A rare case of alpha-thalassaemia intermedia in a Malay patient double heterozygous for α + –thalassaemia and a mutation in α 1 globin gene CD59 (GGC \rightarrow GAC)

ABSTRACT

A rare case of thalassaemia-intermedia involving a non-deletion alpha thalassemia point mutation in the 1-globin gene CD59 (GGC GAC) and a deletion +(-3.7) thalassaemia in which use of high performance liquid chromatography (HPLC) C-gram Hb subtype profile and DNA molecular analysis helped establish the diagnosis.

Keyword: Alpha-thalassaemia intermedia; Non-deletional alpha 1 globin gene CD59; Deletion- 3.7; HPLC; Molecular analysis