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REVIEW

Exploring Public Attitudes and Barriers toward Thalassemia Screening Strategies: A Systematic Review of Qualitative Studies

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Abstract

Background: Over the last decade, several countries have sought to mitigate the rise in thalassaemia through preventive screening policies; however, incidence remains high. While existing studies, predominantly quantitative, have examined thalassaemia screening, limited attention has been given to synthesizing qualitative evidence on public attitudes and barriers. Consequently, a comprehensive qualitative synthesis is needed to inform contextually relevant, effective, and equitable screening strategies.
Purpose: This review systematically explores public attitudes and barriers toward thalassemia screening strategies (TSS).
Methods: A literature search was conducted according to PRISMA guidelines, using four electronic databases–Scopus, PubMed, ProQuest, and ScienceDirect–using specific keywords. The study population included the general public or parents of children with thalassaemia or sickle cell disease, and studies that investigated attitudes or barriers toward thalassaemia screening. A thorough screening of abstracts and full texts was conducted based on predetermined criteria. Overall, 11 of 472 studies were selected for qualitative analysis. The quality of abstracts and full papers was reviewed using the CASP tool, and thematic analysis was conducted line by line with assistance from OpenCode 4.03 software.
Results: The findings were mapped according to attitudes and barriers toward TSS. Five main themes emerged from the thematic content analysis: (1) negative attitudes influenced by cultural and personal factors; (2) inadequate family support and decision-making; (3) limited knowledge of hereditary and genetic conditions; (4) low self and parental awareness of thalassaemia; and (5) poor communication with healthcare professionals and barriers to screening accessibility.
Conclusion: Thalassaemia screening barriers are a complex socio-cultural and behavioural issue, especially at crucial life stages such as premarital life. Education on thalassaemia and genetic inheritance should be incorporated into youth programmes prior to marriage. Family-centred therapies, training in nursing and health communication, and proactive counselling to encourage regular screening and integrate thalassaemia screening into primary care are crucial for the future.

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1. Introduction

Research on thalassemia screening strategies for early detection in high-risk populations has emerged as a critical area of inquiry due to the significant global burden of thalassemia and its impact on nursing prevention care. Thalassemia, a group of inherited hemoglobin disorders, affects millions worldwide, with carrier rates ranging from 1% to over 20% in endemic regions such as South Asia, Southeast Asia, and the Mediterranean (Agarwal et al., 2025; Demirbaş, 2024; Goonasekera et al., 2025). Over the past decades, screening programs have evolved from basic hematological tests to advanced molecular diagnostics, reflecting technological progress and increased understanding of disease epidemiology (Goonasekera et al., 2025; Mensah & Sheth,

2021). The social and economic implications of thalassemia are profound, with annual births of affected children estimated in the tens of thousands in high-prevalence countries, underscoring the need for effective prevention strategies (Akhtar et al., 2025; Lam et al., 2022). Severe thalassemia can lead to significant patient morbidity and economic strain for under-resourced health systems (Xu et al., 2021a).

Over recent decades, various countries have developed screening programs, including premarital and prenatal screening (Hossain et al., 2024; Verma et al., 2011). Carrier screening involves hematological and molecular techniques to detect asymptomatic individuals, while early detection in high-risk groups enables timely genetic counseling and prenatal diagnosis, forming a continuum aimed at reducing disease incidence (Cao, 2002; Corda et al., 2021; Zhang et al., 2025). The evolution of screening strategies reflects growing recognition of the need for early detection and prevention to alleviate treatment costs and improve patient outcomes (Amarasinghe et al., 2022; Bonham et al., 2023)

Cascade, prenatal screening, or genetic counseling as the effectiveness of comprehensive national prevention programs has been evidenced in several countries, for example, Cyprus, Sardinia, several regions of Continental Italy, and Greece (Cousens et al., 2010). Screening is usually performed with a complete blood count (FBC) and hemoglobin analysis, including quantification of HbA2 and hemoglobin F (HbF). The Thalassemia International Foundation recommends genetic testing if both parents have MCH <25 (Lee et al., 2019). Once a screening test detects a health concern, early intervention can begin to minimize the condition's severity or complications (Jane et al., 2012).

However, the incidence of thalassemia remains high despite these attempts. It is estimated that 56,000 severe cases are born each year, predominantly in regions such as South Asia, Southeast Asia, and the Mediterranean (Ghotbi & Tsukatani, 2005; Xu et al., 2021b). At present, thalassemia prevention and screening remain a challenging issue in a few countries, such as Thailand (Xu et al., 2021a), United Arab (Elsadek et al., 2022), Indonesia (Wahidiyat et al., 2021), Pakistan (Ahmed, 2022), and Iran (Hashemi-Soteh et al., 2019). It is evident that, among both individuals knowledgeable about thalassemia and those who lack such knowledge, there is a gap between knowledge and behavior toward thalassemia screening. In Indonesia, 95.7% of young people have never had a thalassemia test (Wahidiyat et al., 2021). They have not yet disclosed their motivation for declining to participate in the screening program. In Bangladesh, nearly all parents (100%) were not examined for thalassemia-related features before marriage, and nearly all parents (97%) were unaware of the disease at the time of their child's diagnosis. Only 5% of parents would dispute prenatal diagnoses back then, although 70% were hesitant (Hossain et al., 2021).

Determining and promoting family health is challenging due to the complexity of families and the diversity of family life across different racial, cultural, and geographical contexts (Pearce et al., 2018). In thalassaemia prevention, people's decisions would help obtain community awareness of this disease and facilitate easier tracking of its spread. Furthermore, thalassaemia screening can reduce the incidence and economic burden of thalassaemia. Environment and context influence how behaviour is interpreted. Health prevention is challenging across diverse ethnic, cultural, and geographic situations (Zaborskis et al., 2021). A few studies have been done to explore thalassaemia screening in the family (Chakravorty & Dick, 2019; Hossain et al., 2021; Xu et al., 2021b) and hemoglobinopathies (Xu et al., 2021a) toward thalassaemia screening. Most of this research is fragmented on barriers, knowledge, or attitudes toward thalassaemia prevention.

Even though early detection is widely recognized as crucial, significant challenges remain in implementing comprehensive thalassemia screening tailored to diverse populations. While existing research has largely focused on measuring screening uptake and outcomes, less attention has been given to understanding how public attitudes and perceived barriers shape participation in screening initiatives. This systematic review therefore, aims to critically explore public attitudes and barriers toward the implementation of thalassemia screening strategies. By synthesizing qualitative evidence across diverse countries and population settings, this review is important for informing policy and nursing practice, addressing existing public attitudes, and enhancing thalassemia prevention efforts. Effective participation in screening is essential for identifying and tracking carriers, reducing the likelihood of carrier–carrier marriages, and preventing the birth of children with thalassemia. Insights into public attitudes toward thalassemia screening can

provide a foundation for nurses to develop targeted nursing care plans and interventions to adapt, modify, or improve public engagement in thalassemia prevention.

2. Methods

2.1 Research design

This study is a systematic review because it identifies, selects, evaluates, and synthesizes high-quality research relevant to the research topic (Pollock & Berge, 2018). It is a qualitative systematic review with thematic synthesis focusing on public and family attitudes and barriers toward thalassemia screening, synthesizing qualitative evidence to address the sociocultural and behavioral dimensions of screening participation.

2.2 Search methods

This systematic literature review was conducted and reported in accordance with the recommendations of the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) criteria. This approach ensured methodological rigor throughout the review process. Four databases—SCOPUS, PubMed, ProQuest, and ScienceDirect—were searched for studies published within the last ten years up to July 2, 2023. This study used a qualitative systematic review approach with the Sample, Phenomenon of Interest, Design, Evaluation, Research type (SPIDER) framework to guide the literature search and selection strategy. The Sample in this study included adolescents to adults, premarital couples, parents, and health workers involved in thalassemia screening; the Phenomenon of Interest focused on various barriers to the implementation and acceptance of thalassemia screening; Design included studies using in-depth interviews, focus group discussions, or observations; Evaluation was directed at barriers, attitudes, and sociocultural factors influencing screening decisions; while Research type was limited to qualitative or mixed-method studies. Keywords used in the search included “attitude” AND “barrier” AND “thalassemia” AND “screening” AND “prevention”. To further mitigate bias and enhance the reliability of the results, a systematic protocol was implemented for study identification, screening, eligibility assessment, and data extraction.

