Health and Quality of Life Outcomes

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Health-related quality of life and associated factors among children with Transfusion-dependent β-thalassaemia: a cross-sectional study in Guangxi Province

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Abstract

Background Transfusion-dependent β-thalassemia (TDT) is a severe inherited disorder. Without regular treatment, patients with TDT experience complications that can significantly shorten life expectancy and severely impact both their quality of life and that of their families. The condition has attracted significant attention in global health discussions. Due to the challenges of blood supply shortages, the high costs of iron chelation therapy, and hematopoietic stem cell transplantation (HSCT), TDT presents a serious health risk to patients and imposes a substantial burden on families and society. However, research on the health-related quality of life (HRQoL) of thalassemia patients in China remains limited. This study evaluated the factors affecting the HRQoL of these patients, with the goal of developing strategies to improve their quality of life.

Methods In this cross-sectional study, children with TDT were recruited from five treatment centers in Guangxi, a province with a high prevalence of thalassemia in China. Structured questionnaires were employed to gather relevant data on sociodemographic variables, disease characteristics, treatments, and associated costs. The HRQoL was assessed using the Transfusion-Dependent Quality of Life (TranQoL) questionnaire, with a proxy version for patients aged 0–11 years and a child version for those aged 12–18 years.

Results The study included 418 participants, yielding an overall TranQoL score of 60.6 ± 16.3 among thalassemia patients. Multiple linear regression analysis revealed a negative correlation ($P < 0.05$) between overall TranQoL scores and several factors: increasing patient age, the presence of multiple thalassemia patients within a family, and undergoing HSCT. Conversely, adherence to regular treatment was positively correlated with higher TranQoL scores ($P < 0.05$).

Conclusion The study demonstrates that HRQoL among Chinese patients with TDT is at a low level. Age, treatment adherence, family support, and socioeconomic status were identified as key determinants influencing HRQoL. It is

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essential to further enhance and optimize health insurance policies and medical services to support comprehensive treatment strategies for these patients.

Keywords Transfusion-dependent thalassaemia, Health-related quality of life, Patient-reported outcomes, TranQoL

Background

Thalassemia is a hereditary blood disorder caused by autosomal mutations that disrupt the synthesis of globin chains. These genetic defects lead to the insufficient production or complete absence of one or more types of globin chains, resulting in the premature destruction of red blood cells [1, 2]. The global movement of populations through immigration has spread thalassemia beyond its traditional epicenters in the Mediterranean, Africa, and Asia to regions including Europe, the Americas, and Australia, making it a global public health concern [3, 4]. It is estimated that approximately 1.5% of the global population are carriers of the β-thalassemia allele, while 5% carry the α-thalassemia allele [5, 6]. In China alone, an estimated 30 million individuals carry thalassemia genes. According to the International Thalassaemia Association's 2023 report, the prevalence of β-thalassemia gene carriers in China ranges from 1 to 8%, with higher rates observed in southern regions compared to northern areas. Guangxi Province has the highest prevalence [7], establishing thalassemia as a significant public health issue related to birth defects in southern China.

The management of thalassemia requires a tailored approach based on the patient's specific diagnosis and overall health status. The cornerstone of treatment typically includes regular blood transfusions and iron chelation therapy to manage excess iron accumulation. According to the Chinese Guideline for the Diagnosis and Treatment of Transfusion-Dependent β-Thalassemia (2022 Edition) [8], hematopoietic stem cell transplantation (HSCT) remains the only curative option for severe β-thalassemia. Research indicates that while the life expectancy of patients with transfusion-dependent β-thalassemia (TDT) has improved over recent decades, their average lifespan is still significantly lower than that of the general population. This disparity highlights the persistent severity of thalassemia as a critical threat to patients' lives and health. The prolonged treatment required for TDT adversely impacts patients' emotional, social, and academic functioning, leading to a marked decline in their health-related quality of life (HRQoL) and work productivity. Additionally, the disease progressively leads to complications such as skeletal deformities, growth retardation, and delayed puberty, further exacerbating the challenges faced by adolescent patients, particularly in terms of their growth, development, and HRQoL [9–12]. Numerous studies have shown that TDT patients have a poorer HRQoL compared to the general population [13–15]. Previous research has identified that factors such as patient age, the number of comorbidities, and the number of prescribed medications negatively impact HRQoL [16, 17]. In various surveys assessing children's HRQoL, TDT patients consistently report lower scores across physical, emotional, social, academic, and psychological domains [16, 18, 19]. The impact of different treatment modalities on HRQoL also varies significantly. For example, transfusion and iron chelation therapies have been shown to negatively influence HRQoL, even in the absence of overt disease-related complications [20]. Moreover, the shortage of blood resources in China, combined with the high cost of transplantation surgery, limits treatment options, imposes a significant burden on β-thalassemia patients and their families, and leads to a decline in patients' HRQoL.

