A Teenager's Guide to Turner Syndrome

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Introduction

You may have recently been told that you have Turner Syndrome (TS), or you may have known this for a while. In either case, you have probably wondered what the future will hold...how TS may affect your life as a teenager and as an adult. There is every reason to feel positive about your future – with modern medical help, your life should be very similar to other girls and women, in most ways. You should enjoy good health, happiness and a normal lifespan.

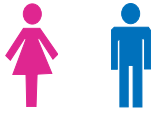
However, there are some aspects of your life that will be different because of TS, and this booklet will help you to understand these issues, and understand ways of dealing with challenges you may meet along the way.

We hope this guide will help you to feel positive and confident about life with TS. Armed with this information, we are sure you can achieve your potential and live life to the full.

What is Turner Syndrome?

Turner Syndrome (TS) is a genetic disorder associated with low growth rate. Without treatment, the most obvious sign of TS is being unusually short *(however, with modern treatment, girls with TS can experience an increased growth rate and an improved final height)*. TS is caused by lack of information from the X chromosome, and it only affects females.

To explain this in a bit more detail, the chromosomes are the parts of our cells that 'map out' our individual characteristics - everything from hair colour to height and physical type.



X X X Y

There are 46 human chromosomes, including the sex chromosomes - called **X** and **Y**. The sex chromosomes determine your gender - if you have an **X** and a **Y** chromosome you will be **male**; if you have two **XX** chromosomes you will be **female**.

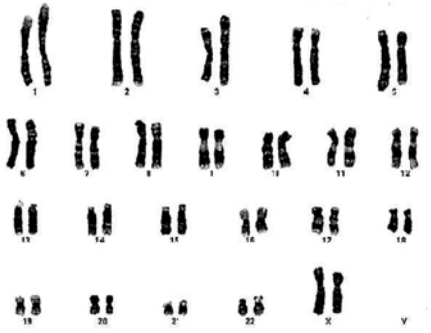
In TS part or all of an X chromosome is missing or altered. The sex chromosomes also carry genes that code for other physical characteristics. Loss of part or all of an X chromosome can therefore lead to a number of different physical characteristics – short stature being the most obvious.

TS is a highly variable syndrome, affecting different people in different ways - so don't be overwhelmed by the list of potential physical characteristics described in this booklet - in some cases, TS is mild, and it even remains undiagnosed in many people.

In addition, with modern medicine many aspects of TS can be effectively treated if necessary, particularly if a preventive approach is taken. Probably one of the most difficult aspects of TS is infertility, but these days, techniques such as in vitro fertilisation (IVF) have allowed many TS women to become mothers.

So in summary, although it's important to be aware of potential symptoms, don't be alarmed because with proper treatment, Turner Syndrome is a manageable condition that is just one aspect of an otherwise healthy life.

Unaffected human chromosomes



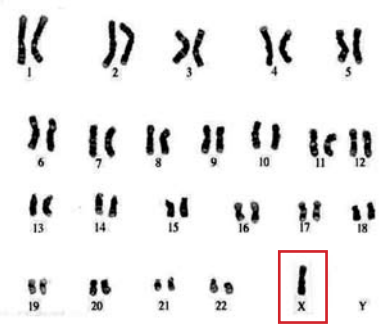
Diagnosis and its effects

TS can be diagnosed at any age - from birth to adulthood. You may have been diagnosed during early childhood, if your growth slowed down, and your height was obviously less than that of your friends. It is better to diagnose TS early, because the earlier that treatment can be started, the better the outcome of treatment may be. The diagnosis can be made for different reasons - you may have been diagnosed with TS when you were a baby if you had difficulties with feeding, or puffy hands and feet. Or you may not have been diagnosed until your teens, when you wondered why your periods hadn't started. *(See page 5 for a full list of symptoms)*.

To confirm a suspected diagnosis of TS, a genetic test is performed. This is because, as mentioned earlier, TS is a genetic disorder, caused by loss of all or part of one of the X chromosomes. This can usually be detected using a blood test. In some cases this may not be conclusive, and skin is tested using a 'scratch' test.

