CME CREDIT

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Thoracic aortic aneurysm: How to counsel, when to refer

ABSTRACT

Thoracic aortic aneurysm (TAA) is usually clinically silent and progresses slowly until a tipping point is reached, after which the aortic diameter can expand more rapidly and the condition can potentially end in aortic dissection or rupture. Causes include bicuspid aortic valve and genetic syndromes (Marfan, Loeys-Dietz, and Ehlers-Danlos syndromes) and familial associations, but many cases are idiopathic. Clinicians should therefore be alert for clues on chest imaging, and consider screening in first-degree relatives of patients known to have aortic disease. Early referral to a cardiologist specializing in aortic disease is key.

KEY POINTS

Screening and referral depend on clinical context. A size-based model to determine screening, referral, follow-up, and management serves most cases but should be modified in the context of connective tissue disease or family history of aneurysm and dissection.

Medical management involves strict blood pressure and heart rate control with beta-blockers and angiotensin-converting enzyme inhibitors or angiotensin II receptor blockers. Activity modifications should be tailored to the individual, although extreme isometric exercises and heavy lifting should be discouraged.

Patients with TAA should be followed up annually, unless the patient is presenting for initial evaluation or significant changes are seen with dedicated imaging.

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THORACIC AORTIC ANEURYSM (TAA) needs to be detected, monitored, and managed in a timely manner to prevent a serious consequence such as acute dissection or rupture. But only about 5% of patients experience symptoms before an acute event occurs, and for the other 95% the first "symptom" is often death.[1] Most cases are detected either incidentally with echocardiography, computed tomography (CT), or magnetic resonance imaging (MRI) during workup for another condition. Patients may also be diagnosed during workup of a murmur or after a family member is found to have an aneurysm. Therefore, its true incidence is difficult to determine.[2]

With these facts in mind, how would you manage the following 2 cases?

Case 1: Bicuspid aortic valve, ascending aortic aneurysm

A 45-year-old man with stage 1 hypertension presents for evaluation of a bicuspid aortic valve and ascending aortic aneurysm. He has several first-degree relatives with similar conditions, and his brother recently underwent elective aortic repair. At the urging of his primary care physician, he underwent screening echocardiography, which demonstrated a "dilated root and ascending aorta" 4.6 cm in diameter. He presents today to discuss management options and how the aneurysm could affect his everyday life.

Case 2: Marfan syndrome in a young woman

A 24-year-old woman with Marfan syndrome diagnosed in adolescence presents for annual follow-up. She has many family members with the same condition, and several have undergone prophylactic aortic root repair. Her aortic root has been monitored annually for progression of dilation, and today it is 4.6 cm in diam-

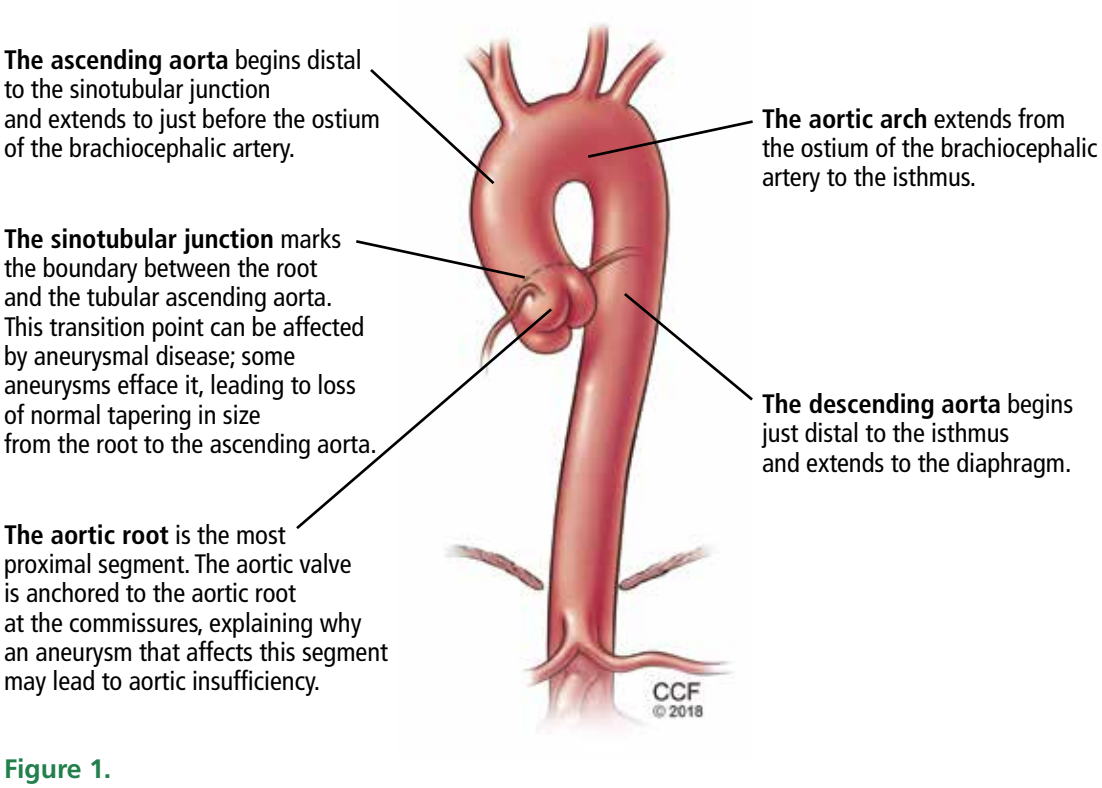


Figure 1.