2.3 Inclusion and exclusion criteria

The criteria for the publications evaluated included those involving individuals, family members, or community views and experiences with thalassemia or hemoglobinopathy screening in the forms of cascade, premarital, prenatal, and genetic screening; using qualitative methods and published in the English language. Studies using quantitative methods, reviews, conference abstracts, and final project reports within mixed-method designs were excluded from this review. In this study, a ten-year time constraint was applied to enhance the applicability of the findings to contemporary thalassemia practices and policies, while reducing bias due to outdated contexts that are no longer relevant to current thalassemia screening procedures and regulations.

2.4 Screening of articles

The study selection and screening process followed the PRISMA guidelines and is presented in Figure 1. Title and abstract screening were independently conducted by two reviewers, with discrepancies resolved through discussion. Only studies with a qualitative design were included in the review. Studies involving sickle cell disease were included when attitudes and barriers overlapped with hemoglobinopathy prevention and when they investigated attitudes or barriers toward thalassemia screenings. The included studies reported barriers or attitudes toward thalassemia screening or prevention.

2.5 Data extraction

During the data extraction step, the following features of each study were recorded: author(s) and year of publication, country, study objectives/aims, participants, methods of data collection and analysis, and thematic findings. Data extraction was conducted independently by two reviewers using an automatic tool provided by each database. The extracted data were then cross-checked between reviewers, and any discrepancies were resolved through discussion to ensure accuracy and completeness. During the data extraction process, the researchers reported the findings of each article as presented in the original publications. The preliminary comparative data from each article were subsequently analyzed alongside the thematic findings identified in

the study. Upon completion of the extraction process, a comprehensive analysis was performed to investigate barriers to thalassemia screening as identified across the included articles.

2.6 Quality appraisal

The Critical Appraisal Skill Program (CASP) Qualitative Checklist was used to appraise methodological quality. The checklist consists of two screening questions and eight detailed questions addressing research value, findings, data analysis, ethical issues, reflexivity, sampling, and study design. Previous research has shown that articles are categorized as high, medium, or poor quality based on CASP scoring if they meet at least eight of the ten criteria, five to seven criteria, or four or fewer criteria, respectively (Kanavaki et al., 2016). This appraisal aimed to guide the interpretation and weighting of data rather than to exclude studies based on quality scores. All studies meeting the inclusion criteria were included, including those of lower methodological quality, to ensure a comprehensive review and retention of potentially useful contextual information. Findings from studies with methodological constraints were carefully interpreted, and their influence on the overall thematic synthesis was examined closely to ensure transparency and analytical rigor while maintaining methodological inclusivity in qualitative evidence synthesis. FI, KM, and MN conducted the methodological appraisal using the CASP tool.

2.7 Data analysis

The selected studies were analyzed using thematic analysis. Line-by-line coding was applied using OpenCode 4.03 software from Umea University and conducted by FI. An inductive thematic synthesis approach was utilized to analyze qualitative findings related to barriers to thalassemia screening. Participant statements were coded line-by-line to identify significant units, with categories developed from codes appearing in at least two articles. Initial themes were proposed by FI and subsequently reviewed by MN and KM as public health and qualitative research experts, while AAW and TTW ensured clarity and accuracy of the language. The thematic analysis focused on people's views and barriers across screening strategies. To enhance rigor and sociocultural relevance, reviewers communicated frequently, compared findings iteratively, and conducted contextual language assessments.

3. Results

3.1 Search results

A total of 472 records were identified from database searches (Scopus, PubMed, ProQuest, and ScienceDirect). After removing 34 duplicates, 438 records were screened by title and abstract, of which 425 were excluded for not meeting the inclusion criteria. Thirteen full-text articles were assessed for eligibility, and 2 were excluded due to irrelevance to the study objectives or lack of focus on screening experiences. Consequently, 11 studies were included in the final qualitative synthesis (Figure 1).

3.2 Characteristics and methodological quality of the selected studies

The included studies were conducted across diverse geographical settings, including Brazil (n = 1), Saudi Arabia (n = 1), Iran (n = 1), the United Kingdom (n = 1), Thailand (n = 1), Malaysia (n = 1), the Netherlands (n = 2), Taiwan (n = 1), Africa (n = 1), and the Republic of Maldives (n = 1). Across the 11 studies included, the number of participants ranged from 8 to 86, while reported participant ages ranged from 18 to 68 years. Data were collected primarily through in-depth or semi-structured interviews and focus group discussions, and most studies employed thematic or content analysis to identify key attitudes and barriers related to thalassemia or hemoglobinopathy screening (Table 1, Appendix A). Regarding critical appraisal, ten studies were rated as high quality (CASP scores 8–10), and one study was rated as moderate quality (score = 6) (Table 2).

3.3 Thematic findings of attitudes and barriers toward thalassemia screening

Five themes emerged from the thematic analysis, representing barriers at the individual, familial, and healthcare system levels toward thalassemia screening: (1) negative attitudes influenced by cultural and personal factors; (2) inadequate family support and decision-making; (3) limited knowledge of hereditary and genetic conditions; (4) low self and parental awareness of thalassaemia; and (5) poor communication with healthcare professionals and barriers to screening accessibility.

