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THALASSEMIA AMONG ADOLESCENTS

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ABSTRACT
Thalassemia is a genetic blood disorder that affects hemoglobin production, leading to anemia, fatigue, and other complications. Among adolescents, thalassemia can impact physical and emotional development, academic performance, and social relationships. As they transition from childhood to adulthood, adolescents with thalassemia face unique challenges, including managing chronic illness, maintaining treatment adherence, and coping with body changes and emotional stress. Early diagnosis, comprehensive care, and support are crucial to help adolescents with thalassemia navigate these challenges and achieve optimal health, well-being, and quality of life. Raising awareness and promoting education are essential to support affected individuals. The abstract on thalassemia among adolescents focuses on the impact of this genetic blood disorder on the health and well-being of young individuals. Thalassemia, characterized by abnormal hemoglobin production leading to anemia, presents challenges for adolescents, including symptoms like fatigue, weakness, and the need for regular medical interventions such as blood transfusions and iron chelation therapy. This abstract highlights the importance of proper management strategies, including treatment adherence, lifestyle adjustments, and emotional support for adolescents living with thalassemia. By raising awareness, providing education, and fostering a supportive environment, adolescents with thalassemia can navigate the complexities of their condition and lead fulfilling lives. The abstract emphasizes the significance of a holistic approach to care that addresses the physical, emotional, and social aspects of thalassemia in adolescents, ultimately aiming to improve their quality of life and overall well-being.

Key Words: *Thalassemia, adolescents, genetic blood disorder, anemia, symptoms, treatment, management, challenges, support, awareness, education, lifestyle, well-being, quality of life..*

INTRODUCTION

Thalassemia is a genetic blood disorder that affects the production of hemoglobin, leading to anemia. Among adolescents, thalassemia can have a significant impact on their health and quality of life. Adolescents with thalassemia often experience symptoms such as fatigue, weakness, pale skin, and jaundice due to the decreased production of healthy red blood cells. This condition requires lifelong management through blood transfusions, iron chelation therapy, and sometimes even bone marrow transplants. Living with thalassemia as an adolescent can be challenging as it may affect their physical health, emotional well-being, and social life. Regular medical check-ups, adherence to treatment plans, and a healthy lifestyle are crucial for managing thalassemia effectively. Additionally, support from family, friends, and healthcare providers is essential in helping adolescents cope with the demands of the condition. Educating adolescents about thalassemia, its treatment, and the importance of adherence to medical recommendations is vital for empowering them to take control of their health. By raising awareness, providing support, and promoting self-care practices, adolescents with thalassemia can lead fulfilling lives despite the challenges posed by this genetic disorder. Thalassemia major (TM) is India's most inherited haemolytic disease due to disruption of beta-globin biosynthesis and less commonly alpha alpha-globin defect. The disease may be thalassemia major or intermedia [Quirolo K, Vichinsky E (2016)]. Carriers of this disease are symptomatic and may pass on the disease in its severe form to their children. According to global statistics, 5% of people of Mediterranean and Southeast Asia countries are carriers of the thalassemia gene [Piel FB, Weatherall DJ,2014]. In India, the incidence of this inherited disorder is 3–4% with an annual estimate of around 8000 to 10,000 new births with major disease, contributing to around 10% of the annual world incidence [Madan N, Sharma S, Sood SK, Colah R, Bhatia HM,2010; Mohanty D et al,2013]. Thalassemia is a hereditary blood disorder resulting in chronic haemolytic anemia disease due to defective hemoglobin synthesis. Thus, erythrocytes of patients with thalassemia have a shorter lifespan (less than 120 days). (Adam S, Afifi H, Thomas M, Magdy P, El-Kamah G,2017).

It is the most common genetic disease worldwide and has been declared a world health problem. (Telfer P,2005) According to the World Health Organization (WHO), thalassemia is the most common genetic disease in the world which has now been declared a world health problem. (Sheikh KA et al ,2014; Dhirar N et al 2016)
Approximately 7% of the world's population has the thalassemia gene. Based on WHO data, about 250 million people in the world (4.5%) carry the thalassemia gene, while 80–90 million of them carry the thalassemia gene. β-Thalassemia major is considered the commonest hemoglobinopathy in the Mediterranean area particularly Egypt with an estimated carrier rate of 9-10.2% [Saboor M et al 2014]. Registered cases of homozygous β-thalassemia in big centres of Egypt in 2006 up to Sept 2007 (n = 9912) [El-Beshlawy A et al 2009]. From about 10,000 registered β-thalassemia cases and more than 20,000 non-registered cases; 95%of them are β-thalassemia major, and 5% are thalassemia intermediate or haemoglobin H disease β-thalassemia is a chronic condition, which put the huge psychosocial burden on the patient and his family. [El-Gindi HD et al 2015]