Aortic dissection can often be mistaken for an acute myocardial infarction or other acute event

eter, a 3-mm increase from the last measurement. She has grade 2+ aortic insufficiency (on a scale of 1+ to 4+) based on echocardiography, but she has no symptoms. She is curious about what size her aortic root will need to reach for surgery to be considered.

LIKELY UNDERDETECTED

TAA is being detected more often than in the past thanks to better detection methods and heightened awareness among physicians and patients. While an incidence rate of 10.4 per 100,000 patient-years is often cited,[3] this figure likely underestimates the true incidence of this clinically silent condition. The most robust data come from studies based on in-hospital diagnostic codes coupled with data from autopsies for out-of-hospital deaths.

Olsson et al,[4] in a 2016 study in Sweden, found the incidence of TAA and aortic dissection to be 16.3 per 100,000 per year for men and 9.1 per 100,000 per year for women.

Clouse et al[5] reported the incidence of thoracic aortic dissection as 3.5 per 100,000 patient-years, and the same figure for thoracic aortic rupture.

Aneurysmal disease accounts for 52,000 deaths per year in the United States, making it the 19th most common cause of death.[6] These figures are likely lower than the true mortality rate for this condition, given that aortic dissection is often mistaken for acute myocardial infarction or other acute event if an autopsy is not done to confirm the cause of death.[7]

RISK FACTORS FOR THORACIC AORTIC ANEURYSM

Risk factors for TAA include genetic conditions that lead to aortic medial weakness or destruction such as Loeys-Dietz syndrome and Marfan syndrome.[2] In addition, family history is important even in the absence of known genetic mutations. Other risk factors include conditions that increase aortic wall stress, such as hypertension, cocaine abuse, extreme weightlifting, trauma, and aortic coarctation.[2]

DIAMETER INCREASES WITH AGE, BODY SURFACE AREA

The thoracic aorta consists of the root and the ascending, arch, and descending segments (**Figure 1**); the abdominal aorta consists of

TABLE 1

Aortic diameters: Upper limits of normal[a]

		Ascending aorta (mm)		Descending aorta (mm)	
Age (years)	BSA (m^2)	Women (n = 1,147)	Men (n = 1,805)	Women (n = 736)	Men (n = 1,195)
< 45	< 1.70	33.8	33.0	23.0	NA
	1.70–1.89	34.4	36.3	24.6	26.6
	1.90–2.09	35.0	36.3	22.7	26.7
	> 2.1	NA	38.3	NA	28.3
45–54	< 1.70	35.2	38.6	24.3	24.2
	1.70–1.89	37.2	38.1	25.4	27.5
	1.90–2.09	38.9	39.7	27.2	29.2
	> 2.1	40.6	40.6	28.3	29.6
55–64	< 1.70	36.9	36.3	25.9	26.1
	1.70–1.89	37.0	39.7	27.1	28.6
	1.90–2.09	39.0	41.2	27.8	29.9
	> 2.1	42.0	43.1	31.7	31.6
≥ 65	< 1.70	37.5	38.5	27.0	NA
	1.70–1.89	39.2	41.0	27.4	32.4
	1.90–2.09	42.7	42.2	29.0	31.0
	> 2.1	NA	42.4	29.8	32.5

[a]Upper limits of normal are 2 standard deviations above the mean. Not calculated if there were fewer than 6 patients in a group.
BSA = body surface area; NA = not available

Information from Wolak A, Gransar H, Thomson LE, et al. Aortic size assessment by noncontrast cardiac computed tomography: normal limits by age, gender, and body surface area. J Am Coll Cardiol Cardiovasc Imaging 2008; 1(2):200–209. doi:10.1016/j.jcmg.2007.11.005

the suprarenal and infrarenal segments.[8,9] These divisions are useful, as aneurysmal disease can be confined to specific locations along the length of the vessel, and the location can affect the clinical presentation and management decisions and lend insight into the pathogenesis.

