Case Report

A Silent Hemolysis Behind a Failing Heart: Hereditary Spherocytosis Associated With SLC4A1 Mutation Presenting with Secondary Hemosiderosis and Acute Decompensated Heart Failure

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Hereditary spherocytosis (HS) is an inherited hemolytic anemia that usually presents with anemia, jaundice, and splenomegaly, but atypical presentations like heart failure may delay diagnosis. A 67-year-old man presented with progressive breathlessness, frothy cough, bilateral leg swelling, and a history of recurrent jaundice. Examination showed pallor, icterus, tachypnea, bilateral pitting edema, and basal crepitations. Initial hemogram showed anemia with leukocytosis, macrocytosis, elevated mean corpuscular hemoglobin concentration, and mild thrombocytopenia. Electrocardiography revealed complete heart block, and echocardiography showed global hypokinesia with reduced ejection fraction. Hemolysis workup demonstrated a negative direct Coombs test, a positive osmotic fragility test, normal thyroid function, and marked hyperferritinemia with elevated transferrin saturation. Subsequent cardiac magnetic resonance imaging (MRI) showed preserved myocardial iron with recovered ventricular function, while liver iron quantification demonstrated moderate hepatic iron overload. Genetic analysis identified a homozygous SLC4A1 variant. With this, a diagnosis of HS with secondary hemosiderosis presenting as acute decompensated heart failure was made. The patient improved with supportive management and follow-up evaluation. This case highlights the value of sequential hematologic, cardiac, radiologic, and genetic assessment in adults with unexplained hemolysis.

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Introduction

Hereditary spherocytosis (HS), a type of congenital hemolytic anemia, is the most common inherited disorder caused by defects in red blood cell membrane proteins. Due to mutations involving membrane and cytoskeletal proteins responsible for maintaining erythrocyte structural integrity, the red blood cells lose their normal biconcave shape and become spherical and fragile, leading to premature destruction in the spleen.[1] The clinical manifestations vary depending on disease severity and commonly include hemolytic anemia, jaundice, and splenomegaly, with presentations ranging from severe neonatal hyperbilirubinemia requiring transfusions to mild chronic asymptomatic anemia.[2]

Chronic hemolysis can result in iron overload due to continuous release of hemoglobin-derived iron into the circulation, exceeding the body's iron-binding and storage capacity. Progressive iron deposition in vital organs leads to secondary hemosiderosis and may involve the liver, pancreas, endocrine glands, and heart.[3]

Iron overload may occur secondary to repeated blood transfusions, chronic hemolytic anemias, alcohol-related liver disease, ineffective erythropoiesis, and hereditary hemochromatosis caused by mutations in the high iron Fe gene (HFE) and non-HFE genes, including transferrin receptor 2 (TFR2), hemojuvelin (HJV), hepcidin antimicrobial peptide (HAMP), and solute carrier family 40 member 1 (SLC40A1). Accurate evaluation of iron overload is important for early diagnosis, appropriate treatment, and prevention of irreversible organ damage.[4]

Mutations in the SLC4A1 gene cause defects in the AE (anion exchanger-1) membrane protein, leading to loss of red blood cell membrane integrity, formation of spherocytes, and the development of hereditary spherocytosis through increased splenic destruction of erythrocytes.[5]

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Table 1: General examination and vital signs

Parameter	*Findings*
General condition	Conscious and oriented
Pallor	Present
Icterus	Present
Dyspnea	Present
Tachypnea	Present
Bilateral pedal edema	Present
Lymphadenopathy	Not significant
Blood pressure	160/100 mmHg
Pulse rate	54/min
SpO_2	96% on room air
Capillary refill time	< 2 seconds
Respiratory system examination	Bilateral crepitations and wheeze are present with diminished breath sounds in both upper lung fields
Cardiovascular system examination	Complete heart block with atrioventricular dissociation and ventricular escape rhythm
ECG findings	Complete heart block with atrioventricular dissociation and ventricular escape rhythm at 39 beats/minute without significant ischemic ST-segment or T-wave abnormalities
Echocardiography findings	Global left ventricular hypokinesia with severely reduced left ventricular systolic function and ejection fraction of 32%, with mild pulmonary hypertension
Central nervous system examination	Normal
Abdominal examination	Splenomegaly present
Other systemic examination	Non-significant

