A review of thoracic aortic aneurysm disease

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Abstract

Aortic diseases may be diagnosed after a long period of subclinical development or they may have an acute presentation. Acute aortic syndrome is often the first sign of the disease, which needs rapid diagnosis and decision making to reduce the extremely poor prognosis. Aortic dilatation is a well-recognised risk factor for acute events and can occur as a result of trauma, infection, or, most commonly, from an intrinsic abnormality in the elastin and collagen components of the aortic wall. Over the years it has become clear that a few monogenic syndromes are strongly associated with aneurysms and often dictate a severe presentation in younger patients while the vast majority have a multifactorial pathogenesis. Conventional cardiovascular risk factors and ageing play an important role. Management strategy is based on prevention consisting of regular follow-up with cross-sectional imaging, chemoprophylaxis of further dilatation with drugs proved to slow down the disease progression and preventative surgery when dimension exceeds internationally recognised cut-off values for aortic diameters and the risk of rupture/dissection is therefore deemed very high.

Introduction

The finding of a mildly dilated aorta is not uncommon on routine imaging of the heart. Examination of the aortic root, ascending aorta and aortic arch is part of the standard reporting for echocardiographers. This paper reviews aortic disease with specific focus on diagnosis, management and further investigations. It will review the genetic basis of hereditary aortic disease and summarise the presentation and management of acute aortic presentations.

There is no accepted definition for aortic diseases, or aortopathies, and the term is broadly applicable to both inherited and acquired conditions. They include thoracic and abdominal aneurysms and their risk of rupture or dissection. They account for significant cardiovascular morbidity and mortality worldwide. Recent European studies reflect this with incidence of aortic dissection in developed countries of 6 per 100,000 (1).

Aortic aneurysms can develop at any location but the majority happen in the abdominal aorta and more specifically between the renal arteries and the common iliac bifurcation. While the physiopathology has common features with abdominal aneurysm, cardiologists are far more involved in the management of thoracic aortic aneurysms. Therefore, the definition of aortopathy for the purpose of this paper relates to any condition where there is enlargement of the thoracic aorta, which will be the focus of this review.

In the past decades, more consistent surveillance and advances in understanding the genetic background and clinical presentation have been made so that there are now a number of new entities in which the main concern is around vascular dilatation and/or ruptures. It is also known that many congenital heart defects are associated with aortic dilatation, especially cono-truncal...
abnormalities, where it is felt a significant hemodynamic role with stretch to the proximal aorta is played by a longstanding right-to-left shunt in utero (i.e. tetralogy of Fallot).

**Presentation of aortopathy**

Patients with aortopathy present in a number of ways: 1) as an incidental finding as part of a routine examination such as transthoracic echocardiography (TTE), computerised tomography (CT) of the chest, cardiac magnetic resonance imaging (CMR) or on a routine chest radiograph (CXR), 2) as an acute presentation with thoracic aortic dissection 3) as part of cascade screening for a relative of an individual presenting with aortic disease and 4) as part of a recognised congenital cardiac condition.

Echocardiography descriptors used for the aorta will usually include the dimensions of the aorta at the aortic valve annulus, the aortic root, the sino-tubular junction, the ascending aorta, the mid arch and the proximal descending aorta, with aortic valve morphology and function being described and the velocity in the proximal descending aorta given (Table 1).

A comment on flow reversal in the aortic arch will be made for patients with aortic incompetence. The presence of diastolic tailing on continuous wave Doppler (CW) is made if coarctation of the aorta is suspected. TTE will give all of these measurements but the ability to define accurate dimensions and to produce three-dimensional reconstructions of the aorta is best achieved using CT scanning or MRI.

