Hereditary Elliptocytosis - A Case Report

Nanda J Patil¹, Shrutika D Dhawan², Avinash Mane³, Manish Barnawal²

¹Professor, ²Tutor, ³Lecturer,
Department of Pathology, Krishna Institute of Medical Sciences, Karad, Maharashtra.

Corresponding Author: Shrutika D Dhawan

Received: 01/08/2015   Revised: 20/08/2015   Accepted: 25/08/2015

ABSTRACT

Hereditary elliptocytosis is a disorder of red cell membrane which results from mutation in genes encoding various red cell membrane and skeletal proteins. Peripheral smear of these patients reveal elliptical RBCs. Usually the patients are asymptomatic and rarely present with hemolysis. We report a case of hereditary elliptocytosis in a 16 year old female patient diagnosed accidentally when she was admitted for dengue fever.

Key Words: Hereditary elliptocytosis, red cell membrane disorder.

INTRODUCTION

Hereditary elliptocytosis is one of the membrane disorders of red cell membrane where red blood cells show elliptical shape instead of biconcave disc shape. Patients present with anemia or hemolytic episodes. We present a case of hereditary elliptocytosis in a 16 year old female patient who was admitted for dengue infection.

CASE HISTORY

A 16 year female, presented with fever with chills and generalised bodyache for which she was admitted in medical ward. Complete blood count revealed mild anemia (Hb- 11gm/dl). Red cell indices were within normal range and platelet count was reduced (30,000/mm³). Peripheral smear revealed elliptical red cells which were more than 90% (Fig 1) with evidence of polychromasia and occasional nucleated RBC. Differential count revealed lymphocytosis. Ultrasonography showed mild hepatosplenomegaly while serological test for dengue was positive.

Fig 1. Photomicrograph of peripheral blood smear showing elliptical red cells (> 90%) (Leishman,400X).
DISCUSSION

Hereditary elliptocytosis is a red cell membrane disorder which results from mutations in various membrane or skeletal proteins and leads to decreased red cell deformability, reduced life span of red cells and their premature removal from the circulation. The disorder is autosomal dominant and both sexes are equally affected. [1] Elliptocyte was first introduced by Dresbach in 1904. Elliptocytosis was first recognised as hereditary condition in 1932 by Huaer. The incidence is 1 in 5000 to 10000. The condition is common in Africa and mediterranean areas. [2]

Hereditary elliptocytosis is usually discovered as an incidental finding during blood smear examination. In these cases red blood cells are elliptical instead of routine biconcave disc. Presence of more than 25 % elliptocytes is generally considered as a requirement for diagnosing the condition. [3] The cases of hereditary elliptocytosis fall under these categories:

1. The cases with no signs of hemolysis.
2. The cases having compensated hemolysis.
3. The cases with non-compensated hemolysis. [4]

Though majority of cases are asymptomatic, 10-12 % of cases reveal hemolysis. [1,5]

The disorder presents with a wide clinical features. The patients may be asymptomatic or reveal vague symptoms like malaise, abdominal pain as in our case. Few patients present with moderate to severe anemia and are prone to usual complications of chronic hemolysis like cholelithiasis. [6,7] Overt hemolysis may manifest first time with stressful conditions like surgical procedures and infections. Patients with significant hemolysis have splenomegaly. [6] Patients with non-compensated hemolysis are transfusion dependent.

Diagnosis of hereditary elliptocytosis mainly depends on RBC morphology, that is, elliptical RBCs on peripheral blood smear, panel of osmotic fragility test and eosin -5-maleinide (EMA) binding is a flow cytometric method. [8,9] We made diagnosis with RBC morphology.

Treatment of these cases is based on supportive measures like folate therapy for moderate to severe haemolytic anemia. Severely anemic patients may be transfusion dependent. [1] Splenectomy is indicated for severe form of elliptocytosis after 5 years of age which increases the red cell life span and haemoglobin levels. [10]

Our case was diagnosed incidentally during the episode of dengue fever with evidence of hemolysis. Thrombocytopenia and lymphocytosis were consequence of dengue fever.

CONCLUSION

Hereditary elliptocytosis is mostly asymptomatic, rarely presenting with hemolysis.

Microscopic examination of the blood film plays an important role in the diagnosis. The case is presented to highlight the wide spectrum of clinical presentation and severity of disease.

REFERENCES


How to cite this article: Patil NJ, Dhawan SD, Mane A et al. Hereditary elliptocytosis – a case report. Int J Health Sci Res. 2015; 5(9):610-612.