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**RESEARCH ARTICLE**

# A CASE REPORT ON HBE HOMOZYGOUS HAEMOGLOBINOPATHY

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## INTRODUCTION

Haemoglobin within the red blood cells is vital for existence, being the means of transporting oxygen to tissues. Haemoglobin (Hb) abnormalities are the most frequent genetic disease, affecting approximately 7% of the world population [1].Haemoglobin E is a β chain variant, which is common in south-east Asia[2]. Chernoff and colleagues first described it in 1954 [3]. The prevalence of HbE in India is about 0-3.5% with a increased clustering in Kolkata (22%) and Assam (50-80%)[1].

### Case Report

A 49-year-oldasymptomatic patient, anortheast descent,who is a known case of type II diabetes, came to the medicine OPD for routine investigations. Fasting and postprandial plasma glucose levels were raised.

Thus, Glycosylated Hb was advised to study the glycemic index. On performing HbA1c by HPLC, the values remain undetected and an unknown variant window covering nearly 88% at R.T. 1.58 was observed. (Figure 1). In view of this, Complete blood count and High Performance Liquid Chromatography was advised to the patient. CBC findings revealed haemoglobin of 12 gm/dl, total count of 8200 cells/cumm with a relative normal leukocytic distribution and a platelet count of 1.72 lacs/cumm.

Mean corpuscular volume (MCV), Mean haemoglobin concentration (MCH) and mean corpuscular haemoglobin concentration (MCHC) was found to be 69.0 fL, 19.7 pg, 28.6 g/dL respectively. Red cell distribution width was raised to 19.2%. Peripheral smear reviewed showed predominantly microcytic hypochromic RBCs with occasional tear drop cells (Figure 2). High performance liquid chromatography (HPLC) performed on D-10 by Bio-Rad showed Hb F value of 4.6%, HbA1c of 3.4%, HbA0 of 7.2% and HbA2 of 124.7%, values suggestive of HbE homozygous disease. (Figure 3). The patient was advised family screening in order to study genetic make up. It was observed that both his parents were HbE trait.

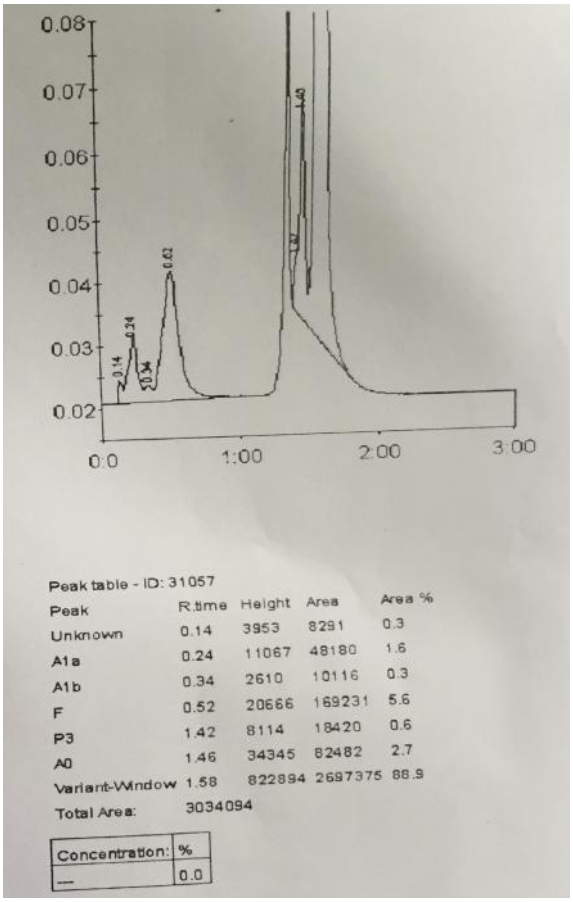


**Figure 1** Chromatogram showing eluted HbA1c

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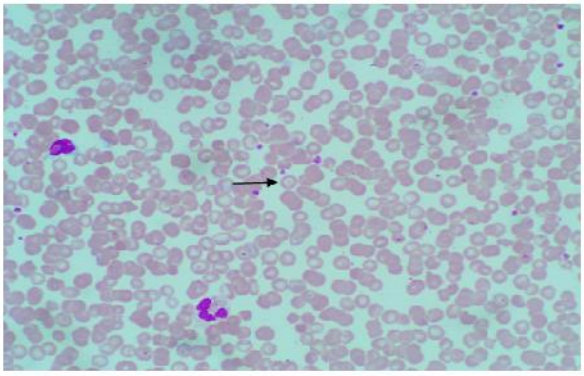

