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Clinical and Reproductive Implications of Beta Thalassemia Trait in Women: Insights from a Diagnostic Screening Program in Chandigarh, India

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Abstract

Background: Beta Thalassemia Trait (BTT) is a common genetic disorder in India, typically presenting as a clinically silent condition. However, its implications for women's reproductive health remain underexplored.

Objective: This study investigates the clinical and reproductive profiles of women diagnosed with BTT during a diagnostic screening program in Chandigarh, with the goal of assessing the potential health and reproductive risks associated with the trait.

Methods: A cross-sectional study was conducted among 35 women identified with BTT out of a cohort of 500 patients screened for anemia. Data were collected on clinical symptoms, hematological parameters, and reproductive history. Diagnosis was confirmed using complete blood count, peripheral blood smear, serum ferritin, and High-Performance Liquid Chromatography (HPLC). Descriptive statistical analysis was performed.

Results: The majority of participants were women of reproductive age (26–34 years), with 65.7% being asymptomatic. Mild symptoms such as fatigue and pallor were reported by one-third of the participants. Hematological profiles revealed microcytic hypochromic anemia with elevated HbA2 and normal ferritin levels. Among those with a history of pregnancy, 29.2% reported miscarriages, and 20.8% experienced complications. Despite the reproductive implications, only 16.7% had received genetic counseling, and none had undergone partner screening.

Conclusion: Although often asymptomatic, Beta Thalassemia Trait can contribute to reproductive challenges in women. The absence of structured genetic counseling and partner testing indicates a critical gap in preventive care. Integrating thalassemia screening into antenatal and reproductive health services is essential for reducing the burden of Beta Thalassemia Major and improving maternal outcomes.

Keywords: Beta Thalassemia Trait, Women's Health, Reproductive Outcomes, Genetic Counseling, India, Hemoglobinopathy

1. Introduction

Beta Thalassemia is one of the most common hereditary hemoglobinopathies

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worldwide, with India bearing a significant burden of the condition (Colah et al., 2010). It results from mutations in the β-globin gene, leading to reduced or absent synthesis of the β-globin chains of hemoglobin. Beta Thalassemia Trait (BTT), also known as the carrier state or β-thalassemia minor, is usually asymptomatic and characterized by mild microcytic anemia. However, despite its seemingly benign nature, the clinical and reproductive implications of BTT in women, especially in the Indian context, remain underexplored.

The prevalence of BTT in India varies regionally, with studies reporting rates between 3% and 17%, depending on geographic, ethnic, and social factors (Madan et al., 2010; Balgir, 2019). Women of reproductive age often remain unaware of their carrier status until they undergo routine antenatal investigations or fertility evaluations. In such cases, the presence of unexplained anemia, history of recurrent miscarriages, or adverse pregnancy outcomes may lead to further hematological assessment. The condition can have significant reproductive consequences when both partners are carriers, resulting in a 25% risk of giving birth to a child with Beta Thalassemia Major (Cappellini et al., 2014).

While BTT is clinically mild, it may contribute to subtle hematological imbalances that affect women during pregnancy, including increased fatigue, poor iron absorption, and in some cases, complications such as fetal loss (De Sanctis et al., 2013). There is also evidence suggesting that undiagnosed BTT may lead to unnecessary iron supplementation in women, which can be harmful if not appropriately managed (Kattamis et al., 2013).

In India, where nutritional anemia is common and antenatal care coverage is still improving, distinguishing BTT from iron deficiency anemia is a public health priority. Early detection and counseling are crucial, especially in urban areas like Chandigarh, where women may present late for antenatal visits. Despite national guidelines encouraging routine screening, awareness and implementation remain inconsistent (ICMR, 2018).

This study explores the clinical and reproductive implications of BTT among women identified through a diagnostic screening program in Chandigarh. It examines their presenting symptoms, hematological parameters, and reproductive histories, with the aim of highlighting the need for integrated thalassemia screening and genetic counseling in women's healthcare services.

2. Methodology

Study Design and Setting

This was a cross-sectional diagnostic study conducted among female patients referred for anemia screening at a tertiary care center in Chandigarh, India. The study population included women aged 18 to 45 years who were either symptomatic or undergoing routine antenatal investigations.

