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CONGENITAL HEART DISEASE

Structure and function of the aorta in inherited and congenital heart disease and the role of MRI

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The aorta is not simply a tube or conduit but a highly complex part of the vascular tree, originating from the left ventricular (LV) outflow tract and aortic valve and extending to its major thoracic and abdominal branches. Passing blood from the heart to the limbs and major organs is one functional aspect of the aorta; of equal importance is its capacity to distend and recoil in response to pulsatile flow, thereby reducing afterload for the LV and facilitating diastolic perfusion of the coronary arteries.

Intrinsic aortic wall abnormalities have been described in inherited connective tissue disorders such as Marfan syndrome and bicuspid aortic valve (BAV) disease, but recent reports indicate similar aortic involvement in classical congenital heart disease (CHD) entities such as coarctation of the aorta, tetralogy of Fallot (ToF) and transposition of the great arteries (TGA).^{1,2} The aortic media consists of smooth muscle and an extracellular matrix that is composed of ground substance, in which elastic fibres and collagen are embedded in a hydrated gel.^{1,2} Smooth muscle cells govern vasodilatation and vasoconstriction, collagen provides inert aortic wall strength, and elastin permits distension and recoil in response to pulsatile flow.¹ Current focus on the origin of aortic wall abnormalities has shifted towards these specific medial constituents and their crucial role in the structural and functional integrity of the aorta (figure 1).^{1,2}

In prototypical diseases such as Marfan syndrome and BAV disease, loss of fibrillin-1 microfibrils has been reported to dissociate smooth muscle cells from the medial matrix components, resulting in accelerated cell death and matrix disruption.³ Matrix metalloproteinases (MMPs)—endogenous enzymes that degrade the matrix components—become activated in fibrillin-1 deficient tissues, degrading the structural support of the aorta.³ Similar focal abnormalities within the aortic media have recently been described in coarctation, ToF, and TGA.^{1,2} Whether these aortic wall changes result from an intrinsic medial abnormality or are secondary to haemodynamic states before and after surgical repair (or both) is unknown.^{1,2} Whatever the aetiology of aortic wall pathology given this heterogeneity, aortic dilatation and reduced aortic elasticity will evolve in all entities when loss of structural support of the aortic wall progresses.^{1,2}

With advancing age, the aortic wall structure undergoes unfavourable changes resulting in a decline in aortic elasticity and an increase in aortic circumference.^{1, w1} Arterial hypertension, atherosclerosis, and other cardiovascular risk factors such as smoking, hypercholesterolaemia, and diabetes may all lead to atheroma formation that may progress to aortic dissection and rupture due to similar degenerative processes in the extracellular matrix and the vasa vasorum. Therefore, the negative sequelae related to ageing and these cardiovascular risk factors will have an additional impact on already existing aortic wall abnormalities, and adversely affect the long term prognosis in affected CHD patients.

Both dilatation and reduced elasticity due to loss of (elastic) properties of the aorta have a detrimental effect on aortic valve function, especially if the aortic valve is already structurally malformed (bicuspid).^{w2} Decreased aortic root elasticity increases leaflet stress and therefore predisposes for aortic valve dysfunction.^{w2} Aortic dilatation contributes to aortic valve dysfunction by loss of coaptation of the aortic valve leaflets.^{3, w2} Moreover, with reduced aortic elasticity, the cushioning 'windkessel' effect of the aorta is impaired. This leads to increased systolic blood and pulse pressure, which in turn will increase myocardial oxygen demand.^{w3} Arterial stiffening is also associated with impaired coronary perfusion, the presence of coronary artery disease, and LV dysfunction^{w4} (box 1).

As the life expectancy of CHD patients has much improved over the past decades, cardiologists will be increasingly confronted with the challenge of concomitant aortic sequelae (figure 2). Therefore, non-invasive monitoring of aortic dimensions and elasticity, aortic valve competence, and LV function is clinically highly desirable in all affected CHD patients. Echocardiography is widely used in daily practice, but has drawbacks as acoustic windows generally become limited with increasing patient age. In contrast, MRI provides unlimited field-of-view for accurate assessment of aortic and cardiac anatomy and function.^{4–9, w2, w5–w8} Although MRI is more time consuming and expensive, implementation in the follow-up of patients with aortic sequelae may be cost effective, as its imaging quality and reproducibility may allow for better selection of patients for intervention.^{w9}

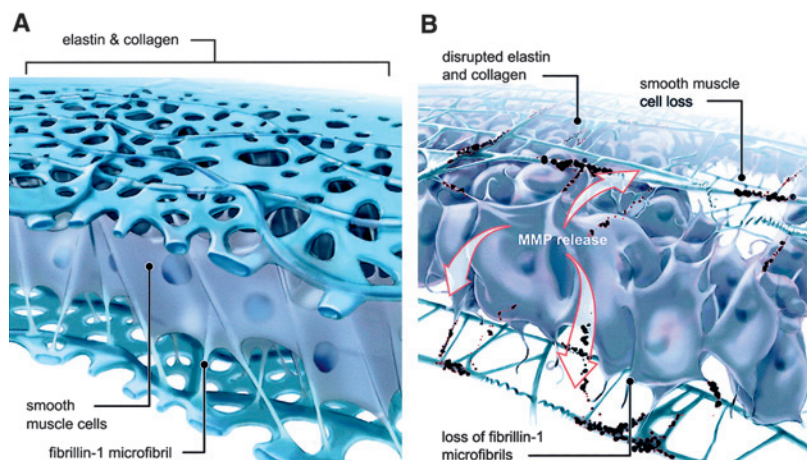


Figure 1 The elastic laminae of the aortic media provides structural support and elasticity to the aorta. In normal tricuspid valve patients (A), fibrillin-1 microfibrils tether smooth muscle cells to adjacent elastin and collagen matrix components. In patients with a bicuspid aortic valve (B), deficient microfibrillar elements result in smooth muscle cell detachment, matrix metalloproteinases release, matrix disruption, cell death, and a loss of structural support and elasticity. Reproduced with the permission of Elsevier from Fedak *et al.*³

