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LIVED EXPERIENCES OF ADULT THALASSEMIA PATIENTS VISITING A TERTIARY CARE HOSPITAL IN PESHAWAR

Original Research

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ABSTRACT

Background: Thalassemia is an autosomal recessive blood disorder characterized by defective hemoglobin synthesis leading to chronic anemia and dependence on lifelong medical care. Beta thalassemia major, the most severe form, not only imposes significant medical challenges but also disrupts education, social participation, and overall quality of life. Understanding the lived experiences of patients is essential for informing holistic care strategies and policy responses.

Objective: The study aimed to explore the challenges encountered by adult patients with beta thalassemia major in managing their treatment, educational activities, and health-related quality of life.

Methods: This qualitative study employed a hermeneutic phenomenological approach to capture the lived experiences of affected individuals. Purposive sampling was used to recruit 13 participants (8 males and 5 females) from a tertiary care hospital in Peshawar. Participants were aged 18 years and above and diagnosed with beta thalassemia major. Data were collected through semi-structured, in-depth interviews conducted in the native language for clarity and comfort. All interviews were audio-recorded, transcribed verbatim, and translated into English. Thematic analysis was performed following Braun and Clarke's six-step framework to generate themes and subthemes. Ethical approval and informed consent were secured prior to data collection.

Results: Analysis revealed three main themes: treatment-related challenges, education-related challenges, and health-related quality of life. Under treatment challenges, 11 participants reported difficulties in arranging regular blood donors, 9 highlighted frequent shortages of chelation or supportive medications, and 10 described severe financial hardships due to out-of-pocket treatment costs. Educational barriers were noted by 8 participants, with discontinuation or poor performance linked to physical weakness, cognitive burden, or lack of parental support. Regarding health-related quality of life, 12 participants reported limitations in physical wellbeing, 7 expressed psychological stress such as anxiety or depression, while most described diminished social functioning due to dependency and social isolation.

Conclusion: The study concluded that adult patients with beta thalassemia major face interconnected medical, educational, financial, and social challenges. These findings emphasize the urgent need for comprehensive interventions, including sustained medication supply, organized donor networks, financial assistance, educational support, and culturally appropriate psychosocial care to improve the quality of life of this vulnerable population.

Keywords: Adult, Anemia, Beta Thalassemia, Blood Transfusion, Chelation Therapy, Quality of Life, Socioeconomic Factors.

INTRODUCTION

Thalassemia is an inherited autosomal recessive blood disorder characterized by chronic anemia resulting from the body's inability to synthesize adequate amounts of hemoglobin, a protein essential for oxygen transport in red blood cells (1). Mutations in the genes responsible for hemoglobin synthesis lead to defective or absent globin chains, and the condition is transmissible from parents to offspring (2). Among the different types, beta thalassemia major represents the most severe form, where either markedly reduced or absent beta-globin production causes life-threatening anemia requiring lifelong medical care (3). The genetic risk is particularly high when both partners are carriers, as there is a 25% chance that their child will inherit the disease (4). This inheritance pattern underscores the importance of preventive strategies in populations with high carrier frequencies. Globally, the burden of thalassemia is substantial, with an estimated 80–90 million people, or about 1.5% of the population, carrying beta thalassemia genes. The highest prevalence is observed in the Middle East, Mediterranean regions, Central Asia, and the Indian subcontinent, while Southeast Asian countries, including Indonesia, Thailand, and India, account for nearly half of all global carriers and affected births. In contrast, developed regions such as Europe and North America contribute a smaller proportion, approximately 10–13% of carriers (5,6). According to the World Health Organization, around 4.5% of the global population, amounting to 250 million individuals, is affected by thalassemia or other inherited hemoglobin disorders (7). In Pakistan, the prevalence of beta thalassemia carriers is estimated between 5–7%, translating to more than 10 million individuals. Each year, approximately 5,000 new cases of beta thalassemia major are diagnosed, placing a significant strain on the healthcare system (8).

