BMJ Public Health

Experiences of family caregivers of children living with thalassaemia-major in Karachi: a phenomenological study

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To cite: Baqar M, Ahmed JM, Asim M, *et al*. Experiences of family caregivers of children living with thalassaemia-major in Karachi: a phenomenological study. *BMJ Public Health* 2024;**2**:e001359. doi:10.1136/bmjph-2024-001359

► Additional supplemental material is published online only. To view, please visit the journal online (https://doi.org/10.1136/bmjph-2024-001359).

Received 22 April 2024
Accepted 10 October 2024

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ABSTRACT

Introduction *Thalassaemia* major, a global health burden, presents a growing challenge in Pakistan's strained healthcare system. This study explores how caregivers of children with beta-thalassaemia major navigate healthcare services, aiming to identify facilitators and barriers to accessing optimal care.

Methods A qualitative interpretative phenomenological approach was employed. In-depth interviews with 18 purposively sampled caregivers from Karachi thalassaemia centres were conducted. Thematic analysis using a combined inductive-deductive approach identified themes within the interview data.

Result The study revealed significant challenges for caregivers at individual, interpersonal and organisational levels. Lack of awareness about carrier states, limited disease knowledge, financial constraints and concerns about the child's future emerged as primary hurdles. Parents felt helpless due to the absence of emotional and social support for their children's treatment. Obstacles such as complicated registration processes at thalassaemia-care centres, high costs of chelating agents and blood unavailability were major barriers to seeking care and caregiving. Additionally, the study highlighted the absence of guidelines for thalassaemia-carrier detection among mothers during antenatal care.

Conclusion The study emphasised the importance of implementing premarital screening programmes due to the lack of knowledge about the disease and carrier state. To prevent the disease, it is crucial to include thalassaemia-carrier detection for mothers in antenatal guidelines and provide counselling at the primary level. Additionally, caregivers encountered treatment accessibility issues, prompting the establishment of a satellite thalassaemia centre linked to a top-tier tertiary care hospital in the public sector. This initiative addressed treatment challenges and improved overall care for patients with thalassaemia and their caregivers.

WHAT IS ALREADY KNOWN ON THIS TOPIC

⇒ Existing research primarily focuses on thalassaemia clinical management, with limited understanding of family caregiver experiences, especially in accessing and navigating healthcare services for children with thalassaemia in Pakistan.

WHAT THIS STUDY ADDS

⇒ This study offers unique insights into the challenges faced by family caregivers and identifies barriers to accessing healthcare services. It also highlights the critical role of caregiver support in managing the disease and emphasises the need for comprehensive support.

HOW THIS STUDY MIGHT AFFECT RESEARCH, PRACTICE OR POLICY

⇒ The study findings have implications for future research on developing and evaluating interventions to address the identified barriers to care. Healthcare providers and policy-makers should prioritise developing caregiver support programmes and improving access to essential services for children with thalassaemia. It underscores the need for comprehensive support, improved healthcare access, and preventive measures like premarital and prenatal screening for patients with thalassaemia and their caregivers.

INTRODUCTION

Thalassaemia stands as one of the most common inherited blood disorders, impacting males and females equally worldwide.[1] This condition follows an autosomal recessive pattern,[2] stemming from abnormal gene mutations that lead to reduced or absent production of alpha (α) or beta (β) globin chains, resulting in either alpha (α) or beta (β) thalassaemia.[3]

Individuals with Cooley's anaemia, known as β-thalassaemia (major), typically present severe clinical symptoms, requiring lifelong red blood cell transfusions and chelation therapy to manage iron overload. Conversely, beta-thalassaemia intermedia or Mediterranean anaemia often does not require transfusions but may show signs of anaemia and iron overload later in life.[4] Those with beta-thalassaemia minor or carriers usually remain clinically asymptomatic.

Beta-thalassaemia is predominantly prevalent in regions like the Mediterranean, Italy,

Greece, Africa, the Middle East, the Far East and South Asian countries.[2] Its occurrence is not common in North America and Europe.[5] Asia has the highest prevalence, comprising 95% of α-thalassaemia and β-thalassaemia births in Southeast Asia.[6] Low-income and middle-income countries witness an estimated 50 000–100 000 deaths annually due to this condition.[7 8] In Middle Eastern Asian countries like Iraq, Jordan, Lebonan, Saudi Arabia, UAE, Oman and Qatar,[8] consanguineous marriages contribute to high beta thalassemia carrier rates.[5] South and Southeast Asian countries also exhibit high carrier rates, up to 12%.

Thalassemia is a growing public health concern in Pakistan despite being preventable. With a population exceeding 220 million, the country lacks a national thalassemia registry and crucial epidemiological data on its incidence, prevalence and mortality rates regarding thalassaemia and other blood disorders. Estimates suggest a 5%–7% carrier rate, resulting in 8–10 million β-thalassaemia carriers and around 100 000 individuals with beta-thalassaemia major. Annually, 5000–9000 new β-thalassemia cases are added to Pakistan's population due to a trend of marriages among carriers. Despite improvements in literacy, many remain unaware of the increasing population affected by beta-thalassaemia major.