In this article, the five most common entities of inherited connective tissue disorders and classical CHD with intrinsic aortic wall abnormalities will be discussed, including Marfan syndrome, BAV disease, coarctation of the aorta, ToF, and TGA, with description of the potential role of MRI in their evaluation and management.

MRI OF THE AORTA: TECHNIQUES AND PROTOCOL

An overview of a typical imaging protocol is listed in table 1. The focus of the protocol is on assessment of aortic dimensions and vessel wall condition (expressed by elasticity), aortic valve competence, and LV function.

Cardiac and aortic anatomy

A whole heart, three dimensional (3D) steady state free precession (SSFP) sequence can be acquired to generally outline the cardiac and cardiovascular anatomy.⁸ For this, a 3D volume is obtained covering the cardiovascular structures between the level of the diaphragm and the neck.⁸ Instead of multiple series of two dimensional (2D) slices requiring highly individualised planning procedures,

Box 1 Deleterious effects of aortic wall abnormalities

- ▶ Frequent aortic dilatation and even aortic dissection.
- ▶ Loss of aortic elasticity.
- ▶ Increased systolic blood and pulse pressure.
- ▶ Increased LV afterload.
- ▶ Increased myocardial oxygen demand.
- ▶ Impaired coronary perfusion.
- ▶ Development of coronary artery disease.
- ▶ LV dysfunction.

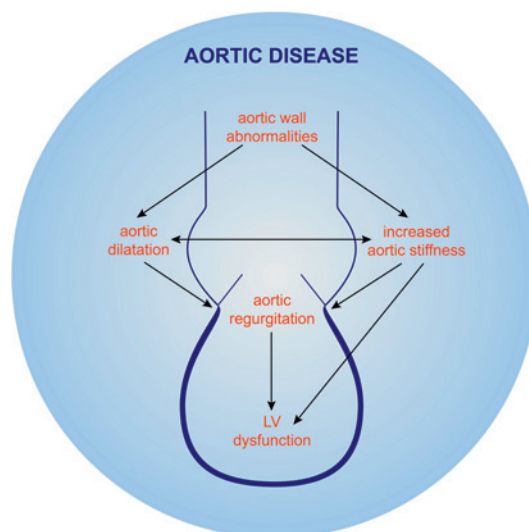


Figure 2 Overview of the aortic wall pathology cascade, indicating the close relationship between aortic dimensions and elasticity, and aortic and left ventricular function.

all cardiac information can be contained in this single operator independent acquisition without breath-holds.⁸ During subsequent post-processing, arbitrarily reformatting of any desired imaging plane can be performed without loss of resolution.⁸ Contrast enhanced 3D MRI angiography (MRA) (using gadolinium) may be used additionally to visualise aortic dimensions at any level and the potentially complex nature of a coarctation lesion (figure 3), as well as aortic complications such as aortic aneurysms.^{7 10} Aortic wall sequelae such as dissection and intramural haematoma can be assessed with 2D black blood spin echo images.

Aortic elasticity

Aortic wall degeneration is not only associated with dilatation, but also with loss of elastic properties.^{4-7 w2 w4 w6} The resultant increased aortic stiffness is represented by locally reduced aortic distensibility and increased regional pulse wave velocity (PWV).^{4-7 w2 w4 w6} Aortic root distensibility (mmHg^{-1}) can be measured at the level of the sinotubular junction, for which two separate images are acquired for the maximal and minimal aortic lumen area (A_{max} and A_{min}) measured at the peak of aortic systolic flow and before the systolic flow curve (coinciding with the isovolumetric contraction phase), respectively.^{w2} Distensibility (mmHg^{-1}) can be calculated using the following formula: $([A_{\text{max}} - A_{\text{min}}] / [A_{\text{min}} \times (\text{systolic blood pressure} - \text{diastolic blood pressure})])$.^{w2}

Regional PWV can be acquired for the aortic arch and the descending aorta (figure 4).^{4-7 w6} For this, 2D flow MRI is applied at the levels of the ascending aorta, proximal descending aorta, and the abdominal aorta for calculation of the propagation velocity of the systolic flow wave.^{4-7 w6} A higher PWV represents a stiffer aorta, providing important prognostic information as increased PWV predicts

