Center for Management Science Research
ISSN: 3006-5291 3006-5283 Volume 3, Issue 6, 2025

DEMOGRAPHICS, CLINICAL PROFILES, AND HEALTHCARE UTILIZATION OF PATIENTS WITH BETA THALASSEMIA MAJOR: A MULTI-CENTER STUDY

Maria Ishrat Siddiqui[*1], Dr. Muhammad Ashaab Siddiqui[2], Dr. Muhammad Ali Siddiqui[3]

Department of Public Administration University of Karachi
MBBS, MRCGP (UK), Consultant Family Medicine St Richard's Road Surgery Kent England
Internal Medicine Resident Valley Bapist Medical Center Harlingen Tx

DOI: https://doi.org/10.5281/zenodo.17383688

Keywords
Beta Thalassemia; Demographics; Age; Weight; Burden

Article History
Received: 28 August 2025
Accepted: 06 October 2025
Published: 18 October 2025

Corresponding Author: *
Maria Ishrat Siddiqui

Abstract

Thalassemia is an inherited autosomal recessive blood disorder that occurs due to abnormal form of hemoglobin in the blood. It is an autosomal recessive condition caused by decreased formation of alpha or beta chains of hemoglobin. Pakistan continues to suffer from a high thalassemia burden with statistics rising every year. With around 10 million carriers across the country, it is reported that each year, around 5000 children are diagnosed with thalassemia. The aim of this study is to bridge the gap between the problems faced by thalassemia patients in Pakistan and the facilities not available to them as compared to the developed countries.

Method: *A cross- sectional study was conducted with the duration of this study was 7 months. The data was collected from six tertiary care hospitals and eight Thalassemia centers, with the selection based on the accessibility of data.*

The inclusion criteria for the study was: a clinical diagnosis of Beta Thalassemia Major and age group between 1-18 years, irrespective of gender. The exclusion criteria included patients who were over 18 years of age and had comorbidities or complications other than Beta Thalassemia Major, as well as those who did not give consent.

A questionnaire was circulated to record the data. It had two parts. The first part included demographics and the second part had close-ended questions related to the treatment of Thalassemia.

Results: *There were 255 participants whose percentage of males and females was 47.8 and 52.2 respectively. It is established that age was positively correlated with visits to hospitals that patients with Beta Thalassemia made that as age increases, patients with the condition had more visits to hospitals ($r = 0.25$, $p < 0.05$, 95% CI [0.12, 0.38]). Another positive correlation between the weight and frequency of blood transfusion in B-thalassemia major patients ($r = 0.22$, $p < 0.05$, 95% CI [0.15, 0.35]) as well as between the weight and liver function tests that were performed per year in thalassemia patients ($r = 0.28$, $p < 0.05$, 95% CI [0.15, 0.41]) were also found.*

Conclusion: *These findings indicate how parties to BTM patients, whether regarding age and weight, have increased into a healthcare issue that needs greater initiatives by the government, preventive measures, and equal access to care in Pakistan as compared to the developed nations.*

data from six tertiary care hospitals and eight Thalassemia centers. A questionnaire was used to record data on demographics and close-ended questions related to thalassemia treatment. Positive correlations were observed between age and hospital visits, liver function tests, viral profiles, and CBCs conducted. Weight was positively correlated with the frequency of blood transfusions and liver function tests conducted per year.

INTRODUCTION

Thalassemia is a hereditary group of disorders associated with defective synthesis of alpha and beta subunits of hemoglobin [1]. Beta-thalassemia arises from a multitude of over 200 distinct point mutations and, infrequently, from deletion events which occurs on chromosome 11[1, 2]. Most common gene mutation in Pakistan is found to be IVS 1-5 (G-C) along with Fr 8-9, IVS 1-1 (GT), Cd -30 (G-C) being the other common mutations [3]. Beta-thalassemia (β- thalassemia) is characterized by absent or reduced synthesis of the hemoglobin subunit beta chain, that results in microcytic hypochromic anemia and has two clinically significant forms i.e. Beta thalassemia major and Beta thalassemia intermedia. Individuals with Beta thalassemia major present between 6 to 24 months of life and require regular blood transfusions whereas those with Beta Thalassemia Intermedia present later in life and do not require frequent blood transfusions [4].

Globally, 40,000 infants are anticipated to be born with Beta thalassemia major annually according to a 2008 report of World Health Organization, while around 1.5% of the population exhibits carrier status [5,6]. In Pakistan, the carrier prevalence varies from 5% to 8%, resulting in an estimated total of 9.8 million carriers within the population. Additionally, approximately 5,000 children are annually diagnosed with beta-thalassemia major in Pakistan [7].

