Molecular basis of transfusion dependent beta-thalassemia major patients in Sabah

ABSTRACT

Beta-thalassemia is one of the most prevalent inherited diseases and a public health problem in Malaysia. Malaysia is geographically divided into West and East Malaysia. In Sabah, a state in East Malaysia, there are over 1000 estimated cases of ϐ-thalassemia major patients. Accurate population frequency data of the molecular basis of ϐ-thalassemia major are needed for planning its control in the high-risk population of Sabah. Characterization of ϐ-globin gene defects was done in 252 transfusion dependent ϐ-thalassemia patients incorporating few PCR techniques. The study demonstrates that ϐ-thalassemia mutations inherited are ethnically dependent. It is important to note that 86.9% of transfusion-dependent ϐ-thalassemia major patients in Sabah were of the indigenous population and homozygous for a single mutation. The Filipino ϐ0-deletion was a unique mutation found in the indigenous population of Sabah. Mutations common in West Malaysia were found in 11 (4.3%) patients. Four rare mutations (Hb Monroe, CD 8/9, CD 123/124/125 and IVS I-2) were also found. This study is informative on the population genetics of ϐ-thalassemia major in Sabah

Keyword: ϐ globin gene mutations; Beta thalassemia; Filipino ϐ0-deletion; Indigenous population; Sabah