Normal dimensions for the aortic segments differ depending on age, sex, and body surface area.[8,44,45] The size of the aortic root may also vary depending on how it is measured, due to the root's trefoil shape. Measured sinus to sinus, the root is larger than when measured sinus to commissure on CT angiography or cardiac MRI. It is also larger when measured leading edge to leading edge than inner edge to inner edge on echocardiography.[10]

TAA is defined as an aortic diameter at least 50% greater than the upper limit of normal.[8]

The aorta increases in diameter by 0.7 to 1.9 mm per year if not dilated, and larger-diameter aortas grow faster.[11] In addition, men have a larger aortic diameter than women.[10] Size-based criteria and indices are useful for defining and monitoring aneurysmal progression, since larger patients tend to have a larger aorta.[10] **Table 1** lists upper limits of normal

Depending on the initial size of the TAA, refer patients to a cardiologist with expertise in aortic disease or to an aortic surgeon

TABLE 2

Common causes of thoracic aortic aneurysm

Idiopathic

Bicuspid aortic valve

Genetic syndromes
Marfan syndrome
Loeys-Dietz syndrome
Ehlers-Danlos syndrome
Familial thoracic aortic aneurysm and dissection
Autosomal dominant polycystic kidney disease
Turner syndrome

Inflammatory vasculitis
Takayasu arteritis
Giant cell arteritis
Behçet arteritis
Ankylosing spondylitis

Other
Atherosclerosis
Syphilis
Previous dissection
Infectious aortitis
Trauma

Risk factors
Hypertension
Smoking
Advanced age

Beta-blockers are recommended for patients with TAA

values for the ascending and descending aorta by age, sex, and body surface area obtained by Wolak et al in a study using noncontrast CT.[10]

Geometric changes in the curvature of the ascending aorta, aortic arch, and descending thoracic aorta can occur as the result of hypertension, atherosclerosis, or connective tissue disease.

HOW IS TAA DIAGNOSED?

TAA is asymptomatic in most patients and is usually detected on imaging. However, it should be actively looked for in patients who have a family history of Marfan, Loeys-Dietz, or Ehlers-Danlos syndrome or a family history of aortic aneurysm or dissection (not necessarily in a first-degree relative, but more significant in a first-degree relative or in multiple family members across generations), and in patients with a bicuspid aortic valve or autoimmune disease such as Takayasu or giant cell arteritis (**Table 2**). **Table 3** lists the common genetic disorders with their associated mutations and clinical features.

Some patients present with chest pain that may be related to local compression due to the aorta's large size. Hoarseness, dysphagia, or chronic cough may be a presenting symptom, particularly in patients with descending aortic aneurysm or congenital aortic anomaly.

An abnormal chest radiograph with a prominent aortic shadow or mediastinal widening should prompt further evaluation for TAA. In addition, patients with known abdominal aortic aneurysm should have the rest of the aorta imaged as well to rule out associated TAA.

Imaging tests

TAA can be diagnosed with several imaging tests, each with advantages and disadvantages (**Table 4**).[12] Most commonly used in its diagnosis and follow-up are transthoracic echocardiography (**Figure 2**), cardiac-gated computed tomographic angiography (CTA), and MRI (**Figure 3**). Transesophageal echocardiography is not routinely used in diagnosing TAA but is invaluable during surgery to assess aortic valve function and immediate results of aortic repair.

Which imaging test to use depends on the clinical context as well as the availability and expertise at each institution. For example, screening of first-degree relatives of a patient with thoracic aortic disease typically begins with transthoracic echocardiography and can be escalated to CTA or MRI if an abnormality is detected. Alternatively, patients with connective tissue disease with a particularly severe vascular phenotype such as Loeys-Dietz syndrome should undergo screening with dedicated aortic imaging such as CTA, since this disease can affect the entire aorta and its branch vessels.

The aortic diameter should be measured perpendicularly to the centerline of flow, which is now easier with dedicated aortic imaging technology and widely available, user-friendly, 3-dimensional reconstruction software (**Figure 4**).[2]

It is particularly important to obtain a gated CTA image in patients with aortic root aneurysm to avoid motion artifact and possible erroneous measurements. Gated CTA is done with electrocardiographic synchronization

TABLE 3

Inherited connective tissue diseases and thoracic aortic aneurysm

Inherited condition	Implicated gene	Clinical features
Marfan syndrome	*FBN1*	Aneurysm of aortic root, dilation of pulmonary artery, and aortic dissection
Ehlers-Danlos syndrome	*COL5A1, COL5A2, COL3A1*	Arterial mid-sized rupture, especially involving thoracic vasculature
Loeys-Dietz syndrome	*TGFBR1, TGFBR2, SMAD3, TGFB2, TGFB3*	Premature and aggressive aneurysm and dissection; aneurysm may involve aortic segments other than the root
Familial thoracic aortic aneurysm and dissection	*ACTA2, MYH11, PRKG1, MYLK, TGFBR2*	Thoracic aortic aneurysm and dissection; associated vascular disease (eg, patent ductus arteriosus)
Bicuspid aortic valve	Unknown (may be associated with *ACTA2, MYH11*, syndromic connective tissue diseases)	Aortic dilation typically involving the aortic root and ascending aorta
Autosomal dominant polycystic kidney disease	*PKD1, PKD2*	Dilation of the aorta and thoracic aortic dissection
Turner syndrome	45,X	Thoracic aortic aneurysms and dissections, bicuspid aortic valve, aortic coarctation

Reprinted from Cury M, Zeidan F, Lobato AC. Aortic disease in the young: genetic aneurysm syndromes, connective tissue disorders, and familial aortic aneurysms and dissections. Int J Vasc Med 2013(2013); 2013:267215. doi:10.1155/2013/267215

Our blood pressure goal is < 130/80 mm Hg; heart rate goal is ≤ 70

and allows for image processing to correct for cardiac motion.