Considering management, thalassemia is broadly classified into Transfusion-dependent (TD) and Transfusion independent (TI) [8]. According to fourth edition guidelines of Thalassemia International Federation, management of Transfusion Dependent Thalassemia requires blood transfusions on Hb <7 dg/L on 2 different occasions at least two weeks apart without any contributory cause or Hb level more than 7 dg/L with significant symptoms of anemia, failure to thrive and clinically significant extra medullary hematopoiesis [9].

In TDT, the complications of iron overload and Transfusion Transmitted Infections (TTIs) are unavoidable to some degree because of the numerous blood transfusion needed and causing the development of iron overload complications. The present multi-centered research was targeting pediatric patients to reduce the effects of adult comorbidity and to capture early patterns of utilization.

1. Material and methods

The purposive sampling was by selecting sites that were accessible and represented a variety of public/ private facilities in Karachi to guarantee a variety of patient profile and minimize selection bias. A structured questionnaire was used to collect the data and included the elements of demographics and the set of close-ended questions on the diagnosis, management and investigations during a period of 7 months, which was pilot-tested on 20 patients to ensure the face-validity (Cronbachs alpha = 0.82) and was given to the respective guardians in face to face interviews to eliminate recall bias. Existing medical records were cross-verified when responding. The population of the study covered Beta- thalassemia major (BTM) patients. The informed consent was taken in writing, as per the ethics of the study, by way of permission of the participants and their guardians.

The inclusion criteria included all Beta thalassemia major patients between 1 and 18 years, regardless of gender, to concentrate on the patterns of management of the disorder in pediatric settings and exclude adults to avoid the heterogeneity of the long-term problems. Beta-thalassemia major patients who had attained an age above 18, those who refused to give informed consent, those with known co-morbidities or those with other pathologies irrelevant to the research were ruled out to ensure the sample homogeneity. Note: BTM-related complications (e.g.,

iron overload) were considered in case they were not comorbid with other illnesses.

Ethical approval was obtained from the Institutional Review Board of Children Hospital Karachi IRB Letter No. CH-0427 Dated: 12/06/2024 Declaration of Helsinki.

After data were collected, statistical analysis of the variables was done using SPSS version 26. Frequencies and percentages of the nominal variables were the content of the descriptive analysis. Pearson correlation analysis of continuous variables was used to analyze the relationship between the various variables in an inferential analysis. Thepower of 80 to be able to detect a moderate correlation (r=0.3) at 0.05 means that a sample size of 255 was adequate. Various comparisons were corrected by Bonferonni. All the analyses were conducted on SPSS version 26.

2. Results

The total number of participants were 255: 133 (52.2%) females and 122 (47.8%) males. All participants were diagnosed as Beta thalassemia-major patients (100%). The average age was 11 years ± 5.2 years. The mean weight was 26.44 kg ± 12.6 kg, and the mean interval days between transfusion frequency was 17 days ± 7 days. The mean hospital visits per month were 2 days ± 1 day.

Variable	Mean ± SD	n (%)
Age (years)	11 ± 5.2	255
Weight (kg)	26.44 ± 12.6	255
Interval between transfusions (days)	17 ± 7	255
Hospital visits per month	2 ± 1	255
Sex (Female)	-	133 (52.2%)
Sex (Male)	-	122 (47.8%)
Diagnosis (BTM)	-	255 (100%)

In individuals with beta-thalassemia major, a positive correlation was found between age and number of hospital visits (p=0.05), age and LFT(p=0.05), age and CBC(p=0.05), age and viral profile(p=0.05), which suggests that as age increases, there is an increase in the number of hospital visits, LFTs and need for annual viral profiling.

There is a statistically significant positive correlation (p=0.05) between the weight of beta-thalassemia major patients and both the frequency of blood transfusions and the frequency of Liver Function Tests (LFTs) conducted per year.

An indeterminate correlation was observed between CBC tests conducted per month and a decrease in transfusion frequency among B-thalassemia major patients.

