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Original Article

Dilation of the ascending aorta in childhood: 4 cases without obvious predisposing disease

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Abstract Dilation of the ascending aorta is rare in childhood. When seen, it is usually associated with some form of connective tissue disease or predisposing cardiac malformations, especially an aortic valve with two leaflets. We describe four children in whom significant dilation of the ascending aorta was encountered as an incidental finding. No patient had any sign of an associated connective tissue disease, nor did we detect any predisposing cardiac anomalies. One patient had undergone surgical ligation of the arterial duct in infancy, whilst another had undergone repair of aortic coarctation, also in infancy. A third child has had repair of an atrioventricular septal defect with exclusively atrial shunting, whereas the fourth patient had a structurally normal heart. The aortic valve had three leaflets, and was functionally normal in all. The dilation of the ascending aorta was progressive in all patients, and finally surgical treatment was recommended, relying on the guidelines established for the management of patients affected with the Marfan syndrome.

Keywords: Aortic aneurysm; children; Marfan's syndrome

Dilation of the aortic root, with or without accompanying dilation of the ascending aorta, is a feature commonly observed in children with an underlying predisposing disease such as the different forms of systemic abnormalities of the connective tissue. The typical, and most frequently observed, case is a patient with Marfan's syndrome, where up to four-fifths of the patients show aortic enlargement during the course of the disease.[1] Aortic dilation has also been recognized as being associated with bifoliate aortic valves, with or without concommitant coarctation.[2] Besides these predisposing factors, dilation of the ascending aorta is observed but rarely in children. We describe here a small series of children with an unusual presentation of aortic dilation eventually requiring surgical treatment.

Summary of presentations

Relevant clinical characteristics of the patients are outlined in Table 1.

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The four patients, 3 girls and one boy, all came to our attention because of aortic dilation during an eight-year period ending in 1999. The observed dilation of the aortic root and ascending aorta was an incidental finding in all. It was discovered during routine outpatient visits for follow-up of other known cardiovascular disease in 3 patients, and during hospitalisation because of cranial trauma in the last patient. All the patients underwent serial echocardiography at 6-monthly intervals, with an example of the dilation encountered being shown in Figure 1. Measurements of the aorta were compared to standard centile curves established for the follow-up of patients with Marfan's syndrome.[3] The evolution of the diameters of the ascending aorta with increasing time is shown in Figure 2.

All the children had clinical genetic examination by a specialist, and were thoroughly evaluated according to the currently recommended clinical diagnostic criterions for Marfan's syndrome.[3] None of the children had any of the known clinical signs, such as skeletal, ocular or dermal manifestations. Molecular genetic testing was not routinely performed.

Treatment consisted of valve-sparing supracoronary replacement of the ascending aorta,

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Table 1. Relevant clinical data

	Patient 1	Patient 2	Patient 3	Patient 4
Age at diagnosis of aortic dilation	8.5 years	1 year	9 years	5.5 years
Time of observation	1 year	4.5 years	5 years	9.5 years
Max. aortic diameter/	35 mm	38 mm	40 mm	39 mm
aoratic valvar diameter	15 mm	13 mm	19 mm	17 mm
Associated cardiac disease	None	Persistent duct	Coarctation	AVSD with atrial shunt
Previous surgery	None	Ductal ligation	Coarction repair	Correction of AVSD
Associated systemic disease	None	None	Pulmonary emphysema	Fetal alcohol syndrome
Familiarity of aortic disease	neg.	neg.	neg.	neg.
Familiarity of connective tissue disease	neg.	neg.	neg.	neg.
Aortic valve (at surgical inspection)	Tricuspic	tricuspid	tricuspid	tricuspid
Aortic regurgitation	none	none	trivial	none
Mitral valvar prolapse	no	no	no	no
Surgical procedure	Asc. Aorta replacement	Asc. Aorta replacement	Asc. Aorta replacement	Asc. Aorta replacement
Histology of aorta:				
– specific	wall thinning	wall thinning	wall thinning	patchy fibrosis
– cystic media necrosis	negative	negative	rare, patchy	negative
– inflammatory cells	none	none	none	none

