

Research Article

Public Awareness, Genetic Counseling, And Uptake of Thalassemia Carrier Screening: A Multicenter Study in Pakistan

Aleeza Sana¹, Durr-e-shahwar², Abdul wahid³, Muhammad Younas Siddiqui⁴, Asma Ali⁵, Annie Talpur⁶

¹Ministry of National Health Services Regulations and Coordination. Deputy Director. Islamabad.

²Community Medicine. PUMHS. Peoples University of Medical and Health Sciences. Nawabshah.

³Assistant Professor Haematology. Pathology Department. Jhalawan Medical College. Khuzdar.

⁴Medical Superintendent. Rai Foundation Teaching Hospital. Sargodha.

⁵Assistant Professor. Community Medicine. Rai Medical College. Sargodha.

⁶Lecturer. Physiology Department. Sindh Medical College. Jinnah Sindh Medical University. Karachi.

Corresponding Authors: Aleeza Sana

Ministry of National Health Services Regulations and Coordination. Deputy Director. Islamabad.

Email: ¹aleezasana128@gmail.com, ²durreshahwarlanjar@gmail.com, ³dr.wahid55@gmail.com, ⁴younassaddiqui@gmail.com, ⁵drasmanabeel@gmail.com, ⁶annie.talpur@jsmu.edu.pk

Received: 29.08.26, Revised: 24.09.26, Accepted: 12.10.26

ABSTRACT

Background: Thalassemia prevention in Pakistan depended on public understanding, accessible genetic counseling, and timely carrier screening. However, awareness of the disease did not necessarily translate into testing.

Objective: To assess awareness, changes following counseling, screening completion, and barriers among adults attending the hospital-linked services in Punjab.

Methods: A multicenter prospective before-and-after study included 360 adults aged 18-45 years between January and December 2025. Participants completed a ten-item knowledge assessment before and immediately after structured counseling. Free carrier screening was offered, and completion was verified within 30 days. Paired comparisons and Wilson confidence intervals were calculated.

Results: The mean knowledge score increased from 3.73 ± 1.48 to 7.73 ± 1.33 , with a mean improvement of 4.00 points (95% confidence interval: 3.83-4.17; $p < 0.001$). Willingness to undergo screening increased from 47.5% to 83.9%. Screening was completed by 252 participants (70.0%; 95% confidence interval: 65.1-74.5%). Laboratory findings were consistent with beta-thalassemia trait in 21 screened participants (8.3%; 95% confidence interval: 5.5-12.4%). Among 108 participants without completed screening, travel or work commitments, perceived lack of need, and concerns about marriage prospects were the leading barriers.

Conclusion: Counseling was followed by improved knowledge and greater willingness to screen, although testing completion remained lower. Accessible sampling, confidential communication, and follow-up support emerged as priorities for strengthening prevention services. The findings informed service delivery rather than establishing a causal reduction in disease incidence.

Keywords: Thalassemia, Genetic Counseling, Public Awareness, Disease Prevention.

INTRODUCTION

Thalassemia represented a substantial continuing challenge for child health and community medicine because impaired globin-chain production caused chronic anemia, while its more severe forms required sustained specialist care. The clinical spectrum extended from an often unrecognized carrier state to transfusion-dependent disease, making prevention and treatment closely connected responsibilities.[1] Regular transfusions, iron chelation, and monitoring for complications placed substantial demands on families and

health services, particularly where access to coordinated care remained uneven.[2]

Pakistan carried a substantial burden of beta-thalassemia, although published national estimates relied on heterogeneous sources rather than a recent comprehensive population survey.[3] A Pakistani review described important prevention services, including carrier testing, genetic counseling, and prenatal diagnosis, but also identified gaps in their availability and implementation.[4] These gaps mattered because families needed understandable information and a clear route

to testing before reproductive decisions became urgent.

Public familiarity with the name of the disease did not necessarily indicate an understanding of inheritance or screening. Among Pakistani non-medical university students, knowledge of prenatal screening remained limited despite generally favorable views about premarital testing.[5] Research involving parents of affected children also documented incomplete understanding of transmission and available preventive measures.[6] Another Pakistani study found that many parents remained unaware of antenatal screening, underscoring the need for communication that accommodated different educational backgrounds.[7] These findings suggested that information delivered during clinical contact required careful explanation rather than an assumption that repeated hospital attendance ensured understanding.

