

## RARE BUT DEADLY FINDINGS DON'T MISS THESE

# Late Presentation of Massive Aortic Aneurysm with Aortic Regurgitation



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### INTRODUCTION

Aortic regurgitation (AR) is a valvular heart disease characterized by the backflow of blood from the aorta to the left ventricle in diastole.<sup>1</sup> The clinical manifestations of this disease can range from asymptomatic to clinical symptoms and signs of congestive heart failure, with various etiologies.<sup>1</sup> Here, we describe a case of asymptomatic severe AR caused by marked aneurysmal dilation of the ascending aorta.

### CASE PRESENTATION

A 55-year-old man with a medical history of tobacco abuse (20 pack-years) presented to the cardiology clinic after being referred by his primary care physician for a murmur. The patient stated that he had never been told he had a murmur; however, he did not see doctors regularly. He denied any prior personal or family history of heart disease. He denied any symptoms. His vital signs were stable, with a blood pressure of 141/65 mm Hg and a heart rate of 81 beats/min. On physical examination, there was an apical heave, grade 3/6 systolic and early diastolic murmur heard loudest at the right upper sternal border, prominent carotid pulsation, bounding pulses, and visible capillary pulsations with light compression of the nail bed (Quincke's sign). Transthoracic echocardiography was performed, revealing a large aortic aneurysm measuring 7 cm at the sinus of Valsalva (Figure 1) with severe torrential AR (Video 1). The pressure half-time was measured at 248 msec (Figure 2), and there was holodiastolic reversal of flow in the ascending aorta (Figure 3). There was severe left ventricular (LV) enlargement (Video 2), with LV internal diameter in diastole of 7.5 cm (or 3.61 cm/m<sup>2</sup> when indexed to body surface area), LV end-diastolic volume of 395 mL, LV end-systolic volume of 225 mL, and a reduced LV ejection fraction (LVEF) of 43% as obtained using the Simpson biplane method. LV measurements were suggestive of eccentric hypertrophy given an LV mass index of 179 g/m<sup>2</sup> and relative wall thickness of 0.27 on the basis of American Society of Echocardiography chamber quantification guidelines.<sup>2</sup> Marked aneurysmal dilation of the ascending aorta was also seen in the apical views (Videos 3 and 4).

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The aortic valve short-axis views obtained (Figure 4, Video 5) were probably off axis despite multiple attempts, but bicuspid aortic valve cannot be excluded given that concomitant aortopathy and the eccentric closure of the aortic valve (Figure 5) increase the likelihood of bicuspid aortic valve. Furthermore, the patient did not have any definitive association with other potential causes of aortopathy, such as Marfan syndrome, Turner syndrome, or Ehlers-Danlos syndrome. Computed tomographic angiography showed aneurysm of the ascending aorta at the sinus of Valsalva, and the maximal ascending aortic dimension was 7.2 cm, with no evidence of dissection (Figure 6). Cardiothoracic surgery was consulted given his large aortic aneurysm (>5.5 cm) with concomitant decreased LVEF (<55%) and chronic severe AR while asymptomatic, which meets class I recommendations for surgical intervention with a Bentall procedure.<sup>3</sup> Left heart catheterization was scheduled before surgery to evaluate for coronary artery disease; however, the patient declined further interventions and did not follow up. Despite several attempts, we were unable to reestablish contact with the patient.

### DISCUSSION

Severe AR can present acutely with advanced heart failure and increased risk for early mortality if untreated.<sup>4</sup> However, as seen in our patient, most cases of severe AR are chronic and present late after many years of being asymptomatic.<sup>5</sup> The prevalence of chronic severe AR is unknown, but a study done in the United States showed the prevalence of AR to be about 4.9%, occurring mostly in men and with the incidence increasing with age, with most cases seen at age 50 years and older.<sup>5</sup>

AR can be caused by a variety of diseases, one of the most common causes in the United States being ascending aortic disease, in which aneurysmal dilatation causes noncoaptation of the aortic valve leaflets.<sup>3,5</sup> Ascending aortic aneurysm also occurs more commonly in men and with increasing age, with most cases being asymptomatic.<sup>6</sup> The risk for dissection or rupture of the aneurysm increases with increasing diameter, with greater risks occurring at  $\geq 5.5$  cm.<sup>6</sup> Our patient had a large aneurysm measuring 7.2 cm, thus increasing his risk for morbidity and mortality.