Table 1 MRI scan protocol

Number	MRI sequence type	Scan duration (min)
1	Scout images single phase and parallel imaging reference scan	2
2	Whole heart 3D sequence (FB), morphology heart and aorta	5–10
3	Whole heart 4D flow sequence (FB), flow across 4 intracardiac valves	5–10
4	Aorta 4D flow sequence (FB), optional, flow within thoracic aorta	5–10
5	Contrast enhanced 3D MRA (FB), morphology thoracic aorta	5–10
6	Two sets of scout cine images of the aortic root (BH)	2
7	2D minimal and maximal lumen area at sinotubular junction (BH), for aortic distensibility	3
8	2D multislice multiphase short axis (BH), for systolic biventricular function	6
9	Oblique sagittal single slice scout of the aorta (BH)	1
10	2D flow sequence in ascending aorta and proximal descending aorta (FB), for PWV	3
11	2D flow sequence in descending aorta (FB), for PWV	3
12	2D flow sequence in descending aorta at level of diaphragm (FB), optional	3
Total scan duration time (min)		43–58

BH, breath hold; FB, free breathing; MRA, magnetic resonance angiography; PWV, pulse wave velocity.

progressive dilatation in Marfan patients.^{4 7} The addition of 2D flow MRI at the level of the diaphragm can be used to quantify collateral flow in coarctation patients.^{w5} Recently, flow sensitive four dimensional (4D, time resolved 3D) sequence protocols have been described in which a double-

oblique sagittal 3D volume covers the entire thoracic aorta (figure 5), which may be used to identify pathological aortic flow patterns predisposing to aneurysm development, as well as PWV assessment of even shorter, arbitrarily chosen aortic segments.¹¹

Aortic valve and LV function

Accurate calculation of aortic valve regurgitation (AR) fraction (as a percentage of aortic forward flow) can be performed by quantification of flow at the level of the aortic valve. Velocity encoded MRI also allows for peak flow velocity assessment across stenotic lesions such as aortic valve stenosis or coarctation, and thus estimates peak gradients using the simplified Bernoulli equation. Systolic LV function can be assessed using an SSFP cine sequence in the short axis or axial orientation.

FIVE ENTITIES WITH AORTIC WALL ABNORMALITIES

Marfan syndrome

Marfan syndrome is a heritable connective tissue disorder and is caused by a mutation of the *FBN1* gene on chromosome 15 that codes for fibrillin-1, in the absence of which elastin is more readily degraded by MMPs and smooth muscle cells will dissociate from the medial matrix components.¹ As

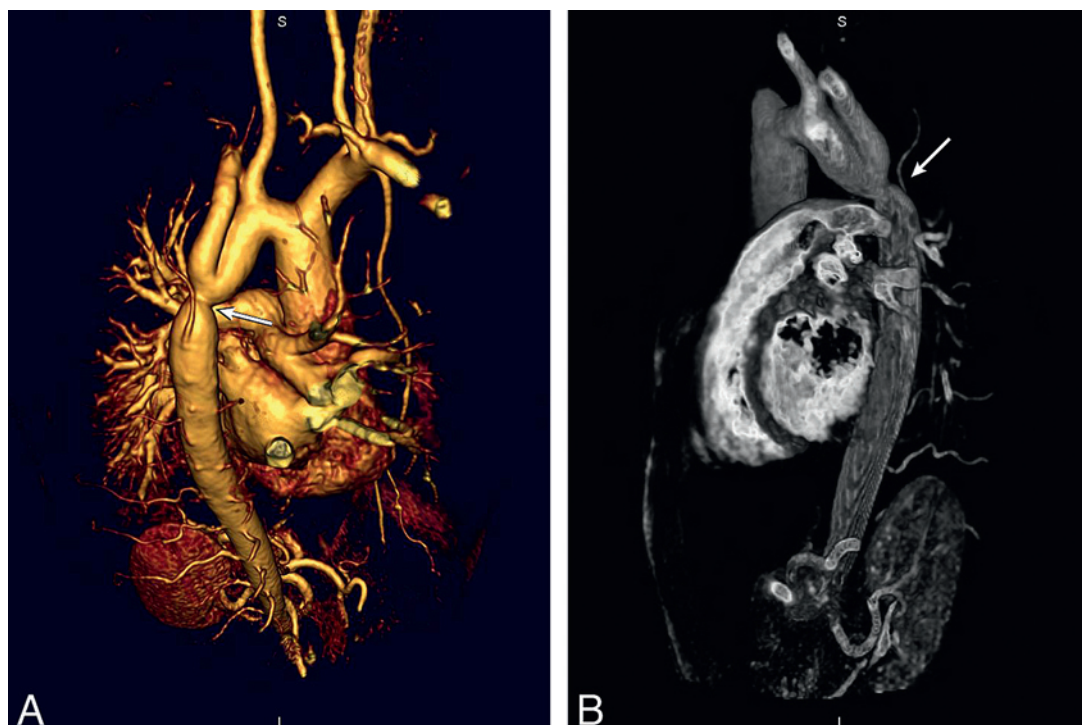


Figure 3 Gadolinium-chelate enhanced MRA images in a 34-year-old man after coarctation repair. Shaded surface full volume rendering display image (A) and selected volume rendering display image (B). Flow speed was significantly increased at the level of stenosis at residual/re-coarctation (arrow, A, B). MRI phase contrast flow volume was 7.2 litres/min as measured immediately distal from the level of the stenosis, and 6.0 litres/min measured at the level of the diaphragm. This indicates flow decrease from the proximal to the distal descending thoracic aorta that excludes haemodynamically significant collateral flow. Note the relative normal size of the intercostal arteries; no major collaterals were observed at MRA. Reproduced with the permission of Elsevier from Ho V, ed. *Cardiovascular imaging*, 2010.