Social stigma, misconceptions about religious beliefs related to abortions and family marriages, and limited awareness of genetic diseases have exacerbated the problem. Ethnic, social and cultural factors contribute to the high prevalence of transfusion-dependent thalassaemia in Pakistan. Promoting awareness about premarital and antenatal screening appears to be the primary solution to address this.

Pakistan faces challenges in implementing national policies and public programmes for premarital screening, counselling and early detection of thalassaemia in families. Although some provinces have passed laws on premarital screening for thalassaemia, these laws remain largely unimplemented due to government disinterest, lack of public awareness and the cost of screening tests.

The country's weak health system, limited resources and inadequate accessibility to supportive treatments such as regular transfusions with chelation therapy and multidisciplinary care further compound the issue. Patients with thalassaemia in Pakistan suffer from a significantly reduced life expectancy of 10–12 years, primarily due to complications arising from iron overload and substandard treatment.[9–11]

The majority of patients with transfusion-dependent thalassaemia come from low socioeconomic backgrounds. Public sector hospitals have limited resources to manage chronic diseases like transfusion-dependent thalassaemia. Welfare organisations and non-governmental organisations, through thalassaemia centres, step in to provide essential services such as monthly transfusions and chelation therapy when required. However, these centres face challenges due to the growing number of patients, with around 5000 new cases annually. Despite their efforts, these centres often fall short of delivering comprehensive care. They are understaffed and lack clinicians and ancillary staff with the expertise needed for optimal management. Most patients receive oral chelation therapy, and only a few with chronic conditions get intravenous chelation therapy, typically administered once or twice a month during their transfusion visits.

Most NGOs offer supportive transfusion services to patients, but only a few provide chelation therapy. For emergency services, patients turn to tertiary hospitals. In Karachi, four NGOs assist patients with regular screenings, hormonal checkups and cardiac health assessments.

The absence of national programmes for thalassaemia guidance, screening and family counselling underscores the necessity to comprehend the experiences of parents and caregivers involved in their children's lifelong healthcare. Hence, this study aimed to explore the challenges and experiences of parents and family caregivers of children with beta-thalassaemia (major) in Pakistan, identifying the obstacles and facilitators affecting their access to healthcare services.

Understanding parents' perspectives and lived experiences is crucial for effective interventions, health advocacy and policy improvements in tackling this pressing health issue.

MATERIALS AND METHODS

This study, conducted in Karachi, Pakistan, employed a descriptive phenomenological qualitative approach. Karachi offers diverse healthcare services through public, private and NGO thalassaemia centres, serving not only its residents but also those in surrounding regions like interior Sindh and nearby areas of Balochistan. Eight private/NGO thalassemia centres in Karachi are focused on providing crucial daycare services, particularly to patients from remote areas. Eighteen caregivers, primarily parents of children with thalassaemia, were purposively sampled from these centres for in-depth interviews until saturation. Interview guide was initially prepared in English and translated into Urdu, ensuring consistency via a pilot test and the involvement of bilingual experts in meticulous steps like back translation and expert reviews. Interviews were conducted between June and August 2022. Transcripts, verbatim in Urdu, were later translated into English for analysis, maintaining precision through bilingual experts' involvement, as per the requirements of the master's thesis of Aga Khan University, which is accessible to other scholars, colleagues and teachers in the university repository (given the university's international recognition). This has also opened up opportunities for publishing the research. The interview guide is attached as an online supplemental file.

Conceptual framework of the study

This study used the socioecological framework, making specific thematic adjustments to understand parental or

