

Carta ao editor / Letter to editor

Thalassemia screening in Brazil – Results for 20 years *Talassemia no Brasil – Resultados de 20 anos de estudo*

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To editor

Brazil is the largest country in South America, with a population, according to the government consensus of 2000, of approximately 180 millions. The original inhabitants were native Indians, but five hundred years ago the continent was colonized by Europeans, specifically by the Portuguese, with some Spanish and Italians. Slaves were also brought from Africa. Thus, the population is an admixture of races: in the north there is a predominance of native Indians, in the northeast there is a high percentage of Negroes and their descendents and in the south and southeast there is a great influence of Caucasians. More recently there was substantial immigration from Asia.¹ Some works with genetic markers in different parts of Brazil show a trihybrid population with components of Indians, Negroes and Caucasians.² Specifically for Caucasians the main components are Italians, Portuguese and Spanish. This genetic diversity is seen in hemoglobinopathies with genotype associations and many phenotypes, in particular for thalassemia, where the high frequency of alpha and beta thalassemic traits very frequently cause an association of alpha and beta thalassemys as well as other situations.^{3,4} So, for thalassemia studies in Brazil it is very important to consider the make up of the population, the method of analysis applied, the type of thalassemia inherited and the influence of the environment and other genetic diseases.

During the last twenty years the hemoglobin laboratory in Unesp, has performed hemoglobin screening programs in different populational groups, including students and blood donors. This analysis using both electrophoretic and chromatographic procedures has shown a consistent picture of thalassemia in Brazil with the analysis of 41,235 blood samples.⁵⁻⁸

Alpha thalassemia is the most frequently inherited disease in the world, including the Brazilian population, affecting mainly Asian descendents and some African groups. Data related to this thalassemia in the Brazilian native Indian population, who are descendents of Asiatic races, is scarce and requires further investigation.⁹ Until ten years ago, due to inadequate diagnostic methods, the frequency of alpha thalassemia was underestimated. Some studies show a frequency of 10-12% in some regions with 25% in specific groups.^{4,5} In part, this difference seems to be related to the admixture of the population. In a study of alpha thalassemia traits diagnosed by classic methodologies in the state of Sao Paulo, molecular analysis shows a high prevalence of the alpha 3.7 deletion.⁴

For beta thalassemia, populational studies to determine the real prevalence in Brazil are scarce and are related to specific groups. We do not know the real prevalence for each state of Brazil. In Caucasian descendents in the state of Sao Paulo the rate is estimated at almost 3%.¹⁰ Thalassemic syndromes more frequently affect people in the southern and southeastern regions, especially because of the racial characteristics of these populations. Many hemoglobin variants have been described in Brazil, and with the significant admixture, the existence of interactions among thalassemic forms and variants are frequent.^{7,10} The most common beta thalassemia mutation, predominant in Mediterranean populations including the Italian, Spanish and Portuguese, is common in inhabitants from the state of São Paulo. Molecular studies in this population show the high frequency of Mediterranean mutants such as the CD39 mutation. In the same study, with samples from the northeastern region, we observed a high frequency of a mutation which was rarely seen in the southeastern population, the IVS1-6 mutation.

In many Brazilian states access to health services is difficult because of the long distances involved and information about thalassemia is restricted to a few centers. It is necessary to bring up-to-date the mechanisms of the services provided in all the states of Brazil, including population screening programs for thalassemia with the aim of diagnosing heterozygotes. As a result of these policies, it will be possible to provide education for these individuals and to give assistance to homozygous families who do not have correct diagnoses or clinical management. It is important to remember that Brazil has a special characteristic: "There are many small countries inside this big country" and that thalassemia screening programs need to consider the characteristics of each specific population due to the differences in the language, interpretation and access to health facilities.

The national neonatal screening program for hemoglobinopathies in Brazil mainly focuses on Hb S. In Sao José do Rio Preto, a small city in the state of São Paulo, we have been developing a pilot neonatal screening program for all hemoglobinopathies including thalassemia. We analyzed 6.050 cord blood samples of neonates in a school hospital during six years, using classic

methodologies that included electrophoretical procedures, cytological tests and HPLC. We found 15% of hemoglobinopathy carriers with high frequencies of alpha thalassemia and Hb S. Beta thalassemia was found in 1.96% of the samples tested. All cases were confirmed after six months.¹⁰ With this program it was possible to provide genetic counseling, to establish social and clinical management and to give support to the families. Using this screening program education for these individuals can be provided and assistance to homozygous families who do not have correct diagnoses or clinical management.

This study also shows the internal migration within Brazil that was reflected in the phenotypes of hemoglobinopathies observed over the years. The Northeastern region of the state of São Paulo received many groups from other areas of Brazil to work in sugar cane plantations in 1999 and 2000. It is important to remember that Brazil has special populational characteristics and the thalassemic screening programs need to consider the characteristics of each specific population due to differences in the access to health facilities.

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