INSIGHTS-JOURNAL OF
HEALTH AND REHABILITATION

INSIGHTS-JHR
INSIGHTS-JOURNAL OF HEALTH AND REHABILITATION

# EVALUATING CURRENT ATTITUDES REGARDING PARENTAL GENETIC TESTING FOR BETA THALASSEMIA AMONG PREGNANT WOMEN IN PAKISTAN

*Original Research*

**Iqra javaid[1]*, Abdul Mannan[1], Sabiha Khalid[2], Aroosa Farooq[3], Muhammad Danish[4], Rabia Basri[3], Muhammad Akram[5], Maheen Farooqi[6]**
[1]Punjab Thalassaemia & Other Genetic Disorders Prevention and Research Institute, Pakistan.
[2]Bs(hons) Biotechnology, University of Veterinary & Animal Sciences, Pakistan.
[3]M. Phil Human Genetics & Molecular Biology at University of Health Sciences. Pakistan.
[4]BSN Generic Student, Shahida Islam Nursing College, Lodhran, Pakistan.
[5]Bds.Mds Oral and Maxilofical surgeon Zaheer Hospital sheikhupura, Pakistan.
[6]Stanford University, USA.
**Corresponding Author:** Iqra javaid, Punjab Thalassaemia & Other Genetic Disorders Prevention and Research Institute, Pakistan. javaidiqra77@gmail.com
**Acknowledgement:** The authors thank the Pakistani Society of Medical Genetics and the participating patients for their cooperation.

Conflict of Interest: None
Grant Support & Financial Support: None

## ABSTRACT

**Background:** Thalassemia is one of the most common inherited hemoglobinopathies, resulting from reduced or absent synthesis of alpha (α) or beta (β) globin chains. In countries like Pakistan, where consanguineous marriages are common, the risk of transmitting such genetic disorders increases. Prenatal genetic testing, particularly chorionic villus sampling (CVS), offers early diagnosis but may contribute to maternal psychological distress, which remains underexplored in local populations.

**Objective:** To assess the knowledge, attitudes, and psychological responses of pregnant Pakistani women undergoing CVS for thalassemia screening.

**Methods:** This cross-sectional study was conducted between April and June 2024 at the Thalassemia Department of Bahawalpur Victoria Hospital, Punjab, Pakistan. A total of 48 pregnant women, between 11–14 weeks of gestation and confirmed thalassemia carriers, were enrolled. Data were collected using a structured, interviewer-administered questionnaire covering demographic details, clinical history, knowledge of thalassemia, attitudes toward CVS, and psychological experiences. Descriptive statistics were used to summarize responses, and ordinal logistic regression was performed to explore associations between gestational age, emotional responses, and willingness to undergo testing.

**Results:** Among the 48 participants, 54.2% expressed willingness to undergo CVS again in future pregnancies. A majority (72.9%) reported favorable attitudes toward genetic testing. About 60.4% cited active partner support during the diagnostic process. Women with previously affected children showed greater acceptance of CVS. Sleep disturbances and anxiety were more pronounced in early gestation, while overall psychological strain remained minimal due to familial and clinical support systems.

**Conclusion:** Prenatal CVS testing was generally well-tolerated among thalassemia-carrier pregnant women. Support from partners, family, and healthcare providers played a pivotal role in alleviating maternal anxiety and enabling informed decision-making.

**Keywords:** Anxiety and Depression, Genetic Counselling, Genetic Disorder, Hemoglobin, Pregnant Women, Prenatal Testing, Thalassemia.

INSIGHTS-JOURNAL OF
HEALTH AND REHABILITATION

INSIGHTS-JHR
INSIGHTS-JOURNAL OF HEALTH AND REHABILITATION

## INTRODUCTION

Thalassemia is a hereditary blood disorder inherited in an autosomal recessive manner, characterized by the impaired synthesis of hemoglobin chains—either alpha or beta—leading to ineffective erythropoiesis and chronic anemia. Globally, thalassemia affects approximately 4% of newborns among every 10,000 births, while in Pakistan, it poses a substantial public health burden, with an estimated 5–7% of the population acting as asymptomatic carriers (1–3). In the absence of early diagnosis and intervention, affected individuals face lifelong dependence on blood transfusions and iron chelation therapy. Advances in prenatal diagnostics, including procedures such as chorionic villus sampling (CVS) and amniocentesis, have made it possible to detect such genetic anomalies during early pregnancy, typically around 12 to 13 weeks of gestation (4,5). These procedures, although valuable, are associated with a miscarriage risk ranging from 0.2% to 1%, and they frequently provoke heightened levels of maternal anxiety and psychological distress (6,7). Concerns about potential fetal anomalies, the anticipation of diagnostic results, and the perceived invasiveness of testing contribute to a significant emotional toll on expectant mothers (8). Cultural and religious beliefs also deeply influence how prenatal testing is perceived and accepted, particularly in countries like Pakistan where Islamic values guide ethical decisions regarding reproduction and termination of pregnancy. In such settings, pregnancy termination may be considered permissible before the 120th day of gestation, corresponding with the Islamic concept of ensoulment, or in circumstances where maternal life is jeopardized (9,10). Despite the availability of prenatal diagnostic tools, their utilization remains uneven, hindered by limited public awareness, social stigma, and inconsistent healthcare infrastructure (11).

