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# Prevalance of HBE Disease in North-East Region of India

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***Abstract***

*HbE(Beta26Glu—lys)is an abnormal haemoglobin in which there is a single point mutation in beta chain.There is a change in amino acid, from glutamic acid to lysine at position 26.It is the most common Haemoglobin variant in South-East Asia and the second most common worldwide.However in India, it is prevalent in North-East region & West Bengal, but relatively less in rest of the country.At birth babies Homozygous (two abnormal alleles)for HbE allele do not present symptoms, because they still have HbF.After the disappearance of HbF, the amount of HbE increases & now the patient starts developing mild Beta Thalassemia with mild Haemolytic Anaemia & mild splenomegaly.*

*Heterozygous HbE occurs when the gene for HbE is inherited from one of the parent(known as HbE Trait) & is not a Disease as patients are asymptomatic and they don't have any health problem, although they may have low MCV and abnormal red blood cells (target cells) on peripheral smear but clinical relevance is mainly due to potential of transmitting HbE.*

*This paper reports total 200 cases(from the month of January'23 to april'23)from North-East region & West Bengal out of which 11 cases are of HbE Trait and 2 cases are of HbE Disease.*

*Lab investigation is based on RBC indices & HPLC parameters.This study has been done in a Redcliffe Diagnostics Lab on 200 patients from the month of January-April'23.*

*The main aim of this study is to create awareness among the people of North-East region of this rare disorder so that it can be included in the differential diagnosis of Thalassemia Intermedia & Thalassemia Major.Due to which people may undergo Prenatal Diagnosis & Genetic Counselling.*

## Introduction

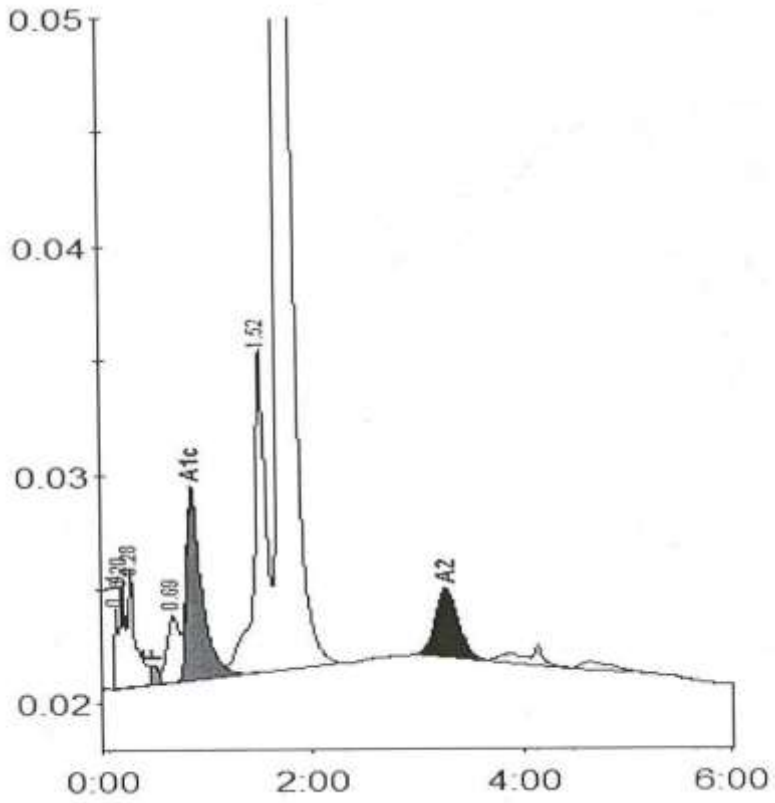
It has been estimated that approximately 70 lakhs children born each year either have some congenital anomaly or have some genetic disorder. Among these 25% of these babies suffer from only five disorders out of which two belong to inherited disorders of haemoglobin. Thalassemias are worldwide genetic disorders with defective globin chain synthesis. HbE results from glutamate to lysine substitution in codon#26 of the beta globin gene which produces structurally abnormal haemoglobin. As well , Hb is synthesised at a reduced rate & behaves as a mild form of Beta Thalassemia. In the Indian subcontinent, HbE is restricted to the North-Eastern states( West Bengal, Assam, Nagaland, Manipur, Tripura & Meghalaya).Identification of of HbE is very important as double heterozygous patients may present clinically as Thalassemia Major and if not treated may lead to dreadful complications. HPLC is one of the best methods for the screening and detection of various Haemoglobinopathies and provides rapid, reproducible & precise results.The present study attempted to find out the occurrence of HbE in the population of North-East region & West Bengal so that a definite plan of action regarding the diagnostic, preventive & therapeutic strategies can be formulated to minimise disorders in future generations.