The consequences of thalassemia extend beyond medical complications, encompassing psychosocial, emotional, and financial challenges for both patients and their families. Many affected children come from socioeconomically disadvantaged backgrounds, where families are unable to afford the lifelong costs of treatment, including regular blood transfusions and iron chelation therapy. This economic disparity contributes to higher morbidity and mortality rates, particularly in low- and middle-income countries where an estimated 5–10 million children die annually from beta thalassemia and related complications (9). Preventive strategies such as premarital screening programs have shown promise in reducing new cases; however, in Pakistan such initiatives remain limited to provincial levels in Sindh, Khyber Pakhtunkhwa, and Balochistan, with no coordinated nationwide program yet implemented (8). Despite the advances in diagnostic and therapeutic approaches, there is a pressing need to understand the lived experiences of patients with thalassemia major. Chronic illness management extends beyond medical care, encompassing challenges in education, employment, social integration, and mental health. While considerable epidemiological data exist on prevalence and genetics, there remains a gap in literature exploring how individuals cope with the condition in their everyday lives. Addressing these dimensions is critical for developing holistic care strategies that go beyond the biomedical model. The present study was therefore designed to explore the multifaceted challenges faced by adult patients living with beta thalassemia major, with the aim of providing insights that can inform patient-centered care and targeted interventions.

METHODS

The study was conducted at the Hematology Day Care Unit of Hayatabad Medical Complex, a tertiary care hospital in Peshawar, where adult patients with beta thalassemia major routinely receive treatment and follow-up care. An interpretive phenomenological approach was employed, as this design is particularly suited to exploring the lived experiences of individuals and allows for a deeper understanding and interpretation of their subjective realities (10). Purposive sampling was used in line with interpretive phenomenological analysis guidelines, and data were collected from 13 participants until thematic saturation was achieved (11). Among these participants, eight were male and five were female, all of whom were 18 years of age or older and diagnosed with beta thalassemia major. Patients who were hemodynamically unstable or undergoing psychiatric treatment at the time of data collection were excluded from the study to minimize confounding factors and ensure participant safety. Data were obtained through in-depth face-to-face interviews complemented by observation. An interview guide consisting of open-ended questions was used to encourage participants to narrate their experiences freely. The interviews were conducted in Pashto, the native language of the participants, to ensure comfort and clarity, and were audio-recorded with permission. The recordings were later transcribed verbatim and translated into English for analysis. The timing of

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interviews was mutually agreed upon with participants to minimize inconvenience. In-depth interviewing was chosen as it provides the researcher with an opportunity to obtain rich, detailed insights into participants' lived experiences (12).

Ethical principles were strictly observed throughout the research process, ethical approval was obtained from the relevant Institutional Review Board (IRB). Participation was voluntary, confidentiality and anonymity were ensured, and informed consent was obtained from all participants before enrollment. Approval for data collection was secured from the hospital administration, and the study was conducted in compliance with established ethical guidelines. Data collection and analysis were conducted simultaneously to ensure reflexivity and to refine emerging insights during the process. Thematic analysis was applied following the six-step iterative framework outlined by Braun and Clarke, which included familiarization with the data, generating initial codes, identifying and reviewing themes, defining and naming themes, and incorporating illustrative quotations from participants to substantiate findings (13). Open, axial, and selective coding strategies were used in sequence. Initially, open codes were identified, which were then organized through axial coding into nine subthemes. These subthemes were subsequently condensed into three overarching themes: treatment-related challenges, education-related challenges, and issues related to health-related quality of life. Rigor and trustworthiness of the study were ensured by adhering to four key criteria: credibility, transferability, dependability, and confirmability (14,15). Credibility was established by triangulating data collection methods and validating findings with participants and subject experts. Transferability was supported by providing thick descriptions of the context and participants, allowing readers to assess applicability to other similar settings. Dependability was achieved through an inquiry audit, ensuring consistency of findings if the study were repeated under similar conditions. Confirmability was maintained through reflexivity and triangulation, reducing the risk of researcher bias. The transcripts were revisited multiple times to enhance familiarity with the data, and supportive quotes were incorporated into the analysis to ensure that the voices of participants were authentically represented.

