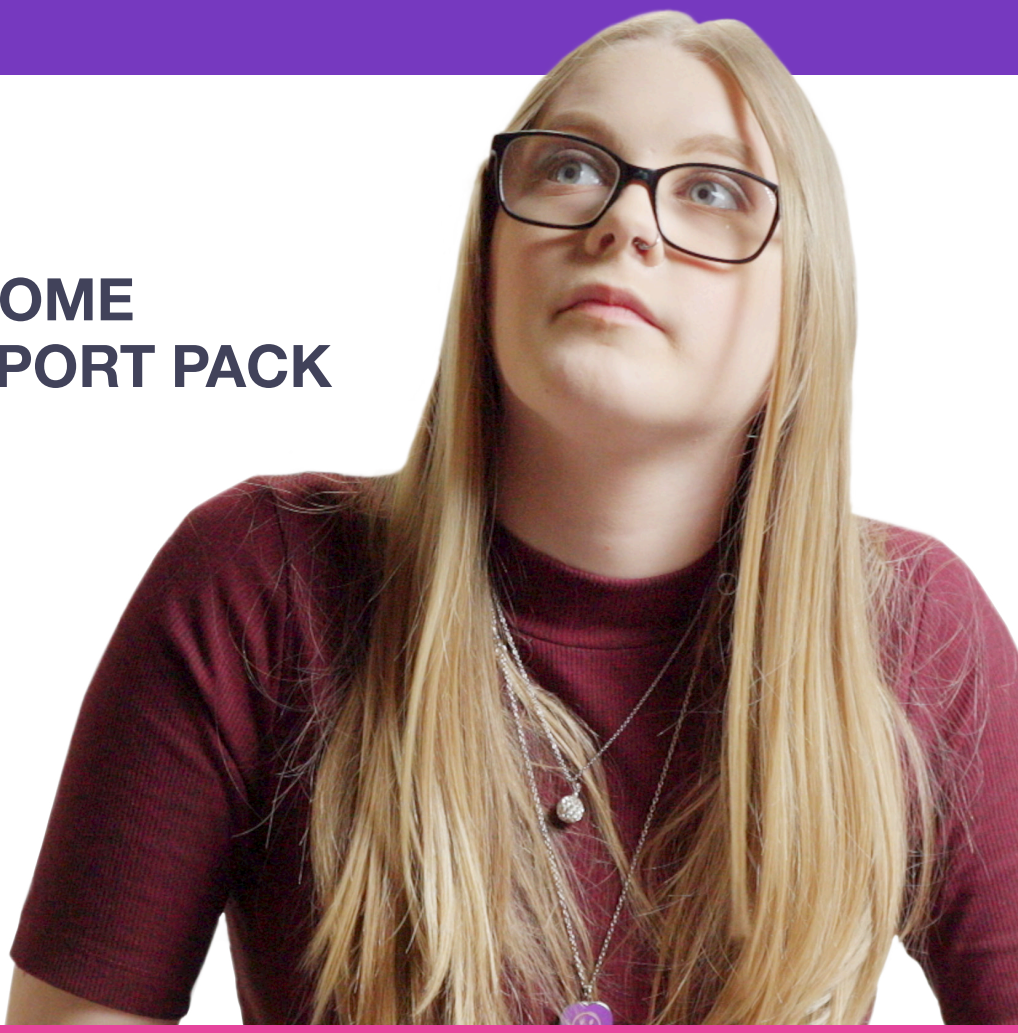
Missing an x.
But still exceptional.

TURNER SYNDROME DIAGNOSIS SUPPORT PACK



You have downloaded this pack because you think that you, your daughter or someone you know may have Turner Syndrome. So here is some more information about this condition.

This pack should provide you with more information and help support a conversation with your GP who can then offer help and advice about the next steps.

> When I found out Freya had Turner Syndrome, I thought 'What's that?!' I didn't know what it was. It was a bit scary, but it's not now.

Emma, mum of Freya, 7 (diagnosed during pregnancy)

What is Turner Syndrome?

Turner Syndrome is a complex genetic condition that only affects girls and is characterised typically by short stature and delayed or absent sexual development amongst other physical and behavioural symptoms.

Although Turner Syndrome varies in severity, it is always important to seek treatment as early as possible in order to give a girl with Turner Syndrome the best opportunity to live a healthy and fulfilling life.

How common is Turner Syndrome?

Turner Syndrome is classified as a rare genetic condition, although it is in fact the most common chromosomal defect affecting girls. It occurs in approximately 1 in 2000 live female births.

Although many girls with Turner Syndrome are diagnosed at birth, there are many who remain undiagnosed into their teens and even into adult life. An earlier diagnosis can lead to better management of the symptoms of Turner Syndrome and can help aid a girl with Turner Syndrome in living a normal life.

YOUR NOTES:

What causes Turner Syndrome?

Females are usually born with two X sex chromosomes, XX, however in Turner Syndrome one or part of an X chromosome is missing.

This occurs randomly when the baby is conceived in the womb. There are a few different karyotypes (chromosome analysis patterns) that can lead to Turner Syndrome and this is part of the reason why Turner Syndrome can look very different depending on the girl.

> “Being diagnosed with something as complex and intricate as Turner Syndrome … No two girls are the same. One girl’s diagnosis can be completely different to another girl’s. So don’t compare your journey to anyone else’s.”

Meg, 22 (diagnosed at 10)

YOUR NOTES:

How is Turner Syndrome diagnosed?

The term ‘syndrome’ is used to describe a collection of symptoms that originate from one cause, but it doesn’t mean that all are present.

