A CASE REPORT OF SEVERE HEMOLYTIC ANAEMIA WITH ELLIPTOCYTOSIS

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INTRODUCTION

Hereditary elliptocytosis is a rare, autosomal dominant haematological disorder[1]. It is a hemolytic anaemia due to membrane defect, which produces a broad spectrum of disease that ranges from fully compensated hemolysis to life threatening anaemia. Red cells with uniform elliptical shape come under the category of hereditary elliptocytosis (HE) however homozygous or compound heterozygous for HE present with distorted morphology termed as hereditary pyeropoikilocytosis (HPP)[2].

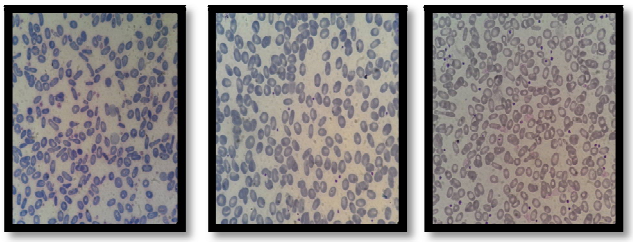
We reported a case of HE with associated beta thalassaemia trait due to its rarity and severe presentation.

CASE REPORT

A male 7 months of age, resident of Azad Kashmir, presented with complaint of progressive pallor since 3 months of age, high grade fever and cough for the last 7 days. He was referred to haematology dept. Armed Forces Institute of Pathology to find out the cause of anaemia. He had a past history of mild jaundice off and on. He was a offering of a consanguineous marriage and his elder brotherhad died at the age of 6 months due to anaemia and recurrent infections, however parents had no complaints. Patient was transfused 1 unit RCC 2 months back. There was no known family history of thalassaemia.

On examination, the child was moderately pale and icteric. His vital signs were within normal limits. On abdominal examination liver was palpable 2 cm below right costal margin and spleen tip was palpable. Rest of the systemic examination was unremarkable.

On haematological examination his hemoglobin (Hb) was 6.1g/l with MCV 58.4 fl and MCH 17.4 pg, total leucocyte count (TLC) was $11.2x10^9$/l; differential leucocytes count: neutrophils 34%, lymphocytes 60%, monocytes 05% and eosinophil 01%. Peripheral smear showed elliptocytes which were more than 50%, there was marked anisocytosis, poikilocytosis and red cell fragmentation (fig-1a). Reticulocyte count was 20%. Haemoglobin studies were done which showed Hb F 0.9%, Hb A 95.4% and Hb A_2 3.7%. Osmotic fragility was increased and thermal sensitivity testing was also done which showed increased haemolysis at temperature of 45^oC. Serum total bilirubin was 88 umol/l, indirect bilirubin was 82.7 umol/l and direct bilirubin was 5.37 umol/l. Serum ferritin was 83 ng/ml. Coombs test both direct and indirect were negative.



(a) (b) (c)

Figure-1: Blood film of (a) Child (b) Father (c) Mother.

Family study was done in which CBC of father showed normal counts, Hb 15.6 g/dl with MCV 85.5fl and MCH 29.8 pg. Peripheral smear showed marked elliptocytosis (fig-1b). CBC of mother showed low Hb 10.3 with MCV 60.4 fl and MCH 18.0 pg. Peripheral smear showed

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marked elliptocytosis with hypochromic microcytic blood picture (fig-1c). A haemoglobin study of mother was done which showed Hb F 0.9%, Hb A 95.2% and Hb A_2 3.9% and diagnosis of beta thalassaemia trait with heterozygous hereditary elliptocytosis was made. PCR was done further to identify and confirm presence of common beta thalassaemia mutation and found heterozygous Fr 8-9 mutation in child and motherand no mutation in father. On the basis of history, examination, laboratory investigations and family study final diagnosis of hereditary pyerpoikilocytosis with beta thalassemia trait was made in this child.

DISCUSSION

HE is a rare entity that sometimes present with other haemoglobin disorders like beta thalassaemia, sickle cell anaemia and Hb C. Its incidence is highest in west and central Africa but it is rare in our part of the world.

The disorder is caused by abnormality in horizontal links of the cytoskeleton, which in 80% of cases occur due to alpha spectrin deficiency[3]. In rest of the cases there is deficiency of protein 4.1, beta spectrin and rarely glycophorin C. Heterozygotes present with mild clinical picture with fully compensated hemolysis whereas homozygotes and compound heterozygotes like hereditary elliptocytosis in combination with beta thalassaemia trait present with severe hemolytic anaemia due to mutual enhancement of involved genes[4]. HE and HPP both can present in the same family. In neonates clinical and haematological picture is more severe due to increase level of Hb F as it increases the level of 2,3-DPG which destabilizes spectrin-actin interaction thus exacerbating the defect.

HE/HPP are identified by careful examination of blood film of patient and parents and as many first degree relatives as possible. Other acquired causes of elliptocytosis should be investigated which include iron deficiency anaemia, folate or B_{12} deficiency, thalassaemia, etc.

Most of the patient present with mild clinical picture and should be given folate supplements however few may present with severe picture and splenectomy is indicatedin these cases as it will increase red cell life span and reduce severity of anaemia[5]. Blood transfusions can be given to improve anaemia. In case of neonates condition improves once Hb F decreases and adult haemoglobin take over.

CONFLICT OF INTEREST

This study has no conflict of interest to declare by any author.

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