SICKLE CELL ANEMIA: A DIAGNOSTIC AND TREATMENT OVERVIEW

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Sergiane de Jesus Rocha Mendonça[1], Ronice Reis da Silva[2] and Mirela Sofia Mendonça de Araújo[3]

ABSTRACT

Sickle cell disease, a genetic and hereditary condition with the highest prevalence in the black population, is characterized by a change in the morphology of erythrocytes. The objective of this study was to outline the diagnostic and treatment strategies employed in patients with Sickle Cell Anemia treated at the Hematology and Hemotherapy Center of São Luís - MA. As a methodology, a literature survey and field study were carried out, in which data were collected through questionnaires applied to patients undergoing treatment. Data analysis revealed a predominant age range between 5 and 10 years in patients with the disease, with a majority consisting of females. Regarding the diagnosis, approximately half of the participants had the condition identified early, before the first year of life, through the heel prick test. Regarding therapeutic management, it was observed that about 79% use a combination of antibiotics, analgesics and folic acid, and that 75% have already required blood transfusions. The most common clinical manifestations of the interviewed patients were severe joint pain, skin pallor, jaundice, fever, and fatigue. It was found that more than half of the patients do not live in the capital of Maranhão, who also mentioned as the main obstacles in treatment the difficulty in free access to medicines and the absence of treatment options in their cities of origin. Thus, it becomes evident the need to expand the network of Hematology and Hemotherapy Centers in Maranhão, aiming to serve the population of patients with Sickle Cell Anemia residing outside São Luís and, consequently, promote an improvement in the quality of life of these individuals

Keywords: Sickle cell disease. Hemoglobin S. Neonatal screening.

[1]Dr. in Biodiversity and Biotechnology
Federal University of Maranhão
sergiane.jrm@ufma.br
https://orcid.org/0000-0002-7393-9858
http://lattes.cnpq.br/7136199043365924
[2]Graduated in Natural Sciences – Biology
Federal University of Maranhão
ronicereisdassilva@hotmail.com
[3]Undergraduate student in Psychology
Federal University of Maranhão
mirela.sofia@discente.ufma.br

INTRODUCTION

Sickle Cell Disease, a hereditary condition with significant occurrence in Brazil, originated in the African continent, being introduced in the Americas by the migratory flow. This genetic hemoglobinopathy, characterized by high morbidity and mortality, manifests as a severe chronic hemolytic anemia, resulting from a mutation in hemoglobin A, which leads to the production of hemoglobin S, with distinct physicochemical properties (BRUNETTA *et al*., 2010).

According to data from the National Neonatal Screening Program in Brazil, the prevalence of children diagnosed with Sickle Cell Disease had an incidence of 3.75 per 10,000 live births, between 2014 and 2020 (BRASIL, 2025), with an estimated number of patients in Brazil with the disease between 60,000 and 100,000.

According to the National Health Surveillance Agency (ANVISA, 2001), although Sickle Cell Disease is more common in individuals of African descent in Brazil, population miscegenation has made this pathology frequent also in people with white or brown skin in the national territory, with a higher incidence in the Northeast region.

In terms of the number of cases of Sickle Cell Disease, the highest incidence is in the Northeast, with a predominance in Bahia (BRASIL, 2025). In the state of Maranhão, after diagnostic confirmation, patients with this condition receive treatment at the Hematology and Hemotherapy Center of Maranhão, with about 1700 patients regularly seen (ARANHA, 2023).

The term sickle cell anemia encompasses a group of inherited hemolytic anemias that share the presence of hemoglobin S (HbS) in red blood cells. HbS represents an alteration of normal hemoglobin, hemoglobin A (HbA), and is caused by a genetic mutation that affects one of the nitrogenous bases of deoxyribonucleic acid (DNA) responsible for encoding beta globin (CASTILHOS, 2015).

Sickle cell anemia is transmitted when an individual inherits a mutated gene for hemoglobin S production from each parent. If the inheritance is from only one mutated gene (maternal or paternal), the individual will have only the sickle cell trait. Carriers of sickle cell trait do not develop the disease, being asymptomatic and, therefore, not requiring specialized treatment. It is essential that these individuals are aware of their diagnosis to avoid the risk of having children with another carrier of the trait, which could result in offspring with sickle cell anemia, trait or without the genetic alteration (ZAGO; PINTO, 2007).

The prevention of the disease is carried out through neonatal screening, ideally between the second and seventh day of life. The heel prick test, the sickling test, and the spot test are screening tests, while hemoglobin electrophoresis by isoelectric focusing electrophoresis (IEF) or high-performance liquid chromatography (HPLC) is the confirmatory test of the diagnosis for Sickle Cell Disease (Brasil, 2024).

