Research Square

Preprints are preliminary reports that have not undergone peer review.
They should not be considered conclusive, used to inform clinical practice,
or referenced by the media as validated information.

Living with beta-thalassaemia major: stigma, emotional avoidance and relationships to care. A qualitative study among adult patients in France.

Emilie COSTE
Université Grenoble Alpes: Universite Grenoble Alpes
Mélanie ASTIE
CHU Grenoble Alpes: Centre Hospitalier Universitaire Grenoble Alpes
Caroline MAKOWSKI
CHU Grenoble Alpes: Centre Hospitalier Universitaire Grenoble Alpes
Damien Oudin Doglioni
damien.oudin-doglioni@univ-grenoble-alpes.fr

Universite Grenoble Alpes https://orcid.org/0000-0003-3624-7251

Research Article

Keywords: Transfuso-dependent beta thalassaemia, stigma, trauma, treatment adherence, patient – provider relationship, coping strategies

Posted Date: April 27th, 2026

DOI: https://doi.org/10.21203/rs.3.rs-8810004/v1

License: This work is licensed under a Creative Commons Attribution 4.0 International License.
Read Full License

Abstract

Background

Transfusion-dependent β-thalassaemia (β-TDT) is a rare inherited disorder requiring lifelong red blood cell transfusions and iron chelation. While medical advances have improved survival, the treatment remains invasive and time-consuming. Patients face significant psychosocial burdens, including anxiety, depressive symptoms, and social stigma. Current literature suggests a reliance on emotion-focused coping, which may hinder adherence. However, qualitative data exploring the subjective experience of adult β-TDT patients remain scarce in French-speaking contexts.

Objectives

This study aimed to (i) explore the lived experience of adults with β-TDT in France, focusing on identity, stigma, and coping strategies, and (ii) examine how patients negotiate their relationship with long-term care to identify clinical levers for improving adherence and psychological support.

Methods

We conducted an exploratory qualitative study at Grenoble Alpes University Hospital (CHUGA). Seven adult patients (aged 30–68 years) were purposively recruited. Data were collected via individual semi-structured interviews (36–95 minutes), conducted in person or via secure videoconference. Analysis followed a thematic approach inspired by constructivist grounded theory, involving iterative coding and constant comparison to ensure reflexivity and trustworthiness. The study adhered to GDPR and ethical standards (Declaration of Helsinki).

Results

Two overarching themes emerged. (i) Fighting against stigma: Participants reported a long-standing sense of bodily difference and internalised shame. Normalisation strategies – such as hiding the disease or skipping treatments to ‘live like others’ – were used to preserve identity but often compromised self-care. Family secrecy and cultural norms regarding blood further reinforced feelings of disgrace. (ii) An ambivalent relationship to care: Early experiences of invasive procedures were often perceived as objectifying or traumatic. The hospital environment triggered anticipatory anxiety and hypervigilance. However, patients also recognised treatments as lifesaving, expressing a desire for shared decision-making and the recognition of their ‘experiential knowledge’.

Conclusions

Adults with β-TDT oscillate between a quest for normality and the chronic intrusiveness of treatment. The narratives suggest a potential traumatic impact of repeated medical procedures, echoing literature linking PTSD symptoms to poorer adherence. Clinical implications include adopting trauma-informed care in haematology, systematic screening for trauma-related symptoms, and utilising the Common-Sense Model to address illness representations. Valuing experiential knowledge through collaborative therapeutic education is essential for enhancing adaptive coping and long-term adherence.

1 Background

β-thalassaemia major (β-TDT) is a haemoglobinopathy of global prevalence (1), predominantly affecting populations originating from the Mediterranean Basin, Africa, and Southeast Asia (2). It is estimated that approximately 60,000 new cases occur annually (3). In France, the prevalence is estimated at 1/100,000, representing nearly 800 patients (4).

As a genetic disorder with autosomal recessive inheritance, β-TDT is characterised by a quantitative deficit in β-globin chain synthesis (2, 5). The resulting imbalance between α and β chains leads to ineffective erythropoiesis and chronic haemolysis (6). This pathophysiology culminates in severe anaemia during early infancy, necessitating regular – and typically lifelong – red blood cell transfusions (7). Concurrently, iron homoeostasis is disrupted by both increased gastrointestinal absorption and repeated transfusion intake (8). If not effectively managed, excess circulating free iron and subsequent tissue sequestration (secondary haemochromatosis) result in cellular damage and multi-organ dysfunction (6, 8).

