Picture Perfect: Imaging the Aorta in Marfan Syndrome

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It is important to look at how Marfan syndrome affects different parts of the body. The most important concern is how it affects your heart and blood vessels, especially the main artery that takes blood from the heart to the body – the Aorta. The majority of those with Marfan syndrome (60-80%) will develop problems with their aorta at some point in their lifetime. This includes aortic regurgitation (where blood leaks from the aorta back into the heart), aortic aneurysm (enlargement of the width of the aorta) or aortic dissection (tearing of the inner layers of the aortic wall).

Thanks to better early-stage diagnosis, careful monitoring, proper management and timely preventative aortic surgery, the life expectancy of people with Marfan syndrome is now nearing that of the general population.

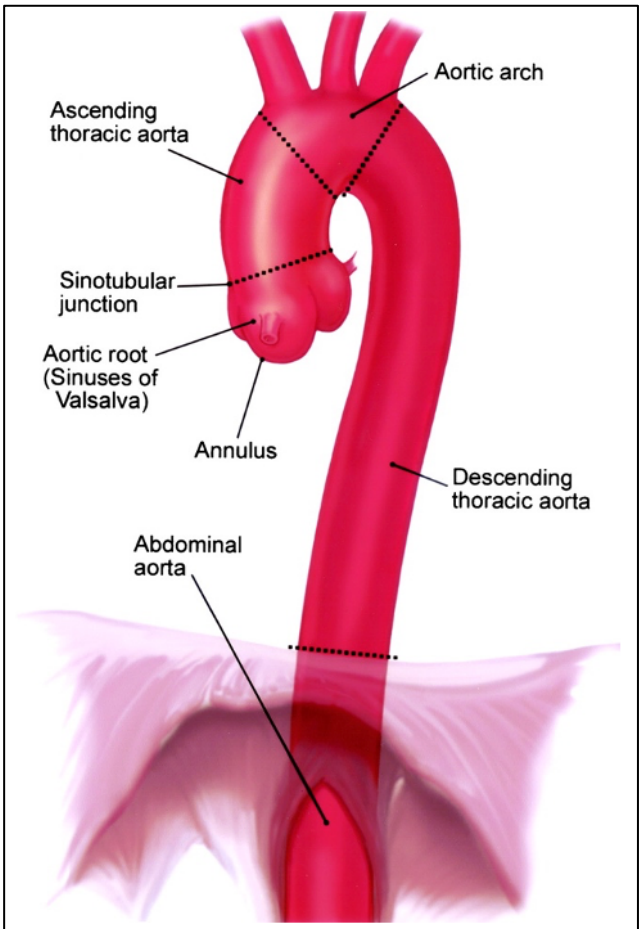

Figure 1: Segments of the aorta. Taken from (1).

The aorta is divided into five segments, and the part that connects to the heart is called the aortic root (Figure 1). A special measurement called a Z-score compares the size of the aortic root to what is expected for someone's body size and age. A Z-score above 2.0 indicates that the aortic root is enlarged, which can be a sign of Marfan syndrome. Adults can check their z score with the following: https://marfan.org/dx/z-score-adults/.

The Z-score, along with aortic root diameter is very useful to clinicians. The latter remains the major determinant of aortic complications such as an aneurysm or dissection. The latest guidelines suggest that the risk of a serious tear, known as an aortic dissection, increases when the aortic root's size exceeds 50 mm, or 45mm in the presence of risk factors. These include a family history of aortic problems, rapid aortic growth (more than 3 mm per year in adults), pregnancy, and uncontrolled high blood pressure. Recent research is also showing that certain genetic differences may influence a person's risk of developing aortic complications.

There are various ways in which we can measure the aorta. This includes echocardiography, CT scans, MRI scans and doppler ultrasound scans. Each type of imaging technique has different value in terms of its ability to predict an aortic event such as dissection. Here is an overview of how each one is used:

Echocardiography

Echocardiography is an ultrasound scan of the heart that uses sound waves to create pictures of your heart valves and chambers. There are two main types of echocardiography: transthoracic echocardiography (TTE) and transoesophageal echocardiography (TOE). TTE involves placing an ultrasound probe directly on the chest, while TOE uses a probe inserted into the oesophagus (the food pipe), which allows for more detailed images, especially of areas that are harder to see with TTE. Although TOE provides more accuracy, it is more invasive and requires sedation, so it is often used during surgery or at the bedside, rather than for long-term monitoring. Doppler echocardiography is a specific type of echocardiography that focuses on visualising blood flow through the heart's valves and chambers.