RESULTS

The findings revealed that the majority of participants demonstrated an adequate level of knowledge regarding their disease. Except for a small number, most participants recognized thalassemia as a genetic blood disorder, commonly associated with consanguineous marriages. However, awareness about premarital screening for carrier detection was markedly low. The results were organized into three overarching themes with associated subthemes.

Treatment related Challenges

Medicines related Challenges: Shortage of medicines, especially iron chelation therapy, was one of the most critical issues reported. Although the hospital provided some medications free of cost, occasional shortages were noted, and patients were often required to purchase additional prescribed drugs themselves. Due to financial constraints, most patients reported using fewer medications than prescribed. One participant stated, *"Another thing is… sometimes, tab disperone [chelation therapy] is not given to us from here. And we also use a number of other medicines for pain, which are also not provided from here. So, when there is a shortage of medicine, we go to buy it in the market with our own money"* (R.2). Another remarked, *"Some of the medicines are provided by the hospital, and others, we buy it with our own money. Although many medications are prescribed [by the physician] for me, I take few of them"* (R.13).

Blood Transfusion related Challenges: All participants required periodic blood transfusions, with the frequency depending on age and activity level. While the hospital provided blood free of cost during alternate visits, patients were expected to arrange donors on subsequent occasions. This requirement posed significant psychological and practical difficulties. A participant shared, *"I start asking my father fifteen days earlier than the prescribed date of transfusion to search the donor. He starts requesting people for donation. When the assigned date is reached, sometimes we are successful in finding a donor and sometimes we fail to do so…Blood arrangement is a big problem"* (R.6). Another noted, *"It is a very difficult job to find a blood donor. We start searching for donor a month earlier than the assigned date of blood transfusion"* (R.2). While relatives were often initial donors, over time they became reluctant, and students from universities and madrassas were identified as more generous donors.

Financial Challenges: The majority of participants reported severe financial hardship. Many families survived on the income of low-paid laborers, making it impossible to afford out-of-pocket treatment costs. The problem was further compounded in families with more than one thalassemic child. One participant shared, *"These medicines are very costly. If I take all the prescribed medications it costs about Rs. 50,000 per month. My little sister is also thalassemic. So, we don't buy medicines from outside"* (R.3). Another reported, *"I cannot do any work and laying all the day at home. If I try to do a little work… my breathing gets worse"* (R.9). Many linked their

struggles to the rising inflation, as expressed by one respondent, *"The prices of everything is so high. Everything is so costly… floor, ghee, pulses, vegetables, everything is beyond above the financial level of poor man. I alone receive free medications amounting to 45000–48000 every month. So just think …how can I buy it from the market?"* (R.5).

Education related Challenges

Physical Weakness: Chronic fatigue and low hemoglobin levels were cited as major barriers to attending school regularly. A participant described, *"It happens that when the level of my blood is decreased, then I feel weakness and I also lose interest in doing things. Then, neither, I want to eat anything, nor I wish to go outside of my home"* (R.1). Another added, *"I am not able to walk too much. When I walk my heart starts beating fast and then my breathing also deteriorates. I could not continue schooling because of my illness. We couldn't pay for transportation to school, so I stopped schooling"* (R.3).

Cognitive Burden: Participants also reported mental exhaustion and difficulty concentrating due to disease-related symptoms. *"Because of my illness, I could not go to school. I used to feel dizziness and over-burdened due to school work. When I was in grade one, I discontinued my schooling"* (R.13). Another participant explained, *"After passing the fifth class, I discontinued my schooling, because I felt suffocated in school due to noise"* (R.12).

