Caracterización clínica y epidemiológica de población pediátrica costarricense con alfa-talasemia [

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text (article)

Analítica

Objective: Describe the clinical and epidemiological characteristics of patients with alpha thalassemia in the Hematology Service of a national pediatric hospital. Methods: Cross-sectional descriptive observational study. 60 patients from the Hematology Service of the National Children's Hospital "Dr. Carlos Sáenz Herrera", Costa Rican Social Security Fund, from January 1, 2018 to January 31, 2019, with hematometric indices suggestive of alphaThalassemia, with AA standard hemoglobin electrophoresis with normal or decreased HbA2 with ages between 0 months and 12 years 11 months old. Molecular analysis: Identification of 21 mutations and deletions that includes the detection of the most frequent deletions/mutations for the alpha globin gene: 3.7, 4.2, 20.5, MED, FIL, SEA, THAI, anti-3.7 tripling, HbConstant Spring, HbQuonSze, Hb Adana, HbKoya Dora, HbIcara, HbPakse, a2 poli A-1/2, a2-cd142, a1-cd14, a2-init-cd, a2-cd19, a2-IVS1, a2-cd59. The reverse hybridization PCR amplification method of genomic DNA in peripheral blood leukocytes of patients isused. Results: Of the 60 cases studied, in 44/60 (73%) cases the disease is confirmed. The average age at diagnosis for these cases is 4.9 years (SD 3.0), the female sex predominated in 52.3% of the cases. Guanacaste reported the highest prevalence of the disease. The deletional genetic defect -3.7 Kb was the most frequent genotype and the phenotype in 77.2% of the cases was he silent carrier of alpha thalassemia. In 84.1% of subjects positive for alpha thalassemia, it correlated with hypochromia, microcytosis, and erythrocytosis in the initial blood count. 9% of the cases showed the coexistence of alpha thalassemia and iron deficiency anemia. Conclusions: This study demonstrates that the findings of erythrocyte indices that indicate hypochromia and microcytosis with increased erythrocyte count; normal iron indices and a normal hemoglobin (AA) electrophoresis pattern suggest that they should be studied molecularly for alpha t
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