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ORIGINAL ARTICLE

Quality of life in transfusion dependent thalassemia patients at national institute of child health.

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ABSTRACT… Objective: To determine the quality of life of children suffering from transfusion dependent thalassemia. **Study Design:** Case-control study. **Setting:** Department of Pediatrics, National Institute of Child Health, Karachi, Pakistan. **Period:** March 2024 to August 2024. **Methods:** Children of either gender, aged between 6 months to 14 years, and having transfusion dependent thalassemia were included as cases (n=95). For controls (n=95), healthy volunteers without chronic disease, matched in terms of gender and age were included. At the time of enrollment, demographical data like age, gender, height, and weight were noted. Relevant laboratory investigations were performed and evaluated. Children of both groups along with their parents were interviewed about quality of life administering WHOQOL-BREF questionnaire. **Results:** In a total of 190 children (95 children in each group), 104 (54.7%) were male and 86 (45.3%) female. Physical health scores were 35.62±6.68 in children with transfusion dependent thalassemia vs. 74.92±7.92 in controls (p<0.001). Psychological health scores were 38.16±8.12 vs. 76.68±6.66 (p<0.001) among transfusion dependent thalassemia children versus controls, respectively. Social relationships were 41.19±6.14 vs. 77.50±7.01 (p<0.001) among transfusion dependent thalassemia children versus controls, respectively. Environmental scores were 43.51±6.93 vs. 71.59±6.06 (p<0.001), among transfusion dependent thalassemia children versus controls, respectively. **Conclusion:** Our study highlights the significant negative impact of transfusion-dependent thalassemia on the quality of life of affected children across all domains of the WHOQOL-BREF questionnaire.

Key words: Physical Health, Pshychological Health, Social Relationship, Thalassemia, WHOQOL-BREF.

INTRODUCTION

Thalassemia encompasses a diverse group of blood disorders caused by mutations in hemoglobin genes, leading to ineffective erythropoiesis.[1] It is a genetic condition characterized by mutations in the genes involved in hemoglobin synthesis, resulting in reduced production of alpha or beta globin chains.[2,3] β-thalassemia refers to a hereditary blood disorder marked by either a reduction or complete absence of β-globin chain production, leading to decreased hemoglobin levels, impaired red blood cell production, and anemia.[4] Clinically, β-thalassemia is divided into two categories as “”transfusion-dependent thalassemia (TDT), and “non-transfusion-dependent thalassemia (NTDT)”.[5] TDβT represents the most severe form, characterized by significant anemia and requiring lifelong, regular blood transfusions to maintain adequate hemoglobin levels.[6] Globally, β-thalassemia impacts a substantial proportion of the population, with an estimated 270 million individuals being carriers of the condition, including 70 million specifically carrying β-thalassemia.[7] The global prevalence is estimated to include 80–90 million carriers, accounting for roughly 1.5% of the population.[8] In Pakistan, the prevalence of β-thalassemia is around 6%, with a gene carrier rate of 5–7%. More than 50,000 patients in the country are currently receiving treatment at specialized thalassemia centers.[9,10]

Thalassemia-related complications are widely recognized for their negative impact on quality of life. According to the “World Health Organization

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(WHO)", quality of life refers to an individual's perception of their position in life, influenced by cultural and value systems, as well as their personal goals, expectations, and concerns.[11] Among children with TDT, these complications often result in significant psychological challenges, emotional stress, feelings of hopelessness, and difficulties with social integration, ultimately diminishing their quality of life.[12-15] A study by Alzahrani RA and colleagues evaluated the quality of life in transfusion-dependent thalassemia patients using the WHOQOL-BREF tool. The findings revealed that the mean quality of life score for these patients was 82.4 ± 5.54, significantly lower compared to healthy control patients, who had a mean score of 87.86 ± 12.9, indicating a better quality of life.[12] Another similar study by Mettananda S, et al. was also reported the lower mean score 72.9 in TDT group and higher mean score 91.5 in healthy patients that shows low quality of life in TDT patients.[13] Despite development in diagnosis and management of TDT in children, quality of life is one of the most neglected aspect throughout the world resulting in decreasing the standard of living. Very little research has been done on quality of life of children suffering from TDT and need much more research. Therefore, this study was conducted in order to determine the quality of life of children suffering from TDT.

