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Case Report

A Rare Clinical Case Report of Hereditary Spherocytosis in a 17-Year-Old Female

Hemalatha Ganjala*, Ramya Jyothi Padigala, Jayasri Dimmiti, Ram Nageswara Rao Gajula, Mohana Kollipara

Department of Pharmacy Practice, Nirmala College of Pharmacy, Acharya Nagarjuna University, Atmakur, Mangalagiri, Guntur, Andhra Pradesh, INDIA.

ABSTRACT

A congenital hemolytic condition called Hereditary Spherocytosis (HS) is caused by a lack of Red Blood Cells' (RBCs') plasma membrane protein. The hereditary condition is characterized by splenomegaly, jaundice, and anemia. We describe a case of a 17-year-old female patient who came to the surgical gastrointestinal department complaining of two days of abdominal pain. The patient also had a cold and cough for a few days, but these symptoms were unrelated to the main diagnosis. Laboratory investigations confirmed the diagnosis of hereditary spherocytosis. The patient underwent laparoscopic splenectomy and cholecystectomy as definitive management. Postoperatively, her symptoms resolved, and all the abnormal laboratory parameters normalized at the time of discharge. This case underscores the significance of early recognition and surgical management in enhancing outcomes for patients with hereditary spherocytosis.

Keywords: Hereditary spherocytosis, Jaundice, Plasma membrane protein, Congenital hemolytic condition.

Correspondence:
Ms. Hemalatha Ganjala
Pharm. D, Department of Pharmacy Practice, Nirmala College of Pharmacy, Atmakur, Mangalgiri, Guntur-522503, Andhra Pradesh, INDIA.
Email: ganjalahemalatha30@gmail.com

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INTRODUCTION

Hereditary spherocytosis is a common hemolytic disease characterized by a defect or deficiency in one or more proteins that make up the red blood cell membrane. Consequently, red blood cells are abnormally shaped, have increased metabolic demands, and are prematurely sequestered and destroyed in the spleen (He *et al*., 2018). Membrane abnormalities are the most common cause of hereditary spherocytosis. These abnormalities make erythrocytes less deformable and more susceptible to degradation in the spleen. The genes most commonly affected are those encoding spectrin, band 3, and ankyrin, which are membrane proteins (Delaunay 2007). Oskar Minkowski first characterized this illness and published his findings on familial clusters in 1900 (Kar *et al*., 2009). Hereditary spherocytosis is characterized by splenomegaly, anemia, jaundice, and family history. Consequences include pigment gallstones, aplastic, hemolytic, and megaloblastic crises, stunted growth, skeletal abnormalities, and, less frequently, skin ulceration and persistent dermatitis. Physical examination, complete blood cell count, reticulocyte count, and other measures can all serve as the foundation for a logical diagnosis. Because no single test detects all types of hereditary spherocytosis, it is advised to use two test methods (ideally the AGLT and EMA tests) (Bolton-Maggs *et al*., 2012). For moderate to severe cases of hereditary spherocytosis, splenectomy is considered the conventional surgical procedure. It is recommended for patients with severe aplastic crisis, cholelithiasis, developmental delay, substantial splenomegaly, and recurrent hemolytic crisis—especially children (Eber *et al*., 1992).

CASE REPORT

A 17-year-old female patient presented to the surgical gastrointestinal department with 2 days of abdominal pain. She also had a few days of cold-like symptoms and cough, which were incidental and unrelated to the primary diagnosis. Birth history revealed postnatal jaundice soon after delivery, suggestive of hemolysis due to hereditary spherocytosis. She experienced recurrent jaundice every 2 years since childhood. At age 17, the condition was formally confirmed. Developmental history was normal for age, with no reported delays. Immunization history was complete according to the national schedule. There was no family history of hemolytic conditions or HS. Physical and systemic examination was normal. Laboratory investigations revealed decreased hemoglobin, hematocrit, red blood cell count, lymphocytes, eosinophils, and serum sodium. Increased values were observed in red cell distribution width, total white blood cell count, reticulocyte count, absolute neutrophil count, platelet count, total bilirubin, direct bilirubin, indirect bilirubin, SGOT, PTT, and APTT. Table 1 shows the key diagnostic markers.