Decompensated heart failure (DHF) is a clinical syndrome in which structural or functional cardiac abnormalities impair the heart's ability to pump or accommodate blood within normal physiological pressure limits, resulting in significant functional limitation and requiring urgent medical intervention.[6] Chronic anemia and systemic iron overload associated with hemolytic disorders may contribute to cardiovascular complications, including arrhythmias, conduction abnormalities, and cardiac dysfunction.

In this case report, we present a 67-year-old male with hereditary spherocytosis due to an SCL4A1 mutation and secondary hemosiderosis who presented with acute decompensated heart failure, highlighting the diagnostic challenges and importance of early recognition of chronic hemolytic disorders associated with systemic iron overload.

Case Presentation

A 67-year-old male presented with complaints of breathlessness and bilateral lower limb swelling for 3 days, associated with orthopnea. The breathlessness was also associated with a cough with frothy sputum. The patient was a known case of congenital atrioventricular malformation with moderate tricuspid regurgitation and severe pulmonary hypertension, complete heart block, and had a preserved ejection fraction of 60%. There was no significant family history, personal history, or other relevant past history. There was no history of diabetes mellitus, hypertension, or chronic kidney disease. There was also no history of previous similar complaints or prior hospital admissions.

On general examination, the patient was conscious and oriented. Pallor and icterus were present. The patient was dyspneic and tachypneic. Bilateral pedal edema was present. No significant lymphadenopathy was noted. The recorded vital signs showed a blood pressure of 160/100 mmHg, pulse rate of 60/min, SpO_2 of 96% on room air, and capillary refill time of less than 2 seconds. On respiratory system examination, bilateral crepitations and wheeze were present, with diminished breath sounds in both upper lung fields. Spleenomegaly was present on abdominal examination. Electrocardiography revealed complete heart block with atrioventricular dissociation and a ventricular escape rhythm at 39 beats/minute, without significant ischemic ST-segment or T-wave abnormalities. Initial transthoracic echocardiography demonstrated global left ventricular hypokinesia with severely reduced left ventricular systolic function and an ejection fraction of 32%, along with mild pulmonary hypertension, consistent with acute decompensated heart failure (Table 1).

Initial laboratory investigations showed leukocytosis with a white blood cell count of 22.2 × 10^3/μL, anemia with hemoglobin of 5 g/dL and red blood cell count of 2.96 × 10^6/μL, macrocytosis with mean corpuscular volume (MCV) of 101.4 fL, and mild thrombocytopenia with platelet count of 129 × 10^3/μL. Differential leukocyte count showed neutrophilic leukocytosis. In view of severe anemia with hemoglobin of 5 g/dL, peripheral smear examination was performed, which showed spherocytic red blood cells. Vitamin B12 and folate levels were within normal limits. Reticulocyte analysis showed

Table 2: Baseline laboratory investigations of the patient at presentation

Investigation	*Findings*	*Reference range*
Hb	5 g/dL	13–17 g/dL
TLC	22.2 × 10^3/μL	4–11 × 10^3/μL
RBC count	2.96 × 10^6/μL	4.5–5.9 × 10^6/μL
Platelet count	129 × 10^3/μL	150–450 × 10^3/μL
MCV	101.4 fL	80–100 fL
Differential count	Neutrophilic leukocytosis	Neutrophils 40–75%
Peripheral smear	Predominantly normocytic normochromic RBCs without significant anisopoikilocytosis	Normal morphology
Direct antiglobulin test	Negative	Negative
Osmotic fragility test	Positive	Negative

a markedly elevated absolute reticulocyte count of 3.49 × 10^5/μL, with a corrected reticulocyte count of 7.9% and a reticulocyte production index (RPI) of 4, indicating a hyperproliferative type of anemia with increased red blood cell production. To rule out autoimmune causes of hemolysis, a direct Coombs test was performed and was negative. Further evaluation with osmotic fragility testing was positive, supporting the diagnosis of hereditary spherocytosis (Table 2).

