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A comparative sociological examination of the challenges faced by Thalassaemia patients at PGIMS, Rohtak, and PGIMER, Chandigarh.

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Abstract

The Presented research paper outlines the causes, symptoms, and social challenges faced by individuals with Thalassaemia, a genetic disorder prevalent in regions such as Africa, the Mediterranean Basin, the Middle East, the Indian Subcontinent, South East Asia, Melanesia, and the Pacific Islands. Approximately 3% of the global population are beta-thalassaemia carriers, with India having the highest number of carriers. Alarming statistics show that one in every 30 children is born with the Thalassaemia trait/mutated gene, posing a significant risk of the gene being passed on to the next generation. About 20% of the world's population is affected by alpha and beta Thalassaemia, with India bearing a heavy burden of 40 million Thalassaemia patients, including 1 lakh Thalassaemia major patients requiring regular blood transfusions. Tragically, approximately 3000 children aged 10 to 20 years die annually due to undiagnosed iron overload. Prenatal chorionic villus sampling during the 11-13th week of pregnancy offers a means to prevent the birth of affected children.

Keywords: Thalassaemia, genetic disorder, blood transfusions, iron overload, pre-natal chorionic villus sampling.

Introduction:

Thalassaemia is a genetic blood disorder characterised by inadequate haemoglobin production, leading to anaemia (Silberstein et al., 2014), (Cooley, 1925). Thalassaemia is a genetic disorder characterised by abnormal production of haemoglobin, the protein in red blood cells responsible for oxygen transport throughout the body. The term "thalassaemia" originates from the Greek word "Thalassa", which translates to "the sea." This nomenclature reflects its historical identification among populations in regions near the Mediterranean Sea, where the condition was first documented. Individuals with thalassaemia typically experience a range of health challenges due to the reduced efficiency of their haemoglobin, leading to anaemia and other related complications (Betts et al., 2020). Professor Cooley, an American paediatrician, first described it. Thalassaemia is a genetic blood disorder that is inherited from parents. Approximately 0.44 in 1,000 children worldwide are born with Thalassaemia major. In the Indian subcontinent, approximately 10% of the global Thalassaemia population is affected by this chronic illness due to a lack of awareness. The disorder is caused by mutations in the genes responsible for producing the beta-globin protein (Rund & Rachmilewitz, 2005).

This leads to the dysfunctional synthesis of normal haemoglobin, resulting in the different clinical manifestations of Thalassaemia, including Minor, Intermedia, and Major forms (Galanello & Origa, 2010). Thalassaemia exists in several forms, with Thalassaemia minor being the least severe. This condition is characterised by one mutated beta-globin gene and one normal gene (Shafique et al., 2023). Individuals with Thalassaemia minor typically maintain normal iron levels and exhibit minimal clinical symptoms. In contrast, Thalassaemia

intermedia, or non-transfusion-dependent Thalassaemia, presents with more pronounced symptoms but requires less frequent transfusions than the Major form. Thalassaemia major, called homozygous Cooley's anaemia, is the most severe variant resulting from the inheritance of two mutated beta-globin genes. This leads to severe anaemia, necessitating regular blood transfusions to sustain adequate patient haemoglobin levels (Sanchez-Villalobos et al., 2022; Musallam et al., 2023).

The complications associated with major thalassaemia are significant. The reduced lifespan of red blood cells, coupled with the need for frequent transfusions, leads to iron overload in the body (Taher & Saliba, 2017), which can have detrimental effects on vital organs such as the liver, kidney, and heart (Basu et al., 2023). This can result in conditions like cirrhosis of the liver and an increased risk of liver cancer (Lin et al., 2023). Iron accumulation can lead to health issues, including weak bones and an increased risk of osteoporosis and fractures after age 20. More than one lakh Thalassaemia patients across the country die before the age of 20 (Rao et al., 2024). The awareness and understanding of Thalassaemia have significantly improved since its initial description in 1925, and early detection and intervention have become crucial in managing the disease (Ali et al., 2021).

Genomic technologies have enabled preconception and prenatal screening for Thalassaemia traits, allowing for timely interventions to prevent the birth of children with the disorder (Garcia-Herrero et al., 2020). More than one lakh Thalassaemia patients across the country die before the age of 20 (Isalkar, 2018). It is essential for healthcare professionals, especially gynaecologists, to advocate for and perform appropriate screening tests, such as HB Electrophoresis or HBA2 testing, during pregnancy to identify Thalassaemia traits in expectant mothers (Baxi et al., 2012). This can enable the implementation of preventive measures and informed decision-making regarding managing the disorder in the offspring (Cao & Kan, 2013). The overall goal of managing Thalassaemia is to improve the quality of life for affected individuals by minimising the disorder's impact through early detection, appropriate medical interventions, and supportive care (Cappellini et al., 2018).

