

A Case of Hereditary Elliptocytosis

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Here are three images from peripheral blood smear from a patient with Hereditary Elliptocytosis. The patient presented for a general check up when his CBC showed only mild anemia with slightly increased reticulocytes. The diagnosis was made incidentally when blood smear was examined.

Hereditary Elliptocytosis:

Hereditary Elliptocytosis (HE) is a congenital hemolytic disorder in which the red blood cells are either elongated (cigar or oval shape) or exhibit irregular degrees of poikilocytosis. This abnormality is a result of a defect in one of the skeletal proteins in the red blood cells membrane. It is transmitted as an autosomal dominant trait with at least 4 genetic loci implicated in the pathogenesis.

The elliptical erythrocyte form is acquired in the circulation and that is why the reticulocytes and bone marrow red blood precursors are normal in shape (1).

The true incidence is unknown because of it is asymptomatic in the majority of the cases, but generally it is estimated to be present in 1 per 2000-5000 individuals.(1) Morbidity ranges from asymptomatic cases most of the time to severe transfusion dependent disease, rare fatalities have been documented. While Hereditary Elliptocytosis has no sex predilection, it is more common in African and Mediterranean descents.

Clinical Findings:

Most patients with heterozygous (HE) are asymptomatic (common HE), however homozygous patients with Hereditary pyropoikilocytosis (HPP) may suffer from severe hemolytic anemia and may become transfusion dependent (2)(3). Stomatocytic Elliptocytosis is a benign disorder in which red blood cells have a broad and oval shape with a presence of stomatocytes occasionally (more common in Southeast Asia). The most common complications that patients with HE face are gallstones and hemolytic crisis and to a lesser extent, leg ulcers and/ or dermatitis. (3)

Laboratory findings:

The diagnosis is made based on the findings on the peripheral blood smears (elliptocytes)(1). Elliptocytes can be seen in other diseases like in megaloblastic and iron

deficiency anemias, however the elliptocytes in these conditions are less than 25% of the erythrocytes, in contrast to more than 25% in Hereditary Elliptocytosis Figs (1-2-3) and sometimes up to 60%. (1)

Controlled thermal stress test (not required to confirm the diagnosis) is also helpful. CBC findings of anemia (not common) with increased reticulocyte count (up to 4% may get higher in severe cases)(1) assist in making the diagnosis as well. In addition to osmotic fragility tests (normal) and DNA testings that are not required but can play important roles in confirming the diagnosis.

Treatment:

Treatment is not indicated except in severe cases where occasional transfusion might be required. (2) Daily Folate is recommended for patients with severe hemolysis. Splenectomy may be considered in patients with severe anemia and significant symptoms.

Key Words & Abbreviations: HE: Hereditary Elliptocytosis, HPP: Hereditary pyropoikilocytosis, hemolytic anemia

