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Case Report

Silent Threat: Ascending Aortic Dissection Triggering Congestive Heart Failure in A Young Indonesian Woman Suspected of Marfan Syndrome – A Case Report

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ABSTRACT

Background: A considerable number of thoracic aortic aneurysms (TAAs) remain unidentified or misdiagnosed owing to the silent nature of the disease and the lack of a screening program. The diagnostic process is frequently intricate, leading to potentially catastrophic outcomes. **Case summary:** A 32-year-old woman presented with shortness of breath and swelling in both legs, resembling symptoms of heart failure. Despite the absence of known cardiovascular risk factors, echocardiography unveiled a critical condition; painless ascending aortic aneurysm with dissection, posing a life-threatening risk. **Conclusion:** Initially, the patient's susceptibility to the disease was not readily apparent. Interestingly, aortic dissection patients presenting with heart failure symptoms were less inclined to experience chest pain compared to those without such symptoms. This disparity might triple the time required for diagnosis and elevate mortality risk to 33%. Thus, this case report aims to increase awareness in the medical community about rare and subtle cases that might be overlooked or misdiagnosed due to atypical presentations.

Highlights:

1. Thoracic Aortic Aneurysm is not frequently discussed. However, this study analyzed it and pointed out how life-threatening a disease with silent nature could be.
2. It shows how screening for patient needed to be conducted thoroughly to ensure there is no potential threat left undiagnosed.

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Introduction

Most cases of thoracic aortic aneurysms (TAAs) are found at the age of more than 65 years and are degenerative [1]. TAAs can develop without symptoms until they rupture, resulting in a mortality rate exceeding 90% [2]. The combined incidence and prevalence of TAAs were 5.3 per 100,000 individuals per year, with ruptured aneurysms having an incidence of 1.6 per 100,000 individuals per year [2]. The prevalence of asymptomatic TAAs ranges from 0.16% to 0.34% [3]. Incidentally observed dilation of the ascending aorta (4-5 cm) has been documented in 2.7% of the general population [3]. There are several risk factors for the growth and rupture of ascending TAAs. These include a family history of the disease, older age, hypertension, chronic obstructive pulmonary disease, cigarette smoking, male sex, and increased aortic diameter [1]. Marfan syndrome is among the genetic syndromes linked to TAAs and aortic dissection (AoD), alongside Loeys–Dietz syndrome, Ehlers–Danlos syndrome, arterial tortuosity syndrome, Shprintzen-Goldberg syndrome, and cutis laxa [4]. In East Asia, Marfan syndrome is perceived as a rare condition; however, researchers suggest that this perception may be due to substantial underestimation and subsequent underdiagnosis of the disease in the region.[5]

Managing patients with inconspicuous and contrasting risk factors poses a significant challenge, particularly in developing countries with limited awareness and resources. This predicament can result in misdiagnosis and delayed treatment, contributing to heightened mortality rates. This case report details the clinical scenario of a young Indonesian woman with a painless ascending aortic dissection, exhibiting symptoms of heart failure and suspected, yet under-diagnosed, Marfan syndrome.

Case Presentation

A 32-year-old woman presented to the emergency room following a referral from another hospital, diagnosed with congestive heart failure and suspected aortic dissection. The suspicion arose after an echocardiography revealed a dilated aortic root with flaps extending from the left coronary cusp to the arcus, accompanied by severe aortic regurgitation. The patient's chief complaint was increasing shortness of breath, particularly intensified over the past 5 days prior to her visit to the initial hospital. Additionally, she reported experiencing swelling in both legs for the last 2 months, progressively worsening. Although she denied chest pain, she acknowledged occasional discomfort in her chest over the past month. The patient had no history of previous medical illnesses, and there were no similar complaints or connective tissue disorders among her family members.

Upon examination, vital signs were stable but there were audible diastolic murmurs in aorta and crackles at the lung bases bilaterally. There was also an increase in jugular vein pressure and bilateral lower extremity edema. ECG showed sinus rhythm with left atrium enlargement, left ventricular hypertrophy, and T inversion in leads II, III, and aVF (Figure 1). Laboratory results showed a very high D-dimer value (9690) and elevated alanine and aspartate aminotransferase.