Chronic severe AR leads to an increase in LV volume, which over time results in LV dilation and hypertrophy. However, if left untreated, the left ventricle decompensates because of LV volume overload, resulting in systolic dysfunction and a decrease in LVEF. Although patients may remain asymptomatic initially, the LV decompensation will lead to elevated filling pressures, decreased cardiac output, and eventually heart failure symptoms.<sup>1,3</sup> Our patient, though asymptomatic, had already developed LV systolic dysfunction, thus increasing his risk for morbidity and mortality even further.

Aortic root dilation can be due to various etiologies, and when observed, possible differential diagnosis such as bicuspid aortic valve, Marfan syndrome, Turner syndrome, and Ehlers-Danlos syndrome

**VIDEO HIGHLIGHTS**

**Video 1:** Two-dimensional transthoracic echocardiographic parasternal long-axis view demonstrating a severe aortic regurgitant jet with a dilated aortic root and gross LV enlargement.

**Video 2:** Two-dimensional transthoracic echocardiographic apical four-chamber view demonstrating a grossly enlarged left ventricle with mild systolic dysfunction with an LVEF of 40% to 45%.

**Video 3:** Two-dimensional transthoracic echocardiographic apical five-chamber view imaged off axis showing a visualized portion of the proximal ascending aorta with aneurysmal dilation (*orange star*).

**Video 4:** Two-dimensional transthoracic echocardiographic apical three-chamber view demonstrating aneurysmal dilation of the ascending aorta.

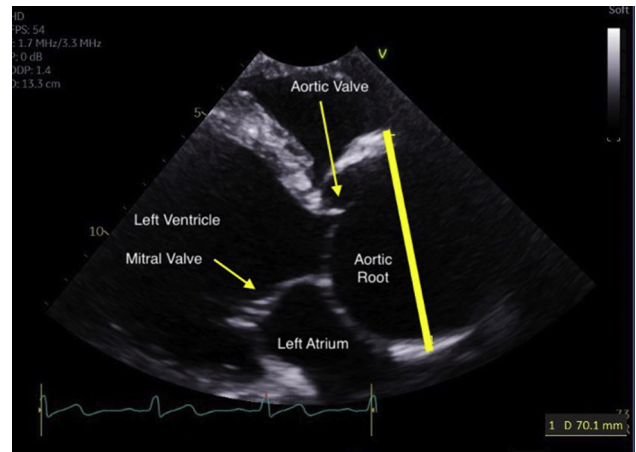
**Video 5:** Two-dimensional transthoracic echocardiographic parasternal short-axis view of the aortic valve demonstrating a markedly dilated aortic root (*orange star*).

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type 4, among others, should be considered.<sup>1,7</sup> During evaluation of these patients, certain features may point toward a certain diagnosis. For example, a patient with Marfan syndrome may be tall and thin and have long extremities, scoliosis, enophthalmos, and dental crowding.<sup>8</sup> In our patient, these features were not present, but echocardiography was suggestive of a possible bicuspid aortic valve.

Echocardiography is vital for the diagnosis of valvular heart diseases such as AR<sup>9</sup> and, as in this case, also useful in the early diagnosis of ascending aortic aneurysm. Transthoracic echocardiography is readily available, is painless, and yields quick and accurate diagnosis of enlargement of the aortic root and ascending aorta.<sup>10,11</sup> Furthermore, it is helpful in the assessment of AR severity, such as the demonstration of holodiastolic flow reversal in the aortic arch and abdominal aorta as a specific sign of severe AR.<sup>9</sup> In our case, the ascending aorta was used to demonstrate this because of technical limitations with pulsed-wave Doppler of the descending aorta. Although computed tomographic angiography is a mainstay in the diagnosis of ascending aortic aneurysm, echocardiography is a reliable tool for screening and monitoring.<sup>6</sup> Magnetic resonance angiography is also accurate in the diagnosis of ascending aortic aneurysm, but it is not widely available.<sup>6</sup> Cardiac magnetic resonance imaging and cardiac catheterization are useful diagnostic tools in evaluating the severity of AR, LV volumes, LV mass, LVEF, and coronary artery anatomy, particularly before planning treatment strategies.<sup>3,9,12</sup>

Severe asymptomatic AR is treated medically with vasodilator therapy by controlling hypertension, but surgical valve replacement is indicated with symptoms and/or LV systolic dysfunction and severe LV dilation.<sup>3,5,12,13</sup> In cases such as ours in which there is concomitant ascending aortic aneurysm, aortic valve replacement along with surgical repair or replacement of the ascending aorta is indicated if the diameter is  $>5.5$  cm.<sup>3,5</sup> The prognosis of severe asymptomatic AR is improved if LV systolic dysfunction and LV dilation are promptly recognized and surgery is performed, thus decreasing mortality.<sup>14</sup> The rate of progression to symptoms and LV systolic dysfunction is



**Figure 1** Two-dimensional transthoracic echocardiographic parasternal long-axis view zoomed in on the aortic valve and root. The leading edge-to-leading edge technique was used to measure the aortic root at just over 7 cm.