The role of parental carrier screening becomes crucial in identifying at-risk pregnancies, guiding reproductive choices, and minimizing the birth of children with thalassemia (12). Genetic counseling services serve as an essential component of this process, offering couples scientifically accurate, culturally sensitive information that supports informed, value-aligned decision-making (13). Furthermore, studies have shown that women are often motivated to pursue prenatal testing due to a strong desire for clarity regarding fetal health, a generally favorable attitude toward testing, and, in some cases, a willingness to consider termination following adverse findings (14). However, the psychological impact of ambiguous or inconclusive results is significant. Such uncertainty may lead to sustained anxiety, prompting debates around whether incomplete or non-actionable results should be disclosed at all, given the emotional burden they may impose and the ethical implications of misleading autonomy (15,16). Given the complex interplay of psychological, cultural, and ethical considerations, it becomes imperative to understand how pregnant women in Pakistan perceive and respond to prenatal testing—especially CVS in the context of thalassemia risk. This study aims to explore the knowledge, attitudes, and decision-making processes of expectant mothers undergoing prenatal testing, with a specific focus on psychological responses to CVS in thalassemia screening. The objective is to generate evidence that can inform more patient-centered genetic counseling approaches within resource-constrained, culturally diverse settings.

## METHODS

This cross-sectional study was conducted to assess pregnant women's attitudes toward prenatal genetic testing for thalassemia. The research was carried out at the Thalassaemia Department of Bahawalpur Victoria Hospital (BVH) between April 1, 2024, and June 30, 2024. Ethical approval for the study was obtained from the Institutional Review Board of Bahawalpur Victoria Hospital and informed verbal and written consent was obtained from all participants prior to enrollment, ensuring voluntary participation and data confidentiality. Participants were recruited from among pregnant women who were confirmed carriers of thalassemia and were scheduled for prenatal diagnostic testing via Chorionic Villus Sampling (CVS) during their 11th to 14th week of gestation. These women were identified through hospital records and approached after their initial visit at 8–10 weeks of gestation. Eligibility criteria included being currently pregnant with a gestational age between 11 and 14 weeks, confirmed carrier status of thalassemia, and having a partner also screened for carrier status. Women who were not currently pregnant, had previously undergone prenatal testing in earlier pregnancies, or were single mothers were excluded from the study. A hospital administrative assistant generated a list of eligible couples, which was used by genetic counselors to contact families telephonically. Counselors explained the objectives, procedures, and potential benefits of the study in culturally appropriate and sensitive language. Prenatal CVS testing was offered free of cost to all eligible female carriers at the hospital. Couples were also counseled about the 25% risk of disease inheritance when both parents are carriers.

INSIGHTS-JHR
INSIGHTS-JOURNAL OF HEALTH AND REHABILITATION

A structured questionnaire was designed and administered to participants in person. The questionnaire comprised four sections. The first section included fourteen demographic questions covering age, marital status, education level, and consanguinity, with categorical options such as “under 20,” “20–29,” etc. The second section assessed clinical characteristics, such as the number of children affected, living or deceased offspring, and history of fetal death, using categorical variables. The third section explored participants’ knowledge, awareness, and attitudes toward prenatal diagnosis, employing a Likert scale to measure familiarity and attitudes. The final section included seven questions evaluating women's psychological responses, such as anxiety, sleep disturbances, and concern about testing, using multiple-choice and scaled responses (15-17). Data were reviewed for completeness and entered using a computerized database. Statistical analysis was performed using the Statistical Package for the Social Sciences (SPSS), version 24. Descriptive statistics, including frequencies and percentages, were used to summarize demographic, clinical, and attitudinal data. Two ordinal logistic regression models were developed to investigate factors influencing participants’ decision-making and psychological outcomes. Model 1 evaluated predictors of willingness to undergo CVS, including the number of affected children, prior history of prenatal testing, gestational age, and education level. Model 2 assessed psychological disturbances (e.g., sleep disturbance, agitation, sensitivity to criticism) in relation to gestational age. Model fit was assessed using -2 Log Likelihood values, Chi-square statistics for model improvement, and pseudo R-squared indices, including Cox and Snell, Nagelkerke, and McFadden values. Parameter estimates, odds ratios (OR), and 95% confidence intervals (CI) were calculated. Statistical significance was set at $p < 0.05$. In Model 1, Cox and Snell, Nagelkerke, and McFadden values were 0.350, 0.468, and 0.312 respectively, indicating that approximately 35% of the variability in willingness to undergo CVS could be explained by covariates such as fetal death, education, and prior testing history.