Initial clinical manifestations typically emerge at approximately six months of age, including febrile episodes, pallor, irritability, jaundice, failure to thrive, hepatosplenomegaly and, in the absence of transfusion therapy, craniofacial and long-bone deformities (7). Initiating a transfusion protocol before the age of two is imperative; without regular intervention, life expectancy is limited to approximately fifteen years (7). Iron overload preferentially affects the liver, myocardium, and endocrine glands, leading to hormonal insufficiencies (hypogonadism, thyroid disorders, diabetes, adrenal insufficiency) and organic sequelae (cardiac, renal, and hepatic complications) (2, 5). Myocardial involvement remains the primary cause of mortality (9). Furthermore, hormone replacement therapy is frequently required to facilitate puberty (10). An increased risk of thromboembolic events (10), cerebrovascular complications, and malignancies (11) has also been documented in β-TDT patients.

Management relies on the dual regimen of regular transfusions and iron-chelating agents, which are indispensable for controlling iron overload (10). Regarding curative interventions, haematopoietic stem cell transplantation remains a therapeutic option for specific paediatric cases (10). Gene therapy has been under clinical exploration since 2007 (12), and several molecules aimed at correcting globin chain imbalance are currently in use or under development (7). These advancements, alongside a more nuanced understanding of erythropoiesis and progress in diagnostic imaging and chelation protocols, have significantly extended life expectancy – now exceeding 50 years (7). However, precise longitudinal

data for the youngest generations, who benefit from early access to modern therapies, are yet to be established. Nevertheless, the burdensome and chronic nature of these treatments renders therapeutic adherence a critical challenge (4).

β-TDT profoundly impacts the biopsychosocial quality of life (QoL) of affected individuals (13). Patients frequently cite the intrusiveness of care, the disease's dominance over daily life (13), and the ramifications of endocrine damage, particularly regarding pubertal development (14). Numerous studies indicate an increased vulnerability to depression, anxiety (15), body image disturbances, impaired self-esteem, emotional dysregulation, and social functioning limitations (16, 17). Chronic pain, frequently reported, further exerts a deleterious effect on mental health (18). Moreover, several works highlight the subjective experience of disability, stigmatisation, and discrimination stemming from physical limitations, the sense of 'otherness', and experiences of social rejection or systemic misunderstanding (19–21).

As with any severe chronic condition, living with β-TDT necessitates the implementation of adaptive mechanisms. However, in this population, certain strategies prove maladaptive. Processes of normalisation are frequently observed, whereby patients seek to maintain a positive identity by conforming to societal norms (22). Their efficacy remains relative, contingent upon life events and the reactions of the social environment (20). The drive to 'lead a normal life' may manifest as the minimisation of the illness or treatment non-adherence, with potential risks to clinical outcomes (20). Furthermore, emotion-focused and avoidance-oriented coping strategies, identified as predominantly in β-TDT patients, are associated with heightened somatic and psychological distress (16, 23, 24). Specifically, Abbasi et al. (23) demonstrate that a reliance on emotion-centred responses (rumination, withdrawal, guilt) correlates with a deterioration in overall QoL and physical health, whereas problem-solving strategies are associated with superior outcomes. Similarly, in young adults, Messina et al. (16) highlight a predominance of escape-avoidance coping alongside a high frequency of somatisation, depression, and anxiety. Finally, Parviniannasab et al. (24) show that defensive strategies (emotional and evasive) are linked to lower resilience, suggesting fragile psychological adaptation to somatic constraints. The emotional processing of stressful experiences, which facilitates a return to baseline functioning (25), often appears incomplete (26).

Consequently, the potentially traumatic nature of β-TDT must be considered. Firstly, the chronic illness itself possesses traumatogenic potential (27): the cumulative burden of adversity arising from successive medical challenges (28) can generate stress levels consistent with post-traumatic stress disorder (PTSD) (27). Secondly, avoidant coping and incomplete emotional processing are established risk factors for PTSD (29–31). The quest for normality, which may lead to the delay or interruption of treatment, further exposes patients to life-threatening medical crises: without regular transfusions, β-TDT remains almost universally fatal before adolescence (7); with transfusion but without chelation, historical cohorts show a median mortality at 17 years, primarily due to iron overload complications (32). Ultimately, the mastery of iron overload (hepatic and myocardial) is the primary determinant of longevity (8). Thus, dependence on blood supplies and chelators confronts patients with the traumatogenic reality

of treatment availability, where social or public health crises may be perceived as direct existential threats.

International research regarding the psychological functioning of individuals with β-TDT remains sparse and, to our knowledge, French data are currently non-existent. In this context, we have chosen to employ a qualitative methodology to explore the lived experiences and representations that patients maintain regarding their illness and therapeutic regimens.

