

Molecular characterisation of Haemoglobin Constant Spring and Haemoglobin Quong Sze with a combine-amplification refractory mutation system.

Abstract

Background: The interaction of the non-deletional α +- thalassaemia mutations Haemoglobin Constant Spring and Haemoglobin Quong Sze with the Southeast Asian double α -globin gene deletion results in non-deletional Haemoglobin H disease. Accurate detection of non-deletional Haemoglobin H disease, which is associated with severe phenotypes, is necessary as these mutations have been confirmed in the Malaysian population. Methods: DNA from two families with Haemoglobin H disease was extracted from EDTA-anticoagulated whole blood and subjected to molecular analysis for α -thalassaemia. A duplex polymerase chain reaction was used to detect the Southeast Asian α -globin gene deletion. Polymerase chain reaction-restriction fragment length polymorphism analysis was then carried out to determine the presence of Haemoglobin Constant Spring and Haemoglobin Quong Sze. A combine-amplification refractory mutation system protocol was optimised and implemented for the rapid and specific molecular characterisation of Haemoglobin Constant Spring and Haemoglobin Quong Sze in a single polymerase chain reaction. Results and Conclusions: The combine- amplification refractory mutation system for Haemoglobin Constant Spring and Haemoglobin Quong Sze, together with the duplex polymerase chain reaction, provides accurate pre- and postnatal diagnosis of non-deletional Haemoglobin H disease and allows detailed genotype analyses using minimal quantities of DNA.

Keyword: Combine-ARMS; Hb Constant Spring; Hb Quong Sze; Medical sciences.