Individuals with Sickle Cell Disease may manifest a variety of symptoms, with varying intensity, such as **fever, prostration, toxemia, hypoxia, vomiting, dehydration, respiratory symptoms, and pain.** Some patients, even following the appropriate treatment, may have recurrent acute crises, such as intense pain in the bones, abdomen and other parts of the body, as well as greater susceptibility to pneumonia and other infections. These crises can progress to more severe conditions such as septic shock and death, due to immunological impairment with risk of death if there is no appropriate medical intervention (BRASIL, 2024).

The sickle cell anemia treatment protocol recommends the prophylactic use of penicillin up to five years of age, aiming to prevent infections such as pneumonia, meningitis, and osteomyelitis. For this reason, strict surveillance by the clinical team is essential, given the possibility of septicemia occurring within a 24-hour period (KIKUCHI, 2007).

The treatment of sickle cell anemia involves the use of medications such as folic acid, analgesics and antibiotics, as well as blood transfusions in cases of very low hemoglobin levels. Patients with this condition undergo medical follow-up with various specialties, including hematologists, pediatricians, general practitioners and other health professionals (ARAÚJO, 2004).

Early diagnosis of Sickle Cell Disease is essential, as it allows the immediate institution of treatment, which contributes to the attenuation of crises, improves life expectancy and quality of life, and prevents infant mortality in children under five years of age with the disease. **The present investigation seeks to analyze the diagnostic methods and therapeutic approaches employed in patients with Sickle Cell Anemia at the Hematology and Hemotherapy Center of São Luís - MA, in order to identify strategies that can increase the well-being and quality of life of these individuals.**

METHODOLOGY

The present investigation employed a mixed methodology, comprising both the review of pertinent literature, through the analysis of scientific articles, monographs, dissertations and theses, and the realization of a field study. Primary data collection

occurred through the application of questionnaires to patients diagnosed with Sickle Cell Anemia and undergoing treatment at the Hematology and Hemotherapy Center, located in São Luís, Maranhão.

In conducting this research, the ethical precepts outlined in Resolution 196/96 were strictly observed, ensuring the anonymity and confidentiality of the participants' identity. Prior to their inclusion in the study, the individuals were duly informed about the research objectives and invited to participate by signing the Informed Consent Form (ICF), and were subsequently submitted to an interview and completion of the questionnaire.

Data were obtained through questionnaires answered by patients with Sickle Cell Disease and/or their legal guardians. These instruments contained multiple-choice questions, formulated in accessible and direct language, covering aspects such as the patient's profile, the diagnostic process, the treatment modalities, and the level of knowledge about sickle cell disease.

After the collection phase, the data were submitted to a process of analysis and interpretation. The results obtained were then organized and presented by means of graphs, in order to demonstrate the relevant information in statistical format.

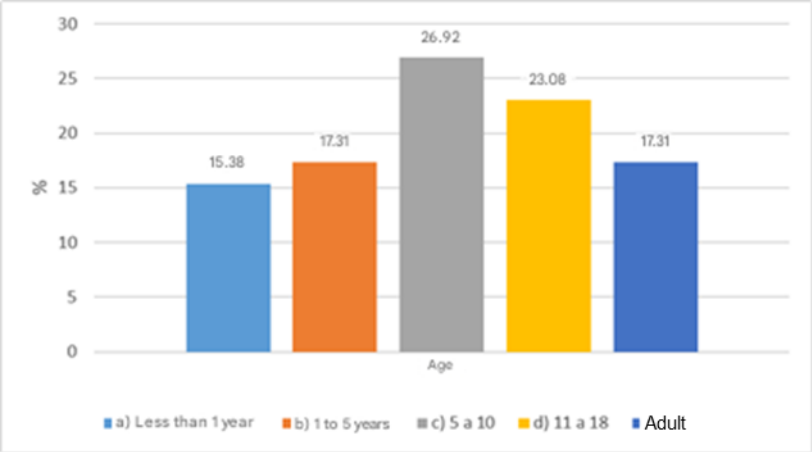
RESULTS AND DISCUSSION

The incidence of sickle cell disease is significantly higher in the Northeast region compared to the South region of Brazil, due to the higher proportion of the population of African descendants in the Northeast, since the disease has a genetic origin linked to African ancestry. According to the Ministry of Health (2023), the Northeast region has 8 to 12 times more cases of Sickle Cell Disease than the South region, which places it as a priority area for public policies for screening, monitoring, and treatment of the disease (BRASIL, 2023)

Among the 52 patients being followed up at the Hematology and Hemotherapy Center, located in São Luís, MA, there was a higher concentration of cases of Sickle Cell Anemia in the age group of 5 to 10 years, representing 26.92% of the total number of interviewees. On the other hand, the lowest frequency was observed in children under one year of age (15.38%), as illustrated in Figure 1.

