarian function, other sexual and reproductive organs (uterus and vagina) are normal. The physical features associated with Turner Syndrome are shown below.



It must be emphasised that some girls may have only one or two mild features of the syndrome while others may have several that are easily recognized. This is because the X chromosome is missing in some cells but not others, a

condition referred to as "Turner mosaicism". The cause of the missing or damaged X chromosome in Turner Syndrome is not known and there is also no known increased risk of recurrence in any future pregnancy.

How is Turner Syndrome Diagnosed?

Although the diagnosis is initially based on the characteristic physical signs, it must be confirmed by genetic analysis. Normally, each cell has 23 pairs of chromosomes, making a total of 46 chromosomes. One of these pairs, the sex chromosomes, determines the gender of the foetus; i.e. Will the baby by a boy or a girl? In a boy the sex chromosomes will be an X and Y (46XY) whereas in girls there are two X chromosomes (46XX). In Turner Syndrome there may be a completely or partly missing X chromosome in some or all cells of the body, making 45 chromosomes (45XO, the O representing the missing chromosome). In some girls with Turner Syndrome, only a proportion of their cells have this abnormality. This is known as “Turner mosaicism”. In a small proportion (about 1%) of girls with Turner Syndrome, a particle of a Y chromosome is sometimes found. This tiny fragment causes a significant increase in the risk of developing a tumour of her ovary. If a particle of a Y chromosome is identified, the ovaries should be removed.

When is Turner Syndrome Diagnosed?

Diagnosis is possible at birth, or even before the baby is born by a process called amniocenteses or chorionic villus sampling. These tests allow examination of the chromosomes of the foetus in the womb. Often, a girl with Turner Syndrome is not diagnosed until early childhood when growth progressively slows down. Diagnosis may

sometimes be even later when puberty fails to occur or slows and stops during teenage years.

Problems in Infancy

Baby girls with Turner Syndrome may have puffy hands and feet when they are born. This is probably due to poor development of part of the lymphatic system in these areas, which drains away body fluids through small vessels under the skin. This puffiness usually disappears soon after birth, although in some girls it can remain or recur at the time of puberty. Some girls with Turner Syndrome may be born with a heart defect which may require surgery during infancy. There may be difficulties with sleeping patterns. Problems may also arise with some babies because of poor sucking and difficulties with chewing and swallowing. A speech therapist can be of great support with feeding problems. Early feeding problems are very common but do improve and do not lead to any serious disorders.

Problems in Childhood

Hearing and Vision

Middle-ear infections are common and hearing can be impaired. As the girls progress to pre-school age, recurrent ear infections may become troublesome and some may require grommets, which are small tubes inserted into the eardrum to drain fluid away from the middle ear. Hearing tests need to be done regularly to check for any hearing loss. Eyes need to be tested for short-sightedness, squints and ptosis (drooping eyelids).

Growth

Short stature is the most common feature in Turner Syndrome. Girls with Turner Syndrome are often small at birth but most retain a normal growth rate until any time for 3–7 years, when growth slows. The average height of a woman with untreated Turner Syndrome is around 147cm, but this also depends on the height of her parents. The cause of poor growth in Turner Syndrome is due to several factors including poor growth in the womb, the absence of the growth spurt at puberty in the untreated girl and possible skeletal abnormalities. Girls with Turner Syndrome usually have normal levels of growth hormone however, treatment with growth hormone to improve growth and height is usually given in childhood. The final height can be increased by approximately 5-7cm with the use of growth hormone.

Schooling & Development

Intelligence in girls with Turner Syndrome falls across the normal range and there is no increase of intellectual disability. Progress at school is generally good and there are many girls who excel, although some do have specific learning difficulties. Reading age is often advanced, whereas writing age is sometimes delayed. Difficulties experienced with spatial skills in some girls may result in particular difficulties with mathematics and geometry. Some girls with Turner Syndrome have difficulties with activities involving dexterity and co-ordination such as catching a ball.

<u>Problems During Adolescence</u>

The adolescent years can be a difficult time and apart from maybe being smaller than their friends, other emotional problems may arise**.** A main characteristic of Turner

Syndrome is the failure of ovaries to function properly and therefore the failure of sexual development in these girls. Normally, the ovaries perform two functions; storage of eggs and production of the female sex hormones – oestrogen and progesterone. Oestrogen is the hormone required for feminising a girl at puberty and maintaining feminisation throughout life. It is also necessary to build and maintain bone strength and to create a healthy profile of cholesterol and other fats in the blood. In girls with Turner Syndrome, the number of eggs in the ovaries gradually diminishes during childhood and the ovaries usually stop functioning properly well before the age that puberty would normally begin. Without replacement oestrogen therapy, puberty will either not occur or may start with a small amount of breast development, but then just stops. At the appropriate age for starting puberty, treatment with oestrogen will initiate breast development and later, with a treatment combination of oestrogen and progestogen, regular withdrawal bleeds or 'periods' occur. Up to 30–40% of girls with Turner Syndrome enter puberty spontaneously, 4% achieve menarche (onset of periods) and 1% are spontaneously fertile. In those women with Turner Syndrome who do start periods spontaneously, the ovaries are likely to stop functioning in early adult life. Infertility is a common problem in women with Turner Syndrome due to the non-functioning ovaries.

