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AB089. Prevalence of the most common β-globin gene mutations in Filipino β-thalassemia patients

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Background: Beta (β)-thalassemia is an autosomal recessive disorder characterized by reduced or absent β-globin chain synthesis resulting in decreased hemoglobin in red blood cells (RBC) and consequently, microcytic, hypochromic anemia with varying degrees of severity. The disease is a result of mutations in the human β-globin (*HBB*) gene. This genetic disorder is considered an emerging global health burden, especially in Southeast Asia, as 50% of world carriers of β-thalassemia, corresponding to 40 million people, are found in this region alone. Thus, prevalence of *HBB* gene mutations should be identified in our population. This study aimed to determine the prevalence of common β-globin gene mutations in Filipino β-thalassemia patients.

Methods: Fifteen [15] patients underwent DNA extraction and Beta globin strip assay mutational analysis.

Results: The HbE (20%) was the most common allele detected using the strip assay method. Interestingly, a suspected unnamed deletion was detected in 13.33% of patients tested. A high number of normal genotype was also detected which composed majority (60%) of the patients.

Conclusions: The result of the study suggests the limitation of strip assay method in the detection of Filipino *HBB* gene mutations. Further analysis of the samples tested via the sequencing method is recommended.

Keywords: Beta thalassemia; Filipino; mutations

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