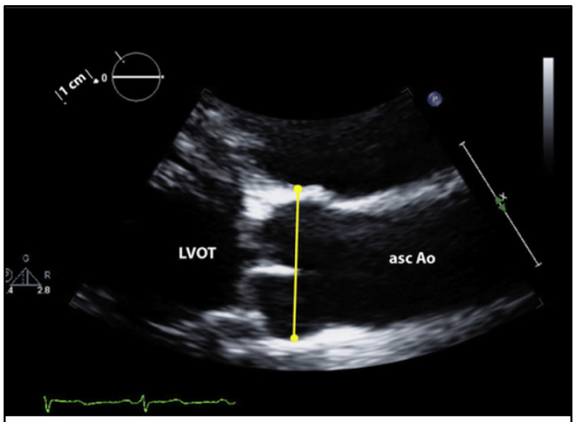

Figure 2: Measurement of the aortic root with echocardiography. Taken from (2)

Echocardiography is the most affordable and widely available type of heart scan, making it the first choice for evaluating aortic diseases. However, while echocardiography provides valuable information about the upper part of the aorta, it may not accurately measure the aortic arch (the curved part) or the descending aorta. In those cases, a CT scan or MRI is recommended for more precise imaging.

Cardiovascular Computed Tomography (CCT)

Cardiovascular CT (CCT) is a widely available and reliable imaging method commonly used to diagnose, monitor, and plan treatment for aortic diseases. It works by taking multiple X-ray images of the chest, which are combined to create detailed cross-sectional views. This is especially helpful in emergency situations as it provides results that are almost 100% accurate in detecting aortic conditions.

During a CT scan, contrast dye is often injected into the bloodstream, highlighting blood vessels, organs, and tissues, which makes them easier to examine. Some CT protocols can evaluate not only the aorta but also the pulmonary (lung) and coronary (heart) arteries simultaneously. To prevent blurring or measurement errors, especially around the aortic root and ascending aorta, the scan is synchronised with the heart's electrical activity, improving the accuracy of the images.

A standard CCT protocol involves several steps:

- A non-contrast scan to check for calcifications (hard spots where calcium has built up, which can sometimes mean there's damage), blood clots, or surgical material.
- A contrast-enhanced scan to visualise blood flow and the aorta more clearly.
- A late scan to detect leaks or signs of inflammation or infection in the aortic wall.

Although contrast dye improves image clarity, it can sometimes pose risks, such as kidney toxicity or allergic reactions. It's also important to note that CT scans expose you to radiation, so doctors aim to limit radiation, especially for pregnant women and younger individuals, when using CT for long-term monitoring.

Mastering the Cloverleaf: The Challenge of Aortic Root Measurements

Measuring the aortic root can be tricky because it has a unique cloverleaf shape (Figure 3) and is often uneven, especially in patients with aortic root enlargement. Standard measurement methods sometimes underestimate how large this area actually is, which could lead to delays in treatment. Two main methods are used: one that measures across the deepest points of the sinuses (called "sinus-to-commissure" – Figure 3a) and another that measures across the widest part ("sinus-to-sinus" – Figure 3b). The sinus-to-sinus method usually gives a more accurate size and helps ensure that important issues, like aortic enlargement, aren't missed, so surgery can be done in time if needed.

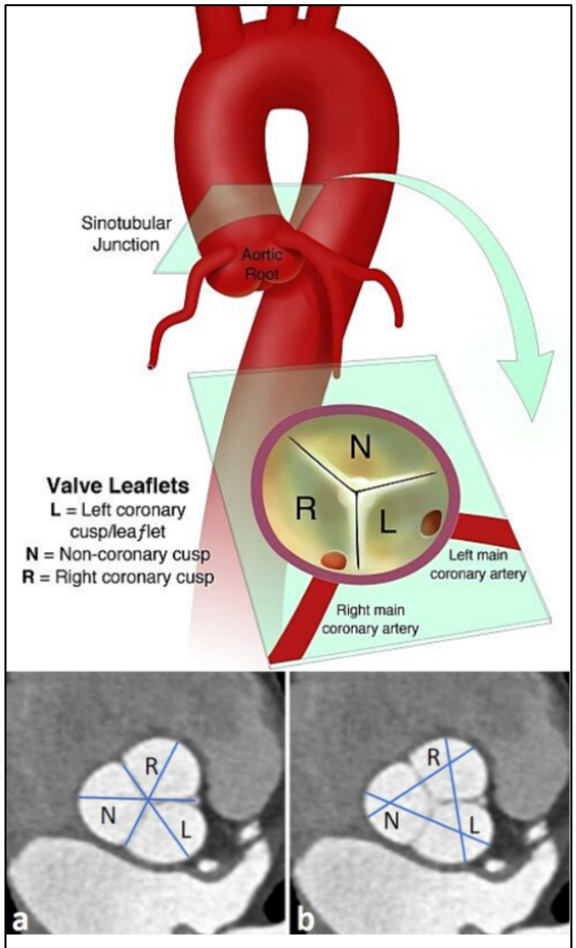

Figure 3: Cross-section of the aortic root. (a) shows the sinus-to-commissure method. (b) shows the sinus-to-sinus method). (3,4)