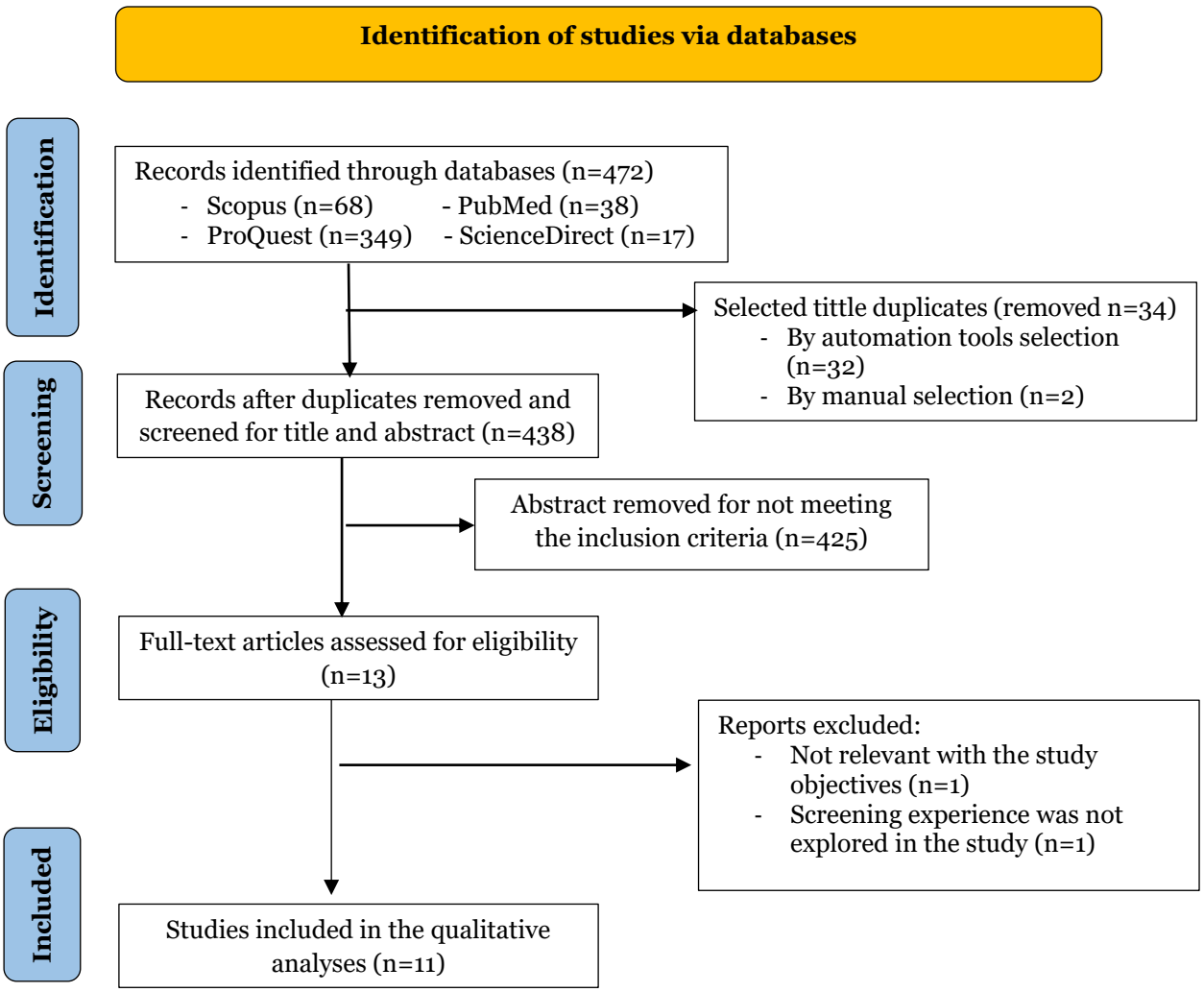

Figure 1. Screening and selection of studies for the review

3.3.1 Negative attitudes affected by cultural and personal factors

The thematic synthesis revealed that perceptions about Thalassaemia Screening Services (TSS) significantly influence screening uptake. Adverse perceptions towards screening were influenced by cultural and individual characteristics. Screening was frequently regarded as redundant or in contrast with dominant social norms, with worry about stigma, familial reputation, and conventional ideas concerning marriage and heredity (S2, S4, S6, S7, S9). On a personal level, negative attitudes were exhibited as dread of adverse outcomes, denial of perceived risks, and hesitance towards medical testing, which subsequently inhibited the willingness to engage in screening (S1, S2, S6) (Figure 2).

"*The whole family [was] traumatized … and I, together…*" (Study 2) (Cohen-Kfir et al., 2020)

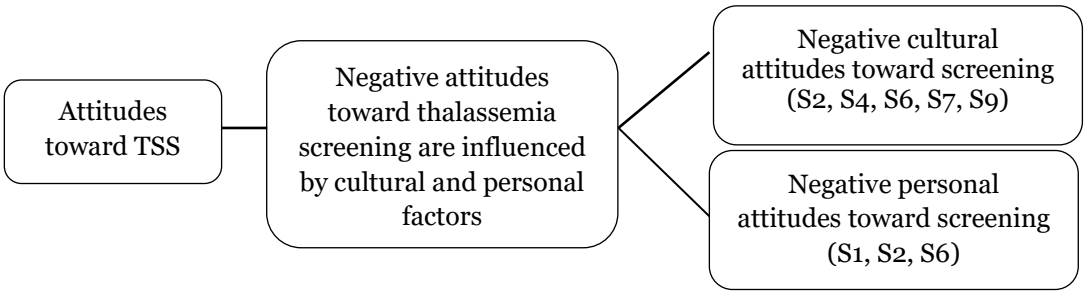

Figure 2. Synthesis tree view of attitudes toward TSS

Table 2. Quality appraisal of the selected articles based on the CASP tools

Criteria	Rocha et al., 2022	Cohen-Kfir et al., 2020	Hosoya, 2017	Boardman & Hale, 2019	Xu et al., 2021a	Jenet et al., 2022	Holtcamp et al., 2017	Chen & Cheng, 2020	Verdonk et al., 2018	Archer et al., 2022	Waheed et al., 2016
1. Was there a clear statement of the aims of the research?	+	+	+	+	+	+	+	+	+	+	+
2. Is a qualitative methodology appropriate?	+	+	+	+	+	+	+	+	+	+	+
3. Was the research design appropriate to address the aims of the research?	+	+	?	+	+	+	+	+	+	+	+
4. Was the recruitment strategy appropriate to the aims of the research?	?	+	+	+	+	-	+	+	?	+	+
5. Was the data collected in a way that addressed the research issue?	+	+	+	+	+	+	+	+	+	+	+
6. Has the relationship between the researcher and participants been adequately considered?	-	?	-	?	?	-	?	+	?	?	?
7. Have the ethical issue taken into considerations?	+	+	?	+	+	+	+	+	+	+	+
8. Was the data analysis sufficient rigorous?	+	+	?	+	+	+	+	+	+	+	+
9. Is there a clear statement of findings?	+	+	+	+	+	+	+	+	+	+	+
10. How valuable is the research?	+	+	+	+	+	+	+	+	+	+	+
Score /10	8	9	6	9	9	8	8	10	8	9	9

Notes. +: Yes, currently met; – : No, clearly not met; ?: Not known.

3.3.2 Inadequate family support and decision-making

This theme underscores the significant impact of family dynamics on decisions regarding thalassaemia screening. Multiple studies indicated ambivalence in family decision-making, characterised by divergent perspectives, uncertainty, or internal conflicts, which hindered or obstructed screening uptake (S2, S8, S9, S11). Moreover, communication obstacles with relatives hindered the transmission of precise information concerning screening advantages and protocols (S1). The lack of emotional and instrumental support was also shown by the lack of family support, which showed that the family was not committed to preventative screening (S1, S6).

3.3.3 Low knowledge about hereditary and genetic conditions

Limited understanding of thalassaemia as a hereditary genetic condition emerged as a significant barrier to screening participation. Several studies reported inadequate information concerning thalassaemia screening, particularly regarding its objectives, scheduling, and preventive benefits (S1, S2, S5, S6, S8, S11). This theme reflects information deficits associated with medical and genetic aspects of thalassemia, highlighting the insufficient comprehension of hereditary characteristics, inheritance mechanisms, screening advantages, and the ramifications of genetic counseling. These knowledge deficits were illustrated by several participant statements across studies, as shown below:

> *"…Now I don't understand so much [regarding genetic counseling]. If there was a lecture, I would go [to it] … the head of genetics."* (Study 2) (Cohen-Kfir et al., 2020)

> *"Exactly what it was, I had never seen anybody with thalassemia; I didn't even know it ran in the families at all."* (Study 5) (Boardman & Hale, 2019)

Moreover, inadequate understanding of familial genetic risk fostered the belief that screening was superfluous, especially in the absence of conspicuous symptoms (S1, S2, S6, S7). This information deficit substantially compromised risk assessment and informed decision-making.

3.3.4 Low self- and parental awareness of thalassemia

This category indicates a lack of awareness among individuals and parents regarding thalassaemia and its long-term consequences. Barriers included inadequate family planning, wherein genetic factors were infrequently incorporated into reproductive choices (S1, S6), and insufficient premarital planning, where thalassaemia screening was not regarded as a priority before marriage (S1, S6). Moreover, insufficient self-awareness concerning individual career status (S4, S7) and enduring biases about screening, such as the assumption that screening is needless in the absence of symptoms, further decreased involvement in screening (S2, S3, S4, S6, S9, S11). This theme refers not simply to knowledge or ignorance but also to a diminished sense of relevance and perceived vulnerability. Individuals or parents may be aware of thalassemia yet may not regard it as significant in premarital planning, reproductive choices, or the future of their children.

3.3.5 Poor communication with HCP and barrier accessibility toward screening.

The fifth category encompasses structural and systemic barriers within healthcare services. Many studies found obstacles to receiving screening services, such as restricted availability, financial limitations, and geographic inaccessibility (S1, S5, S6, S7, S8, S10). Furthermore, inadequate communication between healthcare practitioners and the population limited potential for effective counselling and education related to thalassaemia screening (S1, S2, S6, S7, S8). The insufficient prioritisation of thalassaemia screening in healthcare systems has worsened its underutilisation as a preventative public health strategy (S5, S7, S10). Participants stated:

> *"I waited for a long time but didn't get call to do the screening."* (Study 6). (Jenet et al., 2022)

> *"Very helpful to have some kind of a practical tool or, if necessary, a website with concise and reliable information."* (Study 7). (Holtkamp et al., 2017)

The barrier mapping for thalassemia screening strategies (TSS) is shown in Figure 3, and the thematic findings on attitudes and barriers from each study are presented in Table 3.