## Materials & Methods

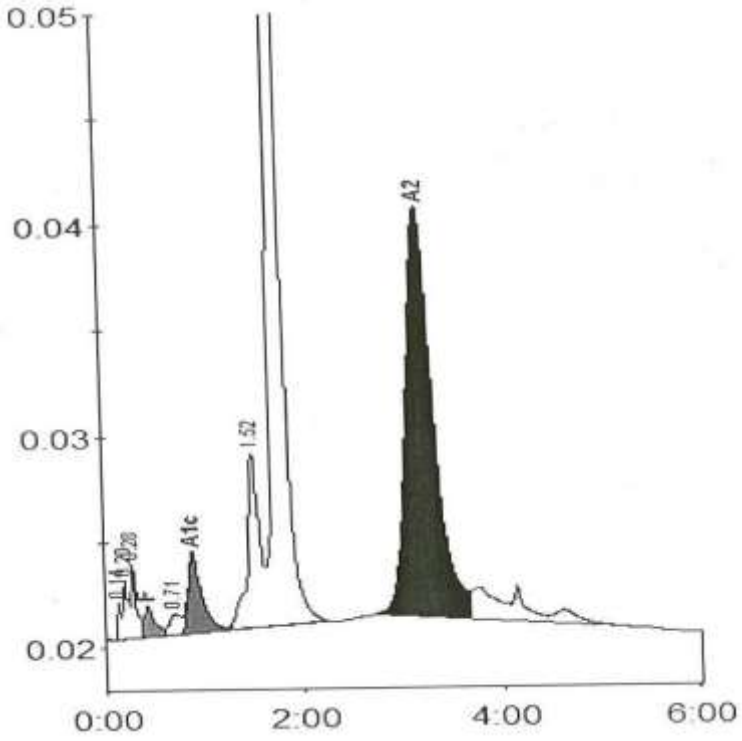
This study was conducted in Redcliffe Diagnostics Lab(Pathology Department)over a period of three months from January ‘23 to March’23. Patients who came for HPLC TEST (pregnant females, small children & adults with low haemoglobin)were included in the study. Detailed clinical history and family history along with past history of blood transfusion, if present was noted. Blood samples were collected in EDTA vials and analysed with Horiba Yumizen 1500 cell counter for complete blood counts & Biorad D-10 analyser for HPLC analysis. HbE was identified by its retention time and quantified by computing the area under the corresponding peak in the elution profile. Reports with Hb, RBC count, PCV, MCV, MCH, MCHC, RDW(CV), RDW-SD, HbF, HbA2, HbAO, HbD, HbS, HbC, HbE along with graphs were generated.

## Results

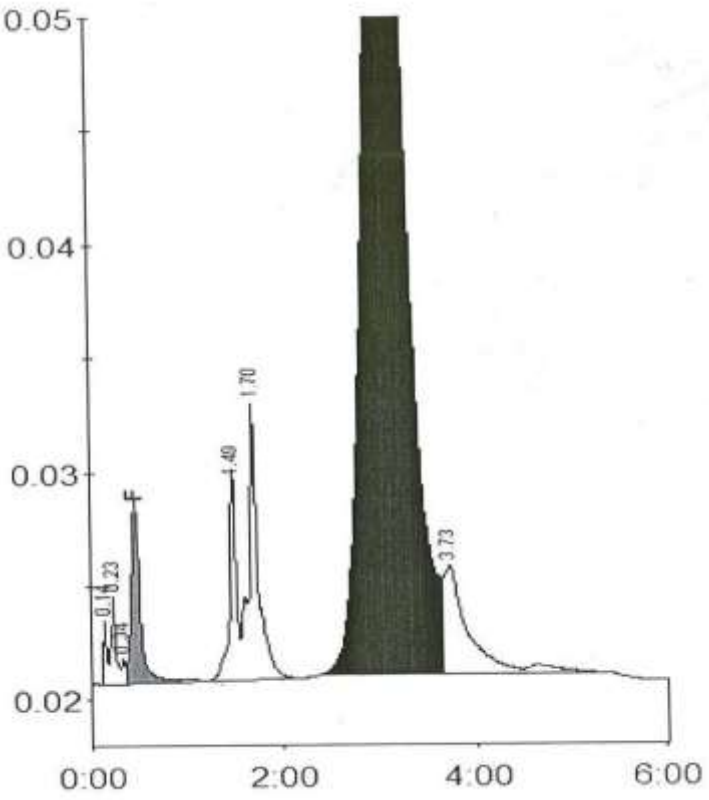
During the period of three months ,among approximately 200 patients from North-East region & West Bengal the most common abnormality in haemoglobinopathy detected was HbE.11 cases were of HbE Trait (heterozygous)& 2 cases were of HbE Disease.(homozygous)




**Normal HPLC Graph**




**HbE TRAIT(HETEROZYGOUS)**




**HbE(HOMOZYGOUS)**

## Conclusion

11 out of 13 cases came out to be HbE Trait while 2 cases were of HbE Disease. HbE Trait is a heterozygous state, clinially silent associated with microcytosis, slight erythrocytosis and target erythrocytes; no anaemia or reticulocytosis is seen. Lower proportions of HbE in carriers indicate the presence of co-inheritance of Alpha Thalassemia or iron deficiency anaemia. Diagnosis is based on the Hb pattern analysis by electrophoresis or HPLC test.

Homozygosity for HbE shows prominent microcytosis, target cells, leptocytes but little or no anaemia and slight Splenomegaly. On quantitative analysis HbE accounts for 70-95% of the Hb; HbF is normal or is only slightly increased. Red cell survival is slightly reduced while they heterozygous patients are asymptomatic. The awareness of HbE, a relatively rare Hb variant in this part of India may have utility in clinical management and genetic counselling. The main aim of this study is to create awareness among the people of North-East region of this rare disorder so that it can be included in the differential diagnosis of Thalassemia Intermedia & Thalassemia Major. Due to which people may undergo Prenatal Diagnosis & Genetic Counselling.

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