

Ascending Aortic Aneurysm in an Asymptomatic Young Woman Without Risk Factors



Amrin Kharawala, MD, Diego Barzallo, MD, Jincy Thankachen, MD, and Rosy Thachil, MD, *Bronx, New York*

INTRODUCTION

Ascending thoracic aortic aneurysm (asc TAA) is defined as localized dilation of the aorta to more than 50% of its predicted size.¹ The average size of the ascending aorta is 28.6 mm on a chest x-ray, while the normal range of the aortic root is 36 to 39 mm in men and 35 to 37 mm in women as measured on chest computed tomography (CT).¹ Thoracic aortic aneurysms (TAAs) can be classified based on their location (which may involve 1 or more aortic segments): aortic root and/or ascending aorta (60%), aortic arch (10%), or descending aorta (40%).² Ascending aortic aneurysms usually occur due to cystic medial degeneration, which causes weakening of the aortic wall leading to dilation and aneurysm formation. It can occur due to aging, and hypertension is known to accelerate the process.² Sporadic asc TAA is usually seen in patients by 60 years of age due to reduced elasticity and compliance predisposing to dilation.³ In the younger age group, asc TAA can be familial, associated with connective tissue disorders like Marfan, Ehlers Danlos, Loeys-Dietz, and Turner syndrome, or secondary to bicuspid aortic valve or untreated tertiary syphilis.² Takayasu arteritis is another cause that can rarely lead to asc TAA and tends to affect women more commonly than men, with the mean age of diagnosis being approximately 30 years of age.⁴ We present a 39-year-old woman who was incidentally found to have a 65-mm large asc TAA despite the absence of predisposing conditions or clinical symptoms on the initial visit.

History of Presentation

A 39-year-old woman with no cardiovascular risk factors presented to the clinic for atraumatic wrist pain. Review of systems included palpitations. The patient denied chest pain, shortness of breath, dizziness, and syncope. On the follow-up visit, the patient mentioned experiencing palpitations lasting for 30 minutes that were associated with mild intermittent substernal chest pain radiating to the back and neck, which improved at rest. Initially, the patient had ignored these symptoms and attributed the palpitations to panic attacks. The patient was of average build with no marfanoid characteristics. Vital signs

were normal, with a temperature of 98.9 °F, heart rate of 68, respiratory rate of 18, and blood pressure of 129/61 mm Hg. Cardiovascular exam was significant for a grade 4 diastolic murmur, best heard in the right upper sternal border, along with thrill over the precordium. The patient had an uncomplicated pregnancy 4 years ago after 2 miscarriages 15 years ago. There was no history of rheumatic heart disease, syphilis, childhood illnesses, congenital disorders, or connective tissue disorders. Family history was negative for inherited disorders, sudden death, or aneurysms. There was a remote history of smoking 1 pack-year without any alcohol or drug use. Differential diagnosis included paroxysmal arrhythmias including atrial fibrillation, cardiac angina, valvular heart disease like aortic or pulmonary regurgitation, mitral or tricuspid stenosis, and aortic root dilation with ascending aortic aneurysm. Laboratory tests were only significant for a weakly positive anti-citrullinated peptide antibody. Anti-nuclear antibody and extractable nuclear antigen antibody panel, syphilis, thyroid-stimulating hormone, rheumatoid arthritis factor, serum complement levels, infectious workup, and inflammatory markers were negative. Chest x-ray was negative for aortic abnormalities (Figure 1), electrocardiogram showed normal sinus rhythm, and Holter monitor revealed no abnormality. A transthoracic echocardiogram (TTE) was performed due to the presence of a diastolic murmur and nonspecific cardiac symptoms, which showed moderate to severe aortic regurgitation with ascending aortic aneurysm and aortic root dilation to 65 mm (Figure 2, Video 1). Aortic regurgitation was graded as moderate to severe as the vena contracta was 8 mm, pressure half time was 364 ms, and jet diameter/left ventricular outflow tract was 0.60 (Figure 3, Video 2). Chest CT showed that the proximal ascending aorta is dilated to 63 mm. The aortic root was dilated to 49 mm (Figures 4 and 5). As the aortic root diameter was well over 60 mm, the patient underwent Bentall's surgery with replacement of the ascending aorta including the aortic root and valves¹ (Figure 6). The surgical pathology report showed myxoid degenerative changes in both the aortic valve and ascending aorta.