4. Discussion

This review aimed to systematically explore public attitudes and barriers toward thalassemia screening strategies (TSS). The findings were mapped according to attitudes and barriers related to TSS, yielding five themes: (1) negative attitudes influenced by cultural and personal factors; (2) inadequate family support and decision-making; (3) limited knowledge of hereditary and genetic conditions; (4) low self and parental awareness of thalassemia; and (5) poor communication with healthcare professionals and barriers to screening accessibility. Each theme is discussed below.

4.1 Attitudes toward TSS

4.1.1 Negative attitudes affected by cultural and personal factors

The description of attitude appears in four research studies, encompassing negative personal and cultural attitudes toward screenings. Due to religious and cultural factors, as experienced by minority people in genetic counseling or genetic testing, prenatal screening policies related to pregnancy termination cannot be adopted. This is consistent with other research in Arabia, where 75% of students agreed that thalassemia fetuses should not be aborted (Elsadek et al., 2022). Parents have difficulty deciding whether this procedure is safe for the baby. They believe this is no longer important due to parents with normal conditions. According to one study, families have unpleasant occurrences and experiences associated with pregnancy termination that they do not want to repeat.

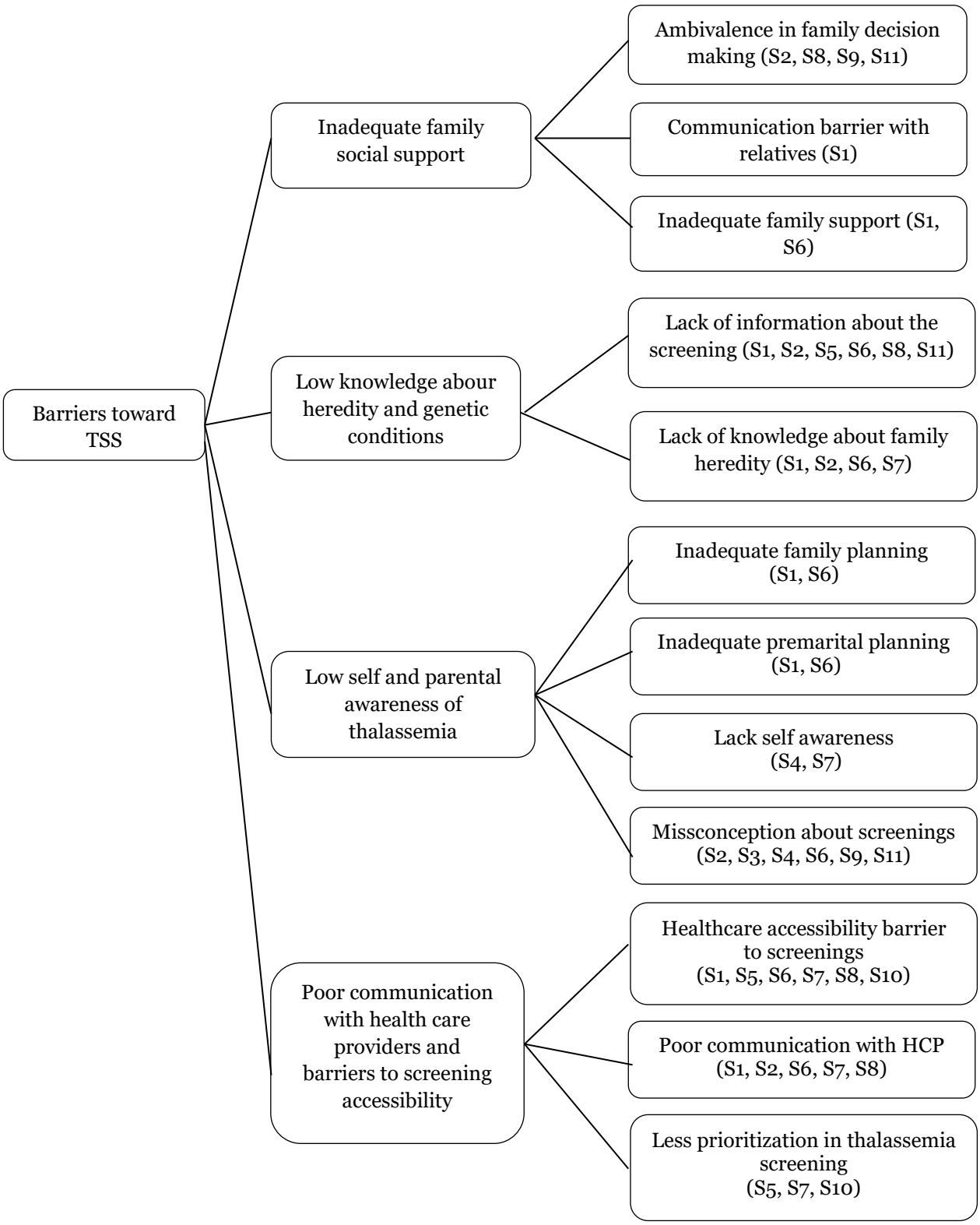

Figure 3. Synthesis tree view of barriers toward TSS

Roche et al. (2022) reported disappointment or past trauma resulting from diagnostic procedures. In contrast, three investigations revealed that participants were rejected for private reasons, such as concerns regarding genetic counseling (Cohen-Kfir et al., 2020). This is unlike other findings that show a mix of factors between individuals and the impact of beliefs, especially religious ones, in Taiwanese and Malaysian mothers related to abortion (Chen & Cheng, 2020; Jenet et al., 2022). They believed that termination of pregnancy did not fit their beliefs.

Table 3. Thematic analysis distribution of the findings

Themes	Rocha et al., 2022	Cohen-Kfir et al, 2020	Hosoya,2017	Boardman & Hale, 2019	Xu et al., 2021a	Jenet et al., 2022	Holtcamp et al., 2017	Chen & Cheng, 2020	Verdonk et al.,2018	Archer et al, 2022	Waheed et al.,2016	Total
Attitudes toward TSS												
Negative Attitudes Influenced by Cultural and Personal Factors												
Negative cultural attitude toward screening		+		+		+	+		+			5
Negative personal attitude toward screening	+	+				+						3
Barriers toward TSS												
Inadequate Family Social Support												
Ambivalence in family decision-making		+	+					+	+		+	5
Communication barrier with relatives	+											1
Inadequate Family Support	+					+						2
Low Knowledge about Hereditary and Genetic Conditions												
Lack of information about the screening	+	+			+	+		+			+	6
Lack of knowledge about family hereditary	+	+				+	+				+	5
Low Self and Parental Awareness of Thalassemia												
Inadequate family planning	+					+						2
Inadequate premarital planning				+			+					2
Lack self-awareness		+	+	+		+			+		+	6
Misconception about screenings					+							1
Poor Communication with Health Care Providers and Barriers to Screening Accessibility												
Healthcare accessibility barrier to screening	+				+	+	+	+		+		6
Poor communication with HCP	+	+				+	+	+				5
Less prioritization in thalassemia screening					+		+			+		3

Pregnancy termination is one way the condition manifests itself, although information on refusal to cascade screen based on religion is limited. In other research, abnormal genetic results were detected in 50 of 173 cases (28.9%) with a termination indication of structural deformity who agreed to invasive genetic testing (Eyisoy et al., 2023). Being diagnosed with a chronic illness is a traumatic life event (Van Wilder et al., 2021). Primary missed diagnoses and diagnostic errors have a modest occurrence but have significant consequences for patients, hospitals, and medical systems (Moonen et al., 2017). Planned follow-up following proper explanation can aid in preventing diagnostic error and detecting initial missed diagnoses, minimizing the delay to the final diagnosis, and the risk of medicolegal action. These findings collectively suggest that views

towards thalassaemia screening are profoundly influenced by sociocultural contexts and individual perspectives, highlighting the necessity for culturally sensitive and person-centred strategies to enhance screening acceptance.