**Table 1** Echocardiographic assessment of the aorta (data from 2010 EAE guidelines (37)).

<table>
<thead>
<tr>
<th>View</th>
<th>Part of the aorta</th>
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<tbody>
<tr>
<td>Transthoracic aorta</td>
<td>Ascending + descending thoracic</td>
</tr>
<tr>
<td>Parasternal long + short axis</td>
<td>Descending thoracic</td>
</tr>
<tr>
<td>Apical four-chamber</td>
<td>Descending thoracic</td>
</tr>
<tr>
<td>Apical two-chamber and/or long axis</td>
<td>Descending thoracic</td>
</tr>
<tr>
<td>Suprasternal</td>
<td>Arch, descending + ascending thoracic</td>
</tr>
<tr>
<td>Subcostal</td>
<td>Abdominal (+ascending thoracic)</td>
</tr>
<tr>
<td>Transoesophageal echo</td>
<td>Ascending aorta</td>
</tr>
<tr>
<td>Upper oesophageal – long/short axis</td>
<td>Descending thoracic and arch</td>
</tr>
<tr>
<td>Aortic – long/short axis</td>
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</table>

**Common categories of aortopathy disease**

**Degenerative aortic disease**

The aorta normally possesses a high degree of elasticity, which aids in the propulsion of blood downstream and this is due to the wall structure, especially components of the tunica media. It is the smooth muscle cells, elastin and collagen, which impart the elastic properties and strength of the aorta. It is alteration of the quantity and/or architecture of these fibers that leads to the mechanical and functional changes associated with aortic disease (2).

Physiological changes to the aortic anatomy occur throughout life but are generally asymptomatic until middle age. Age-related changes occur, with the tunica media, gradually stiffening due to fracturing of elastin fibers and loss of smooth muscle cells, which are responsible for synthesizing elastin within the aorta. Following this decline in elasticity, the aorta will dilate and the mechanical forces of expansion will be transferred to collagen within the aortic wall. This process leads to progressive remodelling, resulting in a stiffer and less compliant vessel (3). This results in diminished aortic resilience and tensile strength, culminating in aortic wall thinning, dilation and increased wall stress, all of which may result in dissection and/or rupture.

Thoracic aortic dilatation is therefore a common finding with increasing age and is often seen in patients with known atherosclerotic disease, in particular in those patients with systemic hypertension. The distribution of aortic dilatation varies from being confined to the aortic root and ascending aorta to gross dilatation of root, ascending, arch and descending aorta, the so-called mega aortic syndrome.

Patients will frequently present as an incidental finding and routine monitoring, blood pressure control and secondary prevention of atherosclerosis is the management strategy.

Whilst the first diagnosis may be with TTE 2D imaging, initial sizing and subsequent surveillance of the aorta is best carried out using CT or MRI and will determine the management strategy.

**Bicuspid aortic valve disease**

This is a common congenital abnormality with a prevalence estimated at 1% of the general population. It is also a common associated feature of a number of congenital abnormalities including aortic coarctation, Shones syndrome and Turner syndrome.
Transthoracic echo is ideally suited to define the morphology of the valve and to measure annular and sinus dimensions. It is worth noting that the aortic valve annulus may be small and the aortic root and ascending aorta may also be smaller than normal – this is important to recognise as surgery in such conditions may require enlargement of the annulus and root to prevent patient prosthesis mismatch.

When assessing the aortic valve, subtypes relating to the valve morphology are described with true bileaflet valves and combinations of fusion patterns of the right, left and non-coronary cusps, with the commonest form being fusion of right and left coronary cusps around 80%.

It is described that certain valve types are more likely to develop ascending aortic dilatation. Recent 4D MRI studies of aortic flow in BAV demonstrate marked differences in wall stress associated with differing valve morphology. The R-L fusion pattern shows a highly eccentric flow, which is associated with aortic dilatation (4).

Proteomic analysis of explanted aortic tissue following surgery has demonstrated a key difference between bicuspid and non-bicuspid aortic valve aortas. Proteins involved in the inflammatory response are upregulated in the non-bicuspid aortopathy, where proteins involved in repair mechanisms are seen to be induced in the bicuspid aortopathy tissue (5), suggesting that the pathophysiology of BAV-associated aortopathy differs from other forms of the disease where intrinsic aortic disease may exist.