Cardiovascular magnetic resonance (CMR)

CMR is a non-invasive imaging technique that uses powerful magnets and radio waves to create detailed pictures of the heart and blood vessels. It works by aligning hydrogen atoms in the body with a magnetic field, then using radio waves to disturb them. As they realign, the MRI detects the energy released and converts it into images. It is the most detailed imaging method for the aorta and can examine the aorta's size, shape, and tissue characteristics, such as inflammation, infection, plaque build-up (substance building up in the arteries, which can lead to blockages and cause heart problems), or bleeding. CMR can also assess how well your heart valves and chambers are working and measure blood flow.

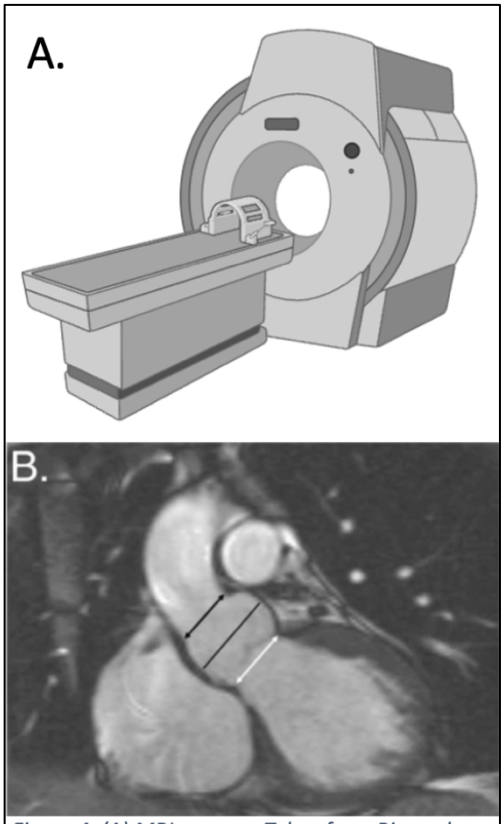

Figure 4. (A) MRI scanner. Taken from Biorender. (B) Outflow of the left ventricle (chamber) of the heart. Taken from (5)

CMR is especially useful for young people, women, and pregnant patients because it doesn't involve radiation or iodine-based contrast dyes. However, if kidney function is very low, special care is needed when using gadolinium-based contrast agents. CMR is generally safe for people with pacemakers or defibrillators with proper monitoring, though it's not recommended for those with certain implants like cochlear or brain clips. In emergency situations, CMR is less commonly used because it's not always available, takes longer, and can be difficult to use on patients who are unstable.

Although CMR can assess the heart's function and blood flow in ways that help doctors better understand a patient's risk of complications, CCT, with its higher image clarity, may be recommended for detailed planning before surgery or when there are discrepancies in previous measurements.

Which Scan is Right?