To understand the genetics in a bit more detail, first consider the standard human chromosomes. A female has 46 chromosomes, including 2 X chromosomes *(the sex chromosome that confers gender, among other things)*. This is written as 46 XX. In 'classical' TS, the X chromosome is missing in all cells - this is written as 45X0 *(this means that there are 45 chromosomes, rather than 46, and only one X chromosome is present, rather than two)*. In some cases of TS, the X chromosome is not actually missing, but is altered in some way.

TS chromosomes



There is also another variant of TS called 'mosaicism' where the X chromosome is only missing in some cells – this is described as Turner Mosaicism *(written as 46XX/45X0)*. In this case, the physical characteristics of TS may be quite minor, as fewer cells are affected.

The genetics of TS are quite complex and hard to take in at a time when you may still be feeling shocked by the diagnosis. Don't hesitate to ask your parents for more information, or speak to your specialist or GP if there is anything that is not clear.

Medical characteristics of Turner Syndrome

Before looking at some of the possible features of TS *(described below)*, it is important to stress again that although you have TS you can expect to live a long and healthy life. You may have only a few of the features, and medical treatments can often be used to alleviate these. The overall outlook for those with TS is good. BUT an awareness of possible symptoms allows you to take a preventive approach and be aware of how things may develop. The term 'syndrome' may sound scary but it is just a collection of features that can point to the diagnosis.

The two main clinical features of TS are:

- Short stature
- Non-functioning ovaries *(associated with lack of sexual development and infertility)*

Other features that MAY occur with TS

*(**Please note:** girls with TS may only have a few of them)*

- Puffy hands and feet
- Broad chest and widely spaced nipples
- Droopy eyelids
- Low hairline
- Low-set ears
- Short fourth toe and short fingers
- Soft fingernails that turn up at the end
- Arms that turn out slightly at the elbows
- Lots of small brown moles on the skin
- Webbing of the skin of the neck
- Small jaw and narrow, high-arched palate, which may cause feeding difficulties or dental complications

Growth and shot stature

You may have been slightly small at birth, and by the age of 2-3 years, your growth rate probably slowed down - you have probably always been one of the smallest in your class. You may well have been diagnosed with TS in early childhood and started on growth hormone therapy. This will have increased your growth rate. You may take longer than your friends to reach full adult height. However, with hormone treatment, you should reach close to your potential height, which will be linked to the heights of both of your parents.

Puberty and fertility

In TS the ovaries do not function properly, and do not produce the hormones that cause puberty. But with help from hormone treatment, you will be able to go through puberty, develop a womanly shape and start having periods. Although some girls with TS (about 20%) start puberty on their own it will not progress normally without the help of hormone treatment.

Because your ovaries do not work in the normal way, it is unlikely that you could conceive a child naturally (less than 1% of women with TS do so). The good news though is that your other reproductive organs are not affected by TS - most importantly your uterus is normal and able to nurture a growing baby. With the help of hormone treatment you can enjoy a normal sex life, and there are ways in which you may be able to have babies.

Osteoporosis

As well as being involved in fertility and sexual maturation, the hormone oestrogen is also involved in maintaining healthy bones - this is why women after the menopause are more prone to osteoporosis - weakening of the bones, and potential fracturing. In TS, the risk of osteoporosis arises early, because of the lack of oestrogen throughout adulthood. Therefore, a preventive approach to osteoporosis is recommended earlier than in others. Hormone therapy will mimic the natural production of oestrogen, and help to maintain healthy bones. Exercise and maintaining a good intake of dietary calcium can help.

Hearing

Ear infections are common in childhood generally, but particularly so in girls with TS, and it is important to keep an eye on your hearing - tell your parents if you think you have problems hearing, particularly in the classroom. Grommets (small tubes) may need to be inserted to help drainage within the ear. Again, it is a matter of taking a preventive approach and tackling this issue at an early stage. On the positive side, ear infections do not usually continue in adulthood.

