A Parent’s Guide to Turner Syndrome
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You may have recently been told that your daughter has Turner Syndrome (TS), or you may have known this for a while. In either case, we hope you will find this booklet a useful source of information and advice. Before reading further, it is important to bear in mind a few key points. First, above all, be reassured that TS was not caused by anything you or your partner did – it is caused by a random genetic mutation. Secondly, there is every reason to feel positive about your daughter’s future – with modern medical help she should enjoy good health, happiness and a normal lifespan.

This booklet contains not only practical information, such as symptoms and medical treatments, but also wider issues, like talking to your daughter about TS, and other social and psychological aspects of TS.

We hope this guide will help you to feel positive and confident about dealing with TS. But always remember that all children are individuals, and you know your daughter better than anyone else. With your knowledge, and the resources available to you, your child can achieve her 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).

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.

There are 46 human chromosomes, including the sex chromosomes - called X and Y. As shown in the diagram, it is these sex chromosomes that make someone male or female - XX is female and XY is male.

In TS, part or all of one of the X chromosome is lacking or altered, and this leads to the physical characteristics of TS. 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 – your daughter may only have some of them. TS even remains undiagnosed in many people. In addition, with modern therapeutic approaches 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.*
Diagnosis and its effects

TS can be diagnosed at any age – from birth to adulthood. Many girls are diagnosed during early childhood, when growth slows down, and their height is obviously less than that of children of the same age. 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 – in a baby it may be because of difficulties with feeding, or puffy hands and feet. In childhood, lack of growth may lead to the diagnosis. In some girls, diagnosis is delayed until teens, when they consult their doctor because they haven’t started their periods (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 test 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 two X chromosomes (the sex chromosome that confers gender, among other things). This is written as 46XX.

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.

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 return to your specialist or contact the Turner Syndrome Support Society (TSSS) if there is anything that is not clear.
Receiving a diagnosis of TS is a major turning point for you and your child. Many parents feel a sense of relief that they know what’s wrong after years of nagging doubt. For children who are old enough to understand, the news that they may be able to catch up in height with treatment is often a hugely positive thing. However, it may also take time to come to terms with the diagnosis. It may also take a while to digest the potential symptoms and even to understand the complex genetics.

Many parents may ask – was it something we did? (the answer is NO! – it’s just a random chance that causes certain chromosomes to develop faults). The most important thing about the diagnosis is that it allows you to move forward and make full use of the treatments available to ensure your child achieves their best possible outcome. The issue of whether to tell your daughter about TS on diagnosis can be quite sensitive. For instance, is she old enough to understand the basic facts about TS? It is also important to consider how much to tell her, and what language to use, as some aspects of TS are complex. Do talk to your GP, genetic counsellor or other health professionals about the best way to talk to your daughter about TS.

In general, the following tips may be helpful:

- Be open with facts as much as possible, as children will often pick up on ‘secrets’ and would rather know what’s going on. They may guess that something is wrong anyway
- Encourage your daughter to share any concerns with you and to ask questions about TS
- Always maintain a positive attitude to TS, emphasising that you can overcome challenges together
Medical characteristics of Turner Syndrome

Before looking at some of the potential features of TS *(described below)*, it is important to stress again that those with TS can expect to live a long and healthy life. Many only have 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 short stature

Girls with TS are often slightly small at birth, and by the age of 2-3 years, their growth rate usually slows down, so they are small for their age throughout their childhood. It is at this point in early childhood that many children are diagnosed and can begin growth hormone therapy. Without growth hormone therapy, they will continue to grow at a slow rate, with low expectation of adult height (average height, 4'8”/142cm – about 8”/20cm shorter than normal average height). At puberty many girls will need further hormone therapy. In addition, girls with TS who do not receive hormone treatment will not enter puberty, and thus will miss out on the associated growth spurt, further reducing their height potential. In many cases, it also takes longer to reach full adult height – for instance the late twenties, rather than late teens. However, with hormone treatment, it is possible for most girls to reach close to their potential height, which will be linked to the heights of both of their parents.

Puberty and fertility

In TS the ovaries do not function properly, and do not produce the hormones that cause puberty. Therefore, without help from hormone treatment, girls with TS will not 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 of the non-functioning ovaries, very few girls with TS can conceive a child naturally (less than 1%). The good news though is that the other reproductive organs are not affected by TS – most importantly the uterus is normal and able to nurture a growing baby. With the help of hormone treatment, women with TS can enjoy a normal sex life. They may be able to conceive using egg donation and IVF (in vitro fertilisation).
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. Hearing tests are recommended if there is any doubt – particularly as impaired hearing can be linked to learning difficulties. 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. Some 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, and have regular check ups with a podiatrist.

- 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
- For babies, make sure sleepsuits, babygrows etc do not constrict the feet
Growth hormone

Growth hormone has changed the outlook for many girls with TS, enabling them to achieve a normal adult height (often over 5 feet). The growth spurt associated with starting growth hormone treatment 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 those with TS. The final height that a girl with TS will reach will depend on a number of factors, including the time of starting therapy, and also her 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. For some, they may need help from an early stage, for instance some babies with TS need help with feeding - some toddlers may need speech therapy. Other babies and toddlers with TS don’t have obvious developmental problems.

There can be issues with behaviour – from difficulty in concentrating to spatial awareness or hearing difficulties. Do not hesitate to seek help at an early stage – get as much help as you need – from the GP, the health visitor or the Turner Syndrome Support Society (TSSS).

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 – this can be addressed by making the teacher aware of the situation. Some girls with TS 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.

