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Case report: giant ascending aorta at a young age

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Abstract

Introduction: The aneurysms of the thoracic aorta are most common in the VI–VII life decade. In this case report we present a young female with an aneurysm of ascending aorta and severe aortic valve regurgitation**.**

Case report: A 33-year-old woman with shortness of breath and other typical symptoms of the aneurysm of the aorta and a severe regurgitation of the aortic valve had underwent several tests to find the main cause of this mentioned disease. The patient was treated surgically - the David Procedure was performed, and the aortic valve was repaired.

Conclusion: This case report shows that the aneurysm of ascending aorta could be present at a young age without any specific obvious predisposing factors such as congenital bicuspid aortic valve, phenotypic signs of Marfan syndrome or known familiar history.

KEYWORDS: thoracic aorta aneurysm; aortic valve regurgitation; familial aneurysms syndrome.

1. INTRODUCTION

Degenerative aneurysms of the thoracic aorta are the most common in the VI–VII life decade, mainly in males with other risk factors for atherosclerosis (1). Conversely, when an aortic disease occurs in a young patient, further evaluation is needed to detect rarer causes. The majority of patients with aortic aneurysms have a long latent period before clinical symptoms appear (2). In many cases during an imaging examination on the chest, for example, chest XR, echocardiography, the aortic aneurysms can be detected incidentally. Symptoms from compression of adjacent structures can be the presenting feature: chronic chest pain, cough, dysphagia can result from pressure on the sternum, trachea, or esophagus respectively (2). However, in 75% of patients, the first presentation is an acute sudden severe tearing chest pain (2). We present a case report of a young female with an aneurysm of ascending aorta and severe aortic valve regurgitation.

2. CASE REPORT

A 33-year-old woman was admitted to the emergency department due to severe shortness of breath which lasted for 3 days. The dyspnea occurred especially at night and weakened only after changing the position to sitting or standing. Furthermore, headaches appeared in the temporal field and the patient felt palpitation. The patient denied other comorbidities, usage of any drugs, or previous cardiovascular diseases in her family. She was a long-term smoker.

During the first physical examination, the general condition of the patient was stable. The heart rhythm was regular with 70 beats/min. The blood pressure was 110/44 mmHg in the left arm and 116/48 mmHg in the right. Cardiac auscultation revealed a decrescendo systolic blowing fourth-degree murmur, best heard on the left lower sternal border, spreading to Botkin-Erb's point, neck, between the shoulder blades and diastolic fourth-degree murmur at the Botkin-Erb's point spreading to the apex. Breathing sounds were clear. Systolic pulse with the diastolic wave was felt on both sides of the tibialis posterior arteries. No other objective significant abnormalities were found during physical examination.

For further treatment, the patient was hospitalized in the Department of Cardiology.

Total blood count, creatinine level, electrolytes, glycaemia, coagulation parameters were normal. Erythrocyte Sedimentation Rate (ESR) was 6 mm/h (normal range (NR) 0-11mm/h), C-reactive protein (CRP) was 5 mg/l (NR 0-5 mg/l), hemoglobin (Hb) – 124 g/l (NR 120-155 g/l). NT-pro- BNP was slightly elevated (267 ng/l) (NR 0-125 ng/l).

The electrocardiogram showed sinus rhythm, left ventricular hypertrophy, high symmetrical T wave in thoracic derivatives and ischemic changes: T wave depression in aVL derivations, q waves in III, aVF derivations.

Chest X-ray showed a small venostasis in the lungs without infiltration, increased size of the aorta. (Figure 1)

2D echocardiography revealed evidence of a severe dilatated proximal part of ascending aorta (86 mm) with a severe regurgitation of the aortic valve. A large aneurysm of the aortic root and ascending part was visible - about 7 cm in length from the annulus of the aortic valve. The aortic valve was tricuspid. The left ventricular (LV) was dilated, especially in the middle part up to 74 mm. LV systolic function was normal (ejection fraction (EF) – 55%) with eccentric LV remodeling. (Figure 2,3)

An urgent computed tomography (CT) scan (CT aortography) was performed (Figure 4). There were no signs of dissection. Accurate diameters of the thoracic and abdominal aorta were measured.