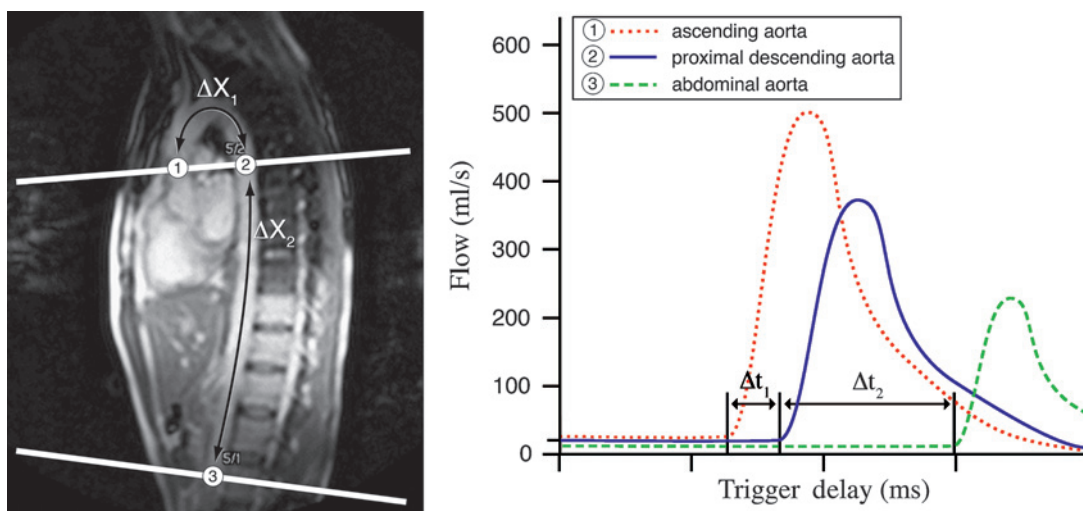


Figure 4 Left panel shows an oblique sagittal scout covering the full course of the aorta, indicating the sites for the pulse wave velocity (PWV) assessment: the ascending aorta (1), the proximal descending aorta just distal to the aortic arch (2), and the most distal level of the abdominal aorta depicted in the oblique sagittal scout (3). Determination of the onset of the three systolic flow waves for PWV at the measurement sites is depicted in the right panel. The distance between these sites and the transit time (Δt_1 and Δt_2) between the individual onsets of the systolic flow waves determine the PWV.

as a result, the aortic wall will exhibit a highly variable degree of aortic medial degeneration with a high risk of progressive aortic dilatation and subsequent aortic dissection or rupture.^{7–10} Numerous reports indicate that Marfan syndrome serves as a prototype for intrinsic aortic wall

pathology and concomitant aortic and LV sequelae in other CHD entities.^{1–3 12 13 w10 w11}

Aortic dilatation is the most common complication in patients with Marfan syndrome (60–80% of adult patients) (figure 6).^{w12} The relative abundance of elastic fibres in the ascending aorta,