## RESULTS

A total of 79 couples were contacted for participation, of which 48 pregnant women met the inclusion criteria and consented to be enrolled in the study. All participants were between 11 and 14 weeks of gestation at the time of enrollment. The majority of the women were aged between 20–29 years (47.9%) and 30–39 years (47.9%), with only 2.1% each under 20 years or aged 40 and above. All participants were married, and 70.8% reported consanguineous marriages with first cousins. Regarding education level, 50.0% had less than primary education, while only 10.4% held a bachelor’s or master’s degree. The predominant ethnic group was Saraiki (64.6%), followed by Punjabi (37.1%). All pregnancies were singleton, with 91.7% of women falling between 12–20 weeks of gestation. A history of fetal death was reported by 39.6% of the participants, and 18.8% had at least one deceased child. The majority had at least one thalassemia-affected child, with 60.4% reporting one affected child, 14.6% having two, and 2.1% reporting three or more. Only 22.9% had no affected children. Regarding living offspring, 31.3% had one or two living children each, while 6.3% had none. Concerning familiarity with prenatal genetic screening, only 12.5% were strongly familiar, while 45.8% reported slight familiarity. Despite limited procedural awareness, 81.3% strongly agreed that knowing the fetal beta-thalassemia status before birth was important. Willingness to undergo CVS again in future pregnancies was observed in 54.2%, while 45.8% declined. Emotional responses varied: 22.9% experienced continuous fatigue, 27.1% reported panic attacks, and 22.9% felt sleeplessness related to the procedure. A total of 43.8% managed stress by talking with partners or family, whereas 16.7% sought professional counseling.

Partner support was confirmed by 60.4% of the women. Concerns regarding the accuracy and reliability of testing were voiced by 47.9%, while 12.5% feared potential pregnancy risks, and 10.4% cited emotional or psychological impacts. Appetite changes attributed to CVS-related stress were reported in 33.3% of participants. Complications following CVS included miscarriage in 16.7% and pregnancy loss followed by recovery in 12.5%. Ordinal regression analysis revealed that willingness to undergo CVS was significantly associated with the absence of previously affected children (estimate = 11.387, $p = 0.000$) and the presence of one affected child (estimate = 11.387, $p = 0.000$). The model fit was acceptable with Cox and Snell $R^2 = 0.350$, Nagelkerke $R^2 = 0.468$, and McFadden $R^2 = 0.312$. In the second model, early gestational age (<10 weeks) was negatively associated with sleep disturbances and stress (estimate = -48.501, $p = 0.000$). Slight agitation (estimate = -40.909, $p = 0.000$) and sensitivity to criticism (estimate = -2.701, $p = 0.045$) also showed negative associations with CVS willingness. Model 2 demonstrated good explanatory power (Nagelkerke $R^2 = 0.791$). Regarding decision-making, 79.2% of couples reported discussing screening results with a clear plan of action. The most cited factors influencing decisions to proceed with screening included medical advice (39.6%), religious beliefs (10.4%), and emotional readiness (4.2%). Additionally, 47.9% desired professional counseling or therapy as their preferred form of support, while 27.1% sought emotional support from family and friends.

INSIGHTS-JHR
INSIGHTS-JOURNAL OF HEALTH AND REHABILITATION

**Table 1: Demographic characteristics of the study participants (N = 48)**

| Studied Variables | Groups | Frequency (%) |
|---|---|---|
| Age (Years) | Under 20 | 01 (2.1) |
| | 20-29 | 23 (47.9) |
| | 30-39 | 23 (47.9) |
| | 40 and above | 01 (2.1) |
| Marital status | Married | 48 (100) |
| | Divorced | - |
| Highest level of education | Less than Primary School | 24 (50.0) |
| | Primary School | 09 (18.8) |
| | High School | 05 (10.4) |
| | College | 05 (10.4) |
| | Bachelor degree | 02 (4.2) |
| | Master degree | 02 (4.2) |
| | Doctorate | 01 (2.1) |
| Consanguinity | First Cousin | 34 (70.8) |
| | Unrelated | 11 (22.9) |
| | Second cousin | 03 (6.3) |
| Ethnicity | Saraiki | 31 (64.6) |
| | Punjabi | 13 (37.1) |
| | Balochi | 02 (4.2) |
| | Pushto | 01 (2.1) |

**Table 2: Clinical characteristics and number of offspring affected, alive, deceased, and fetal death**

| Studied Variables | Groups | Frequency (%) | N |
|---|---|---|---|
| Pregnancy | Single | 100 | 48 |
| | Twins | - | 0 |
| Gestational age | Less than 1 week | 4.2 | 2 |
| | 12-20 weeks | 91.7 | 44 |
| | 21-30 weeks | 4.2 | 2 |
| Affected children | None | 22.9 | 11 |
| | One affected | 60.4 | 29 |
| | Two affected | 14.6 | 7 |
| | Three or more affected | 2.1 | 1 |
| Number of offspring alive | None | 6.3 | 3 |
| | One | 31.3 | 15 |
| | Two | 31.3 | 15 |
| | Three | 20.8 | 10 |
| | Four | 6.3 | 3 |
| | Five | 4.2 | 2 |
| Number of offspring deceased | None | 66.7 | 32 |
| | One | 18.8 | 9 |
| | Two | 12.5 | 6 |
| | Three | 2.1 | 1 |
| Fetal death in utero | None | 60.4 | 29 |