Family-based screening offered an important opportunity to reach relatives who shared genetic risk. Work in Punjab developed a decision support resource that participants considered acceptable for discussions about cascade testing.[8] Nevertheless, carrier identification required an appropriate diagnostic pathway: red-cell indices helped identify possible abnormalities, but hemoglobin analysis and, where indicated, molecular testing were needed to clarify carrier status.[9] Laboratory guidance emphasized reliable detection of clinically important variants and accurate quantification of hemoglobin fractions.[10] Screening therefore depended on both public engagement and technical quality; neither component could replace the other.

Genetic counseling provided the link between a laboratory result and its meaning for an individual or couple. Contemporary guidance supported explaining reproductive risk, the limitations of screening, and available choices in a manner that respected personal preferences.[11] Recent literature also described screening across premarital, prenatal, and newborn settings, while emphasizing differences in resources and implementation.[12] Experience from Bangladesh highlighted the importance of joining education, accessible testing, and continuing support within a coordinated prevention strategy.[13] The relevance of that experience lay in comparable service constraints, rather than an assumption that all

South Asian populations had identical beliefs or needs.

More recent international recommendations further emphasized culturally appropriate communication and informed reproductive choice.[14] For Pakistani services, an important unanswered question was what happened after counseling: whether adults understood the information, accepted testing, and completed screening. This study therefore assessed baseline awareness, immediate changes in knowledge and screening willingness, laboratory screening completion within 30 days, and reported barriers among adults attending the hospital-linked services in Punjab. Its emphasis was on the practical steps connecting community education with prevention opportunities for families.

MATERIALS AND METHODS

A multicenter prospective before-and-after study was conducted through community medicine and pediatric services linked to the teaching hospitals in Pakistan, between January and December 2025. Hospital outpatient departments and affiliated outreach sessions provided access to adults from urban and rural catchment areas. Recruitment ended on 30 November, allowing the final 30-day screening assessment to be completed in December. All participants received counseling; there was no untreated comparison group. The same counseling content, assessment timing, and laboratory referral procedures were applied across all the participating services throughout the recruitment period. Ethical approval was obtained from the institutional ethics committees of the participating hospitals. Written informed consent was obtained from all participants before interviews, counseling, and blood sampling. Participation was voluntary, and confidentiality was maintained throughout the study.

Adults aged 18–45 years who had no previously documented thalassemia carrier result were eligible. Healthcare professionals, medical students, individuals with known thalassemia disease, and people unable to complete an interview were excluded. One adult per household was enrolled to reduce duplication of family experiences. Consecutive eligible attendees were approached during designated recruitment sessions. The sample target was based on estimating a screening proportion near 50% with approximately 5.5% absolute precision at 95% confidence,

requiring about 318 observations; allowing for incomplete participation increased the target to 360. This calculation addressed precision, rather than power for demonstrating an intervention effect.

Age, sex, education, residence, marital status, and family history of thalassemia were recorded using a standardized interview form. A ten-item knowledge assessment covered inheritance, carrier health, reproductive risk, diagnostic testing, counseling, and prevention. Each correct answer received one point; incorrect and uncertain answers received zero, yielding scores from zero to ten. Questions were reviewed for content and language by community medicine and pediatric clinicians and tested for comprehension in 20 adults outside the main sample. Urdu versions were used, with spoken Punjabi or Saraiki explanations when needed. This preliminary work assessed usability; it did not establish full psychometric validation.

Before counseling, participants completed the knowledge assessment and four separate questions about screening willingness, family communication, counseling acceptance, and confidentiality. Trained clinicians then delivered an approximately 20-minute session using a common script, simple inheritance diagrams, and a short printed information sheet. The session explained that carriers were generally healthy and that, when both partners carried beta-thalassemia variants, each pregnancy carried a 25% probability of an affected child. Participants were encouraged to ask questions and explain key

messages back in their own words. Discussions emphasized voluntary decisions and avoided blame or instructions about whom participants should marry.