The postoperative course was complicated by stroke with left hemiplegia, hemineglect, and right gaze preference. Head CT revealed a subdural hematoma, and video electroencephalogram showed frequent right frontocentral spike and slow waves indicating increased cortical hyperexcitability without any seizures. The patient also developed a new complete heart block, complicated by poor epicardial lead capture in the transvenous pacemaker. This led to bradycardia and asystole, requiring cardiopulmonary resuscitation, which resulted in seizure-like activity. Antiepileptic medications were started prophylactically, and the patient underwent permanent pacemaker implantation, after which the patient was sent for in-patient rehabilitation. Five months after the surgery, the patient was asymptomatic without any cardiopulmonary limitations. The permanent pacemaker is functioning without any complications. Metoprolol 25 mg and warfarin for the mechanical aortic valve were continued. Transthoracic

From the Jacobi Medical Center/Albert Einstein College of Medicine, Bronx, New York.

Keywords: Ascending aortic aneurysm, Aortic root dilation, Women, Asymptomatic

Correspondence: Amrin Kharawala, MD, Jacobi Medical Center, 2433 Fenton Avenue, Bronx, NY 10469. (E-mail: Amrinkharawala95@gmail.com).

Copyright 2022 by the American Society of Echocardiography. Published by Elsevier Inc. This is an open access article under the CC BY-NC-ND license (<http://creativecommons.org/licenses/by-nc-nd/4.0/>).

2468-6441

<https://doi.org/10.1016/j.case.2022.11.005>

VIDEO HIGHLIGHTS

Video 1: Two-dimensional TTE, parasternal long-axis view, demonstrates the severely dilated, 65-mm aneurysmal ascending aorta. Also seen is a moderately dilated left ventricle with normal global systolic function.

Video 2: Two-dimensional TTE, parasternal long-axis view with color flow Doppler, demonstrates the moderate-severe functional aortic regurgitation (vena contracta, 8 mm; jet diameter/left ventricular outflow tract ratio, 0.60; pressure half time, 364 ms) that is associated with the asc TAA.

View the video content online at www.cvcasejournal.com.

echocardiography showed normal biventricular systolic function with a left ventricular ejection fraction of 55% and normal mechanical aortic valve function (Figure 3). The patient underwent rehabilitation, which helped them to regain power in their extremities, and has remained seizure free on levetiracetam.

DISCUSSION

The incidence of ascending aortic aneurysm is around 10 per 100,000 person-years.⁵ The usual age of diagnosis of TAA in women is by 60 to 70 years of age, and it is deemed that women have worse prognosis with a higher rate of aneurysm growth compared to men.^{5,6} Ascending aortic aneurysm can be clinically silent or present with classic symptoms like chest or upper back pain, dyspnea, or symptoms related to compression of nerves or vessels. Additionally, diastolic murmurs are often missed on physical exam when the patient does not present with cardiac complaints. However, based on the knowledge that diastolic murmurs are always pathological, we pursued further investigation in our patient.

On an in-depth query of the literature, we found a similar presentation of asc TAA in a 29-year-old man with a history of murmur and family history of bicuspid aortic valve who was incidentally diagnosed to have chronic type A aortic dissection, despite a lack of clinical symptoms. Even in this case, the physical exam findings of aortic regurgitation led to further investigations and eventual surgical repair with favorable outcomes.⁷ In another similar case of a large asc TAA in a 45-year-old woman without risk factors, medical management was pursued with symptomatic improvement for at least 2 years post-follow-up.⁸

Our patient did not have any signs of an overt connective tissue disorder and is relatively young to have a sporadic aneurysm. Marfan syndrome was ruled out based on the revised Ghent criteria.⁹ Coady *et al.*¹⁰ found that at least 19% of patients in their TAA database had a family history of TAA and presented at significantly younger ages. Our patient could possibly have familial thoracic aortic syndrome, which tends to have an autosomal dominant mode of inheritance; however, the family history was negative for asc TAA.² The 3p24.2-25 mutation is known to cause both isolated and familial TAAs, and hence genetic testing along with histological evidence of cystic medial degeneration can aid in diagnosing the etiology.² In our case, due to the unavailability of genetic testing, we were unable to confirm the cause; however, it can be postulated that the variability in the gene expression and penetrance could have led to a negative family history.

