Thalassemia screening and diagnosis: Strategies for early detection and effective management.

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Introduction

Thalassemia is a group of inherited blood disorders characterized by reduced production of hemoglobin, the protein in red blood cells responsible for oxygen transport. The condition results in anemia, and depending on its severity, can significantly impact an individual's quality of life. Effective screening and diagnosis are crucial for early detection, which can lead to timely management and treatment, thereby improving patient outcomes [1].

Thalassemia encompasses various genetic disorders, primarily thalassemia alpha and thalassemia beta, each caused by mutations in the genes responsible for hemoglobin production. These mutations lead to the production of abnormal hemoglobin or reduced hemoglobin synthesis, resulting in anemia and other related symptoms. The severity of thalassemia can range from mild to severe, with the latter often requiring intensive medical intervention [2].

Newborn screening is a critical strategy for the early detection of thalassemia. Many countries have implemented routine screening programs that test blood samples from newborns for abnormal hemoglobin levels. This initial screening helps identify infants at risk of thalassemia before symptoms develop [3].

Once thalassemia is suspected, hemoglobin electrophoresis is a key diagnostic tool. This laboratory test separates different types of hemoglobin based on their electrical charge and size, enabling the identification of abnormal hemoglobin patterns characteristic of thalassemia [4].

Genetic testing plays a crucial role in diagnosing thalassemia, particularly for determining the specific mutations involved. Molecular genetic tests can identify mutations in the alpha or beta globin genes, confirming the diagnosis and providing information on the type and severity of thalassemia. Genetic counseling is also essential for families, as it offers insights into inheritance patterns, risks for future pregnancies, and available preventive measures [5].

Family screening is recommended for individuals with a family history of thalassemia, especially in regions where the condition is more prevalent. Screening relatives can help identify carriers and those at risk of having affected children. Prenatal diagnosis, including chorionic villus sampling (CVS) and amniocentesis, allows for the detection of thalassemia in the fetus, providing expectant parents with information to make informed decisions about their pregnancy [6].

Diagnosing thalassemia can be challenging due to its overlap with other types of anemia and the variability in clinical presentation. In regions with diverse populations, distinguishing thalassemia from other hemoglobinopathies and anemia disorders requires a thorough clinical evaluation and a combination of diagnostic tests. Additionally, mild forms of thalassemia may be asymptomatic and go undetected without routine screening [7].

Effective management of thalassemia involves a multidisciplinary approach that includes regular monitoring, blood transfusions, and iron chelation therapy. For individuals with severe thalassemia, regular blood transfusions are necessary to maintain adequate hemoglobin levels and prevent complications [8].

Educating patients and their families about thalassemia is an integral part of effective management. Understanding the nature of the condition, treatment options, and potential complications helps patients adhere to treatment plans and make informed decisions about their care [9].

Ongoing research continues to advance our understanding of thalassemia and improve treatment options. Recent developments include gene therapy and new drug treatments that aim to correct the underlying genetic defects or reduce the need for frequent transfusions. These innovations hold promise for better management and potentially curative treatments in the future, offering hope for improved outcomes for individuals with thalassemia [10].

Conclusion

Thalassemia screening and diagnosis are essential for early detection and effective management of this inherited blood disorder. By utilizing a combination of new-born screening, haemoglobin electrophoresis, genetic testing, and family screening, healthcare providers can accurately diagnose thalassemia and implement appropriate treatment strategies.

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***Received:** 02-Sep-2024, Manuscript No. AAHBD-24-148127; **Editor assigned:** 04-Sep-2024, PreQC No. AAHBD-24-148127(PQ); **Reviewed:** 16-Sep-2024, QC No. AAHBD-24-148127; **Revised:** 20-Sep-2024, QC No. AAHBD-24-148127(R); **Published:** 27-Sep-2024, DOI: 10.35841/aahbd-7.3.192*

***Citation:** Wulandari J. Thalassemia screening and diagnosis: Strategies for early detection and effective management. Hematol Blood Disord. 2024;7(3):192*

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Citation: *Wulandari J. Thalassemia screening and diagnosis: Strategies for early detection and effective management. Hematol Blood Disord. 2024;7(3):192*