Here is a list of some of the features that can be associated with Turner Syndrome. You may want to circle any areas of concern.

- ✖ Shorter than your friends/family
- ✖ Delayed or absent breast development compared to peers
- ✖ Delayed or absent pubertal development compared to peers, late or sporadic periods
- ✖ Social, emotional, behavioural difficulties and some specific learning difficulties
- ✖ Cardiovascular conditions
- ✖ Fertility problems
- ✖ Recurrent ear infections, hearing loss
- ✖ Increased numbers of moles on the skin
- ✖ Lymphoedema swelling of the feet and hands
- ✖ “Webbing” of the skin on the neck, a shorter wider neck
- ✖ A low hairline
- ✖ A high arched palate
- ✖ Low set ears
- ✖ Droopy eyelids
- ✖ Short sighted
- ✖ Spoon-shaped nails, ingrown toe nails
- ✖ Broad chest
- ✖ Increased carrying angle of the elbow

YOUR NOTES:

How is Turner Syndrome diagnosed?

Turner Syndrome may be suspected in pregnancy during a routine ultrasound scan if heart or kidney abnormalities, or swelling in the body's tissues are detected.

Later on in life, a suspected diagnosis can usually be made due to the presence of some of the physical features discussed above, especially being shorter than peers. Sometimes, it may be that one of the conditions associated with Turner Syndrome, such as hearing problems or ear infections, may lead to a suspected diagnosis.

If needed, your GP can refer you to an Endocrinologist who is a specialist in growing and hormones and who will carry out a test called 'karyotyping'. This simple blood test analyses chromosome patterns in human cells and will be used to confirm a diagnosis.

YOUR NOTES:

> "If you're diagnosed early, it's a given that there are going to be less complications. If you start growth hormone early you are going to get the benefit from it. You will also start oestrogen at the right time. Getting treatment as early as possible is almost like mimicking nature's course."

Hannah, 14 (diagnosed at birth)

How are girls with Turner Syndrome treated?

Although there is no cure for Turner Syndrome, there are treatments available that target the associated symptoms and the condition will usually be managed by a multi-disciplinary team of healthcare professionals.

Growth hormone therapy is initiated as soon as possible after diagnosis providing the girl hasn't yet reached final height (at about 16). This involves taking a daily injection and will help her grow towards a normal height in adulthood.

Hormone replacement therapy is normally initiated at the start of when puberty would normally occur (around 11 or 12) and will be taken until menopause is reached (around 50) as a tablet, patch or gel. Oestrogen, which is the female sex hormone, is not produced in girls with Turner Syndrome and so oestrogen replacement therapy is needed to begin the changes that would normally occur during puberty, such as breast growth. It also protects the womb and the bones, preventing osteoporosis. Progesterone is usually started after oestrogen and helps start regular monthly periods.

Throughout life, girls with Turner Syndrome are closely monitored to ensure that sound health is maintained, and they may visit a number of specialists to ensure all the associated conditions can be managed effectively.

YOUR NOTES:

Why should I book a visit with a GP?

If you, your daughter or someone you know shows some of the characteristics discussed in this resource pack, it is important that a healthcare professional is consulted as soon as possible.

It may be that the girl in question doesn't have Turner Syndrome but may have something else associated with similar symptoms. It is important to emphasise that she may not have Turner Syndrome and may just be experiencing growth and sexual development at a different pace to her peers. However it is better to visit a GP for peace of mind and the knowledge that if it is Turner Syndrome, it can be treated and managed effectively if diagnosed.

It is likely that the GP may refer on to an endocrinologist (a hormone specialist) who will have more knowledge of Turner Syndrome.

YOUR NOTES:

> "I run a clinic for women with Turner Syndrome and if I had one piece of advice for anyone thinking they, or someone they may know might have Turner Syndrome, it would be to go and visit a GP. Explain your concerns and worries and they will be able to help you understand if you do have Turner Syndrome and direct you to someone who can help you further."

Dr Helen Turner, Consultant Endocrinologist

Prepare a list of any relevant symptoms that you can think of. The more information that you can give your GP the better their understanding will be.

- Explain your concerns that it may be Turner Syndrome and why it is important to you to have a diagnosis if your concerns are correct.
- Don't be afraid to show your knowledge if you have done some research. This isn't something that GPs will come across every day!
- Remember that it may not be Turner Syndrome, but the GP will be able to let you know if there is anything to be worried about.
- Try and keep a note of what you talked to your GP about in your consultation as it is normal to feel anxious in these situations and forget parts of the discussion. This will help later if you are referred onto another healthcare professional.
- Girls show a wide variation in the age at which they develop pubertal changes. However, puberty is considered to be delayed if there are no signs of development when a girl reaches her 13th birthday. If you have concerns, it's important that further tests are performed to discover if there is a problem such as Turner Syndrome.

If you would like to find out more about Turner Syndrome or find someone that you can talk to about it, even before you visit your GP, go to **www.tss.org.uk***

The Turner Syndrome Support Society is an active society set up to support those with Turner Syndrome who live in the UK. On their website there are plenty of resources available to help you understand Turner Syndrome further and there are ways that you can get in contact by email or phone.

TSSS

*Links to external websites are provided as a resource to the viewer. This website is neither owned or controlled by Pfizer Ltd. Pfizer accepts no responsibility for the content or services of the linked site.

This material forms part of the **www.missinganx.com** campaign which is developed and funded by Pfizer. The information provided on this site is intended for general information and education and is not intended to be a substitute for advice provided by a doctor or other qualified healthcare professionals.

PP-GEN-GBR-1539. August 2022.

Pfizer