The chest x-ray, as depicted in Figure 2, demonstrated conclusive cardiomegaly and suggested thoracic scoliosis. Clinically, the patient presented with tall stature, long limbs, and thin fingers. Although the thumb sign was negative, the wrist sign was positive. The patient's height was measured at 170 cm, with a weight of 61 kg. Notably, she wore glasses with a prescription of -5.00 D and reported experiencing blurry vision since elementary school. Unfortunately, no further diagnostic examinations were conducted to confirm the suspicion of Marfan's syndrome.

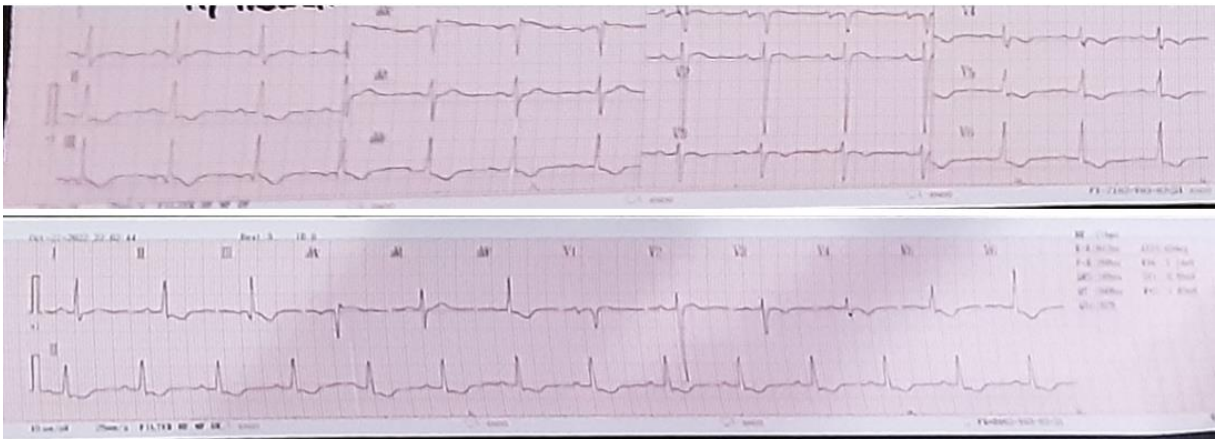

Figure 1. The patient's initial 12-lead electrocardiogram.

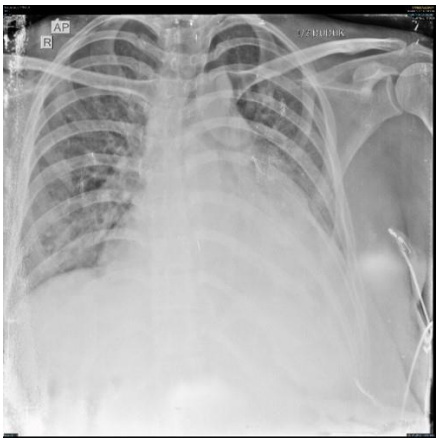

Figure 2. Chest x-ray showing cardiomegaly and thoracal scoliosis.

The patient underwent collaborative treatment with an internist, and the assessment indicated hepatic venous congestion with suspected cardiac cirrhosis. During the treatment period, episodes of bigeminy premature ventricular contractions were detected on the ECG on the 2nd (Figure 3) and 4th days. Subsequent echocardiography, as illustrated in Figure 4-5, and initial abdominal ultrasound and CT angiography, shown in Figure 6, were conducted.

The echocardiography results revealed an ascending aortic aneurysm with dissection, severe aortic regurgitation due to aortic dilation, mild mitral regurgitation, mild tricuspid regurgitation, normal left ventricular systolic function with an ejection fraction of 64%, good right ventricular contractility, and a high likelihood of pulmonary hypertension. The CT angiography findings suggested a fusiform aneurysm originating from the aortic valve, approximately 8.02 cm in length and 5.72 cm in diameter, with dissection occurring near the aortic valve.