HOW IS TAA CLASSIFIED?

TAA can be caused by a variety of inherited and sporadic conditions. These differences in pathogenesis lend themselves to classification of aneurysms into groups. **Table 3** highlights the most common conditions associated with TAA.[13]

Bicuspid aortic valve aortopathy

From 1% to 2% of people have a bicuspid aortic valve, with a 3-to-1 male predominance.[14,15] Aortic dilation occurs in 35% to 80% of people who have a bicuspid aortic valve, conferring a risk of dissection 8 times higher than in the general population.[16–18]

The pathogenic mechanisms that lead to this condition are widely debated, although a combination of genetic defects leading to intrinsic weakening of the aortic wall and hemodynamic effects likely contribute.[19] Evidence of hemodynamic contributions to aortic dilation comes from findings that particular patterns of cusp fusion of the bicuspid aortic valve result in changes in transvalvular flow, placing more stress on specific regions of the ascending aorta.[20,21] These hemodynamic alterations result in patterns of aortic dilation that depend on cusp fusion and the presence of valvular disease.

Multiple small studies found that replacing bicuspid aortic valves reduced the rate of aortic dilation, suggesting that hemodynamic factors may play a larger role than intrinsic wall properties in genetically susceptible individuals.[22,23] However, larger studies are needed before any definitive conclusions can be made.

TABLE 4

Imaging studies for aortic aneurysm

Imaging study	Advantages	Disadvantages
Cardiac-gated multidetector computed tomographic angiography	Highly specific and sensitive Can diagnose vascular anomalies, tortuosity	Large doses of ionizing radiation and contrast
Magnetic resonance angiography	Highly specific and sensitive Can be accurate without using contrast Ability to assess valve disease	Prolonged scanning time Difficult for those with claustrophobia
Transthoracic echocardiography	Noninvasive, readily available Can assess valve disease	Limited visualization of ascending aorta Accuracy reduced with "oblique" measurements
Transesophageal echocardiography	Highly specific and sensitive Ability to assess valve disease	Requires skilled personnel Invasive, requires procedural sedation
Angiography	Highly sensitive and specific	Invasive Requires contrast

Based on Smith AD, Schoenhagen P. CT imaging for acute aortic syndrome. Cleve Clin J Med 2008; 75(1):7–17. pmid:18236724

Heavy lifting can increase blood pressure for short periods and, in theory, initiate dissection or rupture

HOW IS ANEURYSM MANAGED ON AN OUTPATIENT BASIS?

Patients with a new diagnosis of TAA should be referred to a cardiologist with expertise in managing aortic disease or to a cardiac surgeon specializing in aortic surgery, depending on the initial size of the aneurysm.

Control blood pressure with beta-blockers

Medical management for patients with TAA has historically been limited to strict blood pressure control aimed at reducing aortic wall stress, mainly with beta-blockers.

Are angiotensin II receptor blockers (ARBs) beneficial? Studies in a mouse model of Marfan syndrome revealed that the ARB losartan attenuated aortic root growth.[24] The results of early, small studies in humans were promising,[25–27] but larger randomized trials have shown no advantage of losartan over beta-blockers in slowing aortic root growth.[28] These negative results led many to question the effectiveness of losartan, although some point out that no studies have shown even beta-blockers to be beneficial in reducing the clinical end points of death or dissection.[29] On the other hand, patients with certain *FBN1* mutations respond more readily than others to losartan.[30] Additional clinical trials of ARBs in Marfan syndrome are ongoing.

Current guidelines recommend stringent blood pressure control and smoking cessation for patients with a small aneurysm not requiring surgery and for those who are considered unsuitable for surgical or percutaneous intervention (level of evidence C, the lowest).[2] For patients with TAA, it is considered reasonable to give beta-blockers. Angiotensin-converting enzyme inhibitors or ARBs may be used in combination with beta-blockers, titrated to the lowest tolerable blood pressure without adverse effects (level of evidence B).[2]

The recommended target blood pressure is less than 140/90 mm Hg, or 130/80 mm Hg in those with diabetes or chronic kidney disease (level of evidence B).[2] However, we recommend more stringent blood pressure control: ie, less than 130/80 mm Hg for all patients with aortic aneurysm and a heart rate goal of 70 beats per minute or less, as tolerated.

Activity restriction

Activity restrictions for patients with TAA are largely based on theory, and certain activities may require more modification than others. For example, heavy lifting should be discouraged, as it may increase blood pressure significantly for short periods of time.[2,31] The increased wall stress, in theory, could initiate dissection or rupture. However, moderate-intensity aerobic activity is rarely associated with significant elevations in blood pressure and should be encouraged. Stressful emotional states have been anecdotally associated with aortic dissection; thus, measures to reduce stress may offer some benefit.[31]

Our recommendations. While there are no published guidelines regarding activity restrictions in patients with TAA, we use a graded approach based on aortic diameter:

- 4.0 to 4.4 cm—lift no more than 75 pounds
- 4.5 to 5 cm—lift no more than 50 pounds
- > 5 cm—lift no more than 25 pounds.