Lack of Interest: Some participants admitted that personal or parental lack of motivation contributed to discontinuing education. *"We used to go to school but personally I didn't like school. So I told my father that I didn't want to continue my education. My father respected my opinion and at that time I stopped going to school"* (R.6). Conversely, a few participants persevered and achieved higher education despite their illness. One respondent reported, *"Until passing my secondary school certificate examination, things went quite good, but when I entered the college, the college staff was not that much cooperative… then I continued my studies as private student, and recently I took my final examination of master program in humanities"* (R.5).

Health related Quality of Life

Physical Wellbeing: Persistent fatigue and pain restricted daily functioning and recreational activities. *"I cannot play any game. All the day and night I stay at home and I have no friends"* (R.3). Another participant shared, *"When our blood level drops, we feel difficulty in walking, and when we walk, we develop difficulty in breathing"* (R.6).

Psychological Wellbeing: Participants frequently reported depression, anxiety, and distress related to hospital visits. One participant explained, *"Initially, I was enrolled both in school and madrassa, and at that time I was suffering from severe depression… I used to be feeling fine for a month and then for the following two or three months, I used to be suffering from depression"* (R.2). In contrast, some participants described acceptance and resilience, such as, *"I have no psychological problems, not at all. Every human is destined to die a day, and therefore, we don't worry about these things. This is what Allah has decided for us and we accept it happily"* (R.11).

Social Functioning: Many participants experienced impaired social interactions and limited opportunities for employment or marriage planning. *"I cannot do any heavy work or business. I have two goats; I take them for grazing and keep myself busy that way"* (R.11). Another participant revealed, *"Currently I do nothing. But yes, I intend to establish my own business in future. I want to start business of dealing in clothes, because, the business of clothes keeps on running throughout the year"* (R.2). In contrast, a minority reported maintaining active social and occupational roles. One participant expressed, *"My life is so good, I perform all activities in life. I perform my prayers, cook the meals, and when I wish I also sew the clothes. I also have my own boutique and make handy crafts… My relationship with friends is also good. I don't feel to be suffering from disease"* (R.6).

