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ISSN: 0976-3031

International Journal of Recent Scientific Research
Vol. 6, Issue, 5, pp.3856-3858, May, 2015

International Journal of Recent Scientific Research

RESEARCH ARTICLE

A RARE CASE REPORT OF HEREDITARY ELLIPTOCYTOSIS WITH HEMOGLOBIN D TRAIT

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ARTICLE INFO

Article History:

Received 5th, April, 2015
Received in revised form 12th, April, 2015
Accepted 6th, May, 2015
Published online 28th, May, 2015

Key words:

Hereditary elliptocytosis, haemolytic anaemia, hemoglobinopathies, Hemoglobin D trait

ABSTRACT

Hereditary elliptocytosis is an autosomal dominant condition wherein the erythrocytes are cigar or oval shaped. It is a type of haemolytic anaemia caused due to defects in red cell membrane cytoskeletal proteins. We present a case of a patient with branchial cyst whose peripheral blood smear examination for anaemia revealed elliptocytes and a high reticulocyte count. Further work up with high pressure liquid chromatography showed that the patient had Hb D trait also. The co-existence of these two conditions is an extremely rare entity.

INTRODUCTION

The incidence of hereditary elliptocytosis varies worldwide from region to region and is not exactly known because of the asymptomatic nature of this condition. However, in the US population, it is approximately 3-5 per 10,000 [1].It causes symptoms of haemolytic anaemiaonly in a few affected patients.

Hemoglobinopathies are hereditary single gene disorders that affect the alpha or beta glob in chains of the haemoglobin molecule. Depending on the chain and protein moiety affected, there are a wide variety of haemoglobin variants which may or may not cause clinically significant disease. Hb D isone of them which has many variants- Hb D Punjab, Hb D Los Angeles, Hb D Iran to name a few. In India, its frequency is highest among Sikhs of Punjab (3.6%), followed by Jammu & Kashmir (3.3%), Uttar Pradesh (2.3%) whereas in Western India it is about 1%[2,3,4].

Case Report

A 25 year old female patient from Nanded presented with chief complaint of swelling in the left lateral side of neck since birth, easyfatigability accompanied by breathlessnesson exertion since 6 monthsand fever on &off since 5 days. She gave a past history of 2 blood transfusions, 10 years and 3 years back respectively and a single episode of seizure. In herfamily, she gave a history of anaemia in mother. On examination, she was poorly built, had severe pallor with mild icterusand moderate splenomegaly. There was no evidence of clubbing, oedema or lymphadenopathy. The neck swelling was diagnosed to beprobablybranchial cyst on fine needle aspiration cytology. Ultrasonography confirmed the splenic enlargement and demonstrated multiple gall stones.

Her Complete Blood Count showed Haemoglobin of 4.9g/dl, Mean Corpuscular Volume (MCV) 85fl, Mean Corpuscular Haemoglobin (MCH) 28pg, Mean Corpuscular Haemoglobin Concentration (MCHC) 36.9g/dl, Total Leucocyte Count 2870 cells/cu.mm and Platelet Count 1.50lakhs.Reticulocyte

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countwas raised to 5%. The peripheral blood smear examination stained with Field's A and B stain showed about 70% elliptocytes, few tear drop cells, schistocytes with marked anisopoikilocytosis. Total leucocyte count was reduced with normal morphology of the WBCs and platelets. Sickling test was negative. The liver function testsrevealedraised bilirubin of 6.45mg/dl with indirect bilirubin being 5.67mg/dl with normal AST and ALT levels and mild hypoproteinemia of 5.91g/dl.

In view of family history of anaemia, past history of blood transfusions and more than 25% elliptocytes on peripheral smear, a diagnosis of hereditary elliptocytosis was given and ancillary tests advised. High Pressure Liquid Chromatography was done. It showed Hb F of 1%, Hb A1c4.4%, Hb A0 64.5%, Hb A2 3.8% and an unknown peak corresponding to Hb D of 22.5%. Hence shewas diagnosed to have Hb D trait. Unfortunately, the patient took discharge against medical advice and hence further work up could not be done.

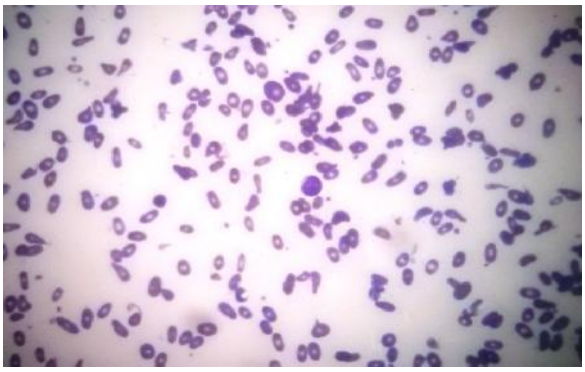

Fig 1 Peripheral Smear Field's satin (X40) showing more than 70% elliptocytes and anisopoikilocytosis