INSIGHTS-JHR
INSIGHTS-JOURNAL OF HEALTH AND REHABILITATION

| Studied Variables | Groups | Frequency (%) | N |
|---|---|---|---|
| | One | 22.9 | 11 |
| | Two | 6.3 | 3 |
| | Three | 8.3 | 4 |
| | Four | 2.1 | 1 |

**Table 3: Assessment of parental attitudes regarding prenatal genetic screening**

| Statements | Disagreement | Slightly | Moderately | Strongly/ Yes |
|---|---|---|---|---|
| How familiar are you with prenatal genetic screening procedures? | 9 (18.8%) | 22(45.8%) | 11 (2.9%) | 6 (12.5%) |
| How familiar are you with prenatal genetic screening procedures? | 5 (10.4%) | 21(43.8%) | 17 (35.4%) | 5 (10.4%) |
| How important is it to you to know if your baby has beta thalassemia before birth? | 2 (4.2%) | 7(14.6%) | - | 39 (81.3%) |
| Do you consider prenatal genetic screening important for beta-thalassemia disorder? | 1 (2.1%) | 7(14.6%) | 20 (41.7%) | 20 (41.7%) |
| Do you feel good about CVS test is good or not for diagnosing thalassemia disorder in the upcoming fetus? | 4 (8.3%) | 22(45.8%) | 5 (10.4%) | 17 (35.4%) |
| Have you ever felt agitated about the CVS test attempt or not? | 27(56.3%) | 10(20.8%) | 5 (10.4%) | 6 (12.5%) |
| Are you sensitive to the criticism or feedback of others regarding CVS procedure or beta thalassemia diagnosis? | 21(43.8%) | 10(20.8%) | 11 (22.9%) | 6 (12.5%) |
| Do you feel continuous fatigue before or after CVS? | 35 (72.9%) | 1 (2.1%) | 1 (2.1%) | 11 (22.9%) |
| Do you feel sleepless due to the upcoming CVSprocedure? | 34 (70.8%) | 1 (2.1%) | 2(4.2%) | 11 (22.9%) |
| Are you comfortable about the potential outcomes of prenatal genetic screening, such as further diagnostic tests? | 3(6.3%) | 1(020.8%) | 24(50.0%) | 11(22.9%) |
| Do you have any panic attacks just because of the CVS test | 35 (72.9%) | - | - | 13 (27.1%) |
| Are your partner is supportive about the genetic screening and potential results? | 4(8.4%) | 9(18.8%) | 6(12.5%) | 29(60.4%) |
| Are you agreeing to attempt a CVS test again for your upcoming fetus? | 22(45.8%) | - | - | 26(54.2%) |
| Did you feel morning sickness just because of the CVS procedure? | 14 (29.2%) | - | - | 34(70.8%) |
| Have Extended Family Screening done? | 18(37.5%) | - | - | 30(62.5%) |

**Table 4: Model Fitting Information of Ordinal Regression**

| | Model ftting criteria | Likelihood ratio tests | | | | Model ftting criteria | Likelihood ratio tests | | |
|---|---|---|---|---|---|---|---|---|---|
| Model 1 | -2 Log Likelihood | Chi-Square | Df | p value | Model 2 | -2 Log Likelihood | Chi-Square | Df | p value |
| Intercept Only | 57.444 | - | - | -- | Intercept Only | 30.309 | - | - | - |
| Final | 36.733 | 20.711 | 6 | 0.002 | Final | 6.287 | 24.022 | 5 | 0.000 |
| Pseudo R-Square | | | | | Pseudo R-Square | | | | |

| | Model ftting criteria | Likelihood ratio tests | | Model ftting criteria | Likelihood ratio tests |
|---|---|---|---|---|---|
| Cox and Snell | Nagelkerke | McFadden | Cox and Snell | Nagelkerke | McFadden |
| 0.350 | 0.468 | 0.312 | 0.394 | 0.791 | 0.726 |

**Table 5: Parameter Estimates of theOrdinal Logistic Regression with willness to attempted CVS test asthe dependent variable.**
**Model 1**

| Predictors | Estimate | P-Value | 95% Confidence Interval | |
|---|---|---|---|---|
| | | | Lower Bound | Upper Bound |
| No Affected Children | 11.387 | .000 | 9.192 | 13.583 |
| One Affected Child | 11.387 | .000 | 10.781 | 14.738 |

**Model 2: Attempted CVS test asthe dependent variable.**

| Predictors | Estimate | P-Value | 95% Confidence Interval | |
|---|---|---|---|---|
| | | | Lower Bound | Upper Bound |
| Sleepless due to upcoming CVS test | 2.028 | 0.079 | -.236 | 4.293 |
| Slightly Agitated | -40.909 | 0.000 | -45.397 | -36.422 |
| Moderate Agitation | -43.279 | . | -43.279 | -43.279 |
| Highly Agitated | -21.536 | . | -21.536 | -21.536 |