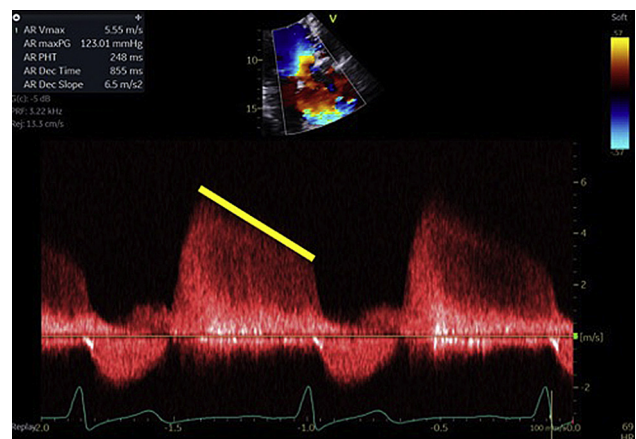
reported to be 3% to 6% per year,<sup>13</sup> and the rate of growth of ascending aortic aneurysms has been reported at 0.07 cm/y.<sup>15</sup>

**CONCLUSION**

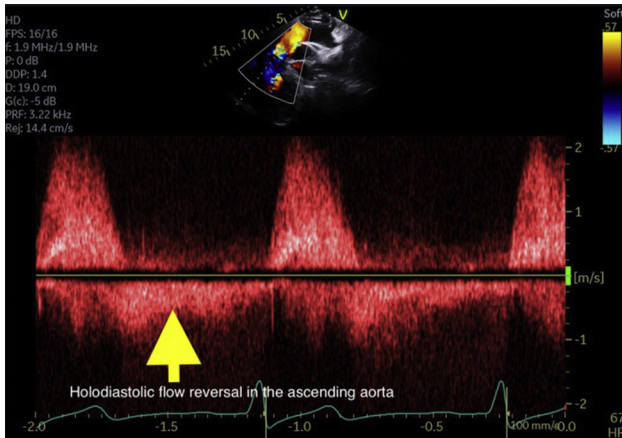
Severe AR, a possible sequela of marked aneurysmal dilation of the ascending aorta, may be asymptomatic but not benign. Its chronicity leads to LV dilation and dysfunction, causing significant morbidity and mortality if left untreated. Transthoracic echocardiography is a vital tool for early diagnosis.

**ACKNOWLEDGMENT**

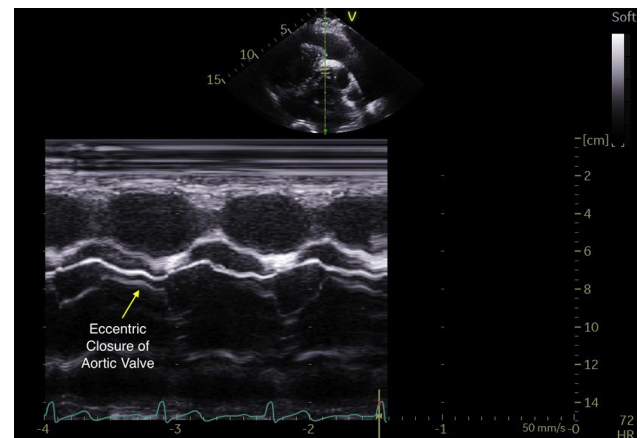
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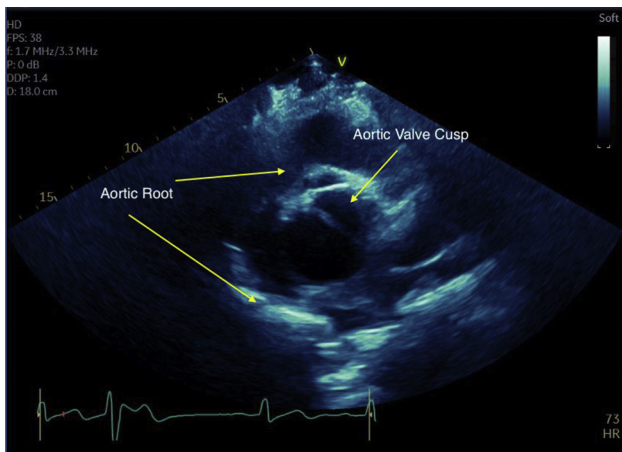
**Figure 2** Two-dimensional transthoracic echocardiographic apical long-axis view with continuous-wave Doppler across the aortic valve demonstrating a regurgitant pressure half-time of 248 msec.