The assessment was repeated immediately after counseling. Free screening was offered regardless of post-session answers, with same-day sampling or a scheduled appointment. A reminder was provided when sampling had not been completed within two weeks. Screening completion required a laboratory record within 30 days; verbal willingness alone did not meet this definition. Participants without a completed test were asked to identify their single most important barrier. Declining testing did not affect access to routine clinical services.

Screening included a complete blood count and high-performance liquid chromatography for every participant who provided a sample. Findings were reviewed by laboratory clinicians. A pattern consistent with beta-thalassemia trait required raised HbA2 at or above the laboratory threshold of 3.5%, interpreted alongside red-cell indices and clinical history. Borderline or discordant findings were recorded separately and referred for further assessment, including iron studies or molecular testing when indicated. A result without a beta-thalassemia trait pattern was not treated as excluding every alpha-globin disorder or rare hemoglobin variant. Participants with a carrier pattern received individual post-test counseling and an offer of partner assessment. Operational definitions were summarized in Table 1.

Table 1. Operational Definitions and Assessment Schedule

Measure	Definition	Timing
Knowledge score	Correct answers across ten items; range 0–10	Before and immediately after counselling
Screening willingness	Affirmative answer to accepting carrier testing	Before and immediately after counselling
Screening completion	Blood count and haemoglobin analysis documented	Within 30 days
Carrier pattern	HbA2 $\geq 3.5\%$, interpreted with indices and history	Laboratory review
Screening barrier	One principal reason for non-completion	Day 30

Records used coded identifiers, and results were communicated privately. Continuous measures were summarized as mean $\pm$ standard deviation and categorical measures as frequency and percentage. Knowledge scores were compared using a paired t-test, and paired binary responses using exact

McNemar tests. Wilson 95% confidence intervals were calculated for screening completion and carrier-pattern frequency. Item-level comparisons were exploratory. Two-sided p-values below 0.05 were considered statistically significant.

RESULTS

Of 402 adults approached, 22 did not meet eligibility criteria and 20 declined participation. The remaining 360 completed both assessments and received counseling. Screening status was established for all participants at 30 days, including those without a completed laboratory test. The analysis therefore used the full enrolled sample for awareness and screening completion, while laboratory findings were

restricted to the 252 participants who underwent testing.

Participant characteristics were summarized in Table 2. Urban and rural residents and different educational backgrounds were represented. A family history of thalassemia was reported by one fifth of participants, indicating that the sample included adults with direct family experience as well as those without it.

Table 2. Participant Characteristics (N=360)

Characteristic	Category	n (%)
Age, years	18–24	120 (33.3)
	25–34	154 (42.8)
	35–45	86 (23.9)
Sex	Female	198 (55.0)
	Male	162 (45.0)
Residence	Urban	204 (56.7)
	Rural	156 (43.3)
Education	Primary or less	108 (30.0)
	Secondary	150 (41.7)
	Higher secondary or university	102 (28.3)
Marital status	Married	216 (60.0)
	Unmarried	144 (40.0)
Family history	Present	72 (20.0)
	Absent	288 (80.0)

Baseline responses showed that recognition of thalassemia as a blood disorder was more common than accurate understanding of reproductive risk. Correct responses increased across all ten knowledge items after

counseling (Table 3). The mean score rose from 3.73 ± 1.48 to 7.73 ± 1.33 . The paired mean improvement was 4.00 points (95% CI: 3.83–4.17; $p < 0.001$). Retention was unmeasured.

Table 3. Correct Knowledge Responses Before and After Counseling (N=360)

Knowledge item	Before, n (%)	After, n (%)
Thalassemia affected hemoglobin production	238 (66.1)	312 (86.7)
Thalassemia was inherited	143 (39.7)	275 (76.4)
Carriers could remain healthy	123 (34.2)	267 (74.2)
Two carriers had a 25% affected-child risk per pregnancy	72 (20.0)	261 (72.5)
Unrelated couples could both be carriers	130 (36.1)	284 (78.9)
Blood testing could identify a carrier pattern	156 (43.3)	288 (80.0)
Screening before pregnancy supported informed choices	98 (27.2)	269 (74.7)
Genetic counselling helped interpret reproductive risk	96 (26.7)	263 (73.1)
Thalassemia was not spread by ordinary social contact	180 (50.0)	288 (80.0)
Prenatal diagnosis differed from parental carrier screening	105 (29.2)	275 (76.4)

All item-level paired comparisons: $p < 0.001$; these comparisons were exploratory. Responses concerning testing and communication also became more favorable after counseling (Table 4). Willingness to

undergo screening increased by 36.4 percentage points. Nevertheless, stated willingness exceeded documented completion. Confidentiality remained relevant despite favourable views of prevention services.

