Hereditary Spherocytosis in a 17 year girl: A case report

Dr. Ramesh Chandra[1], Dr. Mayank Gupta[2§], Dr. Gaurav Chauhan[3], Dr. Sunil Mahavar[4], Dr. Swati Srivastava[5], Dr. Raman Sharma[6]

[1,3]Junior Resident, Department of Internal Medicine, S.M.S Medical College, Jaipur(Rajasthan)India
[2]Senior Resident, Department of Internal Medicine, SMS Medical College, Jaipur(Rajasthan)India
[4]Assistant Professor, Department of Internal Medicine, SMS Medical College, Jaipur(Rajasthan)India
[5]Professor, Department of Internal Medicine, SMS Medical College, Jaipur(Rajasthan)
[6]Senior Professor, Department of Internal Medicine, SMS Medical College, Jaipur(Rajasthan)
[§]Corresponding author's

Abstract— *Hereditary spherocytosis (HS) is a familial hemolytic disorder with marked heterogeneity of clinical features, ranging from an asymptomatic condition to a fulminate haemolytic anaemia. Although a positive family history of spherocytosis increases the risk for this disorder, it may be sporadic in some cases. Seventeen years old girl presented with mild anaemia, jaundice and moderate splenomegaly. Her haematological parameters supported diagnosis of hereditary spherocytosis. Appropriate treatment was started with an advice to patient to undergo splenectomy as an active part of management of the disorder. Mother and other siblings were normal.*

Keywords: Anaemia, Jaundice, Splenomegaly, Hereditary, Spherocytosis.

I. INTRODUCTION

Hereditary spherocytosis (HS) is a rare inherited red blood cell (RBC) membrane disorder that is characterized by spherically shaped RBCs on peripheral blood smear[1, 2, 3]. Hereditary spherocytosis was first described in 1871.[4] T

The exact data is not available but it seems quite uncommon in India. The incidence is 1:4500 in Caucasians[2, 3] (North Africa, West Asia, and Europe). It is inherited as autosomal dominant but approximately 25% of cases are discovered in persons who have no family history of HS; these cases may represent spontaneous mutations or recessive forms of the disease.[4]

Splenectomy almost cures anemia but should be deferred till 5 to 6 years of age due to fear of fulminant sepsis.[1, 2, 3] This case is reported in view of the rarity of this disorder in India and it is hoped that this report will bring increase awareness on the existence of HS and hence an increase index of suspicion to avoid unnecessary use of blood transfusion.

II. METHODOLOGY

A case who was presented with with complaints of generalised weakness, yellowish discoloration of eyes and skin since 6 months and decreased appetite, non-documented weight loss since four months at SMS Medical College, Jaipur (Rajasthan) India. On investigation it came out to Hereditary spherocytosis (HS) which is a very rare disease. So case was studied thoroughly and case report was prepared to publish this rare case.

III. CASE REPORT

A Seventeen years old girl reported from a town, Suratgarh in Rajasthan to SMS Hospital on 8th May 2017 with complaints of generalised weakness, yellowish discoloration of eyes and skin since 6 months

and decreased appetite, non-documented weight loss since four months. She also had exertional breathlessness and easy fatigability sine 6 months.

She had significant h/o recurrent episodic jaundice at the age of 2 years; 6 years & at 12 years. Her father (42 years old) suffered from jaundice 20 years back at the age of 19 years for which he had received treatment and cured but no records were available. Other four siblings were normal. No past history of blood transfusion or hospitalization was there.

Examination revealed a young lady, with a small and short stature; mildly pale and moderately jaundiced (Figure 1). The spleen was enlarged by 6cm below the right costal margin but the liver was not palpably enlarged. Other systems were normal.

On routine investigation-

i. Haemogram – hemoglobin 9 gm/dl.
ii. Peripheral blood smear- RBCs showed anisopoikilocytosis, few spherocytes, mild polychromasia, no nucleated RBCs seen, hypochromasia of mild degree, No hemoparasite seen. WBCs and platelets were normal (Figure 2).
iii. Red cell indices - MCHC- 40 %, MCV -74.3 fl, RDW – 22.8%
iv. Reticulocyte count – 11.45%
v. Osmotic fragility –started at 0.70% of NaCl and ended at 0.45% of NaCl. (Markedly increased)
vi. Bone marrow- showed erythroid hyperplasia consistent with hemolytic anemia
vii. Hemoglobin electrophoresis- normal
viii. Direct coomb's test- negative
ix. Liver function tests- S.bilirubin- total 5.1 mg/dl and indirect 3.4 mg/dl (unconjugated
x. hyperbilirubinimia).
xi. Abdominal sonography revealed Moderate Splenomegaly (16.4 cms). Liver, Gall Bladder, Pancreas and Both Kidneys were normal.