After diagnosing a severe aneurysm of the ascending aorta, in the absence of a traumatic origin or bicuspid aortic valve with possible aortopathy, immunological tests were performed suspecting possible aortitis related to autoimmune, connective tissue disease. Antinuclear antibodies (ANA) and antineutrophil cytoplasmic antibodies (ANCA) were negative. Treponema pallidum IgG/IgM antibodies were investigated on suspicion of syphilitic aortitis as a possible cause of aneurysm (it was not found). In research of possible causes, antibodies against hepatitis C virus (HCV) and hepatitis B virus surface antigen (HBsAg) had also been tested and returned negative.

Medicinal treatment with beta adreno-receptor blockers (metoprolol) was prescribed to decrease the risk of aneurysm rupture. Anamnesis, results of laboratory test, echocardiography, CT scan were discussed in Heart team and decided to perform open-heart surgery – replacement of ascending aorta and aortic valve – was done while the risk of the operation was low EuroSCORE II (3) - 1.2%).

The patient was directly transferred to the Department of Cardiac Surgery. The surgical approach was performed through the median-thoracotomy, followed by cannulation of the ascending aorta, inferior vena cava and right atrium (RA), clamping down on the ascending aorta (for 70 minutes). Infusion of the cardioplegic solution to the heart ostia was initiated. The operation was performed using the Artificial Blood Circulation System (ABCS) - cardiopulmonary bypass (for 93 minutes). During Valve Sparing Root Replacement (also called the David Procedure) the ascending part of the aorta was exchanged with a 30 mm prosthesis, and the aortic valve was repaired and reconnected to a new section of aortic tissue. By preserving the native aortic valve, patients avoid the need for lifelong anticoagulation therapy (4,5). Aortic tissue pathology showed degenerative changes only. After the operation, a larger secretion from the drain was observed. Coagulation rates were adjusted by 2 pieces of fresh frozen plasma and 6 pieces of cryoprecipitates. The patient

underwent urgent re-sternotomy and second review of sutures, operation field. After re-opening of the sternum, a solid hematoma around the aortic root was observed, about 150 ml of clot was removed. Active bleeding from the prosthetic connections was not observed.

After a successful suspension of bleeding, restoration of hemostasis the patient was treated at the Intensive Care for 4 days.

Intravenous heparin (dose adjusted according to ADTL), then warfarin (a dose adjusted according to clotting rates) were given to prevent thrombosis.

Eight days after surgery, echocardiography was repeated. The aortic valve was functioning well, mild central regurgitation was observed, no additional masses on the valves were visible, LV diameter was decreased (48 mm) and LV EF was normal (55%).

Further treatment with warfarin was recommended for up to 3 months, following aspirin for up to one year.

The follow-up echocardiography after 3 months demonstrated a well-functioning prosthesis of ascending aorta, mild aortic and more significant reduction of LV mass.

