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Case Report

CASE REPORT ON HEREDITARY ELLIPTOCYTOSIS

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ABSTRACT
Hereditary elliptocytosis is a condition where the RBC's in the peripheral blood is elliptical in shape. Usually the patients are asymptomatic and occasionally present with hemolysis. In this we describe the details of two cases where one patient presented with haemolytic anemia at very early age of life and the other presented in the adulthood. The first case was transfusion dependent and the second case did not required transfusion. The peripheral smear of both patients showed elliptocytes.

Key Words: *Elliptocytes, Hemolysis, Transfusion, Anemia*

INTRODUCTION
Hereditary elliptocytosis is a condition where the red blood cells are elliptical in shape rather than biconcave disc shape. It is one of the membrane disorders of red blood cell which predisposes to haemolytic anemia. The incidence and the severity of hemolysis differ depending on the type of elliptocytosis and the agent causing hemolysis.

CASES
Case 1
A 17 year old male presented to our hospital with history of fever, bilious vomiting and diarrhoea for 3 days. Past History revealed the presence of icterus since childhood. Patient had noticed worsening of icterus and tiredness since 1 week. He was admitted to the hospital and investigated with routine tests. Peripheral smear revealed more than 40% elliptocytes. Sickling test was done to rule out sickle cell disease and was found to be negative. On further going into the details of the history patient's mother informed that at the age of 2 months, he was diagnosed to have elliptocytosis incidentally. It was also noted that the severity of icterus increased with infections. The patient received transfusion once in two years then and the frequency of blood transfusion decreased as he grew older. In 2001 patient had an attack of parvovirus infection following which he had an aplastic crisis. In 2008 patient had obstructive jaundice due to cholelithiasis for which he underwent cholecystectomy. Presently, the patient was admitted for fever, vomiting and increasing jaundice. On examination, patient was icteric, with dry oral mucosa and had hepatosplenomegaly. Laboratory investigations included complete haemogram, peripheral smear and liver function test.
Peripheral smear showed 40% elliptocytes. Sickling test was negative, thus ruling out sickle cell disorder. Osmotic fragility test was performed to rule out spherocytosis. Liver function test showed increased levels of both conjugated and unconjugated bilirubin. Since his Haemoglobin levels were low he received packed cell transfusion.
This case is presented because of the rarity of the disorder by itself and the case being symptomatic. Hereditary elliptocytosis is asymptomatic in about 90% of the cases. This patient is symptomatic in that patient has been having jaundice since childhood which worsened with episodes of infection and the presence of anemia necessitating packed cell transfusion.
Case 2
A 23 years old male patient with complaints of jaundice since 6 months. Routine investigations revealed unconjugated hyperbilirubinemia. Peripheral smear revealed the presence of 40% elliptocytes. His haemoglobin was maintained at the lower limit of normal range and WBC & platelets were within normal limits.

Case Report

This case is presented because of its presentation as unconjugated hyperbilirubinemia. The disease by itself is rare and this case presented with clinically detectable level of jaundice.

DISCUSSION

Elliptocytosis was first described by Dresbach in the year 1904 and first recognised as a hereditary condition in the year 1932 by Hunter. The incidence of hereditary elliptocytosis is hard to determine because more than 90% of the people with elliptocytosis are asymptomatic (Hoffman, 2000). These categories of cases never come to medical attention. The incidence is 1 in 5000. HE is common in people of Africa and Mediterranean region (Ravindranath, 1994). Since elliptocytosis are resistant to malaria, some subtypes of elliptocytosis are more prevalent in areas where malaria is endemic (Gallagher, 2004).

The inheritance of hereditary elliptocytosis is autosomal dominant and both sexes are equally affected. The most important exception is pyropoikilocytosis where the inheritance is autosomal recessive. On the basis of red cell morphology HE can be classified into three types of elliptocytosis. Common hereditary elliptocytosis, spherocytic elliptocytosis and south East Asian elliptocytosis (Hoffbrand *et al.*, 2011).

Most common form of elliptocytosis is common hereditary elliptocytosis, which is extensively studied and researched. 5-10% cases present with haemolytic anemia. The most common defect in the common hereditary elliptocytosis is mutation in α or β chains (Conby, 1991).

In this study the first case was presented as haemolytic anemia at early infancy requiring frequent transfusions. A case of hereditary elliptocytosis in a day old baby with severe jaundice was reported by Morioko Ichiro *et al.*, from Japan (Morioka *et al.*, 2000). This baby presented with severe unconjugated hyperbilirubinimia underwent exchange transfusion and phototherapy. The RBC morphology at the one day showed haemolytic blood picture with elliptocytes. The baby's mother was diagnosed as hereditary elliptocytosis with the electron microscopic studies and the infant was also diagnosed similarly at the age of 3 months.

The second case presented with jaundice and severity of jaundice decreased when the patient was treated with hepatic enzyme inducing drugs like phenobarbitone. Similar case report was done by Radlovic *et al.*, (2011) a 15 years old boy presented with jaundice (unconjugated hyperbilirubinemia) without anaemia. Patient had a previous history of similar episode and the peripheral smear showed elliptocytosis and mother had similar finding in the blood (Redlovic, 2011). Another case of a one year old child evaluated for fever, was found to have elliptocytosis with neutropenia and thrombocytopenia. On further evaluation of this child at the age of two years revealed partial albinism, otitis media and enlarged liver and spleen. At this time the peripheral smear showed elliptocytosis with giant granulation of neutrophils, monocytes and lymphocytes. The case was diagnosed as hereditary elliptocytosis with Chediak-Higashi syndrome (Islam, 2001).

Conclusion

Hereditary elliptocytosis is mostly asymptomatic with rare presentation as haemolytic anemia. Both the cases are presented to highlight the varied clinical presentation and severity of the disease entity. The first case manifested as haemolytic anemia requiring frequent blood transfusion and was diagnosed at very young age. The second case presented with jaundice but not associated with anemia.

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