LOW COST SCREENING METHOD FOR IDENTIFICATION OF HAEMOGLOBIN E/β THALASSAEMIAS

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Haemoglobin E/β thalassaemia are the double heterozygote state for Haemoglobin E and the β thalassaemia and have a variable phenotypic expression. Even in the siblings with same genotype there is a variable phenotypic expression rendering some patient's transfusion dependent and others asymptomatic. The factors make them transfusion dependent are not well understood. By identifying these factors and inducing these in transfusion, dependent patient should be able to convert them back to transfusion independent patient. A population survey is necessary for identification of these patients. The usual methods of identification of these patients need haemoglobin electrophoresis, which is very expensive. The aim of this study is to device a low cost method to identify E/β thalassaemia cases.

We studied 143 patients with anaemia due to common causes including Hb E syndromes and 20 normal individuals. The diagnoses of these cases were confirmed by using standard biochemical and haematological investigations for each of the condition. Peripheral blood samples were collected into heparinised capillary tubes and packed cell volume, plasma bilirubin level and results of dichlorophenol indophenol test were measured and used to prepare a three dimensional graph to identify the clusters. Four clusters were identified indicating Hb E disease, Hb E trait, Hb E/β thalassaemia and another cluster including IDA, anaemia of chronic disorder, β thalassaemia trait and enzymopathies. The differences between each of those categories were identified statistically using discriminant analysis.

The results indicate that, by using these three simple measurements cases of Hb E disease can be identified 100% correctly, Hb E trait can be identified 89% correctly and Hb E/β Thalassaemias can be identified 88% correctly. The number of misclassifications for the total group was only seven. The misclassifications in Hb E/β Thalassaemias are due to the presence of unusual cases of Hb E trait with transfusion dependency due to coinheritance of other haematological diseases

The results indicate that this combination of three simple tests, which cost few rupees, can predict the cases of Hb E syndromes correctly categorizing them into homozygous, heterozygous and double heterozygocity with β thalassaemia and can be used as reliable method to identify these conditions in population screening.