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Peripheral smear examination revealed anisocytosis with normocytic, hypochromic red blood cells, polychromatophils, and 7-8 nucleated red blood cells per 100 white blood cells. White blood cell morphology showed neutrophilic leukocytosis with a left shift. Bedside ultrasonography of the abdomen demonstrated hepatomegaly with mildly altered parenchymal echotexture, echogenic gallbladder sludge with diffuse edematous wall thickening, splenomegaly, and minimal ascites.

After splenectomy and cholecystectomy, the patient was advised to receive Amoxicillin-Clavulanate injection (1.2 g) intravenously every 8 hours and Piperacillin-Tazobactam injection (4.5 g) intravenously every 8 hr to prevent postoperative infections. Pantoprazole injection (40 mg) was administered intravenously once daily for gastric protection. Paracetamol injection (1 g) was administered intravenously thrice daily to treat postoperative pain and fever. Table 2 shows the patient's therapeutic approach. At discharge, the patient was prescribed Piptaz (piperacillin + tazobactam) injection (4.5 g) intravenously thrice daily for three days to prevent postoperative infections. Pantoprazole tablet (40 mg) was prescribed once daily, preferably in the morning, to protect the stomach from acid and prevent ulceration. Paracetamol tablets (650 mg) were advised thrice daily for 5 days as an analgesic and antipyretic. Table 3 shows the patient's discharge medications.

DISCUSSION

HS is a hereditary condition that affects structural proteins in erythrocyte membranes, most often band 3, α- spectrin, β-spectrin, protein 4. 4.2, and ankyrin- 1 (Christensen and Henry 2010). These abnormalities result in spherical, hyperdense, poorly deformable Red Blood Cells (RBCs) with a short lifespan that are eventually destroyed in the spleen, leading to spherocyte formation and extravascular hemolysis (Christensen *et al*., 2015). While HS can affect people of any race or ethnicity, it typically affects 1 in 2000 white neonates with Northern European heritage (Christensen *et al*., 2015). About 65% of affected newborns have a parent with HS, indicating that HS is most frequently inherited in an autosomal dominant form. However, a negative family history should not be used as an excuse to ignore HS, because up to 30% of people with the condition have de novo mutations or recessive inheritance (Gallagher 2021). The traditional trio of HS in neonates (anemia, splenomegaly, and jaundice) is uncommon. A newborn may present with hydrops fetalis or be asymptomatic. Jaundice in newborns is the most typical sign of HS (Coramusi *et al*., 2025). Splenomegaly is uncommon in newborns with HS and is usually observed in older children and adults. A third of blood smears from affected newborns do not show spherocytes, despite their being characteristic of the condition. Although spherocytes can be observed in DAT + ABO incompatibility, their presence does not usually indicate HS. However, if spherocytes are consistently observed in a hyperbilirubinemic ABO- incompatible DAT-negative neonate, HS should be seriously investigated (Cortesi *et al*., 2025). In this case, the patient presented with chief complaints of abdominal pain. The patient also experienced a few days of unrelated symptoms, including a cold and cough. The patient had a past medical history of recurrent jaundice since childhood, including postnatal jaundice, suggestive of chronic hemolysis. Notably, there was no significant family history of similar illness in this patient. However, the absence of family history does not confirm HS, as approximately 30% of patients may exhibit autosomal recessive inheritance or de novo mutations.

Physical examination, total red blood cell count, reticulocyte count, medical history, and selected tests-particularly the EMA test (eosin-5-maleimide binding) and AGLT (acidified glycerol lysis time)-can be used to make the diagnosis. Hyperbilirubinemia, reticulocytosis, and anemia are common indicators of hemolytic disease. RDW is an indicator of RBC anisocytosis. Wide variations in RBC size, or increased RDW, are a sign of HS (Achenjang *et al*., 2025). In this instance, the patient's laboratory investigations showed anemia with reticulocytosis, hyperbilirubinemia, and increased red cell distribution width, consistent with hemolysis. Peripheral smear examination revealed anisocytosis, polychromatophils, and nucleated red blood cells, although spherocytes were not explicitly noted. The

Table 1: Key laboratory findings in this patient.