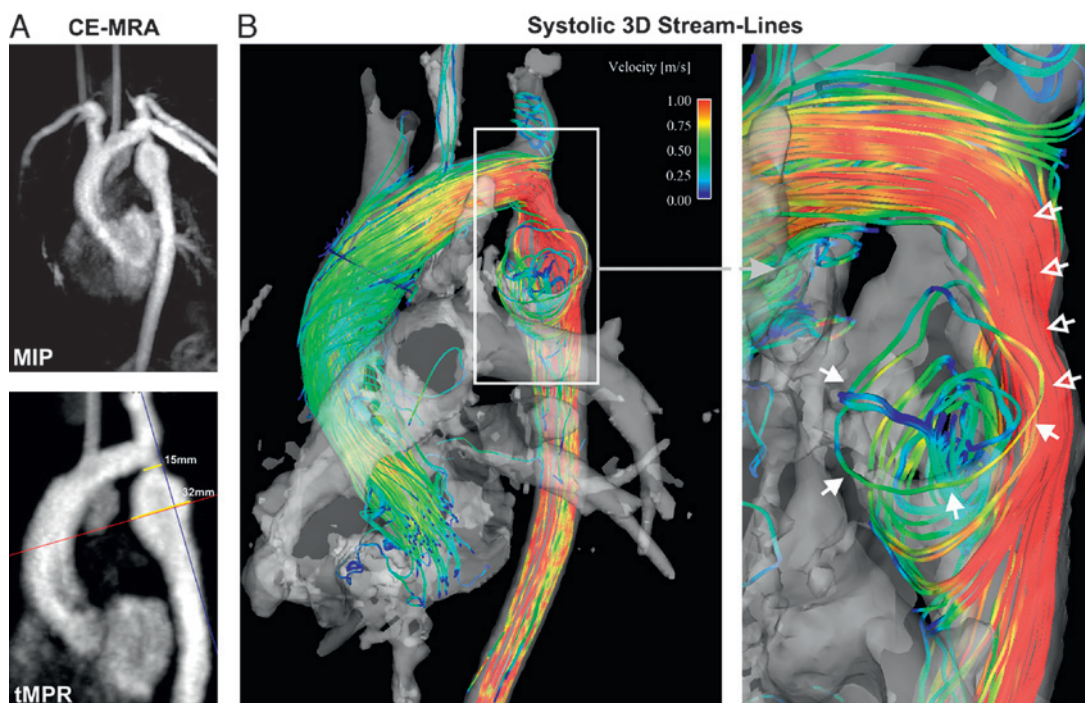


Figure 5 Vascular geometry and systolic flow characteristics as assessed with 4D flow MRI. A shows a maximum intensity projection (MIP) and thick multiplanar reformat (tMPR) showing coarctation and post-stenotic dilatation of the proximal descending aorta. B shows systolic 3D streamlines in the entire thoracic aorta and a magnified region encompassing the pathological site. CE-MRA indicates contrast-enhanced magnetic resonance angiography. Reproduced with the permission of Wolters Kluwer/Lippincott from: Markl M, Arnold R, Hirtler D, *et al.* Three dimensional flow characteristics in aortic coarctation and poststenotic dilatation. *J Comput Assist Tomogr* 2009;**33**:776–8.

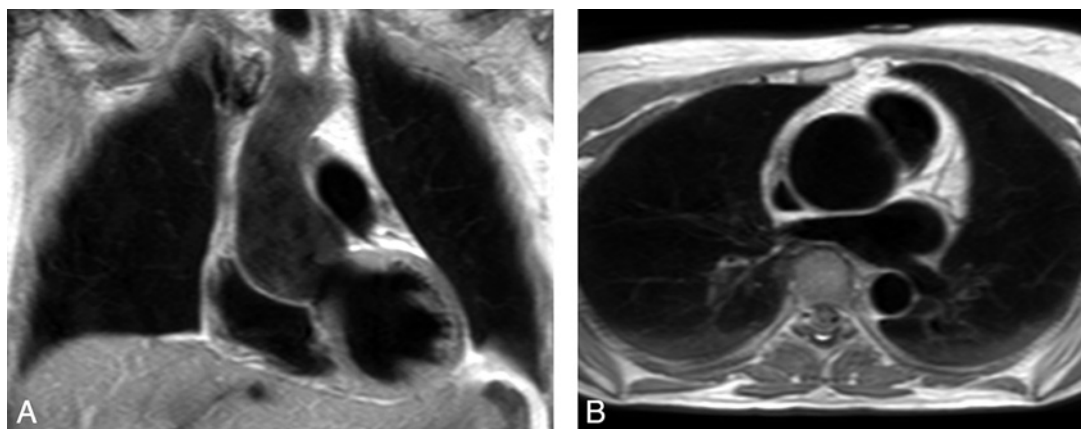


Figure 6 Coronal (A) and axial (B) black blood turbo spin-echo MRI images in a 46-year-old woman with Marfan syndrome, showing a pear shaped aortic root (A) with dilatation up to 5.1 cm. Reproduced with the permission of Elsevier from Ho V, ed. *Cardiovascular imaging*, 2010.

coupled with the repetitive stress of LV ejection, probably account for the aortic dilatation that usually occurs primarily in the aortic root.^{w12} Therefore, the majority of patients present with enlargement of the ascending aorta or a type A dissection; only rare cases present with a type B dissection involving the descending aorta.¹⁰ Replacement of the aortic root has been recommended before the diameter exceeds 5.0–5.5 cm.^{7 10} Independent predictors of progressive aortic dilatation that will prompt the recommendation for surgery when the aorta is <5.0 cm include rapid growth of the aortic diameter (>1 cm/year), a family history of premature aortic dissection (<5 cm), and the presence of greater-than-mild AR.^{7 10} AR may result from distortion of the aortic valve cusps' coaptation by the enlarged aortic root and occurs in 15–44% of patients.¹⁰

Recent MRI reports indicate that reduced aortic elasticity is an independent predictor of progressive aortic dilatation and adverse long term outcome.⁷ As elastin fragmentation in the aortic media is scattered in an irregular pattern along the aorta, regional distensibility as assessed with 2D flow MRI may be sensitive in the detection of regional variations in aortic stiffness.⁷ Recently developed 4D flow MRI appears to be even more advantageous, as it allows for PWV assessment of even shorter, arbitrarily chosen aortic segments at particular risk for progressive dilatation. For

optimal risk stratification, aortic stiffness may be taken into account in combination with aortic dimensions and the previously mentioned clinical predictors of progressive aortic dilatation.^{7 w12} MRI has been recommended for routine assessment of aortic diameters and stiffness, as well as for the follow-up of aortic complications such as intramural haematoma and aortic aneurysms.^{7 10} Evaluation of aortic dilatation should be performed every 6 months to determine the rate of progression, which can be extended to annual evaluation when the aortic size is stable over time.¹⁰ MRI can also be used to adequately monitor the beneficial effect of β -blocker administration on the progression rate of aortic dilatation¹⁰ (box 2).

