Phenotypic spectrum of thalassaemia major in West Java Province: implications for the future health burden

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Abstract

Thalassaemia major is the most common monogenic diseases with the highest frequency in Indonesia. According to Indonesian Thalassemia Foundation-The Parent Association of Thalassemia Patients (YTI-POPTI), West Java has the largest number of people with thalassemia which involves 1751 out of 5501 patients. Complex characteristic appearance with a wide spectrum in beta thalassaemia patients has resulted in the clinician’s difficulty in predicting prognosis. Accurate phenotype spectrum data are needed for predicting prognosis in the high risk West Java Province. This research is a descriptive quantitative study with a cross sectional design. One hundred and sixty consecutive samples of thalassemia patients was taken from two geographical areas of West Java Province. The spectrum of thalassemia was analysed with descriptive statistics for both characteristics and scoring parameters, which was developed by Thalassemia Research Center, Mahidol University. This scoring has six independent parameters: haemoglobin level, age at disease presentation, age at receiving first blood transfusion, requirement for transfusion, spleen size, growth and development. This score was able to separate patients into three distinctive severity categories: mild, moderate, and severe. The data were taken from medical record and questionnaires. There was a high prevalence (61%) of moderately severe beta thalassaemia patients. The severe group had a mean haemoglobin content around 4.66 ± 1.50 g/dL. Most of the population in this area were first diagnosed as younger than 3 years and received the first transfusions when they were under 5 years old. Overall, 95.92% from moderate degree had routine transfusions every 4 weeks. All patients had spleen size under 3 cm. There was similarity in the proportion of growth status in moderate degree between <3rd percentile and 3-25th percentile, which is 31.63%. The phenotypic spectrum of patients with thalassemia major in West Java is mostly moderate degree, this poses a challenge for clinicians with appropriate therapy which may result in a better prognosis.

Keywords: phenotype, scoring, thalassemia major

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