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Molecular and Haematological Characterisation of Hb Malay in Malaysian Population

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Abstract

Haemoglobin (Hb) Malay is a variant haemoglobin with a mutation in the globin gene causing substitution of serine for asparagine at codon 19 of the beta globin chain. This variant Hb was first described in 1989 and was designated after the ethnic origin of the first case - Malay. It is characterised as beta^{+} thalassaemia phenotype. This study aims to determine the mutation spectrum of Hb Malay and its coinheritance with other thalassaemia/haemoglobinopathies in our population. A retrospective analysis was performed on 2413 diagnostic samples received by our laboratory in 2017. A total of 132 cases of Hb Malay were identified. Their demographic data including age, race, gender, red blood cell indices and Hb analysis findings were recorded. Mutation analyses of alpha and beta globin genes were performed using various methods which include multiplex ARMS PCR, GAP PCR and DNA sequencing. Our study showed that prevalence of Hb Malay in our population was 5.5%. Majority of the cases were Malays, 127/132 (96.2%), followed by Dusun, 2/132 (1.5%), Chinese, 1/132 (0.8%), Bajau, 1/132 (0.8%) and Orang Asli, 1/132 (0.8%). Ninety one were female. Their age ranges from 6 months to 53 years old with the mean age of 19. Four out of 132 patients were homozygous Hb Malay, 83/132 were heterozygous Hb Malay, 27/132 were compound heterozygous Hb Malay/Hb E, 8/132 were compound heterozygous Hb Malay/beta^{+} thalassaemia, 7/132 were compound heterozygous Hb Malay/beta^{0} thalassaemia followed by compound heterozygosity of Hb Malay/delta beta^{0} thalassaemia (1/132), Hb Malay/gamma delta beta^{0} thalassaemia (1/132), Hb Malay/Hereditary Persistence Foetal Haemoglobin (1/132) and Hb Malay/Hb Khon Kaen (1/132). Hb analysis findings showed wide ranges of HbA2 and Hb F level in high performance liquid chromatography and capillary zone electrophoresis depends on their types of inheritance. Molecular identification is mandatory for definitive diagnosis of Hb Malay since Hb analysis was not able to differentiate Hb A and Hb Malay, as it co-migrates. Identification of this variant haemoglobin is important in the mass screening programme for genetic counselling to prevent birth of thalasaemia children and also for clinical management of the patients.

Keywords: Hb Malay, thalassaemia, haemoglobinopathies

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