BAV disease

BAV disease is the most common CHD, occurring in 1–2% of the population.^{w14 w15} BAV is the result of abnormal aortic cusp formation due to inadequate production of fibrillin-1 during valvulogenesis and is likely the result of a complex developmental pathology rather than simply the fusion of two normal cusps.^{3 w13} AR is the most frequent (80%) complication and usually occurs from cusp prolapse, fibrotic retraction or dilation of the sinotubular junction, in many cases requiring aortic valve replacement.³ BAV is also present in the majority of elderly patients with significant aortic valve stenosis, reflecting the propensity for premature fibrosis and calcium deposition.³ Stenotic BAVs create a high velocity jet that increases wall shear stress on the anterolateral portion of the ascending aorta.¹³ Despite the fact that echocardiography remains the routine investigation of choice, MRI is reported to be superior in distinguishing normal and BAVs, as well as for assessment of the dynamic properties of the valve.^{w14}

The vascular complications of BAV are less well understood and are associated with significant morbidity and mortality.^{3 w13} The histology of the ascending aortic wall in patients with BAV shows strong similarities with the fibrillin-1 deficient aortas of Marfan patients¹ (figure 1). As

Box 2 Marfan syndrome

- ▶ Caused by *FBN1* gene mutation.
- ▶ As a consequence, a variable degree of aortic media degeneration is present.
- ▶ High risk for progressive aortic dilatation and dissection.
- ▶ Primarily type A dissection.
- ▶ Independent predictors of aortic dilatation are rapid growth and reduced elasticity.
- ▶ β -blockers may prevent progressive aortic dilatation.
- ▶ Annual MRI is recommended when aortic size is stable over time.

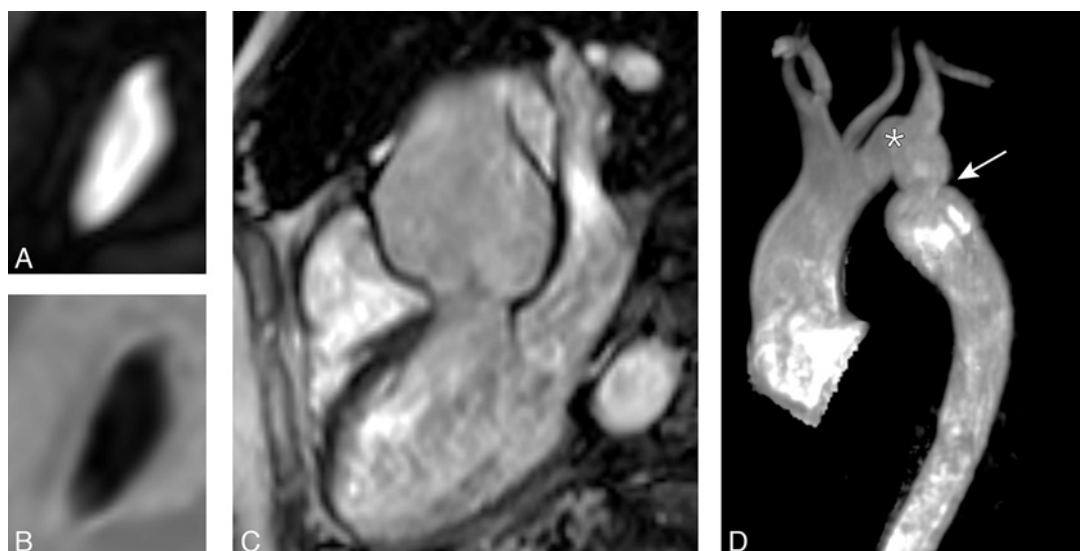


Figure 7 Phase contrast modulus (A) and phase (B) images of the bicuspid aortic valve, gradient echo image of the aortic root (C), and gadolinium-chelate enhanced MRA image of the thoracic aorta (D) in a 54-year-old woman with Turner syndrome. Note the combination of the slit-like bicuspid aortic valve with slit flow (A, B) and post-stenotic dilatation that measured 4.6 cm (C), together with other aortic pathology; aortic kinking (*, D) and pseudo coarctation (arrow, D). Reproduced with the permission of Elsevier from Ho V, ed. *Cardiovascular imaging*, 2010.

a consequence of abnormal aortic wall composition and increased local wall shear stress due to distorted aortic flow patterns, serious complications such as progressive aortic root dilatation (50–60% of all patients with BAV) (figure 7) and/or aneurysm formation may finally result in aortic dissection (5% of all patients with BAV).^{2 w13} Despite this lower incidence of aortic dissection than occurs in Marfan syndrome (40%), BAV is by far the more common aetiology in aortic dissection as Marfan syndrome is a rare entity (0.01% vs 1–2% of patients with BAV).³ In light of the asymptomatic nature of aortic dilatation and the high mortality rate associated with aortic dissection, at least annual follow-up is recommended until the rate of progression of aortic dilatation is determined.² A recent 4D flow MRI study demonstrated specific pathological flow patterns within the aorta in BAV, indicating specific aortic areas with increased wall shear stress that may be at risk of aortic aneurysm formation.¹¹ Aortic root replacement is generally more aggressively recommended for BAV (ie, 4–5 cm) than for those of patients with a tricuspid valve (ie, 5–6 cm).^{2 3} Evaluation of the elastic properties of the ascending aorta by MRI may be

useful to identify patients who are at risk of progressive aortic dilatation analogous to Marfan patients.^{6 7} Increased aortic stiffness as assessed with MRI is also associated with LV hypertrophy in BAV, as a result of increased LV afterload.⁶ As LV hypertrophy may lead to diastolic LV dysfunction and heart failure, it poses a future risk for LV function in BAV.⁶ As many BAV patients will require cardiac surgery during their lifetime, close monitoring of aortic dimensions, aortic elasticity, aortic valve competence, and LV function is mandatory during follow-up, especially when progressive aortic dilatation is present⁶ (box 3).