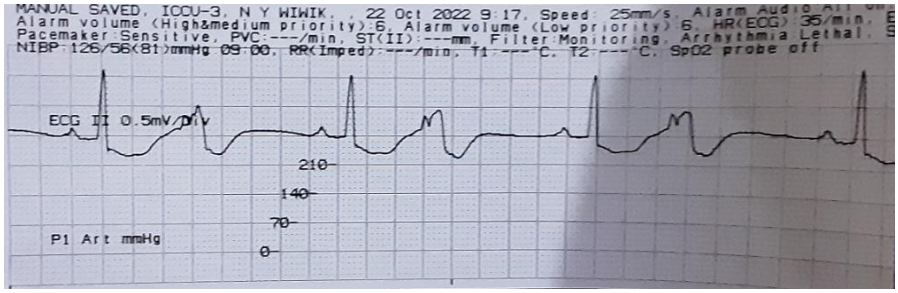

Figure 3. Lead II of the electrocardiogram taken on day 2 reveals bigeminy premature ventricular contractions.

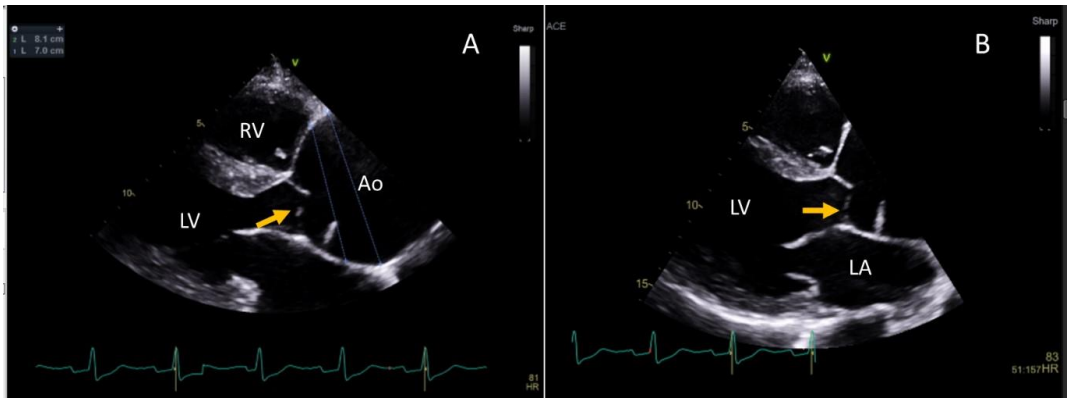

Figure 4. A transthoracic echocardiogram, parasternal long-axis view.

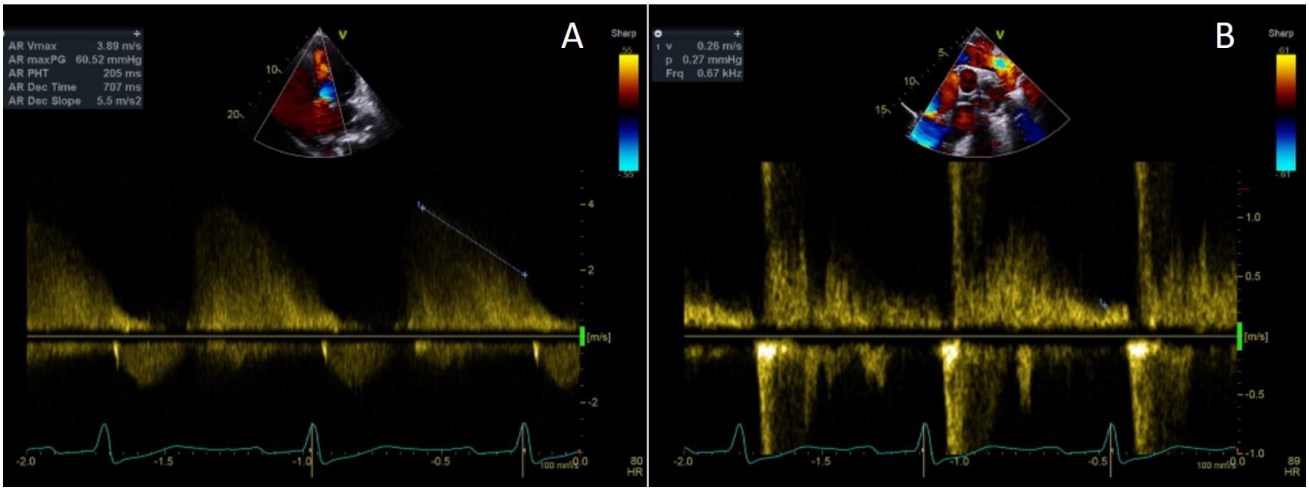

Figure 5. Echocardiogram showing severe aortic regurgitation.