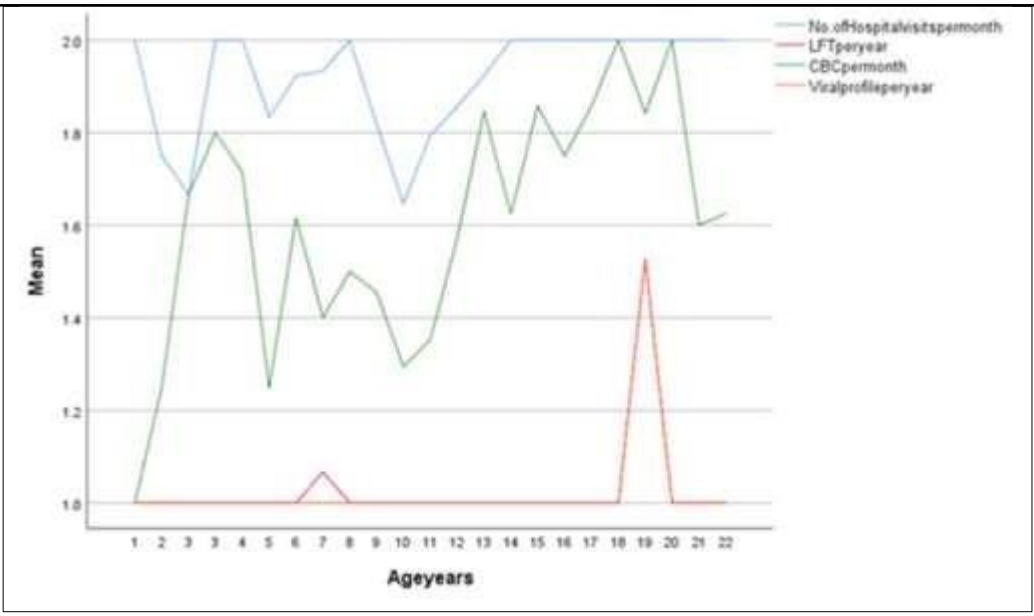

Figure 1 Scatter graphs with positive correlations of age with (A) mean number of hospital visits per month, (B) LFTs done per year, (C) viral profiles done per year and (D) weight with transfusion frequency.

The above graph shows a positive correlation between age and number of hospital visits, LFTS per year, CBC per month and Viral profiles per year.

3. Discussion

This is a severe health problem because the beta-thalassemia major is a monogenic blood disorder that is a major health challenge in the country. Unfortunately, there are very few studies on the area of holistic assessment in Pakistan. Our study fills this gap by measuring the relationships of the usage of healthcare and demonstrating how age and weight become the origins of increased monitoring needs. We conducted a study to comprehensively analyze the various aspects of beta-thalassemia major and its treatment applying correlations between different variables in these patients, including age, weight, interval days between transfusions, hospital visits, liver function tests, CBC tests, and viral profiles and the significance of these factors in contributing to the burden of these patients.

The management of Beta thalassemia includes blood transfusion and iron chelation, which when regularly administered, can significantly improve patients' lives. However, iron buildup in organs may require splenectomy to avoid iron overload complications; nevertheless, splenectomy has its own fatal risks of sepsis, hypercoagulability, and others.

Thalassemia patients also undergo annual viral profile tests due to the risk of contracting HBV and HCV from regular blood transfusions and as the age increases exposing patients to chronic transfusions, the risk of acquiring hepatitis increases. A study showed HCV prevalence of 29.8% and HBV of 4.13% in affected patients in Pakistan. [10]

It is concerning that a study in Islamabad showed that 76% of the families spend more than 80,000pkr monthly for which majority had to sell their livelihood, compromise on education or other children's health if they don't have any service entitlement. [11] Thus, many of these families suffer through financial burden and cannot afford their basic needs and treatment.

Beyond financial strain, there is a clear need for awareness regarding transmission and treatment of Thalassemia. This need for awareness is emphasized by a study conducted in Karachi indicating that even the parents of thalassemia children had inadequate knowledge regarding thalassemia, its prevention and treatment. [12] The propagation of Thalassemia is alarming due to strict inter-marriage rules in some

castes. A study regarding psychosocial burden was conducted in Pakistan which suggests that consanguineous marriages, level of education of parents, belief in superstition causing thalassemia, religious restriction regarding prevention and termination of pregnancy, social stigmatization are the major predictors of causing psychosocial burden [13].

These complications further contribute to a social burden, as patients may require additional time away from work and receive less support from co-workers. Enacted stigma referring to discriminatory behaviors and felt stigma referring to personal perception of discrimination affect the personal and social life of these patients leading to social isolation. [14]

To improve the lives of these patients, a holistic approach is vital, addressing all aspects of their well-being extensively. While organizations working for the provision of treatment at low or no cost should be supported, public campaigns should be run to spread awareness about pre-marital screening, prenatal screening, proper treatment, and the disadvantages of consanguineous marriages.