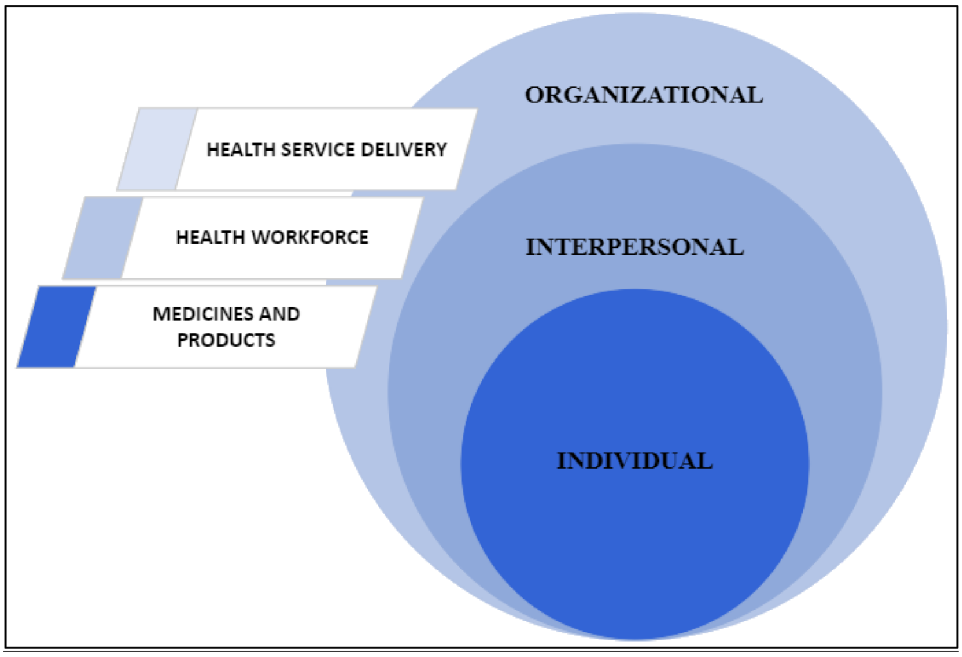

Figure 1 The modified socioecological and WHO building block framework.

family caregiver challenges. This framework delves into individual behaviours influenced by various levels of factors, as described in prior research.[12 13] It highlights the interplay among individual, interpersonal, organisational, community and policy-level elements, crucial in preventing public health issues.[14] This model is centred on health, focusing on its determinants.

For this research, the socioecological framework was adopted with tailored adaptations at the organisational level. Specifically, three components from the WHO health system building block framework were integrated at this level. The study aimed to explore the experiences of parents and caregivers seeking treatment for their children. The study used a modified socioecological framework, to uncover individual, interpersonal and organisational challenges.

Figure 1 shows the modified socioecological model employed in the study.

Patient and public involvement statement

The caregivers of patients with thalassaemia, including parents and guardians, were actively involved in this study. They participated through interviews, providing valuable insights and experiences that shaped the research findings. Although they were not involved in the design or conduct of the study, their input was essential in understanding the challenges faced by patients with thalassaemia and their families. The study outcomes will be disseminated with relevant stakeholders, including healthcare providers and advocacy groups, to ensure the findings benefit both patients and their caregivers.

Study procedure

Before interviews, participants were briefed about the study's scope and assured confidentiality. Each participant gave written consent, and interviews, conducted in private spaces, ensured privacy. Participants explicitly agreed to audio recording, transcription and translation afterward. Interviews lasting between 30 and 45 min, conducted at thalassaemia centers, adhering to the ethical principles of the Declaration of Helsinki.

Data analysis

Participants consented to an audio recording before interviews. Notes were taken and cross-checked with tape recording transcripts. Audio files were securely transferred to password-protected laptops with unique IDs. The thematic analysis involved translation, transcription and identifying themes and subthemes from data. Both inductive and deductive approaches were applied, with codes extracted and themes generated from transcripts. Two researchers independently coded and resolved discrepancies. In-depth interview data underwent triangulation from various thalassaemia centres to ensure reliability.

Researcher reflexivity and bracketing

In descriptive phenomenology, bracketing helps researchers set aside their preconceptions to impartially analyse parents' experiences with children with thalassaemia.[15] Reflexivity involves consciously separating personal values from data analysis, reducing bias.[9 11] This approach was used before data collection to ensure unbiased analysis. The study also followed the Consolidated criteria for Reporting Qualitative research (COREQ) checklist, ensuring research validity across qualitative domains[16] and boosting the study's reliability.

Table 1 Demographic profile of the study participants

Caregivers	Total (n=18)
Age of parents	
25–40 years	10
41–55 years	7
55 and above	1
Gender	
Male	7
Female	11
Academic qualification	
None (illiterate, no formal school education)	8
Primary (grade 5)	2
Matric (grade 10)	5
Bachelors (undergraduate degree)	3
Employment status	
Daily wage	8
Regular employment	9
Retired	1
Relationship with spouse before marriage	
Cousin	10
No relation	8
Number of children with thalassemia major	
One	15
More than one	3

RESULTS

The demographic profile in table 1 shows that the study involved caregivers—mothers, fathers or both—seeking treatment for children with thalassaemia major. At NGO *thalassaemia* centres, 18 participants (7 males, 11 females) aged 27-75 were interviewed. Many had little education, with most being illiterate. They often married cousins and earned a living through jobs and daily wages.

Table 2 illustrates three main themes derived from a modified socioecological framework. These themes and their related subthemes emerged from the analysis of in-depth interviews

Details of table 2 with significant sentences and meaning are attached as online supplemental table 3.

Individual level

The individual-level theme explores the unique experiences, perspectives and challenges faced by caregivers looking after individuals dealing with disease. It focuses on how these caregivers understand the condition, manage the situation and meet the specific care requirements of affected children. This theme emphasises the caregiver's journey in navigating the disease alongside the affected individuals, shedding light on their struggles and caregiving responsibilities.