METHODS

This case-control study was conducted at the department of pediatrics, National Institute of Child Health, Karachi, Pakistan from March 2024 to August 2024. For sample size calculation, online Open Epi sample size software was used taking stats from Alzahrani RA, et al., who reported the mean QOL score of 82.4±5.54 in TDT versus 87.86 ± 12.9 in healthy group patients[12], by taking confidential interval 95%, power 80% and ratio of sample size 1. The sample size was calculated as 190 (95 in each group). Non-probabality, purposive sampling technique was used. The inclusion criteria were children of either gender, aged between 6 months to 14 years, and having TDT (case group). For controls, healthy volunteers without chronic disease, matched in terms of gender and age were included. The exclusion criteria were presence of any co-morbid conditions unrelated to β-thalassemia or its complications. Children whose parents/guardians were not willing to participate in this study were excluded. A child who underwent regular blood transfusions and received at least one blood transfusions per 4-6 weeks were labeled as having TDT. Approval from "Institutional Ethical Committee" was acquired (letter number: IERB-40/2021, dated: 02-02-2022). Informed and written consents were acquired from parents/guardians.

At the time of enrollment, demographical data like age, gender, height, and weight were noted. Relevant laboratory investigations were performed and evaluated, including complete blood count, serum ferritin and iron levels. Children of both groups along with their parents were interviewed about quality of life administering WHOQOL-BREF questionnaire. It consists of 26 questions, which are categorized into four main domains as Physical Health (7 items), Psychological Health (6 items), Social Relationships (3 items), and Environment (8 items). Raw scores are obtained by summing the scores for each item in a domain, and then raw scores are transformed to a scale of 0 to 100, where a higher score reflects a better quality of life. Data were recorded on a pre-designed proforma.

After collection of data, the analyses was conducted using IBM-SPSS Statistics, version 26.0. The mean and standard deviation were calculated for quantitative variables. Frequency and percentages were calculated for qualitative variables. Independent sample t test and chi-square test were applied for comparing data between groups. Bivariate correlation analysis was performed applying Pearson's correlation. For all inferential statistics, $p<0.05$ was considered as significant.

RESULTS

In a total of 190 children, 104 (54.7%) were male and 86 (45.3%) female, representing a male to female ratio of 1.2:1. The mean age was comparable between TDT children and controls (8.24±2.17 vs. 8.30±2.14 years, p=0.840). TDT children had significantly lower height (110.90±12.16 cm

vs. 115.37±13.0 cm, p=0.015), and statistically similar weight (23.35±5.69 kg vs. 24.78±5.97 kg, p=0.092). Hemoglobin levels were significantly lower among TDT children (9.36±1.12 g/dl vs. 12.50±1.02 g/dl, p<0.001). TDT children had lower platelet counts (252.41±48.58 vs. 276.57±41.84, p=0.001) but higher white blood cell counts (7.72±1.18 vs. 6.61±1.59, p<0.001). Serum ferritin (1466.27±529.41 ng/mL vs. 101.49±20.55 ng/mL, p<0.001 and iron 182.55±28.73 μg/dl vs. 119.49±28.56 μg/dl, p<0.001) levels were elevated in TDT children (Table-I).

Physical health scores were 35.62±6.68 in children with transfusion dependent thalassemia vs. 74.92±7.92 in controls (p<0.001). Psychological health scores were 38.16±8.12 vs. 76.68±6.66 (p<0.001) among TDT children versus controls, respectively. Social relationships were 41.19±6.14 vs. 77.50±7.01 (p<0.001) among TDT children versus controls, respectively. Environmental scores were 43.51±6.93 vs. 71.59±6.06 (p<0.001), among TDT children versus controls, respectively. Details about the quality of life, measured using the WHOQOL-BREF questionnaire across all domains among children of both study groups are shown in Table-II.