Figure 1. Chest X-ray

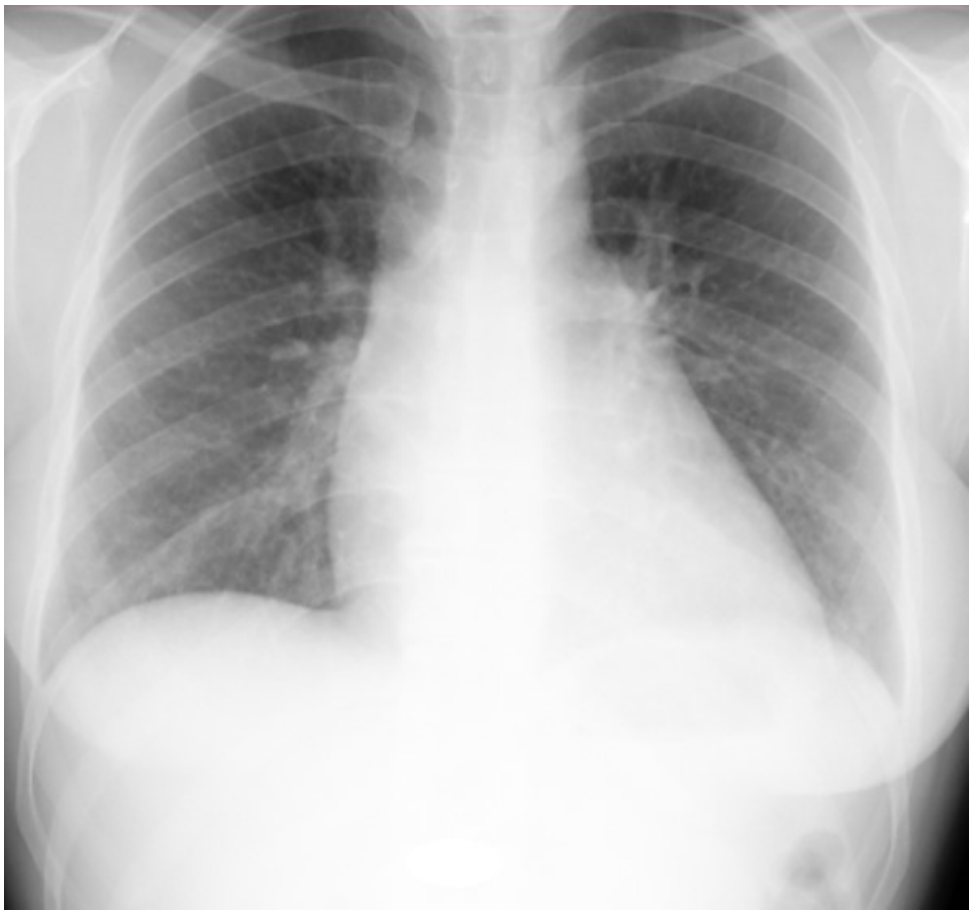
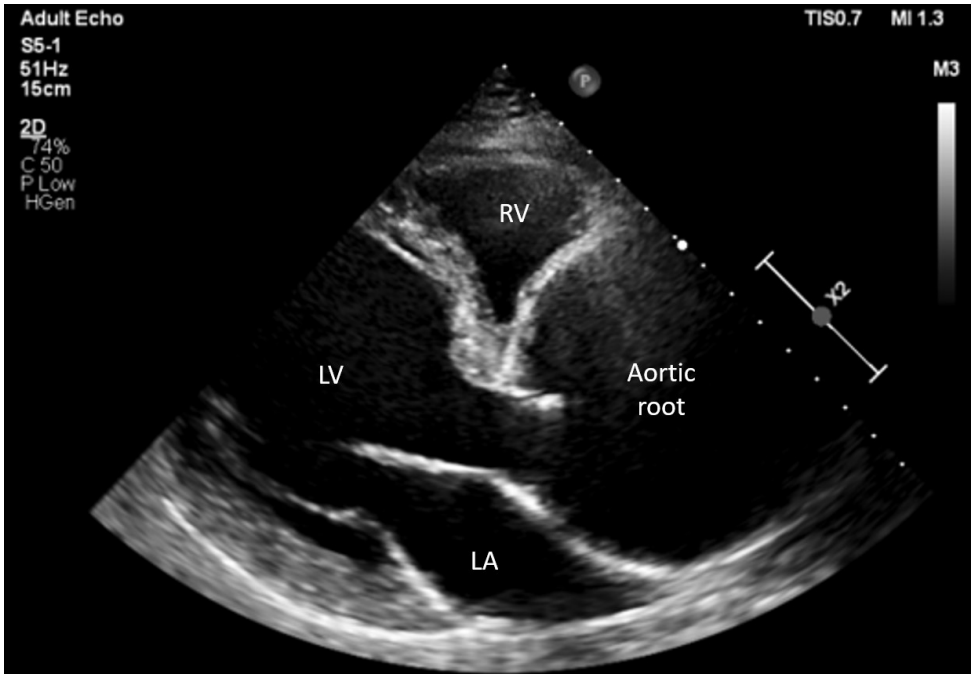
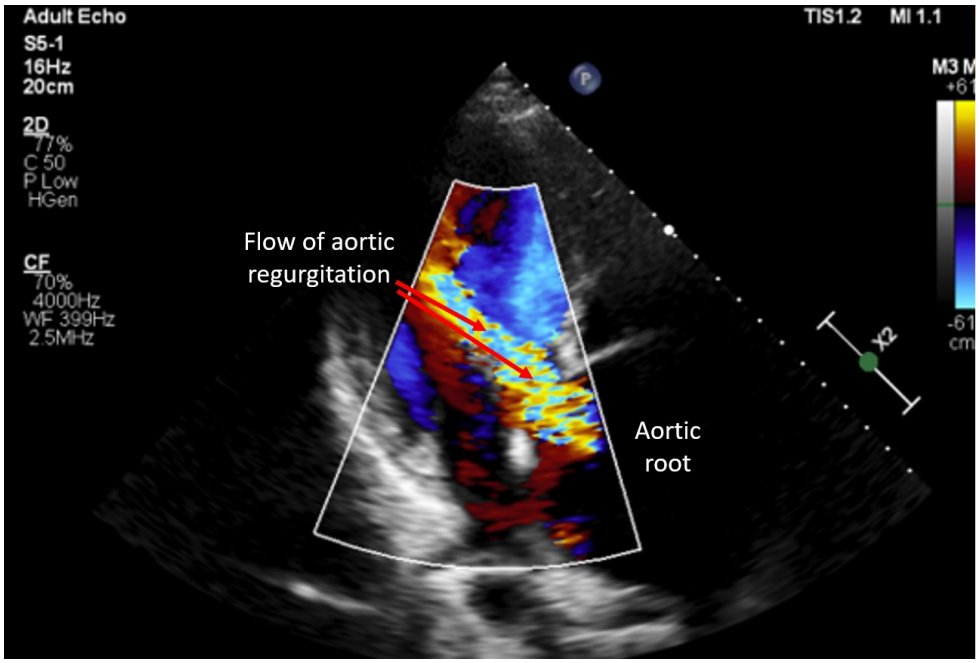