REVIEW OF LITERATURE

Madan N, Sharma S, Sood SK, Colah R, Bhatia LH et al 2010 conducted a study on Frequency of β-thalassemia trait and other hemoglobinopathies in northern and western India. Indian J Hum Genet. 2010 Jan;16(1):16-25. Doi: 10.4103/0971-6866.64941. PMID: 20838487; PMCID: PMC2927789. The overall gene frequency of βTT in Mumbai and Delhi was 4.05% 2.68% and 5.47% in children of the two cities respectively. In Mumbai, the gene frequency was evenly distributed. Most children with βTT from Mumbai were from Marathi (38.9%) and Gujarati (25%) speaking groups. Gene frequency was >5% in Bhatias, Khatris, Lohanas, and Schedule Castes. In Delhi, a higher incidence was observed in schoolchildren of North and West Delhi (5.8-9.2%). The schoolchildren of North and West Delhi are predominantly of Punjabi origin compared to children in the South of the city (2.2%, 2.3%). When analysed state-wise, the highest incidence was observed in children of Punjabi origin (7.6%) and was >4% in several other states. The majority of the traits from Mumbai were anaemic (95.1% male and 85.6% female). The prevalence of anemia was lower (62.7% male and 58.4% female) in children with βTT from Delhi. This was a reflection of the higher prevalence of anemia in children without hemoglobinopathy in Mumbai than in Delhi. Nutritional deficiency was probably more severe and rampant in children in Mumbai. The gene frequency of Hb D was greater in schoolchildren from Delhi (1.1%) than in Mumbai (0.7%). The Hb S trait (0.2%) was observed exclusively in children from Mumbai. A low incidence of Hb E trait (0.04%) was seen in children in Mumbai. A higher incidence is reported from the East. The number of cases studied from the eastern region was small as the data from the East (Kolkata) could not be included in the analysis.
Cappellini MD, Robbiolo L, Bottasso BM, Coppola R, Fiorelli G, Mannucci AP,2000 conducted a study on Venous thromboembolism and hypercoagulability in splenectomised patients with thalassaemia intermedia. Results: A high prevalence of thromboembolic events was found, particularly in splenectomised patients with thalassaemia intermedia (29%). These patients had high plasma levels of markers of coagulation and fibrinolysis activation. Furthermore, thalassaemic red cells and erythroid precursors from splenectomised patients with thalassaemia intermedia had an enhanced capacity to generate thrombin. To evaluate the role of splenectomy per se on procoagulant activity, we evaluated the capacity to form thrombin in healthy individuals who had been splenectomised for trauma. They produced the same amount of thrombin as non-splenectomised controls. In conclusion, the results of this study show the existence of a hypercoagulable state in splenectomised patients with thalassemia intermedia and that their red and erythroid cells are capable of acting as activated platelets in thrombin generation.
Adam S, Afifi H, Thomas M, Magdy P, and El-Kamah G, 2017 conducted a study on Quality-of-Life Outcomes in a Paediatric Thalassemia Population in Egypt. Hemoglobin.

RESULTS:

The Arabic version of SF36 tool was used to assess HRQoL outcomes. Socioeconomic data were collected by patient and parent interviews. Clinical data were collected by review of medical records. One hundred and thirty patients and 60 controls were enrolled, with a mean age of 5.4 ± 3.2 years and 6.3 ± 3.0, respectively. The HRQoL outcome scores were lower in all domains in the thalassemia group compared to the control group ($p = 0.0001$). Transfusion-dependent (TD) patients had lower HRQoL scores compared to non-transfusion-dependent (NTD) patients ($p = 0.0001$). Patient education and maternal education were independently associated with better HRQoL scores ($p = 0.007$, $p = 0.028$, respectively). Residents of rural areas reported lower scores compared to urban residents ($p = 0.026$).

CONCLUSION

In conclusion, thalassemia among adolescents poses significant physical, emotional, and social challenges. Effective management requires a multidisciplinary approach, including regular blood transfusions, iron chelation therapy, and monitoring for complications. Adolescents with thalassemia need support to maintain treatment adherence, cope with emotional stress, and navigate transitions to independence. With comprehensive care, education, and support, adolescents with thalassemia can lead active, productive lives and reach their full potential. Increased awareness and advocacy are crucial to improve healthcare services, reduce stigma, and enhance the quality of life for adolescents with thalassemia. Early intervention and support can make a significant difference.

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