Coarctation of the aorta

Coarctation of the aorta accounts for 5–10% of all CHD and is associated with a significantly increased cardiovascular morbidity even after successful surgical correction.^{7 14 w3 w15} Structural aortic wall abnormalities with reduced aortic elastic properties proximal and distal to the site of coarctation imply that coarctation is a systemic vascular disease.^{w15} The presence of concomitant BAV in 20–85% of coarctation patients, and the strong histological similarity of aortic wall abnormalities between both entities, are also suggestive of an inherited origin of aortic wall pathology.¹

After initially successful repair, complications may occur such as persistence of hypertension, recoarctation, aortic dilatation, and aneurysm formation (figure 3).^{w15} Persisting resting and exercise induced hypertension have been reported in 10–46% and 30–60% of cases, respectively.^{7 w3} Hypertension is accompanied by an increase in aortic medial collagen and a decrease in smooth muscle that may persist after successful repair.¹ Late hypertension may be caused by functional recoarctation at the site of surgical repair due to

Box 3 Bicuspid aortic valve disease

- ▶ Most common congenital heart disease entity.
- ▶ Similar histological aortic wall abnormalities as in Marfan syndrome.
- ▶ Frequent aortic valve stenosis and/or insufficiency.
- ▶ Aortic wall abnormalities are associated with aortic root dilatation and even dissection.
- ▶ Reduced aortic elasticity may predict (progressive) aortic dilatation.
- ▶ Reduced aortic elasticity leads to increased LV afterload and LV dysfunction.

decreased distensibility.^{w16} Aortic arch geometry as assessed by MRA also appears to be a strong predictor of sequelae: a so-called 'gothic arch' with a triangular shape of the aortic arch is associated with vascular thickening (figure 3), reduced aortic distensibility, and late hypertension when compared to a 'Romanesque' or rounded aortic arch.¹⁵ Impaired LV function, increased LV mass, and concomitant adverse LV remodelling even in postoperative patients with normal blood pressure can be attributed to reduced aortic elastic properties, resulting in increased LV afterload.^{w17}

Adults after surgical repair of coarctation should be closely monitored for detection of recoarctation (7–60% during follow-up), progressive aortic dilatation, and even dissection.^{w15} Risk factors for these long term aortic complications are the presence of BAV, advanced age, and hypertension.² Systematic MRI screening has been proposed as an effective approach for the follow-up with early detection of recoarctation.^{w2 w9} 2D spin echo MRI is essential to detect abnormalities of the aortic wall abnormalities, while 3D MRA provides a highly accurate view of the entire reconstructed aorta, obviating the need for invasive x-ray angiography by catheterisation for planning of treatment (figure 3).¹⁴ A combination of narrowest aortic cross-sectional area and mean flow deceleration in the descending aorta obtained by MRI is able to predict a catheterisation peak-to-peak gradient ≥ 20 mm Hg, considered to be the reference standard for haemodynamic severity of (re-)coarctation.¹⁴ 2D and 4D flow MRI are also useful to determine the presence and haemodynamics of collateral vessels (which maintain distal aortic perfusion) and to identify pathological flow patterns across the site of coarctation (figure 5), respectively.^{11 w5} Finally, promising utilities are MRI guided balloon angioplasty and MRI guided deployment of stents to repair aortic coarctation with a significant reduction of radiation exposure^{w18} (box 4).

ToF

ToF is the most commonly encountered cyanotic CHD entity.¹⁶ Patients after repair frequently encounter longstanding pulmonary regurgitation as a result of right ventricular (RV) outflow tract reconstruction and associated impaired RV func-

tion after repair.¹⁶ LV dysfunction is due to a multifactorial process which is explained by preoperative cyanosis, perioperative sequelae, and adverse RV–LV interaction: longstanding pulmonary regurgitation will lead to RV dilatation, which is associated with increased LV end-systolic volumes and impaired septal contractility, which have an adverse effect on LV systolic performance.⁵

Progressive aortic root dilatation has frequently been described after ToF repair (15–88%).^{12 16} Two hypotheses have been postulated: increased blood flow from both ventricles to the overriding aorta before surgical repair may result in increased stress on the aortic wall,^{1 12 16} while strong histological similarities of the aortic media as in Marfan syndrome have been described (although direct linkage to gene mutation(s) encoding for fibrillin-1 has not been cleared yet).^{1 16 w10} Whether aortic wall pathology results from an intrinsic medial abnormality inherent to ToF itself or is secondary to the antecedent volume load through the aorta before repair (or perhaps a combination of the two) remains difficult to determine.⁵

The potential for complications of aortic root dilatation that may necessitate surgical intervention is increasingly recognised.¹⁶ A recent study reported the progressive nature of aortic dilatation in ToF, as aortic dilatation increased at a rate of 1.7 mm/year, in contrast to 0.03 mm/year in healthy controls.¹² Aortic dissection late after ToF repair in adults whose aortic roots exceeded 6 cm indicate that monitoring of aortic dimensions is mandatory, especially when a (progressively) dilated ascending aorta is present.^{2 16 w19} Aortic root surgery may be considered in the case of progressive AR and aortic root dilatation exceeding 5.5 cm, particularly when the primary indication for surgery is pulmonary valve replacement and both procedures may be combined.^{w20} Aortic wall pathology also initiates a cascade of events, with aortic dilatation and increased aortic stiffness leading to mild degrees of AR (15–50%), while the subsequent LV volume overload is an independent risk factor for LV systolic dysfunction.⁵ Augmented aortic wave reflections and increased aortic and peripheral arterial stiffness have also been reported, contributing to pulsatile load on the LV and adversely affecting LV ejection.¹⁷ Aortic wall pathology may therefore represent a separate mechanism leading to AR and LV dysfunction, in addition to adverse RV–LV interaction.⁵ As LV dysfunction is considered to be a strong predictor of impaired clinical status and the occurrence of major adverse events in ToF, aortic sequelae may be of significant prognostic value.^{w21} Close monitoring of the aortic–LV cascade using MRI is therefore recommended during follow-up and should be part of already routine MRI of RV function in patients with ToF^{5 w10} (box 5).