2 Methods

This study employed an exploratory, cross-sectional qualitative design. The research was conducted at Grenoble Alpes University Hospital (CHUGA) within the framework of the Rare Disease Competence Centre for Haemoglobinopathies (Fil Rouge — MCGRE). The study design and reporting were guided by the Consolidated Criteria for Reporting Qualitative Research (COREQ) checklist to ensure transparency and methodological rigour.

The Clinical Research Directorate of the CHUGA approved this non-interventional study (n°VST3 — 7 February 2025). All procedures were performed in accordance with French national regulations (we followed the MR03 reference methodology of the French Data Protection Authority), the Declaration of Helsinki (33), and the European General Data Protection Regulation (GDPR). Prior to the interviews, each participant received comprehensive oral and written information regarding the research objectives, the voluntary nature of participation, data anonymisation, and the right to withdraw at any time. Informed consent was obtained for both participation and the use of anonymised verbatim excerpts in scientific publications.

2.1 Participants and Inclusion Criteria

Eighteen adult patients residing in the Isère department and receiving care at the CHU in Grenoble or Voiron for transfusion-dependent thalassaemia were identified as potentially eligible. Ultimately, seven patients agreed to participate and were included in the study. While the sample size (n = 7) reflects the rarity of the condition within the regional catchment area, the recruitment followed the principle of ‘information power’ (34). The depth of the semi-structured interviews, the high specificity of the patient experiences, and the application of a robust theoretical framework ensured sufficient internal validity to explore the core phenomena.

The inclusion criteria were: (1) aged ≥ 18 years; (2) diagnosis of transfusion-dependent thalassaemia; (3) currently receiving iron chelation therapy; (4) proficient in the French language; and (5) capable of providing free and informed consent.

2.2 Data Collection

Patients were recruited via written flyers distributed in the day hospital and through email correspondence from the medical team. Interested individuals contacted the investigator (a

psychologist in training) directly to schedule an interview based on their availability.

Three interview modalities were offered to ensure participant comfort:

- In-person: in a dedicated, confidential room at the day hospital, typically during a transfusion session.
- In-person: within a private hospital room.
- Remote: via a secure videoconferencing platform (GCS Sara: the Auvergne-Rhône-Alpes teleconsultation platform, CHUGA site).

The individual, semi-structured interviews followed a thematic guide exploring: (i) somatic and emotional experiences of the disease since childhood; (ii) educational, professional, and familial trajectories; (iii) social relationships and perceived stigmatisation; (iv) representations of the illness, treatments, and biological markers; (v) relationships with the healthcare system and providers; and (vi) adjustment strategies and expectations regarding psychological support.

Interviews lasted between 36 minutes and 1 hour 35 minutes. They were audio-recorded and transcribed verbatim using the Tadddam software (35), developed at the University of Grenoble Alpes (UGA). In compliance with GDPR, the software performed automatic speaker recognition and initial anonymisation. Transcripts were subsequently reviewed, corrected, and fully anonymised by removing all potentially identifying information.

A reflexive stance was maintained throughout the data collection process. The primary interviewer, a psychologist in training, utilised her clinical background to foster a safe and empathetic environment, facilitating the disclosure of sensitive emotional material. To mitigate potential bias, the interviewer maintained a research diary to document personal impressions and ensure a distinction between clinical support and data collection.

2.3 Data Analysis

Data were analysed using a constructivist-informed grounded theory approach, as described by Glaser and Strauss (35), focusing on the subjective meanings constructed by the participants. The analysis followed several iterative stages:

- Immersion: preliminary reading of verbatim transcripts to achieve data familiarity.
- Open coding: line-by-line segmentation of transcripts and assignment of descriptive codes (e.g. ‘body shame’, ‘pervasive fatigue’, ‘fear of blood shortage’, ‘desire for normality’).
- Constant Comparison: ongoing comparison between codes and across interviews to group-related codes into broader categories.
- Thematic Development: construction of central themes and subthemes by articulating the relationships between categories, with frequent returns to the source text to ensure validity.

- Theoretical Formulation: Integration of the themes with established health psychology models (illness representations, coping, stigmatisation, and trauma).

Data collection and analysis were conducted iteratively. Preliminary thematic saturation was achieved when the final interviews yielded redundant conceptual information, suggesting that the core thematic structure was robust enough to address the research objectives.

The analysis was conducted by the primary investigator under the supervision of the principal investigator and in collaboration with the CHUGA clinical teams, allowing for the triangulation of interpretations. To ensure the dependability and credibility of the findings, a process of peer debriefing was employed. The coding frame and emerging themes were cross-examined by the supervising researcher and the clinical team to resolve any interpretative discrepancies and to enhance the confirmability of the final thematic map. The findings are presented below according to the two primary thematic categories identified.