Therefore, scientifically evaluating and managing the factors that influence the quality of life in patients with TDT is of significant practical and clinical importance. Currently, both in China and globally, research on β-thalassemia primarily focuses on clinical and epidemiological aspects, such as genotype screening and blood analyses in high-prevalence areas. However, studies addressing the socioeconomic impacts, including patient quality of life, are limited. In-depth investigations into the socioeconomic impact on TDT patients, particularly regarding their quality of life, are still lacking in systematic data support and thorough analysis, which, to some extent, limits our comprehensive understanding of TDT patients' HRQoL. The objective of this study is to achieve a more comprehensive understanding of HRQoL among children with TDT and to identify the key influencing factors. The insights gained from this research are intended to guide future research agendas and support the development of effective preventive and management strategies by relevant departments and organizations.

Methods

Study design and population

This hospital-based questionnaire survey was conducted at five centers in the cities of Nanning, Guilin, and Liuzhou, Guangxi Province, China. Children with TDT who were undergoing treatment were recruited for the study. The inclusion and exclusion criteria were collaboratively established by the researchers and clinicians. The main inclusion criteria were: (i) a diagnosis of transfusion-dependent β-thalassemia, (ii) patients receiving routine transfusion and iron chelation therapy, (iii) patients between the ages of 2 and 18 years, and (iv) patients or primary caregivers capable of understanding

and answering the questionnaire. Patients with TDT who exhibited cognitive impairment or were unable to respond to the questionnaire due to an acute illness were excluded from the study. A cohort of 422 patients and their parents consented to participate in the study after being fully informed. Data collection took place from August to October 2023. This study was approved by the Medical Research Ethics Committee, School of Public Health, Fudan University (approval no. 2022-07-0978).

Measures

The Transfusion-Dependent Quality of Life questionnaire (TranQoL) was used to measure the HRQoL of children with TDT. This validated, disease-specific instrument evaluates the HRQoL of patients with TDT [18] and comprises four domains: physical health, emotional health, family functioning, and school functioning. Scores range from 0 to 100, with higher scores indicating better HRQoL. Prior to employing this questionnaire, a user agreement was established with the Mapi Research Institute in Lyon, France.

Given that all participants were children with varying levels of disease awareness and comprehension of the questionnaire, the study utilized two versions of the instrument: a proxy version and a child self-report version. In the absence of an age restriction for either version, and based on relevant literature [21] and the Chinese context, the research team decided that preschoolers and primary school students (under 12 years of age) or those unable to complete the questionnaire independently would use the parental proxy version. Conversely, children in junior high school and above would use the self-administered version. Previous research has validated the internal consistency between these two versions of the scale [22]. The age-appropriate questionnaires were administered to patients and their parents during routine visits or blood transfusions by trained investigators.

The questionnaires also included questions about the patients' personal characteristics, disease conditions, utilization of health services, and healthcare costs. Key personal characteristics assessed included demographic details such as age, sex, weight, and socio-structural factors like ethnicity. Socio-economic aspects were explored through queries regarding the occupational status and marital status of the patient's parents, family income, the type of health insurance coverage, and whether the family had any indebtedness. Disease-related questions focused on the duration of the patient's treatment, complications experienced, and the patient's self-assessment of their health. Health service utilization was assessed through questions about hospital admissions, blood transfusions, and medication use. Healthcare costs were categorized into direct and indirect costs. Direct medical expenses included outpatient expenses, blood transfusion costs, and drug costs, while indirect medical expenses encompassed transportation costs and lost wages.

Data collection

The study was thoroughly explained to each participant and their caregivers by uniformly trained researchers before the questionnaire was administered. To ensure the quality of the survey, investigators accompanied patients throughout the questionnaire completion process, with data collection conducted on a one-to-one basis. The questionnaire was completed either by the patients themselves (ages 12–18 years) or by their parents (for those under 12 years). In cases where the patient was unaware of specific treatment details or healthcare costs, the attending physician was consulted. Additionally, the study included interviews with patients and clinicians to gather their perceptions on the current state of thalassemia management, treatment deficiencies, and unmet needs.

