Sickle cell case series

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Abstract

Haemoglobin S (HbS) is caused by GAG→GTG mutation on codon 6 of β-globin gene. They are uncommon in Malaysia, mainly seen in immigrants, Indians and rarely Malays. This study reviewed the laboratory findings of patients with HbS. Seven patients were reviewed. HbS was quantified by HPLC and/or Capillary Electrophoresis. Molecular analysis was performed using Multiplex Ligation-dependent Probe Amplification (MLPA), Flow-through Hybridization (FTH) and Sanger Sequencing. Two African, three Malay and two Indian individuals aged between 2 to 31 years were selected. Five patients had homozygous HbS, one had S/β thalassemia and one had HbS trait. Haemoglobin level of homozygous and S/β thalassemia patients ranged from 5.8-10.9 g/dL and was normal in HbS trait. HbS, HbF and HbA2 levels without hydroxyurea ranged from 58.3-94.7%, 1.5-35.5%, 1.1-3.8% respectively among homozygotes; 67%, 27.2%, 4.2% in S/β thalassemia and 38.6%, 0.1%, 2.8% in the HbS trait. MLPA detected presence of HbS mutation (HBB:c.20A>T, rs334) in all cases. FTH performed on four cases showed one heterozygous HbS mutation, two homozygous HbS mutation and one compound heterozygous for HbS and Codon 41/42 (-TCTT) β-gene mutations. DNA sequencing confirmed the homozygosity, compound heterozygosity and carrier states of HbS mutation in all cases. HbS level varies widely between homozygous HbS patients, as it is affected by HbF level. MLPA detected presence of HbS mutation and FTH further identified the zyosity and concomitant beta-thalassemia.

Keywords: Haemoglobin S; MLPA; FTH

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