3 Results

3.1 Sociodemographic Characteristics

The seven participants had a mean age of 53 years (range: 30–68 years), were predominantly female (n = 5/7), and all resided in the Isère department. Six patients lived with β-TDT major, while one had β-thalassaemia intermedia that had become transfusion-dependent. Marital and family situations varied significantly (single without children, cohabiting with or without children, and single parents). Four participants were professionally active (salaried or self-employed), and three were unemployed at the time of the study, reflecting a diversity of life trajectories.

3.2 Theme 1: Patients Struggling Against Stigmatisation

This primary theme encompasses discourse related to bodily difference, shame, social exclusion, and the strategies employed to maintain an acceptable identity.

3.2.1 Identity Issues and Self-Perception

All participants reported, with varying intensity, a sense of having been ‘different’ since childhood. The participants’ narratives suggest a ‘biographical disruption’ (37), where the illness is not merely a medical condition but an ontological assault on the self. The ‘stigma of difference’ was often crystallised through physical markers (e.g. short stature, splenectomy scars), which functioned as ‘discrediting’ attributes in social interactions. This resulted in a persistent tension between the actual social identity (the sick self) and the virtual social identity (the desired normal self), forcing participants into a state of constant identity negotiation. These elements often fuelled a sense of vulnerability and a perceived assault on femininity or masculinity: ‘I knew I wasn’t quite like the others… I didn’t have the same strength…’

Several narratives detailed explicit experiences of rejection or stigmatisation at school, including bullying, peer exclusion, and disparaging remarks from teachers. Some participants spontaneously utilised the term ‘disability’ to characterise their limitations and care dependency, often accompanied by a sense of debt or guilt towards their families. Stigmatisation was also rooted in cultural and familial representations of blood and illness. For one participant from an Italian village, transfusions remained a long-standing family secret, associated with shame and the notion of being ‘a pariah’. Such experiences facilitated the internalisation of stigma and self-deprecating thoughts, echoing findings from other cultural contexts.

However, identity was not solely defined by fragility. Several participants described a journey toward illness acceptance, often through association work, participating in blood drives, or supporting other patients – activities that became significant sources of pride and meaning.

3.2.2 Potentially Traumatic Somatic Consequences

Chronic fatigue, the cardinal symptom of β-TDT, was described as pervasive – limiting leisure, employment, and parenting – and often leading to social disengagement. Repeated hospitalisations, complications (infections, surgical interventions), and certain painful care experiences were recounted in emotionally charged terms, occasionally explicitly described as ‘traumatic’.

Beyond mere dissatisfaction, the narratives revealed elements of iatrogenic suffering. Participants described early paediatric care through a prism of ‘medical trauma’, characterised by a perceived loss of bodily integrity and agency. The use of terms such as ‘mistreatment’ reflects a deep-seated sense of objectification, where the clinical necessity of the procedure overshadowed the psychological safety of the child. This cumulative adversity appears to underpin current patterns of medical avoidance and anticipatory anxiety observed in adulthood.

In some cases, elements of re-experiencing and physiological reactivity (hypervigilance, bodily tension, and avoidance of hospitals or examinations) were detectable in the narratives, although no formal PTSD diagnosis was established within the scope of this study.

3.2.3 Normalisation Mechanisms and Social Dynamics

Participants expressed a profound desire to ‘live like everyone else’ and to avoid reducing their existence to their pathology. This manifested in normalisation strategies: minimising symptoms, refusing academic or professional accommodations, socially camouflaging the illness, and selectively disclosing their condition. These strategies align with the findings of Atkin and Ahmad (20) regarding young people with thalassaemia major, highlighting the symbolic importance of ‘normality’.

In the present study, the desire to protect others (spouses, children, and friends) from the burden of the disease occasionally led to voluntary isolation, which was subsequently experienced as painful loneliness. Conversely, several patients reported a progressive ‘thinning’ of their social circles, prioritising ‘authentic’ bonds with individuals capable of non-judgmental acceptance.

Finally, some verbatim excerpts suggested that the effort to maintain a ‘normal’ role (at work or within the family) sometimes occurred to the detriment of health – for instance, delaying a transfusion to avoid a work absence or temporarily interrupting chelation during holidays – concurring with observations from other qualitative studies.

3.3 Theme 2: An Ambivalent Relationship With Healthcare

The second theme addresses the complexity of the bond with the healthcare system and therapeutic regimens: an interplay between gratitude and rejection, trust and suspicion, and vital dependency versus a desire for autonomy.