Hereditary aortopathy

It has long been recognised that there are families in which multiple members have suffered a thoracic aortic dissection, often with dissection occurring at young ages. There are many recognised hereditary aortopathies, which broadly fall into the syndromic and non-syndromic categories (Fig. 1).

Marfan syndrome (MFS), which has been considered the prototype of monogenic syndromic disease, is known to increase the risk of TAAD. Since the early 1990s, it has been demonstrated that the majority of cases of MFS are due to mutations in the Fibrillin 1 gene (FBN1) (6). Since then, though, improvements in the process of mutation detection has allowed a number of mutations to be identified as the cause of TAAD in hereditary aortopathy conditions other than MFS. The most common display an autosomal dominant pattern of inheritance with highly variable penetrance. Extensive clinical and imaging studies found that involvement of the arterial vasculature is far more extensive than just the thoracic aorta in some of these conditions. Also, genetic mutations that were originally described with syndromic presentation have been found to be present in families with familial thoracic aortic aneurysms and dissection (FTAAD) where the only manifestation is vascular with no other systems involved.

Familial aortopathy with known genotypes typically relate to mutations in the proteins forming the lamellar unit of the aortic wall and currently these known genotypes fall into three areas: mutations in genes coding for the extracellular matrix, those coding for smooth muscle protein and those that code for transforming growth factor beta (TGF beta) signalling (Fig. 2).

These known genetic variants account for around 40% of familial aortopathies and far less in those presenting with thoracic aortic dissection for the first time in their family. There is ongoing collaborative work looking for novel variants and to define what polygenic interactions may account for this otherwise unexplained aortic dilatation.

Marfan syndrome

First described clinically in 1896, Marfan syndrome is the most common of the inheritable condition with significant aortic disease. It is a highly penetrant...
autosomal dominant disease with variable expressivity involving a number of systems mainly cardiovascular, ocular and skeletal. Prevalence is as much as 17 cases per 100,000 population and ~25–30% mutations are de novo.

Marfan syndrome is caused by mutations in the FBN1 gene on chromosome 15q21 encoding fibrillin-1, a glycoprotein in the extracellular matrix. A deficiency of fibrillin leads to weakening of the extracellular matrix and upregulation of transforming growth factor beta signalling (TGFB) and altered transcription of target genes. More than 1800 mutations have been identified, almost all unique to an affected family (7).

Significant morbidity and mortality are typically related to the cardiovascular manifestations and complications and in particular dilatation of the aortic root at the sinus of Valsalva, which is present in many patients. Other manifestations include mitral valve prolapse and rarely dilated cardiomyopathy irrespective of vascular disease. Mean survival of untreated patients is 40 years but advances in understanding of the disease have brought a lower threshold for preventative surgery and systematic medical therapy, so that life expectancy is currently not dissimilar to the general population when appropriate management is in place.

The diagnosis of Marfan syndrome is established using the revised Ghent criteria in suspected cases (8). Briefly, the diagnosis is made in an individual who has an FBN1 pathogenic variant known to be associated with Marfan syndrome and aortic root enlargement (Z-score ≥2.0) or ectopia lenti. In the absence of genetic testing a clinical diagnosis can be made with demonstration of aortic root enlargement (Z-score ≥2.0) and ectopia lenti or a defined combination of features throughout the body, yielding a systemic score ≥7 according to the revised Ghent criteria (Table 2).

Lifelong follow-up is mandated and for aortic root measurements, transthoracic echocardiography is easy and reproducible. We recommend an echocardiogram every 2 years in stable aorta and more frequent imaging when progression is documented and in the paediatric group. Should there be any serial growth CT or MRA is warranted to facilitate discussion with the aortic surgical team. Below are two TTE of the aortic root of an FBN1 mutation positive Marfan patient who failed to attend follow-up for many years with significant interval change.

