Importance of early diagnosis of thalassemia and implications in its treatment: literature review

Importância do diagnóstico precoce da talassemia e implicações no seu tratamento: revisão da literatura

Importancia del diagnóstico precoz de la talasemia e implicaciones en su tratamiento: revisión de la literatura

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ABSTRACT
Thalassemia is a hereditary genetic disease that is characterized by changes in the conformation of hemoglobin. This pathology includes different types of blood changes that are characterized by lower levels or the absence of normal globin chains in hemoglobin. With the aim of contributing to the reduction of damage caused by thalassemia, the present study aims to promote awareness about the importance of early diagnosis of thalassemia, aiming to reduce health complications and improve patients' quality of life through medical interventions and support. To carry out this literature review on thalassemia, we will adopt an integrative analytical approach of searching and analyzing relevant studies. Initially, databases relevant to the area were selected, such as PubMed, Scopus, Web of Science. This study highlights the significance of early diagnosis of thalassemia as a crucial measure in the effective management of this genetic condition. Early identification not only allows for the implementation of appropriate medical interventions and treatments, but also helps reduce health complications and improve patients' quality of life.

Keywords: Hemoglobin. Diagnosis. Quality of Life. Public Health.

RESUMO
A talassemia é uma doença genética hereditária que se caracteriza por alterações na conformação da hemoglobina. Esta patologia inclui diferentes tipos de alterações sanguíneas que se caracterizam por níveis mais baixos ou pela ausência de cadeias normais de globina na hemoglobina. Com o objetivo de contribuir para a redução dos danos causados pela talassemia, o presente estudo visa promover a conscientização sobre a importância do diagnóstico precoce da talassemia, visando reduzir complicações de saúde e melhorar a qualidade de vida dos pacientes por meio de intervenções e apoio médico. adequado. Para realizar esta revisão de literatura sobre talassemia, adotaremos uma abordagem analítica integrativa de busca e análise de estudos relevantes. Inicialmente foram selecionadas bases de dados relevantes para a área, como PubMed, Scopus, Web of Science. Este estudo destaca a importância do diagnóstico precoce da talassemia como uma medida crucial no manejo eficaz desta condição genética. A identificação precoce não só permite a implementação de intervenções e tratamentos médicos adequados, mas também ajuda a reduzir complicações de saúde e a melhorar a qualidade de vida dos pacientes.


RESUMEN
La talasemia es una enfermedad genética hereditaria que se caracteriza por cambios en la conformación de la hemoglobina. Esta patología incluye diferentes tipos de alteraciones sanguíneas que se caracterizan por niveles más bajos o ausencia de cadenas de globina normales en la hemoglobina. Con el objetivo de contribuir a la reducción del daño causado por la talasemia, el presente estudio pretende promover la conciencia sobre la importancia del diagnóstico precoz de la talassemia, con el objetivo de reducir las complicaciones de salud y mejorar la calidad de vida de los pacientes a través de intervenciones y apoyo médicos. adecuado. Para llevar a cabo esta revisión de la literatura sobre la talassemia, adoptaremos un enfoque analítico integrador de búsqueda y análisis de estudios.
relevantes. Inicialmente se seleccionaron bases de datos relevantes para el área, como PubMed, Scopus, Web of Science. Este estudio destaca la importancia del diagnóstico temprano de la talasemia como medida crucial en el manejo eficaz de esta condición genética. La identificación temprana no sólo permite la implementación de intervenciones y tratamientos médicos adecuados, sino que también ayuda a reducir las complicaciones de salud y mejorar la calidad de vida de los pacientes.

**Palabras clave:** Hemoglobina. Diagnóstico. Calidad de Vida. Salud Pública.

1 INTRODUCTION

Thalassemia is a hereditary genetic disease that is characterized by changes in the conformation of hemoglobin. This pathology alters the formation of this blood component and is also called Mediterranean disease, since the first recorded cases predominated in natives who were close to this region. This led to the first descriptions and explanations of thalassemia around 1932 Shafique et al., 2023).