Figure 1
Pallor and icterous Eye

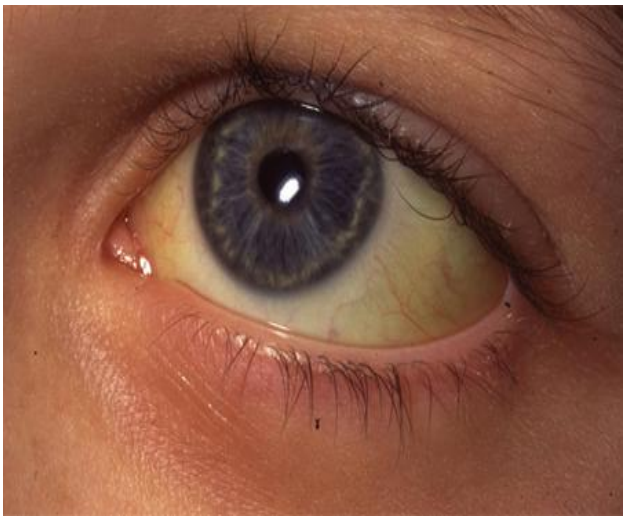
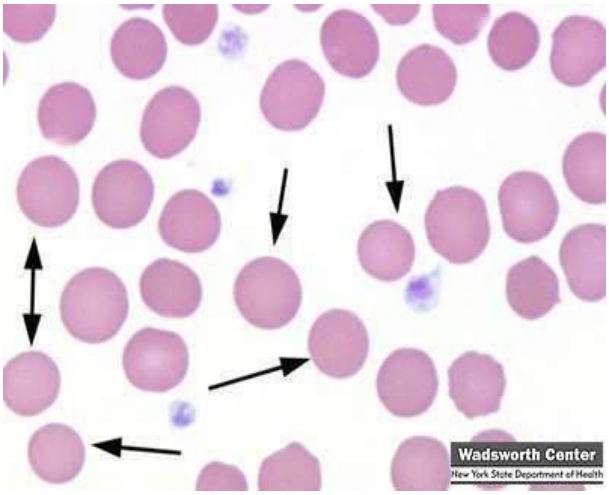


Figure 2
Peripheral Blood Smear



Based on family history, clinical findings and investigations the diagnosis of hereditary spherocytosis was confirmed. She was put on folic acid supplement 5 mg daily. Girl is under consideration for total splenectomy.

IV. DISCUSSION

Hereditary Spherocytosis is the commonest inherited red cell membrane disorder. It is common in west Asia, North Africa and Europe with incidence of 1: 1000 to 1:4500[2, 3]. It is quite rare in India but exact data is not available. First clinical description of HS was given by Vanlair and Mesius in 1871[4]. HS has wide spectrum of severity from asymptomatic disease without anemia with minimal hemolysis to severe hemolytic anemia requiring frequent blood transfusions to sustain life. It is usually transmitted as autosomal dominant trait and less frequently as autosomal recessive trait. As many as 25% have no family history and are fresh mutants.[4]

Hereditary Spherocytosis results from cytoskeleton defect in primary red cell membrane[3]. Most cases of HS are heterozygous because homozygous states are lethal[1-4]. In pedigrees that have a dominant defect, affected family members tend to have similar degrees of hemolysis and clinical severity.[5, 6, 7] Four abnormalities in red cell membrane proteins have been identified and include 1) Spectrin deficiency alone, 2) Combined spectrin and ankyrin deficiency, 3) Band 3 deficiency, and 4) Protein 4.2 defects. Ankyrin gene mutation is the m/c defect in HS. Each is associated with a variety of mutations that result in different protein abnormalities and varied clinical expression. A deficiency in spectrin, ankyrin, or protein 3 causes defects in vertical stabilization of the phospholipid bilayer of the RBC membrane cause separation of the spectrin –phospholipid bilayer. Portions of the phospholipid bilayer form vesicles and are lost from the RBC surface resulting in decreased surface area and spherocytosis.

In the first few postnatal months, anemia can develop in children who do not mount an adequate reticulocyte response[4]. In later childhood, HS can present with anemia, jaundice, and splenomegaly[4-6]. Affected patients may have mild, moderate, or severe anemia. Children who have moderate to severe anemia may have poor exercise tolerance, poor growth, and academic difficulties[4-6]. Older individuals develop bilirubin stones and may present with cholecystitis.[4-6]

People with mild HS may remain undiagnosed for decades and only be discovered when spherocytes are seen when a blood count is done for another indication, or the person is found incidentally to have an enlarged spleen.

V. CONCLUSION

Although HS is rare it does occur in our environment and when suspected haematological assessment is necessary to avoid diagnostic pitfalls and mismanagement.

CONFLICT OF INTEREST

None declared till now.

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