We also recommend not lifting anything heavier than half of one's body weight and to avoid breath-holding or performing the Valsalva maneuver while lifting. Although these recommendations are somewhat arbitrary, based on theory and a large clinical experience at our aortic center, they seem reasonable and practical.

Activity restrictions should be stringent and individualized in patients with Marfan, Loeys-Dietz, or Ehlers-Danlos syndrome due to increased risk of dissection or rupture even if the aorta is normal in size.

We sometimes recommend exercise stress testing to assess the heart rate and blood pressure response to exercise, and we are developing research protocols to help tailor activity recommendations.

WHEN SHOULD A PATIENT BE REFERRED?

To a cardiologist at the time of diagnosis

As soon as TAA is diagnosed, the patient should be referred to a cardiologist who has special interest in aortic disease. This will allow for appropriate and timely decisions about medical management, imaging, follow-up, and referral to surgery. Additional recommendations for screening of family members and referral to clinical geneticists can be discussed at this juncture. Activity restrictions should be reviewed at the initial evaluation.

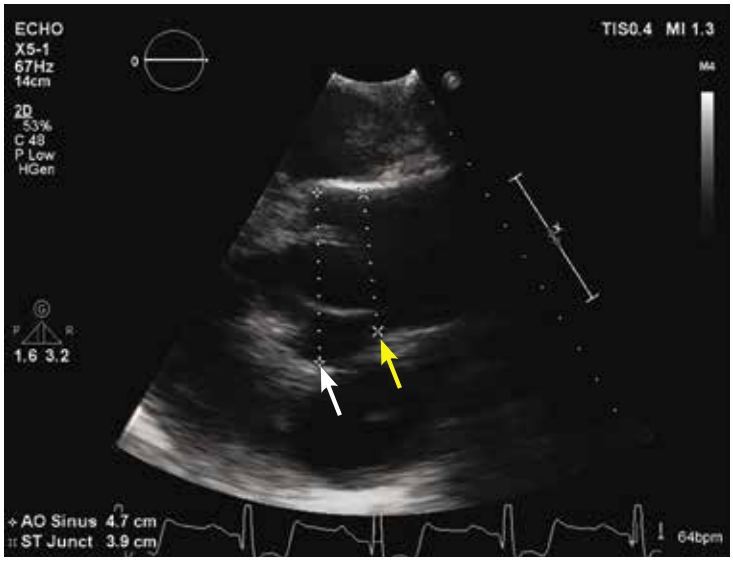


Figure 2. Echocardiographic image of an aneurysmal aortic root (white arrow) that tapers to normal dimensions at the sinotubular junction (yellow arrow) and ascending aorta. See videos (this image and a bicuspid aortic valve).

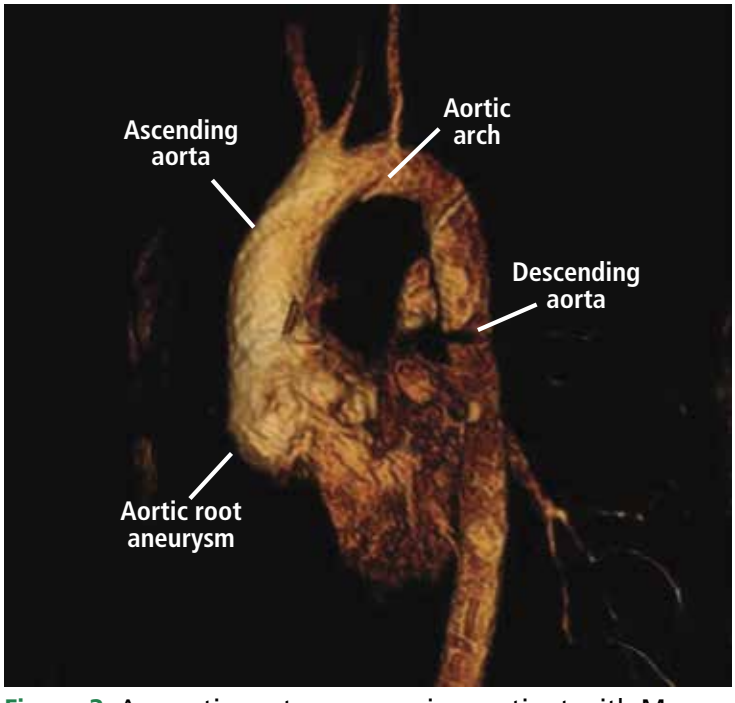
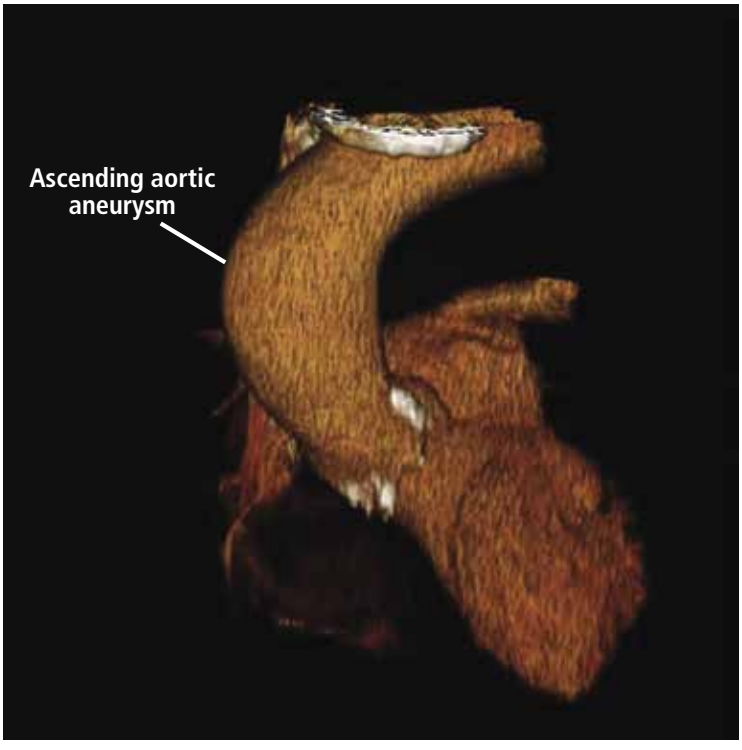