Heart

For the majority of girls with TS, the heart is normal. But about 10% of girls with TS are born with a condition called coarctation of the aorta. This means an abnormal narrowing of the main blood vessel out of the heart. Other girls have a minor abnormality of one of the heart valves. Fortunately, in most cases these conditions are minor - but some cases may need surgery. To be on the safe side, it is advisable for all girls who are diagnosed with TS to have their heart examined by a cardiologist.

Kidneys

Kidney disease is more common in TS than in the general population. This is partly because the shape of the kidney may be slightly altered (e.g. horseshoe kidney), and partly because of a tendency to increased blood pressure.
A preventive approach can be taken by paying careful attention to blood pressure. For many girls, their kidneys will not be a major issue - maybe just occasional treatment may be needed to clear a kidney infection.

Thyroid

The thyroid is a small gland in the neck which produces a hormone called thyroxine. This is important for general wellbeing and to maintain good energy levels. A minority of girls with TS have an underactive thyroid (only about 10% of those with TS). This can occur at any age, so regular testing is recommended, using a simple blood test. An underactive thyroid can cause lethargy and weight gain, but fortunately this is very simply treated with a tablet of thyroxine - this completely restores the body's thyroid function.

Blood pressure

High blood pressure is more common in people with TS than in the general population so blood pressure should be checked and monitored regularly. Some forms of Hormone Replacement Therapy (HRT) can also contribute to high blood pressure and weight gain. Luckily this is an area where it is easy to start to make a difference - by maintaining a healthy weight through exercise and good diet - this will also benefit overall health and wellbeing.

Coeliac disease

There is an increased chance of girls with TS developing coeliac disease (an intolerance to gluten in the diet). This can cause symptoms such as bloating or diarrhoea or may be asymptomatic. It can be detected by a blood test. Treatment is with a modified diet.

Feet

Some girls with TS get swollen feet. For this reason, it is important that girls with TS pay good attention to footwear.

- Always have both feet measured for length and width
- Also make sure the girth of the shoe is correct (the measurement around the foot)
- Choose firmly-fitting shoes - not slip ons, but laces, straps or Velcro
- Keep heels low - less than 4cm and wide rather than spiky (no stilettos!)
- Choose leather if possible for healthy ventilation
- Ensure socks and tights are well fitting and comfortable

Treatment approaches

Growth hormone

Growth hormone has dramatically changed the outlook for those with TS. Bearing in mind that the average final height of a TS woman without growth hormone was 4' 8" *(142 cm)*, most girls with TS will grow faster with growth hormone, and many reach a normal adult height *(often over 5 feet)*, with the biggest height increase occurring during the first 12 months of treatment. As you may have experienced, this 'growth spurt' is often welcomed with great delight by children who have coped with the experience of being unusually short. Although increased height is not a medical need, the effect of these extra inches is very valuable to most girls. The final height that you will reach will depend on a number of factors, including the time of starting therapy, and also your other height genes inherited from both parents.

Hormone Replacement Therapy (HRT)

Most girls with TS can now experience a normal puberty, thanks to the introduction of HRT. This simply means taking the sex hormones, oestrogen and progesterone, to promote breast development, menstruation etc. The decision about the best age to start is dependent on individual factors, but is often around the age of 12 years. Treatment with oestrogen is gradually increased over 2-3 years (this mirrors the natural situation and promotes a normal rate of development). Towards the end of puberty, progesterone is added to the oestrogen treatment - again this reflects the normal hormone production from a functioning ovary.

Living with Turner Syndrome

Again it is important to emphasise that life is different for each girl with TS - some have more challenges to face than others. If you ever have any concerns about your condition and its impact on your life, make sure you talk about it, whether to your parents, a teacher, your GP or health visitor. The Turner Syndrome Support Society (TSSS) is also a great source of information and support - check out their website on www.tss.org.uk.

School and educational issues

Although many girls with TS do really well at school, there are some who do have specific difficulties (in general, girls with TS have average intelligence). For instance, as mentioned, hearing difficulties can occur - if so, you and your parents must make sure your teachers are aware of the situation.

