A rare hemoglobin variant, Hb Arya in a Malay woman

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Abstract

Inherited hemoglobin disorder has emerged as one of the most important genetic diseases in the world. These abnormalities can be categorised as thalassemia or hemoglobin variants. We report a rare case of hemoglobin variant in a 16 years old girl from Negeri Sembilan, who was asymptomatic with hemoglobin of 13.5 g/L, MCV 78.4 fl and MCH 26.3 pg. Hb analysis using High Performance Liquid Chromatography (HPLC) and Capillary Electrophoresis (CE) were done followed by DNA sequencing. The HPLC result showed Hb A2 3.4%, Hb F 0.3% and Hb A 64% with 19.6% of unknown Hb (X) and 0.6% of unknown Hb (Y) eluted at RT 4.57 min and RT 4.18 min respectively. CE showed a prominent peak at Hb S zone (20%) and small peak at Hb D zone (1.5%). DNA sequencing of the whole blood sample demonstrated a copy of an α globin gene variant in the HBA2 gene, identified as heterozygous state of α2 Codon 47 (GAC>AAC), Hb Arya. Hb Arya was first described in a family from Iran. It moves with Hb S on electrophoresis (alkaline pH), perfectly mimicking electrophoretic mobility pattern of Hb S carrier. As conclusion, the clinical significant of Hb Arya is indeterminate. The majority of hemoglobin variants fortuitously discovered is of minimal clinical interest; however, identification of this variant is important to avoid misdiagnosis of Hb S carrier and also to eliminate possibility of interaction with other thalassemic syndrome in the future.

Keywords: Hemoglobin variant; Hb Arya; HPLC; CE; DNA sequencing

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