This pathology includes different types of blood changes that are characterized by lower levels or the absence of normal globin chains in hemoglobin. In this sense, normal hemoglobins bind to oxygen collected in the lungs and distribute it to the tissues. However, when faced with a change in these transport proteins, gas distribution is reduced. In addition to this, this blood element is not only involved in the transport of oxygen, but also has a role in transporting other gases, such as carbon dioxide, which is collected from tissues, after cell metabolism, and subsequently taken to the lungs to be deleted. Therefore, the change in hemoglobin reduces the concentration of gases in the body (Safiri et al. 2021).

This disease is part of a set of changes that are identified as hemoglobinopathies, which are a set of genetic pathologies that affect hemoglobin. They are divided into two large groups: thalassemia syndromes, which result in severe anemia and other serious complications, and structural hemoglobin anomalies, which result in hemoglobin malfunction. Both are caused by defects in the alpha or beta genes (Shafique et al., 2023).

Faced with these issues, the present study aims to address the importance of early diagnosis of thalassemi, aiming to reduce health complications and improve patients' quality of life through medical interventions and adequate support.
2 METHODOLOGY

This work follows the study methodology proposed by Gil (2008). This is a bibliographical review defined by the author as “developed from already prepared material, consisting of books and scientific articles”. In this work, an integrative literature review on the early diagnosis of thalassemia is proposed. To carry out the integrative review, the following steps will be followed: 1) definition of the research question; 2) search in scientific databases; 3) selection of relevant studies; 4) data extraction and analysis; 5) presentation and discussion of results. To carry out this literature review on thalassemia, we will adopt an integrative analytical approach of searching and analyzing relevant studies. Initially, databases relevant to the area were selected, such as PubMed, Scopus, Web of Science. The search terms "thalassemia", "diagnosis", "early" and their variations were used, combined with Boolean operators. After screening the titles and abstracts, relevant studies were selected for detailed analysis. The synthesis of results will be carried out through a qualitative analysis, identifying trends and discrepancies between studies.

3 RESULTS AND DISCUSSIONS

Thalassemia is a genetic hemoglobinopathy generated by genetic alterations in globin genes (alpha or beta), which generate a reduction or absence of synthesis of one or more globin chains that form hemoglobin. These changes can generate several different clinical effects (Ministry of Health, 2016).

Thalassemia can be divided into two major types, alpha and beta. Each type can manifest itself at different levels, namely minor, intermediate and major. Considering this, thalassemia minor can generate mild manifestations that may not be noticeable, characterizing a more asymptomatic condition. In the intermediate degree, there is a moderate deficiency in the production of hemoglobin, therefore, the clinical manifestations are more pronounced, however, they are not yet serious. Thalassemia major is the most serious form of the pathology, therefore, if the patient does not receive appropriate treatment, clinical manifestations may involve severe anemia, hepatomegaly, splenomegaly and bone changes (Ministry of Health, 2016).
One of the main clinical changes that arise due to thalassemia is heart disease. Cardiovascular diseases lead to a high frequency of morbidity and mortality, reaching around 75% of deaths. Some of the common heart diseases are ventricular hypertrophy, dilation of the heart chambers, diastolic dysfunction, mitral and tricuspid insufficiency and pulmonary hypertension. This ends up developing more severe conditions that can lead to death when not treated correctly (Lopes et al., 2022).

Early diagnosis of thalassemias is crucial to improving the prognosis and quality of life of patients affected by this genetic condition. These blood disorders, characterized by abnormal production of hemoglobin, can result in serious anemia if not identified and treated early. Early detection allows doctors to implement appropriate interventions, such as transfusion and iron chelation therapies, which help prevent serious complications such as heart failure and damage to vital organs (Ali et al., 2021).

Furthermore, early diagnosis of thalassemias enables genetic counseling for families at risk, allowing them to make decisions about future pregnancies and family building, becoming important in areas where the disease is more prevalent, contributing to reducing the global burden of the disease and to prevent its transmission to future generations. Genetic counseling can also help identify asymptomatic carriers, who can be informed about the risk of transmitting the disease to their offspring (Braid; Batten; Sparks, 2022).

