

Rheumatic Heart Disease

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Abstract

Rheumatic heart disease is an important and preventable cause of mortality and morbidity worldwide, particularly in school-aged children and young adults. It can lead to potentially life-threatening sequelae, including heart failure, endocarditis, atrial fibrillation, and embolic stroke. Permanent valvular damage can require surgical repair or replacement. Rheumatic heart disease is largely preventable with antibiotic prophylaxis. Two-dimensional echocardiography remains the gold standard for evaluation, though cardiac MRI and cardiac CT can provide useful diagnostic information.

Keywords: Cardiac, Infection, Autoimmune

Case Summary

A tweenaged girl presented with sudden onset of purposeless movements of the limbs and face described as “jerking” by the family. Her physical exam was notable for a systemic blood pressure of 100/45 mm Hg and a II/IV diastolic decrescendo murmur at the right upper sternal border.

Imaging Findings

Transthoracic echocardiogram (Figure 1) showed focal thickening with restricted motion of the right aortic valve leaflet and associated severe aortic valve insufficiency. The anterior leaflet of the mitral valve is thickened with restricted tip motion during diastole and associated mild mitral valve insufficiency.

Diagnosis

Rheumatic heart disease.

The differential diagnosis includes nonrheumatic aortic or mitral valve insufficiency such as functional mitral valve insufficiency associated with viral myocarditis, bacterial endocarditis, and mitral valve prolapse due to degenerative myxomatous disease.¹

Discussion

Rheumatic heart disease is estimated to affect up to 40 million people worldwide and contributes to half a million deaths per year.² During an episode of acute rheumatic fever (ARF), rheumatic carditis may present as valvulitis or pericarditis. About 70% of these patients may progress to rheumatic heart disease. It is the cardiac sequelae of ARF, an autoimmune response that can occur 2-3 weeks following untreated *Streptococcus pyogenes* pharyngitis.³ Clinical manifestations of ARF can include carditis, arthritis/arthralgia, Sydenham chorea, erythema marginatum, and subcutaneous

nodules. Secondary findings include fever, elevated inflammatory markers (eg, C-reactive protein), and a prolonged PR interval.

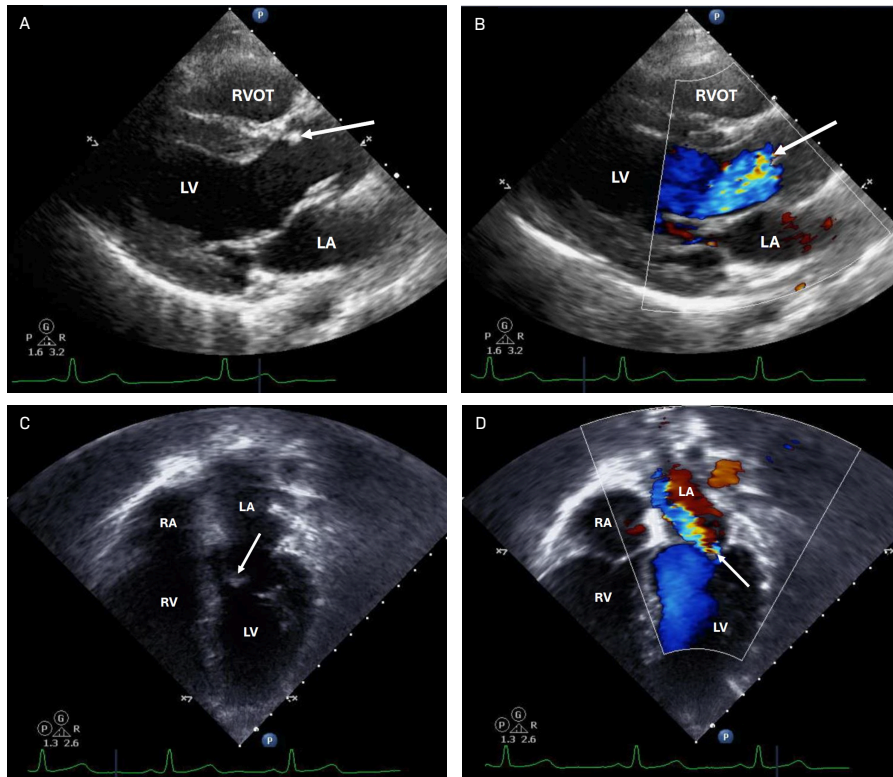
Rheumatic heart disease is the most common cardiovascular disease worldwide in those under 25 years of age.⁴ It is an important cause of morbidity and mortality, particularly in school-aged children and young adults, and those in resource-poor areas globally.⁵ While it is largely preventable with antibiotic (penicillin) prophylaxis, rheumatic heart disease can lead to complications, including heart failure, endocarditis, embolic stroke, and atrial fibrillation.⁵ The pathogenesis of ARF and rheumatic heart disease is complex and not fully understood but involves both host and bacterial factors, molecular mimicry, and aberrant innate and adaptive immune responses from the host that result in the loss of self-tolerance and subsequent cross-reaction with host tissues.⁶

