Abstract

This study aimed to determine the association between the hematologic parameters such as Hb, MCV and MCH values and alpha globin mutations in Filipino Alpha-Thalassemics. Highly suspected Filipino Alpha-Thalassemics (n=61) ages 1 to 59 years old with MCV and MCH values below 80 fl and 27 pg, and with marginal/below marginal Hb levels were included in the study. The patients underwent DNA extraction and Alpha-Globin-StripAssay mutational analysis. The three-gene deletions, (-α 3.7/--SEA), (-α 3.7/--FIL) and (-α 4.2/--SEA), the two-gene deletions, (--FIL/αα), (--SEA/αα), (-α 3.7/ α 3.7), and (-α 3.7/ α 4.2), the one-gene deletion (--α 3.7/αα), and the (α2 cd 59/αα) were found in 18.03%, 72.13%, 3.28%, and 1.64% of the patients tested, respectively. Fifty-eight (58, 95.08%) patients with low levels of MCV and MCH were found positive for the alpha globin mutations. It was also found that patients with three gene deletions (11/11, 100.00%) consistently have low levels of Hb in comparison with patients with two gene deletions (37/44, 84.09%). These results suggest that (--FIL/αα), and (--SEA/αα) are the most prevalent mutations in the patients tested and are highly associated with low levels of MCV and MCH. Sequencing analysis is advised for the three patients with normal genotypes. Further investigation is recommended to check the usability of Hb levels, in combination with MCV and MCH levels, in suggesting the number of affected alpha globin genes.

Keywords: alpha-thalassemia; red cell indices; hemoglobin levels; Filipinos

*Author for Correspondence