3.3.1 Instrumental and Objectifying Relationships with Providers

Many participants retrospectively described childhood care experiences where they felt like ‘objects’ of technical procedures rather than subjects of care. Repeated punctures, prolonged infusions (e.g. nocturnal subcutaneous deferoxamine cycles), a lack of age-appropriate explanations, and inadequate pain management continue to fuel critical discourse, occasionally marked by the use of the term ‘mistreatment’.

This experience of objectification – where the body becomes a ‘thing of care’ (38) – was described as psychologically taxing, particularly when not mitigated by the recognition of the patient’s subjectivity. For several respondents, the mere prospect of a hospital visit triggers significant anxiety, agitation, or insomnia, even when current relationships with adult specialist teams are described as satisfactory.

3.3.2 Burdensome, Intrusive, and Anxiety-Inducing Treatment

Treatments were unanimously perceived as burdensome and intrusive, dictating the structure of daily life: the rhythm of transfusions, systematic pre-transfusion biological monitoring, daily chelation, and side-effect management. Participants used metaphors like ‘having a ball and chain’, from which they found it difficult to imagine being liberated.

Beyond the practical burden, patients expressed significant concerns about the future. Some feared that treatments might no longer be reimbursed or that blood availability could be compromised by health or geopolitical crises. These concerns echo documented issues in the literature regarding the security and availability of blood product supplies.

3.3.3 Coherence, Control, and Experiential Knowledge

Participants manifested a strong need for comprehension and coherence regarding biological data (haemoglobin, ferritin levels) and therapeutic goals. Some developed personal interpretations that

diverged from the biomedical model (e.g. an excessive focus on a specific ferritin threshold, perceived as inherently alarming).

Viewed through Leventhal's Common-Sense Model (39), these results represent illness and treatment representations structured around cognitive (perceived causes, consequences, controllability, duration) and emotional (fear, anger, shame) dimensions. When global coherence is lacking or medical information is insufficiently linked to lived experience, some patients resort to avoidance (not asking questions, not checking results, missing appointments) or forms of catastrophising that sustain anxiety.

Simultaneously, all participants expressed a desire for greater agency: deciding on transfusion schedules, adjusting treatment modalities, and co-constructing priorities with providers. They asserted the legitimacy of their experiential knowledge, demanding that it be recognised alongside biomedical expertise.

4 Discussion

This qualitative study, involving seven adults living with transfusion-dependent β-thalassaemia (β-TDT) in France, highlights two central dynamics. First, a continuous struggle against stigmatisation and an ongoing effort to preserve an acceptable identity, which may lead to normalisation strategies that carry significant health costs. Second, an ambivalent relationship with the healthcare system, characterised by the tension between vital dependency on transfusions and chelation, and the rejection of practices perceived as objectifying or traumatic. These dynamics unfold within the context of a life-threatening chronic illness beginning in infancy, necessitating successive identity adjustments and confronting patients with enduring uncertainty regarding the future.

While many psychosocial studies in β-TDT originate from regions with high prevalence (e.g. the Middle East or South America), our study provides unique insights into a highly centralised European healthcare system. In France, the structure of the 'Centres de Référence et de Compétence' (such as the MCGRE network) offers a paradox: while it ensures high-quality biological standardisation, our results suggest it may inadvertently reinforce the 'institutionalisation' of the patient's life. This underscores the need for these expert centres to integrate dedicated psychological and social support pathways that match their haematological excellence.

4.1 Identity, Stigma, and the Quest for Normality

Our findings corroborate several observations from international research within the French context. Regarding identity, narratives of bodily difference, pubertal delay, school bullying, and professional integration challenges align with numerous studies describing the impact of β-TDT on self-image and social participation (15). The burden of stigma – whether external or internalised – appears as a major determinant of psychological distress.

The desire for ‘normality’ observed in our interviews echoes the concept of normalisation described by Atkin and Ahmad (20), where individuals attempt to maintain an ‘ordinary’ daily life by comparing themselves to seemingly healthy peers. This can lead to the rejection of treatments perceived as too visible or stigmatising. Similarly, qualitative studies in Brazil and Iran demonstrate how adults with thalassaemia constantly negotiate the balance between therapeutic constraints, the desire for autonomy, and quality of life (21, 40).

Our findings extend the sociological understanding of ‘identity work’ in rare diseases. The participants’ experiences of physical difference (short stature, scars) align with Goffman’s theory of the ‘discredited’ identity, where the individual’s stigma is visible and must constantly be managed in social interactions (43). The normalisation strategies identified – such as the conscious concealment of treatment – represent a significant ‘biographical labour’ aimed at maintaining a ‘discreditable’ rather than ‘discredited’ status. This highlights that for β-TDT patients, treatment adherence is not merely a biological necessity but a social risk that must be negotiated daily.

