Prevalence of sickle cell trait and sickle cell anemia in two Brazilian quilombola communities from Southwest Bahia State

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Abstract—Sickle cell disease (SCD) is the most common hereditary hematologic disease in the world and prevalent among people of African descent. Large part of this prevalence is found in remaining quilombola populations, which still face difficulties related to health because of eventual discrimination and/or marginalization for ethnic, cultural and social reasons. Thus, the objective of the present study was to evaluate the prevalence of SCD - sickle cell trait and sickle cell anemia - in the population aged 10 years or older of the Lagoa de Maria Clemência and Oiteiro communities, Vitória da Conquista, Bahia, Brazil. To this end, a cross-sectional study was carried out with 149 individuals. Blood samples were collected by venipuncture and put in tubes containing EDTA. Initially, part of the blood was incubated with sodium metabisulphite to observe the occurrence of sickling under optical microscope. Samples with sickled erythrocytes were referred for hemoglobin electrophoresis with High Performance Liquid Chromatography. Data analysis showed that 4.1% of the population has SCD; of these, 3.5% have sickle cell trait and 0.7% have homozygous hemoglobin S, and 66.7% were female and 33.3% male. The prevalence of SCD in the quilombola communities studied was similar to those found in other communities of northeastern Brazil. Because these individuals have greater hereditary susceptibility to SCD, and also because of their low socioeconomic conditions, constant monitoring of these populations is necessary to identify individuals with SCD and inform them of the possible risk of birth of children with sickle cell anemia resulting from the marriage between individuals with sickle cell trait.

Keywords—sickle cell disease, sickle cell anemia; sickle cell trait; quilombola communities; black population.

I. INTRODUCTION

Sickle cell disease (SCD) is the most common hereditary hemoglobinopathy in the world. The worldwide prevalence of hemoglobin disorders is 7%, largely represented by thalassemia and sickle cell disease (WANG, 1999; WEATHERALL, 2001). This disease involves a range of different genetic modifications, ranging from mild, sometimes asymptomatic, to severe forms with a high mortality rate. The mutation occurs on chromosome 11 and results in the replacement of glutamic acid by valine at position 6 of the N-terminal end in the β-chain of globin, generating hemoglobin S and thus causing a structural and functional alteration in the protein. Individuals with homozygous hemoglobin S (HbSS) have a more severe condition; in turn, those with heterozygous hemoglobin S (HbAS) tend not to present clinical manifestations (DI NUZZO, 2004).

Red blood cell sickling occurs in conditions of low blood oxygen tension, such as those present in physical and mental stress, infection, dehydration, and changes in body temperature. When erythrocytes are sickle-shaped, they lose their elasticity and tend to cause vaso-occlusion.
in the microcirculation and, consequently, induce ischemia and/or tissue necrosis (OLIVEIRA et al., 2013). Clinical presentations resulting from this process can be mild to severe pain, lower limb ulcers, acute chest syndrome, priapism, retinopathies, stroke, renal failure, among others (FÉLIX, SOUZA and RIBEIRO, 2010).

Historically, SCD originated in the African continent and was brought to America by the time of colonization, through about 3-4 million African slaves (OLIVEIRA et al., 2013). Despite miscegenation throughout Brazil, there is a higher prevalence of SCD in the Northeast due to the increased number of black ancestors. Large part of this prevalence is found in remaining quilombola populations, which still today face difficulties related to health because of eventual discrimination and/or marginalization for ethnic, cultural and social reasons ZAGO et al., 2002). Furthermore, epidemiological information specifically pertaining to quilombola people in health information systems is scarce, demonstrating the importance of conducting epidemiological studies with this population group (GOMES et al., 2013).

In this context, the objective of the present study was to investigate the prevalence of sickle cell trait (SCT) and sickle cell anemia (SCA) in two quilombola communities in Vitória da Conquista (Bahia, Brazil), called Lagoa de Maria Clemência and Oiteiro.

### II. MATERIAL AND METHODS

This is an experimental cross-sectional study conducted with the residents of the Lagoa de Maria Clemência and Oiteiro Quilombo Remnants, Vitória da Conquista, Bahia, Brazil. This article is part of a larger project entitled: Genetic counseling for patients with sickle cell trait and identification of chronic diseases as well as their risk factors in quilombola residents of southwest Bahia, whose activities were submitted and approved by the Research Ethics Committee of the State University of Southwest Bahia, under CAAE number 73479917.6.0000.0055.

All residents were invited and spontaneously decided to participate in the project. All the people who wished to participate were included in the study of SCT and SCA, except those under 10 years of age. These individuals signed the Informed Consent Form (Appendix 1) or the Consent Form in the case of underage (Appendix 2) (in the case of those who could not write, fingerprints were collected). Volunteers were also interviewed about their socioeconomic status and risk factors for SCT and SCA (Appendix 3).