Auscultation is often used to screen for rheumatic heart disease. However, this exam method is neither sensitive nor specific. Studies have shown that echocardiography is 5- to 50-fold more sensitive than auscultation.⁷ Because of its improved sensitivity, two-dimensional (2D) echocardiography is the gold-standard diagnostic test used to diagnose rheumatic heart disease. While making

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Figure 1. (A) Two-dimensional transthoracic echocardiogram parasternal long-axis view at peak systole. There is thickening and restricted motion of the right cusp of the aortic valve (arrow). LA, left atrium; LV, left ventricle; RVOT, right ventricular outflow tract. (B) Color Doppler transthoracic echocardiogram parasternal long-axis view at end diastole. There is severe aortic valve insufficiency present (arrow). LA, left atrium; LV, left ventricle; RVOT, right ventricular outflow tract. (C) Two-dimensional transthoracic echocardiogram 4-chamber view at end diastole. The anterior leaflet of the mitral valve is thickened with restricted tip motion creating the appearance of a “hockey stick” (arrow). LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle. (D) Color Doppler transthoracic echocardiogram 4-chamber view at early systole. There is mild mitral valve insufficiency (arrow). LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle.



the diagnosis, 2D echocardiography can identify and measure the severity of acute valvular disease, assess cardiac function, monitor disease progression, and determine the suitability and plan for intervention. Color Doppler is an essential component of echocardiographic assessment of rheumatic heart disease used to identify and quantify pathological flow regurgitation.²

Key findings on echocardiogram in patients with rheumatic heart disease include morphological and pathological abnormalities of the mitral or, less commonly, aortic valves. Mitral regurgitation is the most common abnormality in patients with rheumatic heart disease. When the regurgitation is severe, the left atrium can be

enlarged. Mitral stenosis may develop due to fibrosis and valvular thickening secondary to chronic inflammation. Morphologic abnormalities of the mitral valve may also be present. The mitral valve can appear thickened with doming and restricted mobility, producing a characteristic “elbow” or “dog leg” deformity of the anterior mitral valve leaflet. Aortic stenosis or regurgitation may also be present.

Cardiac MRI can assess the impact of valvular disease on the myocardium and predict postsurgical outcomes. Cardiac MRI can be utilized to determine the etiology of left ventricular dysfunction, allow detailed assessment of regurgitant valves (including the volume of regurgitation), and estimate left ventricle

myocardial fibrosis. An advantage of MRI over echocardiography is its ability to calculate mitral regurgitation severity regardless of jet shape or direction.² Cardiac MRI can demonstrate dilated cardiac chambers, myocardial inflammation, pericardial inflammation, and pericardial effusion.⁸

Cardiac CT can provide additional information on the integrity and mobility of valves and aid in the identification of paravalvular pathology. CT also facilitates a detailed assessment of valve thickness, calcification, and annular fibrosis, in addition to information on the subvalvular structures and left ventricle.^{2,6,8} CT findings include valvular or pericardial calcification and dilated ventricles and atria.