4.2 Coping Strategies and Psychological Adjustment

In terms of adjustment strategies, our results are consistent with the systematic review by Ahmadian et al. (41), which identifies a predominance of emotion-focused and avoidance-oriented coping in thalassaemia patients, linked to increased psychological distress. Several verbatim excerpts illustrate forms of avoidance (delaying care, minimising symptoms, social withdrawal) and catastrophising (rumination, fearful anticipation), which may conflict with the requirements of rigorous clinical adherence.

4.3 The Healthcare Relationship and Traumatogenic Potential

Narratives of early invasive procedures, inadequate pain management, and a sense of being objectified illuminate the traumatogenic potential of care pathways in β-TDT. Multidisciplinary management across various specialities may hinder the integration of the disease into a coherent unit, increasing the risk of perceiving β-TDT as a collection of disjointed complications rather than a global pathology (39). These observations align with research suggesting that chronic illnesses requiring repeated, painful interventions from childhood can constitute cumulative traumatic experiences, carrying a risk of PTSD symptoms. The meta-analysis by Taggart-Wasson et al. (42) further emphasises that PTSD is associated with increased treatment non-adherence across several chronic conditions, a correlation that likely extends to β-TDT.

A significant contribution of this study is the characterisation of early paediatric care as a potential source of cumulative medical trauma. The narratives suggest that the repetitive, invasive nature of transfusions and chelation, when experienced in an objectifying clinical environment, may lead to ‘iatrogenic psychological suffering’. This trauma is not merely a side effect of the illness but a product of the healthcare encounter itself. By framing these experiences through the lens of trauma-informed care,

we argue that haematology departments must move beyond technical proficiency to recognise the ‘relational safety’ of the patient as a primary clinical outcome.

Furthermore, the participants’ desire to understand biological markers and participate in decision-making aligns with contemporary models of care that insist on the recognition of experiential knowledge and the co-construction of treatment plans. The illness representations observed (e.g. fatalistic beliefs or specific focus on ferritin thresholds) fit within the dimensions of Leventhal’s Common-Sense Model (37) and help explain specific health behaviours. The observed focus on specific biological markers, such as ferritin thresholds, illustrates a ‘cognitive-emotional dissonance’ within the Common-Sense Model. While clinicians view ferritin as a longitudinal safety indicator, patients often interpret it as an immediate emotional barometer of ‘failure’ or ‘danger’. This misalignment suggests that catastrophising occurs when the biomedical data lacks ‘lay coherence’. Clinical interventions should, therefore, focus on ‘illness perception mapping’ – explicitly discussing how a patient’s internal model of their blood and iron levels influences their willingness to maintain a rigorous chelation regimen.

4.4 Clinical Implications

Several clinical implications can be drawn for haematology and liaison psychiatry/psychology:

- Trauma-Informed Care: Integrate a trauma-informed perspective into thalassaemia services. This includes systematic screening for anxiety, depression, and post-traumatic symptoms (e.g. via standardised questionnaires) and sensitising staff to the psychological effects of objectification and repeated invasive procedures. Priority should be given to pain management, informed consent, and the promotion of patient self-efficacy and empowerment.
- Addressing Illness Representations: Utilise the Common-Sense Model (39) to explore patient beliefs regarding the causes, progression, consequences, and controllability of β-TDT. Co-elaborating explanations of biological markers (e.g. ferritin) that bridge medical knowledge and lived experience can reduce catastrophising and misunderstandings that hinder adherence.
- Supporting Adaptive Coping: Proposed psychotherapeutic interventions focused on emotional regulation, tolerance of uncertainty, and the reduction of avoidance (e.g. Cognitive Behavioural Therapy, group therapy, or therapeutic education), building upon recent work on emotional intelligence training in β-TDT adolescents (44).
- Valuing Experiential Knowledge: Involve patients and patient associations in the design of therapeutic education programmes and care pathways. Creating ‘reflective spaces’ (groups or workshops) allows for the sharing of experiences regarding stigma, reinforcing a sense of belonging and legitimacy.

4.5 Limitations and Conclusion

This study has several limitations. The small sample size (n = 7) and recruitment from a single centre limit the generalisability of the findings. Participants may also represent a more engaged or expressive

subset of the patient population. Furthermore, the lack of systematic clinical psychiatric data prevents a direct diagnostic link between the traumatic elements in the narratives and formal PTSD.

