Case Report

Rare case of hereditary spherocytosis with rheumatic mitral stenosis

Alok Mathur[1,2], J B Gupta[1,4]*, Mohammad Izhar Joad[1], Veda Shree Ravte[3], Priyansha Jain[1], Ajit Singh Brar[1]

From [1]Department of Internal Medicine, [2]Department of Preventive Cardiology, [3]Department of General Medicine, [4]Department of Clinical Research, Eternal Hospital, Jaipur, Rajasthan, India

ABSTRACT

A rare case of hereditary spherocytosis (HS) and rheumatic mitral stenosis coexisting in a patient having severe stenosis, atrial fibrillation, and symptoms of the left ventricular dysfunction, along with hemolytic anemia attributed to HS. We present the case of a 58-year-old lady who presented to the emergency department with complaints of increasing shortness of breath for the past week. She was examined to have atrial fibrillation with a fast ventricular rate. On investigations, she was found to have severe rheumatic mitral stenosis with evidence of hemolytic anemia. Further, evaluation of the cause of her anemia revealed HS. This case highlights the importance of the evaluation of anemia in patients with valvular heart diseases. If a treatable cause is found, anemia can be treated to reduce the cardiac burden.

Key words: Hemolytic anemia, Hereditary spherocytosis, Rheumatic heart disease, *Streptococcus pyogenes*, Valvular heart disease

Hereditary spherocytosis (HS) is an autosomal dominant inherited extravascular hemolytic disorder. The classical clinical features are anemia, jaundice, and splenomegaly. However, there is marked heterogeneity in the clinical presentation with some cases having asymptomatic disease while others having fulminant hemolytic anemia [1]. The disease may be diagnosed early in life if severe or may go unnoticed until later in adult life if the hemolysis is mild. Rheumatic fever, despite its declining prevalence, is a significant contributor to mitral stenosis. Women are more likely to have rheumatic fever. It occurs as a result of an autoimmune response that follows a Group A *Streptococcus* (GAS) *pharyngeal* infection. In the heart, the mitral valve is the most affected structure, with leaflet thickening, the fusion of the commissures, and the shortening of the chordae [2,3].

We present a case, where both the entities, HS, and rheumatic mitral stenosis concomitantly exist. This case highlights the importance of the evaluation of anemia in patients with valvular heart diseases. If a treatable cause is found, anemia can be treated to reduce the cardiac burden.

CASE REPORT

A 58-year-old lady was evaluated in the emergency department for progressively increasing shortness of breath and generalized weakness for the past week. She felt short of breath on mobilizing from one room to the other in the house and was able to perform some household activities. Her dyspnea fell in Class 3 of the New York heart association classification. The patient history revealed that she is being treated for hypothyroidism. She has had multiple sick episodes during her childhood. She has a history of caesarian delivery of her daughter. She denies any tobacco, alcohol, or substance abuse.

The patient was afebrile and demonstrated atrial fibrillation with a fast ventricular rate of 120 beats per minute on her electrocardiogram. Lung auscultation revealed bilateral crackles at the lung bases with no rales or wheezing. Cardiac examination revealed an irregularly irregular pulse with a mid-diastolic rumbling murmur heard best at the cardiac apex with no radiation. She demonstrated elevated jugular venous pressure of 10 cm and bilateral pitting edema of the lower extremities.

2D echocardiogram performed for this patient showed enlargement of the right and left atrium, rheumatic heart disease with severe mitral stenosis (mitral valve area of 1.03 cm^2), moderate to severe mitral regurgitation, severe tricuspid regurgitation, pulmonary arterial hypertension, and normal left ventricular function (Fig. 1). The patient was also found to have anemia (Table 1). She also had an elevated indirect bilirubin level (5.60). Iron studies were normal. Her serum lactate dehydrogenase was elevated with negative direct and indirect coombs tests. Her abdominal ultrasound revealed moderate splenomegaly with the presence of multiple cholelithiasis.

Access this article online	
Received - 15 September 2022 Initial Review - 30 September 2022 Accepted - 22 December 2022	Quick Response code
DOI: 10.32677/ijcr.v8i12.3628	

Correspondence to: Dr. J B Gupta, Senior Consultant, Department of Internal Medicine and Director, Department of Clinical Research, Eternal Hospital (EHCC) and Research Institute, 3A, Jagatpura Road, Near Jawahar Circle, Jaipur - 302020, Rajasthan, India/Diabetes Care Centre, Jaipur, Rajasthan, India. E-mail: drjbgupta@gmail.com.