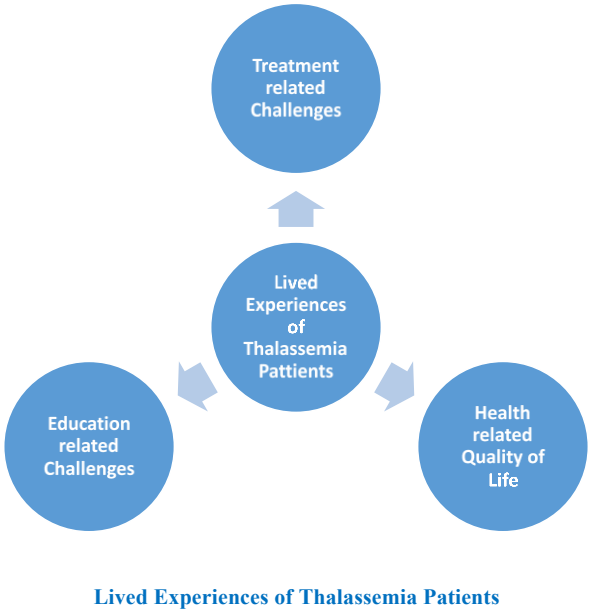

Figure 1 Lived Experiences of Thalassemia Patients

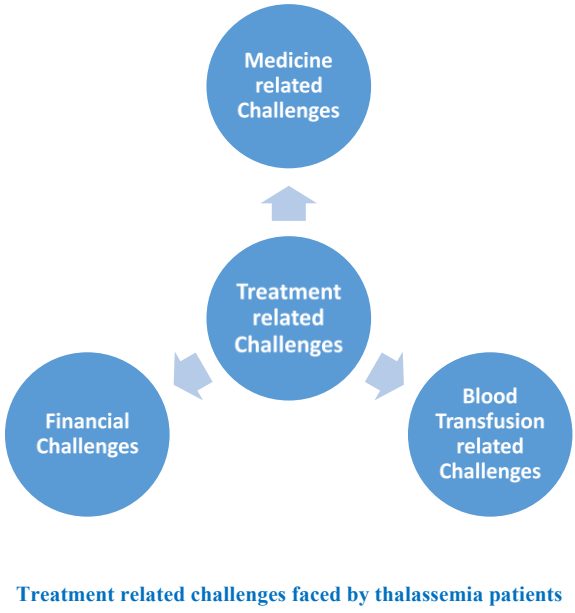

Figure 2 Treatment Related Challenges Faced by Thalassemia Patients

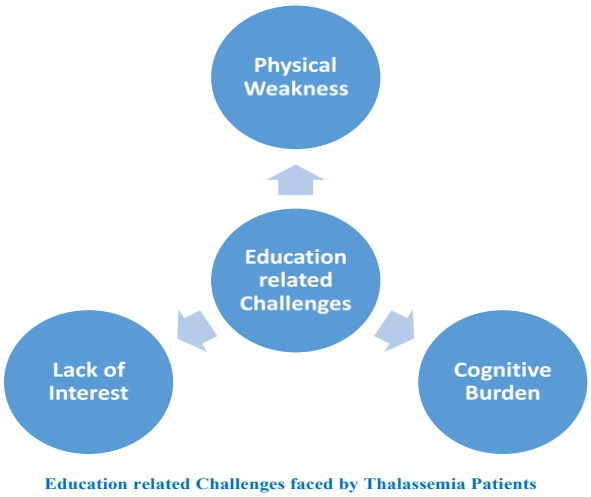

Figure 3 Education Related Challenges Faced by Thalassemia Patients

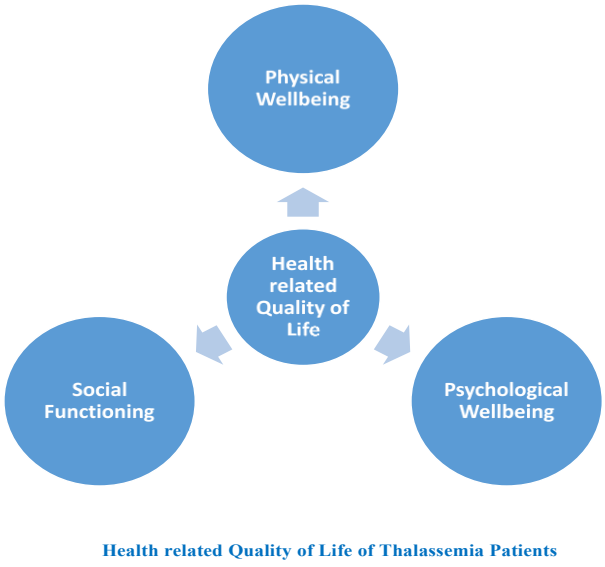

Figure 4 Health Related Quality of Life of Thalassemia Patients

DISCUSSION

The study offered a nuanced account of how adult Pakistanis living with beta thalassemia major navigated everyday life, situating their experiences within a hermeneutic phenomenological frame that foregrounded meaning, context, and lived realities. Three interlocking domains—treatment, education, and health-related quality of life—captured the breadth of challenges described by participants and illuminated where existing services and policies fell short. Treatment-related burdens clustered around the practicalities of medication access, the logistics of recurring transfusions, and the steady financial erosion from long-term care. These findings converged with prior reports that transfusion pathways carry risks and recurrent obstacles, including reactions and exposure to infections, underscoring the fragility of transfusion-dependent care systems (16,17). The persistent financial strain described by participants also aligned with literature that framed thalassemia management as a high-cost condition whose sustainability depends on coordinated policy, supply-chain reliability, and targeted social protection (18). A distinctive contribution of the present work was the recurrent mention of

