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DIAGNOSIS AND TREATMENT OF PATIENTS WITH SICKLE CELL ANEMIA: AN INTEGRATIVE REVIEW OF THE LITERATURE

Marielle Flávia do Nascimento Araújo*[1], Tayane Moura Martins[2], Aline Girliane de Lima Santana[3], Williani Maria da Silva Morais[1], Rosângela Silva de Medeiros[4], Maria Mylenna Costa Ferreira[1], Bruna Batista da Silva Santos[1], Jessyca Juliana da Silva Barros Burgos[1], Ariany Thauan Pereira de Holanda[1], Isabella Korina dos Santos Barbosa[1], Rebecca Ferreira da Paixão[1] and Aridan Maria Pereira de Holanda[5]

[1]Centro Universitário Mauricio de Nassau; [2]Universidade Universidade Federal do Pará; [3]Faculdade de Ciências Humanas e Exatas do Sertão do São Francisco, FACESF; [4]Faculdade de Ciências Humanas, ESUDA; [5]Facultad Interamericana de Ciências Sociais Paraguay

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Corresponding author:
Marielle Flávia do Nascimento Araújo

ABSTRACT

Objective: The study presented here aims to identify, through scientific research, the ways of diagnosis and treatment of sickle cell anemia. Methodology: Integrative literature review of evidence present through research, method in critical evaluation and synthesis, helping to firm the existing health practices. The search was made in LILACS, IBECS, MEDLINE and PUBMED databases. The studies and search were performed in articles from the last 10 years (2012-2022) in order to obtain updates and more recent articles in the literature**. Results and Discussion:** The first scientific evidence of the disease in Brazil appeared in the mid-nineteenth century precisely in 1835, through the doctor named José Martins da Cruz Jobim, in the city of Rio de Janeiro, when discussing and exposing the diseases that most affected the low-income population in Brazil. In its physiopathological process, sickle-cell anemia is a hereditary genetic disease that has characteristics arising from a mutation in position 6 of the beta globin chain on chromosome 11, where the bases (GAG) are replaced by the bases (GTG), thus resulting in the replacement of glutamine acid by valine. **Conclusion:** Because it is a hereditary disease and originated from a mutation in the beta chain of hemoglobin, this disease has high morbidity, especially in the north and northeast, with emphasis on people of African descent. In order to reduce mortality, it is extremely important that sickle cell anemia be diagnosed and treated early to avoid further complications and disease progression. Among these methods, there are tests that help diagnose the patient.

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INTRODUCTION

Characterized as an autosomal hematological disease, sickle cell anemia exists from a mutation in chromosome 11, generating modifications in the hemoglobin structure, resulting in the substitution of a glutamic acid for valine, with the resolution of hemoglobin S (Rocha et al., 2022). This protein is responsible for modifying the erythrocytes, transforming the red blood cells into sickle-shaped red blood cells. These red blood cells become increasingly rigid, generating complications such as vaso-occlusion in small vessels, which consequently causes tissue inflammation and ischemia, causing the carrier to experience intense pain, increased likelihood of acquiring infections, and even damage to human body organs. Among the complications that may occur in patients with sickle cell anemia, the renal diseases called sickle cell nephropathy are the most common, since they include glomerular and tubular changes (Rocha et al., 2022 and Campos, 2022). Sickle cell anemia is the most prevalent hereditary disease in Brazil, with great psychological and social damage, and it affects about 0.1% to 0.3% of the black population, due to the high level of miscegenation. It is estimated that 5-6% of the population has a hemoglobin S (HbS) gene, reaching a number of 700 to 1,000 new cases per year (Campos, 2022 and FIGUEIREDO, 2014). With an estimated 4% of the population having some sickle cell trait and 25,000 to 50,000 having homozygous or compound or double heterozygous status, sickle cell anemia is more prevalent in the northern and northeastern regions, between 6% and 10%. While in the South and Southeast, only two to three percent of the population may be affected, the justification for this is due to the aplotypes with respect to transmission by the black population coming from Africa. The clinical protocol and therapeutic

