Case Report

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A Case of Hereditary Spherocytosis with Associated Hypersplenism

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Abstract

Hereditary spherocytosis is a less common blood disorder where the cytoskeleton of red blood cell is defective due to mutation in their structural proteins and consequently the cell becomes spherical and fragile and destructed earlier than their usual life span. The case was a 25 year old male who had been suffering from continuous lethargy, weakness and abdominal pain for 2 years. After thorough clinical and laboratory exams he had spherical red blood cells in peripheral blood film and elevated fragile osmotic test, therefore he diagnosed with hereditary spherocytosis. Hereditary spherocytosis an infrequent red blood cell disorder where the cells become round and easily destructed and osmotic fragile test helps for confirmed diagnosis.

Keywords: Hereditary spherocytosis; Hemolytic anemia, RBC; Spectrin; Ankyrin

Introduction

Hereditary spherocytosis (HS) is the commonest hereditary abnormalities of erythrocyte's cell membrane. In this condition, red blood cell (RBC) becomes spherical and loses its flexibility. The most common manifestation is hemolytic anemia, however, in severe condition it leads to splenomegaly, and jaundice. For the first time, HS was explained in 1871 and now its prevalence is 1/5000 Americans while its prevalence in China was projected to be 1.27 cases in each 100,000 males and 1.49 cases in each 100,000 females [1,2]. Meanwhile, in 75% of cases it is inherited in autosomal dominant manner and the remaining cases are either due to new mutations or recessive fashion [2]. The exact prevalence in Afghanistan is not known and its diagnosis is still demanding this country. We expect that this report would raise awareness among Afghan medical society to diagnose this condition earlier and treat it correctly.

Case Report

A 25 year old male patient had been admitted in Badakhshan provincial hospital, a remote and less accessible area of northern Afghanistan one and half months ago. He was transfused one unit of blood and referred to Kabul, capital city of Afghanistan for further investigation and finally he was admitted in Ali Abad university hospital at 2 April 2021. He has been complaining of progressive fatigue, weakness and abdominal pain for 2 years which was located in right upper quadrant and epigastric areas and radiating to the right side of neck. In physical examination, he had pale conjunctiva, yellow sclera, splenomegaly measured 5 finger tips below the costal margins, hepatomegaly, right upper quadrant tenderness and systolic murmur on heart auscultation. For definitive diagnosis we ordered a series of laboratory and imaging investigations where the hemoglobin concentration was 8.2 gr/dl, platelets account was 100,000 per microliter, leukocytes count was 2900 per microliter, mean corpuscular volume (MCV) was 77.7 fl, mean corpuscular hemoglobin (MCH) was 28.5 pg and mean corpuscular hemoglobin concentration (MCHC) was 36.6 g/dl, red cell distribution width (RDW) was 16.8%. Peripheral blood film showed mild anisocytosis and spherocytes (**Figure 1**).

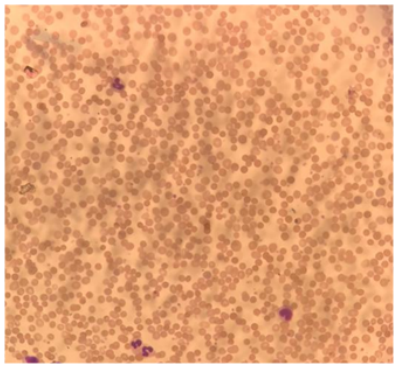

Figure 1 Microscopic image of peripheral smear.

Direct coombs test was negative and bone marrow aspiration revealed increased erythropoiesis, normal cellularity, myelopoiesis, megakaryocytes, lymphocytes, plasma cells. Abdominal sonography showed splenomegaly 19 cm, hepatomegaly, and multiple stones in gallbladder and echocardiography study was normal. Based on the clinical feature, laboratory and imaging studies the diagnosis of hereditary spherocytosis and associated

hypersplenism was made. The patient treated with folic acid supplementation and pneumococcal, meningococcal and H influenza vaccination was administered. Then, he was referred to surgery for elective splenectomy and possibly cholecystectomy.

Discussion

Hereditary spherocytosis a familial hemolytic anemia. In majority of cases the mode of inheritance is autosomal dominant but around one quarter of cases are inherited as autosomal recessive in which clear family history may not be found. In HS, there is defect in the cytoskeleton of red cell's membrane causing red cell's membrane fragile. Because of the latter effect some small vesicles separate from the RBCs membranes leading to spherical shaped (spherocytes) red blood cell which is more fragile and rapidly destroyed in spleen (extravascular hemolysis). Hereditary spherocytosis is caused by mutation in erythrocyte's membrane proteins such as α-spectrin, β-spectrin, ankyrin, band 3 or protein 4.2, where, the spectrin deficiency is the most common form amongst these proteins (**Figure 2**) [3,4]. The major proteins in the cell membrane of RBS are α-spectrin (band 1), β-spectrin (band 2), ankyrin (band 2.1), band 4.1, band 4.2 and actin (band 5) which all of them involved in cytoskeleton, band 3 as bicarbonate transporter, band 4.5 (GLUT-1, a glucose transporter), glyceraldehyde 3 phoshate dehydrogenase (band 6), an enzyme of glycolysis and stomatin (band 7) [5]. HS presents with anemia, jaundice, splenomegaly and pigmented gall bladder stones in laboratory examination the hemoglobin concentration is low, reticulocytes count is increased and indirect bilirubin is also raises. Blood film show spherocytes RBCs without central paleness. Osmotic fragility test, flow cytometry for eosin 5 maleimide binding test and osmotic gradient ektacytometry diagnosis.

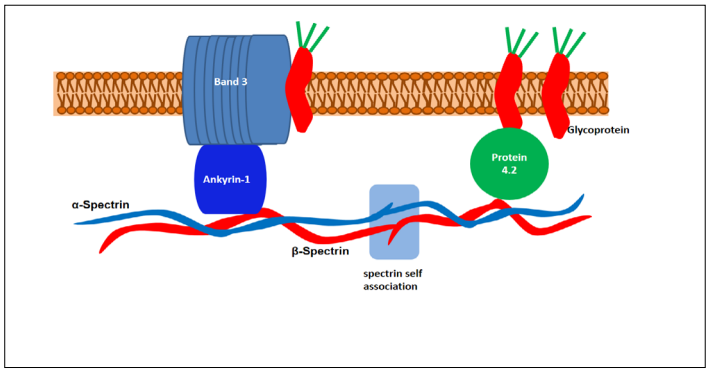

Figure 2 Major RBC membrane proteins which their mutation causes of hereditary spherocytosis.

Conclusion

Hereditary spherocytosis is infrequent blood disorder which is rarely diagnosed in our country because of lack of access to contemporary diagnostic equipment. Clinical manifestation, thorough physical examination and laboratory exams are necessary to diagnose this condition.

Competing Interest

We assert that there is not any competing interest in this case report.

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