Table 3: Liver function tests and renal function tests of the patient during evaluation

Investigation	*Findings*	*Reference range*
Total bilirubin	5.73 mg/dL	0.2–1.2 mg/dL
Direct bilirubin	1.10 mg/dL	0–0.3 mg/dL
AST/SGOT	102 U/L	5–40 U/L
ALT/SGPT	173 U/L	7–56 U/L
ALP	110 U/L	44–147 U/L
Total protein	7.2 g/dL	6–8.3 g/dL
Albumin	4.1 g/dL	3.5–5 g/dL
Blood urea	53 mg/dL	15–45 mg/dL
Serum creatinine	1.12 mg/dL	0.7–1.3 mg/dL
Urine analysis	Albumin positive, ketone bodies absent	Albumin negative, ketones absent

To assess the extent of hemolysis and end-organ involvement, liver and renal function tests were performed. Liver function tests (LFT) showed hyperbilirubinemia with total bilirubin of 5.73 mg/dL and direct bilirubin of 1.10 mg/dL, along with elevated transaminases including aspartate aminotransferase (AST/SGOT) 102 U/L and alanine aminotransferase (ALT/SGPT) 173 U/L. Alkaline phosphatase (ALP) was within normal limits at 110 U/L. Serum total protein and serum albumin were within normal limits at 7.2 and 4.1 g/dL, respectively. Renal function tests (RFT) showed blood urea 53 mg/dL and serum creatinine 1.12 mg/dL with mild electrolyte abnormalities, while urine analysis showed albumin positivity with absent ketone bodies (Table 3).

In view of leukocytosis, blood and urine cultures were performed to look for an infective etiology. Blood cultures showed no growth, whereas urine culture grew multidrug-resistant *Escherichia coli*. Viral markers, including hepatitis B surface antigen (HBsAg), hepatitis C virus (HCV), human immunodeficiency virus (HIV), Epstein-Barr virus (EBV), herpes simplex virus (HSV), and hepatitis A virus/hepatitis E virus (HAV/HEV) serology, were negative.

As the patient had chronic hemolysis and a history of multiple blood transfusions, iron studies were performed to evaluate for iron overload. Iron studies showed serum iron of 115 μg/dL, total iron-binding capacity (TIBC) of 199 μg/dL, transferrin saturation of 58%, ferritin of 12,858.3 ng/mL, and unsaturated iron-binding capacity

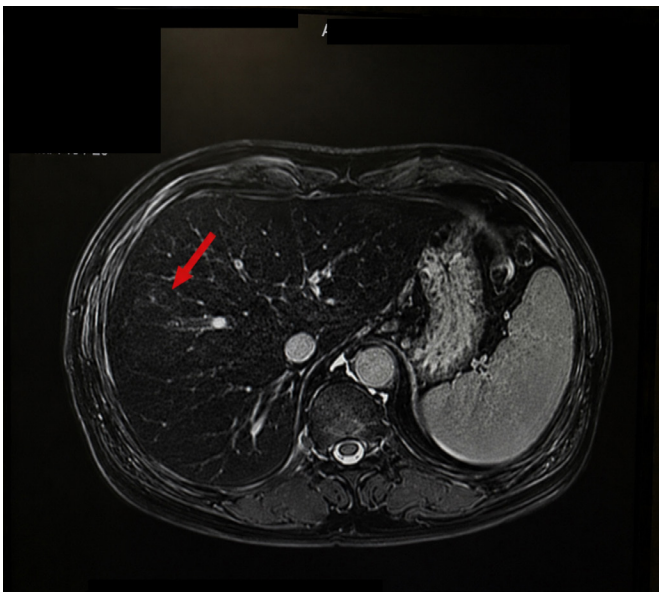

Figure 1: Axial T2-weighted HASTE magnetic resonance image showing diffuse low signal intensity of the liver parenchyma (red arrow), suggestive of hepatic iron deposition

Table 4: Iron studies of the patient, including serum iron, ferritin, total iron-binding capacity, unsaturated iron-binding capacity, and transferrin saturation