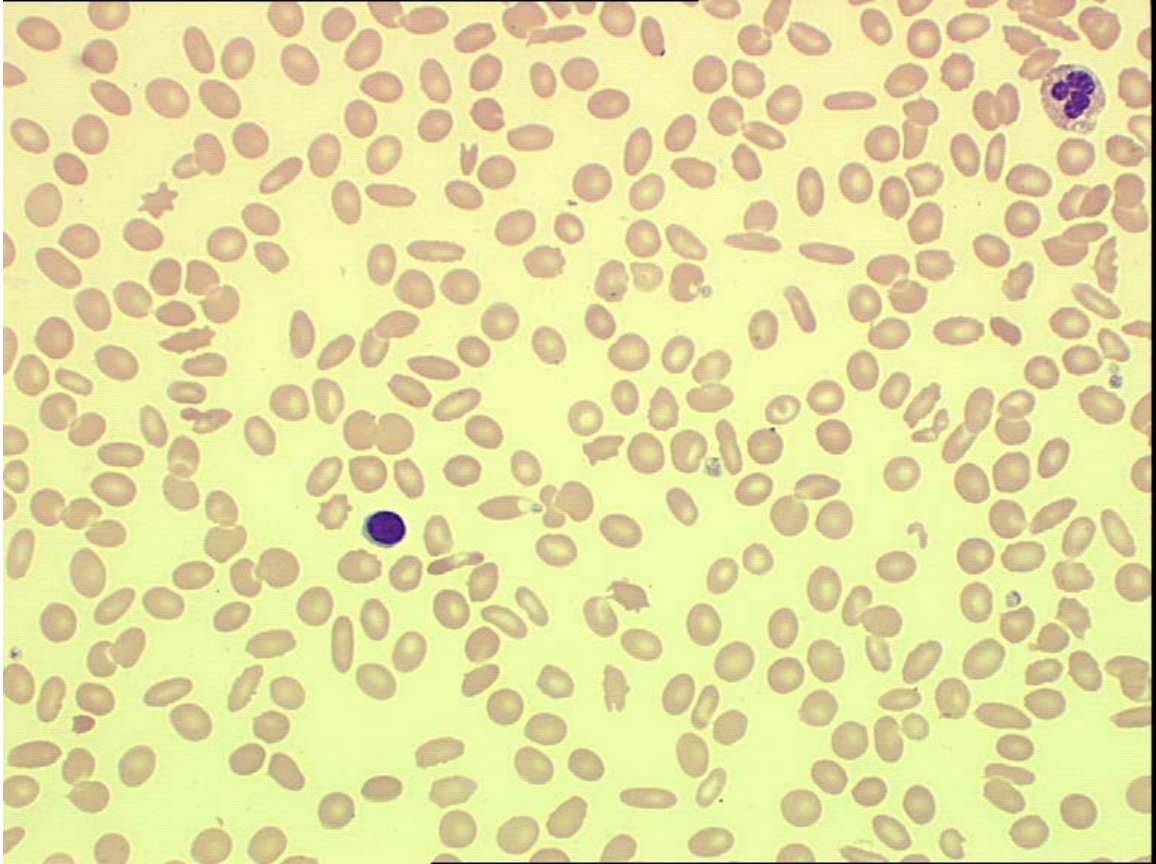


Fig (1): Peripheral Blood Smear with Elliptocytosis (Giemsa 500X)

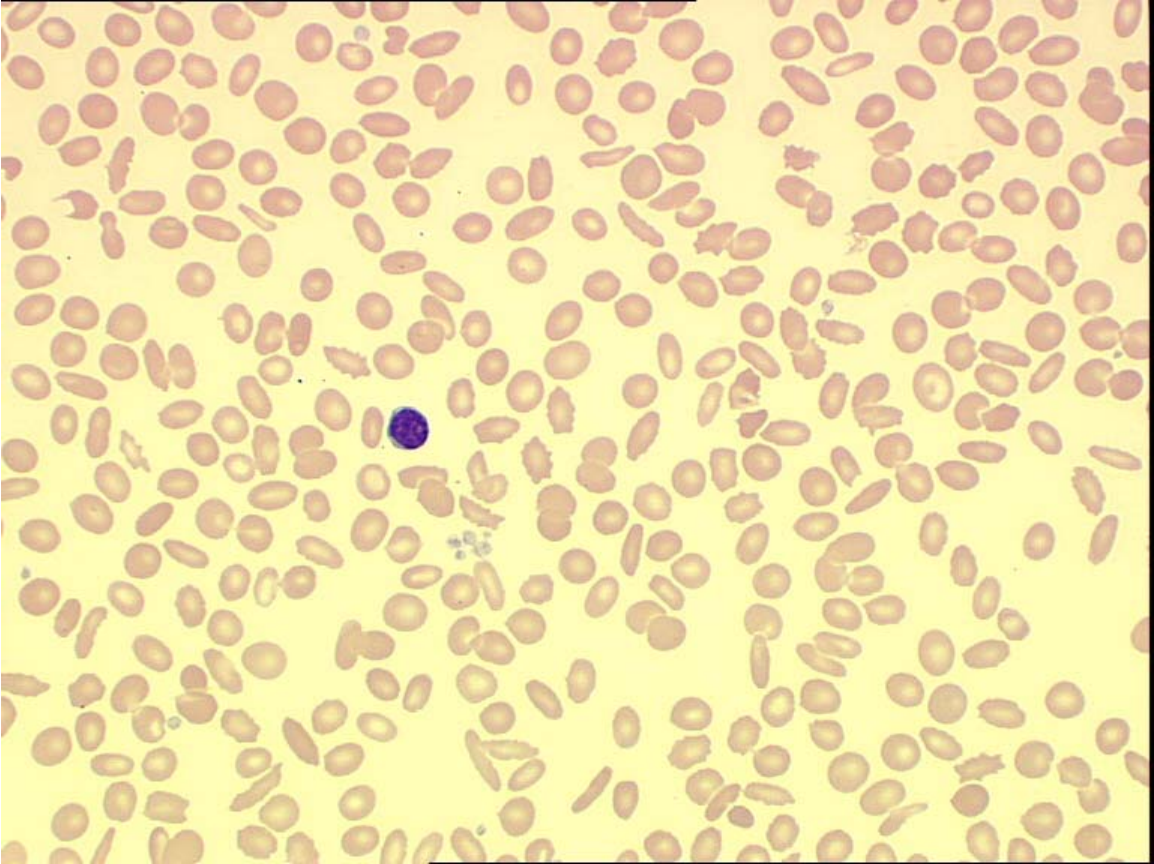


Fig (2): Peripheral Blood Smear with Elliptocytosis (Giemsa 500X)