4. Conclusion

In Pakistan, beta thalassemia major has increasingly clinical and economic burden, as age/weight and healthcare utilization (e.g., transfusion, tests) have a positive correlation ($r > 0.22$, $p < 0.05$) to show an increasing healthcare demand. Multi-center research (n=255) requires interventions at the government level, such as increased screening, low-cost therapies, and educational efforts to prevent and help families to close gaps, which are noticeable in terms of international comparisons.

Compliance with ethical standards

Disclosure of conflict of interest

Authors have declared that no competing interest exist.

References

Eliezer A. Rachmilewitz, Patricia J. Giardina; How I treat thalassemia. Blood 2011; 118 (13): 3479–3488. doi: https://doi.org/10.1182/blood-2010-08-300335

Rund D, Rachmilewitz E. Beta-thalassemia. N Engl J Med. 2005 Sep 15;353(11):1135-46. doi: 10.1056/NEJMra050436. PMID: 16162884.

Ansari SH, Shamsi TS, Ashraf M, Bohray M, Farzana T, Khan MT, Perveen K, Erum S, Ansari I, Nadeem M, Ahmed M, Raza F. Molecular epidemiology of β-thalassemia in Pakistan: far reaching implications. Int J Mol Epidemiol Genet. 2011;2(4):403-8. Epub 2011 Nov 28. PMID: 22200002; PMCID: PMC3243455.

Langer AL. Beta-Thalassemia. 2000 Sep 28 [Updated 2023 Jul 20]. In: Adam MP, Mirzaa GM, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2023.

Modell B, Darlison M. Global epidemiology of haemoglobin disorders and derived service indicators. Bull World Health Organ. 2008 Jun;86(6):480-7. doi: 10.2471/blt.06.036673. PMID: 18568278; PMCID: PMC2647473.

Kattamis, A., Forni, G.L., Aydinok, Y., Viprakasit, V., 2020. Changing patterns in the epidemiology of β-thalassemia.

European Journal of Haematology 105, 692–703.. https://doi.org/10.1111/ejh.13512

Asif, Naghmi, and Khalid Hassan. "Prevention of beta thalassemia in Pakistan." J Islam Med Dent Coll 3.2 (2014): 46-7.

Yardumian A, Telfer P, Constantinou G, Darbyshire P, Darlison M, Kinsey S, Miller E, Modell B, Pollitzer M, Porter

J. Standards for the Clinical Care of Children and Adults with Thalassaemia in the UK. United Kingdom Thalassaemia Society; 2005 Jun 14.

Farmakis D, Porter J, Taher A, Domenica Cappellini M, Angastiniotis M, Eleftheriou A. 2021 Thalassaemia International Federation Guidelines for the Management of Transfusion-dependent Thalassemia. Hemasphere. 2022 Jul 29;6(8):e732. doi: 10.1097/HS9.0000000000000732. PMID: 35928543; PMCID: PMC9345633.

Waheed, U., Saba, N., Wazeer, A., and Ahmed, S. (2021). A Systematic Review and Meta-Analysis on the Epidemiology of Hepatitis B and Hepatitis C Virus among Beta-Thalassemia Major Patients in Pakistan. Journal of laboratory physicians, 13(3), 270–276. https://doi.org/10.1055/s-0041-1731110

Qamar, M. K., and Shaikh, B. T. (2022). Out Of Pocket Expenditure On Thalassemia Major And Its Implications On The Household Economics. Journal of Ayub Medical College, Abbottabad : JAMC, 34(1), 91–94. https://doi.org/10.55519/JAMC-01-8332

Arif, F., Fayyaz, J., and Hamid, A. (2008). Awareness among parents of children with thalassemia major. JPMA. The Journal of the Pakistan Medical Association, 58(11), 621–624.

Ul Hassan Rashid, M. A., Abbasi, S.-U.-R. S., and Manzoor, M. M.. (2020). Socio-religious Prognosticators of Psychosocial Burden of Beta Thalassemia Major. Journal of Religion and Health, 59(6), 2866–2881. https://doi.org/10.1007/s10943-020-01069-6

Kumar, N., Turbitt, E., Biesecker, B. B., Miller, I. M., Cham, B., Smith, K. C., and Rimal, R. N. (2019). Managing the need to tell: Triggers and strategic disclosure of thalassemia major in Singapore. American journal of medical genetics. Part A, 179(5), 762–769. https://doi.org/10.1002/ajmg.a.61107.