**Table 6: Responses of Pregnant Women related to Prenatal Genetic Testing. Data are presented as number and percentage. (An in-depth examination)**

| **Serial No.** | **Attitude and concerns toward Prenatal Genetic Testing and Thelassemia Disorder** | **Response of Women N (%)** |
|---|---|---|
| 1- | What concerns do you have about prenatal genetic screening for beta-thalassemia disorder? | |
| | Accurately and reliability of the test | 23 (47.9) |
| | Potential risks to the pregnancy | 06 (12.5) |
| | Emotional and psychological impact | 05 (10.4) |
| | Accuracy and reliability of the test, potential risks to the pregnancy, emotional and psychological impact, and financial cost | 01 (2.1) |
| | Emotional and psychological impact and financial cost | 01 (2.1) |
| | Accuracy and reliability of the test and emotional and psychological impact | 01 (2.1) |
| | Accuracy and reliability of the test, emotional and psychological impact, and financial cost | 01 (2.1) |
| | Accuracy and reliability of the test and financial cost | 08 (16.7) |
| | Potential risks to the pregnancy and financial cost | 01 (2.1) |
| | Accuracy and reliability of the test and potential risks to the pregnancy | 01 (2.1) |
| 2- | Any complications because of the CVS test? | |
| | Got complications regarding pregnancy loss but recovered by gynecologist | 06 (12.5) |
| | Got miscarriage by this test | 08 (16.7) |

| Serial No. | Attitude and concerns toward Prenatal Genetic Testing and Thelassemia Disorder | Response of Women N (%) |
|---|---|---|
| | Got a miscarriage due to depression or some careless things | 04 (8.3) |
| | Not attempt this test | 22 (45.8) |
| | No complications | 08 (16.7) |
| 3- | Do you feel any changes in appetite due to the CVS procedure? | |
| | Overeating | 05 (10.4) |
| | Lack of food intake | 11 (22.9) |
| | Normally taking meal | 30 (62.5) |
| | Sometimes overeating and sometimes lacking in food | 02 (4.2) |
| 4- | How do you manage stress and anxiety related to your pregnancy and the potential genetic disorder? | |
| | Talking to my partner/family/friends | 21 (43.8) |
| | Seeking professional counseling or therapy | 08 (16.7) |
| | Keeping busy with work or hobbies | 10 (20.8) |
| | Other | 01 (2.1) |
| | Seeking professional counseling or therapy and keeping busy with work or hobbies | 03 (6.3) |
| | Talking to my partner/family/friends and keeping busy with work or hobbies | 01 (2.1) |
| | Talking to my partner/family/friends and Using relaxation techniques (e.g., meditation, yoga) | 01 (2.1) |
| | Keeping busy with work or hobbies and do not share with anyone | 01 (2.1) |
| | Talking to my partner, using relaxation techniques, and keeping busy with work or hobbies | 01 (2.1) |
| | Sleep | 01 (2.1) |
| 5- | What kind of support do you feel you need during this time (e.g., emotional support, information, counseling)? | |
| | Emotional support from family and friends | 13 (27.1) |
| | Professional counseling or therapy | 23 (47.9) |
| | Detailed information and resources about beta thalassemia | 03 (6.3) |
| | Emotional support from family and friends and professional counseling or therapy | 05 (10.4) |
| | Professional counseling or therapy and detailed information and resources about beta thalassemia | 02 (4.2) |
| | Emotional support from family and friends and detailed information and resources about beta thalassemia | 01 (2.1) |
| | Professional counseling or therapy and Financial | 01 (2.1) |
| 6- | Have you and your partner discussed the implications of the screening results? If so, what was the outcome of that discussion? | |
| | Yes, and we have a clear plan of action | 38 (79.2) |
| | No, we haven't discussed it yet | 03 (6.3) |
| | We discussed it but didn't reach a conclusion | 07 (14.6) |
| 7- | What factors will influence your decision to proceed with the screening or any further steps if beta thalassemia is detected? | |
| | Medical advice from healthcare providers | 19 (39.6) |
| | Emotional readiness and personal beliefs | 02 (4.2) |
| | Support from family and friends | 04 (8.3) |

