

Factsheet 13

Turner Syndrome **and your heart**



TURNER SYNDROME
TSSS
SUPPORT SOCIETY [UK]

Understanding the possible problems you may have with your heart is important, as women with Turner Syndrome (TS) are more likely to be born with a heart problem (congenital), and/or develop an abnormality with the heart during their lifetime when compared with girls and women without TS. All girls, teenagers and women with TS will have investigations to help assess and monitor these conditions and where necessary make sure that the right treatment is given if needed.

Congenital Heart Problems in TS

Bicuspid aortic valve (BAV)

The aortic valve is the main outlet valve in the heart, and usually has 3 small flaps or 'cusps' that open and close as the blood flows through. Around 20% of women with TS are born with 2 cusps rather than 3 - this is a 'bicuspid aortic valve', and is also found in around 1% of the general population. The bicuspid valve often functions normally but in some women, it can over time, become narrowed ('stenosed') or can leak, so you need to be seen in clinic because of this. The bicuspid aortic valve is normally diagnosed with an echocardiogram, which is an ultrasound scan of the heart. Women with a narrowed or leaky valve that becomes more severe over time may need to undergo a surgical operation to replace the valve.

Meg and her heart check up

"Yesterday I had a heart echocardiogram and scan.

They used an ultrasound machine to look closely at my heart, which is a small probe that they press on my chest that looks through my skin and creates a picture on the computer for the doctor to look at. They attached stickers to my chest and hooked up wires to measure my heart beats.

To any girl with Turner Syndrome heart health is so important. This was completely painless and I urge all of you, no matter what your age, to go and get checked out. There is nothing to be frightened about."

With a bicuspid aortic valve there is an increased risk of infective endocarditis (an infection of the heart valve). The symptoms of endocarditis are a persistent temperature or flu like illness, which does not settle, and of feeling generally unwell. Please contact your specialist if you have any of these symptoms.

Coarctation of the aorta

The aorta is the large blood vessel that takes oxygen rich blood from your heart to the rest of your body. Coarctation is a narrowing in part of your aorta which stops the lower half of your body receiving enough blood. This is normally diagnosed in childhood with an echocardiogram, however if the narrowing is not severe, it can present later in life with patients having high blood pressure. Women with

TS are more likely than the general population to have a coarctation.

Coarctation can be successfully treated either by surgery or by a keyhole operation.

Patients do need to be reviewed regularly to check their blood pressure and the site of the coarctation repair.

Occasionally the coarctation site can become enlarged or narrow again, and this can be seen either on an echocardiogram, however you may need a CT scan or an MRI scan.



Anomalous Pulmonary Venous Drainage

A small number of women with TS (around 10%) have anomalous pulmonary venous drainage, where oxygen rich blood returns to the right side of the heart, not the left side. Women can have no symptoms, or can be slightly short of breath when they exercise. The diagnosis is made with an echocardiogram and either a CT or an MRI scan. The cardiologist will review the scan to look for this, and sometimes women may need surgery to correct this. Others may simply need regular monitoring.

Acquired Heart Conditions (can develop during your lifetime)

Dilatation of the ascending aorta and Aortic Dissection

Women with TS are at increased risk of enlargement of the ascending aorta, which can lead to a condition called aortic dissection. The aorta is the main blood vessel coming out of the heart, and this can get bigger over time. Conditions that can worsen this include having a high blood pressure and having a bicuspid aortic valve.

Women with TS need to be reviewed regularly with an echocardiogram (ultrasound) and if needed an MRI or CT scan to assess their aorta. Very rarely the aorta can enlarge and then suddenly dissect or tear, which can be a life threatening event. The symptoms that women describe with a dissection are a sudden severe pain in their chest, but they can also feel very unwell or collapse. An aortic dissection requires emergency surgery and therefore you or your relative/friend/work colleague would need to dial for an ambulance immediately and get to hospital. It can occur in pregnancy, and therefore it is important that you are reviewed by a

cardiologist before/during and after pregnancy. As it is a rare event, we advise all women with TS to carry a warning card to help medical professionals think of and to make the diagnosis.

High Blood Pressure/ Hypertension (see Factsheet 12)

The heart works by pumping blood around your body to deliver oxygen and nutrients to your organs. Your blood pressure is the force your heart uses to pump blood around your body through the arteries, which are your blood vessels. You need some pressure to keep your blood moving around your body. Your blood pressure naturally goes up and down, especially when moving or exercising. If your blood pressure is always high, even when you are resting, then you have developed hypertension or high blood pressure.

Women with TS are more at risk of high blood pressure and it is important to monitor for this and treat if required.



Carolyn and her surgery to prevent Aortic Dissection

"As part of my regular endocrine checks I had regular echocardiogram and MRI scans. It was noted I had a dilated aortic root, which was monitored closely. It slowly increased in size and the idea of surgery to prevent dissection was put to me. After considering the options given to me, meeting the surgeon, bombarding him with questions, and the support of my family I decided to go ahead. A decision I've never regretted, and 10 years on I'm leading a good life, still here to tell the tale."

Ischaemic Heart Disease IHD/ Coronary Artery Disease

Ischaemic heart disease is when your coronary arteries, the arteries supplying blood to your heart become narrowed by a build-up of fatty material within their walls. The arteries may become so narrow that they can't carry enough oxygen rich blood to your heart, causing symptoms including chest pain (angina) or breathlessness. If a piece of atheroma breaks off, it can cause a blood clot to form. This clot can block your coronary artery and cut off the supply of blood and oxygen to your heart muscle. This is known as a heart attack. There are several risk factors that can increase the likelihood of developing IHD. These include having a high blood pressure, high cholesterol, diabetes, smoking, being overweight, not doing enough physical activity, a family history of heart attacks and your ethnic background. The risk of IHD increases with age. It is commoner in women with TS, and therefore it is crucial to try and reduce your risk. It is also important to monitor for any symptoms that might be a sign of ischaemic heart disease or cerebrovascular disease CVD (narrowing of the blood vessels to the brain which could cause a stroke). Living a healthy lifestyle can help lower your risk of developing both IHD and CVD.

Tests on your heart

1. **Echocardiogram.** An Echocardiogram looks at the structures of your heart, and gives information on how well your heart is working and on the function of your heart valves. A gel is used with the scan to help the sound waves reach your heart. It feels cold and sticky, but is otherwise harmless.
2. **ECG.** An ECG records the rhythm, rate and electrical activity of your heart by attaching sticky electrodes to your body
3. **A BP monitor.** Your blood pressure is measured using a blood pressure cuff placed on your arm. When you have your blood pressure measured, your reading is written as two numbers. The first is when the pressure is at its highest (or systolic pressure), for example in adults 120mmHg and the second is when it is at its lowest (or diastolic pressure), for example 80mmHg. Your blood pressure should be under 140/90 mmHg in adults. For children, there are age-related ranges that are used.
4. **An MRI scan.** A cardiac MRI scan is a non-invasive test that uses an MRI machine to create magnetic and radio waves to show detailed pictures of the inside of your heart.
5. **CT scan.** A computerized tomography scan (CT scan) uses computers and rotating X-ray machines to create cross-sectional images of the body.

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Ref: Clinical Practice Guidelines, European Journal of Endocrinology (2017) 177, G1-G70, www.tss.org.uk.

This factsheet was produced thanks to a grant from the Society of Endocrinology.

TURNER SYNDROME



SUPPORT SOCIETY [UK]

Further information about Turner Syndrome can be obtained from: Arlene Smyth - Executive Officer
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Spring 2021 • Photo courtesy of TSSS members