De Novo Haemoglobin Sabine Presented With Chronic Haemolytic Anaemia: A Case Report

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Abstract

Haemoglobin (Hb) Sabine is an unstable Hb variant that causes moderate haemolytic anaemia in heterozygous state. This haemoglobin is the result of a point mutation of β-globin gene at codon 91(CTG)>(CCG). This study describes a two-years-old boy who presented with chronic haemolytic anaemia. To the best of our knowledge, this is the first case report of heterozygous Haemoglobin Sabine in Malaysia. The patient presented at the age of nine months with moderate anaemia, pallor and dark coloured urine. He suffered a haemolytic episode during a course of infection. Clinical examination revealed moderate hepatosplenomegaly. He had received transfusion twice during the course of his illness. Full Blood Picture showed moderate anaemia with evidence of haemolysis and presence of basophilic stippling. High Performance Liquid Chromatography showed increased HbF level (17.3%) with normal HbA2 level. Two small peaks were seen at retention time (RT) 4.56min (1.6%) and RT 4.87 min (5.4%). Capillary electrophoresis showed a small peak at zone 6 (1.1%) and another small peak at zone 4 (4.0%). Gel electrophoresis at alkaline pH showed a faint band just catodal to S band. DNA analysis for β–globin gene identified Heterozygous state of Codon 91 (CTG>CCG) Hb Sabine. A family study showed negative finding for similar mutation. High index of suspicion for suspected unstable haemoglobin is important to reach to the diagnosis. This case highlighted de novo presentation of unstable haemoglobin variant. Molecular analysis is needed for correct characterization of Hb Sabine.

Keywords: Haemoglobin Sabine, Unstable Haemoglobin, Haemolytic anemia

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