Table 2 Themes, subthemes and categories derived from analysis

Theme	Subthemes	Categories
Individual level	Insight into the disease process	1. Knowledge and understanding of disease
		2. Detection of the carrier state
	Coping with the disease process	1. Increased responsibilities
		2. Neglected kids
		3. Financial constraints
		4. Surviving the misery and grievance
		5. Complication of disease
	Demanding care and nursing needs of children	1. Mood swings and Aggressive behaviour
		2. Close monitoring of children
		3. Difficult time
Interpersonal level	Social support network	1. Behaviour of the family and relatives
		2. Workplace problems
Organisational level	Obstacles in seeking care at public and private healthcare organisations	1. Behaviour of the health workforce
		2. Thalassemia service delivery
		3. Registration at the thalassemia centres
		4. Blood availability and arrangements
		5. Medicines availability and affordability

Insight into the disease process

Insight into the disease process refers to how well caregivers understand the disease. It covers their grasp of how the disease develops, its effects on the body, the symptoms it causes, possible complications and how it progresses in affected individuals. This aspect examines how much caregivers know about the disease—its causes, impacts and medical details. It involves their awareness and understanding of the disease's traits, helping them handle its challenges and impacts on themselves and their children.

Caregivers expressed a lack of detailed knowledge about thalassaemia when questioned. While familiar with the term, many lacked specific information about its prevalence or available treatments. For instance, one person mentioned recognising the name but not understanding how common the condition is or the available treatment options. This suggests a limited understanding of the disease's prevalence and management.

Additionally, misconceptions existed within communities regarding the causes of thalassaemia major. One participant highlighted a prevalent belief that "the

condition might be caused by fever, immunization, or injuries, although in reality, there is no such connection." (PC 003)

In-depth interviews revealed that none of the participants were aware of being carriers of the disease until their child received a diagnosis. Despite visiting private hospitals for prenatal care, none was informed about their carrier status during these visits. This lack of awareness persisted until their child was diagnosed with thalassaemia major.

A female caregiver mentioned "she remembered undergoing blood tests during pregnancies but was never informed about being a carrier until her child was diagnosed with the disease." (PC 001). Similarly, a male caregiver highlighted, "Despite multiple pregnancies and consistent anemia in his wife, they only discovered their carrier status when their child fell ill." (PC 002). The mothers were not tested for thalassaemia carrier state during pregnancy, although regular blood routine checkups were done to screen for iron deficiency. These accounts underscore the significant gap in caregivers' knowledge about their carrier status and the disease, highlighting the need for improved information dissemination within healthcare systems.

Coping with the disease process

Coping involves managing difficulties or challenging situations. Caregivers mentioned various life changes while caring for their children with thalassaemia. These challenges include increased responsibilities, overlooked needs of other children, financial struggles due to out-of-pocket expenditures dealing with emotional pain (acceptance, grief and hopelessness) and handling the complexities of the disease.

Caring for children with thalassaemia significantly changed caregivers' lives across various dimensions. First, these individuals shouldered increased responsibilities, managing household chores and sharing economic burdens with their partners. Many lacked support, as depicted by one caregiver, who shared, "I have to manage house chores early in the morning before coming to the center because it takes a whole day for the transfusion. There is no one to look after the house. I have started stitching clothes on wages to have some savings for my son as my husband is a daily wager (earning US$ 3.4/day)". (PC 007)

Second, this commitment led to emotional turmoil, with caregivers feeling guilty about neglecting their other children. Some expressed regret for not adequately focusing on their younger children's needs. One caregiver said, "I feel that I have neglected my younger one. I was too busy with the elder one that I did not even have vaccinations of my younger one on time." (PC 012)

Financial challenges loomed large for these families. High transportation costs to thalassaemia centres, primarily through taxis due to a lack of private vehicles, imposed significant burdens. Additionally, the expenses associated with medications were considerable, forcing families to rely on charity or request discounts at care centres. One caregiver shared, "The disease was undiagnosed till the age of 3 years, and none of the public hospitals accepted him. We had to take him to the private hospital when his condition deteriorated multiple. We ended up in heavy debt. We have no savings, and sometimes we do not have Rs 100 (US$ 0.36/36 cents) to bring him to the center for transfusion, so it gets delayed by 7–10 days". (PC 004) This financial strain often led to accumulating debts and delays in necessary treatments.

