

HBG2, BCL11A and HBS1L-MYB as Potential Modifier for Blood Transfusion Requirement in Beta Thalassemia Major Patients

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Beta thalassemia is an inherited hemoglobinopathy in which the production of one or both beta globin chains is impaired that has the highest prevalence globally. In Indonesia the number of thalassemia patients at 2009 rose to 8.3 percent of 3,653 registered patients in 2006. Persons with beta thalassemia treated in hospitals often show diverse clinical manifestations. Based on previous research, people with thalassemia had common mutations showing damage on beta globin gene (HBB). The molecular basis for this HBB mutations turned out to vary from one population to other populations. In other words, the distribution of HBB mutation which causes thalassemia, related to ethnic, HBG2, BCL11A, and HBS1L-MYB is a modifier for hemoglobin F in hemoglobinopathies disorder. In addition, there is complexity of the genetic background associated with a variety of phenotypes presented among patients. Genetic heterogeneity related to fetal hemoglobin (HbF) production has been reported as an influencing phenotypic factor of β -thalassemia. The beta thalassemia major is one of hemoglobinopathies disorder that required regular blood transfusions. The risk for complications such as hemosiderosis, infections and other transfusion reactions are increased in patients with more frequent transfusion. Previous research has shown that the levels of hemoglobin F associated with blood transfusion requirements in patients with β thalassemia major. However, the potential modulator HBG2, BCL11A and HBS1L-MYB as a prognostic factor for blood transfusion requirement in patients with beta thalassemia major of Indonesian population, is not yet known. This situation can detain early diagnosis, effort to reduce morbidity and the quality of life improvement. As part of the thalassemia belt, molecular heterogeneity of beta thalassemia in West Java is believed to vary and can be a genetic marker. Upon this, prognostic factors HBG2, BCL11A and HBS1L-MYB related with blood transfusion requirement in patients with beta thalassemia major will be analyzed.

Keyword: Faktor Prognostik, Transfusi Darah, Talassemia

Correlation between HBA Mutation Spectrum and Phenotype Scoring in Thalassemia Patients

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α -Thalassemia is an autosomal recessive genetic disorder characterized by symptoms of hemolytic anemia that is caused by a deletion in the gene HBA1 and HBA2 so that the formation of α -protein globin becomes disturbed. 1-3 α -thalassemia clinical picture varies depending on the magnitude of disruption of the gene HBA1 and HBA2. 1,2,4 α -Thalassemia are found a lot in the tropical region, especially among Asian race. 5-7 Research by Eijkman Molecular Biology Institute in Jakarta has been carried out on five ethnic populations. Molecular defect found in many ethnic Chinese are SEA (58%) and α -globin gene deletions 3.7-kb (27%). Not many studies, looking for correlations between genotype and phenotype in α -thalassemia, have been reported. 8-10 Determination of types of mutations needs to be examined as a basis of knowledge on the types of mutations that underlie clinical variation and severity of α -thalassemia. 2,4,11 Such information can be used in early diagnosis efforts to reduce morbidity and improve quality of life. Research on the description of the type and frequency of HBA1 and HBA2 mutations, phenotype mapping, and correlation of genotype and phenotype in people with α -thalassemia, in West Java.

The research sample include people with thalassemia who came to the outpatient treatment at selected hospitals in West Java. This study is a retrospective observational cohort, which is analytical in nature. This is a multi-year study, with the first year devoted to finding data of the types of HBA1 AND HBA2 mutation in people with thalassemia, selected as the subjects in the research sample. Still in the same year, identification will be conducted of clinical variation and severity before observations on HBA1 and HBA2 mutations. In the following year, further identification will be carried out of the data as regards phenotypes, that is, clinical symptoms and severity levels at the end of the observation period.

Keyword: HBA Mutation, Scoring, Thalassemia Patients