The correlation matrix showed significant positive correlations between all domains, with physical health correlating most strongly with social relationships (r=0.785, p<0.01). Psychological and social relationship scores were also highly correlated (r=0.750, p<0.01), as shown in Table-III.

Characteristics		Transfusion Dependent Thalassemia (n=95)	Controls (n=95)	P-Value
Gender	Male	52 (54.7%)	52 (54.7%)	1
	Female	43 (45.3%)	43 (45.3%)	
Age in years, Mean±SD		8.24±2.17	8.30±2.14	0.840
Height in cm, Mean±SD		110.90±12.16	115.37±13.0	0.015
Weight in Kg, Mean±SD		23.35±5.69	24.78±5.97	0.092
Hemoglobin in g/dl, Mean±SD		9.36±1.12	12.50±1.02	<0.001
Platelets (10^3/ul)		252.41±48.58	276.57±41.84	0.001
White blood cells count (10^3/ul)		7.72±1.18	6.61±1.59	<0.001
Serum ferritin in ng/mL, Mean±SD		1466.27±529.41	101.49±20.55	<0.001
Iron (ug/dl), Mean±SD		182.55±28.73	119.49±28.56	<0.001

Table-I. Comparison of demographical and laboratory characteristics of children (N=190)

Domains	Transfusion Dependent Thalassemia (n=95)	Controls (n=95)	P-Value
Physical health , Mean±SD	35.62±6.68	74.92±7.92	<0.001
Psychological health, Mean±SD	38.16±8.12	76.68±6.66	<0.001
Social relationship, Mean±SD	41.19±6.14	77.50±7.01	<0.001
Environment, Mean±SD	43.51±6.93	71.59±6.06	<0.001

Table-II. Comparison of quality of life by using WHOQOL-BREF questionnaire of children (N=190)

Domains	Physical Health Score	Psychological Health Score	Social Relationships Score	Environmental Score
Physical Health	1	0.751**	0.785**	0.687**
Psychological Health	0.751**	1	0.750**	0.702**
Social Relationships	0.785**	0.750**	1	0.749**
Environment	0.687**	0.702**	0.749**	1

**Correlation is significant at the 0.01 level (2-tailed).

Table-III. Correlation Matrix analyzing domains across WHOQOL-BREF questionnaire

DISCUSSION

Our findings demonstrate a significantly impaired quality of life in children suffering from TDT compared to healthy controls across all domains measured by the WHOQOL-BREF questionnaire. The mean physical health score of children with TDT was significantly lower than that of healthy controls (35.62 ± 6.68 vs. 74.92 ± 7.92, $p < 0.001$). This difference is substantial and reflects the considerable physical burden that TDT places on children. These results align closely with findings by Roghani et al., who reported a similar negative effect of TDT on physical health, with a mean score of 36.05 ± 13.80 in their cohort.[16] Alzahrani et al., also reported similar findings but they utilized the WHOQOL-BREF tool.[17] Regular blood transfusions, iron overload, and the complications associated with these treatments contribute to poor physical health.[18] The impaired physical health in TDT patients is likely attributable to chronic fatigue, frequent hospitalizations, and the burden of managing iron chelation therapy.

The psychological health of children with TDT in our study was significantly lower than that of healthy controls ($p<0.001$). Children with TDT often face psychological stress due to the chronic nature of their illness, the need for regular transfusions, and concerns about their future. Our findings are in line with those reported by Shafie et al., who demonstrated that psychosocial health was significantly affected in children with TDT.[19] Roghani et al., reported a low mean emotional impact score in TDT patients, indicating the widespread emotional burden experienced by these children.[16] Maheri et al., explored the predictors of psychological health and found that anxiety, depression, and perceived barriers were significant negative predictors of quality of life.[20] These findings align with our study's results, as children with TDT often face anxiety related to their illness, the frequent need for medical procedures, and their limited physical abilities compared to their peers. The significantly lower psychological health scores in our study reflect these challenges.