The emotional toll of managing thalassaemia was profound. Acceptance of the disease varied, with some finding solace in religious beliefs while others focused on embracing reality early on. However, the fear of losing their child lingered, causing deep emotional distress. Hopelessness regarding their children's futures prevailed, rooted in the continuous necessity for treatment and witnessing other children's suffering. One male caregiver whose three elder daughters had already passed away from thalassaemia stated about his son's future, "I think nothing about his future; he has to undergo transfusion till the end of life. There are thoughts about his end too as I have seen the sufferings of my elder daughters which I cannot explain." (PC 006)

Another caregiver of 14 years old boy said, "Life and death are in the hands of Allah; we cannot do anything and think nothing about their future." (PC 015)

Lastly, the challenges extended to their children's education. While caregivers endeavoured to enrol their thalassaemia-affected children in formal schools, disease-related complications frequently disrupted their education. Instances of discontinuation arose due to transfusions or complications like infections. One caregiver desperately said, "She studied for three years in a school but had transfusion reactions every time and transfusion-transmitted infection (Hep C), so she could not continue her studies." (PC 011)

Another caregiver said, "The school was cooperating, and she studied until grade five but had to come to the center for drip daily, so she dropped the school two years back." (PC 005)

Overall, caregivers grappled with multifaceted challenges, encompassing increased responsibilities, financial strains, emotional distress and disruptions in their children's education due to thalassaemia.

Demanding care and nursing needs of children

This subtheme encapsulates the high level of care and specific nursing requirements essential for children affected by thalassaemia. It highlights the specialised attention, medical care and daily support crucial for their well-being.

In essence, the demanding care and nursing need to underscore the intricate and comprehensive support required by children with thalassaemia, encompassing the medical aspects and the daily care essential for their health and well-being.

Caregivers noted how the demands of the disease, like strict dietary needs and frequent transfusions, often led to stubbornness and aggression in their children. Parents went to great lengths to accommodate their wishes to avoid their sense of isolation. One parent shared, "We never force our daughter into anything. We cater to her needs even if they arise late at night." (PC 008)

Parents emphasised the need for continual vigilance regardless of their child's age. They learnt to interpret subtle signs, such as changes in appetite or behaviour, indicating when their child required a transfusion. One caregiver shared her experience by saying, "I gauge his transfusion time through indicators like loss of appetite, fever, excessive sleep, or aggressive behavior. He never tells us directly." (PC 001) Even older children who managed their medications needed monitoring, especially in regulating physical activities that could affect their health. Caregivers of older children shared their experience and said, "They are old enough to take medications and communicate their condition, but we need to keep a watchful eye to prevent them from engaging in strenuous activities like sports and cycling. Otherwise, their hemoglobin levels might plummet." (PC 006) and (PC 013)

The journey of caring for a child with thalassaemia was described as highly challenging. Seeking treatment, managing the disease and witnessing their children in pain or discomfort took a significant emotional toll on caregivers. One female caregiver said, "These 14 years of my life have been incredibly challenging. We have had to rush around, dealing with her pain and fever. She undergoes regular canola placement, enduring immense pain and irritation, yet she does not express it. It is heartbreaking to see her consume more medicines than food." (PC 005)

A lack of proper guidance after diagnosis added to the difficulties, forcing some to rely on their instincts to manage their child's condition. Another caregiver shared her experience and said, "We were not properly guided after his diagnosis, making it incredibly tough to manage his condition. There were times when his condition worsened, and we felt hopeless. We navigated his care based on our understanding. I try to console him, telling him he needs the pump, but he cries in agony, saying it is unbearably painful. Managing him in this state is a real challenge." (PC 010)

These accounts shed light on the emotional and physical strain experienced by caregivers. They often found themselves managing multiple children with thalassaemia or dealing with their children's reactions during medical procedures, presenting significant challenges in their caregiving journey.

Interpersonal level

The interpersonal level theme centres on the social support network experienced by caregivers, encompassing interactions with siblings, relatives, neighbours and workplaces while seeking treatment for their children. It highlights the community and social experiences during this journey.

Social support network

This subtheme refers to the help and support caregivers receive from different people in their social circles while they care for their children with thalassaemia. It includes assistance, advice and relationships with family, friends, neighbours and colleagues as they tackle the difficulties of managing this condition.

Within the participants' social circles, the behaviour of family and relatives played a crucial role in their caregiving experiences. While some caregivers received significant support, including emotional and financial assistance, from relatives, others faced unsupportive attitudes and severed relationships. Those surrounded by familial support felt emotionally uplifted and financially aided, finding solace in sympathetic inquiries about their child's health. For instance, one participant mentioned, "My parents and siblings are remarkably caring and supportive. They have gone above and beyond to provide both financial and emotional assistance. Their empathy and comforting words serve as our pillar of strength." (PC 003) Their primary strength is the remarkable care and financial assistance their parents and siblings provide.

Conversely, some caregivers encountered hurtful comments, blame and a lack of empathy from relatives. One participant shared her distressing experience, "Our family, including siblings, never extended help or support. Relatives did not accept the disease, and despite my daughter being 14 now, they persistently attribute her condition to our wrongdoing. We are subjected to their bitter and mocking remarks." (PC 005) These negative experiences included hurtful remarks attributing the child's condition to the family's actions, leaving lasting emotional distress.

