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Case Report

Hereditary Spherocytosis: A Case Report

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ABSTRACT:

Introduction: Hereditary spherocytosis is a red cell membrane disorder that causes hemolytic anemia. Due to defective cell membrane, red cells are spherical shaped and result in their early lysis. Osmotic fragility of spherocytic red cell is increased. **Case report**: A 22 year old female presented with chief complain of abdominal pain. Initially she was diagnosed as cholelithiasis. Under laboratory evaluation she was found to be anemic with reticulocytosis. In peripheral blood smear, spherocytes were moderately distributed. Antihuman globulin test was negative but osmotic fragility was high. Hence, she was confirmed as case of hereditary spherocytosis. **Conclusion**: Hereditary spherocytosis is a rare red cell disorder and its diagnosis can be made by osmotic fragility test.

Keywords: anemia • hereditary spherocytosis • osmotic fragility test • red blood cell

INTRODUCTION:

Hereditary spherocytosis (HS) is an inherited red cell membrane disorder causing hemolytic anemia. About 75% of cases are inherited in autosomal dominant pattern while 25% occur sporadically. The prevalence of HS in Northern Europe and North America is about one in approximately 2,000.[1,2] Its prevalence in Nepal is unknown and there are only a couple of case reports from our country. Diagnosis and management of HS is still a challenge. We hope this report will bring awareness on existence of HS in our community and will help in early diagnosis with proper management.

CASE REPORT:

A 22 year old woman presented to our hospital with the chief complain of abdominal pain. She had yellowish discoloration of skin at the time of presentation. She had history of recurrent jaundice and pallor since her childhood. She also had history of multiple blood transfusions. For diagnosis of disease, clinician sent her for ultrasound (USG) evaluation of abdomen and laboratory examination of blood. On USG evaluation, she was initially diagnosed as cholelithiasis with fatty liver.

Laboratory investigation showed hemoglobin (Hb) of 9.5 g/dl with normal total white blood cell (WBC) and platelet counts. Red blood cell count (RBC) was low at 3.0 million/µl. Mean cell volume (MCV) was 83 fl and mean cell hemoglobin (MCH) was 32 pg both of which were normal. Mean cell hemoglobin concentration (MCHC) was 40% which was slightly raised. Reticulocyte count of 20.1% was markedly increased. Biochemical investigation showed an increased total bilirubin of 3.5 mg/dl and direct bilirubin of 0.9 mg/dl while other liver function parameters like alkaline phosphatase (ALP), aspartate transaminase (AST), and alanine transaminase (ALT) were normal. Hematological and biochemical finding suggested hemolytic anemia.

Peripheral Blood smear revealed a predominance of normocytic normochromic with few microcytic red blood cells. Spherocytes were

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moderately distributed (Figure 1). Direct antihuman globulin (DCT) test was negative. Glucose-6-phosphate dehydrogenase (G6PD) test was normal. Hemoglobin Electrophoresis showed normal hemoglobin fractions. However, osmotic fragility test was markedly increased as compared to that of normal healthy individual (Figure 2).