4.2 Barriers toward TSS

4.2.1 Inadequate family support and decision-making

Major components include ambivalence in family decision-making, communication barriers with relatives, and insufficient family support. Six research studies found ambivalence in screening decision-making. This occurs when there is no dominating decision-maker in the family or each party has different screening preferences. In one study, extended families did not participate in genetic screening programs due to the difficulty of communicating with other family members about diseases and cascade screening programs; in reality, families only learned about the hereditary condition after a child died (Rocha et al., 2022). Furthermore, this study shows that communication barriers with extended family members and a lack of understanding about this hereditary illness result in a lack of positive behavior among family members who engage in screening. In another study, the lack of family support shown by parents with children with thalassemia is nevertheless pressured to have another child, expecting that the next child will be born healthy, by in-laws or other family members (Jenet et al., 2022).

Parents and partners are more involved in the decision-making process for thalassemia screening. However, in one study by Cohen-Kfir et al. (2020), the expectant mother chose not to terminate her pregnancy on her initiative, drawing lessons from her mother's previous misstep, which was that she had followed her husband's advice. Merely imprecision in their decision-making process might transform screening and planning choices within the family into disobedient actions. This situation indicates that decision-making processes and who makes them can significantly impact health status and results.

Couples' collaborative decision-making may reflect pressure from a spouse to portray a united front to outsiders when one partner (typically the man) makes the decisions, and there is no indication of respect or engagement of the other partner (Osamor & Grady, 2018). Health workers can provide decision-support interventions to families with difficulty making health decisions (Shepherd et al., 2021). Extra time or information load for family members could be a barrier. However, this was weighed against the need to make informed decisions regarding including people who lack capacity.

4.2.2 Low knowledge about hereditary *and genetic conditions*

Lack of information about genetic disorders and hereditary diseases in the family was cited as a barrier to screening in 63.6% of the studies. The primary situations of this condition include ignorance of genetic disorders, such as not knowing if one's family or oneself is a carrier of the trait, lacking knowledge about the disease and screening information (particularly genetic counselling and treatments), and failing to recognize the possible risks for thalassemia patients or those with haematology genetic disorders (Cohen-Kfir et al., 2020; Jenet et al., 2022; Rocha et al., 2022; Waheed et al., 2016). Only one study found that the family understood screening since they had received information from health providers (Holtkamp et al., 2017), but this was insufficient to fully comprehend genetic disorders in the family. From the health workers' perspective, this emerges since no standard information should be presented to families.

These results are in line with a cross-sectional study in Malaysia on hereditary disorder diseases, in which 24.5% of people are unwilling to undertake genetic testing because they believe it interferes with nature and contradicts religion and their beliefs (Chin & Tham, 2020). Effective preconception or premarital preventive initiatives lead to few cases detected postnatally (Angastiniotis & Lobitz, 2019). The need for more information regarding preventative actions that must be performed linked to prenatal screening and genetic counseling, particularly the location of hospitals that might offer this service, and how they can obtain this process, is a challenge explicitly faced in screening.

4.2.3 Low self and parental awareness of thalassemia

The theme includes four points, which are as follows: inadequate family planning, inadequate premarital planning, lack of self-awareness of thalassemia, and misconceptions about the screening. Failure of family planning in couples with carrier features is caused by two factors: an

absence of preparation and a failure to follow through on the process outlined in the arrangements for having children. Parents stated that they were late in screening because the gestational age was already growing up, that they neglected to use contraceptive medication, or that their behavior caused them to stop taking contraception, and that this produced an impact on the development of pregnancies having children with thalassemia (Hosoya, 2017; Jenet et al., 2022). Two investigations revealed that parents who knew they were thalassemia carriers did not engage in family planning (Jenet et al., 2022; Rocha et al., 2022). This might occur because of their ignorance of the significance of the carrier issue (Boardman & Hale, 2019)

Lack of self-awareness in thalassemia patients shows in the significant study (n=6) that it happens particularly before marriage; even though they are aware that they are survivors, this disease leads to careless behavior while selecting a partner. They are uncomfortable when they have to question possible partners for screening, believe that discussing pregnancy is taboo, and believe that it is too late to find another partner because the relationship is already profound. The studies further indicated that people did not want to be screened since they felt well and fine and did not have symptoms of sickness like other people or family members who were sick.

In another study, many teenagers and young adults are unaware of their thalassaemia status, especially premaritally, when screening is commonly delayed despite family awareness (Fatkhiyah et al., 2025). Unmarried youth and university students with inadequate information and low perceived personal risk are less likely to participate in premarital thalassaemia screening, lowering partner selection prevention (Pervin et al., 2021). Insufficient understanding of thalassemia's hereditary nature and screening services continues to impact negligent reproductive choices, underscoring the necessity for early, focused genetic education before marriage (Nor et al., 2022)

According to health belief theory, people will not do anything if they cannot feel the benefits (Rew, 2005). This unmet benefit could be owing to a need for more understanding about the necessity of disease prevention and the consequences of being a carrier of the trait. This conduct can also be attributed to the patient's disbelief in past medical examination results (Cohen-Kfir et al., 2020), concerning the ill effects of prenatal testing, and the belief that genetic tests or genetic counseling are unnecessary. Furthermore, patients decide that screening is unimportant (Waheed et al., 2016). Raising awareness is a critical component of any successful prevention strategy for thalassemia. As the previous research suggests, intensive education for health personnel and people (Cao & Kan, 2013), further education on risk-thalassemia traits is needed.

4.2.4 Poor communication with HCP and barriers to screening accessibility

Five studies reported communication barriers between families or patients and HCPs in screening attempts. Although not all studies found a barrier between patients and HCP, the most common situation happened when delivering information about screening, screening service providers, and screening results follow-up. The communication barrier manifests itself in two contexts: the barriers encountered by patients and the barriers faced by health workers. Miscommunication with the HCP, not receiving information about the screening schedule, and variations in test findings can all be patient barriers, leading to dissatisfaction. Meanwhile, the challenges faced by HCPs include limited practical instruments for screening education and difficulties when offering screening to patients.

Predictors of ineffective communication between patients and HCPs include a lack of consultation time (Mira et al., 2014), attention to communication, and environmental factors (Norouzinia et al., 2015). Seven studies explain the barriers to healthcare accessibility in terms of health infrastructure, including the limited tools for screening, particularly in primary care, and the distance from home to the screening location, which is difficult to reach. The perceived difficulties from the patient or family are restricted costs, limited time to participate in screening, and the high cost of screening.

The lack of prioritizing thalassemia screening includes a lack of significant government support for screening funds. Furthermore, in Africa, the government still needs to provide funds for individual screening (Archer et al., 2022). Social support from the local government is essential to increase the coverage of screening. This can also be a way to overcome the notion that screening is not yet necessary, hence eliminating the perception that screening is unimportant.

5. Implications and limitations

This review emphasizes the critical role of nurses in overcoming attitudinal, informational, familial, and structural barriers to thalassemia screening. Nurses can provide culturally sensitive education by addressing personal and religious concerns and by strengthening family-centered nursing practices. They can also support shared decision-making, thereby enhancing screening uptake. The need for community genetic literacy is highlighted through structured educational initiatives in various settings. Targeted interventions, such as premarital counseling and risk-based education, should be integrated into nursing routines. Expanding nurses' competencies in therapeutic communication and their role as health counselors may improve participation in screening programs by addressing misinformation, stigma, and access barriers.

This study, however, has certain limitations. Although methodological quality was appraised using CASP, lower-quality studies were retained to preserve comprehensiveness; consequently, some synthesized findings may reflect methodological weaknesses of the primary studies. Additionally, most included studies were conducted in specific sociocultural contexts, potentially limiting the transferability of findings to regions where thalassemia screening is not mandatory or less established. Variations in study design, participant characteristics, and healthcare systems may also have influenced thematic interpretation despite efforts to ensure analytic rigor.

