STUDY OF ABNORMAL HEMOGLOBINS AND THALASSAEMIAS IN AN ANEMIC POPULATION

A PROJECT REPORT PRESENTED

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ABSTRACT

EFFECT OF DIFFERENT HEMOGLOBINS ON GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY SCREENING TEST

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Glucose-6-phosphate dehydrogenase (G6PD) deficiency and hemoglobinopathies are common causes of hemolytic anemia in Sri Lanka. Even though G6PD enzyme assay procedure is important for diagnosis of G6PD deficiency all government hospitals in Sri Lanka employ Brewer's test for diagnosis. The aim of the study is to observe the effect of different hemoglobins on screening test for G6PD deficiency. Patients and carriers were investigated for commoner hemoglobinopathies (Hb E, Hb S, Hb D) and thalassaemias using G6PD deficiency screening test as well as G6PD enzyme assay. The results show that Hb E present in Hb E/β thalassaemia, Hb E trait and Hb E disease, gives false positive answer with G6PD screening test, even though their G6PD enzyme levels are normal. Two subjects showed low enzyme levels with positive screening test. Only three out of forty nine (6.2%) were negative for G6PD screening test. Hb S (n=5) and Hb D (n=2) did not show any effect on the screening test for G6PD deficiency. The conclusion is that patients with Hb E are likely to be labeled with G6PD deficiency, while having normal enzyme levels, if only the screening test is used.

ABSTRACT

DICHLOROPHENOLINDOPHENOL (DCPI) TEST TO SCREEN Hb E/β THALASSAEMIA

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Thalassaemia and abnormal hemoglobins are common genetic disorders in Southeast Asia. This is not only an important public health problem but also a socio-economic problem of many countries in the region. An approach to deal with this problem is to prevent and control births of new cases. This requires an accurate identification of couples at high risk in having a thalassaemic child. The diagnosis of thalassaemia and abnormal hemoglobin carriers need several tests that are not practical for screening the population at large. In this study we used three simple laboratory tests to screen for Hb E/β thalassaemia individuals.

One hundred and forty five subjects with different types of anemias and hemoglobinopathies were recruited for this study. Cluster analysis was used to understand the distribution, and if possible, to group patients with similar characteristics. In this study, three independent parameters, packed cell volume (PCV), serum bilirubin level and dichlorophenolindophenol (DCPI) precipitation test were plotted for each case

of anemia. Red cell indices, hemoglobin electrophoresis and other laboratory tests were performed to confirm phenotypes of thalassaemia.

The results showed that three-dimensional representation of PCV, bilirubin and DCPI separated Hb E/β thalassaemia and Hb E/trait into a cluster different from the rest of the anemic population. On either side of Hb E/β thalassaemia-Hb E/trait there were two distinct clusters. As expected, it was found that the three clusters had anemics of different genetic character.

ABSTRACT

CHARACTERIZATION OF Hb E/β THALASSAEMIA USING LABORATORY TESTS

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Hemoglobin E/β thalassaemia is the double heterozygote state for hemoglobin E and β thalassaemia. The clinical picture varies from an almost asymptomatic state, to a severe disease, indistinguishable from β°-thalassaemia major. The aim of this study is to characterize hemoglobin E/β thalassaemia by means of clinical, hematological and laboratory tests available in Sri Lanka.

The results showed that Hb E/thalassaemia could be classified in to three categories, with phenotype ranging from transfusion dependent to a complete lack of symptoms. Most patients (50 %) were able to survive for many years with mean hemoglobin between 6.5 and 8.5 g/dl and only infrequent transfusions. Bony changes and mongoloid facial appearance were seen prominently in severe Hb E/thalassaemia, whereas in HbE/trait and Hb E/disease bony changes were not significant. The serum ferritin levels were nearly equal in all three groups of Hb E. The level of sTfR was markedly elevated in all three groups of Hb E. In Hb E/thalassaemia, Hb F had a high value of 20.18 ± 7.59 %

compared to other Hb E groups. The mean Hb E levels were high (48.57 ± 3.23) % in Hb E/thalassaemia and Hb E/trait (42.17 ± 5.32) %, and a wide range of Hb E 13.5 - 57.5 % was seen in Hb E disease. Hb A levels in Hb E/trait and Hb E/disease varied from 57 to 82 %. As the amount of Hb A was quite substantial, signs and symptoms were less severe than in the Hb E/thalassaemia.