Neonatal screening in the state of Piauí: an urgent need - a study on the prevalence of sickle cell disease in newborns

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Regarded as the most prevalent genetic disease in the world, the frequency of sickle cell anemia is estimated at between 25% and 40% in African countries. In the field of public health, the emphasis on sickle cell anemia as an ethnical-racial disease is based on three aspects related to this pathology which characterize a greater susceptibility of the black and mulatto populations: geographical origin, genetic etiology and statistics on prevalence[1-3].

Sickle cell disease involves a series of different genetic alterations that cause from minor to serious symptoms and a high mortality rate. Hemoglobin S (Hb S) is a mutation of the β-globin gene with the exchange of a nitrogen base at position 6, whereby glutamic acid (GAG) is replaced by valine (GTG), resulting in the expression of the β^s gene and a structural change in its function.

In 2001, the Ministry of Health decree N° GM/MS Nº 822 established the National Neonatal Screening Program (PNTN) that made the diagnosis, treatment and monitoring of sickle cell disease and other hemoglobinopathies obligatory throughout the country. According to PNTN data around 3000 children per year are born in Brazil with sickle cell disease and 180,000 have the sickle cell trait. The disease in Brazil is distributed heterogeneously with an incidence of 0.3% of the population; however, in part due to the poor social profile[1], there is a high prevalence in the northeastern region, chiefly in the states of Bahia, Maranhão and Piauí, where the proportion of African descendants is higher[4,5].

It is of great concern that the state of Piauí, the home to the earliest traces of life in the Americas and registered as the state with the fourth highest self-declared black population, has no indicators in relation to the genetic inheritance of hemoglobin S in newborn babies.

As such, a study was performed to provide support for the implementation of phase II of the newborn screening test, thereby strengthening and consolidating the Sickle Cell Disease Program in the state of Piauí.

The ethical aspects of the study were approved by the Research Ethics Committee of the Universidade Federal do Piauí (# 0181.0.045.00008).

Table 1 - Variant hemoglobins in newborns and maternal ethnicity in the city of Teresina, PI in 2008-2009

Variant Hb	Nº	Ethnicity (%)			
		Black	Mulatto	White	unknown
Hb FS	5	0.5	0.6	0.0	0.0
Hb FAS	39	3.8	3.6	5.0	6.0
Hb FAC	9	0.9	0.9	0.7	0.0
Hb FAD	2	0.2	0.2	0.0	0.0
Hb FA	974	94.0	94.0	94.0	94.0
Others	5	0.5	0.6	0.3	0.0
Total	1034	100.0	100.0	100.0	100.0

Hb FS: homozygous for hemoglobin S; Hb FAS: sickle cell trait; Hb FAC: hemoglobin C trait; Hb FAD: hemoglobin D trait; Hb FA: normal hemoglobin

Table 2 - Percentage of variant hemoglobins in newborns in northeastern capital cities

City	Total Sample	Hb FAS (%)	Hb FS (%)	Year of survey
Teresina (PI)	1034	3.8	0.5	2008-2009
Salvador (BA)[6]	590	9.8	0.2	2000
Natal (RN)[7]	1903	1.5	0.05	2001
Fortaleza (CE)[8]	389	3.8	0.2	2001-2002
Recife (PE)[11]	1998	5.1	0.2	1996-1997
João Pessoa (PB)[10]	1006	0.2	-	1993

Hb FS: homozygous for hemoglobin S; Hb FAS: sickle cell trait

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Tests to detect hemoglobins were carried out using high-performance liquid chromatography (HPLC) and the Variant II (BioRad) system.

On including hemoglobinopathies in the PNTN, the Ministry of Health took an important step towards acknowledging the relevance of the matter to public health in Brazil.

Adorno et al. reported the prevalence of variant hemoglobins in 590 newborns in the city of Salvador; 0.2% for Hb FS and 0.9% for Hb FSC, thus demonstrating the high rate of sickle cell disease in the population, as well as the high prevalence (9.8%) of the sickle disease trait[6]. The city of Teresina registered rates of 0.5% for Hb FS and 3.8% for Hb FAS.

A 2006 survey by Pinheiro et al. of 389 newborns in the city of Fortaleza registered figures of 0.2% for Hb SS and 3.8% for Hb AS[8]. A survey conducted of 1903 newborns in the city of Natal revealed a prevalence of only 0.05% for Hb SS and 1.5% for Hb AS[7]. The city of Teresina registered 0.5% of Hb FS and 3.8% of Hb FAS, a fact which may be explained by the fact that the state of Piauí has a significantly high black population.

These figures reflect need for the immediate implementation of phase II of the PNTN in the state of Piauí and the creation of a multi-disciplinary care network for sickle cell patients.

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