Moreover, workplace challenges compounded the difficulties for parents, with some facing job losses due to repeated visits to thalassemia centres. A father, working as a gardener, shared his experience: "I've faced job loss on multiple occasions due to my daughter's frequent visits to the thalassemia center for transfusions. Requesting time off or taking half-day leave was not acceptable to my employers. Sometimes, even after making the trip, the required blood was not available, rendering our efforts futile. As a result, I now need to work even harder to make ends meet and cover her treatment expenses." (PC 008)

Employers' uncooperative behaviour forced parents to choose between accompanying their children for treatment and risking their livelihoods. This struggle led to significant financial burdens, compelling some parents to work harder to cover their children's treatment expenses.

Organisational level

The third theme captured the interactions and encounters of caregivers within healthcare organisations when seeking treatment for their children. This theme focused

on their experiences and engagements at the institutional level while managing their children's thalassaemia. It documented their interactions with healthcare professionals, the challenges faced within healthcare settings, and their overall experiences navigating the healthcare system.

Obstacles in seeking care at public and private healthcare organisations

This subtheme outlines caregivers' challenges when seeking medical care for their children in public and private healthcare institutions. It highlights the difficulties, barriers and hurdles encountered while navigating these healthcare systems. These challenges might include access, quality of care, financial constraints, communication gaps with healthcare providers, or any other impediments that hinder the smooth process of seeking and receiving adequate medical attention for patients with thalassaemia.

Caregivers navigating thalassaemia care encountered challenges within public and private healthcare domains. Their journey traversed hurdles in service delivery, centre enrolments, blood accessibility and medication availability. Public hospitals presented a grim reality marked by logistical woes, erratic blood supply and financial strains, compelling many to seek alternatives due to unmet diagnoses and unresolved issues. One caregiver's frustration with a public hospital was palpable, saying, "The treatment at public hospitals was deplorable as if they were treating our children like animals. I reached a point where I decided that even if it meant my child's death at home, I would not subject him to treatment at a public hospital again." (PC 003) Another caregiver shared his perspective, stating, "Our encounter with public hospitals was disappointing; they failed to diagnose my daughter's condition. Consequently, we chose not to pursue treatment there. I struggled for six months to secure an appointment for my daughter's growth assessment, available only at a public hospital, but it yielded no results. To date, my daughter remains untested." (PC 008)

Enrolling children at specialised thalassaemia centres proved onerous, often met with capacity-related rejections, though some found solutions through referrals or multiple enrolments across centres. One caregiver shared her experience, saying, "For a year, I tried registering my child at various thalassemia centers every six months, only to face consistent refusal. It was through a referral from XXX welfare society that we were directed to this specific thalassaemia center." (PC 010) In some fortunate cases, parents secured registration at multiple centres to access thalassaemia care. One male caregiver highlighted his situation: "Thanks to Dr. ABC's efforts, my daughter was enrolled at the EFG foundation, renowned for comprehensive thalassaemia care. However, my son and two other daughters were not accepted for full enrollment, though they were granted outpatient checkup privileges. We turned to this center for transfusions and medicines, where no fees are charged, and donor arrangements are unnecessary. We maintain enrollment at both the EFG foundation and the ABC center." (PC 006)

Blood scarcities plagued both NGO-based and public facilities, intensifying the burden as caregivers struggled to secure donors, leading to recurring transfusion challenges. A mother grappling with the realities of having two children with thalassaemia major expressed her ongoing struggle, saying, "Over 15 years, we have frequently faced setbacks due to blood shortages. Our two children require four blood transfusions per month, yet we are compelled to arrange one donor replacement each month. Pleading with others to donate blood for our children's sake has become a regular plea." (PC 013) The recurring theme of blood scarcity persisted in caregivers' experiences, with one participant succinctly stating, "Our most pressing challenge is the consistent unavailability of blood. We find ourselves in perpetual shortage, necessitating repeated visits." (PC 001)

Moreover, crucial iron-chelating medications, pivotal for managing thalassaemia, remained elusive at public hospitals, pressuring caregivers to resort to expensive private pharmacies or cheaper, less-effective alternatives, accentuating the risks of inadequate treatment. One caregiver detailed her experience, stating, "Initially, we resorted to a more affordable medication, but it proved unsuitable for her. The alternative iron-chelating medication came with a hefty price tag of Rs 30–35 k (US$110–130) per month, rendering it financially unfeasible. Consequently, we refrained from administering the medication for an extended period, resulting in complications and stunted growth." (PC 012)

These recurring battles with blood deficits, diagnostic lapses and inaccessible medications underscored the multifaceted and persistent obstacles faced by caregivers in accessing comprehensive thalassaemia care.