In addition, it was found that more than half of these patients (greater than 55%) are female.

Figure 1: Age of patients with SCA



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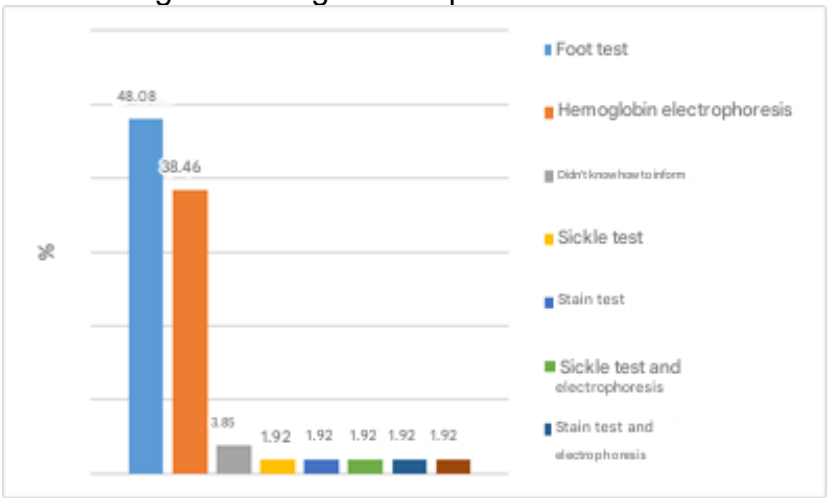
The findings of this research echo the results of Silva (2017) regarding the predominance of female individuals among those with the disease. However, a distinction is observed in relation to the most prevalent age group, since the present study delimited the ages, identifying a higher frequency in children aged 5 to 10 years, while the aforementioned author found a lower occurrence of the disease in children under 12 years of age.

Regarding the diagnostic method, it was found that 48.08% of the patients had the disease identified through the heel prick test. Another 38.46% confirmed the diagnosis through hemoglobin electrophoresis, and a small portion of 3.85% could not inform how the condition was diagnosed. A minority of patients, representing 1.92%, had the diagnosis established by other tests, such as the foil test and the spot test, in addition to the combined use of multiple tests, as detailed in Figure 2.

Figure 2: Diagnosis of patients with SCA



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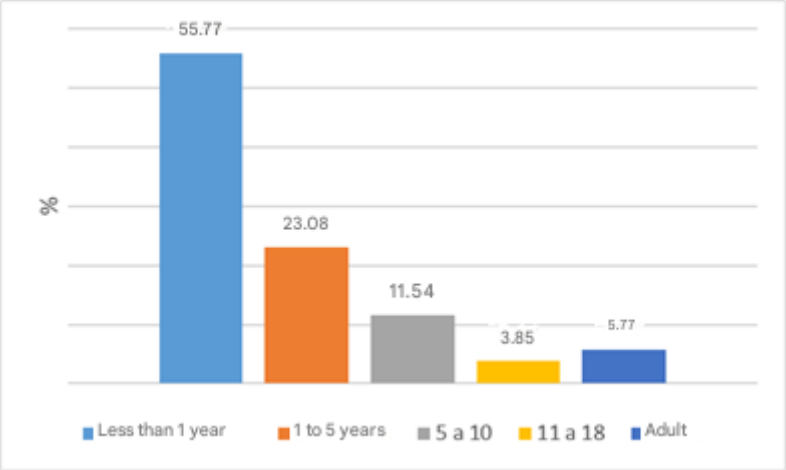
The results obtained in the present investigation are in line with the observations of Watanabe *et al*. (2008), which highlight the prevalence of the heel prick test as the main method for detecting Sickle Cell Anemia. Hemoglobin electrophoresis, a specific laboratory test for the diagnosis of the disease, is recommended for patients who did not undergo the heel prick test in the ideal age group. Other forms of diagnosis identified involved tests such as the stain test and the fuzzing test, often used in conjunction with hemoglobin electrophoresis to confirm the diagnosis.