In this study, data were initially collected from 422 patients. After excluding invalid questionnaires, which exhibited clear signs of arbitrariness, omissions, or errors, a total of 418 valid questionnaires were retained.

Statistical analysis

Missing data, as well as summary and domain scores on the TranQoL, were calculated according to the guidelines provided in the referenced guidebook [23]. Data analysis was performed using the Statistical Package for the Social Sciences (SPSS) software (version 26.0, IBM Inc., Armonk, NY, USA). Baseline characteristics of the participants were presented as percentages and mean ± standard deviation (SD). To identify potential determinants of HRQoL, both univariate and multivariate analyses were conducted. The relationships between each domain and various factors, including demographic, clinical, and health service utilization, were assessed using the chi-square (χ^2) test, analysis of variance, and t-test. A p-value of <0.05 was considered statistically significant.

Results

Demographic characteristics

A total of 418 children with TDT, or their parents, were interviewed in this study. The majority of patients were male (60.53%) with a mean age of 11.13 ± 4.03 years, and a relatively large number were at the primary school level, aged 6 to 11. Given that Guangxi Province has a significant Zhuang ethnic minority population, the proportion of Han and Zhuang patients in this study was nearly equal. A substantial 72.25% of patients' families were found to be in debt, with 66.75% reporting an annual income of less than 50,000 CNY (approximately 7,018 USD). Notably, 47.1% of patients faced annual direct

medical expenses exceeding one-third of their household's annual income. Additionally, 48.8% of respondents rated the financial burden of the disease as severe.

In terms of treatment, 66.03% of patients were able to receive regular care. Most patients (68.90%) were treated with a single iron chelating drug, and 24.88% presented with severe iron overload, indicated by serum ferritin levels exceeding 1,000 μg/L. The study also found that 71.77% of the children experienced complications related to thalassemia, particularly growth retardation and splenomegaly. Regarding surgical treatment, a relatively small proportion of patients underwent splenectomy (16.75%) and HSCT (17.70%) (Table 1). The following factors were found to be correlated with patients' TranQoL scores: age, ethnicity, the number of family members affected by TDT, family poverty, family indebtedness, school dropout, regular treatment adherence, the number of iron-chelator drugs used, the presence of complications, and HSCT.

HRQoL data

Table 2 provides a concise summary of the TranQoL scores within the study cohort, revealing an overall score of 60.6±16.3. The scores for the four domains ranged from 55.6±13.8 for family functioning to 66.9±22.5 for emotional health. The child version identified school functioning as the area with the lowest score (55.7±29.9), while the parent-proxy version indicated family functioning as the lowest (55.3±13.5). Across all versions, emotional health consistently scored the highest. Figure 1 shows the distribution of TranQol scores compared across four domains.

Table 3 presents the analysis of potential factors influencing overall TranQoL scores among the participants. The findings reveal a negative correlation ($P<0.05$) between overall TranQoL scores and several factors, including the patient's age, the number of family members with thalassemia, family indebtedness, and whether the patient had undergone HSCT. Additionally, the analysis identified a significant disparity in TranQoL scores between ethnic groups, with Zhuang patients reporting higher scores than Han patients. Moreover, adherence to regular treatment was positively correlated with higher TranQoL scores ($P<0.05$).

Discussion

In the current landscape, there is a notable lack of research focused on TDT patients in China. To our knowledge, this is the first study to investigate the HRQoL of transfusion-dependent β-thalassemia children in China. A review of the literature reveals a significant discrepancy in HRQoL scores between Chinese children with TDT and those in developed countries. For instance, studies conducted in North America (including Canada and the United States) have reported average HRQoL scores as high as 77, while a study in Singapore recorded a score of 70.3 [22, 24, 25]. This finding positions the HRQoL of Chinese patients with TDT at a low level, indicating the need for effective measures to improve HRQoL. The study revealed that increasing age, the presence of TDT within the family, and undergoing HSCT were significantly associated with lower TranQoL scores. Conversely, a positive correlation was observed between treatment adherence and higher TranQoL scores.