**Figure 3** Two-dimensional transthoracic echocardiographic suprasternal view with continuous-wave Doppler of aortic flow demonstrating holodiastolic flow reversal in the ascending aorta.



**Figure 5** Two-dimensional transthoracic echocardiographic parasternal short-axis view M-mode image demonstrating eccentric closure of the aortic valve.



**Figure 4** Two-dimensional transthoracic echocardiographic parasternal short-axis view of the aortic valve demonstrating a grossly dilated aortic root.

#### SUPPLEMENTARY DATA

Supplementary data to this article can be found online at <https://doi.org/10.1016/j.case.2021.11.004>.

#### REFERENCES

1. Bekerdejian R, Grayburn PA. Valvular heart disease. *Circulation* 2005;112:125-34.
2. Lang RM, Badano LP, Mor-Avi V, Filalo J, Armstrong A, Ernande L, et al. Recommendations for cardiac chamber quantification by echocardiography in adults: an update from the American Society of Echocardiography and the European Association of Cardiovascular Imaging. *J Am Soc Echocardiogr* 2015;28:1-3914.
3. Otto CM, Nishimura RA, Bonow RO, Carabello BA, Erwin JP, Gentile F, et al. 2020 ACC/AHA guideline for the management of patients with valvular heart disease: a report of the American College of Cardiology/American Heart Association Joint Committee on Clinical Practice Guidelines. *Circulation* 2021;143:e72-227.
4. Hamirani YS, Dietl CA, Voyles W, Peralta M, Begay D, Raizada V. Acute aortic regurgitation. *Circulation* 2012;126:1121-6.



**Figure 6** Contrast-enhanced computed tomographic coronal image of the thoracic cavity demonstrating a grossly dilated aortic root compared with the ascending aorta. AA, Ascending aorta; AO, dilated aortic root; LV, left ventricle.

5. Maurer G. Aortic regurgitation. *Heart* 2006;92:994-1000.
6. Lavall D, Schäfers H-J, Böhm M, Laufs U. Aneurysms of the ascending aorta. *Deutsches Arzteblatt International* 2012;109:227-33.
7. Zarate YA, Sellars E, Lepard T, Tang X, Collins RT. Aortic dilation, genetic testing, and associated diagnoses. *Genet Med* 2016;18:356-63.
8. Pyeritz RE. Evaluation of the adolescent or adult with some features of Marfan syndrome. *Genet Med* 2012;14:171-7.
9. Zoghbi WA, Adams D, Bonow RO, Enriquez-Sarano M, Foster E, Grayburn PA, et al. Recommendations for noninvasive evaluation of native valvular regurgitation: a report from the American Society of Echocardiography developed in collaboration with the Society for Cardiovascular Magnetic Resonance. *J Am Soc Echocardiogr* 2017;30:303-71.
10. Evangelista A, Flachskampf FA, Erbel R, Antonini-Canterin F, Vlachopoulos C, Rocchi G, et al. Echocardiography in aortic diseases: EAE recommendations for clinical practice. *Eur J Echocardiogr* 2010;11:645-58.
11. Evangelista A, Maldonado G, Grusso D, Gutiérrez L, Granato C, Villalva N, et al. The current role of echocardiography in acute aortic syndrome. *Echo Res Pract* 2019;6:R53-63.

12. Maganti K, Rigolin VH, Sarano ME, Bonow RO. Valvular heart disease: diagnosis and management. *Mayo Clin Proc* 2010;85:483-500.
13. Borer JS, Bonow RO. Contemporary approach to aortic and mitral regurgitation. *Circulation* 2003;108:2432-8.
14. Dujardin KS, Enriquez-Sarano M, Schaff HV, Bailey KR, Seward JB, Tajik AJ. Mortality and morbidity of aortic regurgitation in clinical practice. A long-term follow-up study. *Circulation* 1999;99:1851-7.
15. Mathur A, Mohan V, Ameta D, Gaurav B, Haranahalli P. Aortic aneurysm. *J Transl Int Med* 2016;4:35-41.