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intermittent shortages of iron chelators and adjunctive medications—an operational gap that had not been emphasized in prior syntheses and that warrants immediate health-system attention. Educational trajectories were shaped by chronic fatigue, mobility constraints, and cognitive load, leading to interrupted schooling and reduced academic performance. This pattern echoed earlier studies linking disease-related immobility, persistent tiredness, and anemia with absenteeism and academic underachievement (19,20). The dataset extended that narrative by drawing attention to the role of motivation and social messaging: some participants reported diminished personal interest in school or parental discouragement, often rooted in fears about health deterioration. This element, seldom foregrounded in previous accounts, suggests that educational outcomes were not only biologically mediated but also socially constructed—shaped by family beliefs, peer attitudes, and the perceived cost–benefit calculus of continued study under chronic illness. Interventions that couple health stabilization with family-centered counseling and school accommodations may therefore hold disproportionate value.

The health-related quality of life findings presented a layered picture. Physical functioning was consistently constrained by breathlessness, pain, and low exercise tolerance, consonant with evidence that physical well-being tracks disease severity and transfusion adequacy (21). Social participation contracted as physical symptoms intensified, with reduced sport, curtailed friendships, and limited work prospects. Notably, the psychological profile diverged from much of the published evidence. Many participants reported relative psychological steadiness or a faith-anchored acceptance of illness, in contrast to studies that documented prominent anxiety, depression, and wider emotional or behavioral sequelae linked to frequent hospitalizations, treatment burden, chelation side-effects, and existential concerns (22-24). Reports of psychological and social maladjustment approaching 80% prevalence in other settings further highlighted the discrepancy (25). Several explanations merit consideration: the community's cultural and religious coping resources may have buffered distress; social desirability and stigma could have suppressed disclosure; and the qualitative focus on meaning may have privileged narratives of resilience over symptom cataloging. The implication is not that distress was absent, but that it may have been under-recognized or differently expressed. These patterns carried practical implications. First, stabilizing the chelation supply chain, integrating analgesia and essential supplements into hospital formularies, and formalizing donor-recruitment support would address the most immediate pressure points. Embedding patient navigators within transfusion services could reduce the monthly scramble for donors while improving safety monitoring for reactions. Second, education-health bridges—transport vouchers, flexible attendance policies, quiet rooms for rest, and individualized learning plans—could keep motivated students in school while mitigating the cognitive and physical load that precipitates dropout. Third, routine psychosocial screening using culturally adapted tools, paired with brief counseling, peer-support groups, and faith-sensitive psychoeducation, would permit early detection of distress without pathologizing adaptive acceptance. Finally, public-health messaging that normalizes premarital carrier screening and strengthens awareness around non-consanguineous risks would complement clinical care and reduce incident cases over time. Methodological strengths reinforced the credibility of these conclusions. The hermeneutic phenomenological design captured depth and nuance; purposive sampling targeted individuals with rich experiential knowledge; interviews in the participants' native language enhanced authenticity; and trustworthiness was supported through triangulation, member checking, thick description, and an audit approach to dependability (14,15). Conducting analysis alongside data collection allowed reflexive refinement and ensured that emergent insights guided subsequent interviews.

Several limitations tempered interpretation. The single-center, small sample constrained transferability and precluded subgroup analysis by age, sex, transfusion frequency, or socioeconomic status. Self-reported experiences remained susceptible to recall and social-desirability biases, particularly for sensitive psychological content. The absence of standardized, validated measures for quality of life or mental health limited comparability with quantitative benchmarks and may partly explain divergence from prior prevalence estimates of psychological morbidity. Future work would benefit from multi-site sampling, integration of validated patient-reported outcome measures, and mixed-methods triangulation that pairs narrative depth with quantifiable frequencies of key challenges. Taken together, the findings affirmed well-described burdens in transfusion-dependent thalassemia while extending the conversation to medication stockouts, donor-arrangement labor, and the social construction of educational withdrawal. They suggested a care agenda that moves beyond biomedical maintenance to encompass supply-chain stewardship, school-system partnership, and culturally grounded psychosocial care. Policy levers that subsidize lifelong chelation, institutionalize donor registries, and mainstream carrier screening would likely yield the greatest population-level gains. Subsequent research should examine implementation models for donor support and chelation logistics, test school-based accommodations in pragmatic trials, and explore culturally specific expressions of resilience and distress to ensure that psychological needs are recognized and met within the lived worlds of patients and families.