Table 4. Affirmative Responses Before and After Counselling (N=360)

Response	Before, n (%)	After, n (%)	p-value
Willing to undergo carrier screening	171 (47.5)	302 (83.9)	<0.001
Willing to discuss screening with family	90 (25.0)	278 (77.2)	<0.001
Would accept individual genetic counselling if needed	170 (47.2)	310 (86.1)	<0.001
Agreed results should be communicated confidentially	206 (57.2)	320 (88.9)	<0.001

Exact McNemar tests were used for paired responses.

Screening completion and laboratory findings were presented in Table 5. Completion was 70.0% (95% CI: 65.1–74.5%). Among tested participants, 21 had findings consistent with beta-thalassemia trait, giving a screening yield

of 8.3% (95% CI: 5.5–12.4%). Six results required further assessment and were excluded from the carrier-pattern count. These findings described the tested subgroup and were not estimates of population-wide prevalence.

Table 5. Screening Completion and Follow-Up Outcomes

Outcome	n/denominator	%
Screening completed	252/360	70.0
Screening not completed	108/360	30.0
No beta-thalassemia trait pattern on initial testing	225/252	89.3
Beta-thalassemia trait pattern	21/252	8.3
Borderline or discordant laboratory findings	6/252	2.4
Post-test counselling among participants with carrier patterns	21/21	100.0
Participants with carrier patterns who were married	17/21	81.0
Partners tested among married participants with carrier patterns	12/17	70.6
Partner tests showing a carrier pattern	2/12	16.7

Both couples with carrier patterns in each partner received referral for specialist counselling. Their subsequent reproductive decisions were not evaluated. Partner results

were additional service outcomes and were not included in the initial 252 screening denominator.

Table 6. Principal Barriers Among Participants without Completed Screening (N=108)

Principal barrier	n (%)
Travel difficulties or work commitments	34 (31.5)
No perceived need because of good health or absent family history	27 (25.0)
Concern about marriage prospects or social stigma	22 (20.4)
Family disagreement	15 (13.9)
Fear of blood sampling	10 (9.3)

Each participant reported one principal barrier; percentages may not total exactly 100 because of rounding.

No participant selected a direct testing fee, because the offered tests were free. However, free laboratory testing did not remove travel requirements or time away from work. These responses explained the remaining service gap without implying that every participant who declined testing lacked understanding.

DISCUSSION

The findings showed a gap between understanding thalassemia and completing carrier screening. Baseline knowledge was particularly limited for inheritance, carrier health, and reproductive risk. A recent study

from Balochistan similarly reported substantial gaps among parents of affected children, supporting the need for clear preventive messages even within families already familiar with treatment.[15] Research among Bangladeshi college students also identified misconceptions despite interest in screening.[16] These comparisons supported targeted education, although their different populations prevented direct comparison of awareness percentages.

Knowledge improved immediately after counseling, but the increase represented short-term learning rather than evidence of lasting behavior change. A nationwide Bangladeshi study reported that favorable attitudes could coexist with incomplete

knowledge and limited preventive practice.[17] In the present study, the higher post-session willingness than documented testing completion reinforced the importance of measuring what participants actually did after receiving information. Screening intention remained useful for understanding acceptability, but it was insufficient as a service outcome.

The counseling approach also addressed family communication and the meaning of carrier status. Pakistani relatives who used a decision support resource described better understanding while continuing to express concerns about stigma and marriage.[18] Healthcare professionals in Punjab reported that structured information helped their discussions with families, suggesting a practical role for simple resources alongside trained staff.[19] Together, these findings supported consistent explanations in familiar languages and opportunities for private questions. They did not justify pressuring adults to test or treating carrier status as a personal failing.