Figure 3. An aortic root aneurysm in a patient with Marfan syndrome using magnetic resonance angiography. See videos (this image, follow-up after surgical repair, and a computed tomographic study).



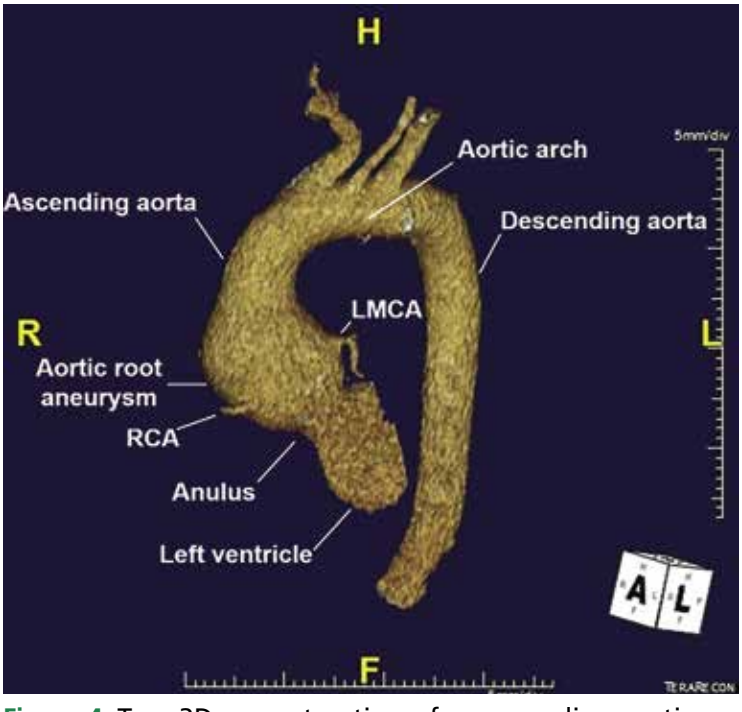


Figure 4. Top, 3D reconstruction of an ascending aortic aneurysm in a patient with a bicuspid aortic valve. See video. Bottom, 3D reconstruction of a young patient with an aortic root aneurysm. Note the tapering from the aneurysmal aortic root to the normal-sized ascending aorta. LMCA = left main coronary artery; RCA = right coronary artery.

To a surgeon relatively early

Size thresholds for surgical intervention are discussed below, but one should not wait until these thresholds are reached to send the patient for surgical consultation. It is beneficial to the state of mind of a potential surgical candidate to have early discussions pertaining to the types of operations available, their outcomes, and associated risks and benefits. If a patient's aortic size remains stable over time, he or she may be followed by the cardiologist until significant size or growth has been documented, at which time the patient and surgeon can reconvene to discuss options for definitive treatment.

To a clinical geneticist

If 1 or more first-degree relatives of a patient with TAA or dissection are found to have aneurysmal disease, referral to a clinical geneticist is very important for genetic testing of multiple genes that have been implicated in thoracic aortic aneurysm and dissection.

WHEN SHOULD TAA BE REPAIRED?

Surgery to prevent rupture or dissection remains the definitive treatment of TAA when size thresholds are reached, and symptomatic aneurysm should be operated on regardless of the size. However, rarely are thoracic aneurysms symptomatic unless they rupture or dissect. The size criteria are based on underlying genetic etiology if known and on the behavior and natural course of TAA.

