32nd World Pediatrics Conference

December 04-05, 2019 | Barcelona, Spain

The first association of HB Knossos: (HBB: c.82G> T) with (HBB: c.118C> T) mutation causes thalassemia homozygous in Algerian children

Belhadi Kamilia[1], **Bendaoud Fadhila**[1], **Benhouda Afaf**[1], **Djaara Hayet**[1], **Gribaa Moez**[2] and **Ben Charfeddine Ilhem**[2]

[1]University of Batna, Algeria

[2]Farhat Hached University, Tunisia

Beta-thalassemia is the most common disease among hemoglobinopathies in Algeria. Mutations found in Algerian beta-thalassemia patients constitute a heterogeneous group, consisting mostly of point mutations. Only in very rare cases did deletions or insertions cause affected or carrier phenotypes. Hb Knossos (HBB: c.82G> T) is a rare variant. In this study, we aimed to investigate the effect of compound heterozygosis for Hb Knossos (HBB: c.82G> T) and (HBB: c.118C> T). To our knowledge, this is the first report of such a combination related with beta-thalassemia major phenotype in a Algerian family, we used the minisequencing assay as a rapid screening procedure to identify most common HBB genetic variants and direct DNA sequencing to detect the rare mutations of HBB gene. Heterozygous inheritance of the mutation results in severe beta-thalassemia phenotype. The proband was a 13-year-old boy when first studied. He was referred because of severe anemia. Hematological analysis of the reveals Hb 7.2 g/dl with microcytosis of 71.1fl, hypochromia 25pg and the number of red blood cells is 2.9, 106/mm^3. In addition, a significantly secondary thrombocytosis and leukocytosis were reported in patient. Electrophoresis of hemoglobin in an alkaline medium shows Hb A2 = 4% HbF = 65% and blood smear confirms microcytosis hypochromia and showing the presence of many dacryocyte with hyper eosinophilia. The combination of these mutations Hb Knossos (HBB: c.82G> T) and (HBB: c.118C> T) causes the beta-thalassemia major phenotype and this is important for genetic counseling.