### Loeys Dietz syndrome

Loeys-Dietz syndrome was first described in 2006 and has a significant clinical overlap with Marfan (9). It can be distinguished by the presence of cleft palate/bifid uvula,

<table>
<thead>
<tr>
<th>Table 2</th>
<th>Systemic score for Marfan syndrome.</th>
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<tbody>
<tr>
<td>Skeletal features</td>
<td>score +3</td>
</tr>
<tr>
<td>Wrist AND thumb sign</td>
<td>score +3</td>
</tr>
<tr>
<td>Wrist OR thumb sign</td>
<td>score +1</td>
</tr>
<tr>
<td>Pectus carinatum deformity</td>
<td>score +2</td>
</tr>
<tr>
<td>Pectus excavatum or chest asymmetry</td>
<td>score +1</td>
</tr>
<tr>
<td>Hindfoot deformity</td>
<td>score +2</td>
</tr>
<tr>
<td>Plain pes planus</td>
<td>score +1</td>
</tr>
<tr>
<td>Reduced US/LS AND increased arm/height AND no severe scoliosis</td>
<td>score +1</td>
</tr>
<tr>
<td>Scoliosis or thoracolumbar kyphosis</td>
<td>score +1</td>
</tr>
<tr>
<td>Reduced elbow extension</td>
<td>score +1</td>
</tr>
<tr>
<td>Imaging features</td>
<td>score +2</td>
</tr>
<tr>
<td>Dural ectasia</td>
<td>score +2</td>
</tr>
<tr>
<td>Protrusio acetabuli</td>
<td>score +2</td>
</tr>
<tr>
<td>Cardiac features</td>
<td>score +1</td>
</tr>
<tr>
<td>Mitral valve prolapse</td>
<td>score +1</td>
</tr>
<tr>
<td>Other features</td>
<td>score +2</td>
</tr>
<tr>
<td>Pneumothorax</td>
<td>score +2</td>
</tr>
<tr>
<td>Skin striae</td>
<td>score +1</td>
</tr>
<tr>
<td>Myopia &gt; 3 diopters</td>
<td>score +1</td>
</tr>
<tr>
<td>Facial features (3/5)</td>
<td>score +1</td>
</tr>
</tbody>
</table>

Dolichocephaly, enophthalmos, downslanting palpebral fissures, malar hypoplasia, retrognathia. Maximum total: 20 points; score ≥7 indicates systemic involvement.
hypertelorism and widespread aortic and arterial tortuosity and aneurysms. Clinical features are frequently present involving vascular, skeletal, craniofacial and cutaneous systems. Incidence is currently unknown and cardiovascular manifestations are more severe and include congenital heart disease and early onset aortic and middle-size artery aneurysm with tendency to rupture at smaller diameter.

Diagnosis is based on the presence of suggestive vascular findings associated with a genetic mutation in one of the six described genes encoding for components of the TGFB pathway. Mutations in cytokines (TGFB2/3), receptors (TGBR1/2) or downstream effectors (SMAD2/3) are also associated with similar clinical presentations, and have led to the classification of Loeys-Dietz type 1 to 6 (10). Median survival was reported at 26 years of age in the early description of the disease and it is heavily dependent on a prompt diagnosis. Catastrophic vascular events have been described as early as 3 months of age and preventative surgery is the only effective management. Routine use of angiotensin II receptor antagonists is recommended based on the known interaction between the two pathways. Such events are also commonly seen in the peri-partum period of pregnancy and many families report death following childbirth.

Aortic dissection can occur even with normal aortic dimensions, as illustrated by the CT scans (Fig. 3). Therefore, education of the affected individual and their family is important, so that they ensure urgent review if they suffer any chest pain. Annual MRA scanning of the aorta is mandated and if there is evidence of serial growth then discussion with the patient and aortic surgical team is reasonable. Replacement of the aortic root will prevent a type A dissection but the long-term risk to the rest of the vasculature remains.