Despite these limitations, this study provides original insights into the subjective lived experience of adult β-TDT in France, specifically regarding stigmatisation, avoidance strategies, and the healthcare relationship. Further qualitative and mixed-method research involving larger, more diverse samples is warranted to explore the impact of cultural origin and socio-economic status. Interventional studies are also necessary to evaluate the efficacy of psychotherapeutic and educational approaches that explicitly integrate these psychosocial dimensions.

5 Conclusion

The findings derived from this qualitative exploration of seven adults living with transfusion-dependent β-thalassaemia highlight the profound psychological labour required to navigate the exigencies of a life-threatening rare disorder and its associated lifelong treatment regimens. A pervasive tension exists throughout these life trajectories, defined by the pursuit of 'normative' biographical goals, the persistent threat of social stigmatisation, and an absolute dependency on high-stakes clinical interventions.

Beyond conventional anxio-depressive comorbidities, the narratives underscore the traumatogenic potential of both the condition and its invasive care pathways. β-TDT must therefore be understood as a significant biographical disruption (37) that leaves a lasting imprint on identity, self-efficacy, and the internalised relationship with the body.

Moving forward, it is imperative that clinical management transcends a purely biomedical focus. Integrating a trauma-informed, medico-psychological framework – one that explicitly addresses the nuances of stigmatisation, emotional avoidance, the need for agency, and the legitimacy of experiential knowledge – is an essential prerequisite. Such a holistic approach is vital not only for improving therapeutic adherence but also for fostering the long-term quality of life and social participation of individuals living with β-TDT.

Declarations

Ethic Approval

The Clinical Research Directorate of the teaching hospital Grenoble Alpes (CHUGA) approved this non-interventional study (n°VST3 – 7 February 2025).

Authors' Contributions.

DOD and CM developed this qualitative research. DOD and EC defined the methodology. DOD and EC defined the qualitative analysis strategy and coordinated the qualitative analyses. DOD, EC, CM, and MA

participated in the qualitative analysis. DOD and EC wrote the first draft of this article. All authors validated the final version of the article.

Declaration of conflict of interest.

All authors declare that they have no conflicts of interest in relation to the conduct of this research that could have influenced the interpretation of the results.

Access to data.

The data may be accessed upon reasoned request to Damien OUDIN (damien.oudin-doglioni@univ-grenoble-alpes.fr).

Acknowledgement.

The authors would like to express their sincere gratitude to the patients who agreed to share their experiences, as well as to the team at the CHUGA for their support in conducting this study.

The authors would like to thank the Grenoble Alpes University Data Platform (PUD-GA) and the PACTE laboratory, with the support of GRICAD and UGA, for providing access to the Tadddam programme (Transformations, Analyses and Developments of Multimedia Data and Archives), which is a platform for automatic transcription and analysis of multimedia data and archives.

References

1. Piel FB, Weatherall DJ. The α-Thalassemias. N Engl J Med. 2014;371(20):1908–16.
2. Muncie HL, Campbell J. Alpha and beta thalassemia. Am Fam Physician. 2009;80(4):339–44.
3. Frangoul H, et al. CRISPR-Cas9 Gene Editing for Sickle Cell Disease and β-Thalassemia. N Engl J Med. 2021;384(3):252–60.
4. Bonello-Palot N, et al. Les thalassémies en 2016. Rev Francophone des Lab. 2016;2016(481):67–75.
5. Rund D, Rachmilewitz E. β-Thalassemia. N Engl J Med. 2005;353(11):1135–46.
6. Kattamis A, et al. The effects of erythropoetic activity and iron burden on hepcidin expression in patients with thalassemia major. Haematologica. 2006;91(6):809–12.
7. Kattamis A, et al. Thalassaemia Lancet. 2022;399(10343):2310–24.
8. Rivella S. Iron metabolism under conditions of ineffective erythropoiesis in β-thalassemia. Blood. 2019;133(1):51–8.
9. Modell B, et al. Improved survival of thalassaemia major in the UK and relation to T2* cardiovascular magnetic resonance. J Cardiovasc Magn Reson. 2008;10(1):42.

