Asymptomatic dissecting aortic aneurysm in a young adult: a case report

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ABSTRACT
Aortic dissection is a rare condition, yet it is potentially fatal. We report here a case of a 37-year-old asymptomatic male who came for a cardiology checkup as he was concerned that seven of his family members had been diagnosed with aortic aneurysm and some of them had progressed to aortic dissection. He had a history of hypertension that was controlled with medications. Although the patient was entirely asymptomatic, the diagnosis of a dissecting aortic aneurysm was confirmed. The aneurysm was confined entirely to the ascending thoracic aorta. The clinical presentation, diagnosis and management of ascending aortic dissection were reviewed. It has to be noted that early and accurate diagnosis and treatment of such a potentially lethal condition are essential for survival. Long term follow up is also required as there is always a risk of recurrence.

Key words: aneurysm, familial, CT aortogram, tranesophageal echocardiography, dissection

Citation

INTRODUCTION
Thoracic Aortic Aneurysms leading to acute dissection (TAAD) are responsible for significant premature mortality and are associated with a wide range of underlying conditions including hypertension, bicuspid aortic valve, and syndromic conditions such as Marfan syndrome (MFS) or Loeys-Dietz syndrome. In the absence of a syndromic cause, it has been estimated that 19 to 20% of TAAD cases have a genetic component leading to familial TAAD (FTAAD). These conditions display interfamilial variability as well as variable penetrance and severity.

Whereas familial thoracic aortic aneurysms are less common than many other cardiovascular conditions, the fact that they can be life-threatening and that even large aneurysms may not produce symptoms makes it all the more important for clinicians to be vigilant in their evaluation of patients at risk. Screening of first degree relatives with Familial Thoracic Aortic Aneurysm (FTAA) and FTAAD is essential.

FTAA and FTAAD could be detected during routine evaluation for non-cardiac reasons or during evaluation of aortic regurgitation murmur.

CASE REPORT
A 37 year-old male patient apparently healthy, reported for routine checkup and evaluation. He had a history of hypertension and family history of ischemic heart disease, diabetes mellitus and hypertension. He was asymptomatic on presentation. He stated that he had seven of his family members (both males and females) diagnosed with aortic aneurysm, and some had progressed into aortic dissection.

Clinical examination revealed a regular pulse rate at 67 beats per minute and a blood pressure 160/75mm Hg that was equal bilaterally. Peripheral pulses were all felt equally bilaterally. On auscultation, there was an early diastolic murmur at the base of the heart consistent with aortic regurgitation (AR). There were no signs of Marfan syndrome or other connective tissue diseases. 12-lead surface electrocardiogram (ECG) showed high-voltage criteria for left ventricular hypertrophy (LVH). Chest X–Ray revealed normal cardiac size and clear lung fields, but there was wide mediastinum. Transthoracic echocardiography (TTE) revealed proximal aortic aneurysm.
and there was a suspicion of the presence of an intimal flap. Moderate AR was detected. CT aortography was performed for full evaluation of thoracic and abdominal aorta. It revealed that the ascending aorta was dilated at its origin (5.3 cm) with a suspicious flap-like shadow at its posterior aspect warranting further evaluation (Figure 1).

Figure 1. CT Aortogram showing the aneurysmal dilatation of the proximal aorta. The dissecting flap is seen.

The aortic arch and descending aorta were of normal diameter down to the aortic bifurcation. Transesophageal echocardiography (TEE) was performed, which confirmed the presence of a dissecting aortic aneurysm that was confined to the ascending aorta (Figure 2).

Aggressive blood pressure lowering with beta blockers was initiated. The patient was advised to proceed for surgical treatment at the earliest. Within a few days, surgery was performed. The aortic valve was repaired, the ascending aorta was replaced with a prosthetic graft and the coronary arteries were re-implanted. We are still following up the patient for more than two years after surgery. He is asymptomatic and all the secondary preventive measures are being followed. In this case, timely referral for early surgical correction saved the life of the patient without complications.