Recent randomized evidence from Indonesia reported improvements in knowledge and screening intentions with digital educational approaches.[20] Those findings supported exploring additional ways to reinforce information, but they did not establish that digital delivery would outperform the present approach in Pakistan. A recent five-country survey linked knowledge and familiarity with more favorable screening attitudes, further emphasizing that educational context influenced responses.[21] For local services, printed materials, spoken explanations, and optional digital reminders could complement one another where access allowed.

The screening pathway identified participants who needed further counseling, while the remaining barriers showed why free tests alone were insufficient. Antenatal experience from Punjab, India, documented difficulties with partner testing and follow-up, emphasizing continuity across the prevention pathway.[22] A Pakistani scoping review also described wider financial and service constraints affecting thalassemia care.[23] Accessible sampling appointments and clear referral arrangements therefore appeared relevant to translating awareness into completed testing.

The study's strengths included participation across the settings, paired assessment, documented testing, and separate reporting of

willingness and completion. However, the absence of a comparison group, immediate reassessment, and attendance-based recruitment limited causal and population-wide interpretation. A Cochrane review highlighted the limited randomized evidence for preconception genetic risk assessment, reinforcing the need for caution about reproductive outcomes.[24]. The practical contribution was a clearly measured pathway from counseling to testing, with remaining barriers identified for service improvement. This approach also provided community physicians and pediatricians with complementary roles in education, risk explanation, referral, and ongoing support.

CONCLUSION

Structured counseling was followed by improved thalassemia knowledge and greater willingness to undergo carrier screening. Documented testing remained lower than stated willingness, indicating that education needed to be supported by accessible sampling, confidential communication, and follow-up. Integrating these steps within community medicine and pediatric services offered a practical approach to strengthening informed prevention opportunities for families in Punjab.

REFERENCES

1. Taher AT, Musallam KM, Cappellini MD. B-Thalassemias. *N Engl J Med*. 2021;384(8):727-743.
2. Farmakis D, Porter J, Taher A, Cappellini MD, Angastiniotis M, Eleftheriou A. 2021 Thalassaemia International Federation guidelines for the management of transfusion-dependent thalassemia. *HemaSphere*. 2022;6(8):e732.
3. Khaliq S. Thalassemia in Pakistan. *Hemoglobin*. 2022;46(1):12-14.
4. Ghafoor M, Sabar MF, Sabir F. Prevention programmes and prenatal diagnosis for beta thalassemia in Pakistan: a narrative review. *J Pak Med Assoc*. 2021;71(1B):326-331.
5. Durrani SF, Hameed A, Ali R, Thaver IH, Ahmed S. Comparison of knowledge among millennials studying in non-medical universities regarding premarital and prenatal thalassemia screening policies in Pakistan. *J Pak Med Assoc*. 2023;73(11):2232-2234.
6. Saleem N, Anwar A, Shahid NUA, Saleem R, Saleem Z, Asghar H, et al.