Treatments Available to Increase Growth Rate and Final Height

Growth hormone is the main treatment for girls with Turner Syndrome to increase the rate of the growth and final height. Even though their levels of growth hormone are normal, additional doses of growth hormone are needed to improve both the rate of growth and final height. Although it is known that treatment with growth hormone does improve

final height, probably as much as 7cm, it is not possible to accurately predict the final height of each individual girl.

Growth hormone has been used for many years with great success in the treatment of children with short stature for various reasons. Biosynthetic growth hormone is manufactured using gene technology and is identical to the growth hormone we produce naturally. The dosage of growth hormone varies according to weight and surface area and will increase as she grows. It is given by an injection once daily. The injection is best given before bedtime to mimic the natural production of growth hormone as closely as possible. Treatment is given until the bones are close to becoming solid.

Like all medicines, growth hormone therapy may occasionally cause unwanted side effects, such as skin reaction at the injection site and less often, headaches, swelling of the arms/legs, visual problems and limping. Children with bone disorders (e.g. hip problems, scoliosis) need to be closely monitored, because rapid growth can aggravate these problems. If you are concerned that your child may be experiencing side effects as a result of growth hormone therapy, you should contact your doctor as soon as possible.

Treatments Available to Initiate Puberty

When there is no ovarian function, puberty will only occur if replacement oestrogen therapy is given. Treatment with the female sex hormone, oestrogen is given to initiate puberty in girls with Turner Syndrome. Treatment should start at a time appropriate for the individual, i.e. it may be necessary to delay puberty for 1–2 years to achieve extra height using growth hormone. However, it is now generally accepted that

commencement of oestrogen should not be delayed beyond 13.5 years where possible as there is no advantage to very late use of oestrogen and height is not increased by such delay. The administration of oestrogen will produce all the female sexual characteristics, such as breast development, change in body shape, maintenance of pubic hair and the associated psychological changes of puberty.

A starting dose of natural oestrogen (Progynova) is usually around 0.5 mg every second day increasing the dose every 6 months to an adult dose of 2mg per day. For the first year or two, only oestrogen treatment is given (that is, oestrogen not combined with another hormone, progestogen). After the dose of oestrogen has been built up to a level sufficient for the first period to occur, or after 24-36 months of oestrogen therapy, progestogen is added (Medroxyprogesterone acetate) for 12-14 days each month. This can also be given every second or third month but 12–14 days of progestogen is necessary for adequate removal of the womb lining (as a period) to keep the uterus healthy. Usually the dose of oestrogen is increased to adult replacement levels over 2½–3 years. At that time it can be made more simple by the use of a pre-packaged oestrogen and progestogen preparation which delivers oestrogen every day with cycles of progestogen to ensure that a period occurs regularly. It is important to have a withdrawal bleed in order to shed the lining of the uterus (womb) and so keep the uterus healthy.

The long term replacement with a ‘natural’ oestrogen without an ‘ethinyl’ component is preferable (eg. Qlaira), as it greatly lowers the risk of high blood pressure. Apart from sexual development, oestrogen is essential for a young woman as nearly 50% of a girl’s bone mass and mineral strength is built for life during puberty. If oestrogen is stopped, osteoporosis will occur, the skin and muscle age

quickly and early heart disease risk is increased. Oestrogen is also important for building psychological good health and confidence. It causes brain maturation in teenage girls and is very important in the social development of a young woman.

Infertility

Due to the non-functioning ovaries, very few women with Turner Syndrome are able to have children without medical help despite having regular 'periods' from cyclical oestrogen/progestogen therapy. However, as women with Turner Syndrome do have a normal uterus it is possible to have a child through egg donation and fertility treatment. An egg can be donated from another woman and fertilised with their partner's sperm. This can take place during the technique of in-vitro fertilisation (IVF) whereby the egg and sperm are put together in a test tube and fertilised. Another method of assisted conception is Gamete Intra-Fallopian Transfer (GIFT). In this technique, the egg and sperm sample are transferred into one of the fallopian tubes to allow fertilisation to take place naturally within the fallopian tube. Successful IVF/GIFT pregnancies have now been reported in many women with Turner Syndrome. During pregnancy the placenta will produce oestrogen and therefore additional hormone treatment will not be required during the pregnancy, after the placenta is established.

