HEREDITARY SPHEROCYTOSIS DIAGNOSED IN 16 YEAR FEMALE- A CASE REPORT

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Abstract:

Hereditary Spherocytosis (HS) is relatively common haemolytic anemia in which basic abnormality is an intrinsic defect of red cell membrane. Clinically patients may asymptomatic to intermittent jaundice,anemia, abdominal pain and splenomegaly. Here we reporting a case of 16-year female presented as recurrent abdominal pain,jaundice ,splenomegaly and anemia. Clinical examination show pallor,jaundice and massive splenomegaly. Investigations confirmed diagnosis of HS, by the presence of peripheral blood smear spherocytes,increased MCHC,reticulocytosis,negative direct coombs test and increased osmotic fragility. She advised to undergo splenectomy and cholecystectomy as active part of management.

Key-words: Hereditary Spherocytosis; Microspherocytes ;Gall stones; Splenomegaly

Introduction:

Hereditary Spherocytosis(HS) is inherited as an autosomal dominant trait, characterised by hemolytic anemia secondary to intracorpuscular abnormality of red cells, which renders them most susceptible to normal mechanism of cell destruction. HS affects male and female equally and is not confined to any particular race,but more commonly encountered in british and northern Europeans[1].Clinical features consist triad of anemia, jaundice and splenomegaly. However such triad not always found in mild hemolysis or compensated hemolysis. Pigment gall stones are frequently seen. Peripheral blood films spherocytosis, Increased MCHC in hemogram and increased osmotic fragility favour diagnosis of HS [2]. No definite treatment yet available for this disease. Present case report of recurrent right sided abdominal pain evaluated and diagnosed as HS, which was undiagnosed until the age of sixteen.

Case report:

A 16-year female, brought by her parents to the emergency department with complaining of recurrent abdominal pain for six years, on & off yellowish discoloration of eyes, weakness for two months. There was no vomiting,no itching over body ,no blood in stool, normal stool colour and no transfusion so far. On general examination she had pallor, jaundice and no lymphadenopathy. Her vitals were normal. Systemic examination revealed tenderness in right hypochondriac region, splenomegaly 10 cm below costal margin with firm ,nontender and irregular margin. Cardiovascular and respiratory examination were normal.

Lab investigation show hemoglobin: 7.0 g/dl (12-18g/dl); hematocrit:20% (37-52%); erythrocytes:1.88 millions/cumm (4.2-5.5); mean corpuscular volume(MCV): 84fl(MCH) (76-96fl);meancorpuscular hemoglobin (MCH)29.4 pg (27-32pg); MCHC 35.1g/dl(29-32g/dl), red

cell distribution width: 23% (11.5 to 14.5); reticulocytes: 20% (0.5-2);leukocytes: 7700 cell/mm3 (400-11000) with normal differential count, platelets: 200 cell/mm3 (150,000-450,000).Peripheral blood smear reveal (Figure1) predominantly microcytic RBCs,moderate anisopoikilocytosis,presenceof many spherocytes (absence of area of central pallor),polychromasia,4 nucleated RBCs/100WBCs. Liver function test showed indirect bilirubin: 3.5 mg/dl (< 0.7mg/dl) and elevated lactic dehydrogenase: 384U/L (120-240U/L). Serum creatinine ,blood urea levels and serum electrolytes were normal. Direct coombs test was negative and HB electrophoresis revealed no abnormal hemoglobin. Sickling test was negative. Increased Osmotic fragility in hypotonic saline solutions seen.