Research findings demonstrated that as TDT patients aged, their quality of life often deteriorated, corroborating previous studies [21, 24, 26–28]. This decline is likely due to the cumulative effects of prolonged transfusion treatments, escalating iron overload, and the development of various complications over time, which significantly disrupt patients' daily lives and their ability to engage in school. Consequently, this places a heavy economic and psychological burden on both patients and their families. Further analyses indicated that families with multiple thalassemia patients consistently exhibited lower TranQoL scores, highlighting thalassemia's systemic impact on both affected individuals and their families. Therefore, preventing thalassemia has profound implications not only for improving patients' quality of life but also for reducing the economic and psychological strain on patients and their families.

However, an unexpected phenomenon was observed: patients who had undergone HSCT scored lower on the TranQoL scale compared to those who had not undergone the procedure, creating a notable discrepancy with previous research findings [29–32]. In-depth interviews with participants and clinical doctors revealed that in China, resources for HSCT are scarce, and the costs associated with surgery and subsequent rehabilitation are daunting. In cases of complications or infections, most families lack the economic means to bear these burdens, which adversely impacts their quality of life. Given these circumstances, a comprehensive re-evaluation of the long-term effects and potential complications associated with this treatment is necessary. This evaluation should consider various factors, including the risk of infection, patients' long-term survival rates, and the incidence of complications. Additionally, there is an urgent need for further improvements and optimizations in health insurance policies and healthcare services to better support this treatment approach.

The differences in TranQoL scores observed between Zhuang and Han patients may indicate underlying disparities in genetics, culture, or socio-economic status, warranting further in-depth investigation. Additionally, the positive correlation between regular treatment and higher TranQoL scores underscores the essential role of consistent and adequate transfusions, along with

Table 1 Baseline characteristics of the children with TDT ($n=418$)

Factors	*N*(%)	TranQoL Score(mean ± SD)	F	*P*
Age (years)			4.616	0.003
< 6	40(9.57%)	69.0 ± 18.4		
6 ~ < 12	161(38.52%)	61.0 ± 14.9		
12 ~ < 15	115(27.51%)	58.5 ± 16.1		
15 ~ < 18	102(24.40%)	59.0 ± 17.4		
Sex			1.496	0.222
Male	253(60.53%)	61.4 ± 16.0		
Female	165(39.47%)	59.4 ± 16.7		
Ethnicity			7.536	0.001
Han	212(50.72%)	57.6 ± 16.2		
Zhuang	203(48.56%)	63.7 ± 15.9		
Other	3(0.72%)	58.9 ± 13.9		
Medical insurance			0.739	0.390
No	32(7.66%)	63.0 ± 15.7		
Yes	386(92.34%)	60.4 ± 16.3		
Number of persons with thalassaemia in the family			10.346	< 0.001
1	284(67.94%)	63.6 ± 16.1		
2	54(12.92%)	54.6 ± 12.9		
3	53(12.68%)	57.2 ± 14.4		
4	20(4.78%)	47.3 ± 17.1		
5	7(1.67%)	46.6 ± 16.1		
Household income (CNY)			0.852	0.466
0 ~ ≤ 25,000	109(26.65%)	60.6 ± 15.9		
25,000 ~ ≤ 50,000	164(40.10%)	61.2 ± 16.1		
50,000 ~ ≤ 75,000	42(10.27%)	61.3 ± 16.6		
> 75,000	94(22.98%)	58.0 ± 17.3		
Recognized as the poor household *			10.862	0.001
No	258(61.72%)	62.6 ± 16.6		
Yes	160(38.28%)	57.3 ± 15.3		
Debt situation			4.390	0.037
No	116(27.75%)	63.3 ± 16.5		
Yes	302(72.25%)	59.6 ± 16.1		
Dropped out of school due to thalassaemia			15.976	< 0.001
No	295(70.57%)	62.6 ± 15.7		
Yes	123(29.43%)	55.7 ± 16.8		
Time to nearest blood transfusion facility (min)			1.537	0.216
≤ 30	238(56.94%)	60.5 ± 16.5		
30 ~ ≤ 60	120(28.71%)	59.2 ± 16.9		
> 60	60(14.35%)	63.7 ± 13.6		
Regular blood transfusions			5.280	0.022
No	122(29.19%)	57.8 ± 16.7		
Yes	296(70.81%)	61.8 ± 16.0		
Regular therapy (excluding transfusion therapy)			16.853	< 0.001
No	92(22.01%)	54.6 ± 16.1		
Yes	326(77.99%)	62.3 ± 15.9		
Amount of iron chelator used			2.724	0.044
0	16(3.83%)	68.3 ± 19.2		
1	288(68.90%)	60.0 ± 15.9		
2	94(22.49%)	59.7 ± 17.1		
3	20(4.78%)	67.7 ± 12.7		
Number of complications (due to thalassaemia)			31.389	< 0.001
0	36(8.61%)	62.8 ± 15.6		
1	196(46.89%)	66.4 ± 15.9		

