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Research Article

**HEREDITARY SPEROCYTOSIS WITH AN ATYPICAL  
PRESENTATION****<sup>1</sup>Dr. Waqas Iqbal, <sup>2</sup>Dr. Ahmad raza, <sup>3</sup>Dr. Maryam Shafique**<sup>1</sup>House Officer Mayo Hospital<sup>2</sup> Medical Officer DHQ Hospital Nankana Sahib<sup>3</sup>Rawlapindi Medical College**Abstract:**

*Hereditary Spherocytosis (HS) is a genetic disorder of RBC membrane proteins; resulting RBCs are spherical in shape. They get distorted passing through capillaries, this difference in shape makes the red blood cells more prone to rupture[\*] , leading to hemolysis. It is characterized by anemia, jaundice and splenomegaly. HS varies in severity from being asymptomatic to causing severe hemolytic anemia. We present here a case of a 22 year old female diagnosed with HS. Patient was undiagnosed for last 7 years. She had no specific symptoms except weakness and lethargy. There was no clinical sign appreciable except pallor yet the laboratory studies suggested it to be HS.*

**Key Words:** *Hereditary Spherocytosis (HS), Hemolytic Anemia, Red Blood Cells (RBCs).*

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**INTRODUCTION:**

Hereditary Spherocytosis (HS) is a familial haemolytic disorder with marked heterogeneity. Clinical features range from being asymptomatic to fulminant haemolytic anaemia. The condition was first described in 1871 (1). HS is the commonest cause of inherited chronic haemolysis in Northern Europe and North America, with a reported incidence of 1 in 5,000 births (2). In 80% of the instances, the inheritance of HS is autosomal dominant and in others autosomal recessive (3) In autosomal dominant form, the deficiency is mild, and hence, the anaemia is mild while in the recessive form, the deficiency is greater, and the anaemia is profound. (4) Patient may remain undiagnosed for years; the more severe the disease the more earlier the presentation. At this point, there exists no cure for the genetic defect that causes hereditary spherocytosis.[5] Acute symptoms of anemia and hyperbilirubinemia indicate treatment with blood transfusions or exchanges and chronic symptoms of anemia and an enlarged spleen indicate dietary supplementation of folic acid and splenectomy,[6] the surgical removal of the spleen. Splenectomy is indicated for moderate to severe cases, but not mild cases.[7].

**CASE SUMMARY:**

A 22 year old female presented to Mayo Hospital, Lahore with complaints of weakness and fatigue and lethargy. Patient was having these symptoms for the almost 7 years. These were gradual in onset, slowly progressing more marked with exertion and relieved to some extent with rest. These were not associated with yellow discoloration of eyes, pruritis, palpitation or pedal edema. Her parents were non-consanguineous and there was no history of any blood disorder in family. There was no significant finding on examination (jaundice, splenomegaly) but a mild pallor. Her CBC showed Hb 11.0, MCV 79 and MCHC 37. ESR was 66. Bilirubin was 1.5 with conjugated being 0.5 n unconjugated fraction was 1.0. Serum LDH level was 305. All these labs were favoring hemolytic anemia. Abdominal scan was unremarkable. Both direct and indirect coombs test were negative. RBC morphology showed presence of spherocytes as well as reticulocytes (11.7%). At the end osmotic fragility test was prescribed that showed increased osmotic fragility with 50% lysis at NaCl concentration of 7-7.5g/l, thus confirming spherocytosis. Since no cure exists for HS so symptomatic treatment was given and patient condition was improved. Folic acid along with a multivitamin was given to patient and she was followed up for 6 months. Her symptoms were relieved and neither required transfusion nor

surgery.

**DISCUSSION:**

On basis of all the laboratory investigations done HS was confirmed to be the cause of anemia in the reported case. Apparently there was no clue about it from the symptoms of patient. Symptoms were not favouring hemolytic anaemia which led the patient undiagnosed for 7 years. Considering hemolytic differentials especially HS which have a range of presentations would help avoid undiagnosed condition in future. An osmotic fragility test can aid in the diagnosis.[8] In this test, the spherocytes will rupture in liquid solutions less concentrated than the inside of the red blood cell. This is due to increased permeability of the spherocyte membrane to salt and water, which enters the concentrated inner environment of the RBC and leads to its rupture.[9] Flow cytometric analysis of eosin-5'-maleimide-labeled intact red blood cells and the acidified glycerol lysis test are two additional options to aid diagnosis.[10] Whenever patient presents with non specific sign and symptoms of anaemia with no hemolytic picture, HS should be kept in mind as a differential diagnosis and a simple osmotic fragility test should be advised.

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