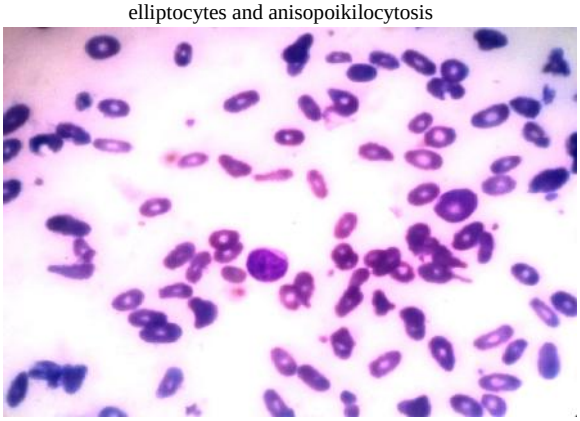

Fig 2 Peripheral Smear Field's stain (X100) showing elliptocytes (at higher magnification)

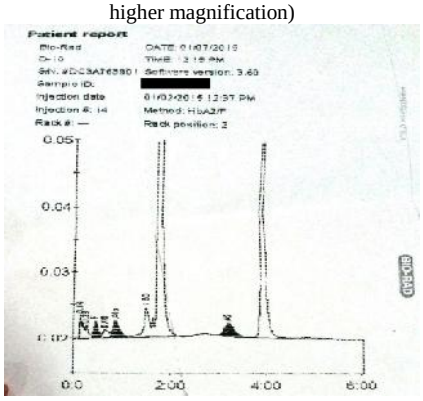

Fig 3 High Pressure Liquid Chromatography showing unknown peak.

DISCUSSION

Hereditary elliptocytosis is an autosomal dominant condition occurring due to mutations ingenes encoding red blood cell cytoskeleton proteins. The red cell is elastic and durable due to its inner specialized membrane skeleton.This is present inner to the plasma membrane. The chief protein componentof the membrane skeletonis spectrin which is composed of two polypeptide chains, alpha and beta intertwined into flexible heterodimers. The spectrin dimers at the head region self-associateto form tetramers and at the tail region associate with actin oligomers. The actin binds to multiple spectrin tetramers creating a two dimensional spectrin actin skeleton that is connected to cell membrane by two distinct interactions-one with protein ankyrin and band 4.2, another with protein 4.1 and glycophorin A.

Hereditary elliptocytosis is mainly caused due to mutations that disrupt the formation of spectrin tetramers. Approximately 65% of cases of hereditary elliptocytosis are the result of mutations of alpha spectrin, 30% are the result of mutations of beta spectrin, and 5% are the result of mutations of protein 4.1[5]. There arethree main forms of hereditary elliptocytosis: a) Common hereditary elliptocytosis. b) Spherocytic elliptocytosis.c) Southeast Asian ovalocytosis.But only 5-20% develops uncompensated haemolysiswith anaemia.

The criterion for diagnosis of hereditary elliptocytosis on peripheral blood smear examination is the presence of at least25% of red blood cells which are elongated into a cigar or oval shape. MCV should be normal, the mean corpuscular diameter below normal, and MCHC increased. Reticulocytosis can also be identified. Molecular genetic testing can also be done to look for the presence of mutations in the specific protein molecules of RBCs.

In our patient, the peripheral blood smear examination showed 70% elliptocytes with the RBC indices also suggestive of elliptocytosis. Pseudoelliptocytosis is a common artefact of peripheral smear preparation. This is ruled out since in pseudoelliptocytosis the blood cells appear stretched and lined up in parallel; this finding is in contrast to true elliptocytosis in which the cells are oriented in different directions (as seen in Fig. 1 & 2)

Hemoglobinopathies are caused due to single disorders in the alpha or beta globin chains of the haemoglobin molecule. Depending upon the gene affected, there are a wide variety of haemoglobin variants and hence hemoglobinopathies. Hb D is the fourth most common haemoglobinvariant encountered first time by Itano in 1951[6,7].It occurs due to substitution of glutamic acid by glutamine at the 121st position of beta globin chain. Hb D basically occurs in four forms: heterozygous Hb D trait, Hb D-thalassemia, Hb S-D disease, and the rare homozygous Hb D disease.

Hb D has an S-like mobility on alkaline electrophoresis but co-migrates with HbA on acid pH. On HPLC, it gives an unknown peak with retention time of 3.9-4.12min just between the peaks of Hb A2and Hb S/ Hb E(both of which elute in the S window).

In our patient with the value of Hb D being 22.5% it was diagnosed to be Hb D trait (Fig. 3)

CONCLUSION

Thus, it was a unique case showing dual defects in RBCs- one related to the defectinred cell membrane and the other a hemoglobinopathy. Both hereditary elliptocytosis and Hb D are rare disorders and the combination of these defects is extremely rare.

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How to cite this article:
Ujwala Maheshwari *et al*., A Rare Case Report of Heredtary Elliptocytosis With Hemoglobin d Trait. *International Journal of Recent Scientific Research Vol. 6, Issue, 5, pp.3856-3858, May, 2015*

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