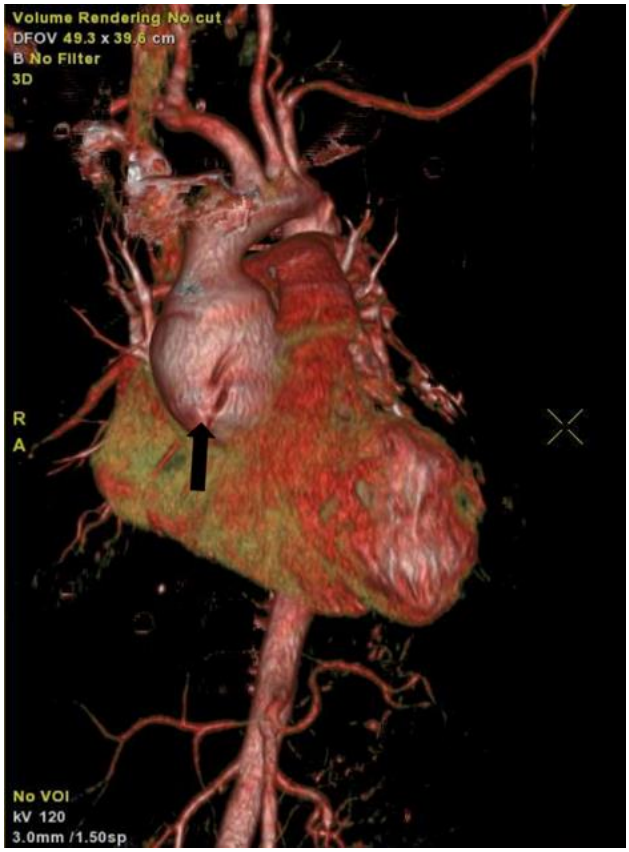

Figure 6. A three-dimensional computed tomography angiography

Throughout the treatment course, the patient was administered a regimen that included bisoprolol (5 mg), atorvastatin, diuretics (furosemide and spironolactone), and isosorbide dinitrate. On the 12th day of treatment, the patient encountered cardiogenic shock, marked by a decline in blood pressure to 93/60 mmHg. To address this critical condition, dobutamine support (3-10 mcg/kg/minute) and norepinephrine (50-200) via a syringe pump were employed for a duration of 6 days.

Following stabilization, the patient was transferred to a larger hospital equipped to perform the Bentall procedure. Unfortunately, the patient succumbed to her condition before the scheduled surgery could take place.

Discussion

Aortic aneurysm and its related complications are prominent contributors to the top 10 causes of global cardiovascular deaths, significantly impacting both health outcomes and resulting in substantial health system costs [6]. People with Marfan syndrome are at up to 250 times greater risk of aortic dissection than the general population [7]. Over the past two decades, genetic studies have revealed that both non-syndromic familial thoracic aortic aneurysm and dissection primarily manifest as single-gene hereditary disorders inherited in an autosomal dominant manner [4]. Syndromic thoracic aortic aneurysm and dissection conditions arise from pathogenic variants in genes associated with extracellular matrix dysfunction and TGF-β signaling.[4]

The International Registry of Acute Aortic Dissection (IRAD) collected 6,424 consecutive patients with acute aortic dissection from January 1996 to May 2017, of which 258 (4%) had Marfan syndrome [8]. It was found that dissection in patients with Marfan syndrome occurs at a significantly younger age than those without (37 vs. 62 years for type A; 40 vs. 64 years for type B). They also found that only about 35% of patients with aortic dissection were female.

According to the 2010 Ghent revisited criteria, the diagnosis of Marfan syndrome is considered possible in the presence of the following combinations: (A) aortic root dilatation/dissection + ectopia lentis; (B) aortic root dilatation/dissection + FBN1 mutation; (C) ectopia lentis + FBN1 mutation, known to be previously associated with aortic root dilatation in the literature or present in the family [9]. Skeletal features, myopia, skin striae, mitral valve prolapse, spontaneous pneumothorax, and dural ectasia are collectively evaluated in the "systemic score" [9]. Each item receives a score, and if the cumulative score reaches seven or more, the systemic score becomes relevant for diagnosis [9]. Referring to the criteria, our patient exhibits an Aortic Z score of 15.46 (Z>2), indicating significant dilation. However, the patient only meets 3 out of the minimum required 7 points for the systemic criteria. Therefore, the current findings are insufficient to establish a diagnosis of Marfan syndrome, although the possibility cannot be ruled out entirely. It has been observed that dissection in patients with Marfan syndrome typically occurs at a significantly younger age compared to those without the condition.[10]

