Hematological Parameter Evaluation in Different Types of Deletional Alpha-Thalassemia in Hospital Universiti Sains Malaysia

Samilawati Mohd Arifin, Marini Ramli, Rosnah Bahar*

Department of Hematology, School of Medical Sciences, Universiti Sains Malaysia, 16150 Kubang Kerian Kelantan, Malaysia

Alpha thalassemia is a common genetic disorder with more than 20% of the world population to be a carrier of some form of α–thalassemia, as estimated by The World Health Organization [1]. It has heterogeneity in its presentation and inheritance and characterised according to their deficient or absent in alpha globin chain involved [2]. The affected individuals may be asymptomatic with hypochromic microcytic anemia or in silent alpha thalassemia may have no clinical signs with normal to mild haematological changes [3]. Current voluntary thalassemia screening programme in Malaysia is mainly based on MCH level of less than 27 before molecular study for alpha thalassemia is done if Hb analysis showed normal results, to exclude alpha thalassemia. Accurate characterization of hematologic parameters is important for selection of appropriate molecular test to determine the carrier genotype, as the test is expensive, time-consuming and not always available. This study was aimed to evaluate the correlation of hematological parameters (Hb, RBC, MCV, MCH, RDW and platelet) with various types of deletional alpha-thalassemia among patients in HUSM.

A retrospective study on 216 samples sent to Molecular Unit, Haematology Laboratory, Hospital Universiti Sains Malaysia from 01.01.2014 till 31.12.2016 was conducted with permission from the Director of HUSM. The data was analysed using SPSS 22 software. Descriptive statistics including median and interquartile range (IQR) were used to look for the correlations. Results obtained from this study showed the median alpha thalassemia are significantly difference in RBC, MCV and MCH ($p < 0.001$) between deletional alpha thalassemia group and normal group. There is also significant difference in Hb, MCV, MCH, RDW and platelet level between different genotypic deletional alpha thalassemia ($p < 0.05$). In alpha thalassemia group, the level of RBC was significantly higher with median of 5.4 $X10^6$/UL, 95% CI 6.0- 4.8 $X10^6$/UL compared to normal group, 4.8 $X10^6$/UL and CI 5.4- 4.2 $X10^6$/UL. Median MCV was 65.4 fL (95% CI 76.9-53.9) lower compared to normal group, 70.8 fL (95% CI 60.9 fL). Similarly, MCH level was lower in alpha thalassemia group,

median 20.8 pg (CI 24.3- 17.3 pg) compared to normal group, median 23.3 pg (CI 27.7- 18.9 pg). One gene deletion patient has normal Hb to mild anemia compared to patient with two and three genes deletion. MCH, MCV is lower in two genes deletion compared to one gene and RDW level is significantly higher in three genes deletion compared to one and two genes deletion. The RBC level shows no significant in between groups of deletional alpha thalassemia patients. We observed that the median Hb for one gene deletion was 12.4 g/dl with a confident interval (CI) of 95% of 9.9-14.5 g/dl, whilst the measure for both two and three gene deletion were 11.3 and 9.3 g/dl, CI 12.8- 9.8 g/dl and 10.1-8.5 g/dl respectively. The level of MCV in one gene deletion was 73.3 fL, (95% CI 84.4- 62.2 fL) whilst for two and three gene deletion were 64.4 fL and 55.4 fL respectively (CI 70.7-53.9 and 65.4- 45 fL respectively). MCH level was higher in one gene deletion compared to two genes with median of 24.4 pg and 20.3 pg respectively (95% CI of 27.7-20.7 pg and 22.7-17.9 pg respectively). Anisopoikilocytosis are marked in two gene deletion compared to one gene deletion with median RDW 16.5% and 14.2 % respectively.

From this observation, the MCH level in patients with deletional alpha thalassemia showed value of less than 25 pg. We can selectively proceed samples with MCH value of less than 25 pg for further molecular test for definitive diagnosis of alpha thalassemia. While the value of RBC and MCV may help as an effective screening for alpha thalassemia cases. The level of Hb, MCV, MCH and RDW may give a rough guide for differentiating between numbers of genes deletion in alpha thalassemia patients. However, these observations should be interpreted cautiously, as possibility of underlying iron deficiency state and co-inheritance of β-thalassaemia and other haemoglobinopathies was not ruled out in these samples.

Keywords: thalassemia screening, haematological parameters, deletional alpha-thalassemia

*** Correspondence:** rosnahkb@usm.my

Acknowledgements

This study was funded by Short Term Grant (304/PPSP/61313113) by Universiti Sains Malaysia.

References:

1. Weatherall, D. and J. Clegg, *Inherited haemoglobin disorders: an increasing global health problem.* Bulletin of the World Health Organization, 2001. **79**(8): p. 704-712.
2. Leung, W., et al. *Alpha-thalassaemia*. in *Seminars in Fetal and Neonatal Medicine*. 2008. Elsevier.
3. Wang, C., et. al. *Measurements of Red Cell Parameters in-Thalassemia Trait: Correlation With the Genotype*. Laboratory Hematology, 2000. **6**(3): p. 163-166.