Screening for ARF is nonspecific based on clinical presentation since fever is diagnostic or specific, and a sore throat is not present in about 1/3 of patients. Auscultation is not specific or sensitive, and as a result, it makes it difficult to support echocardiography screening. As a result, early diagnosis is challenging.

The key to management is secondary prevention using continuous antibiotic prophylaxis to prevent recurrent group A *Streptococcus* infection using benzathine penicillin intramuscularly every 4 weeks (superior to oral penicillin). The duration of prophylactic treatment varies; however, it is often 5-10 years or age 21, whichever is longer. Patients with severe chronic rheumatic fever prophylaxis may be lifelong. Patients with severe valvular heart disease can be treated with catheter-based therapies or surgery.⁹

Outcomes for children with rheumatic heart disease are better in the United States than in developing countries. However, there is still significant risk of disease progression, heart failure, and need for catheter-based therapy or surgery in children who present with moderate-to-severe heart disease.⁹

Conclusion

Rheumatic heart disease is an important and preventable cause of mortality

and morbidity worldwide, particularly in school-aged children and young adults. It can lead to potentially life-threatening sequelae, including heart failure, endocarditis, atrial fibrillation, and embolic stroke. Permanent valvular damage can require surgical repair or replacement. Rheumatic heart disease is largely preventable with antibiotic prophylaxis. 2D echocardiography remains the gold standard for evaluation, though cardiac MRI and cardiac CT can provide useful diagnostic information.

References

- 1) Dass C, Kanmanthareddy A. Rheumatic heart disease. In: *StatPearls* [Internet]. StatPearls Publishing. 2023.
- 2) Seidler S, Zuhair M, Shamsi A, et al. Cardiac imaging in rheumatic heart disease and future developments. *Eur Heart J Open*. 2023;3(2):oeac060. doi:10.1093/ehjopen/oeac060
- 3) Liang Y, Yu D, Lu Q, Zheng Y, Yang Y. The rise and fall of acute rheumatic fever and rheumatic heart disease: a mini review. *Front Cardiovasc Med*. 2023;10:1183606. doi:10.3389/fcvm.2023.1183606
- 4) Remenyi B, Carapetis J, Wyber R, et al. Position statement of the World Heart Federation on the prevention and control of rheumatic heart disease. *Nat Rev Cardiol*. 2013;10(5):284-292. doi:10.1038/nrcardio.2013.34
- 5) Carapetis JR, Beaton A, Cunningham MW, et al. Acute rheumatic fever and rheumatic heart disease. *Nat Rev Dis Primers*. 2016;2(1):15084. doi:10.1038/nrdp.2015.84
- 6) Dooley LM, Ahmad TB, Pandey M, Good MF, Kotiw M. Rheumatic heart disease: a review of the current status of global research activity. *Autoimmun Rev*. 2021;20(2):102740. doi:10.1016/j.autrev.2020.102740
- 7) Dougherty S, Khorsandi M, Herbst P. Rheumatic heart disease screening: current concepts and challenges. *Ann Pediatr Cardiol*. 2017;10(1):39-49. doi:10.4103/0974-2069.197051
- 8) Weerakkody Y, Knipe H, Al Khateeb A, et al. Rheumatic heart disease. Accessed November 17, 2023. <https://doi.org/10.53347/rID-7503>
- 9) Kumar RK, Antunes MJ, Beaton A, et al. Contemporary diagnosis and management of rheumatic heart disease: implications for closing the gap: a scientific statement from the American Heart Association. *Circulation*. 2020;142(20):e337-e357. doi:10.1161/CIR.0000000000000921