Investigation	*Findings*	*Reference range*
Serum iron	115 µg/dL	60–170 µg/dL
TIBC	199 µg/dL	240–450 µg/dL
Transferrin saturation	58%	20–50%
Ferritin	12,858.3 ng/mL	30–400 ng/mL
UIBC	84 µg/dL	111–343 µg/dL
FT3	1.7 pmol/L	3.1–6.8 pmol/L
FT4	1.60 ng/dL	0.8–1.8 ng/dL
TSH	2.300 µIU/mL	0.4–4.0 µIU/mL

(UIBC) of 84 µg/dL, suggestive of secondary iron overload (Table 4).

Legend

TIBC - Total Iron-Binding Capacity, UIBC - Unsaturated Iron-Binding Capacity, FT3 - Free Triiodothyronine, FT4 - Free Thyroxine, TSH - Thyroid-Stimulating Hormone.

As iron studies suggested iron overload with possible iron deposition in organs, MRI with iron quantification was performed. Cardiac MRI showed no significant myocardial iron overload, with myocardial T2 of 45.1 ms and R2 of 22.1, along with recovery of left ventricular systolic function with ejection fraction improving to 62.8%. Liver iron quantification MRI showed diffuse T2 hypointensity of the liver with moderate hepatic iron overload, with liver T2 of 4.3 ms and R2 of 230.9 (Figure 1).

In view of chronic hemolysis with a negative direct Coombs test and positive osmotic fragility test suggestive of a hereditary red blood cell membrane disorder, genetic analysis was performed using phenotype-based sequencing, which identified a homozygous missense variant in the SLC40A1 gene (NM_000342.4), exon 16/exon 20, c.1984T>C, resulting in p.Trp662Arg substitution. The variant was classified as a variant of uncertain significance (VUS) associated with SLC40A1 gene-related disorder (OMIM #109270), with autosomal dominant and autosomal recessive inheritance patterns reported. Correlating the chronic hemolytic picture, negative direct Coombs test, positive osmotic fragility test, iron overload, and genetic findings, a diagnosis of hereditary spherocytosis with secondary hemosiderosis presenting as acute decompensated heart failure was made.

The patient was initially managed for acute decompensated heart failure with complete heart block and acute pulmonary edema. Supportive respiratory care included CPAP/BiPAP support, head-end elevation, strict fluid restriction ranging from 800 mL to 1.5 L/day, and salt restriction of less than 5 g/day. Intravenous furosemide (Lasix) 40 mg twice daily was administered for diuresis and pulmonary congestion, later transitioned to oral torsemide once daily and spironolactone 25 mg once daily as guideline-directed heart failure therapy. He also received intravenous noradrenaline infusion at 1 to 1.5 µg/kg/min and dobutamine infusion at 2.5 µg/kg/min for cardiogenic shock and hypotension, following which blood pressure gradually improved and inotropic support was tapered. Antiplatelet and cardioprotective medications included aspirin 150 mg once daily and atorvastatin 40 mg at bedtime. Enalapril 2.5 mg

Table 5: Differences between hereditary hemochromatosis and hereditary spherocytosis

Feature	*Hereditary hemochromatosis (HH)*	*Hereditary spherocytosis (HS)*
Primary problem	Excess intestinal iron absorption leading to systemic iron overload	Defects in red blood cell membrane proteins causing fragile erythrocytes
Pathophysiology	Impaired regulation of iron metabolism results in iron deposition in organs such as the liver, pancreas, heart, and joints	Abnormal spherical red blood cells are prematurely destroyed in the spleen, causing hemolytic anemia
Main clinical features	Fatigue, abdominal pain, joint pain, diabetes mellitus, skin pigmentation, and organ dysfunction	Anemia, jaundice, splenomegaly, and pigment gallstones
Key laboratory findings	Increased serum ferritin and transferrin saturation	Low hemoglobin, reticulocytosis, increased mean corpuscular hemoglobin concentration (MCHC), and spherocytes on peripheral smear
Diagnostic methods	HFE gene mutation analysis, iron studies, and MRI for iron quantification	Peripheral blood smear, osmotic fragility test, eosin-5-maleimide (EMA) binding test, and family history
Main treatment	Therapeutic phlebotomy and iron chelation therapy in selected cases	Folic acid supplementation, blood transfusion if required, and splenectomy in severe cases

