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(RESEARCH ARTICLE)

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Demographics, clinical profiles and healthcare utilization of patients with beta thalassemia major: A single centered study

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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 thalassemic 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 12 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: The total number of participants were 255 in which 122 (47.8%) were males and 133 (52.2%) were females. A positive correlation was observed between age and hospital visits, showing that an increase in age was associated with an increase in hospital visits among Beta Thalassemia patients (p value< 0.05) (CL:95%). Positive correlation was observed between the age of patients and the number of Liver Function tests. (p-value<0.05) (CL: 95%), viral profile done per year and age among B-thalassemia major patients. (p-value< 0.05) (CL:95%) and between age and cbc conducted per month (p-value < 0.05). Another positive correlation was observed between weight and frequency of blood transfusions in B-thalassemia major patients (p-value< 0.05) (CL:95%) and weight and liver function tests conducted per year among thalassemia patients. (p-value< 0.05) (CL:95%).

Conclusion: This study aims to highlight the problems faced by thalassemia patients in Pakistan and the facilities not available to them compared to developed countries. A cross-sectional study was conducted for 12 months, collecting

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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.

Keywords: Beta Thalassemia; Demographics; Age; Weight; Burden

1. 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 extramedullary hematopoiesis[9].

In TDT, iron overload complications and Transfusion Transmitted Infections (TTIs) are inevitable to some extent due to the frequent blood transfusions required and leading to iron overload complications. To address this gap, we conducted a cross-sectional study in Karachi to observe the factors contributing to the burden of transfusion-dependent beta-thalassemia major patients.

2. Material and methods

We conducted a descriptive cross-sectional study including eight thalassemia centers and six tertiary care hospitals. The data was collected through a structured questionnaire comprising of demographics and close-ended questions about the diagnosis, management, and investigations over a period of 7 months. The study population included Beta-thalassemia major (BTM) patients. In compliance with ethical standards, informed consent was obtained through written permission from the participants and their guardians.

Inclusion criteria consisted of all Beta thalassemia major patients aged between 1 and 18 years, irrespective of gender. Patients with beta-thalassemia major who surpassed the age of 18, those who declined to provide informed consent, individuals with disclosed co-morbidities, or those afflicted by pathologies other than beta-thalassemia major, were excluded from participation to maintain homogeneity of the sample and reduce information bias.

Statistical analysis of the variables was carried out after data collection through SPSS version 26. The descriptive analysis included frequencies and percentages of the categorical variables. Inferential analysis was done to assess the association between different variables.

3. 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.

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.

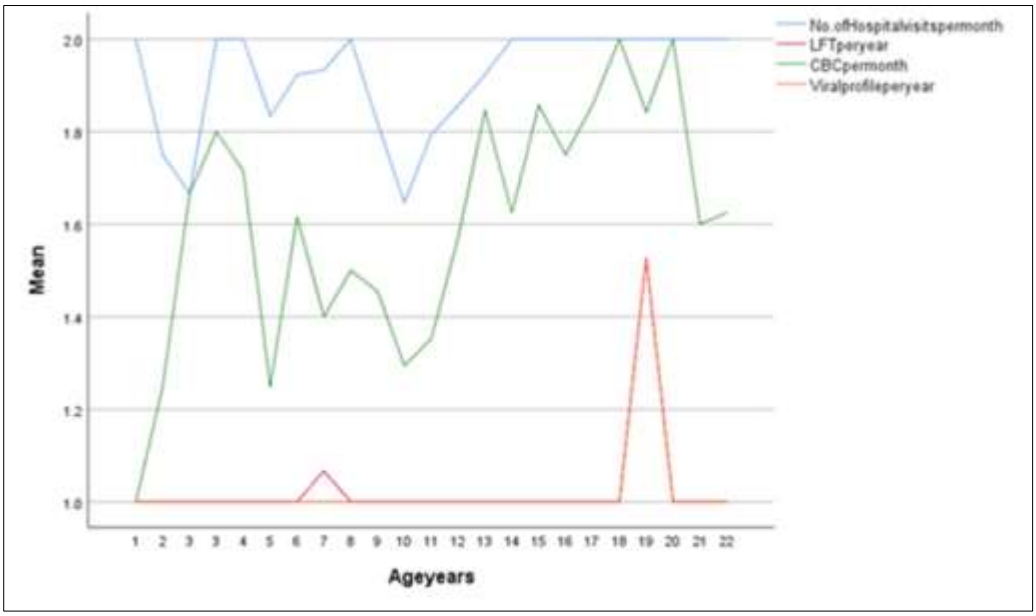

Figure 1 Graph showing correlation between Age and mean No. of hospital visits per month, Weight ,Lfts done per year, Viral profiles done per year

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.

4. Discussion

Beta-thalassemia major is a monogenic blood disorder and poses a significant health challenge nationally. Unfortunately, there are limited studies on holistic assessment of Beta thalassemia major patients in Pakistan. 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.

5. Conclusion

Beta thalassemia major is a hemoglobinopathy characterized by defective synthesis of beta globin chain, this study aims to highlight the point regarding increasing complexity of patients with beta thalassemia major as they age considering different parameters simultaneously like weight ,CBC, viral profiles and Number of hospital visits done per year, which shows positive correlation with increasing age and this signifies the increased demand and need of proper established centers and involvement of government of Pakistan by making major steps towards their wellbeing by provision of basic needs and support and also prevention of disease by various methods.

Compliance with ethical standards

Disclosure of conflict of interest

Authors have declared that no competing interest exist.

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