CASE REPORT

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A Case Report on Hereditary Spherocytosis in a 22-Year-Old Female

Umme Habeeba A Pathan

Department of Pharmacy Practice, Bapuji Pharmacy College, Davangere, Karnataka, INDIA.

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***Correspondence to:**

Ms. Umme Habeeba A Pathan,

Pharm D Intern, Department of Pharmacy Practice, Bapuji Pharmacy College, Davangere-57004, Karnataka, INDIA.

Email id: ummehabeeba777@gmail.com

Abstract

Hereditary spherocytosis is a rare genetically transmitted spherical shaped red blood cell disorder which leads to hemolytic anemia. The symptoms vary from asymptomatic to severe life-threatening anemia. The management includes splenectomy, blood transfusion and vitamin supplements. A 22-year-old female comes to tertiary care hospital with the complaints of fever, yellowish discoloration of eyes and urine. She was diagnosed with hereditary spherocytosis 10 years back and was continuously on blood transfusion. On day 1, her hemoglobin was 6.3 g/dL which falls under life-threatening anemia, total bilirubin was 7.9 g/dL, and direct bilirubin was 0.9 g/dL. She was given with blood transfusion for two days and hemoglobin level raised to 10 g/dL. This case presents life-threatening anemia and icterus associated with severe hereditary spherocytosis. The treatment in hereditary spherocytosis is quite challenging often because of its obscurity.

Keywords: Hereditary spherocytosis, Life-threatening anemia, Icterus, Transfusion.

INTRODUCTION

Hereditary Spherocytosis (HS) is a rare inherited hemolytic anemia mainly affecting red blood cell (RBC) membrane which shows spherical shaped RBC on peripheral smear and cause cell lysis.[1-3] It was first described in 1817.[4] HS is inherited as autosomal dominant pattern in about 75% of cases whereas rest 25% of cases occur sporadically.[5] HS is a product of heterogenous alteration in one of the six genes, most often in the ankyrin gene (ANK1) which encodes for ankyrin.[6,7] Other commonly mutated genes are SPTB, SLC4A1, EPB42 and SPTA1 encoding for spectrin β-chain, anion exhanger 1(band 3), protein 4.2 and the spectrin α-chain respectively.[7,8] The characteristics of HS vary from asymptomatic to fulminant hemolytic anemia.[2] The classical symptoms include anemia, jaundice, splenomegaly and elevated red blood cell osmotic fragility.[9] HS is the most common under-recognized disorder which can present from infancy and throughout the life span.[10]

CASE REPORT

A 22-year-old female visited a tertiary care hospital with the complaints of fever, yellow discoloration of eye and urine since 2 days. She was diagnosed with hereditary spherocytosis at the age of 12 with significant family history of HS. She has been taking blood transfusion since 10 years after visiting to the hospital for regular checkup. She gets to know about her decreased hemoglobin level through routine checkup. On examination her pulse rate was 80 beats per minute, respiratory rate of 22 cycles per minute, blood pressure of 110/70 mm Hg and icterus positive. On day 1, laboratory investigation showed hemoglobin of 6.3g/dL, red blood cells of 2.3 million cells per cubic millimeter, Albumin Globulin ratio (A/G) was 1.8, total bilirubin was 7.9 mg/dL, direct bilirubin was 0.9 mg/dL, and indirect bilirubin was 7.0 mg/dL while other liver function parameters like Aspartate Amino Transferase (AST), Alanine Amino Transferase (ALT), Alkaline Phosphatase (ALP) and Gamma Glutamyl Transpeptidase (GGT) was normal. Urine examination shows pus cells positive, epithelial cells 0-1/hpf and leucocytes 5-6/hpf. On day 3, her hemoglobin was 10 g/dL after blood transfusion for 2 days. Her treatment chart and transfusion drugs are shown in the Table 1 and Table 2 respectively.

DISCUSSION

HS is most common inherited disease with deficiency in red cell membrane proteins like ankyrin and spectrin.[5] Based on the severity of disease, HS is classified into mild, moderate and severe accounting for 10 to 20%, 60 to 75% and 5% of cases respectively.[6] Mild HS is associated with non-anemic, reticulocytosis, splenomegaly or jaundice and undetectable until adolescence; moderate HS is associated with anemia, high reticulocyte count and elevated serum bilirubin and severe HS is associated with marked anemia, hemolysis, hyperbilirubinemia, splenomegaly, and regular blood transfusions.[6] This case belongs to severe HS category due to the life-threatening anemia with total bilirubin levels of 7.9mg/dL and 2 pint of blood transfusion.