Histologic examination of aortic tissue in LDS reveals fragmentation of elastic fibers, loss of elastin content, and accumulation of amorphous matrix components in the aortic media. Structural analysis shows loss of the intimate spatial association between elastin deposits and vascular smooth muscle cells and a marked excess of aortic wall collagen. These characteristics are observed in young children and in the absence of inflammation, suggesting a severe defect in elastogenesis rather than secondary elastic fibre destruction (11).

Vascular EDS

Vascular Ehlers Danlos syndrome is associated with vascular rupture of middle-size arteries and the aorta at normal diameters and hollow organs (12). Clinically, important clues are a history of an arterial/intestinal/uterine rupture and minor features including easy bruising and a typical facial appearance. Guidelines recommend surgical intervention only in the setting of life-threatening complications due to the increased surgical and interventional risk (13, 14) so management of aneurysms requires a multidisciplinary approach and expert team. Lifespan is estimated at a median age of 51 with a wide range and phenotype-genotype correlations are emerging and might allow for a better stratification of risk of events.

Turner syndrome

Turner syndrome, one of the most common sex chromosome aneuploidy syndromes, is present...
in 25–50 per 100,000 females. Diagnosis is made in phenotypic females with a karyotype containing one X chromosome and complete or partial absence of the second sex chromosome, monosomy (45X0) is present in 40–50%. The most important phenotypic features are short stature, gonadal dysgenesis, neck webbing, and an increased incidence of renal and cardiovascular abnormalities. The latter include a wide range of left-sided lesion, more frequently consisting of bicuspid aortic valve (BAV) documented in 14–34%, coarctation of the aorta in 7–14% and thoracic aortic aneurysms reported in 3–42%. Aortic dissection has been estimated to be seven-fold more frequent than in the general population at 40 events in 100,000 patient years with the majority happening in young women in their third or fourth decade. Presence of congenital heart disease, 45X0 genotype, hypertension and pregnancy are considered risk factors. As aortic events do happen at small diameters and considering the short stature related to the syndrome, disease-specific z-scores and surgical thresholds have been suggested (15). Surveillance, even in the absence of overt cardiac disease, is recommended, as acquired cardiac diseases including aortic and coronary disease is prevalent.

**Additional features suggestive of hereditary aortopathy**

**Arterial tortuosity**

Arterial tortuosity of the aortic branch vessels and specifically the head and neck vessels (Fig. 4) is a common finding in a number of hereditary aortopathies and therefore should be screened for with appropriate imaging modality (typically MRA of the whole aorta and head and neck vessels). Whilst tortuosity in itself rarely causes a clinical concern, many conditions also demonstrate small and medium vessel aneurysm formation associated with a significant lifetime risk of morbidity (20). Also the presence of tortuosity has been suggested as a marker for increased risk of aortic events.

**Early onset osteoarthritis**

Early onset osteoarthritis is particularly associated with mutations in SMAD3 and is characterised by early onset of widespread osteoarthritis and osteochondritis dissicans, and by widespread vascular abnormalities including aneurysms and marked tortuosity. It is a highly penetrant autosomal dominant pattern of inheritance. Often the osteoarthritis is the presenting feature and this should be considered in patients with a family history of early cardiovascular death and early onset osteoarthritis (21).
Aortic embryology

The specific location of dilatation which occurs first in the aortic root is thought to be secondary to the high pressures generated by the left ventricle but has been recently felt to be also a consequence of a different embryological origin of the vascular smooth muscle cells in the aortic root (22). In early embryogenesis, vascular smooth muscle cells (VSMC) derive from a number of areas and these appear to be consistent, with aortic root VSMC deriving from the lateral plate mesoderm, the ascending aorta and arch from neural cress origin and the descending aorta from the paraxial mesoderm (23) (Fig. 3). It is of note that the point at which aortic dissection occurs is at the junction of these parts of the aorta (Fig. 5). This is the subject of further investigation but may explain the specific nature of thoracic aortic dissection.