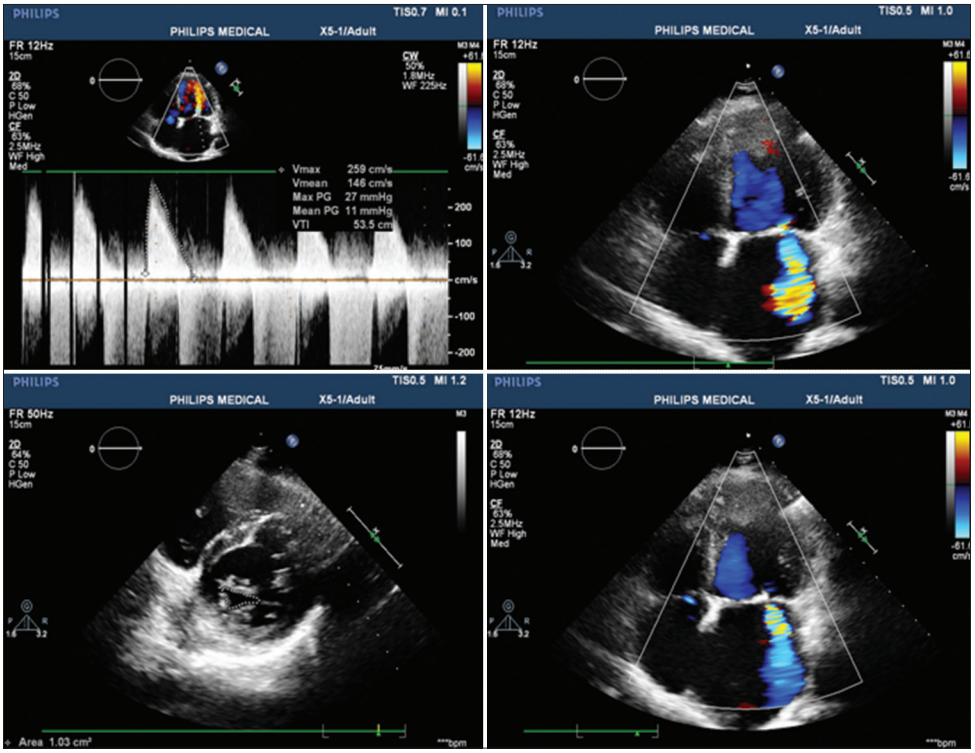

Figure 1: 2D ECHO shows mitral valve area of 1.03 cm², compatible with severe mitral stenosis

Osmotic fragility showed increased lysis of red blood cells (RBCs) in isotonic saline solution. Peripheral blood film was suggestive of reduced red cell mass and anisopoikilocytosis with a variable number of spherocytes/microspherocytes, along with occasional normoblasts (Fig. 2).

The patient was managed with diuretics (torsemide – 10 mg 12 hourly), digoxin − 0.25 mg 24 hourly, and calcium channel blockers (diltiazem – 90 mg 12 hourly) for rate control and for anticoagulation, ecosprin – 75 mg 24 hourly.

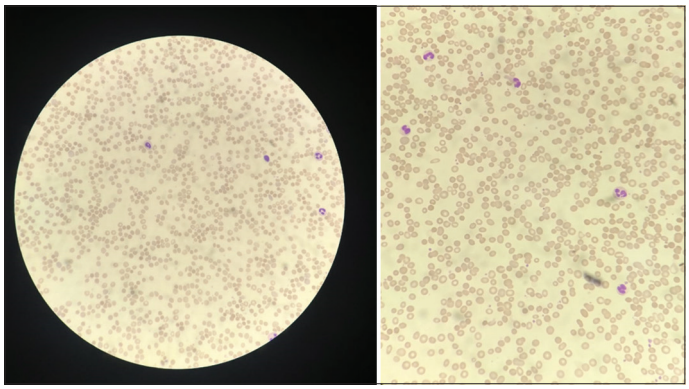

Figure 2: Peripheral blood smear (reduced red cell mass and anisopoikilocytosis with variable number of spherocytes/ microspherocytes along with occasional normoblasts)

Table 1: Laboratory parameters of the patient

S. No	Laboratory parameters	Values
1	Hemoglobin	6.8 g/dl
2	Mean corpuscular volume	81.7
3	Mean corpuscular hemoglobin	25.9
4	Mean corpuscular hemoglobin concentration	31.6
5	Indirect bilirubin level	5.60

DISCUSSION

Rheumatic fever is a delayed consequence of pharyngeal infection of GAS. It is believed to be a result of a diffuse inflammatory process that leads to an autoimmune attack on the body due to the phenomenon of molecular mimicry [4]. Antigenically, the M protein in the GAS cell wall is similar to proteins present in the human body. When antibodies are created against the M protein during the first infection, this also causes the T-cell response against normal human tissues, which results in the long-term sequel of the disease. Rheumatic carditis is the most serious manifestation of the GAS infection. The disease can affect all three layers of the heart and involvement of the valves occurs most often with the mitral valve being the commonest (85% of the patients). Aschoff bodies initially form on the valve leaflets and slowly enlarge due to increased fibrin deposition. This leads to mitral regurgitation or mitral stenosis [5].