ANK1 gene mutation were found in 45% of Asian patients according to the report by Yang *et al.*, in 2018 which is the reason that HS is common in Asia than compared to other continents.[7] Complications of HS are pigmented gallstones, anemia – hemolytic, megaloblastic and aplastic, decreased growth, gout, hematological malignancies, cardiomyopathy and skin disorders.[2,4]

Table 1: Treatment chart.

Brand name	Generic name	Dose	Frequency	Day 1	Day 2	Day 3	Day 4
Inj. Xone	Ceftriaxone	1g	1-0-1	✓	✓	✓	✓
Inj. Pan	Pantoprazole	40 mg	1-0-0	✓	✓	✓	✓
Tab. Dolo	Paracetamol	625 mg	1-1-1	✓	✓	✓	✓
Inj. Ondem	Ondansetron	4 mg	sos	✓	✓	✓	✓

Table 2: Transfusion and related medications.				
Brand name	**Generic name**	**Dose**	**Day 2**	**Day 3**
1 pint PCV	437 ml of blood	437 ml	✓	✓
Before transfusion				
Inj. Lasix	Furosemide	20 mg	✓	✓
Inj. Hydrocort	Hydrocortisone	-	✓	✓
Inj. Avil	Pheniramine maleate	-	✓	✓

According to the diagnosis and treatment guidelines, patient with positive family history of HS, typical clinical features like sphere shaped blood cells in peripheral smear can be diagnosed as HS.[9] Other diagnostics include osmotic fragility test, acidified glycerol lysis test, cryohemolysis test, osmotic gradient ektacytometer and sodium dodecyl sulphate – polyacrylamide gel electrophoresis.[5] Definitive treatment of HS includes splenectomy but is associated with increased risk of infection.[10]

CONCLUSION

This is a case of 22 years old female suffering from severe HS. She had symptoms of severe HS that is life-threatening anemia, elevated levels of bilirubin, and icterus. Her management include 2 pint of blood transfusion with prophylactic usage of antibiotic and antiemetic. To reduce fever and prevent any gastric irritation she was prescribed with paracetamol and pantoprazole. Before transfusion she was given furosemide, pheniramine maleate and hydrocortisone to prevent transfusion related reactions. Splenectomy can be considered in this case to prevent further complications

CONFLICT OF INTEREST

The author declares that there is no conflict of interest.

REFERENCES

1. Park JB, Lee SA, Kim YH, Lee WS, Hwang JJ. Extramedullary Hematopoiesis Mimicking mediastinal Tumor in a Patient with Hereditary spherocytosis: case report. Int J Surg Case Rep. 2017;41:223-5. doi: 10.1016/j.ijscr.2017.10.043, PMID 29096349.
2. Hassan A, Babadoko AA, Isa AH, Abunimye P. Hereditary spherocytosis in a 27-year-old woman: case report. Ann Afr Med. 2009;8(1):61-3. doi: 10.4103/1596-3519.55768, PMID 19763011.
3. Nigwekar P, Shrikhande DY, Niranjan BK, Shah N, Shukla T. Hereditary spherocytosis – A rare case report. Pravara Med Rev. 2010;2(4):25-9.
4. Kalyan M, Kanitkar SA, Gaikwad AN, Kumar H. Hereditary spherocytosis. Med J Dr DY Patil Univ. 2014;7(2):229-31.
5. Koju S, Makaju R. Hereditary spherocytosis. J Lumbini Med Coll. 2018;6(1):41-3. doi: 10.22502/jlmc.v6i1.202.
6. Venkatamurthy M, Kumar BA. A case report of hereditary spherocytosis affecting mother and child with varied severity. Ped Rev. Int J Pediatr Res. 2017;4(7):494-6.
7. Luo Y, Li Z, Huang L, Tian J, Xiong M, Yang Z. Spectrum of ankyrin mutations in hereditary spherocytosis: A case report and review of the literature. Acta Haematol. 2018;140(2):77-86. doi: 10.1159/000492024, PMID 30227413.
8. Agarwal AM. Ankyrin Mutations in Hereditary Spherocytosis. Acta Haematol. 2019;141(2):63-4. doi: 10.1159/000495339, PMID 30602149.
9. Xiaoqiu W, Bingmu F, Jinhong J, Zhigang Q, Guangli M, Yonghua L *et al*. A case report of hereditary spherocytosis with concomitant chronic myelocytic leukemia. Open Med (Wars). 2016;11(1):152-4. doi: 10.1515/med-2016-0029, PMID 28352784.
10. Will A, Henderson CA, Jnah AJ, Newberry D. Hereditary spherocytosis in the neonatal period: A case report. Neonatal Netw. 2017;36(5):280-8. doi: 10.1891/0730-0832.36.5.280, PMID 28847351.