You may need help with maths or writing, but this varies very much from individual to individual. On the positive side, many girls with TS are very good at reading - often ahead of their peers. Take a look on the TSSS site for success stories - it is encouraging to read about women with TS who have happy successful lives, many with university educations.

Being small for your age may also raise issues at school - for instance some types of PE may be difficult for you - always tell the teacher if you find an activity too challenging, particularly ball games such as tennis and squash. On the other hand, many girls with TS are good at gymnastics - where being small and compact can be an advantage!

Another aspect of being short is that being slightly 'different' may make you a target for bullying. Always talk to your parents and teachers if this happens. Most schools have systems in place to control bullying - so make use of the 'buddy' system if your school has one - try to find an older mentor who will step in and stop unacceptable behaviour. It's hard to do but it really is best to ignore bullies and not let them see you are upset. Stick together with your friends - bullies are not worth bothering with.

The years ahead - what to expect as an adult

Again, once you become an adult, it is important that you continue to receive care and health checks that are recommended for those with TS. The points below summarise some of the key issues to think about - this is based on information put together by the Turner Syndrome Support Society (TSSS), and more details can be found on their website.

Ideally women with TS should be under the care of a specialist TS clinic with a multi-disciplinary team equipped to manage the specific medical issues associated with the syndrome. GPs cannot be expected to be experts on all the latest developments in TS treatment.

One of the most important aspects of your health as a woman with TS is oestrogen deficiency, as mentioned earlier. Oestrogen replacement is important from an early age for gynaecological health and prevention of osteoporosis. Some women with TS may be prone to a number of specific medical conditions as well as cardiovascular disease. These can include osteoporosis, thyroid, gastrointestinal and kidney disorders, hearing difficulties and diabetes.

Quality of life in women with TS may be improved with access to better care as adults. Most women with TS require long-term follow-up, and early treatment will ensure they have the best possible quality of life, and that their life expectancy is as good as for women without TS. In many cases, relatively simple preventative measures will address these issues.

Prevention is better than cure - Health checks for an adult with TS

The first clinic visit may include an up-to-date genetic assessment, as modern technology enables more precise testing than previously. There may also be a number of other health checks, and these tests will probably be repeated on a regular basis.

An echocardiograph (heart), bone density scan (DEXA) and audiogram (hearing test) should be repeated every 3 - 5 years.

Women with TS considering an assisted pregnancy should have a full examination first. Because high blood pressure is more common in women with TS this should be monitored and actively treated. The minority of women with TS who are able to consider natural pregnancy should have access to genetic counselling because of a number of possible problems. Ideally women with TS should be cared for in a specialist centre.

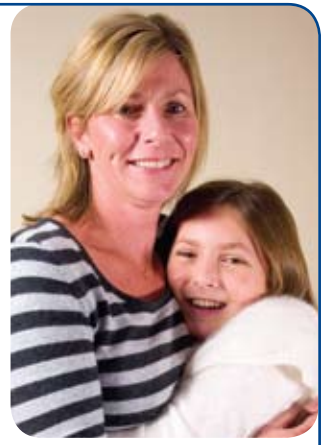
Other possible issues to be considered:

- Genetic counselling - all women with TS should be offered genetic counselling - only their parents may have had the benefit of genetic counselling in the past.
- Psychotherapy - some women with TS benefit from sessions with a psychotherapist/psychologist for help with problems of self-esteem.
- Inflammatory bowel disease (IBD) and coeliac disease - it would appear that there is an increased incidence of both inflammatory bowel disease and coeliac disease in girls and women with TS. The reasons for this are unclear and research continues in this area. Any woman with TS who has unexplained diarrhoea or rectal bleeding should be referred to a gastroenterologist to rule out a diagnosis of ulcerative colitis, Crohn's disease or coeliac disease. Coeliac disease should be screened every 2-3 years
- Girls and women with TS are particularly at risk from in-growing toenails as they often have increased transverse curvature of the nails. Referral to a podiatrist is recommended to prevent infection.
- Swelling of one or more limbs is another area where women with TS often do not currently receive appropriate treatment. Referral to a lymphoedema nurse specialist may be beneficial.