guidelines for sickle cell disease according to the ordinance number 298 of February 9, 2018 according to the Ministry of Health, sickle cell disease is curable through hematopoietic stem cell transplants. However, it is worth emphasizing that these are possibilities (Garioli, 2019 and Lima, 2019). Because of the shortened life span of the sickle cell red blood cells, the patient may manifest chronic hemolysis exposed with pallor, increased level of indirect bilirubin that may consequently result in the formation of cholelithiasis and several other common complications such as spleen destruction and continuous infections. For the adult population that coexists with the genetic trait and the disease, pre-conception genetic counseling is important because there is a high probability of their children having anemia due to genetic issues. This counseling is a necessary tool in the field of this hereditary disease, helping in the management of preventive practices and guidance to parents and newborns (Isaza-López, 2020; Campos, 2022 and Ministério as Saúde, 2018). The disease carries with it several symptoms when it comes to hospital admissions and care, and it is important that the health team, with emphasis on nursing, provides care to ease the pain, infection, complications and aggravations of patients with sickle cell anemia. It is important that this care be continuous throughout the patient's life until finitude (Ministério as Saúde, 2018). In view of the complications related to sickle-cell disease and its heredity, it is necessary to search for complications, diagnosis and treatment of sickle-cell anemia, in order to promote a better quality of life in an effective treatment. The study presented here aims to identify, through scientific research, the ways of diagnosis and treatment of sickle cell anemia.

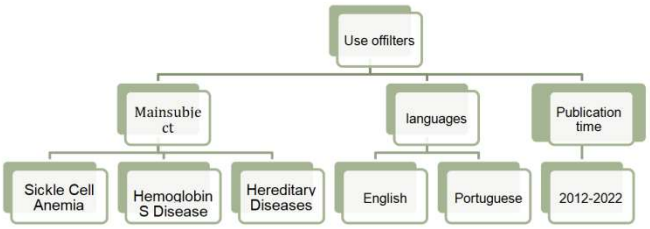
METHODS

This study is an integrative review of the existing literature, which was guided by a guiding question, which compiled the research follow-ups already generated regarding the theme in question. The integrative review has the purpose of gathering and synthesizing research results on a delimited theme or issue, in a systematic and ordered manner, contributing to the deepening of knowledge on the investigated theme. The following steps were followed for this research:

- **Step 1:** Establishment of hypotheses and the research question with the choice and definition of the theme, objectives, identifying the key words and the selected theme with the clinical practice.
- **Step 2:** Sampling with the literature search with establishment of the inclusion and exclusion criteria, use of database and selection of the studies.
- **Step 3:** Categorization of studies with Extraction of information, organization and systematization of information and formation of the database.
- **Step 4:** Evaluation of the studies included in the review with application of analysis, inclusion and exclusion and critical analysis of the selected studies.
- **Step 5:** Interpretation of the results with discussion of the results, proposal of recommendations and suggestions for future research.
- **Step 6:** Presentation of the review with summary of the available evidence, Creation of the detailed document with the description of the review.

The Health Science Descriptors (DeCS) used in the preparation of this article were: Sickle Cell Anemia, Hemoglobin S Disease, Hereditary Diseases. The aim was to answer the following guiding question: What are the ways of diagnosis and treatment for patients with sickle cell anemia?

The inclusion criteria were Articles in available in its integrity, free, available between the years 2016 and 2022, in Portuguese, Spanish and English, restricted to human beings, female and male, adults and elderly, and that answer the question that directs the study and meet the purpose of the same. The articles were searched through electronic libraries: Scientific Electronic Library Online (SciELO) and Virtual Health Library (VHL) with the following databases: Latin American and Caribbean Literature on Health Sciences (LILACS), Medical Literature Analysis and Retrieval System Online (MEDLINE), and PUBMED.