**Figure 2** Peripheral smear showing microcytic hypochromic RBCs(arrow points at target cell, Leishman stain, 400x)

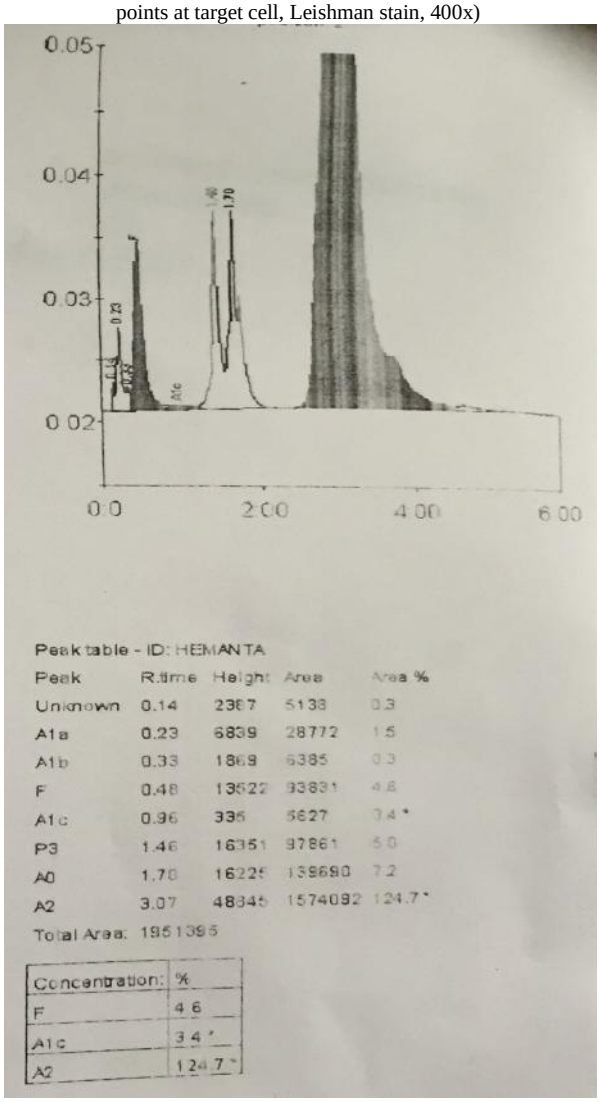


**Figure 3** Chromatogram of HbE homozygous

## DISCUSSION

Nuclear DNA, including the DNA of globin genes, is subject to spontaneous mutations. The site of mutation is critical, different point mutations may give rise to the same variant of haemoglobin. The diagnosis of disorders of haemoglobin chain synthesis usually requires a combination of tests along with detailed clinical history, ethnic background, blood count and peripheral blood film examination. [4].Patients suffering from HbE disease present with chronic anemia without splenomegaly, blood count often resemble βthalassaemia trait. HbE gene is a mutant form of the β-globin (HbE) gene that encodes lysine instead of glutamate at position 26. This β-E chain is inefficiently produced because of a novel cryptic messenger RNA splicer site, leading to thalassaemic RBC indices [5]. Heterozygosity (HbE trait) and homozygosity (HbC-E disease) is clinically mild, where as compound heterozygosity for HbE and HbS (HbSE) and compound heterozygosity for HbE and β-thalassaemia (HbE-β-thalassemia) are clinically severe[6,7,8,9,10].Thus β chain of HbE is synthesized at a reduced rate compared with that of the normal adult haemoglobin (HbA). This results in reduced rate of synthesis of βE chain and therefore of HbE, and consequently heterozygotes, compound heterozygotes and homozygotes show some β thallaesaemic features. [11]. Cation-exchange HPLC is considered as the method of choice to quantify the various normal and abnormal Hb fractions [12]. However it cannot separate HbE from $HbA_2$and hence appears in the A2 window. Thus it has been impossible to precisely determine the quantity of $HbA_2$ in presence of HbE and definitive diagnosis of concomitant thalassemia requires DNA testing.

## CONCLUSION

Thus we report an unusual case of HbE masking HbA1c levels, knowledge of such a condition would help in prevention of misdiagnosis and provide better disease management and patient care. Also, we would like to emphasize of the prenatal screening of neonates for Haemoglobinopathies to prevent further disease burden and be better prepared for tackling any unforeseen complications.

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