Blood Pressure

Women with Turner Syndrome are at risk of developing hypertension (high blood pressure). Blood pressure should be measured regularly, particular during oestrogen therapy. There are several reasons why blood pressure in Turner Syndrome may rise including the possibility of a coarctation

of the aorta (constriction in the large blood vessel leaving the heart), as well as problems with the kidneys or in the blood vessels within the kidney. High blood pressure can result from the use of ‘ethinyl’ oestradiol as found in the oral contraceptive pill. If this occurs, medication should be changed to a non ethinyl containing oestrogen. This change should significantly lower blood pressure, but it may be necessary to additionally treat the high blood pressure with other medications. However, it is very important that a woman with Turner Syndrome receive adequate oestrogen for all the reasons we have mentioned above, so oestrogen should be continued even if blood pressure needs special management.

Heart Problems

Approximately 30–40% of girls with Turner Syndrome have some type of heart problem, most commonly a bicuspid aortic valve. For some of these problems, antibiotics may need to be given when any dental procedure or surgery occurs, to prevent bacteria landing on the abnormal valve. The specialist will advise when this care is needed.

Osteoporosis

Osteoporosis is characterised by reduction in the amount of bone thickness and hence the development of thin, brittle bones and an increase in bone fractures. It is now known that the main bones mass, and thereby strength of the bones, is made during adolescence through the action of the sex hormones (oestrogen and progestogen). Thus, any condition in which the ovaries do not produce enough oestrogens, as occurs in Turner Syndrome, makes the development of osteoporosis more likely. In addition, in Turner Syndrome it has been reported that bones may

possibly be more likely to develop osteoporosis as part of the underlying condition. A good calcium intake, maintenance of normal Vitamin D levels together with regular use of hormone replacement therapy vastly decreases the long term risk of osteoporosis. It is important that women with Turner Syndrome continue with oestrogen/progestogen treatment throughout life until the expected age of menopause. Risks versus benefits will need to be discussed with the specialist.

Autoimmune disorders

Due to a generally increased lifetime risk for autoimmune disorders, routine tests for these problems are usually undertaken every 1–2 years.

Coeliac Disease

5–10% of girls and women with Turner Syndrome may develop this condition, which is a disorder of poor food absorption due to gluten intolerance. A blood test will give an indication if it is present and the specialist will arrange treatment.

Glucose Intolerance

There is an increased risk of women with Turner Syndrome developing diabetes mellitus and it is therefore recommended that glucose levels are monitored annually.

Hypothyroidism

There is an increased risk of women with Turner Syndrome developing an underactive thyroid gland. It is therefore recommended that hormone levels are monitored annually.

Inflammatory bowel disease

Inflammatory bowel disease occurs more commonly in Turner Syndrome. This should be tested if there is chronic abdominal pain, diaorrhoea or bleeding from the bowel.

During your contact with us, it is important that you are happy with your care and treatment. Please speak to a member of staff and/or the ward/department Sister/Charge Nurse if you have any questions or concerns.

MATRON

A Matron is also available during the hours of 9.00 to 5.00 pm Monday to Friday. During these periods, ward/department staff can contact Matron to arrange to meet with you. Out of hours, a Senior Nurse can be contacted via the ward/department to deal with any concerns you may have.

INFECTION CONTROL REQUEST

Preventing infections is a crucial part of our patients' care. To ensure that our standards remain high our staff have regular infection prevention and control training and their practice is monitored in the workplace. We ask patients and visitors to assist us in preventing infections by cleaning their hands at regular intervals and informing staff of areas within the hospital that appear soiled.

As a patient there may be times that you are unsure whether a staff member has cleaned their hands; if in doubt please ask the staff member and they will be only too happy to put your mind at ease by cleaning their hands so that you can see them.

SPECIAL INSTRUCTIONS

None

ANY CONDITION SPECIFIC DANGER SIGNALS TO LOOK OUT FOR

CONTACT INFORMATION:

Your own GP –
Children's Ward – 01695 656912/656612

OTHER USEFUL TELEPHONE NUMBERS/CONTACTS:

NHS 111
Stop Smoking Helpline (Sefton) - 0300 100 1000
Stop Smoking Helpline (West Lancashire) - 0800 328 6297

Please call 01704 704714 if you need this leaflet in an alternative format

Southport and Ormskirk Hospital NHS Trust

Ormskirk & District General Hospital
Wigan Road, Ormskirk, L39 2AZ
Tel: (01695) 577111

Southport & Formby District General Hospital
Town Lane, Kew, Southport, PR8 6PN
Tel: (01704) 547471

FOR APPOINTMENTS

Telephone (01695) 656680
Email soh-tr.appointments@nhs.net

Please remember to complete the **attached** *Friends and Family Test*.
Alternatively, you can complete the *Friends and Family Test* on-line by going to:
southportandormskirk.nhs.uk/FFT
Thank you

Author: Dr May Ng, Consultant Paediatric Endocrinologist
Ref: 13/56
Version: 3
Reviewed: March 2022
Next Review: March 2025