Table 1 (continued)

Factors	*N*(%)	TranQoL Score(mean ± SD)	F	*P*
≥ 2	186(44.50%)	54.1 ± 14.4		
Splenectomy performed			3.801	0.502
No	348(83.25%)	61.3 ± 16.6		
Yes	70(16.75%)	57.1 ± 14.5		
Stem cell transplants performed			19.228	< 0.001
No	344(82.30%)	62.2 ± 16.4		
Yes	74(17.70%)	53.2 ± 13.9		

*As per China's policies, a poor household is defined as a residential household with an income level below the nationally defined poverty line. This definition is administered by the poverty alleviation and development authorities. Typically, a poor household refers to a household with an annual per capita net income of less than 865 CNY.

Table 2 Summary of TranQoL scores for the children with TDT. (mean ± SD)

TranQoL Score	Mean Score for Versions	Child Version(*n* = 132)	Proxy Version(*n* = 286)
Summary Score	60.6 ± 16.3	60.1 ± 17.3	60.8 ± 15.8
Physical Health	61.8 ± 18.9	62.0 ± 18.9	61.7 ± 18.9
Emotional Health	66.9 ± 22.5	65.4 ± 23.6	68.1 ± 22.0
Family Health	55.6 ± 13.8	56.4 ± 14.4	55.3 ± 13.5
School/Career Health	55.7 ± 28.7	55.7 ± 29.9	55.6 ± 28.1

appropriate medication, in sustaining the health and well-being of thalassemia patients. However, the utilization of health services in China remains suboptimal, with medical services not fully meeting the needs of β-thalassemia patients. According to the Blue Book of Thalassemia in China (2020) [33], the treatment of thalassemia patients faces multiple challenges, including a tight blood supply, unstable availability of blood products, and iron overload. Financial constraints have led some patients to discontinue treatment, while a shortage of medical resources, particularly blood supply, has prevented others from receiving timely treatment. Furthermore, a lack of awareness among some patients about the importance of regular treatment also contributes to nonadherence to prescribed treatment regimens.

It is therefore imperative that relevant health departments and policymakers prioritize the healthcare needs of thalassemia patients. This includes enhancing the supply of medical resources, optimizing the distribution of these resources, and improving patients' awareness of regular treatment. Public awareness of the importance of voluntary blood donation must be heightened through increased publicity efforts to motivate greater participation. Improving policies that facilitate barrier-free mutual