once daily was started for left ventricular dysfunction after hemodynamic stabilization. N-acetylcysteine (NAC) 600 mg twice daily and ursodeoxycholic acid 300 mg twice daily were added in view of hepatic dysfunction and hyperbilirubinemia. Nebulized salbutamol was administered for symptomatic respiratory support. The patient was also treated with deferasirox 500 mg orally once daily for 1-week as iron chelation therapy for secondary iron overload. Broad-spectrum antimicrobial therapy included intravenous piperacillin-tazobactam 4.5 g and azithromycin 500 mg for suspected infective etiology during the initial phase of hospitalization. Serial clinical monitoring showed gradual improvement in dyspnea, oxygenation, blood pressure, urine output, and biochemical parameters.

During the initial hospitalization, he developed a hemolytic crisis with a hemoglobin level of approximately 5 g/dL, which likely aggravated the underlying heart failure. Management in the later stages was primarily directed toward refractory heart failure, which was further complicated by complete heart block. Although cardiac hemosiderosis was considered a possible contributing factor, definitive attribution could not be established, as coronary artery disease could not be excluded due to the inability to perform coronary angiography because of the patient's poor general condition.

On follow-up, repeat iron studies showed serum iron of 51 μg/dL, total iron binding capacity (TIBC) of 249 μg/dL, transferrin saturation of 20%, and ferritin of 2002.7 ng/mL. There was a significant reduction in ferritin levels with normalization of transferrin saturation and improvement in iron profile parameters, indicating a good response to treatment.

Discussion

Hereditary spherocytosis is an inherited hemolytic anemia caused by defects in erythrocyte membrane proteins leading to spheroidal red blood cells with reduced deformability and increased splenic sequestration.[1,2]

Chronic hemolysis can result in iron overload due to persistent destruction of red blood cells and continuous release of hemoglobin-derived iron into the circulation. However, in chronic hemolytic states such as hereditary spherocytosis, persistent erythrocyte destruction results in excessive iron turnover that may exceed the body's iron-binding and storage capacity.[3]

Iron overload may occur secondary to repeated blood transfusions in disorders such as thalassemia and sickle cell disease, excessive dietary or supplemental iron intake, alcohol-related liver disease, ineffective erythropoiesis with marrow hyperplasia as seen in myelodysplastic syndromes, chronic anemias associated with increased iron absorption, and porphyria cutanea tarda. Genetic causes include hereditary hemochromatosis due to HFE mutations such as C282Y and H63D, as well as non-HFE mutations involving TFR2, HJV, HAMP, and SLC40A1. Therefore, accurate evaluation of iron overload is essential for timely diagnosis, appropriate treatment, and prevention of irreversible organ damage.[4]

Mutations in the SLC4A1 gene cause defects in the AE1 (anion exchanger-1) membrane protein, leading to loss of red blood cell membrane integrity, formation of spherocytes, and the development of hereditary spherocytosis through increased splenic destruction of erythrocytes.[5] Over time, the increased destruction of red blood cells causes increased intestinal iron absorption and release of iron from hemolyzed erythrocytes. This excess iron may accumulate in organs such as the liver, heart, and endocrine glands, resulting in secondary hemosiderosis (secondary iron overload), even in patients who have never received blood transfusions, as seen in our patient.

Decompensated heart failure (DHF) is defined as a clinical syndrome in which a structural or functional change in the heart leads to its inability to eject and/or accommodate blood within physiological pressure levels, thus causing a functional limitation and requiring immediate therapeutic intervention.[6] In the present case, although the patient developed decompensated heart failure, no significant myocardial iron deposition was identified on evaluation. This suggests that the cardiac dysfunction may have been secondary to chronic anemia, persistent hemolytic stress, or other hemodynamic consequences of hereditary spherocytosis rather than direct iron-mediated myocardial injury.