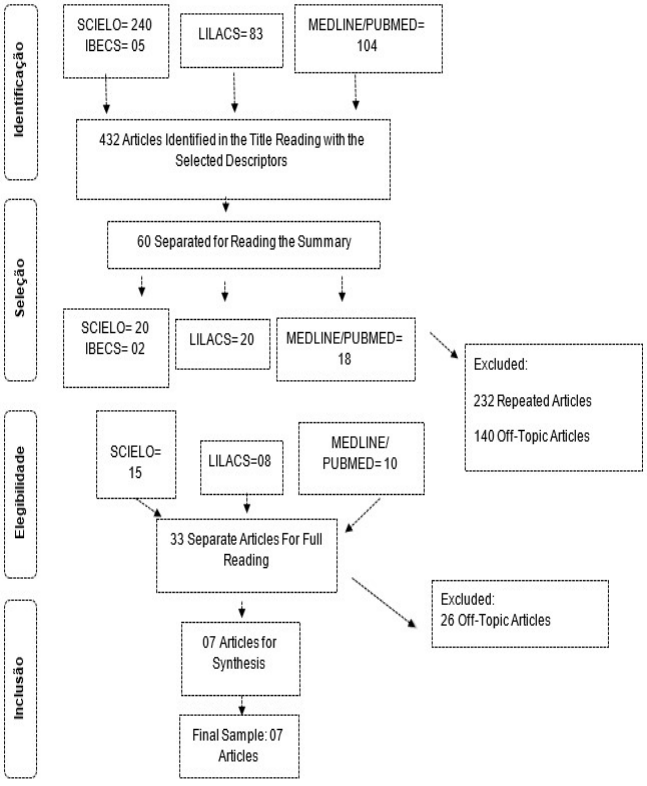


Source: Araújo MFN, et al., 2022

Figure 1. Filters used for research contribution

RESULTS

A total of 432 articles were found in the searched databases. At first the title was read according to the descriptors in each virtual library, and after reading the themes, 60 articles were selected for abstract reading and separated according to the inclusion and exclusion criteria. Next, the dissipation and triage of the full reading of 33 articles was done and those with relevance were admitted, which contained in their total the types of nursing care team, hemodialysis, kidney disease and meeting the study objective, finalizing with the final sample of 10 articles, as shown in Figure 2.



Source: Araújo MFN, et al., 2022.

Figure 2. Schematic representation of the synthesis and analysis of results

The articles were read and analyzed in their entirety. To determine the data, an instrument was prepared with the following variables: article number, title, authors, year of publication, study characteristics, and main results, as shown in Chart 1. According to the literature search, type of study, the most prevalent was integrative review and quantitative, followed by narrative review.

Chart 1. Characterization of the articles in the sample

Article number	Title of the Article	Autor(es) Author(s) and Year	Study characteristics	Main results
1	Anemia falciforme: una revisión sobre elgenotipo de laenfermedad, haplotipos, diagnóstico y estudiosasociados	Díaz-Matallana M, Márquez-Benítez Y, Martínez-Lozano JC, Briceño-Balcázar I, Benavides-Benítez E, Bernal JE, et al.	experimental trial	These include techniques for the determination of hemoglobin and the use of molecular markers, among others.
2	The interaction between Hb B2, Hb S and beta thalassemia: a family case report	Santos R de S, Costa JVM, Leite LAC, Bonini-Domingos CR	case report	They identified that the identification of less common hemoglobin variants requires a thorough analysis with the association of clinical data and different laboratory methodologies.
3	Insight into the complex pathophysiology of sickle cell anaemia and possible treatment	Piccin A, Murphy C, Eakins E, Rondinelli MB, Daves M, Vecchiato C, et al	original study	Address the complex pathophysiology of sickle cell anemia and possible treatment
4	Emerging disease-modifying therapies for sickle cell disease.	Carden MA, Little J.	original study	Address new disease-modifying therapies for sickle cell disease
5	Sickle cell disease: a review.	Kavanagh PL, Fasipe TA, Wun T.	a review.	Address sickle cell anemia as a whole
6	Sickle Cell Disease and laboratory approach: a brief literature review	ALMEIDA R.A; BERETTA A.L	A brief literature review	Address sickle cell anemia as a whole
7	Advances in the treatment of sickle cell disease. Mayo Clinic Proceedings	Kapoor S, Little JA, Pecker LH	Mayo Clinic Proceedings	Address advances in the treatment of sickle cell anemia