The aorta dissection in our patient was chronic and presented as congestive heart failure (CHF), which is a highly atypical feature that may triple the time to diagnosis and increase mortality risk to 33% [11]. Based on the IRAD study, only 6% out of 1069 subjects presented with acute CHF at the time of their AoD. The median times from symptom onset to presentation, diagnosis, or surgical intervention were all consistently longer among patients with CHF. The potential impact of CHF on AoD recognition delay was noted and CHF was an independent predictor of surgical delay [12]. As in our case, patients with CHF at presentation were less likely to have chest pain than those without. Furthermore, when pain was present in patients with CHF, it was often mild and less likely abrupt in onset. Patients with CHF and aortic dissection were less likely to be hypertensive on presentation than those without CHF, but more likely to present in shock.[12]

CHF is usually caused by aortic regurgitation due to aortic valve disease, incomplete closure of the aortic leaflet, or aortic valve disruption. Rupture of an AoD into the right atrium, right ventricle, or main pulmonary artery can lead to a left-to-right shunt and CHF. An additional mechanism of CHF at the time of AoD includes high-output heart failure.[12]

Our patient is also experiencing liver congestion. The fundamental pathophysiology of liver dysfunction involves passive congestion resulting from increased filling pressure or decreased cardiac output, leading to impaired perfusion. Elevated central venous pressure contributes to both direct and indirect elevations in liver enzymes [13]. The d-dimer was elevated remarkably high (19x of normal value). D-dimer levels exhibit high sensitivity but only fair specificity in diagnosing AoD, making them most valuable for ruling out AoD in patients with a low likelihood of the disease [10]. This is particularly useful in cases with atypical presentations, such as our patient.

One of medical therapy given to our patient was bisoprolol. Beta blockers are the initial drug of choice in aortic dissection for lowering blood pressure, ventricular rate, dP/dt, and stress on the aorta [14]. By decreasing left ventricular dP/dt and decreasing shear stress, beta blockers decrease the rate of aortic dilatation.[14]

Subsequently, she was scheduled to undergo a Bentall procedure, a cardiac surgery technique that entails composite graft replacement of the aortic valve, aortic root, and ascending aorta, alongside re-implantation of the coronary arteries into the graft [10]. The major cardiovascular society guidelines from the American College of Cardiology, American Heart Association, European Cardiovascular Society, and Society of Vascular Surgery concur on recommending repair for all symptomatic TAAs, including those that are ruptured, associated with dissection, or causing pain [10,15–17]. Indications for ascending aorta replacement are influenced by the

etiology, diameter, and rate of growth of the aneurysm [10,15,16]. Replacement is recommended for asymptomatic ascending aortic aneurysms with a diameter greater than 5.00 cm, and in Marfan syndrome, the threshold is set at >4.50 cm [10,15,16]. Ideally, our patient should have undergone aortic root replacement at the earliest possible time.

However, the exact underlying etiology responsible for the development of our patient's condition is still undetermined. Unperformed important supporting examinations such as molecular genetic testing and complete skeletal imaging hinder definitive diagnosis. In patients with such remarkable medical history, the presence of an undetected genetic disorder is the greatest possibility that should be suspected. It is better for the patient's family to be screened for the possibility of having the same aneurysm.

Conclusion

The survival rate among patients with aortic dissection hinges on prompt diagnosis and the timely implementation of effective treatment. A stringent and regular life-long follow-up is imperative to identify ongoing dilatation of the ascending aorta and address it preemptively to prevent dissection, thereby fostering a more favorable prognosis. This case report aims to heighten awareness within the medical community regarding the rare and subtle cases that may go unnoticed or be misdiagnosed due to atypical presentations. Future studies should prioritize the development of reliable and feasible screening methods to detect TAAs in patients with Marfan syndrome, particularly in low-income and developing countries.

Acknowledgement

None.

Conflict of Interest

All authors declare that there is no conflict of interest.

Informed Consent

Verbal informed consent was obtained from the patient’s family for publication of this case report.

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