DISCUSSION

This study highlighted a significant lack of understanding among illiterate and educated caregivers regarding thalassaemia. Like studies in Jordan and Iran, parents in this study demonstrated limited knowledge about the disease's inheritance, signs, symptoms and treatment.[17–19] This lack of understanding led to delayed diagnoses and the misinterpretation of symptoms, often confusing thalassaemia-related indications with common childhood ailments like teething. Notably, many parents in Karachi were in consanguineous marriages, increasing the risk of thalassaemia in their children. This lack of awareness about thalassaemia and carrier states among expectant mothers during antenatal care resulted in missed opportunities for prenatal screening and early intervention, a concerning trend noted in private healthcare settings.

This study exposes knowledge gaps about thalassaemia among Karachi caregivers, regardless of education level. Misinterpretations of symptoms and lack of awareness about carrier states during pregnancy led to missed opportunities for early intervention. To combat

this, integrating thalassaemia carrier testing into antenatal checkups and implementing premarital screening programmes, particularly in communities with high rates of early/consanguineous marriage, are crucial. Educational campaigns focused on young adults at colleges and universities can further bridge this knowledge gap.

Caregivers, primarily mothers, demonstrated diverse coping strategies but lacked clear guidelines on crucial aspects like diet, medication and treatment. This knowledge gap increased risks and negatively impacted mortality rates. Tailored guidance and counselling programmes and comprehensive support systems like additional home assistance are essential to reduce caregiver stress, improve family quality of life and prevent neglect of other children. By addressing these knowledge gaps and implementing targeted interventions, we can empower families and healthcare professionals to manage thalassaemia and effectively improve the lives of those affected.

Beyond managing their child's Thalassemia, Karachi caregivers grappled with behavioural changes like aggression and stubbornness, often linked to recurrent treatments. These shifts strained family dynamics, highlighting the need for age-specific guidance and counselling for caregivers. Finances presented another hurdle, particularly for low-income families.[17 20–24] Though accessing healthcare through public centres or NGOs, covering treatment-related costs like transport and medication remained a struggle. Frequent appointments meant lost wages and workplace issues, further deepening financial woes. Addressing these financial constraints, like offering free vocational training for both parents, emerged as crucial to empowering families and effectively managing thalassaemia in their children.

The study highlighted the emotional toll on parents of children with thalassaemia, reporting feelings of helplessness, hopelessness, grief and agony due to the exhaustive treatment regime and lifelong reliance on transfusions and medications without a cure. Parents expressed deep concerns about their children's future and experienced significant distress witnessing their children's suffering. Caregivers grapple with multifaceted challenges on both individual and financial fronts, underlining the critical need for comprehensive support.

Comprehensive education and counselling regarding thalassaemia and its management are crucial to aid parents in coping with their children's condition. Emotional support is pivotal in helping parents navigate these challenges.[20 21 25] The research emphasises the importance of appropriate guidance, including age-specific counselling sessions and financial assistance programmes.

Increasing awareness and premarital screening programmes are essential to bridge the information gap regarding thalassaemia and carrier states in the general population. The study emphasises the healthcare team's crucial role in supporting parents and providing necessary interventions. To address specific challenges faced by parents of children with thalassemia major, the healthcare system should address issues like drug shortages and offer comprehensive support programmes.[26] Healthcare professionals must be sensitive to parents' experiences and challenges, necessitating adequate training and the implementation of tailored support initiatives for parents dealing with thalassaemia major in their children.

In Karachi, caregivers of children with thalassaemia major face a significant uphill battle. Lack of awareness about the disease and carrier states hinders early diagnosis and prevention. Low-income families struggle with high treatment costs, often leading to lost wages and job issues due to frequent appointments. Caregivers desperately need age-appropriate guidance and transparent information on diet, medication and treatment to manage the complexities of thalassaemia. Beyond practical difficulties, emotional and moral support from family, friends and communities is crucial to their mental health and resilience.

Conversely, those lacking such support feel isolated and demoralised. To improve the lives of these families, public awareness campaigns, premarital screening programmes, comprehensive caregiver support with clear guidance, financial aid initiatives and fostering supportive environments are crucial. These targeted interventions can significantly alleviate the burdens on families and improve the well-being of individuals affected by thalassaemia major.

The healthcare team's pivotal role in supporting parents of children with thalassaemia is evident from previous research emphasising the need for qualitative studies in this demographic.[26] The experiences of these parents, rooted in cultural and community contexts, significantly shape their needs and the required support. However, systemic challenges within the healthcare system, like drug shortages and limited capacities in public facilities, intensify the obstacles parents face. These hurdles encompass difficulties in enrolling children at thalassaemia centres, blood scarcity, expensive medications, financial constraints and compromised educational prospects for children with thalassaemia.

Parents face substantial financial burdens, relying on welfare and covering out-of-pocket expenses for hospitalisation and consultations, impacting their ability to provide adequate care. Despite this, they prioritise improved educational outcomes for their children with thalassaemia. Addressing these concerns requires a comprehensive approach that addresses healthcare system challenges and children's educational needs. Policy-makers could consider providing financial support through health insurance to alleviate medical expenses and ensure access to necessary services. Establishing public sector thalassaemia care facilities at tertiary levels would improve specialised care accessibility, reducing travel and expenses. Engaging healthcare teams to offer emotional support and address parental worries is vital, supported by insights from qualitative studies. The healthcare team's role is crucial in assisting parents dealing with thalassaemia,

advocating for resources, and supporting emotional and educational needs.