In terms of medical treatment, the drug most frequently used, beta-blocker, has proven efficacy in TAA in Marfan syndrome; however, beta-blockers' efficacy in the general population is still questionable, with mixed and inconclusive results in clinical trials. Other promising drugs for TTA are angiotensin receptor blockers, matrix metalloproteinase inhibitors, and statins that have shown encouraging results in animal models and observational studies; however, their clinical efficacy has not been consistently probed.¹¹ Further management should include screening the patient's immediate family via TTE to rule out familial causes for asc TAA, and chest CT may be considered for adult men and women beyond child-bearing age.

Aortic root dilation can lead to significantly worse outcomes than mid ascending aorta dilation.¹² It is known that older women with asc TAA tend to have a higher 30-day mortality postsurgery, but

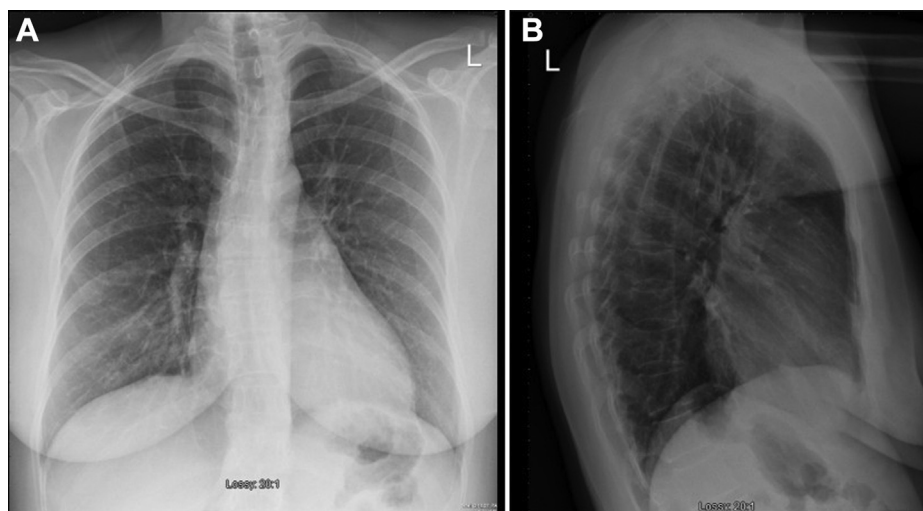


Figure 1 Chest x-ray. (A) Posterior-anterior view of chest x-ray showing a surprisingly normal appearing aorta. (B) Left lateral view of chest x-ray showing a surprisingly normal appearing aorta.

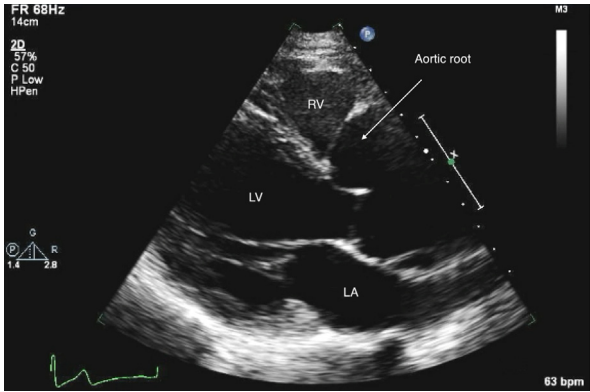


Figure 2 Two-dimensional TTE, parasternal long axis, diastolic phase, demonstrates the ascending aortic aneurysm with severely dilated aortic root to 65 mm. An artifact is seen at the posterior aspect of the aortic root without any evidence of aortic dissection. LA, Left atrium; LV, left ventricle; RV, right ventricle.