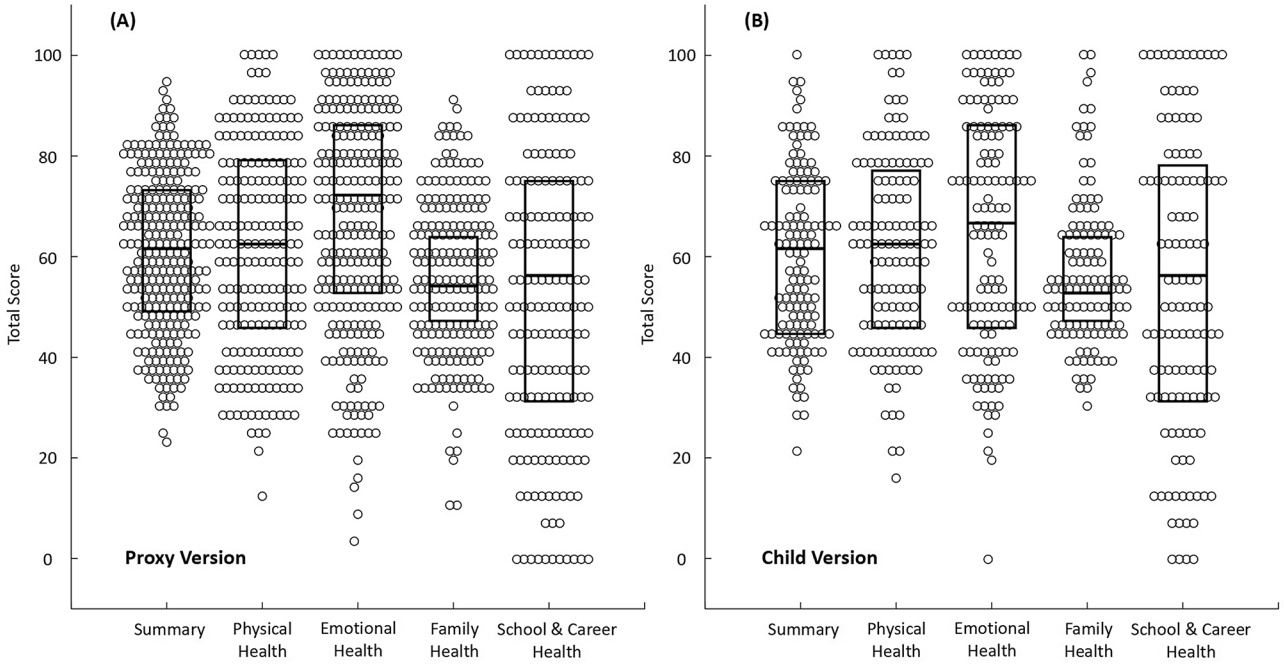

Fig. 1 Beeswarm graphs of (**A**) proxy version baseline TranQol scores and (**B**) child version baseline TranQol scores. The horizontal line represents the median, and the box indicates the 25–75 percentiles

Table 3 Multiple regression analysis of factors influencing HRQoL of children with TDT

Factors	β	SE	t	*P*	95%CI
Age (reference = under 6 years)					
6 ~ < 12	-0.168	2.653	-2.128	0.034	(-10.863,-0.430)
12 ~ < 15	-0.266	2.883	-3.367	< 0.001	(-15.378,-4.040)
15 ~ < 18	-0.224	2.951	-2.902	0.004	(-14.365,-2.761)
Sex (reference = Male)					
Female	-0.076	1.491	-1.704	0.089	(-5.472,0.391)
Ethnicity (reference = Han)					
Zhuang	0.092	1.525	1.967	0.049	(0.002,5.999)
Others	0.026	8.673	0.584	0.560	(-11.988,22.119)
Medical insurance(reference = No)					
Yes	-0.032	2.886	-0.705	0.482	(-7.707,3.641)
Number of persons with thalassaemia in the family (reference = 1)					
2	-0.135	2.272	-2.866	0.004	(-10.981,-2.045)
3	-0.011	2.327	-0.232	0.817	(-5.117,4.036)
4	-0.160	3.429	-3.537	< 0.001	(-18.871,-5.385)
5	-0.087	5.584	-1.964	0.050	(-21.949,0.011)
Household income (CNY)(reference = 0 ~ ≤ 25000)					
25,000 ~ ≤ 50,000	-0.025	1.833	-0.453	0.651	(-4.433,2.774)
50,000 ~ ≤ 75,000	-0.003	2.691	-0.063	0.949	(-5.463,5.121)
> 75,000	-0.040	2.204	-0.707	0.480	(-5.892,2.775)
Recognized as poor families (reference = No)					
Yes	-0.076	1.621	-1.579	0.115	(-5.746,0.628)
Debt situation (reference = No)					
Yes	-0.084	1.691	-1.829	0.068	(-6.420,0.232)
Dropped out of school due to thalassaemia (reference = No)					
Yes	-0.082	1.681	-1.736	0.083	(-6.224,0.386)
Time to nearest blood transfusion facility (min)(reference = ≤ 30)					
30 ~ ≤ 60	-0.041	1.691	-0.888	0.375	(-4.825,1.824)
> 60	0.045	2.206	0.959	0.338	(-2.222,6.453)
Regular blood transfusions (reference = No)					
Yes	0.039	1.845	0.763	0.446	(-2.221,5.035)
Regular therapy (excluding transfusion therapy) (reference = No)					
Yes	0.150	2.045	2.888	0.004	(1.886,9.926)
Amount of iron chelator used (reference = 0)					
1	-0.180	3.743	-1.697	0.091	(-13.712,1.008)
2	-0.071	4.020	-0.695	0.487	(-10.698,5.108)
3	0.051	4.936	0.788	0.431	(-5.816,13.596)
Number of complications due to thalassaemia (reference = 0)					
1	0.074	2.684	0.900	0.368	(-2.860,7.693)
≥ 2	-0.137	2.782	-1.616	0.107	(-9.965,0.976)
Splenectomy performed (reference = No)					
Yes	0.009	2.135	0.185	0.853	(-3.804,4.593)
Stem cell transplants performed (reference = No)					
Yes	-0.127	2.146	-2.519	0.012	(-9.625,-1.187)