Blood samples (about 5 mL) were collected from patients after 10 - 12 hour fast by venipuncture, through a vacuum collection system using a needle (venipuncture) in 1 tube containing EDTA. After collection, the material was stored in refrigerated coolers at 4 °C until analysis at the Medical Biochemistry Laboratory of the Southwest Bahia State University. An aliquot of 50 µl of blood was diluted in 100 µl of physiological solution, followed by the addition of 100 µl of sodium metabisulphite (2%). This mixture was placed on a microscopy slide and spread within 4cm². The slides were prepared with a cover glass, sealed from contact with air, and examined under a 40x objective lens in a microscope after 1 and 24 hours (NAOUM, 1987). The analysis was performed by two independent examiners. When sickling was observed, the blood sample was sent to an outsourced clinical laboratory and analyzed by hemoglobin electrophoresis to identify HbS homozygosis or heterozygosis (CHINELATO-FERNANDES and DOMINGOS, 2006; WOITOWICZ et al., 2010).

### III. RESULTS AND DISCUSSION

The total population of this study was 149 individuals, distributed in 80 (53.7%) women and 66 (46.3%) men. The average age was 42.2 years. The frequency of SCD was higher in females (66.7%) than in males (33.3%) (Table 1).

Regarding ethnicity, 49, 44.3, 4.7 and 2% declared to be black, brown, white and indigenous, respectively. All individuals with SCD were black or brown. None of the white or indigenous participants had SCD (Table 2).

| Table 1. Frequency and percentage of DF in individuals from Quilombos. |
|-----------------|-----------------|------------------|-----------------|
|                 | Sickle cell trait | Sickle cell anemia | No Sickle cell anemia | Total |
| Women           | 4 (80%)          | 0 (0%)            | 76 (53.1%)        | 80 (53.7%) |
| Man             | 1 (20%)          | 1 (100%)          | 67 (46.9%)        | 66 (46.3%) |
| Total           | 5 (3.4%)         | 1 (0.7%)          | 143 (96%)         | 149 (100%) |

The numbers outside and inside the parentheses indicate the number of people for a given analysis and the percentage of the number of people relative to the total for that analysis, respectively.
When asked whether they were aware of their condition, that is, SCT or SCA, 64.4% of the individuals did not know whether they had or not SCD, 33.6% reported that they had not been diagnosed, and 2% had previous knowledge about the diagnosis of SCD. Thus, of the participants with positive samples for SCT, only 33% were aware of this information.

This study showed a prevalence of 3.4% of SCT in the community studied (Table 1), which is similar to surveys in other quilombos of the Northeast region of Brazil, whose prevalence is 4.1% (PEDROSA, FERREIRA and OLIVEIRA, 2004). In Brazil about 2 to 8% of the population has SCT, and the states with the highest prevalence are Bahia (5.3%), Pernambuco (4%) and Rio de Janeiro (4%). On the other hand, the southern region of Brazil has the lowest prevalence, such as the state of Rio Grande do Sul (2%) (CANÇADO and JESUS, 2007).

The prevalence of HbAS in European quilombola communities is moderate compared to African, Asian and Middle Eastern countries. In Africa, as in Brazil, the findings are heterogeneous, probably due to miscegenation. The people affected by SCT in Ghana corresponds to 13.3% of the total population, while in Kenya this figure is 3%. In certain regions of Asia and the Middle East, HbS is found in 25% and 11% of the population, respectively (OFORI-ACQUAH and OHENE-FREMPONG, 2007).

Sickle cell anemia is the most common hereditary disease in Brazil. Its prevalence is higher in the black population, affecting a proportion from 0.1% to 0.3% (RAMALHO, 1986). However, due to miscegenation in the country, there is a growing trend in the number of people affected. In this study, homozygous HbS presented a frequency of 0.7% in the quilombola communities of the Southwest region (Table 1). In the quilombola communities of the state of Piauí, the average found was 0.5% of the inhabitants (SOARES et al., 2017).

In the socioeconomic aspects and risk factors for SCD investigated in the interview, it was observed that 64.4% of the individuals were unaware of the heterozygosis for HbS. A study in the Patioba community of Sergipe state showed that 72.3% of the inhabitants did not have adequate knowledge about SCT and SCA; and another study conducted in quilombola communities in Piauí showed that 81% of the residents did not have knowledge about SCD (MENEZES et al., 2015; SOARES et al., 2017). The lack of knowledge about the health condition reflects directly on the deficiency and/or absence of public policies directed to the screening of SCD.

The information of this population about SCD brings with it a challenge for recognition and medical follow-up. This population has no access to genetic counseling, and therefore, they are unaware of the risk of consanguineous marriage and the morbidity linked to SCA. Therefore, government programs aimed at quilombola communities are needed to disseminate information about SCD and reduce the morbidity and mortality of SCA.

It is noteworthy that the adherence of only 30% of the population of both quilombola communities may be an important limitation of our study. This implies that the probability of selection bias becomes greater, mainly due to the fact that people who participate in this type of activity are usually those with greater knowledge about their health. In addition, the distance between the sample collection site and some farthest residences may favor low adherence.

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