Sl. No.	Laboratory parameters	Observed values	Normal values
	Complete Blood Count (CBC)		
1.	Haemoglobin (Hb)	11.2 g/dL ↓	11.5-16 g/dL
2.	Haematocrit (PCV)	15.8 % ↓	36-46 %
3.	Red Blood Cells (RBC) count	2.99 million/cu.mm ↓	3.8-4.8 million/cu.mm
4.	Red Cell Distribution Width (RDW)	15.7% ↑	11.6-14%
5.	Reticulocyte count	6.4% ↑	0.5-2.5%
	Liver function profile		
6.	Bilirubin - total	1.49 mg/dL ↑	0.00-1.20 mg/dL
7.	Bilirubin - direct	0.56 mg/dL ↑	0.00-0.30 mg/dL
8.	Bilirubin indirect	0.93 mg/dL ↑	0.10-0.80 mg/dL

Table 2: Therapeutic approach.

Sl. No.	Brand name (generic name)	Dose	Roa	Frequency	Category
1.	INJ. Amoxycillin + Clavulanate	1.2 g	IV	8th hourly	Penicillin Antibiotic and Beta-Lactamase Inhibitors
2.	INJ. Piperacillin + Tazobactum	4.5 g	IV	8th hourly	Penicillin Antibiotic and Beta-Lactamase Inhibitors
3.	INJ. Pantoprazole	40 mg	IV	OD	Proton Pump Inhibitor
4.	INJ. PCM (Paracetamol)	1 g	IV	1-1-1	Analgesic, Antipyretic

Table 3: Discharge medications.

Sl. No.	Brand name	Generic name	Dose	Roa	Frequency	Duration
1.	INJ. Piptaz	Piperacillin/Tazobactam	4.5 g	IV	1-1-1	For 3 Days
2.	TAB. Pan	Pantoprazole	40 mg	PO	1-0-0	-
3.	TAB. Dolo	Paracetamol	650 mg	PO	1-1-1	For 5 Days

absence of clear spherocytes does not exclude the diagnosis, as similar findings may vary in mild or early disease. Ultrasound findings of hepatomegaly, splenomegaly, and gallbladder sludge further supported a chronic hemolytic process. The diagnosis in this case was delayed due to the absence of specific characteristics like early age diagnosis, absence of family history, and clear spherocytes. All the laboratory investigations taken together, the classical triad of HS (recurrent jaundice, splenomegaly, and anemia), hyperbilirubinemia, hepatomegaly, gallbladder sludge, and peripheral smear findings are suggestive and confirmatory of the diagnosis of HS in this patient. The diagnosis was formally confirmed at age 17 based on clinical correlation, laboratory findings, and pedigree analysis.

The treatment of HS involves cholecystectomy and splenectomy. For moderate to severe hereditary spherocytosis, splenectomy is the most common surgical procedure. Regular monitoring of hemolysis markers and supportive therapy with folic acid supplementation are required (Manciu *et al.*, 2017). In this case, the patient underwent splenectomy and cholecystectomy under General Anesthesia (GA).

CONCLUSION

This case highlights a patient presenting with HS who had recurrent jaundice since childhood. Despite no family history, the clinical manifestations and all the laboratory tests confirm the HS diagnosis. Careful evaluation of clinical features, laboratory results, and imaging led to a timely diagnosis, underscoring the importance of vigilance in recognizing HS even in atypical presentations. In our case, the patient with hereditary spherocytosis recovered well following splenectomy and cholecystectomy. She was counseled on lifestyle measures, including a balanced diet, folate supplementation, proper hydration, and regular follow-up to monitor for complications.

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ABBREVIATIONS

HS: Hereditary Spherocytosis; **AGLT:** Acidified Glycerol Lysis Time; **EMA:** Eosin-5-Maleimide Binding; **SGOT:** Serum Glutamic Oxaloacetic Transaminase; **PTT:** Partial Thromboplastin Time; **APTT:** Activated Partial Thromboplastin Time; **RBC:** Red Blood Cell; **RDW:** Red Cell Distribution Width; **PCV:** Packed Cell Volume; **IV:** Intravenous; OD**:** Once a Day; **PO:** Per Oral.

CONFLICT OF INTEREST

The authors declare that there is no conflict of interest.

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