INSIGHTS-JHR
INSIGHTS-JOURNAL OF HEALTH AND REHABILITATION

| Serial No. | Attitude and concerns toward Prenatal Genetic Testing and Thelassemia Disorder | Response of Women N (%) |
|---|---|---|
| | Ethical or religious beliefs | 05 (10.4) |
| | Medical advice from healthcare providers and financial considerations and affordability | 03 (6.3) |
| | Financial considerations and affordability and support from family and friends | 02 (4.2) |
| | Emotional readiness personal beliefs andsupport from family and friends | 01 (2.1) |
| | Medical advice from healthcare providers, emotional readiness, financial consideration, religious beliefs | 01 (2.1) |
| | Medical advice from healthcare providers and ethical or religious beliefs | 03 (6.3) |
| | Medical advice from healthcare providers, support from family and friends, and ethical or religious beliefs | 02 (4.2) |
| | Medical advice from healthcare providers and emotional readiness and personal beliefs | 01 (2.1) |
| | Medical advice from healthcare providers and support from family and friends | 01 (2.1) |
| | Massive blood transfusions | 01 (2.1) |
| | Medical advice from healthcare providers and cannot bear the sickness of a child | 01 (2.1) |
| | Financial considerations and affordability and confused | 01 (2.1) |
| | Emotional readiness and personal beliefs and confused | 01 (2.1) |

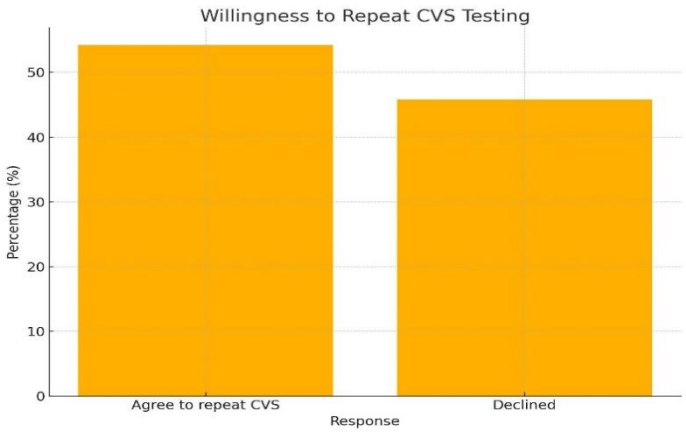


*Figure 1Willingness to Repeat CVS Testing*

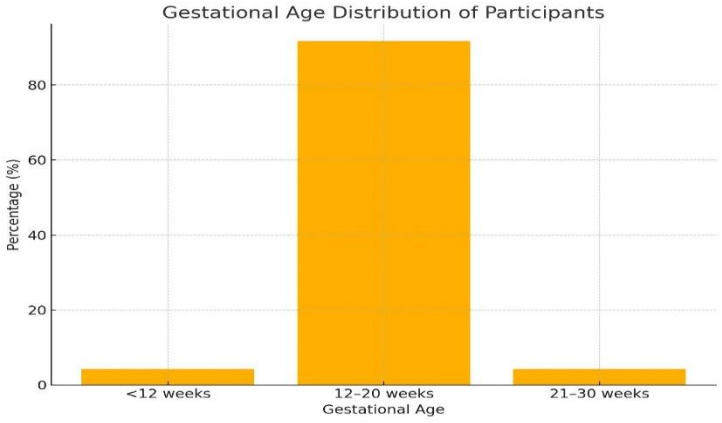


*Figure 2 Gestational Age Distribution of Participants*

## DISCUSSION

This study explored the psychological and emotional dimensions associated with prenatal chorionic villus sampling (CVS) testing among pregnant women, particularly focusing on anxiety, societal influences, and partner involvement. The findings affirm the central role of psychological stressors, such as sleeplessness, agitation, and emotional burden, particularly during the first trimester when most fetal anomalies are typically detected. These observations are consistent with prior research highlighting increased maternal anxiety during early gestation due to the anticipation of adverse fetal outcomes (13). A modest but significant association between gestational age and anxiety-related symptoms reinforces the need for targeted support during this vulnerable period. A notable outcome was the widespread parental support for prenatal testing, with 81% of women favoring early identification of beta-thalassemia. This response mirrors previous findings where early diagnosis is viewed as a gateway to preparing for high-risk pregnancies and managing complex neonatal outcomes (14,15). The willingness of 54.2% of women to consider CVS in future pregnancies highlights an openness to informed reproductive decision-making, although the remaining 45.8% expressed reluctance, underscoring the need for personalized counseling

INSIGHTS-JHR
INSIGHTS-JOURNAL OF HEALTH AND REHABILITATION

approaches to address unresolved fears and misconceptions (16). Importantly, support from healthcare professionals emerged as a key mitigating factor in reducing maternal distress. The beneficial impact of structured, clinic-based counseling, as opposed to informal home-based support, aligned with existing literature reporting improved psychological outcomes when professional guidance is provided during invasive prenatal testing (17). This reinforces the utility of integrating early genetic education into routine antenatal care to mitigate anxiety and improve informed consent processes.