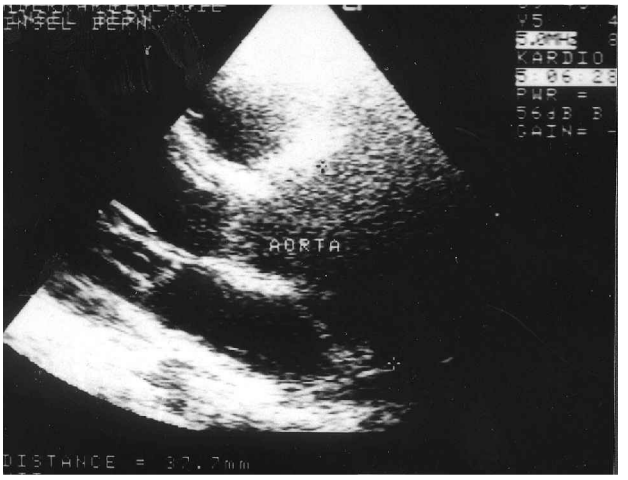

Figure 1.
Echocardiographic image of our second patient, in the parasternal long axis, showing the extent of dilation of the ascending aorta seen preoperatively.

following the technique established by Yacoub and David, using a Vascutek gelweave® vascular prosthesis of adequate size to replace the entirety of the ascending aorta and the aortic root when indicated. All the procedures included an inspection of the morphology of the aortic valve.

All resected aortic specimens were examined histopathologically, with thinning of the medial wall being the most obvious finding in all the cases. In 2 specimens, there was disorganization of the medial muscular cells, while patchy fibrosis was noted in another specimen. The typical findings of cystic media necrosis were seen in only one specimen. In 3 of the patients, we undertook additional specific biochemical typing of the collagen, which excluded most types of Ehlers-Danlos syndrome and osteogenesis imperfecta. Echocardiography was performed in all first degree relatives, and excluded occult aortic disease.

Discussion

Dilation of the aortic root, or of the ascending aorta, or in combination, is a typical lesion in adults, and is typically atherosclerotic or the consequence of cystic medial necrosis.[4] In children, in contrast, arterial and aortic aneurysms are rare. When seen, they are associated with a number of different clinical circumstances, the aorta then being but one of several possible locations for arterial dilation.[5] Isolated dilation of the ascending aorta, in contrast, is exceedingly rare in childhood and, when it occurs, it is usually associated with some form of systemic disease of the connective tissues. Most frequently, it is observed in Marfan's syndrome,[1] but is also described as a feature of the different subtypes of Ehlers-Danlos-syndrome,[6] osteogenesis imperfecta,[7] and even more rarely in the setting of Turner's syndrome.[8] In the latter situation, it is also associated with an aortic valve having two leaflets. It has been demonstrated that dilation of the ascending aorta occurs in patients with bifoliate aortic valves irrespective of, whether

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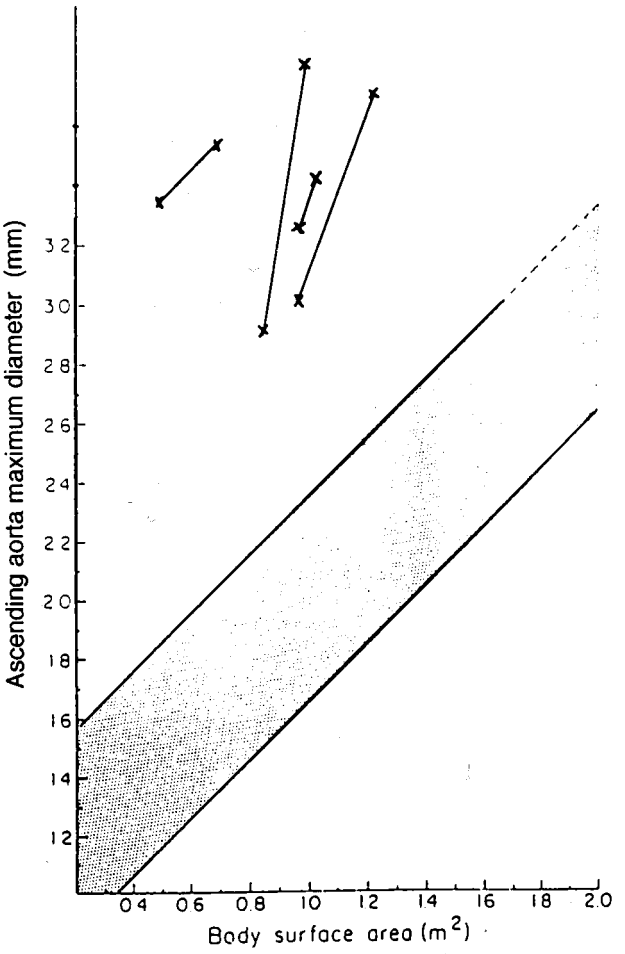