Size and other factors

Treatment should be tailored to the patient's clinical scenario, family history, and estimated risk of rupture or dissection, balanced against the individual center's outcomes of elective aortic replacement.[32] For example, young and otherwise healthy patients with TAA and a family history of aortic dissection (who may be more likely to have connective tissue disorders such as Marfan syndrome, Loeys-Dietz syndrome, or vascular Ehler-Danlos syndrome) may elect to undergo repair when the aneurysm reaches or nearly reaches the diameter of that of the family member's aorta when dissection occurred.[2] On the other hand, TAA of degenerative etiology (eg, related to smoking or hypertension) measuring less than 5.5 cm

in an older patient with comorbidities poses a lower risk of a catastrophic event such as dissection or rupture than the risk of surgery.[11]

Thresholds for surgery. Once the diameter of the ascending aorta reaches 6 cm, the likelihood of an acute dissection is 31%.[11] A similar threshold is reached for the descending aorta at a size of 7 cm.[11] Therefore, to avoid high-risk emergency surgery on an acutely dissected aorta, surgery on an ascending aortic aneurysm of degenerative etiology is usually suggested when the aneurysm reaches 5.5 cm or a documented growth rate greater than 0.5 cm/year.[2,33]

Additionally, in patients already undergoing surgery for valvular or coronary disease, prophylactic aortic replacement is recommended if the ascending aorta is larger than 4.5 cm. The threshold for intervention is lower in patients with connective tissue disease (> 5.0 cm for Marfan syndrome, 4.4–4.6 cm for Loeys-Dietz syndrome).[2,33]

Observational studies suggest that the risk of aortic complications in patients with bicuspid aortic valve aortopathy is low overall, though significantly greater than in the general population.[18,34,35] These findings led to changes in the 2014 American College of Cardiology/American Heart Association guidelines on valvular heart disease,[36] suggesting a surgical threshold of 5.5 cm in the absence of significant valve disease or family history of dissection of an aorta of smaller diameter.

A 2015 study of dissection risk in patients with bicuspid aortic valve aortopathy by our group found a dramatic increase in risk of aortic dissection for ascending aortic diameters greater than 5.3 cm, and a gradual increase in risk for aortic root diameters greater than 5.0 cm.[37] In addition, a near-constant 3% to 4% risk of dissection was present for aortic diameters ranging from 4.7 cm to 5.0 cm, revealing that watchful waiting carries its own inherent risks.[37] In our surgical experience with this population, the hospital mortality rate and risk of stroke from aortic surgery were 0.25% and 0.75%, respectively.[37] Thus, the decision to operate for aortic aneurysm in the setting of a bicuspid aortic valve should take into account patient-specific factors and institutional outcomes.

A statement of clarification in the American College of Cardiology/American Heart Association guidelines was published in 2015, recommending surgery for patients with an aortic diameter of 5.0 cm or greater if the patient is at low risk and the surgery is performed by an experienced surgical team at a center with established surgical expertise in this condition.[38] However, current recommendations are for surgery at 5.5 cm if the above conditions are not met.

Ratio of aortic cross-sectional area to height

Although size alone has long been used to guide surgical intervention, a recent review from the International Registry of Aortic Dissection revealed that 59% of patients suffered aortic dissection at diameters less than 5.5 cm, and that patients with certain connective tissue diseases such as Loeys-Dietz syndrome or familial thoracic aneurysm and dissection had a documented propensity for dissection at smaller diameters.[39–41]

Size indices such as the aortic cross-sectional area indexed to height have been implemented in guidelines for certain patient populations (eg, 10 cm^2/m in Marfan syndrome) and provide better risk stratification than size cutoffs alone.[2,42]

The ratio of aortic cross-sectional area to the patient's height has also been applied to patients with bicuspid aortic valve-associated aortopathy and to those with a dilated aorta and a tricuspid aortic valve.[43,44] Notably, a ratio greater than 10 cm^2/m has been associated with aortic dissection in these groups, and this cutoff provides better stratification for prediction of death than traditional size metrics.[27,28]

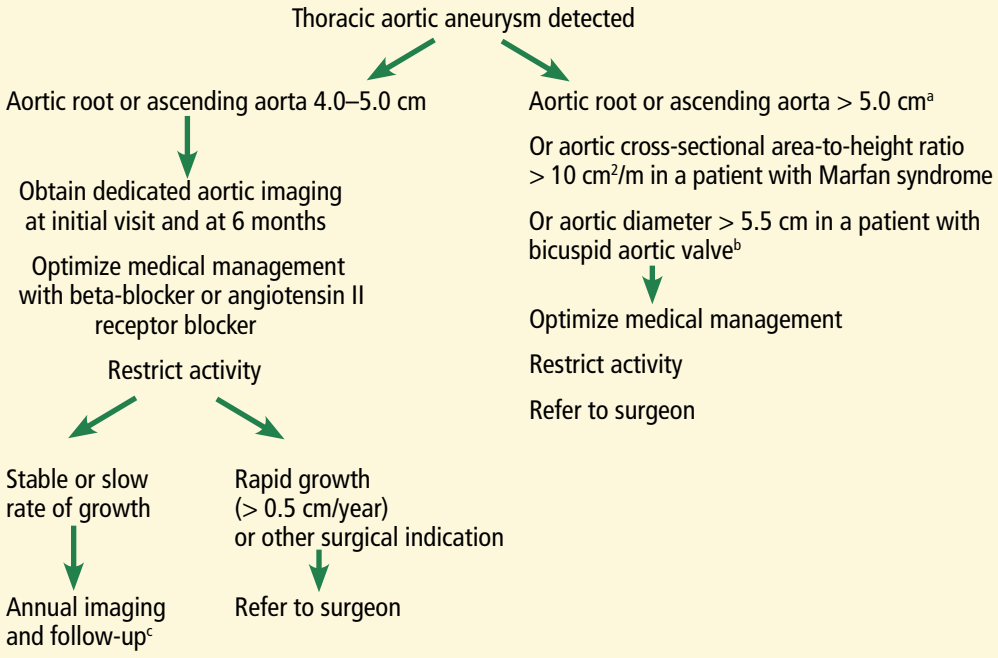
Once the diameter of the ascending aorta reaches 6 cm, the likelihood of acute dissection is 31%