The experience of previously managing thalassemia-affected children significantly influenced parental attitudes toward testing. Women with one or more affected offspring displayed greater acceptance of CVS, likely shaped by firsthand encounters with challenges such as transfusion dependency and chronic health complications (18). However, when diagnostic outcomes were uncertain or complex, hesitancy increased. This reaction echoes earlier research where couples reported emotional overwhelm and reluctance to process detailed medical information prior to definitive results (19,20). Despite receiving preliminary counseling, inconsistent understanding was evident, especially in families with multiple affected children, pointing toward the need for tailored and literacy-sensitive counseling formats. One of the study's distinct findings was the powerful influence of partner support, particularly in low-income, rural communities like southern Punjab. Women who reported emotional backing from their partners demonstrated improved psychological resilience, regardless of educational background. These findings are supported by previous studies that emphasize protective buffering mechanisms and emotional co-regulation among couples undergoing prenatal genetic testing (21,22). In this cohort, 79.2% of women had proactive discussions with their partners regarding testing outcomes, reflecting a strong role of shared decision-making in emotionally stressful scenarios. Despite many women adapting well emotionally, a subset experienced pronounced psychological symptoms, including trauma-like responses. Low educational attainment was a consistent factor associated with concealment of abnormal results and resistance to extended family screening, patterns also documented in similar socio-cultural settings globally (23). These attitudes may stem from stigma, lack of genetic literacy, or fear of social repercussions, suggesting that educational interventions need to be culturally contextualized and accessible.

The study's strengths lie in its focus on a highly specific population—pregnant women undergoing CVS testing in a tertiary care setting—allowing for an in-depth exploration of psychosocial and behavioral factors. Additionally, the use of ordinal regression models added analytical rigor in identifying predictors of willingness and emotional outcomes. However, several limitations must be acknowledged. The sample size was modest, potentially affecting generalizability. The reliance on self-reported data introduces the possibility of recall and response biases. Furthermore, the study exclusively included women who consented to CVS, thereby omitting perspectives of those who declined testing, which could have enriched the understanding of barriers to acceptance. Future research should consider longitudinal designs to track psychological outcomes from pre-test counseling through postpartum, and include male partners and those who opt out of invasive diagnostics. Expanding the sample to encompass diverse geographic and socio-economic groups would enhance the applicability of findings. Incorporating psychometric tools for validated anxiety and stress assessment would also provide greater depth to emotional response analysis. In conclusion, the study highlights the multidimensional nature of psychological experiences during prenatal CVS testing in thalassemia carrier pregnancies. While medical, cultural, and social factors intersect to shape parental decision-making, the critical role of early counseling, professional support, and partner involvement cannot be overstated in promoting maternal well-being and informed reproductive choices.

## CONCLUSION

This study concluded that prenatal genetic testing through chorionic villus sampling (CVS) did not result in lasting psychological strain when adequate emotional and professional support was available. The presence of a supportive partner, family involvement, and access to medical counseling significantly helped in alleviating maternal anxiety. Counseling emerged as the most effective intervention, not only enhancing awareness about thalassemia as a genetic disorder but also empowering pregnant women to make informed, value-based decisions. These findings underscore the importance of integrating genetic counseling services into routine antenatal care and highlight the critical need for broader public health education to encourage timely psychological support for expectant mothers facing difficult reproductive choices.

**AUTHOR CONTRIBUTION**

| Author | Contribution |
|---|---|
| Iqra Javaid* | Substantial Contribution to study design, analysis, acquisition of Data<br>Manuscript Writing<br>Has given Final Approval of the version to be published |
| Abdul Mannan | Substantial Contribution to study design, acquisition and interpretation of Data<br>Critical Review and Manuscript Writing<br>Has given Final Approval of the version to be published |
| Sabiha Khalid | Substantial Contribution to acquisition and interpretation of Data<br>Has given Final Approval of the version to be published |
| Aroosa Farooq | Contributed to Data Collection and Analysis<br>Has given Final Approval of the version to be published |
| Muhammad Danish | Contributed to Data Collection and Analysis<br>Has given Final Approval of the version to be published |
| Rabia Basri | Substantial Contribution to study design and Data Analysis<br>Has given Final Approval of the version to be published |
| Muhammad Akram | Contributed to study concept and Data collection<br>Has given Final Approval of the version to be published |
| Maheen Farooqi | Writing - Review & Editing, Assistance with Data Curation |