Figure 2.
Evolution of the measured diameters of ascending aorta during the period of observation of each individual child, comparing them with the normal range adapted from the experience of DePaepe et al.[3]

or not there is clinically relevant aortic stenosis.[9,10] Dilation also occurs sometimes in the presence of additional aortic coarctation.[2] Cellular apoptosis has recently been postulated to be one factor underlying abnormalities of the aortic wall in patients with congenital malformations of the aortic valve, leading to progressive histologic mural abnormality, and hence dilation.[11] Outside the conditions mentioned above, and rare reports of familial cases,[12] aortic dilation is an exceedingly rare observation in childhood.

Our study focused on the important observation that aortic dilation was observed in a number of children in the absence of any apparent predisposing factor. The children had a normal phenotype, no familiarity for cardiac or connective tissue disease, and none of the established predisposing cardiac malformations. We cannot exclude, however, the possibility that some mild form of connective tissue disease might have remained undiagnosed in our children. For instance, it is known that in relatively well-described clinical make-up of Marfan's syndrome there is a range of overlap between overt disease and the phenotypic normal variation.[3]

Criterions to proceed with surgery in presence of dilation of the aortic root dilation, such as annuloaortic ectasia or aneurysm of the ascending aorta, are well described for the adult population. In children, in contrast, the indications for repair or replacement of the ascending aorta are based mainly on comparative echocardiographic measurements of the dilated segment with the dimensions of the aortic arch and the descending aorta. Surgery should be recommended to prevent rupture or dissection. For our patients, it seemed logical to follow the recommendations established for Marfan's syndrome, advising prophylactic surgical treatment when the aorta reached comparable diameters, since these are the only criterions currently available. It remains to be established whether children such as ours, having aneurysmal aortas in the absence of any recognised syndrome, are at a lesser risk of dissection or rupture compared to those with Marfan's syndrome. Although aortic dilation is exceedingly rare in non-syndromic patients, dissection has been observed.[12] Furthermore, it has been shown, albeit again for those with Marfan's syndrome, that a pattern of generalized dilation of the ascending aorta was a marker of increased risk of dissection as compared to those with isolated dilation of the aortic root.[13] The pattern of generalized dilation was the one observed in all our patients. In a large surgical series of children and adults with Marfan's syndrome, one-eighth of patients were recommended for surgery when the aortic diameters were between 30 and 50 mm, and dissection had occurred in the majority of them, despite a the aortic diameter reached being below the usual cut-off value for prophylactic surgery.[14] When considering the criterions for prophylactic surgery, several groups have stressed the importance of the rate of progression of dilation.[15–17] Such relatively rapid progression of dilation was a marked feature of our patients. Taking into account all these considerations, we recommended surgical intervention to the families of all our patients, although the absolute values of aortic diameter reached during the evolution of the disease might seem relatively low compared to standard recommendations for prophylactic intervention.

In summary, therefore, we draw attention to the fact that some children without obvious

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predisposing conditions undergo clinically significant aortic dilation, which can then be encountered as an incidental finding. These patients should be treated following the criterions established for those afflicted by Marfan's syndrome.

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