Hereditary Elliptocytosis: An Incidental Finding in a 6-year-old Child

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ABSTRACT
A case of hereditary elliptocytosis (HE) in a 6-year-old child diagnosed as an incidental finding has been presented.

Keywords: Hereditary elliptocytosis, Peripheral smear, Sickle cell disease, Thalassemia.


INTRODUCTION
Hereditary elliptocytosis (HE) is an uncommon disorder often picked up as an incidental finding on routine blood film examination. The estimated frequency of this disorder worldwide is 1 in 2000 to 1 in 4000. Here is a case of HE in a child diagnosed as an incidental finding, which has been discussed.

CASE REPORT
A 6-year-old boy presented with fever and diarrhea for 2 days. A routine complete blood count (CBC) and peripheral smear (PS) for red cell morphology and DC was performed. Hemoglobin (Hb) was 12.7 g/dL, mean corpuscular volume (MCV) = 79.6 fl, mean corpuscular hemoglobin (MCH) = 28.2 pg, mean corpuscular hemoglobin concentration (MCHC) = 35.5 g/dL and red cell distribution width = 14.2%. Peripheral smear showed numerous elliptocytes constituting predominant RBC population (90%) with very occasional poikilocytes (Fig. 1). No spherocyte was seen. Reticulocyte count was 5%. Possibility of HE was considered. The serum bilirubin performed subsequently was 0.7 mg/dL. The patient gave history of jaundice 2 years ago. However, no documentation was available. The

Fig. 1: Peripheral smear showed numerous elliptocytes constituting the predominant RBC population (90%) [Wright stain, 400x]

CBC and PS for both the parents were performed which revealed presence of elliptocytes in the father constituting the predominant cell population (90%). The CBC and PS of the mother were within normal limits.

DISCUSSION
Elliptocytes are RBCs with an oval shape. In HE stained smear, the defect in stability is due to failure of proper spectrin dimer–tetramer association. There are four molecular defects which can cause this: (1) Abnormalities in the alpha-chain of spectrin, (2) abnormalities in the B-chain of spectrin, (3) defective protein 4, and (4) defective protein 3.

Hereditary elliptocyte is more common in African and Mediterranean regions. It is transmitted as an autosomal dominant trait, except for a rare Melanesian variant, which has an autosomal recessive mode of inheritance. De novo mutations have been reported in rare cases.

Symptoms of HE vary between members of the same family and in the same individual over time. Most of the patients have a fully compensated hemolytic process with no anemia or splenomegaly. However, the blood film shows 25 to 100% elliptocytes with mild reticulocytosis. 5 to 20% of the patients may present with mild anemia, high reticulocyte count, splenomegaly, gall stones, or chronic leg ulcers. Rarely, crises may occur in these patients secondary to stress or infections. Typically, 50 to 90% (At least 25%) of the RBCs are oval shaped and 10% occur as elongated rod-shaped forms. However, the
degree of hemolysis does not correlate with the number of elliptocytes present.\(^4\) In the instant case, elliptocytes were seen in the PS.

Both autohemolysis and osmotic fragility are normal in nonhemolytic HE. They are increased in patients with spherocytic elliptocytosis and in patients with HE variants and hemolytic disease. Demonstration of the molecular defect in spectrin molecule confirms the diagnosis, however, the required methods for isolation, enzymatic digestion, and electrophoretic analysis are not routinely available in most laboratories.\(^2\)

In addition to the aforementioned tests, patient’s clinical history, characteristics of RBC morphology, and family history are of prime importance in making the diagnosis of HE.\(^2\) In the index case, elliptocytosis was found as an incidental finding in a routine CBC performed for other illness and there was no evidence of hemolysis.

Other causes of elliptocytes in the blood are iron deficiency anemia, macrocytic anemia, sickle cell disease, and thalassemia.\(^2\) The percentage of elliptocytes in these disorders are usually less than 25%, while in HE, they are 90% or more. In the present case, percentage of elliptocytes was found to be 90%.

**CONCLUSION**

Elliptocytosis was discovered as an incidental finding in the patient on routine CBC sent on hospital admission, which was confirmed on positive family study.

**CLINICAL SIGNIFICANCE**

Hereditary elliptocytosis is a rare disease, especially in the Indian population. Family study is advised whenever the pathologist comes across elliptocytes as the predominant cell population.

**REFERENCES**