6. Conclusion

This qualitative synthesis illustrates that negative perceptions and complex barriers significantly impede the implementation of Thalassaemia Screening Strategies (TSS). Obstacles emerge at various levels, such as insufficient individual and parental awareness, restricted understanding of hereditary and genetic risks, inadequate familial social support, and ineffective communication and accessibility within healthcare institutions. Concurrently, adverse cultural and individual attitudes towards screening, including stigma-related fears, misunderstandings regarding genetic testing, and denial of personal risk, intensify reluctance to participate in screening. The study implies that thalassaemia screening surpasses a merely biological concern, presenting a multifaceted socio-cultural and behavioural challenge, especially evident during pivotal life phases like the premarital era. Several strategic proposals are suggested. Initially, early and culturally attuned teaching on thalassaemia and genetic inheritance needs to be incorporated into school, university, and community programs, particularly targeting teenagers and young adults before marriage. Secondly, family-centered treatments are vital for enhancing social support and collaborative decision-making concerning screening, as familial dynamics significantly influence the uptake of preventive services. Third, nurses and healthcare practitioners should undergo training to improve communication skills and proactive counselling, thereby guaranteeing that thalassaemia screening is constantly advocated rather than an elective test. Policymakers should prioritise enhancing accessibility and integration of TSS into primary healthcare and premarital health initiatives, mitigating structural hurdles and normalising screening within conventional public health practices.

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Author contribution

Each author contributed to the preparation of the article. FIH was responsible for manuscript preparation, data collection, and analysis; AAW contributed to content review; KM contributed to data analysis, writing review, and content development; and MN and TTT contributed to manuscript review. All authors approved the final manuscript.

Conflict of interest

There is no conflict of interest in this study; all of the people listed above were involved in the preparation of the document and were notified about their involvement.

Declaration of the use of Artificial Intelligence (AI)

Generative AI, particularly Grammarly and OpenAI's ChatGPT, was employed to enhance linguistic precision and enhance the manuscript's readability. All content was carefully investigated and amended by the authors to guarantee quality and consistency with the study

objectives. The application of AI was confined to language editing and did not affect data analysis, interpretation, or results. The authors accept complete responsibility for the final material and confirm compliance with ethical norms in scientific writing.

References

Agarwal, R. K., Dhanya, R., Parmar, L., Sedai, A., Mani, G., Dhar, A., Hegde, S. Periyavan, S., Erappa, N., Anusha, S., Kumar, S., Rengaraj, B., Mamatha, G. N., Manjunatha, N., Prasad, R. N., Fernandes, R., Makkam, A., & Faulkner, L. (2025). Prenatal hemoglobinopathy screening & prevention in India: A cross-sectional study. *The Indian Journal of Medical Research, 161*(5), 441. https://doi.org/10.25259/IJMR_1003_2024

Ahmed, M. (2022). Healthcare professionals' perceptions of implementing a decision support intervention for cascade screening for beta- thalassemia in Pakistan. *European Journal of Human Genetics, 30,* 795–799. https://doi.org/10.1038/s41431-022-01074-1

Akhtar, S. O., Raza, A. A., Abdullah, Y., & Samadi, A. (2025). Exploring the role of consanguinity in thalassemia prevalence in Pakistan: An in-depth analysis of genetic and cultural factors affecting public health. *Annals of Medicine & Surgery, 87*(7), 4222–4228. https://doi.org/10.1097/MS9.0000000000003404

Amarasinghe, N., Amarasena, A., Thabrew, A., Werawatte, P., Premawardhena, A., Malik, F., Abusayeed, M., & Wickramasinghe, C. (2022). Redesigning new policy options for thalassemia prevention in Sri Lanka. *Thalassemia Reports, 12*(4), 135-142. https://doi.org/10.3390/thalassrep12040018

Angastiniotis, M., & Lobitz, S. (2019). Thalassemias: An overview. *International Journal of Neonatal Screening, 5*(1), 16. https://doi.org/10.3390/ijns5010016

Archer, N. M., Inusa, B., Makani, J., Nkya, S., Tshilolo, L., Tubman, V. N., McGann, P. T., Ambrose, E. E., Henrich, N., Spector, J., & Ohene-Frempong, K. (2022). Enablers and barriers to newborn screening for sickle cell disease in Africa: Results from a qualitative study involving programmes in six countries. *BMJ Open, 12*, e057623. https://doi.org/10.1136/bmjopen-2021-057623

Boardman, F. K., & Hale, R. (2019). "I didn't take it too seriously because I'd just never heard of it": Experiential knowledge and genetic screening for thalassaemia in the UK. *Journal of Genetic Counseling, 28*(1), 141–154. https://doi.org/10.1002/jgc4.1042

Bonham, J. R., Scarpa, M., Schielen, P. C. J. I., Iskrov, G., Angelova, V., Bochev, B., Valchinova, V., Gencheva, T., Dzhuleva, D., Dichev, J., Nedkova, T., Palkova, M., Tyutyukova, A., Hristova, M., Hristova-Atanasova, E., & Stefanov, R. (2023). Neonatal screening prospects for expansion of universal newborn screening in Bulgaria: A survey among medical professionals. *International Journal of Neonatal Screening*, *9* (4), 57. https://doi.org/10.3390/ijns9040057

Cao, A. (2002). Carrier screening and genetic counselling in β-thalassemia. *International Journal of Hematology, 76*(2), 105–113. https://doi.org/10.1007/BF03165098

Cao, A., & Kan, Y. W. (2013). The prevention of thalassemia. *Cold Spring Harbor Perspectives in Medicine, 3*, a011775. https://doi.org/10.1101/cshperspect.a011775

Chakravorty, S., & Dick, M. C. (2019). Antenatal screening for haemoglobinopathies: Current status, barriers and ethics. *British Journal of Haematology, 187*(4), 431–440. https://doi.org/10.1111/bjh.16188

Chen, M. M., & Cheng, B. H. (2020). Understanding Taiwanese women's decisional experiences regarding prenatal screening procedures and diagnostics: A phenomenological study. *Asian Nursing Research, 14*(4), 231–240. https://doi.org/10.1016/j.anr.2020.08.005

Chin, J. J., & Tham, H. W. (2020). Knowledge, awareness, and perception of genetic testing for hereditary disorders among Malaysians in Klang Valley. *Frontiers in Genetics, 11*, 512582. https://doi.org/10.3389/fgene.2020.512582

Cohen-Kfir, N., Bentwich, M. E., Kent, A., Dickman, N., Tanus, M., Higazi, B., Kalfon, L., Rudolf, M., & Falik-Zaccai, T. C. (2020). Challenges to effective and autonomous genetic testing and counseling for ethno-cultural minorities: A qualitative study. *BMC Medical Ethics, 21*, 98. https://doi.org/10.1186/s12910-020-00537-8

Corda, V., Murgia, F., Dessolis, F., Murru, S., Chervenak, F. A., McCullough, L. B., & Monni, G. (2021). Professionally responsible management of the ethical and social challenges of

antenatal screening and diagnosis of β-thalassemia in a high-risk population. *Journal of Perinatal Medicine, 49*(7), 847–852. https://doi.org/10.1515/JPM-2021-0021

Cousens, N. E., Gaff, C. L., Metcalfe, S. A., & Delatycki, M. B. (2010). Carrier screening for Beta-thalassaemia: A review of international practice. *European Journal of Human Genetics, 18* (10), 1077–1083. https://doi.org/10.1038/ejhg.2010.90

Demirbaş, Z. E. (2024). Perspective chapter: Advances in diagnosis of beta thalassemia major. In Šalek, S. Z (Ed.), *Inherited blood disorders - advances in diagnosis and treatment. IntechOpen*. https://doi.org/10.572/intechopen.1007915

Elsadek, A. M., Hassan, N., Shahwan, M., & Abdulrahman Jairoun, A. (2022). Exploring the gap between knowledge and behavior regarding thalassemia among university students: A cross-sectional study in the United Arab Emirates. *Journal of Community Health, 47*, 392–399. https://doi.org/10.1007/s10900-022-01066-8

Eyisoy, Ö. G., Özgökçe, Ç., Uygur, L., Özdemir, M. E., Taşdemir, Ü., Öcal, A., & Demirci, O. (2023). Clinical and genetic aspects of termination of pregnancy; tertiary center experience. *Turkish Journal of Obstetrics and Gynecology, 20*(3), 234–241. https://doi.org/10.4274/tjod.galenos.2023.19677

Fatkhiyah, N., Kartini, A., Nugraheni, S. A., Margawati, A., & Widiasih, R. (2025). Knowledge and attitudes towards thalassemia screening awareness: A study on teenagers from extended families affected by Thalassemia. *Journal of Neonatal Surgery, 14*(4), 163–173. https://doi.org/10.52783/JNS.V14.1762

Ghotbi, N., & Tsukatani, T. (2005). Evaluation of the national health policy of thalassaemia screening in the Islamic Republic of Iran. *Eastern Mediterranean Health Journal, 11*(3), 308-318.