Figure 2. 2D transthoracic echocardiography



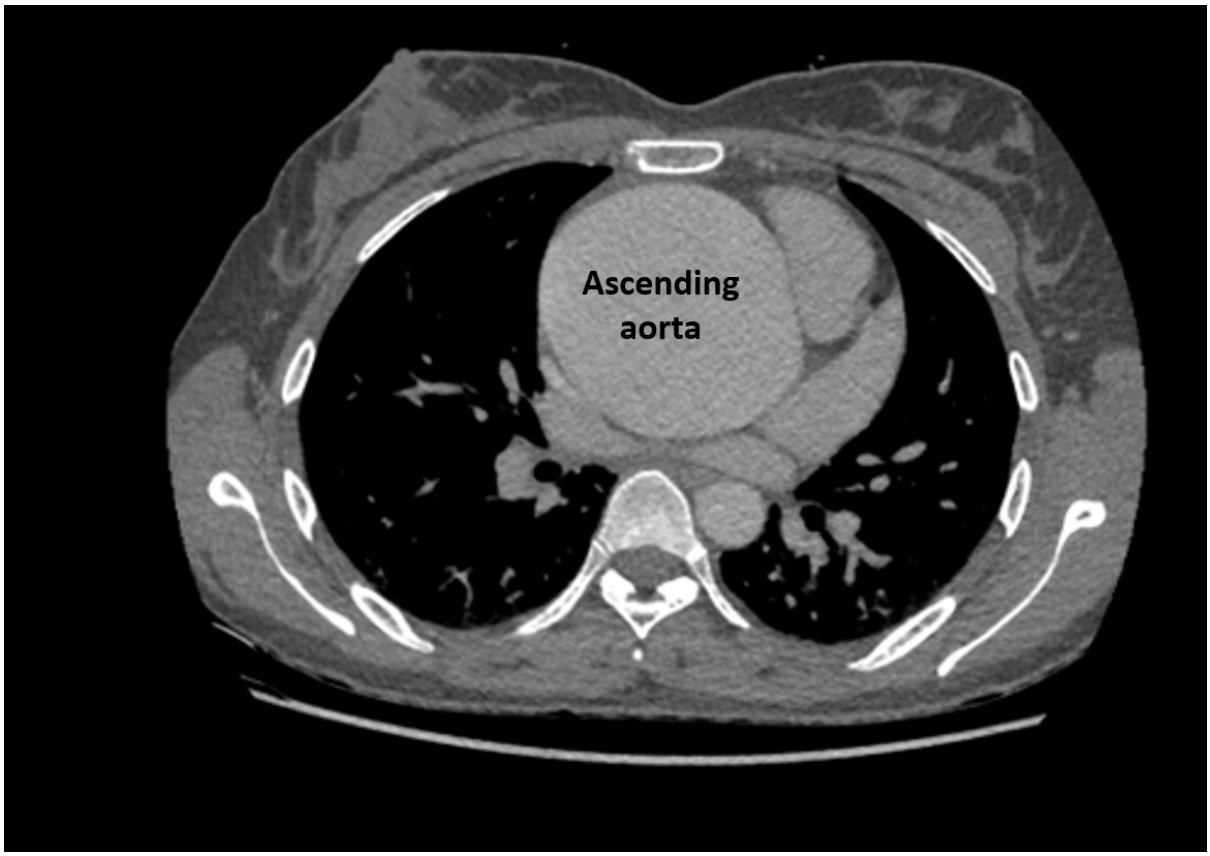
Parasternal long-axis view: severe dilation of the aortic root and dilated LV

