Molecular Genetics, Genomics, Mechanisms of Diseases

AB088. Prevalence of alpha thalassemia mutations in Filipino patients

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Background: Alpha (α)-thalassemia results from the absent or reduced synthesis of the α-globin subunit of hemoglobin. Mutational variants in the *HBA1* and *HBA2* genes, which code for α-globin, have been reported and cause varying degrees of disease severity. These variants are unique for every population. Local data on alpha-globin gene mutations in Filipino alpha-thalassemia is currently lacking.
Methods: This study aimed to identify the prevalence of common α-globin gene mutations in Filipinos highly suspected for α-thalassemia through evaluation of CBC results, red cell indices and upon ruling-out iron deficiency anemia. Sixty five [65] patients underwent DNA extraction and Alpha Globin StripAssay mutational analysis.
Results: The four gene deletion, (--SEA/--SEA), the three gene deletion, (-α 3.7/--FIL), (-α 3.7/--SEA), (-α 4.2/--SEA), the cis two gene, (--SEA/αα), (--FIL/αα), the trans two gene deletion, (-α3.7/-α3.7), the one gene deletion, (-α3.7/αα), and the (α2 cd 59/αα) were found in 1.54%, 21.54%, 56.92%, 1.54%, 6.15% and 1.54% of patients, respectively.
Conclusions: These results suggest that cis two gene deletions identified are prevalent in Filipinos tested. The high prevalence of cis two gene deletion of *HBA1* and *HBA2* in this study is important as these alleles may increase the risk of Hb H and Hb Bart's hydrops fetalis cases in the population.

Keywords: Alpha thalassemia; Philippines; prevalence

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