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CONCLUSION

This study concluded that living with beta thalassemia major profoundly affects multiple dimensions of patients' lives, extending far beyond the biomedical challenges of transfusions and chelation therapy. By using a phenomenological approach, the research highlighted how treatment-related difficulties, educational disruptions, and compromised quality of life intertwine to create a cycle of hardship that is both medical and socio-economic. The insights underscore the urgent need for strengthening health system support, improving access to essential medications, and establishing sustainable blood donor networks. Equally important is the creation of educational and psychosocial support structures that enable patients to pursue meaningful lives despite their chronic illness. These findings emphasize that comprehensive strategies integrating medical, social, and policy interventions are essential for reducing the burden of thalassemia and improving the lived experiences of those affected.

AUTHOR CONTRIBUTION

Author	Contribution
Amanullah Khan*	Substantial Contribution to study design, analysis, acquisition of Data Manuscript Writing Has given Final Approval of the version to be published
Jafaryad Hussain	Substantial Contribution to study design, acquisition and interpretation of Data Critical Review and Manuscript Writing Has given Final Approval of the version to be published
Muzafar Ali	Substantial Contribution to acquisition and interpretation of Data Has given Final Approval of the version to be published
Rahmat Ali Khan	Contributed to Data Collection and Analysis Has given Final Approval of the version to be published

REFERENCES

1. Shafique F, Ali S, Almansouri T, Van Eeden F, Shafi N, Khalid M, et al. Thalassemia, a human blood disorder. Braz J Biol. 2023;83:e246062.
2. Rambod M, Hamidizadeh S, Bazrafshan MR, Parviniannasab AM. Risk and protective factors for resilience among adolescents and young adults with beta-thalassemia major. BMC Psychol. 2023;11(1):231.
3. Musallam KM, Barella S, Origa R, Ferrero GB, Lisi R, Pasanisi A, et al. 'Phenoconversion' in adult patients with β-thalassemia. Am J Hematol. 2024;99(3):490-3.
4. Meloni A, Barbuto L, Positano V, Pistoia L, Spasiano A, Casini T, et al. Pattern and clinical correlates of renal iron deposition in adult beta-thalassemia major patients. Clin Exp Med. 2023;23(7):3573-9.
5. Mensah C, Sheth S. Optimal strategies for carrier screening and prenatal diagnosis of α- and β-thalassemia. Hematology Am Soc Hematol Educ Program. 2021;2021(1):607-13.
6. Zhou BY, Wang YX, Xu SS, Gu H, Li MZ. Molecular Spectrum of α- and β-Thalassemia among Young Individuals of Marriageable Age in Guangdong Province, China. Biomed Environ Sci. 2021;34(10):824-9.
7. Jaing TH, Chang TY, Chen SH, Lin CW, Wen YC, Chiu CC. Molecular genetics of β-thalassemia: A narrative review. Medicine (Baltimore). 2021;100(45):e27522.
8. Taher AT, Cappellini MD, Kattamis A, Voskaridou E, Perrotta S, Piga AG, et al. Luspatercept for the treatment of anaemia in non-transfusion-dependent β-thalassaemia (BEYOND): a phase 2, randomised, double-blind, multicentre, placebo-controlled trial. Lancet Haematol. 2022;9(10):e733-e44.
9. Magrin E, Semeraro M, Hebert N, Joseph L, Magnani A, Chalumeau A, et al. Long-term outcomes of lentiviral gene therapy for the β-hemoglobinopathies: the HGB-205 trial. Nat Med. 2022;28(1):81-8.
10. Calabria A, Spinozzi G, Cesana D, Buscaroli E, Benedicenti F, Pais G, et al. Long-term lineage commitment in haematopoietic stem cell gene therapy. Nature. 2024;636(8041):162-71.