Acute aortic syndromes

Sudden severe chest pain typically radiating through to the back is the classical presentation of patients with a thoracic aortic dissection. The pathological characteristic is separation of the intima from the media of the aorta, creating a new channel, the false lumen. This is termed a thoracic aortic dissection and is typically defined by the point at which the intimal flap occurs and whilst there are a number of classifications used, the simplest is the Stanford classification of type A or type B (Fig. 4). Whenever a thoracic aortic dissection is suspected, urgent CT scanning is mandated to demonstrate the integrity or otherwise of the aortic wall. Recent guidelines suggest that TOE is an alternative to this, but where available CT should be used as it is diagnostic and will demonstrate the full extent of the dissection, allowing the surgeon to plan the surgical intervention (13). Early intervention in this condition is essential to reduce mortality and immediate discussion with the surgical team once the diagnosis is made is crucial.

In the type A dissection (Fig. 4A) the intimal flap originates in the ascending aorta and in the type B it originates in the descending aorta (Fig. 4B). The false lumen typically extends distal to the point of dissection and can extend as far as the iliac vessels in both types. The distinction is important as Type A dissection is associated with significant early mortality, and is managed with emergency open surgical intervention to replace the segment of aorta where the intimal flap has been raised. Without surgery, proximal extension can lead to a cardiac tamponade and/or acute coronary ischaemia when the coronary origins are compromised, or rapid expansion of the aorta leading to rupture. In such cases, death can occur rapidly, with the diagnosis of aortic dissection being made post mortem.

The management of a type B dissection is typically conservative with control of systemic hypertension, with i.v. beta-adrenergic blockade being the mainstay of therapy. In recent times, the developing technology of self-expanding stents has allowed for percutaneous placement of a large covered thoracic aortic stent. Thoracic endovascular aortic replacement (TEVAR) is routinely used in many countries for the acute management of type B dissection but this remains controversial and is not supported by randomised trial data (24). Medical management is associated with excellent outcomes for most patients with type B dissection and remains the standard treatment for all, with additional TEVAR therapy in selected cases.

In selected cases, TEVAR should be considered. In patients post Type B dissection with a rapidly expanding descending aorta, distal perfusion issues, uncontrolled hypertension, ongoing symptoms. These features are more common in patients with a dissection entry point >10 mm, a partially thrombosed false lumen, a descending aortic dimension >35 mm or false lumen diameter >20 mm (25). What is clear, is that if an early intervention (<14 days from presentation) is required in these patients then the outcome is poor regardless of intervention (25).

Routine medical treatment

Systemic hypertension can play a key role in the formation of aortic aneurysm or as a contributing factor.
in patients with congenital cardiac defects or inherited conditions. Aggressive medical treatment is important and a number of drug classes can be used as monotherapy or in combination to achieve optimal blood pressure values (26). Young patients with inherited aortic diseases are very rarely hypertensive but medications that reduce hemodynamic stress on the aortic wall, such as beta blockers (β-blockers) or angiotensin receptor blockers (ARBs), have been tested in Marfan syndrome. Though literature is limited to small studies with some controversial results (27) there is evidence medicines are beneficial and reduce the rate of aortic growth and β-blockade is currently recommended and generally initiated at diagnosis or upon documentation of significant and/or progressive aortic dilatation (28, 29). Beside the haemodynamic stress to the arterial wall, which is an obvious target for treatment, interest in the past decade has moved to possibly modifying the dysregulation of the TGFβ pathway as ARBs have proven effective in mice models (30) and results of the AIMS study are at least partially supporting findings in humans (31). There is evidence in the trials these should be up titrated to the highest tolerated dose and to haemodynamic effect on exercise. No evidence is available in many other rare conditions including scarce data in bicuspid aortic valve aortopathy and medical management is largely extrapolated form Marfan data. A single small randomized trial in vEDS patients showed a reduced rate of vascular events in the group treated with the β-blocker Celiprolol but more data is needed (32). Many centres are now routinely using ARBs in the management of younger patients with aortopathy based on these studies, where beta-blockers remain mainstay therapy in other institution and ARBs are used in conjunction.

