Original Research Article

Screening of thalassemia and other hemoglobinopathies in blood donors by capillary hemoglobin electrophoresis system

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Abstract

Introduction: Hemoglobinopathies are a group of genetic disorders of hemoglobin. Because of consanguinity, caste and area endogamy, some communities show a very high incidence, making the disease a major public health problem. The aim of this study is to screen for thalassemia and abnormal hemoglobinopathies in blood donors by capillary electrophoresis system for early detection, management and referring positive cases for genetic counselling.

Materials and methods: This was a hospital based observational study which was done for a period of one year and EDTA blood samples of anaemic blood donors showing microcytic hypochromic blood picture or low hemoglobin (<7 gm/dl) and low MCV, MCH values were studied. Both voluntary and replacement donors were included in the study. Screening for β thalassemia and other hemoglobinopathies was done in Department of Transfusion Medicine using MINICAP FLEX-PIERCING Capillary haemoglobin electrophoresis system. At the end of the analysis, relative quantification of individual hemoglobin fractions was performed automatically and profiles can be analyzed; the hemoglobin fractions, Hb A, Hb F, Hb A2 were automatically identified.

Results: We had screened 36 blood donors having microcytic hypochromic picture. Out of which 20 turned out to be Beta Thalassemia Trait by capillary hemoglobin electrophoresis system. Therefore, out of the sample size of 36 blood donors, 55.5% were Beta Thalassemia trait.

Conclusion: This study was based to screen thalassemic patients as carriers along with hemoglobinopathies so as to reduce the rate of affected infants and screen even asymptomatic patients for early management and genetic counselling.

Key words

Blood donors, Hemoglobinopathies, Capillary hemoglobin electrophoresis system.

Introduction

The term hemoglobinopathy is used to designate the structural disorders and the term Thalassemia is associated with the quantitative disorder [1]. Every year, 10,000 children with β thalassemia major are born in India, which constitutes 10% of total number in the world. Inherited disorders of hemoglobin synthesis are an important cause of morbidity and mortality worldwide. The curative treatment like bone marrow transplant is costly and so prevention is the cost effective strategy, which includes population screening, genetic counseling and prenatal diagnosis [2]. This has been a success in countries like Greece, Cyprus and Italy [3]. Patients homozygous for β-thalassemia or α thalassemia usually present with the symptoms of the disease whereas carriers for α or β thalassemia are usually found during examination of the relatives of more severally affected patients as part of screening programmes or during the investigation of mild iron refractory hypochromic anemia. Theoretically there are numerous types of thalassemia as there are numerous types of globin chains but clinically most relevant are α and β thalassemia [4].

α Thalassemia result from deletions in 1 or more of the 4 genes responsible for α globin synthesis. 2 genes control α-chain on *Chromosome 16*. β Thalassemia are more common than α thalassemia. Only 1 gene on *Chromosome 11* (2 alleles) is affected [5]. Sickle cell disorders are a group of autosomal recessive disorders, caused by point mutation at the sixth position in β globin chain, valine substituting glutamic acid [6]. Hemoglobin E mutation which causes replacement of glutamate by lysine at 26th position on β-chain [7].

The aim of this study is to screen for thalassemia and abnormal hemoglobinopathies in anaemic blood donors having microcytic hypochromic picture or low haemoglobin (<7 g/dl) for early detection and proper management, to reduce the rate of affected infants and referring the positive cases for genetic counselling.

Materials and methods

This was a hospital based observational study which was done for a period of one year and EDTA blood samples of anaemic blood donors showing microcytic hypochromic blood picture or low hemoglobin (<7 gm/dl) and low MCV, MCH values were studied. Both voluntary and replacement donors were included in the study. Screening for β thalassemia and other hemoglobinopathies was done in Department of Transfusion Medicine using MINICAP FLEX-PIERCING Capillary haemoglobin electrophoresis system. The MINICAP FLEX-PIERCING instrument uses the principle of capillary electrophoresis in free solution. With this technique, charged molecules were separated by their electrophoretic mobility in an alkaline buffer with a specific pH. Separation also occurs according to the electrolyte pH and electroosmotic flow. These Hb fractions were directly detected at a specific absorbance of 415 nm.

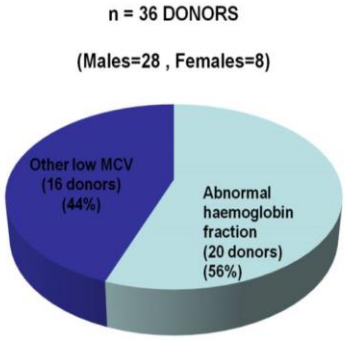
At the end of the analysis, relative quantification of individual hemoglobin fractions was performed automatically and profiles can be analyzed; the hemoglobin fractions, Hb A, Hb F, Hb A2 were automatically identified.

Results and Discussion

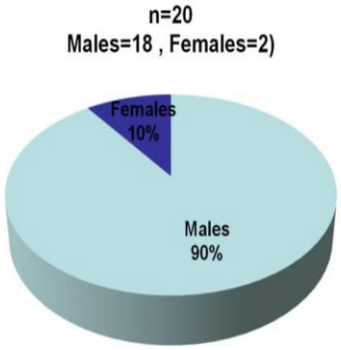
This study was conducted on 36 non remunerated voluntary and replacement blood donors. Of these 20 (55.5%) showed abnormal hemoglobin fraction. The major abnormalities observed were of high HbA2 level and a cut-off of 3.5% was taken for diagnosis of beta-thalassemia trait. It was found that the majority of blood donors had Hb 12.5 g%, HbF levels are within normal limits with the variable reduction in HbA. MCV and

MCH were below normal limits with raised RBC count.

Graph – 1:



Graph – 2:



Majority of donors were males (90%); low number of female donors could be because of local social factors and physical health like anaemia barring them from blood donation. Majority of blood donors under study were in reproductive age (21-45 years) of life. But surely this study concludes the importance of hemoglobinopathy among the so called healthy blood donors.

β-thalassemia trait is probably the most common inherited hemoglobin disorder in the Indian sub-continent. Transfusion of blood obtained from thalassemic trait seems to be one of the possible causes of ineffective transfusion. In a very recent study from Thailand showed that an imbalanced alpha/beta-globin chain as a consequence of either reduction or enhancement of beta-globin chain synthesis can cause abnormal RBC properties in mouse models. This can be extrapolated that RBC of thalassemia trait are defective and hence has short survival as compared to normal β, α globin chain RBC.

Conclusion

Capillary hemoglobin electrophoresis is considered as one of the best methods for screening and detection of various hemoglobinopathies with rapid, reproducible, cheap and precise results. It is recommended for detection of β-thalassemia trait in population and necessary for genetic counseling to reduce the incidence and burden of thalassemia major in the society.

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