Previous reports have described the rare occurrence of severe iron overload and cardiac hemochromatosis in patients with hereditary spherocytosis. Fujino T *et al*. reported a 31-year-old man with hereditary spherocytosis who developed atrial tachycardia, cardiogenic shock, myocardial iron deposition, and refractory heart failure despite iron chelation therapy, eventually resulting in death.[7] Another report described a 24-year-old man with severe heart failure and myocardial iron deposition who showed clinical improvement after initiation of iron chelation therapy.[8] Also, a patient with genetically confirmed HFE-associated hemochromatosis and severe left ventricular dysfunction demonstrated significant

improvement in cardiac function and cardiac MRI T2 values following treatment with deferoxamine and deferiprone.[9] Only a few cases of hereditary spherocytosis associated with iron overload or hemochromatosis have been reported in the literature.[10]

Hereditary spherocytosis is an inherited red blood cell membrane disorder characterized by premature destruction of erythrocytes, leading to chronic hemolytic anemia, whereas hereditary hemochromatosis is an inherited disorder of iron metabolism characterized by excessive intestinal iron absorption and systemic iron overload.[11] Table 1 shows the major differences between hereditary hemochromatosis and hereditary spherocytosis with respect to pathogenesis, clinical manifestations, laboratory findings, diagnostic methods, and management.

Legend

HFE-High Iron Fe gene; MCHC-Mean Corpuscular Hemoglobin Concentration

Understanding the differences between hereditary hemochromatosis (HH) and hereditary spherocytosis (HS) is critical, as misdiagnosis or failure to identify a coexisting condition may lead to inappropriate management and potentially life-threatening complications.[12] The presence of complete heart block, pulmonary edema, elevated ferritin levels, and MRI evidence of iron overload emphasised early diagnosis in our patient. Early recognition of iron overload using serum ferritin and MRI T2 imaging, along with prompt initiation of iron chelation therapy, is essential to prevent irreversible organ damage and improve clinical -outcomes.[13]

Overall, we report a patient presenting with acute decompensated heart failure secondary to iron overload caused by hereditary spherocytosis associated with an SLC4A1 gene mutation. Chronic hemolysis in hereditary spherocytosis can lead to increased intestinal iron absorption and progressive tissue iron accumulation, even without a history of blood transfusions. In our patient, MRI demonstrated moderate hepatic iron deposition with no significant myocardial iron overload on cardiac iron quantification. However, the patient developed acute decompensated heart failure with reduced left ventricular systolic function, which subsequently improved with appropriate treatment. Genetic analysis identified a homozygous SLC4A1 missense variant, supporting the diagnosis of hereditary spherocytosis as the underlying cause of secondary iron overload. This case highlights the need to evaluate for iron overload and its systemic complications in patients with hereditary spherocytosis, including those who have never received blood transfusions.

Conclusion

Hereditary spherocytosis should be considered in adults presenting with recurrent jaundice and hemolytic anemia, even when the clinical presentation is dominated by cardiac manifestations rather than the classical triad. A stepwise diagnostic approach including hemolysis workup, osmotic fragility testing, iron studies, cardiac evaluation, and genetic analysis can help avoid diagnostic delay. Although repeated transfusions are a recognized cause of secondary iron overload, chronic hemolysis and increased intestinal iron absorption may also contribute to iron accumulation in hereditary spherocytosis even in non-transfusion-dependent patients.

In the present case, we report a 67-year-old male who presented with acute decompensated heart failure and was diagnosed with hereditary spherocytosis. The diagnosis was confirmed by a positive osmotic fragility test, peripheral smear demonstrating spherocytes, DAT negativity, and genetic testing identifying an SLC4A1 variant. MRI demonstrated moderate hepatic iron overload without significant myocardial iron deposition. Despite the absence of substantial cardiac iron accumulation, the patient developed heart failure, which was considered multifactorial, likely related to chronic hemolytic anemia, iron overload, and volume overload rather than direct myocardial iron toxicity. The patient was managed with standard heart failure therapy along with iron chelation therapy, following which there was clinical improvement and recovery of left ventricular systolic function.

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