Moderate aerobic exercise is safe and should be encouraged in all patients, but certain activities are not recommended. Patients with a dilated aorta do not compete in impact sports or in activities where sudden deceleration is a risk, they should not lift where straining is required as this may induce a Valsalva manoeuvre associated with a rapid transient increase in systemic blood pressure on release. Extreme sports should be avoided including extreme endurance sports. All recreational drugs should be avoided and smoking cigarettes is discouraged.

Having adequate travel insurance is recommended for all travel, and patients currently should declare they have a dilated aorta and should declare any family history but do not need to declare genetic results. This may change in the future when widespread genotyping may become available and this should be discussed with their clinical genetics team.

Pregnancy is a risk for young women with a dilated aorta. The normal vascular changes that occur in pregnancy predispose to further aortic dilatation and thoracic aortic dissection is not uncommon. Our practice is for all women to undergo pre-pregnancy counselling regarding the risks. Any woman with a dilated aorta will undergo a mid-trimester MRA of the thoracic aorta (33) and if further dilatation has occurred we would discuss a planned caesarian section, prior to the expected date of delivery. Surgery during pregnancy may be indicated in certain cases (34). If the aorta is greater than 4.5 cm prior to pregnancy then the risk of dissection during pregnancy is considered greater than the risk of surgery and patients may discuss prophylactic surgery with their clinical teams. Progestogen-only contraception is recommended and is safe in all women with a cardiovascular condition.

**Elective surgical management**

Indication for elective surgical treatment is commonly discussed to replace the dilated portion of the aorta and address any significant valvular disease or other anatomic findings amenable to surgical repair. While it was once felt the absolute diameter was the only factor influencing the decision making (35) it is clear that many other factors need to be taken into account when assessing these patients’ risk and when trying to prevent acute aortic events. Some of them are incorporated in the most recent guidelines from the American and European societies of cardiology (13, 14), but the most recent genotype–phenotype correlations are not reflected by these documents. Therefore, in rare diseases decision making is extrapolated from data available in similar conditions and thorough assessment of the perceived individual risk and dictate a patient-tailored approach (36).

In Marfan syndrome surgical repair of the aorta is indicated once the maximal measurement approaches 5.0 cm, or the rate of increase of the aortic root diameter approaches 1.0 cm per year or there is progressive and severe aortic regurgitation (13, 14). More aggressive therapy may be indicated in individuals with a family history of early aortic dissection and we would typically offer surgery in such instances once the aorta exceeds 4.5 cm in Marfan syndrome and greater than 4 cm in Loey Dietz syndrome. Many individuals can receive a valve-sparing procedure that precludes the need for chronic anticoagulation.

While there is no agreed-upon absolute size threshold for aortic root surgery in childhood, many centres use
the adult guideline of 5.0 cm given the extreme rarity of aortic dissection in young children or 4.5 cm in the presence of a perceived increased risk. Every effort is made to allow the aortic annulus to reach a size of at least 2.0 cm, allowing placement of an aortic graft of sufficient size to accommodate body growth (18).

Surgery usually consists in replacing the dilated portion of the aorta aiming for a valve-sparing operation when possible in order to avoid the long-term anticoagulation required with mechanical valves (i.e. David/Yacoub operation versus Bentall operation). Novel techniques such as surgical aortic valve repair/plasty and external aortic root support are gaining research interest and results are awaited to assess if they can represent a viable option on a vast scale.

Conclusion
Thoracic aortopathy is a rare condition in which the thoracic aorta dilates, placing the individual at risk of thoracic aortic dissection and rupture. It is a rare cause of sudden death in the younger adult and very occasionally in children, when it is likely to have a genetic cause. However, the majority of such cases remain unexplained and considerable efforts are being made to understand the genetic disorders that underlie aortic dilatation. Imaging is key to diagnosis and long-term surveillance both prior to and following intervention or dissection. In time we may understand the mechanisms underlying the dilatation of the aorta and better therapeutic interventions may be developed.

Declaration of interest
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