HOW SHOULD PATIENTS BE SCREENED? WHAT FOLLOW-UP IS NECESSARY?

Initial screening and follow-up

Follow-up of TAA depends on the initial aortic size or rate of growth, or both. For patients presenting for the first time with TAA, it is reasonable to obtain definitive aortic imaging with CT or magnetic resonance angiography (MRA), then to repeat imaging at 6 months to document stability. If the aortic dimensions remain stable, then annual follow-up with CT or MRA is reasonable.[2]

MRA may be preferable to CT over the long term to limit radiation exposure.[2] Echocardiography should be used if the aortic root or ascending aorta is well visualized, but in



[a] It is reasonable to consider earlier surgical referral once the aortic diameter is greater than 4.5 cm in a patient with Marfan syndrome or bicuspid aortic valve.
[b] Surgery is reasonable in a patient with a bicuspid aortic valve with a diameter 5.0–5.5 cm if additional risk factors are present (rapid growth or family history of aortic dissection), or if the patient is at low surgical risk and the surgery is performed by an experienced surgical team, or with a diameter > 4.5 cm if there is an indication for aortic valve replacement.
[c] Frequency of imaging surveillance may be increased if the aortic diameter is > 4.5 cm in a patient with Marfan syndrome or bicuspid aortic valve.

Figure 5. Initial screening and follow-up of thoracic aortic aneurysm.

We routinely recommend screening of all first-degree relatives of patients with TAA

most patients the view of the mid to distal ascending aorta is limited. Echocardiography also offers evaluation of left ventricular size and function and allows for follow-up of aortic valve disease.

Our flow chart of initial screening and follow-up is shown in **Figure 5**.

Screening of family members

In our center, we routinely recommend screening of all first-degree relatives of patients with TAA. Aortic imaging with echocardiography plus CT or MRI should be considered to detect asymptomatic disease.[2] In patients with a strong family history (ie, multiple relatives affected with aortic aneurysm, dissection, or sudden cardiac death), genetic screening and testing for known mutations are recommended for the patient as well as for the family members.

If a mutation is identified in a family, then first-degree relatives should undergo genetic screening for the mutation and aortic imaging.[2] Imaging in second-degree relatives may also be considered if one or more first-degree relatives are found to have aortic dilation.[2]

We recommend similar screening of first-degree family members of patients with bicuspid aortic valve aortopathy. In patients with young children, we recommend obtaining an echocardiogram of the child to look for a bicuspid aortic valve or aortic dilation. If an abnormality is detected or suspected, dedicated imaging with MRA to assess aortic dimensions is warranted.

BACK TO OUR PATIENT WITH A BICUSPID AORTIC VALVE

Our patient with a bicuspid aortic valve had a 4.6-cm root, an ascending aortic aneurysm, and several affected family members.

We would obtain dedicated aortic imaging at this patient's initial visit with either gated CT with contrast or MRA, and we would obtain a cardioaortic surgery consult. We would repeat these studies at a follow-up visit 6 months later to detect any aortic growth compared with initial studies, and follow up annually thereafter. Echocardiography can also be done at the initial visit to determine if valvular disease is present that may influence clinical decisions.

Surgery would likely be recommended once the root reached a maximum area-to-height ratio greater than 10 cm^2/m, or if the valve became severely dysfunctional during follow-up.

BACK TO OUR PATIENT WITH MARFAN SYNDROME

The young woman with Marfan syndrome has a 4.6-cm aortic root aneurysm and 2+ aortic insufficiency. Her question pertains to the threshold at which an operation would be considered. This question is complicated and is influenced by several concurrent clinical features in her presentation.

Starting with size criteria, patients with Marfan syndrome should be considered for elective aortic root repair at a diameter greater than 5 cm. However, an aortic cross-sectional area-to-height ratio greater than 10 cm^2/m may provide a more robust metric for clinical decision-making than aortic diameter alone. Additional factors such as degree of aortic insufficiency and deleterious left ventricular remodeling may urge one to consider aortic root repair at a diameter of 4.5 cm.

These factors, including rate of growth and the surgeon's assessment about his or her ability to preserve the aortic valve during repair, should be considered collectively in this scenario.

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