Figure 3. Severe aortic regurgitation in 2D transthoracic echocardiography



Apical three chambers view: severe aortic regurgitation

Figure 4. CT angiography



Severe dilation of ascending aorta

3. DISCUSSION

This case report shows that the aneurysm of ascending aorta could be present at a young age without any specific obvious predisposing factors such as congenital bicuspid aortic valve, phenotypic signs of Marfan syndrome or known familiar history. The patient was asymptomatic possibly for a long time. She even gave birth without complications. Fortunately, a huge aneurysm was established before the dissection occur and life-threatening complication was avoided.

Thoracic aortic aneurysms (TAA) are common in cardiovascular practice. The prevalence of TAA is estimated to be 6 persons per 100,000 per year (6). TAA are most often clinically not detectable and can be identified incidentally when an imaging examination is performed prophylactically. Initial symptoms of TAA most often are potentially fatal conditions such as aorta dissection or rupture. Over 95% of thoracic aortic aneurysms are asymptomatic

until the point of dissection or rupture (6). In various studies, TAA occurrence was 0.16–0.34 % (7).

About 20% of patients with aortic dissection are dying before reaching the hospital (8). Without treatment, the mortality rate is 1 to 3 %/hour during the first 24 hours, 30% during 1 week, 80% during 2 weeks, and 90% during 1 year (8). The hospital mortality rate for treated patients is about 30% in cases of proximal dissection and 10% when the distal part is dissected (8). For treated patients who survive the acute episode, the survival rate is about 60% during 5 years and 40% during 10 years (8).

Aneurysms of the ascending thoracic aorta most often result from cystic medial degeneration during smooth muscle cell and elastic fiber loss. Medial degeneration leads to weakening of the aortic wall, which results in aortic dilatation and aneurysm formation. In the long term, it is also very important to determine all possible causes of aneurysms for a better outcome.

The aortic aneurysm can be caused by several diseases. Frequently it is related to connective tissue diseases such as Marfan syndrome, Ehlers–Danlos syndrome, or familial aneurysms syndrome. According to the other researches aneurysms can also occur in congenital diseases such as Turner syndrome, tetralogy of Fallot, bicuspid aortic valve (BAV), Takayasu's arteritis, syphilitic aortitis. In this reported case, according to diagnostic tests – echocardiography, laboratory tests - syphilis, BAV and tetralogy of Fallot were denied. In this case, only a few causes can be considered.