Regarding age at diagnosis of Sickle Cell Anemia, it was revealed that most patients (55.77%) were diagnosed before completing one year of age. A portion of 23.08% received the diagnosis between 1 and 5 years, while 11.54% were diagnosed between 5 and 10 years. The lowest rates were observed in the age groups of 11 to 18 years (3.85%) and in adults (5.77%), indicating, in these cases, a late diagnosis of the condition, as illustrated in Figure 3.

Figure 3: Age at which SCA disease was diagnosed in patients



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The data collected reveal that more than half of the patients being treated at the Hematology and Hemotherapy Center received the diagnosis of the disease before the first year of life, a period considered ideal for the investigation. This early detection represents an advantageous condition for the immediate implementation of treatment, which can minimize the occurrence of severe complications and result in less intense crises. The findings of this research are similar to the results reported by Costa (2021), who found in his research that 71% of newborns between the 3rd and 7th day of life undergo a heel prick test.

The results showed that 75% of the patients participating in the research have already undergone blood transfusion. This information is supported by the data reported by

Félix (2010), who highlights the growing use of blood transfusion in the management of sickle cell anemia. However, the author warns of the significant heterogeneity resulting from iron overload, a complication associated with multiple transfusions. Félix (2010) also emphasizes the importance of simple blood transfusion in cases of hypoxemia, a procedure that can reduce the length of hospitalization and promote a more effective recovery, resulting in improved oxygenation.

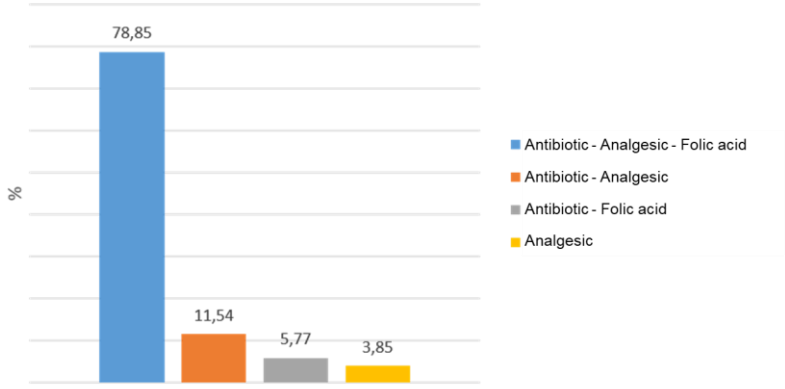
On the other hand, Ferraz (2012) argues that blood transfusion should not be considered a priority in the regular treatment of patients with Sickle Cell Anemia, and is even contraindicated in cases of asymptomatic anemia.

Regarding the most common clinical manifestations reported by patients with Sickle Cell Anemia being followed up at the Hematology and Hemotherapy Center in São Luís, the results point to the following distribution: 26.92% reported intense joint pain associated with edema in the hands and feet, skin pallor and jaundice, as well as fever and fatigue. Another 21.15% reported episodes of ischemic sequestration, in addition to the symptoms previously mentioned. A smaller portion of patients (9.62%) indicated pallor, yellowish color of the eyes, fever and tiredness as the most frequent complaints. The other participants in the research showed a variation between the symptoms already mentioned, with a lower frequency of reports of damage to organs such as the liver, lungs, heart and kidneys, corresponding to 1.92% of the sample.

Patients usually manifest multiple symptoms simultaneously, and the occurrence of only one symptom is less common, which does not imply less intensity or aggressiveness of these clinical manifestations, so the findings of the present investigation are in line with the guidelines of the Clinical Protocol and Therapeutic Guidelines for Sickle Cell Disease regarding pain crises, emphasizing the importance of observing behavioral changes and pain intensity in patients, an assessment that can be facilitated by the use of pain scales, in addition to stimulating hydration, as fluid intake aims to reduce blood viscosity and prevent dehydration (BRASIL, 2024).

Fernandes (2010) reinforces the need for agility in the recognition of crises and in the search for medical care, considering the potential severity of the symptoms of the disease. Among the patients interviewed, the majority (78.85%) follow a therapeutic regimen that includes antibiotics, analgesics, and folic acid. A portion of 11.54% use antibiotics and analgesics, while 5.77% use antibiotics and folic acid. The lowest proportion (3.85%) corresponds to patients who use only analgesics in their treatment, as detailed in Figure 4.