10. Galanello R, Origa R. Beta-thalassemia. Orphanet J Rare Dis. 2010;5(1):11.
11. Farmakis D, et al. The changing epidemiology of the ageing thalassaemia populations. Eur J Haematol. 2020;105(1):16–23.
12. Cavazzana-Calvo M, et al. Transfusion independence and HMGA2 activation after gene therapy of human β-thalassaemia. Nature. 2010;467(7313):318–22.
13. Arian M, et al. Health-related quality of life in beta-thalassemia major patients: a meta-analysis. Qual Life Res. 2019;28(2):321–34.
14. Mattia L, et al. The Quality of Life of Thalassemic Patients: The Role of Endocrine Defect Compensation. EMIDDT. 2021;21(12):2147–58.
15. Tarım HŞ, Öz F. Thalassemia Major and Associated Psychosocial Problems: A Narrative Review. Iran J Public Health. 2022;51(1):12–8.
16. Messina G, et al. Psychosocial aspects and psychiatric disorders in young adult with thalassemia major. Intern Emerg Med. 2008;3(4):339–43.
17. Khurana A, et al. Psychosocial burden in thalassemia. Indian J Pediatr. 2006;73(10):877–80.
18. Oliveros O, et al. Pain over time and its effects on life in thalassemia. Am J Hematol. 2013;88(11):939–43.
19. Barouni M et al. Influence des hémoglobinopathies sur l'aspect psychosocial des patients atteints. Rev Mar Sante Publique. 2018;5(8).
20. Atkin K, Ahmad WIU. Living a 'normal' life: young people coping with thalassaemia major or sickle cell disorder. Soc Sci Med. 2001;53(5):615–26.
21. Ganzella M, Zago MMF. The experience of thalassemic adults with their treatment. Rev Latino-Am Enfermagem. 2011;19(4):968–76.
22. Piga A, et al. Luspatercept improves hemoglobin levels and blood transfusion requirements in patients with β-thalassemia. Blood. 2019;133(12):1279–89.
23. Abbasi S, et al. Quality of Life and Coping Style in Adolescents with Thalassemia. Int J Hematol Oncol Stem Cell Res. 2020;14(1):19–26.
24. Parviniannasab AM, et al. Coping strategies and resilience among adolescents with beta-thalassemia major. Child Adolesc Psychiatr Nurs. 2021;34(4):329–34.
25. Rachman S. Emotional processing, with special reference to post-traumatic stress disorder. Int Rev Psychiatry. 2001;13(3):164–71.
26. Pourmohammad Fahreh F, Shirazi M. Comparison Quality of Life and Emotional Processing among Patients with Major Thalassemia. Jundishapur J Chronic Dis Care. 2020;9(3).
27. Alonzo AA. The experience of chronic illness and post-traumatic stress disorder. Soc Sci Med. 2000;50(10):1475–84.
28. Turner RJ, Lloyd DA. Lifetime Traumas and Mental Health. J Health Soc Behav. 1995;36(4):360.
29. Littleton H, et al. Trauma coping strategies and psychological distress: A meta-analysis. J Trauma Stress. 2007;20(6):977–88.

30. Ehlers A, Clark DM. A cognitive model of posttraumatic stress disorder. Behav Res Ther. 2000;38(4):319–45.
31. Brewin CR. Memory processes in post-traumatic stress disorder. Int Rev Psychiatry. 2001;13(3):159–63.
32. Coates TD, et al. Management of iron overload in hemoglobinopathies. Ann N Y Acad Sci. 2016;1368(1):95–106.
33. World Medical Association. Declaration of Helsinki. 2024.
34. Malterud K, Siersma VD, Guassora AD. Sample Size in Qualitative Interview Studies: Guided by Information Power. Qual Health Res. 2016;26(13):1753–60.
35. Beligné M, Juen P. TADDDAM. Semaine Data-SHS. 2024.
36. Glaser B, Strauss A. Discovery of Grounded Theory. Routledge; 2017.
37. Bury M. Chronic illness as biographical disruption. Sociol Health Illn. 1982;4(2):167–82.
38. Caillol M. L'objectivation nécessaire dans la pratique soignante. Cancer(s) & psy(s). 2019;4(1):133–42.
39. Leventhal H, et al. The Common-Sense Model of Self-Regulation (CSM). J Behav Med. 2016;39(6):935–46.
40. Zeydi AE, et al. Iranian β-Thalassemia major patients' perception of adherence to treatment. Electron Physician. 2017;9(12):6102–10.
41. Ahmadian B et al. Coping Strategies in Patients with Beta-thalassemia and their Parents: A Systematic Review. Int J Pediatr. 2022;10(3).
42. Taggart Wasson L, et al. PTSD and nonadherence to medications: A meta-analysis. J Psychiatr Res. 2018;102:102–9.
43. Goffman E. Stigma: Notes on the Management of Spoiled Identity. Prentice-Hall; 1963.
44. Ahmadian B, et al. The effect of applying emotional intelligence components on coping strategies in adolescents with β-thalassemia major. BMC Pediatr. 2024;24(1):591.