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Hereditary Elliptocytosis - Case Report of a Rare Entity

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Abstract: ***Hereditary elliptocytosis is a rare hemolytic disorder characterized by the presence of elliptical red blood cells. It has heterogenous clinical presentation ranging from asymptomatic carriers to severe hemolytic anemia. Majority of the cases are asymptomatic and are incidentally diagnosed on routine blood examinations. Here we present a case of incidentally diagnosed hereditary elliptocytosis because of its rarity. A 25 year old married female presented with chronic headache to the out patient department with the clinical diagnosis of migraine. Routine peripheral smear examination revealed elliptical red blood cells accounting to 70 %. family screening was done and the mother of the patient revealed elliptocytes in her peripheral smear (60%)and hence the diagnosis of hereditary elliptocytosis was made.***

Keywords: Hereditary elliptocytosis, incidental, elliptocytes

1. Introduction

Hereditary elliptocytosis is an uncommonly inherited disorder characterized by elliptically shaped red blood cells rather than the usual biconcave disc. In 1904 elliptocytosis was first described by Dresbach. Later Hunter established the heritability of elliptocytosis. Hereditary elliptocytosis is commonly seen in African and Mediterranean ethnics. It is much more common in malaria endemics. Hereditary Elliptocytosis has broad spectrum of clinical presentation ranging from an asymptomatic trait with no hemolysis to life threatening severe hemolytic anemia. Because of this varying clinical presentation, the true incidence of Hereditary Elliptocytosis is unknown worldwide.

2. Case Report

A 25 year old female attended the general out patient department with the complaints of on and off headache, generalized weakness, dyspnea and easy fatigue ability. General examination revealed only pallor. No abnormalities were deducted in systemic examination. Routine basic blood investigations were done. **peripheral smear study**:. RBC: normocytic normochromic red cells admixed with elliptocytes>80%No evidence of hemolysis. Wbc : normal in count, morphology and distribution. Platelet: adequate Impression: Features Favour Elliptocytosis (figure 1)

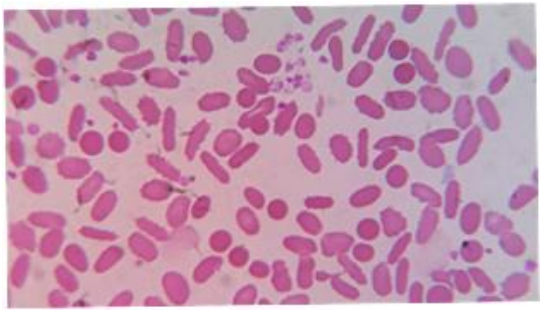

Figure 1: Peripheral smear of the patient

Osmotic fragility test is normal.

Family Screening: Peripheral smear of mother : RBC : normocytic normochromic red cells admixed with elliptocytes>60%. Wbc : normal in count, morphology and distribution. Platelet: adequate

Impression: Features Favour Elliptocytosis (figure 2)

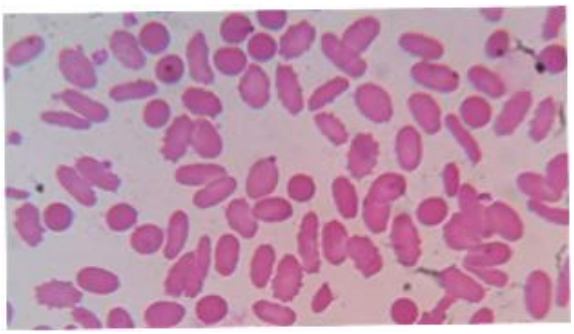

Figure 2: Peripheral smear of the mother

3. Discussion

Ellipticytosis comprises of 3 major categories based on RBC morphology.

1) Common Hereditary Elliptocytosis
2) Spherocytic Hereditary Elliptocytosis
3) South east asian ovalocytosis[1]

In common Hereditary Elliptocytosis, the red cells are characterized by biconcave elliptocytes, rarely rod shaped. A less common condition, Spherocytic Hereditary Elliptocytosis also known as hemolytic ovalocytes, is characterized by round ovalocytes and spherocytes. In South east Asian ovalocytosis red cells are spoon shaped and rigid.

Inheritance: In majority of the cases, Hereditary Elliptocytosis shows autosomal dominant pattern of

inheritance. In few occasional cases autosomal recessive pattern is also noted.

Clinical Manifestation: Hereditary Elliptocytosis is often diagnosed accidentally on routine Peripheral Smear examination if the subjects are asymptomatic carriers. There will be no evidence of hemolysis with normal red cell life span. Such cases may show molecular defects in alpha – spectrin, /beta spectrin/partial deficiency of 4.1 Protein.

Lab Manifestation: Peripheral Smear, Osmotic Fragility Test forms the 2 basic routine investigations for identifying and categorizing Hereditary Elliptocytosis. In mild asymptomatic cases, Peripheral Smear shows elliptocytes and normal value in osmotic fragility test.

Differential Diagnosis: Differential Diagnosis for elliptocytosis includes Iron deficiency anemia, thalasemmia, megaloblastic anemia, myelofibrosisand pyruvate kinase deficiency. In all these conditions the percentage of elliptocytes is not greater than 60%. But the most reliable feature of Hereditary Elliptocytosis is a positive family history and not the percentage of elliptocytes in the peripheral smear examination.(2)

4. Prognosis

Asymptomatic Hereditary Elliptocytosis cases show good prognosis with minimal clinical manifestation. Follow up of the cases is essential. If there is any evidence of hemolysis, splenectomy helps in such situation.

5. Conclusion

We present this case of Hereditary Elliptocytosis to emphasis the significance of routine peripheral smear examination in identifying the rare hematological disorders.

References

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