Natural history suggests that the average time from the onset of rheumatic fever to mitral stenosis is 16.3 years. Further, progression to severe mitral stenosis-related disability from the onset of symptoms is 9.2±4.3 years. The treatment of mitral stenosis depends on the symptoms on presentation. In severe cases, percutaneous balloon valvuloplasty or mitral valve repair has shown similar results and efficacy at 3 years [6].

The most prevalent inherited disorder of the red cell membrane is called HS. While autosomal recessive inheritance is also seen, autosomal dominant inheritance is the most frequent pattern of inheritance. Ankyrin and spectrin, two proteins found in the red cell membrane, are deficient in HS patients. These are the main

proteins that make up a RBC cytoskeleton [7]. RBCs rapidly exit the bloodstream by becoming more spherical in form and having less surface area due to defective membranes.

Pallor, jaundice, and splenomegaly are the main clinical characteristics of HS. Pallor is due to anemia caused by the destruction of red cells in the spleen. Jaundice is due to hyperbilirubinemia caused by the rapid destruction of red cells in the spleen. Splenomegaly is due to sequestration and phagocytosis of spherical-shaped red cells in the spleen. These features may be noted in infancy, childhood, or later in adult age. Most patients with moderate HS are asymptomatic, have modest splenomegaly, mild reticulocytosis, and only a few spherocytes on a peripheral blood smear. About 20–30% of HS cases are mild. Many of these patients go undetected. They might be identified by family history, gallbladder stone testing, splenomegaly, or anemia evaluation. In 60–70% of cases with HS, the moderate type manifests at any age. About 3–5% of cases are severe with life-threatening anemia and need regular transfusions. Such severe HS are inherited in autosomal recessive pattern. Cholelithiasis in HS occurs due to chronic hemolysis [7]. Most of the stones are formed in patients between 10 and 30 years of age. Stones are reported to be present in 21–63% of patients with HS [1].

Clinical history, family history, physical examination, and test results can all be used to diagnose HS. Laboratory findings include hemolysis with reticulocytosis and raised bilirubin level. Laboratory findings include hemolysis with reticulocytosis and raised bilirubin level, normal mean corpuscular volume and mean corpuscular hemoglobin (MCH) values, but a possible elevated MCH concentration [8]. Anisopoikilocytosis and spherocytes are commonly seen in blood smear. Relevant tests indicating the presence of red cell membrane protein defect are required for confirmation of HS [8]. Management involves PRBC transfusions, folic acid supplementation, and splenomegaly in refractory cases.

CONCLUSION

This case highlights the importance of evaluation of anemia in patients with valvular heart diseases. If a treatable cause is found, anemia can be treated to reduce the cardiac burden.

REFERENCES

1. Rutkow IM. Twenty years of splenectomy for hereditary spherocytosis. Arch Surg 1981;116:306-8.
2. Carabello BA. Modern management of mitral stenosis. Circulation 2005;112:432-7.
3. Carapetis JR. Rheumatic heart disease in developing countries. N Engl J Med 2007;357:439-41.
4. Ferri FF. Mitral stenosis. In: Ferri's Clinical Advisor. Netherlands: Elsevier; 2021. Available from: https://www.clinicalkey.com [Last accessed on 2021 Feb 14].
5. Bonow RO. Mitral valve disease. In: Braunwald's Heart Disease: A Textbook of Cardiovascular Medicine. 11th ed. Netherlands: Elsevier; 2019. Available from: https://www.clinicalkey.com [Last accessed on 2021 Feb 14].
6. Cardoso C, Cachado P, Garcia T. Hemolytic anemia after mitral valve repair: A case report. BMC Res Notes 2013;6:165.
7. Costa C, Durão D, Peres M, Leal M. A case of rheumatic valvular heart disease and autoimmune gastritis. BMJ Case Rep 2014;2014:bcr2014207578.
8. Bajoria D, Menon T. The HLA Class II associations with rheumatic heart disease in South Indian patients: A preliminary study. J Clin Diagn Res 2013;7:302-4.

Funding: Nil; Conflicts of interest: Nil.

How to cite this article: Mathur A, Joad MI, Ravte VS, Jain P, Brar AS, Gupta JB. Rare case of hereditary spherocytosis with rheumatic mitral stenosis. Indian J Case Reports. 2022;8(12):403-405.