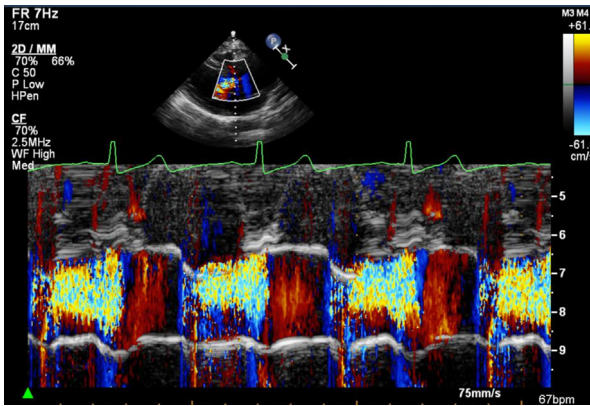


Figure 3 Color Doppler M-mode TTE, parasternal long-axis view of the aortic regurgitation demonstrates a jet diameter/left ventricular outflow tract ratio of 0.6.

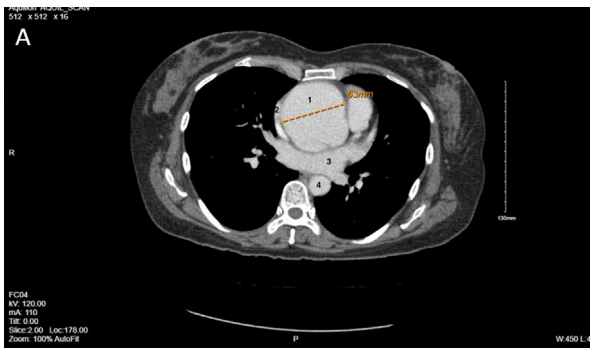


Figure 4 Chest CT with contrast, axial view, demonstrates the (1) 63-mm ascending aortic aneurysm, (2) the right atrium, (3) the left atrium, and (4) the descending aorta.

very little is known about the incidence and prognosis of asc TAA in younger women.¹³ It can be postulated that despite the lack of risk factors, there may be other pathological mechanisms leading to asymptomatic enlargement of the ascending aorta, which needs further

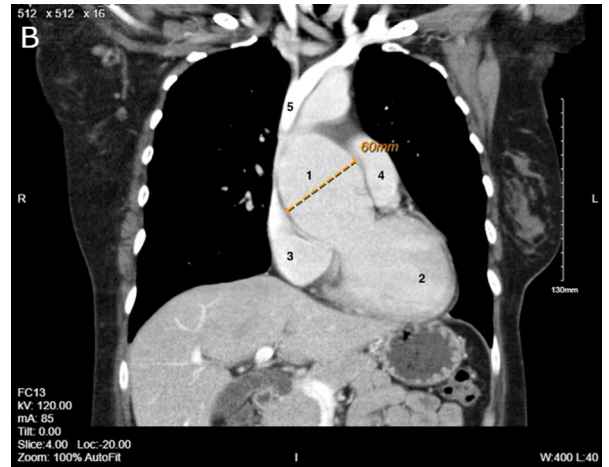


Figure 5 Chest CT with contrast, coronary view, demonstrates the (1) 60-mm ascending aortic aneurysm, (2) the left ventricle, (3) the right atrium, (4) the pulmonary trunk, and (5) the superior vena cava.

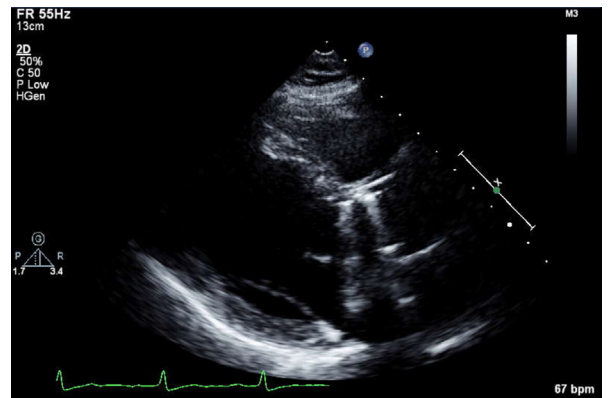


Figure 6 Two-dimensional TTE, parasternal long-axis, diastolic phase, demonstrates postoperative findings with the repaired aorta and the mechanical AVR with associated metallic artifacts. AVR, Aortic valve replacement.

evaluation and research. Although our patient did not appear to have a diagnosis of connective tissue disorder, given the lack of other significant risk factors, it is possible that our patient has an underlying metalloprotease inhibitor abnormality, familial thoracic aortic syndrome, or another undifferentiated connective tissue disease. Genetic testing and further rheumatologic workup may be helpful.