aid in blood donation is also essential. Actively encouraging relatives and friends of patients from severely disadvantaged areas to donate blood without compensation can help ensure the fairness and sustainability of blood supplies. For example, in the Maldives, the National Centre for Thalassemia includes a blood bank where 80% of the blood supply for individuals with severe thalassemia is provided primarily by family members [34]. Moreover, for patients who are able to undergo regular treatment, exploring ways to further solidify treatment effects, minimize complications, and enhance quality of life remains a critical area of research for clinicians and researchers.

The survey highlighted the lowest scores in school functioning as reported by the children and in family functioning according to the proxy questionnaire, revealing discrepancies in focal concerns regarding the

patients' quality of life among different evaluators. Specifically, children appear more vulnerable within the school setting, while parents express greater concern for the familial situation of the patient. Therefore, interventions and support services tailored for thalassemia must carefully address the specific needs of both patients and their families, catering to their multifaceted requirements to enhance overall quality of life.

While China has increasingly focused on the prevention and treatment of thalassemia, Guangxi Province initiated a tailored thalassemia prevention and control program in 2010, adapted to local conditions. This initiative emphasized key components such as premarital healthcare and genetic counselling, prenatal screening and diagnosis, and newborn screening and treatment. It implemented various policies and management measures, strengthened professional expertise, promoted thalassemia prevention knowledge through multiple channels, and explored new technologies for disease prevention and control. Additionally, relevant health insurance policies have significantly reduced the medical cost burden on patients. Between 2010 and 2022, Guangxi Province screened 18,299,700 individuals for thalassemia, with 1,056,200 genetically diagnosed and 102,500 prenatally diagnosed, resulting in a reduction of 15,500 births of children with TDT. This approach has led to the development of the distinctive "Guangxi Model." However, compared to mature prevention and control systems abroad, there remains considerable potential for learning and enhancement. The findings of this study reveal the quality of life and healthcare service utilization among thalassemia patients in China, identifying crucial factors affecting these areas. These insights provide a scientific foundation for refining thalassemia prevention and control efforts in China, optimizing health service utilization, and alleviating the disease burden, demonstrating both innovative and practical significance.

This study has several limitations. It focuses on patients with TDT in Guangxi Province, but due to study constraints, most participants were from the top three cities in Guangxi by GDP, leaving a gap in data from areas with lower economic levels. As a cross-sectional study, it captures data at a single point in time, which limits the ability to track changes in participants over time. Furthermore, while our study used well-established disease-specific scales, most studies continue to use generalized quality of life questionnaires (such as the SF-36), making comparisons with thalassemia patients in other countries and regions challenging. Future research could benefit from employing a combination of generic and specific scales and conducting multi-regional and multi-timepoint studies to expand the sample size, allowing for a more precise investigation of causal relationships between variables.

Abbreviations

HRQoL	Health-related quality of life
TDT	Transfusion-dependent β-thalassemia
HSCT	Hematopoietic stem cell transplantation
TranQoL	The Transfusion-Dependent Quality of Life
SD	Standard deviation

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Author contributions
Q.J.: Study design; investigation; data analysis; and writing(original draft). L.B.: Study design; investigation; data analysis. M.J.: Project design and administration; investigation; and writing (review and editing). Z.X.: Connecting and recruiting patients for research; provision of clinically-relevant advice. W.J.: Study design; investigation. D.Q.: Writing (editing). Z.S.: Investigation. C.Y.: Conceptualization of the study and funding; Supervision; project administration; and writing (review and editing). All authors revised the manuscript and approved the final version for submission.

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Data availability
The datasets used and/or analysed during the current study are available from the corresponding author on reasonable request.

Declarations

Ethics approval and consent to participate
The study was approved by the Medical Research Ethics Committee, School of Public Health, Fudan University (IRB#2022-07-0978). Patients provided written informed consent according to the Declaration of Helsinki.

Competing interests
The authors declare no competing interests.

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