Our stories

Although the experience of each person with TS is different, it can be helpful to hear the stories of others with TS - the ups and downs, and things that have helped them. Below are two stories which are featured in more detail on the TSSS website.

Katie's Mum



In May of 2002, Katie who was nine wanted to go on a school trip for a week. She had a slight problem with bedwetting. I arranged a trip to the doctors for some tablets to help this. She advised us to see a paediatrician 'just as a precaution'.

We had no idea that Katie had Turners, and although we thought she was small, so was her dad and uncle at that age, and her best friends were no taller.

As soon as we walked into our local hospital and met with the paediatrician, he said "don't worry about the bed wetting, she'll grow out of that, she's too small." He ran a test for Turners straight away and while we awaited the results, we looked it up on the internet and spoke to the Child Growth Foundation. We immediately knew that Katie had Turners as we were able to tick many of the characteristics. The test results confirmed it, and our lives were turned upside down, or so we thought.

She started on growth hormone almost immediately, which she has injected herself right from day 1. We were very lucky as she seemed to take things in her stride, and her interest made it easier to explain. We were honest with her from day one and still are. She couldn't go to all the meetings at hospital, and meet with other girls with Turners, if she didn't know the full facts.

That was four years ago. Katie has blossomed into a beautiful happy young lady, (now thirteen). While she struggles sometimes socially, she has made some good friends and is doing well at school. I am extremely proud of her for many reasons, but the main one is the way she just gets on with it. Turners hasn't stopped Katie doing anything, from triathlon to trekking though the Thailand jungles. She'll give almost anything a go.

We have had tremendous support from the TSSS. We have both found comfort from talking to other parents/girls with the condition. We have both made some wonderful friends, with whom we shall be friends for a long time. Support is a must. No one really understands what you're going through, only someone in the same situation. All Turner girls are different and each girl has her own individual problems, but they all share the same caring loving nature that I wouldn't want to change for the world!

Katie's story

In July 2002 I was diagnosed with Turner Syndrome. I was nine years old and I had been to the doctors with a bed wetting problem. I remember the doctor saying that it didn't matter about my bed wetting problem and I would grow out of that soon enough but I was too small. That's all I can remember until my sister and I were going to summer camp in Wales.

We went off all happy and laughing. The week we were away was great. When my mum and dad came to pick us up I was on a real high. We went straight to our nan and grandad's who lived not very far away. In a way I could tell something wasn't right. We stayed at my nan and grandad's for a week and on the Monday my dad said he was taking my sister swimming. I asked if I could go but my dad said no. I remember being quite upset, but later I didn't think very much of it.

After Monday everyone seemed upset - my mum, dad, sister and my nan and grandad. We left to come home on the Saturday a week after we had got there and travelled home. We got in and I went on the computer to play a game and my mum and dad were having a whispered conversation. Then mum and dad both came in and told me to sit down as they had something to tell me. They told me that when I had been at summer camp the test results had come back and it was Turner Syndrome. My first question was "What?"

We must have talked for about two hours. I had all these questions going through my head and I had to get them out. As I was gradually told more I began to make sense of a lot of things. My dad apologised for not taking me swimming but it turned out that he had taken my sister swimming so that he could tell my sister. They had wanted to tell me at home. I went to bed understandably upset, and I don't think I got much sleep. However, when I woke up the next morning, I almost felt that nothing had happened. In September, I started my growth hormone and have been on it ever since.

That was four years ago now and at thirteen I am still under the same doctor as the one who diagnosed me and I wouldn't have it any other way. I am also under a doctor at Great Ormond Street and she is great and very supportive. I love life. I have some terrific friends who are very supportive. I have had trouble with other people at school but that's stopped now. I won't EVER let my TS stop me from doing what I want and it shouldn't stop anyone else. Everyone has their own problems and that's how I see my TS just a little problem.

The people, who have helped me through everything definitely have to be the doctors, but even more than them (sorry!!) are my family. I don't know what I would have done without them. They have been my shoulder to cry on and my trampoline when I fall.