A CASE REPORT "HB E (HOMOZYGOUS) DISEASE"

Garima Bafna[1], R.K. Jain[2]
Professor Physiology, JLN Medical College, Ajmer

Author for correspondence: - Professor Physiology, JLN Medical College, Ajmer Tel.: 0145-2643390: Mobile: 9414009709 Email ID: drgarimabafna@gmail.com

INTRODUCTION

Hemoglobin E is probably the most common structural hemoglobin variant with thalassaemic properties in the world population, reaching its highest frequency in eastern Thailand and Laos.

Hemoglobin E is not only a structural variant but is also synthesized inefficiently as compared with HbA, causing a clinical phenotype of a mild form of ß-Thalassaemia. The interaction of HbE with ß-thalassaemia results in Thalassaemia phenotypes ranging from a condition indistinguishable from Thalassaemia to a mild form of thalassaemia intermedia.

ß-Thalassaemia / Hb E is prevalent throughout south east Asia. It is far more common than homozygous ß-Thalassaemia because the prevalence of Hb E is higher than ß-Thalassaemia. In dealing with ß-Thalassaemia / Hb E one must bear in mind the diversity of phenotypes exhibited by this disease.

CASE REPORT

A 43 yr. old male patient comes to Medical OPD, JawaharLal Nehru Hospital, Ajmer with complain of Yellowish discoloration of eye, pain in joint and bones, Generalized weakness since 10 days on 28.05.2013 on coming to our hospital we have done primary investigation of CBC, PBF, Serum Bilirubin examination, S.Iron, S-TIBC. The Hb was 104, MCV-62.1 fl, MCH-21.5 pg, MCHC-34.7 gm/dl. Reticulocyte count 1.96% S.Iron-110 µ gm% (N-60-180 µgm%)TIBC - 246 µgm% (250-450).

S. Bilirubin Total 2.3 mg/dl Direct 0.6 indirect 1.7 mg/dl. His PBF shows microcytic hypochromic blood picture.

On finding the blood picture we advise Hemoglobin electrophoresis just to rule out possibility of thalassaemia minor.

After the report of Hb electrophoresis came as Hb adult 3.9%, Hb A_2/E-81.1%, Hb F - 5.5% ,the case was diagnosed as Hb E disease (Homozygous) on investigating about family. We came to know out of his three children (2 male & one female child) one male child is also suffering from moderately severe ß-Thalassaemia / Hb E disease.

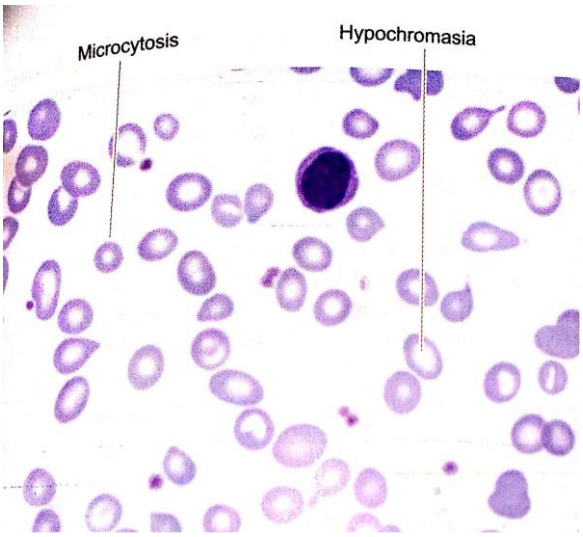
DISCUSSION

HbE is abnormal hemoglobin with a single point mutation in the ß-chain. At position 26 there is a change in the amino acid from glutamic acid to lysine. Hemoglobin E has been one of the less well known variants of normal Hb. It is very common in south east Asia but has a low frequency amongst other races. Hb E can be detected on electrophoresis. The ßE mutation affects ß gene expression creating an alternate splicing site in the mRNA at codons 25-27 of the ß globin gene. Through this mechanism there is a mild deficiency in normal ß mRNA and production of small amounts of anomalous ß mRNA. The reduced synthesis of ß chain may cause ß globin causing instability when there is high amount of oxidant.

Hb E Disease (E)

Hb E Disease results when the offspring inherits the gene for Hb E from both parents. At birth babies homozygous for the Hb E allele do not present symptoms due to Hb F which they still have in the 1st month of life.

Fetal Hb disappears & the amount of Hb E increases, so the subjects start to have mild ß Thalassaemia. People who are heterozygote for Hb E (one normal allele & one abnormal allele) do not show any symptoms (there is usually no anemia or hemolysis). Subjects Homozygous for the Hb E allele (two abnormal allele) have a mild Hemolytic Anemia & mild splenomegaly.



Hb E/ ß Thalassaemia

People who have Hb E / ß Thalassaemia have inherited one gene for Hb E from one parent & one gene for ß Thalassaemia from the other parents. Hb E/ß Thalassaemia is a severe disease and it still has no universal cure. It affects more than a million people in the World. The consequences of Hb E/ß Thalassaemia (when it is not treated) can be heart failure, the enlargement of the Liver, Problems in bones etc.

There are variety of genotype depending on the interaction of Hb E & ß- thalassaemia. The presence of the ß-thalassaemia reduces the amount of HbE usually found in HbE heterozygote. In other case in combination which contain thalassaemia mutation. It provides an increase resistance to malaria (PF).

CONCLUSION

From above case history we came to know that-
(1) It is important to differentiate common iron deficiency Anaemia from ß-Thalassaemia/Hb E to avoid unnecessary iron supplements because they are already at risk of iron overload.
(2) Avoid excessive blood transfusion to ß-thalassaemia / Hb E Patients who are not severely Anaemic and are more or less adapted to lifelong chronic Anaemia.

REFERENCE

1. Chernoff Al, Minirich V Nanakorn S. et al, Studies on Hb E. ± the clinical hematologic and genetic characteristic of Hb E syndromes J. Lab clin med 1956, 47, 455-489.
2. Vichinsky E Hb E Syndromes, ASH Education Book 2007 Vol. 2007 No. : 79-83.
3. Bachiv D & Galecteros F, Hb E disease orpha net Encyclopedia Nov, 2004.
4. http:/www.vrmc.rochest-edu/medicine/genetics/media/documents/hemo E. pdf
5. Kazazian HH JR Wabes PG Bochem CD Lee Ji Antonarakis SE, Fair haul VF, Hb E in Europenas further evidence for multiple origins of the ßE globin. Gene. AmJ Hym. Genet 1984; 36 : 212 - 217.
6. Brain BJ Blood Cells a practical guide 4th ed. Oxford UK : Blackwell 2006.

Disclosure: No conflicts of interest, financial, or otherwise are declared by authors