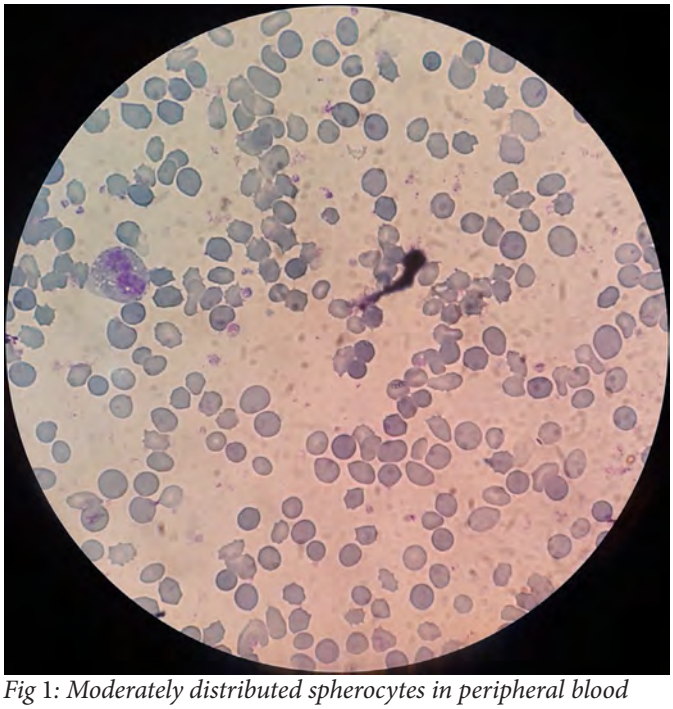

Fig 1*: Moderately distributed spherocytes in peripheral blood smear*

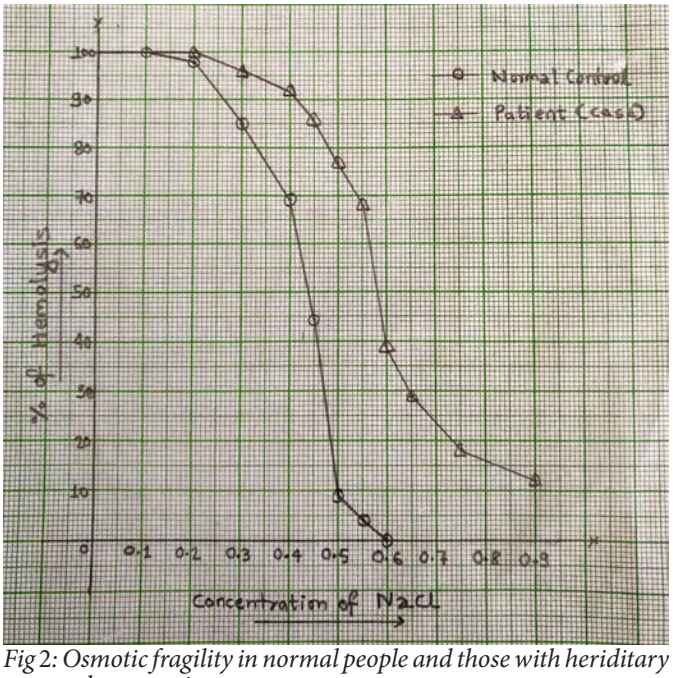

Fig 2*: Osmotic fragility in normal people and those with heriditary spherocytosis*

On basis of the clinical and laboratory investigations, the patient was diagnosed as having hereditary spherocytosis. Patient was counseled for regular follow-up. Family members were counseled for screening for hereditary spherocytosis.

DISCUSSION:

Hereditary spherocytosis (HS) is the most common inherited red cell membrane disorder. It is commonly inherited in autosomal dominant pattern while autosomal recessive inheritance is also seen. Deficiency in red cell membrane protein, namely ankyrin and spectrin, causes HS. These are major proteins that act as cytoskeleton of red blood cell.[1] Due to defective membrane, RBC turns to spherical shaped with decrease in surface area and are rapidly removed from circulation by spleen.

Major clinical features of HS are pallor, jaundice, and splenomegaly. Pallor is due to anemia caused by destruction of red cells in spleen. Jaundice is due to hyper-bilirubinemia caused by rapid destruction of red cells in spleen. Splenomegaly is due to sequestration and phagocytosis of spherical shaped red cells in spleen. These features may be noted at infancy, childhood, or later in adult age. [3] About 20 - 30% of HS are mild and patients are usually asymptomatic with mild splenomegaly, mild reticulocytosis, and few spherocytes in peripheral blood smear. Many such individual are not detected. They may be diagnosed during family study, assessment of gallbladder stone, splenomegaly, or anemia. Moderate form of HS is seen in 60-70% of HS and may present at any age. About three to five percent of cases are severe with life threatening anemia and need regular transfusion. Such severe HS are inherited in autosomal recessive pattern. Cholelithiasis in hereditary spherocytosis occurs due to chronic hemolysis. Most of the stones are formed in patients between 10 to 30 years of age.[1]

Diagnosis of HS can be made from clinical history, family history, physical examination, and laboratory data. Laboratory findings include hemolysis with reticulocytosis and raised bilirubin level. MCV and MCH is normal but MCHC may be raised. 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.[4]

Osmotic fragility test (OFT) is a simple and cost effective test that confirms the presence

of spherocytes. But a normal osmotic fragility test result may be found in 10-20% of the cases. Other tests like acidified glycerol lysis test (AGLT), cryohemolysis test, osmotic gradient ektacytometer, or eosin-5'-maleimide (EMA) test may clinch the diagnosis in these cases. EMA test is more sensitive compared to others and has been used as the reference technique in diagnosis of HS.[5,6] There are few atypical HS which are difficult to diagnose and present with intermittent hemolysis. Such atypical cases can be diagnosed by Sodium Dodecyl Sulfate - Polyacrylamide Gel Electrophoresis (SDS-PAGE) test or molecular techniques.[1,5]

In a study done by European network for rare red cell anemia in 25 European centers, EMA test was used by 60% of the centers, OFT by 50%, cryohemolysis test by less than 20%, and SDS-PAGE and molecular techniques in only atypical cases by few centers.[6] It indicates that even in developed countries there is variation in confirmatory test for hereditary spherocytosis. EMA test has high sensitivity (93%) and high specificity (98%) while osmotic fragility test has sensitivity of 68% and 81% in fresh blood and incubated blood respectively. If EMA and AGLT are performed together, the sensitivity reaches 100%. Hence, this approach is effective diagnostic tool for diagnosis of mild/ compensated HS.[6]

CONCLUSION:

Hereditary spherocytosis is a rare condition and usually remain undetected in many centers of our country as they lack proper diagnostic tools. High index of suspicion and availability of diagnostic investigations are necessary for its diagnosis.

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Conflict of interest:
Authors declare that no competing interest exists.

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