In this study, social relationships were notably impacted in children with TDT ($p<0.001$). This finding is consistent with those of Alzahrani et al., who reported that social functioning was similarly impacted in TDT patients, particularly in males.[17] Roghani et al., also reported that family life and social interactions were significantly affected in their cohort.[16] Our study's lower social health scores may be reflective of cultural and social dynamics in Pakistan, where children with chronic illnesses may face social stigma or isolation. The frequent need for medical care and absence from school likely compounds the challenges faced by these children in maintaining social relationships.[21] The significant impact on social health is also echoed in the study by Shafie et al., who found that school functioning had the lowest score among all domains of psychosocial health.[19] This is consistent with our findings, as many children with TDT in Pakistan struggle with school attendance due to the time-consuming nature of their treatment. Jajhara et al., noted that compliance with blood transfusion and iron chelation therapy improved social and school functioning, a finding that underscores the importance of regular treatment in mitigating some of the social consequences of the disease.[22]

This study showed that children with TDT had lower environmental health scores compared to controls ($p<0.001$). Children with TDT often require frequent medical visits, which can impose a financial burden on families. Roghani et al., reported similar findings, with environmental factors being negatively affected in children with TDT, as the disease often disrupts family routines and increases healthcare expenses. In our study, the environmental domain was significantly correlated with other domains, particularly social health ($r=0.749$, $p<0.01$), indicating that environmental stressors may exacerbate social isolation and poor relationships. Drahos et al., also highlighted the environmental challenges faced by individuals with TDT.[23] Their study on adults with TDT further emphasized the role of healthcare interactions in shaping patients' experiences, with inadequate support contributing to poorer outcomes in environmental health. Our findings suggest that improving healthcare support systems and reducing the financial burden on families could potentially enhance the

environmental quality of life for TDT patients.

This study also highlighted significant differences between cases and controls in terms of hemoglobin levels, platelet counts, WBC counts, serum ferritin, and iron levels. Shafie et al., who noted that hemoglobin levels were a predictor of better quality of life.[19] Higher hemoglobin levels are associated with improved physical and psychosocial health, as children experience less fatigue and better overall functioning. Roghani et al.[16], similarly found that higher ferritin levels were negatively correlated with QOL, and Maheri et al.[20], emphasized the need for effective chelation therapy to mitigate this issue. Our study supports these findings, suggesting that iron overload contributes to poorer physical and psychological health outcomes. These abnormal counts may also reflect underlying splenic dysfunction, chronic inflammation, or bone marrow suppression, all of which are common in children with TDT.[24]

The limitations of this study include the use of a single-center sample, which may limit the generalizability of the findings. The cross-sectional design did not allow for assessing changes in quality of life over time or establishing causality. The reliance on self-reported and parent-reported data could introduce bias, as perceptions of quality of life may vary. Socio-economic factors were not thoroughly assessed, which could have affected quality of life outcomes.

CONCLUSION

Our study highlights the significant negative impact of transfusion-dependent thalassemia on the quality of life of affected children across all domains of the WHOQOL-BREF questionnaire. Our study reinforces the need for comprehensive management strategies that address not only the physical health of TDT patients but also their psycho-social health related quality of life.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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1	Sanam Hussain	Data collection, Drafting, Responsible for data's integrity, approved for publication.	
2	Muhammad Ashfaq	Study concept, Methodology, Proof reading, critical revisions, approved for publication.	
3	Bader u Nisa	Study concept, Methodology, Proof reading, critical revisions, approved for publication.	
4	Hayat Bozdar	Study concept, Methodology, Proof reading, critical revisions, approved for publication.	
5	Aijaz Talani	Data synthesis, data analysis, Proof reading, critical revisions, approved for publications.