TGA

The arterial switch operation (ASO) has become the preferred method of surgery for TGA.^{18 w22} Despite a significant reduction of sequelae,

Box 4 Coarctation of the aorta

- ▶ Frequent structural aortic wall abnormalities.
- ▶ Complicated by persisting hypertension, recoarctation, aortic dilatation, and aneurysm formation.
- ▶ Aortic arch geometry (especially so called 'Gothic' or triangular shaped aortic arch) is a strong predictor of aortic sequelae.
- ▶ Systematic MRI screening allows for early detection of recoarctation.
- ▶ 3D MRA may obviate invasive x-ray angiography.
- ▶ 2D and 4D flow MRI identifies pathological aortic flow patterns and collateral haemodynamics.

Box 5 Tetralogy of Fallot

- ▶ Most common cyanotic congenital heart disease entity.
- ▶ Associated with frequent aortic wall pathology due to volume overload before surgical repair and/or intrinsic aortic wall abnormalities.
- ▶ Concomitant frequent aortic dilatation, loss of aortic elasticity, and even aortic dissection.
- ▶ Aortic wall pathology negatively affects aortic valve and LV function.
- ▶ MRI of aortic—LV cascade should be part of already routine RV function assessment.

Box 6 Transposition of the great arteries

- ▶ Arterial switch operation preferred method of surgery.
- ▶ Frequent aortic wall pathology due to abnormal aorticopulmonary septation, damage to vasa vasorum, and surgical manipulations.
- ▶ Consequent frequent aortic dilatation and aortic valve insufficiency.
- ▶ Concomitant LV volume overload leads to LV dysfunction.
- ▶ Long term outcome not available yet, as arterial switch operation is a relatively new technique of choice.
- ▶ MRI is useful in depiction of aortic sequelae, coronary artery imaging, and LV function assessment.

completion of the ASO may still predispose patients to complications such as LV impairment due to disturbed reimplantation of the coronary arteries, aortic root dilatation, and AR.^{18 19} MRI has proven to be useful in depicting aortic sequelae and coronary artery imaging.^{2 19 20}

Intrinsic aortic wall pathology in TGA has been reported due to abnormal aorticopulmonary septation, damage to the vasa vasorum and surgical manipulations during the ASO, predisposing to

aortic dilatation, aneurysm formation, and even aortic dissection.^{w22 w23} Aortic distensibility may be reduced by impaired aortic elastogenesis as well as by scar formation at the site of anastomosis.^{w22} High grade medial abnormalities in the ascending aorta have already been observed during the neonatal period, suggesting that they are inherited analogously to Marfan syndrome.^{1 w22} Others concluded that aortic wall abnormalities develop due to structural wall differences between the two great arteries, as the former pulmonary arterial wall is exposed to higher systemic pressures after ASO, posing increased stress on the neo-aortic wall.^{w23} Whether medial abnormalities are inherent or acquired therefore remains difficult to distinguish.¹

A high incidence of AR has been reported after ASO (30% at 6 years after ASO) and is probably the result of a multifactorial process for which aortic root geometry, surgical techniques, and preoperative size discrepancy between the two great arteries are involved.^{w24} In addition, AR appears to be functionally correlated with aortic root dilatation and reduced elasticity of the proximal aorta.¹⁸ A recent MRI report described not only frequent aortic root dilatation and AR, but also a cascade of events ultimately leading to LV systolic dysfunction after ASO.¹⁸ Frequent aortic root dilatation and reduced proximal aortic elasticity were associated with the degree of AR, while the concomitant LV volume overload leads to increased LV dimensions and subsequent decreased LV ejection fraction. Therefore, LV systolic dysfunction as the end point in a sequence of events poses a prognostic risk for ASO patients, on top of potential negative perioperative events and disturbed coronary artery reimplantation.¹⁸ Further elucidation of the underlying pathogenic substrate of aortic wall abnormalities and its clinical repercussions after ASO is, however, required considering the relatively new surgical technique of choice, clearly warranting life-long follow-up of aortic and LV parameters¹⁸ (box 6).

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CONCLUSIONS

In this report, we reviewed the five most common entities of inherited connective tissue disorders and classical CHD with intrinsic aortic wall abnormalities, and the potential role of MRI in their clinical evaluation and management. Aortic wall abnormalities are not only limited to a prototypical extreme such as Marfan syndrome, but are also present in a wide range of other CHD entities, each with its own pathogenic substrate and clinical repercussions. Using MRI for assessment of aortic dimensions and elasticity, aortic valve competence, and LV function enables the accurate monitoring of aortic and LV conditions, the effect of interventional measures such as β -blocker administration, and the improved selection of patients who may benefit from surgical treatment.

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