Objective of the research: This research focuses on deepening the understanding of Thalassaemia, a complex blood disorder, while effectively conveying its preventive measures to the general public. The study also aims to uncover the various social, economic, and personal challenges individuals living with Thalassaemia encounter daily. By highlighting these issues, the research aspires to foster greater awareness and support for those affected by the disorder.

Methodology: The research employs a descriptive methodology designed to comprehensively gather data through structured interview schedules with patients diagnosed with Thalassaemia Major, currently undergoing treatment at the Advanced Paediatric Centre in Chandigarh PGI and PGIMS in Rohtak. The data analysis process utilizes sophisticated statistical techniques, particularly on SPSS software, to ensure that the results are presented in a clear, coherent, and standardised format. This approach enhances the clarity of the findings and contributes to a deeper understanding of the patient experience and treatment outcomes associated with Thalassaemia.

Result and Discussion:

The research study collected comprehensive data from 99 individuals diagnosed with Thalassaemia major, comprising 50 males and 49 females. These participants were sourced from various regions, including Haryana, Chandigarh, Punjab, and Uttar Pradesh. The majority of the patients were relatively young, with most falling within the age range of 3 months to 10 years. Diving deeper into the age distribution, the findings revealed that 21% of the respondents were adolescents between 11 and 15 years, while 10% belonged to the 16 to 20 years age bracket. Additionally, 15.9% of the participants were young adults aged 21 to 25. Notably, two patients over the age of 35 also participated in the study, both of whom required regular blood transfusions approximately every 20 days to manage their condition effectively.

The distribution of patients according to their locations reveals a diverse geographical representation. A significant portion, 18%, hailed from Rohtak, while Bhiwani contributed 9% to the patient pool. Panipat and Sonipat accounted for 13% of the patients, highlighting their importance in this demographic. From Chandigarh, 6% of the patients were recorded, and there was a notable presence from Mohali, Patiala, Sangrur, and Fatehgarh Sahib, contributing 12%, 11%, 9%, and 8%, respectively. In contrast, only a tiny fraction, 4%, of patients originated from Uttar Pradesh. It is important to note that thalassaemia patients were identified in other districts across these states; however, there remains a troubling gap in information concerning particular areas, warranting attention and further investigation.

Approximately 40% of parents discovered their child's Thalassaemia diagnosis when the child was just 3 to 6 months old, while the majority had come to terms with the diagnosis by the time their child reached one and a half years of age. The findings of a recent study revealed that around 15% of respondents had children under one year old who required regular blood transfusions, highlighting the critical need for medical intervention at such an early stage. For most of these young patients, transfusions were necessary every 20 days, underscoring the ongoing, diligent care required to manage their condition. Moreover, the study shed light on issues surrounding the awareness and accessibility of the nucleic acid amplification test (NAAT) technique, which is vital for diagnosing and managing Thalassaemia. It was found that nearly 42% of Thalassaemia patients remained uninformed about the NAAT, primarily due to the limited availability of testing services in certain regions. This significant gap in testing not only raises alarms regarding the effective management of Thalassaemia but also poses serious health threats, such as the potential transmission of serious infections like HIV and HCV among patients who may not receive timely and adequate screening.

In the case of blood transfusions, the findings from the study highlighted a significant delay in the process, indicating that it typically takes a minimum of 10 hours from the moment a blood sample is collected until the patient receives the transfusion. This protracted waiting period often leaves patients in uncomfortable positions, as they frequently do not have access to a bed at the medical centre during the wait. At the Rohtak Post Graduate Institute (PGI), where an average of 15 to 20 patients undergo transfusions daily, there is a concerning shortage of resources, with only 7 beds available for these critical procedures. In contrast, Chandigarh PGI is noted for its ample facilities, which alleviate some of the congestion and discomfort experienced by patients.

Moreover, the study shed light on the necessity of iron chelation therapy, mainly due to the elevated ferritin levels commonly found among patients. Citing research from Cianciulli (2009)

and Wang et al. (2010), it was revealed that most patients exhibited ferritin levels ranging from 1000 to 3000 ng/ml, with a striking 28.6% showing levels between 3000 to 5000 ng/ml. Respondents provided valuable insights into dietary and medicinal strategies to manage these elevated ferritin levels. They recommended steering clear of iron-rich foods, adhering to specific medication regimens, and incorporating more citrus fruits into their diets, which could significantly enhance patient outcomes in the long run.

