Clinico-Hematology And Beta Mutation Profiling Of Beta Thalassemia In State Of Kedah - A Pilot Study

Fahmaa Azizi¹, *Rosline Hassan¹, Hafizah Hashim²

¹Department of Hematology, School of Medical Sciences, Universiti Sains Malaysia
²Hematology Unit, Department of Pathology, Hospital Sultanah Bahiyah Alor Setar

roslin@usm.my

Abstract

Beta-thalassemia is a group of hemoglobin diseases caused by a reduction (α+ thalassemia) or absence (α0 thalassemia) in the synthesis of beta-globin chains. More than 400 different types of mutations have been described as being responsible for this disease. Affected individuals can be heterozygous, compound heterozygous, or homozygous for beta-thalassemia, or even have interactions with other hemoglobinopathies. Kedah has a high prevalence of thalassemia (4th highest state), with 20.25/100000 populations. More than 400 different mutations have been reported and identified in the β globin (HBB) gene which are responsible for the development of the β-thalassemia. They generate a wide variety of different clinical phenotypes. The identification of the genotype is important for providing proper counseling to patients and their families. Characterization of these mutations should aid the planning of prenatal diagnosis program for β-thalassemia. To determine the clinical, haematology and beta mutation profiling of beta thalassemia patients in state of Kedah. 63 patients with diagnosis of beta thalassemia were traced through hospital information system. A detailed history with clinical evaluation of all the patients were retrieved. For β-thalassaemia genotyping, 20 different mutations were tested: 19 by M-ARMS and one by simple ARMS technique. Out of 63 patients, 54% were transfusion dependent beta thalassemia with 62% heterozygous beta thalassemia/HbE and 38% are homozygous beta thalassemia. 46% of the studied patients were non-transfusion dependent with 76% were heterozygous beta thalassemia/HbE and the remaining were homozygous beta thalassemia. The most common mutation among transfusion dependent beta thalassemia was compound heterozygous CD26 and CD41/42 (27%) whereas among the non-transfusion dependent thalassemia were compound heterozygous CD26 with IVS1-5 and IVS 1-1 with frequency of 21% and 16% respectively. By characterising the type of mutation among thalassemia patients should aid in planning of the genetic counselling among the newly diagnosed thalassemia patients.

Keywords: beta thalassemia; non transfusion dependent; transfusion dependent; mutation

*Author for Correspondence