Diagnosing thalassemias early can positively influence the development of new therapies and treatment approaches. By identifying patients at early stages of the disease, researchers can better study their progression and response to treatment, which could lead to significant advances in personalized medicine and the search for more effective cures. Early identification also allows patients and their families to receive adequate psychological and social support, helping them to better cope with the physical and emotional challenges associated with the disease (Polainas, 2017).

Diagnosis of thalassemias generally involves a combination of clinical examinations, laboratory analyzes and genetic tests. Initially, the presence of the disease may be suspected based on the patient's symptoms, such as fatigue, paleness and jaundice (Oliveira et al., 2022). Routine laboratory tests, such as a
complete blood count, can reveal anemia and specific blood cell characteristics that suggest thalassemia. Additionally, liver function tests and blood iron levels may be performed to evaluate possible complications. Confirmation of the diagnosis is usually done through genetic tests, such as DNA analysis, to identify mutations in the genes responsible for the production of hemoglobin (Frasão et al., 2022).

In the thalassemia diagnosis process, it is essential to increase access to specialized genetic tests and the training of health professionals in interpreting the results. Neonatal screening programs in areas with a high prevalence of the disease can identify asymptomatic cases early, enabling therapeutic interventions even before the emergence of serious symptoms. These combined approaches have the potential to significantly reduce the time required to diagnose thalassemias, thereby improving clinical outcomes and quality of life for patients (Manganas et al., 2024; Charoenkwan et al., 2024).

In underdeveloped countries, the diagnosis of thalassemias often faces significant challenges due to a lack of medical and financial resources. The specialized genetic and laboratory tests needed to confirm the presence of the disease can be expensive and inaccessible to many underserved communities. Furthermore, the lack of healthcare professionals trained in the identification and management of thalassemias can lead to underdiagnosis and delays in appropriate treatment (Silao et al., 2024).

One promising approach is to invest in simple, low-cost screening methods that can be easily implemented in resource-limited settings. For example, careful clinical assessment of common symptoms and laboratory features of thalassemias can help identify suspected cases, allowing referral for more specific diagnostic testing only when necessary. Furthermore, the development of rapid and portable diagnostic kits based on techniques such as PCR (Polymerase Chain Reaction) or capillary electrophoresis tests can facilitate rapid and accurate detection of the disease in remote and resource-limited areas (Horvei et al., 2021).

Another important strategy is the strengthening of local health systems through training and capacity building of local health professionals in the identification and management of thalassemias. This includes education about the symptoms of the disease, available diagnostic methods, and appropriate treatment options. Additionally, partnerships between governmental, non-governmental, and
academic organizations can help provide resources and technical assistance for implementing screening and diagnostic programs in underdeveloped communities (Hossain et al., 20217).

Strategically adopting integrated approaches that combine effective clinical screening, affordable diagnostic tests and training of local health professionals, it is possible to significantly improve the thalassemia diagnosis process in underdeveloped countries, thus enabling timely access to treatment and improved outcomes for patients affected patients (Tariq et al., 2023).

4 CONCLUSIONS

In view of the above, it is concluded that thalassemia is a genetic disease transmitted by the genetic code and that there is a variation in the type and degree of clinical manifestations. In this sense, it is important that the treatment of patients diagnosed with this condition must be implemented early and completely, to prevent the development of more serious symptoms, such as severe anemia and changes in the liver and spleen.

This study highlights the significance of early diagnosis of thalassemia as a crucial measure in the effective management of this genetic condition. Early identification not only allows for the implementation of appropriate medical interventions and treatments, but also helps reduce health complications and improve patients' quality of life. Therefore, investing in screening and awareness programs is critical to mitigating the negative impacts of thalassemia and promoting better health outcomes for those affected by this disease.

Due to the difficulty of diagnosing this disease in underdeveloped countries, further scientific research with epidemiological research and technological development is necessary to better serve the affected population.
REFERENCES


FRASÃO, G. et al. Thalassemia: find out what the disease is and the importance of early diagnosis. 2022.


