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CASE REPORT

Hereditary Spherocytosis in a young male Report of an unusual case

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INTRODUCTION - Hereditary Spherocytosis (HS) is a familial hemolytic disorder with marked heterogeneity of clinical features, ranging from an asymptomatic condition to a fulminant hemolytic anemia. The etiology of the disease is a deficiency in the membrane proteins which results in instability of the cytoskeleton? Although a positive family history of spherocytosis increases the risk for this disorder, it may be sporadic in some cases. In severe cases the disorder may be detected in early childhood, but in mild cases it may go unnoticed until later in adult life. [1, 2, 3, 4]

CASE REPORT:

A 22 year male reported from a remote village in a tribal area of Nanded to our hospital 16th July 2014 with complaints of abdominal pain, loss of appetite and recurrent jaundice since childhood. He had no history of blood transfusion or any drug intake in the past. He had a positive family history with his elder brother had undergone splenectomy two years back. The reason for which splenectomy done was not available.

On examination he had severe pallor, icterus with lemon yellow tinge; some characteristic changes of hemolytic facies were also noted. Abdominal examination showed mild hepatomegaly and moderate splenomegaly. Other systems were normal. A provisional diagnosis of chronic hemolytic anemia was made and the case was investigated on that line.

The laboratory data were as follows: WBC - 7000 (the morphology of white blood cells and the differential count were normal), Hb 11.5 g/dL, Platelet count - 3lacs, with normal RBC indices. Total-bilirubin 5.1 mg/dL, direct-bilirubin 0.89 mg/dL, LDH 235 U/L, ALT 21 IU/L, AST 61 IU/L. Vitamin B12 levels were normal. The peripheral smear showed polychromasia, anisocytosis and a few spherocytes. Hemoglobinopathies were excluded (Hb electrophoresis was normal). The erythrocytes were assayed for the enzyme G6PD and pyruvate kinase which were normal. The reticulocyte count was 6% and the next test that was performed was osmotic fragility, which showed increased osmotic fragility of the erythrocytes. Flow cytometric analysis of RBCs revealed the membrane defect. Hematological consultation was also obtained and diagnosis of hereditary spherocytosis was confirmed.

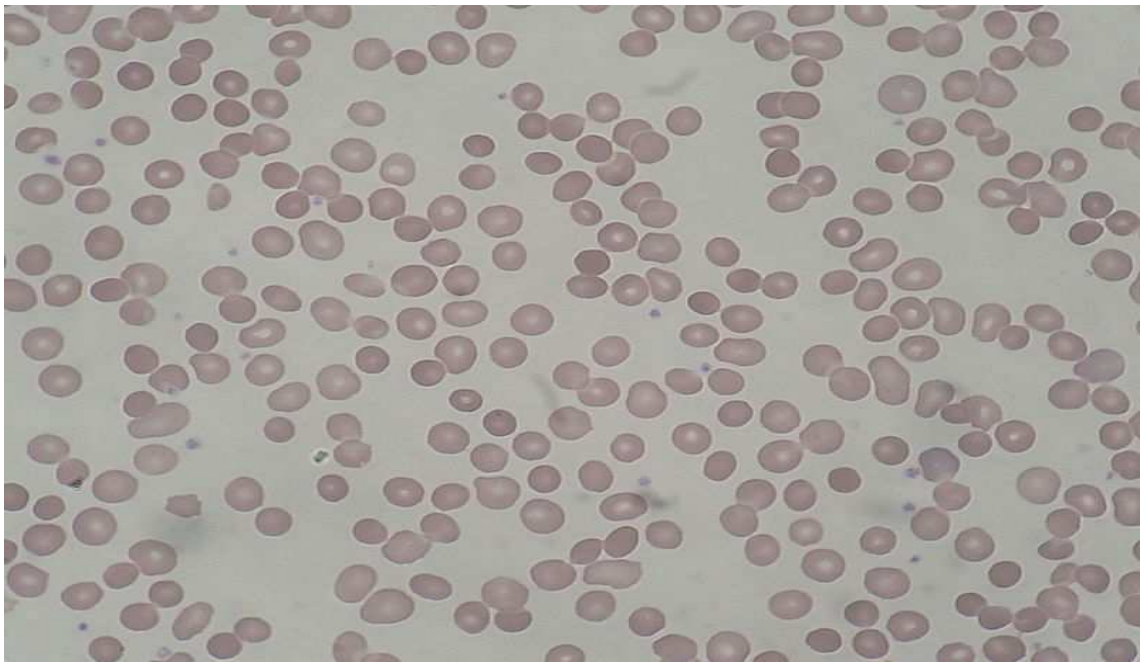

Fig1: Peripheral smear showing polychromasia, anisocytosis and few spherocytes.

DIAGNOSIS AND MANAGEMENT:
Based on family history, clinical findings and investigations the diagnosis of hereditary spherocytosis was confirmed. He was put on folic acid supplement. Patient is under consideration for total splenectomy.

DISCUSSION:
Hereditary spherocytosis is a chronic hemolytic anemia due to defect in red cell membrane. It is common in west Asia, North Africa and Europe with incidence of 1: 1000 to 1:4500. [(5)] It is quite rare in India but exact data is not available. Hereditary spherocytosis 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. The most common molecular defects are of spectrins and ankyrin which are major component of cytoskeleton responsible for RBC shape. Spherical shape of RBC impairs smooth passing from splenic cord to splenic sinuses and the spherocytic RBCs are destroyed prematurely in spleen.

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