

HbE β -Thalassaemia in Malaysia: revisited

Abstract

HbE β – thalassaemia is a public health problem in Malaysia and the most common type of thalassaemia seen in the Malays. It shows considerable diverse phenotypes. Complete molecular analysis to identify primary/ secondary alleles of thalassaemia and gene modifiers are arbitrary predictors of possible outcome of disease. Early diagnosis is important. Patients need to be classified as minor, moderate (TI) and severe. Clinical diagnosis requires careful observations over a period of time with good record keeping of growth, sexual maturation and quality of life. Patients with haemoglobin (Hb) levels less than 7 gm/dl should be treated as transfusion dependent β -thalassaemia major to prevent complications that occur progressively with advancing age. Hb levels less than 7 gm/dl show patients are destined to be short, have splenomegaly and skeletal abnormalities. Pre transfusion mean Hb levels kept between 9-10 gm/dl by transfusion will suppress bone marrow activity and decrease iron absorption through gastrointestinal tract.

Keyword: Thalassaemia; Haemoglobin E; Genetic modifiers; Genotype – phenotype diversity; Natural history; Treatment