Good communication between home and school is obviously vital at all stages. If your child does have learning difficulties, the TSSS has produced an excellent booklet ‘How to help your child survive and succeed at school – A guide for parents and teachers’. Some parents get an independent educational assessment before their child starts school – this helps the teacher to get off on the right foot.
Short stature may also raise issues at school – for instance some types of PE may be difficult for your child: again, a sensitive approach from the teacher is very important. Ball games such as tennis and squash can be challenging. 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’ can make the child a target for bullying. Try to choose a school with a good anti-bullying policy and inclusive ethos – e.g. they may have a buddy system whereby younger ones are mentored by an older child. And again, tackle any issues early before they have a chance to escalate.

**Building self-esteem**

Self-esteem needs to be bolstered if a child feels different through being small. For instance, make sure clothes are appropriate for their age and fit well – it may be a question of making things for them, or altering shop-bought clothes. Personal possessions like bicycles need to be appropriate for their age – not just their size. Don’t make the mistake of treating them younger than their age – encourage them to be independent and do all the things their friends do. Try to find something your child is really good at then she can think ‘I may not be the tallest girl in the class, but at least I’ve got my level 2 at jazz dancing.’
Tips for building self-esteem

- Spend one-on-one time with your child, sharing their interests and listening to them without distraction
- Show plenty of physical affection – hugging, tickling etc
- Give plenty of praise for good behaviour and achievements

Making friends

Some girls with TS are very good at socialising but some need a little extra help in this area. Often it may just take a few pointers, such as emphasising the importance of turn-taking, and listening to other children speaking. Activities like Rainbows or Brownies help all children to interact and develop their social skills.

Tips for making friends

- Give your child plenty of opportunities to invite friends over for tea and play
- Talk to your child about their friendships – advise them on how to get on with friends and resolve arguments
- Explain to them the value of direct communication, such as ‘you made me sad when you wouldn’t play with me’

The TSSS has a great social aspect, where girls with TS can share experiences. The younger girls with TS look up to the older ones, and learn from them - it’s nice to feel they’re not alone.
Our stories

Although the experience of each child 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.

Abigail’s Mum

My name is Jacqui and I was 20 weeks pregnant when I found out that my baby had Turner Syndrome. I found out on Christmas Eve and of course could find no information on Turner Syndrome, particularly due to the time of year.

It was only in the New Year that I could begin finding out about my baby’s condition and what this would mean for her. I had genetic counselling and was put in touch with the Child Growth Foundation and subsequently Arlene of the Turner Group.

It was here, and from Arlene, that I found out the most up-to-date, accurate information and support.

It was, to be honest, a difficult first year with Abigail as she had heart surgery at the age of just 2 days and subsequently quite severe feeding problems. I was in and out of hospital with Abigail for most of the first year of her life, mainly with “failure to thrive” problems. Abi was tube fed for the first 2 years of her life and even so, was still sick a lot of the time. We eventually discovered that Abi was more suited to a pre-digested milk which was gentler on her tummy. After this, things became easier and later, with the help of a speech therapist, we eventually managed to get her to eat and drink normally.

Another feature of Turners is lymphodema of the hands and feet, but particularly the feet. Abi had special shoes as conventional shoes did not fit her feet, but they were lovely and other mums would often ask me where I had bought them! Abi did suffer several ear infections and has suffered particularly with glue ear. She has had grommets inserted 3 times. Abi also has long sight and squint and wears glasses to help with this.

Abi also had some problems with separation, ie. she would not play in a play pen, she would not sleep in a cot and would scream if you left the room.

Finally, I would just like to say that Abigail is now 8 and has blossomed into a chatty, sunny, confident, funny little girl. She is always smiling and laughing; she is always singing and dancing; she never really says anything unkind and is really loving and affectionate. She really is one of the nicest people I know, an absolute joy!
Eleanor’s Mum

Eleanor was born with puffy hands and feet and was looked at by a few paediatricians on 20 December 2004.

We were given the diagnosis of Turner Syndrome when Eleanor was 17 days old. I remember that the first question I asked was if life expectancy was a big factor and was quickly reassured that it wasn’t.

We read up as much as possible on the internet but were a bit disappointed to find that the hospital had a lack of knowledge of the syndrome.

As soon as I found Arlene’s number (at the Turner Syndrome Support Society), I gave her a ring and she’s been a pillar of support ever since. It’s nice to have someone who’s not only a lovely person to talk to but who also has first-hand experience of Turners.

I never really noticed how small Eleanor was for her age until I started a baby group, when she was about 5 months old. Seeing children her own age a lot taller or larger than Eleanor, kind of put her size into perspective, however she is still a very lively and happy little girl.

I do find though, that when we go out, people look at Eleanor and compare her to her sister, Cerys. Cerys is 8 months old. She currently weighs 18lb 13oz. Eleanor is 23 months old and weighs 19lb 2.5oz. Both of them are in clothing for 9 month olds and people think they might be twins. Whereas Cerys is a baby though, Eleanor is a little girl and is talking so people are surprised when they realise. They make comments on how small Eleanor is for her age and I point out that she has a growth problem. It does worry me in the back of my mind sometimes when I am dressing them both after a bath and look how chubby Cerys is and how Eleanor looks so skinny in comparison.

Eleanor started on growth hormone at around 17 months old. I found it rather easy to inject once I had a few goes and Eleanor has taken to it fantastically. No matter what difficulties Eleanor faces in life I still know she is going to be a happy fun loving girl and my little princess.
Contact Information

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