Goonasekera, H. W., Paththinige, C. S., & Dissanayake, V. H. W. (2025). Population screening for hemoglobinopathies. *Annual Review of Genomics and Human Genetics Lanka, 14*(4), 355–380. https://doi.org/10.1146/annurev-genom-091416

Hashemi-Soteh, M. B., Nejad, A. V., Ataei, G., Tafazoli, A., Ghasemi, D., & Siamy, R. (2019). Knowledge and attitude toward genetic diseases and genetic tests among pre-marriage individuals: A cross-sectional study in northern Iran. International journal of reproductive biomedicine, *17*(8), 543–550. https://doi.org/10.18502/ijrm.v17i8.4819

Holtkamp, K. C. A., Vos, E. M., Rigter, T., Lakeman, P., Henneman, L., & Cornel, M. C. (2017). Stakeholder perspectives on the implementation of genetic carrier screening in a changing landscape. *BMC Health Service Research, 17*, 146. https://doi.org/10.1186/s12913-017-2083-9

Hosoya, S. (2017). Changes in attitudes towards marriage and reproduction among people with a genetic illness: A study of patients with thalassemia in Iran. *Anthropology of the Middle East, 12*(2), 28–45. https://doi.org/10.3167/ame.2016.120203

Hossain, M. J., Das, M., & Munni, U. R. (2024). Urgent call for compulsory premarital screening: a crucial step towards thalassemia prevention in Bangladesh. *Orphanet Journal of Rare Diseases, 19*(1), 326. https://doi.org/10.1186/S13023-024-03344-1

Hossain, M. S., Mahbub Hasan, M., Petrou, M., Telfer, P., & Mosabbir, A. A. (2021). The parental perspective of thalassaemia in Bangladesh: Lack of knowledge, regret, and barriers. *Orphanet Journal of Rare Diseases, 16*(1), 315. https://doi.org/10.1186/s13023-021-01947-6

Jane, B., Bindler, R., & Cowen, K. (2012). *Principles of pediatric nursing: Caring for children (5th ed.)*. Pearson Education.

Jenet, G. C., Abd Hamid, I. J., Julaiha, A., & Mangantig, E. (2022). Mothers with multiple β-thalassemia major children in Sabah, Malaysia: A qualitative study exploring the contributing factors. *Journal of Health and Translational Medicine, 25*(Special Issue 1), 144–152. https://doi.org/10.22452/jummec.sp2022no1.16

Kanavaki, A. M., Rushton, A., Klocke, R., Abhishek, A., & Duda, J. L. (2016). Barriers and facilitators to physical activity in people with hip or knee osteoarthritis: Protocol for a systematic review of qualitative evidence. *BMJ Open, 6*(11), e012049. https://doi.org/10.1136/bmjopen-2016-012049

Lam, T. T., Nguyen, D. T., Le, Q. T., Nguyen, D. A., Hoang, D. T. T., Nguyen, H. Du, Nguyen, C. C., Doan, K. P. T., Tran, N. T., Ha, T. M. T., Trinh, T. H. N., Nguyen, V. T., Lam, D. T., Le, M. T., Nguyen, X. T., Ho, T. H. T., Tran, T. H., Ho, V. T., Bui, T. Van, … Nguyen, H. N. (2022). Combined gap-polymerase chain reaction and targeted next-generation sequencing improve

α- and β-thalassemia carrier screening in pregnant women in Vietnam. *Hemoglobin, 46*(4), 233–239. https://doi.org/10.1080/03630269.2022.2096461

Lee, S. Y., Yap, E. S., Lee, E. Y., Goh, J. H., Liu, T. C., & Yip, C. (2019). Evaluation of thalassaemia screening tests in the antenatal and non-antenatal populations in Singapore. *Annals of the Academy of Medicine, 48*(1), 5–15.

Long, H. A., French, D. P., & Brooks, J. M. (2020). Optimising the value of the critical appraisal skills programme (CASP) tool for quality appraisal in qualitative evidence synthesis. *Research Methods in Medicine & Health Sciences, 1*(1), 31–42. https://doi.org/10.1177/2632084320947559

Mensah, C., & Sheth, S. (2021). Optimal strategies for carrier screening and prenatal diagnosis of α- and β-thalassemia. Hematology, *2021*(1), 607–613. https://doi.org/10.1182/hematology.2021000296

Mira, J. J., Guilabert, M., Pérez-Jover, V., & Lorenzo, S. (2014). Barriers for an effective communication around clinical decision making: An analysis of the gaps between doctors' and patients' point of view. *Health Expectations, 17*(6), 826–839. https://doi.org/10.1111/j.1369-7625.2012.00809.x

Moonen, P. J., Mercelina, L., Boer, W., & Fret, T. (2017). Diagnostic error in the emergency department: Follow up of patients with minor trauma in the outpatient clinic. *Scandinavian Journal of Trauma, Resuscitation and Emergency Medicine, 25*(1), 13. https://doi.org/10.1186/s13049-017-0361-5

Nor, M. A. M., Idris, N. S., Zulkifli, M. M., Abu Bakar, R., & Ahmad, I. (2022). Thalassemia screening: Low level of knowledge among unmarried youths in Kota Bharu, Kelantan, Malaysia. *Malaysian Family Physician, 17*(1), 57–65. https://doi.org/10.51866/OA.31

Norouzinia, R., Aghabarari, M., Shiri, M., Karimi, M., & Samami, E. (2015). Communication barriers perceived by nurses and patients. *Global Journal of Health Science, 8*(6), 65–74. https://doi.org/10.5539/gjhs.v8n6p65

Osamor, P. E., & Grady, C. (2018). Autonomy and couples' joint decision-making in healthcare. *BMC Medical Ethics, 19*, 3. https://doi.org/10.1186/s12910-017-0241-6

Pearce, L. D., Hayward, G. M., Chassin, L., & Curran, P. J. (2018). The increasing diversity and complexity of family structures for adolescents. *Journal of Research on Adolescence, 28*(3), 591–608. https://doi.org/10.1111/jora.12391

Pervin, S., Sultana, H., Ahmed, T., Haque, A., Abbas, Md. G., & Hasan, M. M. (2021). Knowledge and awareness regarding premarital screening of β-thalassemia among undergraduate students in bangladesh. *Journal of Current Medical Research and Opinion, 4*(01), 730–737. https://doi.org/10.15520/JCMRO.V4I01.379

Pollock, A., & Berge, E. (2018). How to do a systematic review. *International Journal of Stroke*, *13*(2), 138-156. https://doi.org/10.1177/1747493017743796

Rew, L. (2005). Health belief and health-promotion models. In *Health belief and health-promotion models* (pp. 253-286). SAGE Publications, Inc., https://doi.org/10.4135/9781452233550.n9

Rocha, R., de Souza, T. V., de Morais, R. de C. M., Nascimento, L. de C. N., Do Couto, L. L., & Farias, I. F. de A. (2022). (Lack of) knowledge of mothers about sickle cell trait and disease: A qualitative study. *Revista Brasileira de Enfermagem, 75*(1), e20201217. https://doi.org/10.1590/0034-7167-2020-1217

Shepherd, V., Wood, F., Griffith, R., Sheehan, M., & Hood, K. (2021). Development of a decision support intervention for family members of adults who lack capacity to consent to trials. *BMC Medical Informatics and Decision Making, 21*(1), 30. https://doi.org/10.1186/s12911-021-01390-4