Figure 4 - Treatment used by SCA patients



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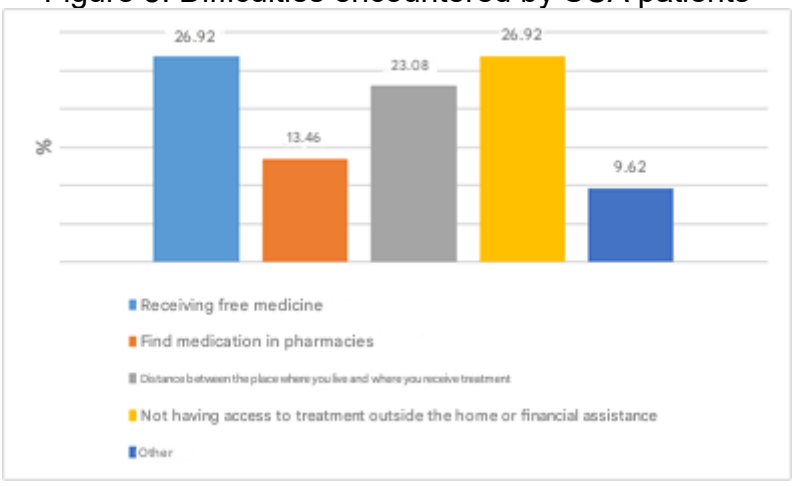
In line with Braga (2007), it is imperative that both patients and their families understand the urgency of early intervention in pain management, through the administration of medications such as antibiotics and analgesics, combined with appropriate nutrition, folic acid supplementation and abundant fluid intake. The author also points to the need, in certain cases, for a combination of different drugs, each with its own specific function: antibiotics for the prevention of infections such as pneumonia, analgesics for pain relief, and folic acid for the formation of blood cells.

Figure 5 illustrates the challenges faced in the treatment of Sickle Cell Anemia. Among the patients interviewed, 26.92% report difficulties in obtaining free medicines, while 13.46% mention the difficulty in finding medication in pharmacies. Another significant obstacle for patients is the displacement from their municipalities of residence to the place of treatment, motivated by the distance and the absence of family members in São Luís (23.08%). An even larger portion, representing 26.92%, emphasizes the financial difficulties resulting from traveling for treatment, since they have to bear the costs of travel, not having access to Out-of-Home Treatment (TFD) and the corresponding financial aid.

Figure 5: Difficulties encountered by SCA patients



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The bibliographic survey indicates that all federative units of Brazil have at least one reference center, usually located in their capitals, intended for the care of individuals with Sickle Cell Anemia. These patients should be included in a comprehensive care program, which includes treatment and the supply of drugs through the Unified Health System (SUS), although the availability of these drugs may be intermittent, representing an obstacle to treatment.

When asked about what Sickle Cell Anemia is, it was found that 67.31% of the patients interviewed stated that they were aware of the disease and its symptoms. On the other hand, 21.15% reported being unaware of the condition, despite having a carrier, and 11.54% reported having limited knowledge about the disease.

Reports presented by Maia *et al*. (2013) showed that patients with knowledge about Sickle Cell Anemia are also able to identify its symptoms. This understanding is not restricted to the pathology itself, but extends to the search for information from family members or close people. However, this knowledge generally does not cover in depth the causes, consequences and characteristics of the disease, but it enables individuals to act in the face of crisis episodes.

In addition, according to the same author, patients who are unaware of Sickle Cell Anemia are more vulnerable to not recognizing its symptoms, consequently not knowing how to inform their family members in times of crisis. Although it represents a non-expressive portion of the knowledge of the disease, the lack of knowledge prevents the identification of its symptoms and crises.

CONCLUSION

Sickle Cell Anemia is a genetic hemoglobinopathy of significant prevalence, being recognized as a public health problem at a global level, so the absence of specialized treatment for Sickle Cell Anemia in some municipalities of the state drives patients to seek the blood center in the capital, evidencing a centralization of care.

The analysis of patient data showed a higher incidence of the disease in the age group of 5 to 10 years, with a predominance of females. A relevant fact is the high proportion of early diagnoses made through the heel prick test, especially in children under one year of age, representing a significant portion of the cases identified in Maranhão.

The difficulties faced by patients in accessing treatment are multifaceted, including the geographical distance between their homes and the referral center, the difficulty in

accessing certain medications, the absence of support for Out-of-Home Treatment and the lack of financial aid to pay for travel.

One positive aspect identified in the survey is the level of knowledge about sickle cell disease among patients, with more than 60% reporting having some understanding of the condition. Given the chronic and severe nature of Sickle Cell Anemia, society's awareness of this pathology is critical. In this sense, genetic counseling for couples with plans to have children, psychological preparation to deal with the disease, and the dissemination of preventive information for sickle cell patients are measures of great relevance for improving quality of life and reducing morbidity and mortality associated with this condition.

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