A rare case of alpha-thalassaemia intermedia in a Malay patient double heterozygous for α+ –thalassaemia and a mutation in α1 globin gene CD59 (GGC → GAC)

ABSTRACT

A rare case of thalassaemia-intermedia involving a non-deletion alpha thalassemia point mutation in the Ŭ1-globin gene CD59 (GGC Ÿ GAC) and a deletion Ŭ+ (-Ŭ3.7) 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-Ŭ3.7; HPLC; Molecular analysis