First of them is Marfan syndrome - a heritable autosomal-dominant disorder caused by mutations in one of the genes for fibrillin-1. Forementioned structural protein is the major component of microfibrils of elastin. The mutations result a decrease of elastin in the aortic wall. Consequently, the aorta loses elastic properties that lead to its stiffness and dilatation. This is considered as the main cause of ascending aorta aneurysms in patients of young age. Up to 80% of patients with Marfan syndrome have dilatation of ascending aorta (9). The data of several studies suggested that although cases of thoracic aortic aneurysms in the absence of overt connective-tissue disorders may be sporadic (10), they are often familial and are referred to as the familial thoracic aortic aneurysm syndrome. In an analysis of their large database of thoracic aortic aneurysm patients, Coady MA and colleagues found that at least 19% of patients had a family history of a thoracic aortic aneurysm, and they presented at significantly younger ages than did those with sporadic aneurysm (10). In this case, the family history of aneurysms is not known but relatives were not fully investigated. It should be done in the nearest future.

The third suspected cause of an aortic aneurysm is Takayasu's arteritis. It is a chronic inflammatory disease of unknown etiology. The disease affects women far more often than men, and the mean age at the time of diagnosis

is 29 years (10). It typically causes obliterative luminal changes in the aorta and other involved arteries. However, in 15% of cases, aortic dilatation may occur and result in aneurysms (10). Therefore, if TAA occurs in a young woman with symptoms of a systemic inflammatory process, the doctor should consider Takayasu's arteritis. In this case – the chronic inflammatory process according to the laboratory tests was denied.

Ehlers–Danlos syndrome (EDS) is one more possible option which could be responsible for aortic dilation. It is characterized by the laxity of the joints and skin disorders. Up to 28% of patients with EDS (all types confounded) present with ascending aorta dilatation (11). A recent retrospective study suggested that these aortas seem to normalize in size when children with EDS become adults (9). However, type IV EDS (autosomal dominant disorder) is characterized by characteristic skin manifestations associated with arterial, uterine, and intestinal dissection and rupture (9). Aneurysm formation is a usual cause for arterial complications, but they can also occur spontaneously. This syndrome is associated with the COL3A1 mutation, and the diagnosis can be made by DNA amplification or by collagen analysis (9).

One more rare but possible cause of an aortic aneurysm is Turner syndrome which is associated with several cardiovascular anomalies, including a bicuspid aortic valve and coarctation of the aorta. Thoracic aortic aneurysms are also found and typically involve the ascending aorta.

It is always–essential to assess risk factors. Moreover, hypertension and smoking appear to accelerate the process by increasing elastolytic enzymes in the aortic medial layer (9,12). In many research hypertension increases ascending aorta dilatation and predisposes of formation of TAA. Unlike inherited forms of ascending aortic aneurysms, hypertension related TAAs complicate at diameters over 6.0 cm and the risk of complications increases exponentially with the further increase in diameter (9).

However, we should consider the possibility of frequent secondary causes. One of them is atherosclerosis, which has long been considered as the second cause of aortic aneurysm formation. The atheromatous plaques destroy small muscle cells and elastic fiber architectures, and as a result, the aortic wall becomes weaker (9).

The main treatment of aneurysms is surgical. Therefore, medical treatment helps to reduce the risk of rupture. However, the medical therapies are quite limited. In a randomized study of adults with Marfan syndrome, Shores et al. found that treatment with propranolol (versus no β-blocker therapy) over 10 years resulted in a significantly slower rate of aortic dilatation, fewer aortic events, and lower mortality (10). Unfortunately, whether these benefits can truly be extrapolated to the non-

Marfan population with thoracic aneurysms remains unknown.

In this presented case the predominant cause of aortic aneurysm was not determined. The patient was referred to outpatient genetic investigations. Genetic findings will not change the tactics of treatment but according to the genetic findings other family members, such as parents, other siblings or the patient's children should also get examined for possible TAA.

During lifetime the disease could progress, and the patient must be monitored.

CONCLUSION

The aneurysm of ascending aorta is a lethal disease and has a potentially high mortality. Lifestyle changes, medical treatment, control of risk factors and serial imaging assessment of aortic aneurysm are also very important in the management of these patients. Comprehensive investigations for possible causes of aneurysms are essential for better outcomes. Results of treatment strongly depend on etiology because an aneurysm could be only one component of specific composite syndromes. Strict and regular life-long follow-up is indicated to detect continuous dilatation of ascending aorta, to assess the severity of aortic regurgitation in order to achieve a more favorable prognosis.

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