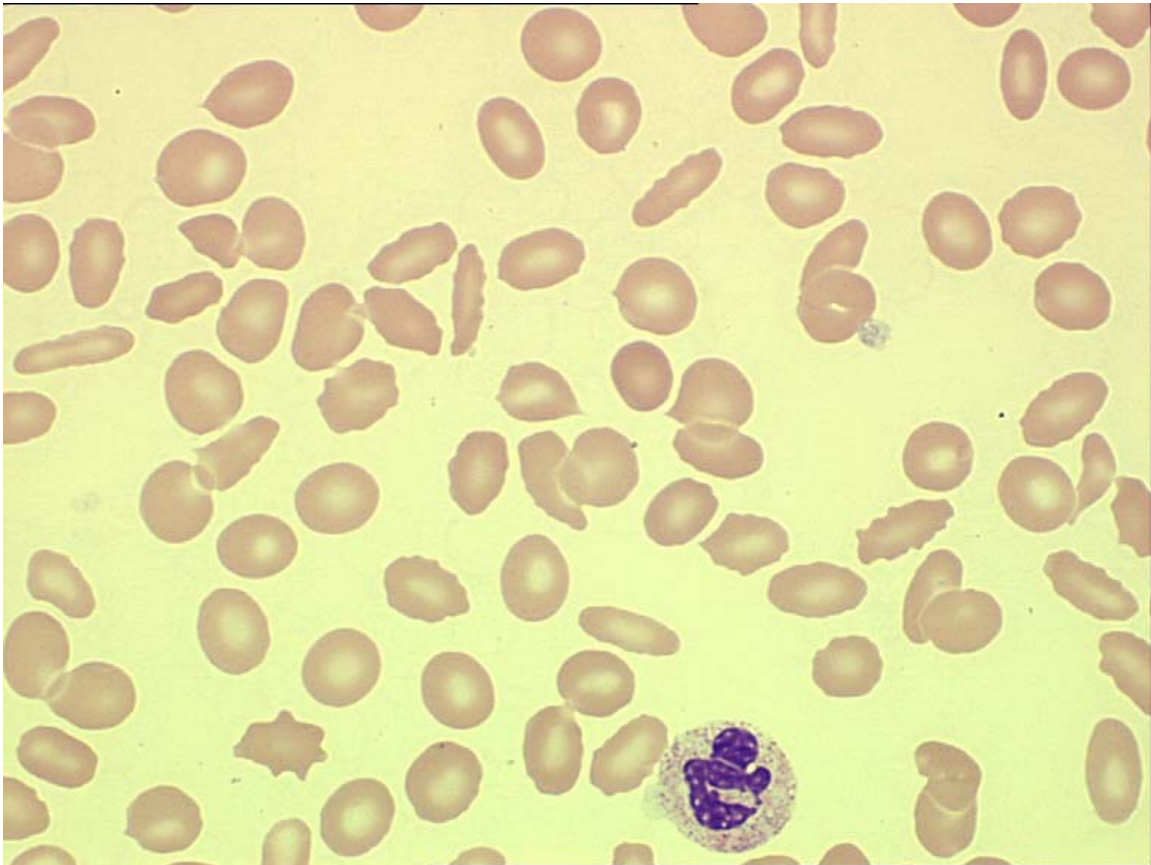


Fig (3): Peripheral Blood with Elliptocytosis (Giemsa 1000X)

References:

1-Shirlyn B McKenzie, Hematology, Second Edition. Hemolytic Anemia Caused by Intrinsic erythrocyte Defects. 1996: 227-229.

2-Mandy Meck, eMedicine, Hereditary Elliptocytosis and Related Disorders. <http://www.emedicine.com/ped/topic987.htm>

3-Ronald Hoffman...[et al.], 3rd Edition. Hematology, Basic Principles and Practice. Red Blood Cells Disorders, Chapter 33. 2000: 587-595