The study underscores the importance of genetic counselling and prenatal testing protocols for Thalassaemia. It reports that some parents engage in genetic counselling prior to trying for a second child and may opt for Chorionic Villus Sampling (CVS) during pregnancy to determine whether the foetus has inherited the Thalassaemia gene (Bhattacharya et al., 2019). Thalassaemia pathophysiology reveals that the bone marrow, responsible for haematopoiesis, plays a crucial role in producing erythrocytes, leukocytes, and platelets. A bone marrow transplant remains the primary therapeutic intervention for Thalassaemia; however, this procedure is intricate and incurs significant costs (Lucarelli et al., 2012). Research has established that optimal outcomes are associated with transplants conducted before age 14 (Caocci et al., 2017). Patients with Thalassaemia frequently encounter challenges in securing blood for transfusions, with some resorting to purchasing blood amid shortages. Supportive measures are facilitated by various Thalassaemia welfare organisations, such as the Haryana Thalassaemia Welfare Society and the National Thalassaemia Welfare Society. These organisations are instrumental in orchestrating blood donation drives, providing medications at subsidised rates, and conducting blood screening camps. Moreover, Thalassaemia has profound implications for patients' educational pursuits, often hindering their ability to engage in academic endeavours.

The government of Haryana provides free bus passes, medicines, and blood transfusions to Thalassaemia patients, but respondents suggest that these facilities should be available in their hometowns. While most patients report no employment issues due to the disorder, the need for regular blood transfusions affects their ability to plan and enjoy outdoor activities. Thalassaemia patients are eligible for benefits under the Disability Bill passed by the Government of India in 2016. However, many are unaware of this bill, and only a tiny percentage have obtained a disability certificate due to administrative complexities. Most respondents emphasised the importance of regular blood transfusions, iron chelation therapy, strict medication adherence, and a well-rounded diet to ensure Thalassaemia patients' good quality of life. There is growing optimism around the potential of gene therapy and CRISPR gene editing (Frangoul et al., 2020) among patients, with many expressing reservations about continuing current medications due to undisclosed risks and benefits.

Respondents' perspectives on strategies to curb the further spread of Thalassaemia Were Examined. A significant consensus emerged advocating for the implementation of the HBA2/HPLC/HB Electrophoresis test. This test, which is crucial for identifying individuals affected by Thalassaemia, should be made freely accessible in all government hospitals. Moreover, there was a strong recommendation for ASHA (Accredited Social Health Activist) workers to actively engage with women who are approximately one month into their pregnancies. They should encourage these women to undergo testing for their Thalassaemia status, thereby taking proactive measures to mitigate the risk of passing the gene onto their offspring. Additionally, respondents proposed an important regulation: Individuals should be required to submit their genetic profiles when applying for their ADHAR cards or enrolling in educational institutions. This initiative could help create a more informed population and assist in the prevention of Thalassaemia transmission among future generations.

Summary and conclusion:

The study conducted an in-depth analysis of 99 individuals diagnosed with Thalassaemia Major across the regions of Haryana, Chandigarh, Punjab, and Uttar Pradesh. Findings indicated a significant frequency of blood transfusions required by paediatric patients, particularly those aged 3 months to 10 years, with some cases necessitating transfusions as frequently as every 20 days. The investigation raised critical concerns regarding the inadequacy of comprehensive testing protocols in certain geographic areas, underlining an urgent need for enhanced awareness and accessibility to nucleic acid amplification testing (NAAT) to mitigate the transmission risk of pathogens such as HIV and HCV. Logistical hurdles emerged as a significant theme, with patients facing challenges, including a deficit of available beds in transfusion centres, leading to protracted waiting periods. The research also stressed the necessity of managing elevated ferritin concentrations through iron chelation therapy, which is crucial in preventing secondary complications.

Moreover, the findings highlighted the multifaceted challenges encountered by Thalassaemia patients beyond clinical aspects, such as their impacts on educational attainment, employment opportunities, and overall quality of life. The study acknowledged the vital contributions of Thalassaemia welfare societies, including the National Thalassaemia Welfare Society and the Panipat Thalassaemia Welfare Society, in delivering support and coordinating essential services for affected populations. Additionally, the study examined the landscape of governmental assistance, advocating for the localization of these facilities within patients' hometowns to improve accessibility. It also pointed out the administrative difficulties that patients face in obtaining disability certifications.

In conclusion, the study recognized the resilience and proactive measures taken by Thalassaemia patients and their families and highlighted critical areas requiring enhancement in healthcare infrastructure, public awareness initiatives, and supportive services.

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