DISCUSSION

Aneurysms of the ascending thoracic aorta most often result from cystic medial degeneration (CMD), which appears histologically as smooth muscle cell dropout and elastic fiber degeneration. CMD leads to weakening of the aortic wall, which in turn results in aortic dilatation and aneurysm formation. CMD occurs normally to some extent with aging, but the process is accelerated by hypertension. CMD is also seen in patients with ascending thoracic aortic aneurysms who do not have overt connective-tissue disorders. Moreover, it is now recognized that although cases of thoracic aortic aneurysms in the absence of overt connective-tissue disorders may be sporadic, they are often familial and are now referred to as the Familial Thoracic Aortic Aneurysm (FTAA) syndrome.

In an analysis of a large database of thoracic aortic aneurysm patients, Coady and colleagues found that at least 19% of patients had a family history of a thoracic aortic aneurysm, and that they tend to present at significantly younger ages than those with sporadic aneurysms. Most pedigrees suggest an autosomal-dominant mode of inheritance, but there is marked variability in the expression and penetrance of the disorder, such that some inherit and pass on the gene but show no manifestation of the disease. Several mutations have been identified. A mutation on 3p24.2–25 can cause both isolated and familial thoracic aortic aneurysms, with histological evidence of CMD. Mutations have also been mapped to two other chromosomal loci (5q13–14 and 11q23.2-q24). The extent of genetic heterogeneity is likely to become more evident as more families with thoracic aortic aneurysms are studied. It was also suggested that this is actually a polygenic condition, thus explaining the variable expression and penetrance. Consequently, at present, it is not possible to perform routine genetic screenings for this syndrome.

Most patients with thoracic aortic aneurysms are asymptomatic at the time of diagnosis, and the
aneurysms are typically discovered incidentally on imaging studies (chest x-ray, CT scan or echocardiography) ordered for other indications. However, the typical manifestations of aneurysms of the root or ascending aorta may include: secondary AR (so a diastolic murmur may be detected on physical examination or, less often, patients present with congestive heart failure), local mass effect (especially with large aneurysms) such as compression of the trachea or main stem bronchus (causing cough, dyspnea, wheezing or recurrent pneumonia), compression of esophagus (causing dysphagia), or compression of the recurrent laryngeal nerve (causing hoarseness of voice). Rarely, chest or back pain may occur with non-dissecting aneurysms as a result of direct compression of other intrathoracic structures or erosions into adjacent bones. The feared consequence of thoracic aneurysm is aortic dissection or rupture (often referred to as an acute aortic syndrome), which is potentially lethal. Typical symptoms of acute aortic syndrome include the abrupt onset of severe pain in the chest, neck, back, and/or abdomen.

The diagnosis of pre-aneurysmal dilatation of the proximal aortic root or ascending aorta is based on measurement of the dimensions of the sinuses of Valsalva and ascending aorta using imaging modalities such as 2D echocardiography, CT scan, MRI, or angiography and comparison with age-appropriate nomograms indexed for body surface area (BSA).

The major diagnostic criteria for familial thoracic aortic aneurysms and aortic dissections are the following:

- Progressive enlargement of the ascending thoracic aorta involving the sinuses of Valsalva, the ascending aorta, or both.
- Exclusion of Marfan syndrome, Loey-Dietz syndrome, Ehlers Danlos syndrome, vascular type and other syndromic causes of TAAD.
- A positive family history of TAAD.

Presently, it is not possible to perform routine genetic screening for this syndrome. Medications that reduce hemodynamic stress on the aorta, such as beta adrenergic blocking agents, are recommended. Because of the high mortality rate associated with acute ascending aortic dissection (~40 to 50%), it is crucial to not only identify patients with enlarged ascending aortas and to attempt to slow aortic growth using medical treatments but also to intervene surgically in an elective fashion before dissection occurs. How, then, is the clinician to determine when to proceed to surgery? Knowledge of the likely risk of dissection for a given condition in an individual patient and aortic diameter become of paramount importance. Surgery is generally recommended in cases in which the ascending diameter exceeds 5.0 to 5.5 cm; however, a large proportion of dissections occur at smaller diameters, and even in patients with no visible enlargement.

CONCLUSION

This case reports on the chance of incidental detection of asymptomatic Thoracic Aortic Aneurysm and Dissection. Diagnosed as being a familial condition in our patient, screening in the first degree relatives was advised. Low threshold for referral for surgical correction saved the patient's life. Blood pressure control and adherence to the cardiovascular risk factor reduction strategies are an essential part of the management plan, both before and after the surgery.

References


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