

α -Thalassemia (3.7 kb deletion) in a population from the Brazilian Amazon region: Santarém, Pará State

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ABSTRACT. The ethnic composition of the Brazilian population favors high frequencies of the $-\alpha 3.7$ deletion, responsible for α -thalassemia, because this mutation is very common in African populations. In spite of its importance, this hemoglobinopathy has been poorly investigated in Brazil, especially at the molecular level. We investigated the prevalence of the $-\alpha 3.7$ mutation in 220 individuals attended at the Municipal Hospital of Santarém, in the State of Pará. These patients were distributed into three different groups: i) 103 individuals with anemia who had microcytosis and hypochromia, ii) 11 individuals without anemia who had microcytosis and hypochromia, and iii) 106 individuals with no hematological alterations. We examined the usefulness of investigating α -thalassemia carrier status for microcytosis. Among the 103 patients with anemia, 20 (19.4%) were heterozygotes ($-\alpha 3.7/\alpha\alpha$) and one (1.0%) was a homozygote ($-\alpha 3.7/-\alpha 3.7$). Among the 11 patients without anemia, one heterozygote ($-\alpha 3.7/\alpha\alpha$) was identified; in the third group, composed of normal individuals (106 samples), deletion $-\alpha 3.7$ was found in seven samples (6.6%), all of which were heterozygotes ($-\alpha/\alpha\alpha$). These frequencies are within the expected range, given available data on the distribution of this hemoglobin disorder in human popu-

lations and the ethnic composition of the population of Santarém. We found that α -thalassemia is a common cause of microcytosis, given that a high proportion (19.2%) of the microcytic population carried α -globin gene deletions.

Key words: Alpha-thalassemia; $-\alpha^{3.7}$ deletion; Brazilian population