CONCLUSION

Ascending aortic aneurysms can be present in young individuals even without the presence of overt risk factors or a positive family history. Therefore, all patients with a diastolic heart murmur and aortic regurgitation should be investigated further with TTE.

ETHICS STATEMENT

The authors declare that the work described has been carried out in accordance with the following guidelines: CARE guidelines for Case Reports.

CONSENT STATEMENT

The authors declare that since this was a non-interventional, retrospective, observational study utilizing de-identified data, informed consent was not required from the patient under an IRB exemption status.

FUNDING STATEMENT

The authors declare that this report did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

DISCLOSURE STATEMENT

The authors report no conflict of interest.

SUPPLEMENTARY DATA

Supplementary data to this article can be found online at <https://doi.org/10.1016/j.case.2022.11.005>.

REFERENCES

- Hiratzka LF, Bakris GL, Beckman JA, Bersin RM, Carr VF, Casey DE Jr, et al. 2010 ACCF/AHA/AATS/ACR/ASA/SCA/SCAI/SIR/STS/SVM guidelines for the diagnosis and management of patients with thoracic aortic disease: a report of the American College of Cardiology Foundation/American Heart Association task force on practice guidelines, American Association for Thoracic Surgery, American College of Radiology, American Stroke Association, Society of Cardiovascular Anesthesiologists, Society for Cardiovascular Angiography and Interventions, Society of Interventional Radiology, Society of Thoracic Surgeons, and Society for Vascular Medicine. *Circulation* 2010;121:e266-369.
- Isselbacher EM. Thoracic and abdominal aortic aneurysms. *Circulation* 2005;111:816-28.
- Martin C, Sun W, Primiano C, McKay R, Elefteriades J. Age-dependent ascending aorta mechanics assessed through multiphase CT. *Ann Biomed Eng* 2013;41:2565-74.
- Procter CD, Hollier LH. Takayasu's arteritis and temporal arteritis. *Ann Vasc Surg* 1992;6:195-8.
- Saliba ES. The ascending aortic aneurysm: when to intervene? *Int J Cardiol Heart Vasc* 2015;6:91-100.
- Cheung K, Boodhwani M, Chan KL, Beauchesne L, Dick A, Coutinho T. Thoracic aortic aneurysm growth: role of sex and aneurysm etiology. *J Am Heart Assoc* 2017;6:e003792.
- Tung RT, Heyns J, Dryer L, Eitutus B. Asymptomatic, chronic type-A dissection of a large ascending thoracic aortic aneurysm in a young patient. *Kans J Med* 2020;13:244-7.
- Huliyurdurga Srinivasasetty NS, Thagachagere Ramegowda R, Kharge J, Bachahalli Krishnanayak G, Patil SS, Raj V, et al. Unusual non progressive idiopathic giant ascending aortic aneurysm-A rarity. *Int J Surg Case Rep* 2016;25:203-6.
- Salik I, Rawla P. In: StatPearls [Internet]. Marfan Syndrome; 2022.
- Coady MA, Davies RR, Roberts M, Goldstein LJ, Rogalski MJ, Rizzo JA, et al. Familial patterns of thoracic aortic aneurysms. *Arch Surg* 1999;134:361-7.
- Chun AS, Elefteriades JA, Mukherjee SK. Medical treatment for thoracic aortic aneurysm—much more work to be done. *Prog Cardiovasc Dis* 2013;56:103-8.
- Kalogerakos PD, Zafar MA, Li Y, Mukherjee SK, Ziganshin BA, Rizzo JA, et al. Root dilatation is more malignant than ascending aortic dilation. *J Am Heart Assoc* 2021;10:e020645.
- Beller CJ, Farag M, Wannaku S, Mesana T, Karck M, Veinot JP. Gender-specific differences in outcome of ascending aortic aneurysm surgery. *PLoS One* 2015;10:e0124461.