Van Wilder, L., Pype, P., Mertens, F., Rammant, E., Clays, E., Devleesschauwer, B., Boeckxstaens, P., & De Smedt, D. (2021). Living with a chronic disease: Insights from patients with a low socioeconomic status. *BMC Family Practice, 22*, 233. https://doi.org/10.1186/s12875-021-01578-7

Verdonk, P., Metselaar, S., Storms, O., & Bartels, E. (2018). Reproductive choices: A qualitative study of Dutch Moroccan and Turkish consanguineously married women's perspectives on preconception carrier screening. *BMC Women's Health, 18*, 79. https://doi.org/10.1186/s12905-018-0574-4

Verma, I., Saxena, R., & Kohli, S. (2011). Past, present & future scenario of thalassaemic care & control in India. *The Indian journal of medical research, 134*(4), 507–521. https://pmc.ncbi.nlm.nih.gov/articles/PMC3237251/

Waheed, F., Fisher, C., Awofeso, A., & Stanley, D. (2016). Carrier screening for beta-thalassemia in the Maldives: Perceptions of parents of affected children who did not take part in screening and its consequences. *Journal of Community Genetics, 7*(3), 243–253. https://doi.org/10.1007/s12687-016-0273-5

Wahidiyat, P. A., Yo, E. C., Wildani, M. M., Triatmono, V. R., & Yosia, M. (2021). Cross-sectional study on knowledge, attitude and practice towards thalassaemia among Indonesian youth. *BMJ Open, 11*, e054736. https://doi.org/10.1136/bmjopen-2021-054736

Xu, J. Z., Foe, M., Tanongsaksakul, W., Suksangpleng, T., Ekwattanakit, S., Riolueang, S., Telen, M. J., Kaiser, B. N., & Viprakasit, V. (2021a). Identification of optimal thalassemia screening strategies for migrant populations in Thailand using a qualitative approach. *BMC Public Health, 21,* 1796. https://doi.org/10.1186/s12889-021-11831-4

Xu, J. Z., Tanongsaksakul, W., Suksangpleng, T., Ekwattanakit, S., Riolueang, S., Telen, M. J., & Viprakasit, V. (2021b). Feasibility of and barriers to thalassemia screening in migrant populations: A cross-sectional study of Myanmar and Cambodian migrants in Thailand. *BMC Public Health, 21*, 1177. https://doi.org/10.1186/s12889-021-11059-2

Zaborskis, A., Kavaliauskienė, A., Eriksson, C., Klemera, E., Dimitrova, E., Melkumova, M., & Husarova, D. (2021). Family support as smoking prevention during transition from early to late adolescence: A study in 42 countries. *International Journal of Environmental Research and Public Health, 18*(23), 12739. https://doi.org/10.3390/ijerph182312739

Zhang, C., Chen, V. C., Osa-Andrews, B., & Cao, J. (2025). Emerging technologies and advanced strategies in hemoglobin defect screening. *Journal Clinical of Medicine, 14*(16), 5690. https://doi.org/10.3390/jcm14165690

Appendix A

Table 1. Characteristics and findings of the selected studies

Author/ Country	Aims	Number and age range of participants, n, years	Data collection, analysis	Thematic Findings
S1 (Rocha et al., 2022)/ Rio De Jeneiro (Brazil)	To analyze the understanding of mothers about sickle cell disease and/or traits of the family from a diagnosed child.	23, unclear	Semi-structured interviews, Thematic analysis	1. Identification of sickle cell disease or trait in the family: (Lack of) knowledge in the face of heredity. 2. The Heel Prick Test result: impact of diagnosis and coping with the situation.
S2 (Cohen-Kfir et al., 2020)/ Saudi Arabia	To explore the general and culture-specific reason for the underutilization of genetic testing and counseling services	28, 26,5-28,8	Individual interviews, Content Analysis	1. Lack of pre-counseling knowledge but desire to learn more. 2. Negative personal and cultural preconceptions about genetic counseling. 3. Culturally based differences in decision-making about pregnancies between Arab subset. 4. Lack of effective communication regarding statistical data from counseling services. 5. Similarities and differences in HCPs versus genetic counselee perceptions.
S3 (Hosoya, 2017)/ Iran	To outline the thalassemia prevention program, which includes carriers' marriage and reproductive choices.	51, 18-49	Semi-structured interviews and questionnaire surveys, unclear	1. Marriage and reproductive choices among people with thalassemia major. 2. Marriage preference among people with thalassemia major.
S4 (Boardman & Hale, 2019)/ United Kingdom	This study explores barriers affecting screening uptake and responses to genetic risk obstacle	15, 25-68	In-depth interviews, Thematic Analysis	1. 'I know exactly how hard it can become…': Experiential knowledge and reproduction in thalassemia families 2. With carriers, it's not a major thing is it? You just carry on normal…": Prenatal screening, experience and reproductive decision-making

Table 1. Continued

Author/ Country	Aims	Number and age range of participants, n, years	Data collection, analysis	Thematic Findings
S5 (Xu et al., 2021a)/ Thailand	To identify key barriers to and facilitators of thalassemia screening and to develop tailored recommendations for providing migrants with access to thalassemia prevention and control.	48, 28-43	28 in-depth interviews and four focus group discussions (FGDs), Thematic Analysis	1. Knowledge barriers 2. Sociocultural factors 3. Structural and systemic factors
S6 (Jenet et al., 2022)/ Malaysia	To explore the reasons that may contribute to mothers in Sabah having multiple children with β-thalassemia major (β-TM)	24, 26-53	In-depth, semi-structured interview, Thematic Analysis	1. Intention to have more children and acceptance towards child's condition 2. Lack of understanding about thalassemia inheritance pattern 3. Barriers in prenatal screening 4. Barriers in abortion 5. Ineffective family planning
S7 (Holtkamp et al., 2017)/ Dutch	To identify general and population-specific barriers and needs reflected by stakeholders regarding the implementation of carrier screening in a changing landscape.	17, unclear	Semi-structured interviews, Thematic Content Analysis	1. Culture: changing the way of thinking (desirability, prioritization) 2. Structure: changing the way of organizing (infrastructure, guidelines, financial structures, challenges during counselling, and training and education.)
S8 (Chen & Cheng, 2020)/ Taiwan	To understand Taiwanese women's decisional experiences regarding prenatal screening procedures and diagnostics.	33, 23-37	Semi-structured interviews. Hermeneutics thematic analysis	1. Accessing health information (Perceived routine information about procedures, Perceived incomprehensible web information) 2. Considering what was best for my baby (Having comprehensive information about desired procedures, Avoiding risks from the test on the baby 3. Considering family finance (Choosing novel screening procedures, Considering affordable test 4. Feeling anxiety posttest (Unprepared for abnormal findings, Anxiety posttest

Table 1. Continued

Author/ Country	Aims	Number and age range of participants, n, years	Data collection, analysis	Thematic Findings
S9 (Verdonk et al., 2018)/ Netherlands	To explore women's perspectives on preconception carrier screening (PCS) and reproductive choices	86, unclear	In-depth Interviews and Focus Group Discussion, Thematic Analysis	1. Partner choice 2. Marriage and having children 3. Healthy children 4. Having your 'own' child: Biological and social parenthood
S10 (Archer et al., 2022)/ Africa	To systematically assess enablers and barriers to the implementation of the NBS program for SCD in Africa	8, unclear	Semi-structured interview, Thematic Analysis	1. Program structure and governance (Health authority endorsement. 2. Technical (Workflow mapping) 3. Funding (Role of government)
S11 (Waheed et al., 2016))/ Republic of Maldives	To explore the reasons for not testing for thalassemia in Maldives before or after marriage.	22,30-53	In-depth interviews, Thematic Analysis	1. Reasons for getting married (Cultural reasons, family reasons, personal reasons) 2. Reasons for not testing (Poor awareness, lack of sufficient knowledge, personal belief, not mandatory)

Notes. S = Study