Strength and limitations

This interpretative phenomenology study provided unique insights into the challenges faced by caregivers of children with thalassaemia, offering recommendations that could inform policy-making regarding crucial interventions. While the study's participants openly contributed, offering valuable perspectives, it primarily focused on caregivers, neglecting input from healthcare providers and policy-makers. Limited resources and time constraints confined the study to two thalassaemia centres in Karachi, omitting other cities in Sindh province. There is a need to broaden research across diverse regions, including those with varying healthcare resources, and conduct studies in regional languages.

Recommendations

Caregivers' experiences shed light on vital recommendations across different levels. At an individual level, antenatal checkups must prioritise screening expectant mothers for the carrier state, addressing their primary concern. On a community scale, promoting awareness and preventing thalassaemia through mandatory screenings during university/college admissions is crucial, alongside campaigns highlighting the advantages of blood donation among youth via public service messages. Health promotion through advocacy and parents' education about the misconceptions of Chronic Villus Screening and abortions will reduce the burden of this disease. Sessions are needed to improve parents' mental health and empower them economically through occupational training and financial support programmes. At an organisational level, establishing a collaborative public–private tertiary care centre in Karachi would streamline treatment for parents seeking comprehensive care. Additionally, government subsidies for medicines, particularly iron chelators, are essential to alleviate financial burdens on caregivers.

Policy implications

The study's findings highlight policy implications at multiple levels. Increasing awareness by mandating carrier-state screenings for youth and students on college or university admission is crucial at individual level. Newly diagnosed carriers should be guided towards premarital testing and family screening, leading to the creation of a thalassaemia registry. At the community level, enforcing the compulsory registration of marriages in the national database, alongside premarital screenings, will strengthen existing laws. The health department should develop strategies to promote awareness, highlight benefits and encourage youth participation in blood donations.

Organisationally, integrating thalassaemia screening into antenatal care in both public and private hospitals is essential. Mothers should be screened for the carrier state, and if confirmed, their spouses should also be tested to provide necessary treatment and reduce the risk of transmitting the disease to the child. Early couple counselling and a database of newly detected carrier families will aid prevention. Additionally, implementing the Sindh Thalassemia programme is vital for comprehensive thalassaemia prevention and care within the province.

CONCLUSION

Parents in the study demonstrated a lack of understanding about thalassaemia's inheritance, emphasising the significance of premarital screening for future prevention. NGO-based centres are the primary healthcare providers for children with thalassaemia in Karachi due to the absence of public care facilities, facing challenges like complications, blood shortages and high expenses. Financial strains include costly treatments and transport. Awareness about blood donation is crucial. Caregivers' experiences underscore various recommendations: ensuring maternal screening during antenatal checkups, creating awareness in universities and promoting blood donation among youth through public campaigns. A collaborative public–private tertiary care centre in Karachi and government subsidies for medicines can streamline treatment. Policy suggestions advocate for compulsory marriage registration with premarital screening and the imperative Sindh thalassaemia programme for prevention and care.

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Acknowledgements This master's thesis, authored by MB, is dedicated to expressing our deep appreciation to MA and all the coauthors for their invaluable guidance and support throughout this research. We are also grateful to all the participants who generously contributed their time and insights, which played a crucial role in the success of this study.

Contributors MB was the principal investigator (PI) who conceptualised the study and was involved in study design. MB and MA designed the study protocol and obtained ERC approval. MB and MA were involved in formulating the study methodology, developing study tools and finalising the data analysis plan. JMA and NP were involved in article writing. MB and MA conducted a review of the literature and were involved in reviewing and editing the study tools. JMA and SA were involved in editing manuscripts. All authors reviewed the manuscript several times and provided critical feedback. The guarantor MB accepts full responsibility for the work and/or the conduct of the study, had access to the data, and controlled the decision to publish. All authors read and approved the final version of the manuscript.

Funding The authors have not declared a specific grant for this research from any funding agency in the public, commercial or not-for-profit sectors.

Competing interests None declared.

Patient and public involvement Patients and/or the public were not involved in the design, or conduct, or reporting, or dissemination plans of this research.

Patient consent for publication Informed consent was obtained from the study participants for research publication (in any journal).

Ethics approval This study received ethical approval from the Aga Khan University (ERC # 2022-7234-2162). All procedures were conducted in accordance with the ethical standards of the Declaration of Helsinki. Participants gave informed consent to participate in the study before taking part.

Provenance and peer review Not commissioned; externally peer reviewed.

Data availability statement All data relevant to the study are included in the article or uploaded as supplementary information.

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