Mutational profile of Beta thalassemic population in the Northeast of Algeria

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Introduction: Beta-thalassemia (β-thal.) is one of the most common recessive monogenic disorders worldwide with a higher prevalence among Mediterranean, Middle Eastern and Indian populations. More than 300 β-thalassemia alleles are currently reported in the HbVar database in order to evaluate the mutational profile of β-thalassemic populations in the Northeast of Algeria.

Materials & Methods: Sixty patients from Dorban hospital, Annaba, Algeria were enrolled in this study. A direct sequencing of the HBB gene of beta globin was performed. The β-thalassemia mutation was identified by automated sequence analysis performed on an ABI Prism 3130 Genetic Analyzer (Applied Biosystems) using the fluorescent dideoxy termination method (Big Dye-Terminator Cycle Sequencing Kit; Applied Biosystems). Statistical analyses were performed with SPSS 22.0 software.

Findings: The three principal β-thalassemic mutations detected were nonsense mutation at codon 39 (HBB: c.118C>T), IVS-I-110 (HBB: c.93-21G>A), IVS-I-1 (HBB: c.92+1G>A). A novel β-thalassemia mutation was characterized for the first time in a patient with white blood cell (WBC) count 11.9 (109/L), Hb 7.3 g/dL, and platelet count 455.0 (109/L). The nonsense mutation codon 39(C>T) is widespread in Algeria with a frequency 25.94% (11) 27.6% (15) and is more common in the west and decreases in center to be predominant in the East. Although, the molecular defects at the origin of this pathology are of extreme diversity, each population is characterized by a group of 4 to 5 mutations that is specific to it.

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