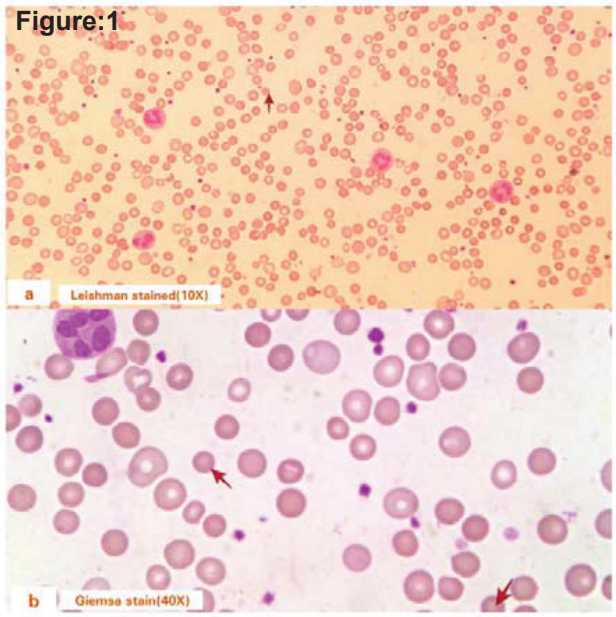

Figure:1 a; Leishman stain blood smear (10X) & Giemsa stain (40X) show many spherocytes (red arrow) with small round,dense RBCs with loss of central pallor.

Chest X ray show normal study.Ultrasound revealed moderate splenomegaly and altered echotexture of liver and multiple gall stones. Based on peripheral blood films findings,high MCHC, reticulocytosis , negative direct coombs test, increased osmotic fragility diagnosis of hereditary spherocytosis was made and suggested supportive treatment and surgical intervention.

Discussion:

Hereditary Spherocytosis (HS) is common red cell membrane disorder, but rarely found in India. Mostly inherited as autosomal dominant but around 25% cases spontaneous mutation or recessive forms seen where there is no family history of HS. In present case there was no family history of anemia or hemolysis.Asymptomattic HS carrier state might exist in 1.4% of the population [3].

The basic defect in HS is defect in one of major protein (spectrin) in cytoskeleton of RBCs membrane resulting in decreased ratio of surface area to volume leading spherocytes [4]. These spherocytes are rigid cells and can not pass through slit like opening of splenic cords and sinuses and prolong stay in spleen resulting in to hypoxia and deprivation of glucose, leading to loss of red cell membrane and this causes further rigid and sphere RBCs and make prone for phagocytosis by reticulo-endothelial cells of spleen. Anemia results when the rate of red cell destruction exceeds the rate of bone marrow regeneration. Functional defect in membrane seen as increased permeability for sodium. An increased rate of passive movement of sodium into the cell is compensated for an increased rate of active transportof sodium out of cell by the cation pump mechanism which require ATP derived from red cell glycolysis The glycolytic rate of the cells greately increased as a compensatory mechanism to provide adequate ATP[4;1].

Clinical features may apparent in early infancy but often escapes detection until adult life. HS presents as anemia,jaundice and splenomegaly and older individuals develop bilirubin stones

and may present with cholecystitis as seen in our case. Hemolytic crisis often precipitated by viral infections and aplastic crises also noted in folic acid deficiency and parvoviruse B19 infections[1]. Haematological investigation usually show evidence of hemolysis which includes spherocytosis(round dense RBCs with loss of central pallor), polychromasia ,nucleated RBCs, reticulocytosis and indirect hyperbilirubinemia. Mean corpuscular hemoglobin concentration (MCHC) usually high (35-38g/dl). MCV and MCH apperas normal or mild elevated. Liver function test show unconjugated hyperbilirubinemia and normal enzymes. In osmotic fragility test,exposure to hypotonic saline causes RBCs to swell and spherocytes are prone for lysis compare to normal biconvex RBCs. However increased osmotic fragility, negative direct coombs test, increased MCHC and peripheral blood smear spherocytosis makes diagnosis of HS and excludes other hemolytic anemia. A rapid flow analysis of eosin-maleimide (EMA) bound to erythrocytes is used as atest for HS and membrane protein deficiency. But this is expensive and not readily available [2].

Medical management and surgical option depends on severity of HS.Medical management includes folic acid supplemenation due to increased erythropoiesis chances of folic acid deficiency are more. Blood transfusion is an optional,depending on Hb concentration. Splenectomy isprincipal form of treatment with cholecystectomy if gall bladder stone are present, as seen in our case. Splenectomy helps to increase the level of hemoglobin, though microspherocytes formed in rest of reticuloendothelial system [5;1]

Conclusion:

To conclude, HS is rare form of hemolytic anemia and can be present at any age. Recurrent abdominal pain,gall stone and splenomegaly in early adult life should be consider for HS. As splenectomy is definitive treatment, early diagnosis helps in hematological or aplastic crisis and complication related after splenectomy.

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