## REFERENCES

1. Yousuf, R., Akter, S., Wasek, S. M., Sinha, S., Ahmad, R., Haque, M., & Salam, M. W. (2022). Thalassemia: A Review of the Challenges to the Families and Caregivers. Cureus, 14(12).
2. Khaliq, S. (2022). Thalassemia in Pakistan. Hemoglobin, 46(1), 12-14.
3. Babker, A. M. (2022). An overview on thalassemia and challenges during COVID-19. International Journal of Health Sciences, (I), 3207-3220.
4. Tesio, N., & Bauer, D. E. (2023). Molecular basis and genetic modifiers of thalassemia. Hematology/oncology clinics of North America, 37(2), 273-299.
5. M. M., Molina, F. S., Rodríguez-Fernández, M., Delgado, J. L., Carrillo, M. P., Jani, J., Plasencia, W., Stratieva, V., Maíz, N., Carretero, P., Lismonde, A., Chaveeva, P., Burgos, J., Santacruz, B., Zamora, J., & De PacoMatallana, C. (2020). New approach for estimating risk of miscarriage after chorionic villus sampling. Ultrasound in obstetrics &gynecology: the official journal of the International Society of Ultrasound in Obstetrics and Gynecology, 56(5), 656–663.
6. Likar, I. P., Jere, K. S., Možina, T., Verdenik, I., & Tul, N. (2020). Pregnancy Loss After Amniocentesis and Chorionic Villus Sampling: Cohort Study. Zdravstveno varstvo, 60(1), 25–29.
7. Hekmat, K., Kamali, F., Abedi, P., & Afshari, P. (2023). Explaining mothers' experiences of performing fetal health screening tests in the first trimester of pregnancy: A qualitative study with content analysis approach. International Journal of Africa Nursing Sciences, 18, 100565.
8. Gibson, L., Ikhile, D., Nyashanu, M., & Musoke, D. (2023). Health promotion research in international settings: A shared ownership approach for north-south partnerships. In Global Handbook of Health Promotion Research, Vol. 3: Doing Health Promotion Research (pp. 263-272). Cham: Springer International Publishing.
9. Garcia, E., Henneman, L., Gitsels-van der Wal, J. T., Martin, L., Koopmanschap, I., Bekker, M. N., & Timmermans, D. R. (2022). Non-invasive prenatal testing (NIPT) and pregnant women's views on good motherhood: a qualitative study. European Journal of Human Genetics, 30(6), 669-675.
10. Mojahed, S., Tabatabaei, R. S., Reihani, F., Dehghani, A., & Khavari, F. (2021). The effect of education on anxiety of pregnant mothers before amniocentesis. Journal of education and health promotion, 10(1), 61.
11. Sheikh, Z. A., & Wahlberg, A. (2023). More than sample providers: how genetic researchers in Pakistan mobilized a prenatal diagnostic service for thalassemia. BioSocieties, 18(1), 197-217.

12. Bedei, I., Wolter, A., Weber, A., Signore, F., & Axt-Fliedner, R. (2021). Chances and challenges of new genetic screening technologies (NIPT) in prenatal medicine from a clinical perspective: a narrative review. Genes, 12(4), 501.
13. Di Mattei, V., Ferrari, F., Perego, G., Tobia, V., Mauro, F., & Candiani, M. (2021). Decision-making factors in prenatal testing: A systematic review. Health psychology open, 8(1), 2055102920987455.
14. Mensah, C., & Sheth, S. (2021). Optimal strategies for carrier screening and prenatal diagnosis of α-and β-thalassemia. Hematology, 2021(1), 607-613.
15. Ngan, O. M. Y., Yi, H., Bryant, L., Sahota, D. S., Chan, O. Y. M., & Ahmed, S. (2020). Parental expectations of raising a child with disability in decision-making for prenatal testing and termination of pregnancy: A mixed methods study. Patient Education and Counseling, 103(11), 2373-2383.
16. Farrell RM, Pierce M, Collart C, Yao M, Coleridge M, Chien EK, et al. Decision-making for prenatal genetic screening: how will pregnant women navigate a growing number of aneuploidy and carrier screening options? BMC Pregnancy Childbirth. 2021;21(1):806.
17. Farrell, R. M., Pierce, M., Collart, C., Yao, M., Coleridge, M., Chien, E. K., ... & Edmonds, B. T. (2021). Decision-making for prenatal genetic screening: how will pregnant women navigate a growing number of aneuploidy and carrier screening options. BMC Pregnancy and Childbirth, 21, 1-10.
18. Liu D, Nong X, Lai F, Nong C, Wang T, Tang Y. Noninvasive Prenatal Diagnosis of SEA-Thalassemia by Combining 1000 Genomes Database and Relative Haplotype Dosage. Hemoglobin. 2024;48(2):71-8.
19. Charoenkwan P, Traisrisilp K, Sirichotiyakul S, Phusua A, Sanguansermsri T, Tongsong T. Noninvasive Prenatal Diagnosis of Beta-Thalassemia Disease by Using Digital PCR Analysis of Cell-Free Fetal DNA in Maternal Plasma. Fetal Diagn Ther. 2022;49(11-12):468-78.
20. Erlich HA, Ko L, Lee J, Eaton K, Calloway CD, Lal A, et al. Non-invasive prenatal testing of beta-hemoglobinopathies using next generation sequencing, in-silico sequence size selection, and haplotyping. Croat Med J. 2024;65(3):180-8.
21. Madgett TE. First Trimester Noninvasive Prenatal Diagnosis of Maternally Inherited Beta-Thalassemia Mutations. Clin Chem. 2022;68(8):1002-4.
22. Jiang H, Zhou C, Yang L, Li S, Liu X. [Carrier screening and prenatal diagnosis for thalassemia-associated mutations in Jiaxing area of Zhejiang]. Zhonghua Yi Xue Yi Chuan Xue Za Zhi. 2023;40(3):295-300.
23. Wu J, Xie D, Wang L, Kuang Y, Luo S, Ren L, et al. Application of third-generation sequencing for genetic testing of thalassemia in Guizhou Province, Southwest China. Hematology. 2022;27(1):1305-11.