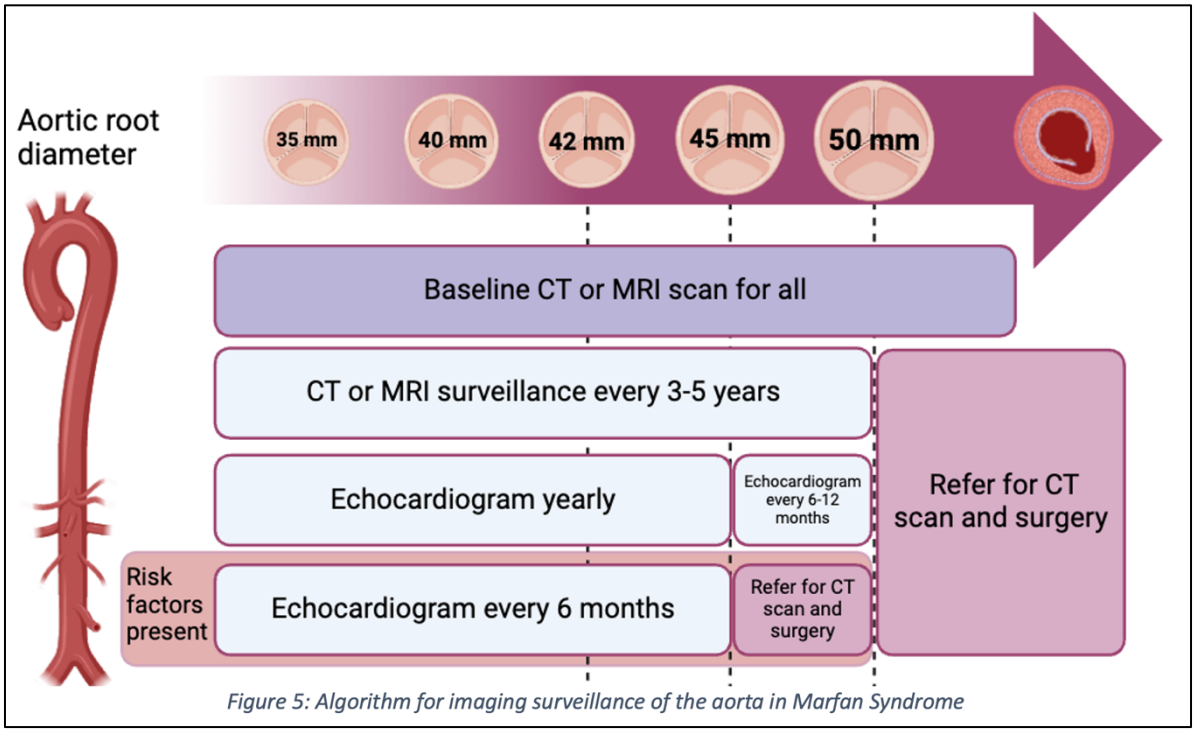

Figure 5: Algorithm for imaging surveillance of the aorta in Marfan Syndrome

The frequency and mode of imaging for Marfan syndrome should be tailored to each patient, taking into account their medical history, risk factors, and prior test results. As shown in Figure 5, the recommended surveillance schedule for aortic root diameter monitoring includes a baseline CT or MRI for all patients, which is repeated every 3-5 years. Additionally, an annual echocardiogram is advised for diameters up to 45 mm, and this increases to every 6-12 months for diameters between 45-50 mm. However, for patients with additional risk factors, such as a family history of aortic problems, rapid aortic growth (more than 3 mm per year in adults), pregnancy, or uncontrolled high blood pressure, echocardiograms are recommended every 6 months for diameters up to 45 mm. Surgery should be considered once the diameter exceeds 45 mm in this subset of patients. Any patient with Marfan syndrome and an aortic diameter greater than 50mm should be referred for a CT scan and evaluated for preventative surgery.

Table 1 provides a comparison of the key imaging techniques: Thoracic Echocardiogram, Oesophageal Echocardiogram, CT scan, and MRI Scan, highlighting their strengths and limitations (6).

	Thoracic Echocardiogram	**Oesophageal Echocardiogram**	**Cardiac CT scan**	**Cardiac MRI Scan**
Availability	++++	+++	++	+
Cost	+	++	+++	++++
Time requirement	+	+++	+++	++++
Radiation	0	0	+++	0
Kidney toxicity	0	0	+++	+
Accuracy	++	++++	++++	++++
Serial examination	++++	++	++	++++
Aortic wall visualisation	++	+++	++++	++++
Aortic valve function	+++	++++	+	++++
Ventricle function	+++	+++	+++	++++
Aortic root assessment	+++	+++	++++	++++
Aortic arch assessment	++	+++	++++	++++
Thoracic aorta assessment	+	++	++++	++++
Abdominal aorta assessment	+++	-	++++	++++

Table 1: Comparison of currently available cardiac imaging techniques.

What does the future hold?

The future of aortic imaging with CT and MRI is moving towards more personalised and precise risk prediction. New technology, such as 4D flow MRI, allows us to visualise blood flow in detail. In cases of aortic dissection, where there is a tear in the aorta, abnormal blood flow can indicate worse outcomes. 4D flow MRI helps to identify these high-risk patterns. Although this technology is becoming more accessible, it is not yet widely available.

Recent innovations in CT scans, like photon-counting CT, provide even sharper images and can detect inflammation and plaque build-up in the aortic wall. Techniques such as vascular deformation mapping, which involves analysing how blood vessels stretch, compress, or change shape in response to blood flow, improve the accuracy of aortic measurements, giving more reliable results.

Artificial Intelligence (AI) is set to play a significant role in aortic imaging. AI enhances the accuracy of complex measurements, such as those from 4D flow MRI, accelerates data analysis, detects anomalies, summarises reports, and predicts outcomes. AI can also segment structures, like estimating aortic wall volume, which is being explored as a potential predictor of dissection in ongoing research at the Royal Brompton. The goal is to provide a more holistic picture of aortic dissection risk, ensuring that only those who truly need surgery get it, while those who do need it can receive it sooner.

Incorporating these advanced techniques into everyday clinical practice will require further research and validation through large, well-designed studies.

5 interesting and current references:

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