11. De Sanctis V, Soliman A, Daar S, Tzoulis P, Yassin MA, Di Maio S, et al. Insulin-Like Growth Factor -1 (IGF-1) and Glucose Dysregulation in Young Adult Patients with β-Thalassemia Major: Causality or Potential Link? Acta Biomed. 2022;93(6):e2022331.
12. Cazzola M. Ineffective erythropoiesis and its treatment. Blood. 2022;139(16):2460-70.
13. Locatelli F, Lang P, Wall D, Meisel R, Corbacioglu S, Li AM, et al. Exagamglogene Autotemcel for Transfusion-Dependent β-Thalassemia. N Engl J Med. 2024;390(18):1663-76.
14. Khandros E, Blobel GA. Elevating fetal hemoglobin: recently discovered regulators and mechanisms. Blood. 2024;144(8):845-52.
15. Ju W, Din G, Huang J, Zheng M, Wang X, Liu L, et al. Efficacy and safety of thalidomide in patients with β-thalassemia intermedia and major. Medicine (Baltimore). 2024;103(43):e40328.
16. Frangoul H, Altshuler D, Cappellini MD, Chen YS, Domm J, Eustace BK, et al. CRISPR-Cas9 Gene Editing for Sickle Cell Disease and β-Thalassemia. N Engl J Med. 2021;384(3):252-60.
17. Pavani G, Fabiano A, Laurent M, Amor F, Cantelli E, Chalumeau A, et al. Correction of β-thalassemia by CRISPR/Cas9 editing of the α-globin locus in human hematopoietic stem cells. Blood Adv. 2021;5(5):1137-53.
18. Gupta V, Kumar I, Raj V, Aggarwal P, Agrawal V. Comparison of the effects of calcium channel blockers plus iron chelation therapy versus chelation therapy only on iron overload in children and young adults with transfusion-dependent thalassemia: A randomized double-blind placebo-controlled trial. Pediatr Blood Cancer. 2022;69(6):e29564.
19. Kwiatkowski JL, Walters MC, Hongeng S, Yannaki E, Kulozik AE, Kunz JB, et al. Betibeglogene autotemcel gene therapy in patients with transfusion-dependent, severe genotype β-thalassaemia (HGB-212): a non-randomised, multicentre, single-arm, open-label, single-dose, phase 3 trial. Lancet. 2024;404(10468):2175-86.
20. Locatelli F, Thompson AA, Kwiatkowski JL, Porter JB, Thrasher AJ, Hongeng S, et al. Betibeglogene Autotemcel Gene Therapy for Non-β(0)/β(0) Genotype β-Thalassemia. N Engl J Med. 2022;386(5):415-27.
21. Zafari M, Rad MTS, Mohseni F, Nikbakht N. β-Thalassemia Major and Coronavirus-19, Mortality and Morbidity: a Systematic Review Study. Hemoglobin. 2021;45(1):1-4.
22. Muthiadin C. Premarital screening and probabilities of genetic disease in premarital screening and probability of genetic diseases in couples preparing for marriage. Trop Genet. 2023 May 31;3(1):20–5.
23. Khaliq S. Thalassemia in Pakistan. Hemoglobin. 2022 Jan 2;46(1):12–4.
24. Zhen X, Ming J, Zhang R, Zhang S, Xie J, Liu B, et al. Economic burden of adult patients with β-thalassaemia major in mainland China. Orphanet J Rare Dis. 2023 Aug 29;18(1):252.
25. Tarım HŞ, Öz F. Thalassemia Major and Associated Psychosocial Problems: A Narrative Review. Iran J Public Health. 2022 Jan;51(1):12–8.