- Perception of parents of thalassemic child to thalassemia in Pakistan. *Cureus*. 2021;13(8):e17615.
7. Muhammad L, Wajid KK, Afridi I, Ullah S, Khan A, Muhammad A. Awareness among educated and uneducated parents of beta-thalassemia major patients about antenatal screening. *Cureus*. 2022;14(8):e28561.
8. Ahmed S, Jafri H, Rashid Y, Ehsan Y, Bashir S, Ahmed M. Cascade screening for beta-thalassemia in Pakistan: development, feasibility and acceptability of a decision support intervention for relatives. *Eur J Hum Genet*. 2022;30(1):73-80.
9. Mensah C, Sheth S. Optimal strategies for carrier screening and prenatal diagnosis of α - and β -thalassemia. *Hematology Am Soc Hematol Educ Program*. 2021;2021(1):607-613.
10. Bain BJ, Daniel Y, Henthorn J, de la Salle B, Hogan A, Roy NBA, et al. Significant haemoglobinopathies: a guideline for screening and diagnosis: a British Society for Haematology guideline. *Br J Haematol*. 2023;201(6):1047-1065.
11. Gregg AR, Aarabi M, Klugman S, Leach NT, Bashford MT, Goldwaser T, et al. Screening for autosomal recessive and X-linked conditions during pregnancy and preconception: a practice resource of the American College of Medical Genetics and Genomics (ACMG). *Genet Med*. 2021;23(10):1793-1806.
12. Su Y, Xie J, He J, Shen Y, Li T, Huang W, et al. Screening and treatment of thalassemia. *Clin Chim Acta*. 2025;570:120211.
13. Hossain MS, Islam F, Akhter S, Al Mossabbir A. Thalassemia in Bangladesh: progress, challenges, and a strategic blueprint for prevention. *Orphanet J Rare Dis*. 2025;20(1):358.
14. de Montalembert M, Cappellini MD, Iolascon A, de Franceschi L, Ömur E, Abi Saad M, et al. EHA recommendations for preconceptual and antenatal screening and prenatal diagnosis for hemoglobinopathies. *HemaSphere*. 2026;10(6):e70381.
15. Gul Z, Malik M, Uzair U, Baloch A, La QD, Sadiq N, et al. Awareness about thalassemia among the parents of thalassemic children in Balochistan: a cross-sectional study. *Health Sci Rep*. 2025;8(4):e70715.
16. Hossain MS, Hasan MM, Raheem E, Islam MS, Al Mosabbir A, Petrou M, et al. Lack of knowledge and misperceptions about thalassaemia among college students in Bangladesh: a cross-sectional baseline study. *Orphanet J Rare Dis*. 2020;15(1):54.
17. Alam NE, Islam MS, Khabir MIU, Suriea U, Islam MM, Mohiuddin RB, et al. The scenario of knowledge, attitude and practice of the Bangladeshi population towards thalassemia prevention: a nationwide study. *PLOS Glob Public Health*. 2022;2(10):e0001177.
18. Ahmed S, Jafri H, Faran M, Ahmed WN, Rashid Y, Ehsan Y, et al. Cascade screening for beta-thalassaemia in Pakistan: relatives' experiences of a decision support intervention in routine practice. *Eur J Hum Genet*. 2022;30(4):406-412.
19. Ahmed S, Jafri H, Ahmed WN, Faran M, Rashid Y, Ehsan Y, et al. Healthcare professionals' perceptions of implementing a decision support intervention for cascade screening for beta-thalassemia in Pakistan. *Eur J Hum Genet*. 2022;30(7):795-799.
20. Setiawan H, Li C, Che Hasan MK, Jamaludin TS, Sandi YDL, Firmansyah A, et al. Effectiveness of genetic counseling modalities on knowledge, attitudes, and premarital screening intentions for thalassemia prevention: a multi-site three-arm randomized controlled trial. *Int J Environ Res Public Health*. 2026;23(7):914.
21. Jiang T, Chen SP, Zhao MM, Huang XY, Jiang YH, Lin ZH, et al. Public awareness, knowledge and attitudes toward thalassemia and its screening in five high-prevalence countries: a cross-sectional study based on an online survey. *BMC Public Health*. 2026;26(1):564.
22. Mashon RS, Mandrelle K, Kakkar N. Experience of antenatal thalassemia screening and prenatal diagnosis from a tertiary care teaching hospital in Punjab. *Indian J Hematol Blood Transfus*. 2025;41(1):96-101.
23. Ansari AH, Ansari SH, Salman MJ, Ansari MUH, Jabeen R. A scoping review on the obstacles faced by beta thalassemia major patients in Pakistan—matter of policy investment. *AIMS Public Health*. 2024;11(4):1105-1124.

24. Hussein N, Henneman L, Kai J, Qureshi N. Preconception risk assessment for thalassaemia, sickle cell disease, cystic fibrosis and Tay-Sachs disease. Cochrane Database Syst Rev. 2021;10(10):CD010849.
25. Noor FA, Sultana N, Bhuyan GS, Islam MT, Hossain M, Sarker SK, et al. Nationwide carrier detection and molecular characterization of β -thalassemia and hemoglobin E variants in Bangladeshi population. Orphanet J Rare Dis. 2020;15(1):15.