DISCUSSION

The first scientific evidence of the disease in Brazil emerged in the mid-nineteenth century precisely in 1835, through a physician named José Martins da Cruz Jobim, in the city of Rio de Janeiro, when discussing and exposing the diseases that most affected the low-income population in Brazil. In the following century, in 1910, the physician James Bryan Herrick presented the first scientific study on sickle cell anemia in an American magazine. He showed the presence of sickle-shaped red blood cells in a young black man who had anemia in its most advanced stage; in addition, this young man had other symptoms such as strong joint pain and jaundice (Ministério da Saúde, 2016 and Díaz-Matallana, 2022). Only after this study did the disease begin to be called sickle cell disease. Its discovery was made through genetic studies using the first molecular pathology in humans, in 1947 the researcher JesséAccioly through research suggested that the disease was hereditary, and only in 1949 the physician James van Gundia Neel proved this theory (Díaz-Matallana, 2021 and Kapoor, 2022). In its pathophysiological process, sickle cell anemia is an inherited genetic disease that has characteristics arising from a mutation in position 6 of the beta globin chain on chromosome 11, where the bases (GAG) are replaced by the bases (GTG), resulting in the replacement of glutamine acid by valine 10.

Normal hemoglobin a is replaced by hemoglobulin S which are from sickle cells thus causing hydrophobic interactions between the hemoglobin molecules. Such polymers thus form crystals called tachyzoites that change the structure of the RBC by modifying its shape commonly called drepanocytes. The intensification of the anemic situation depends exclusively on the amount of hemoglobin GS present and acting in the erythrocytes although people with sickle cell anemia possess hemoglobin is not yes in their cells may also possess hemoglobin a or fetal hemoglobins (Brasil, 2014; Santos, 2021 and Kapoor, 2022). One of the most effective ways to discover sickle cell anemia is genetic counseling performed by a qualified professional with the main objective of informing the family about the disease, risks, complications, diagnosis, and treatment (Santos, 2021 and Piccin, 2022). When the diagnosis is made early in the disease, it can be greatly reduced. The diagnosis can be made through some specific tests, such as a complete blood count (CBC) with red blood cell, white blood cell, and platelet analysis, as well as hemoglobin electrophoresis in alkaline and acidic buffers, which is crucial in detecting both normal and abnormal hemoglobins, Sickle cell test aiming to analyze the morphology of red blood cells with low oxygenation, a technique that allows RBCs to be analyzed under low oxygen concentration by means of a specific solution, microscopic analysis of reticulocytes, and solubility test (Carden, 2022 and Kavanagh, 2022).

The treatment with hydroxyurea was first manufactured in the XIX century in Germany and after almost a century it was approved by the North American FDA for the treatment of neoplastic diseases, and consequently for leukemias and parallel to this, clinical complications of sickle cell anemia. After that, there was an increasing search for drugs that could stimulate the synthesis of gamma globin chains and increase intraerythrocyte synthesis; but only since 1998 has such treatment become part of the therapeutic armamentarium for patients with sickle cell anemia, capable of reducing clinical complications and improving the quality of life of the patient. Despite being an ally, hydroxyurea also has adverse reactions such as low neutrophil and platelet counts, headache, nausea, and even anemia (Kavanagh, 2022; ALMEIDA, 2017 and Kapoor, 2022).

CONCLUSION

Since it is a hereditary disease and originates from a mutation in the Beta chain of hemoglobin, this disease has a high morbidity rate, especially in the North and Northeast regions, with emphasis on people of African descent. In order to reduce mortality, it is extremely important that sickle cell anemia be diagnosed and treated early to avoid further complications and disease progression. Among these methods there are tests that help diagnose the patient. As for the treatment of sickle cell anemia, the transfusion of red blood cells is a great ally to reduce the amount of drepanocytes. In addition, there is another form of treatment such as hydroxyurea, which helps in the production of fetal hemoglobins. It is important that the patient finds out as soon as possible about the disease so that the treatment can be validated and a better quality of life can be achieved.

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