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Sample Size and Sampling Technique

A total of 500 individuals were screened for anemia and hemoglobinopathies, out of which 50 were identified with Beta Thalassemia Trait. From this subset, 35 women were selected for this analysis based on inclusion criteria. A purposive sampling method was used to focus on female patients with confirmed BTT, as diagnosed by hematological parameters and HPLC.

Inclusion and Exclusion Criteria

Inclusion criteria included female patients aged 18–45 years with a confirmed diagnosis of Beta Thalassemia Trait based on hematological and HPLC findings.
Exclusion criteria were women with iron deficiency anemia, chronic illnesses, or co-existing hemoglobinopathies such as HbE disease or Sickle Cell Trait.

Data Collection and Diagnostic Procedures

All participants underwent detailed clinical evaluation, including reproductive history (menstrual regularity, number of pregnancies, miscarriages, and live births), and presenting symptoms such as fatigue, pallor, and general well-being. Blood samples were collected to perform Complete Blood Count (CBC) using an automated hematology analyzer. Peripheral Blood Smear (PBS) examination was conducted to assess red blood cell morphology. Serum ferritin was measured to rule out iron deficiency.

High-Performance Liquid Chromatography (HPLC) was conducted using the Bio-Rad Variant II system to detect abnormal hemoglobin fractions. BTT was confirmed by HbA_2 levels >3.5% in the presence of normal or borderline HbF levels and absence of iron deficiency (as indicated by normal ferritin).

Ethical Considerations

Ethical clearance for the study was obtained from the Institutional Ethics Committee, Desh Bhagat University. Written informed consent was obtained from all participants before data collection. Confidentiality and anonymity were ensured throughout the study process.

Data Analysis

Descriptive statistics such as frequencies, means, and standard deviations were used to analyze clinical and reproductive data. Hematological parameters were compared across subgroups based on reproductive outcomes (e.g., history of miscarriage vs. no miscarriage). Data analysis was performed using Microsoft Excel and SPSS version 21.

3. Results

3.1 Demographic Profile

Out of the 50 individuals diagnosed with Beta Thalassemia Trait in the larger screening cohort, 35 were females and included in this sub-analysis. The age distribution of the

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participants ranged from 18 to 45 years, with a mean age of 30.4 years (SD = 5.9). The majority of women (n = 20; 57.1%) were in the age group of 26–34 years.

Table 1: Age Distribution of Female Participants with BTT (N = 35)

Age Group (Years)	Frequency (n)	Percentage (%)
18–25	8	22.9
26–34	20	57.1
35–45	7	20.0
Total	**35**	**100**

The results indicate that the reproductive age group of 26–34 years was most commonly affected, reflecting the pattern of incidental detection during antenatal visits and fertility evaluations.

3.2 Clinical Presentation

Most women with BTT (n = 23; 65.7%) were asymptomatic at the time of diagnosis. However, some reported mild, nonspecific symptoms such as fatigue (n = 9; 25.7%) and pallor (n = 6; 17.1%). No cases of severe anemia or organomegaly were reported.

Table 2: Presenting Symptoms Among BTT Female Participants

Symptom	Frequency (n)	Percentage (%)
Asymptomatic	23	65.7
Fatigue	9	25.7
Pallor	6	17.1
Headache	3	8.6
Breathlessness	1	2.9

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The majority of women had only mild clinical manifestations, consistent with existing literature indicating BTT is usually a silent carrier state. However, in symptomatic cases, the symptoms were largely nonspecific and could be mistaken for nutritional deficiencies.

3.3 Hematological Profile

Hematological analysis revealed a mild reduction in hemoglobin levels with microcytic and hypochromic indices. The mean hemoglobin level was 9.5 g/dL (SD = 0.6), mean corpuscular volume (MCV) was 68.2 fL, and mean corpuscular hemoglobin (MCH) was 21.5 pg. RBC count was elevated in most participants, and serum ferritin levels were within the normal range.

Table 3: Hematological Parameters of Female BTT Carriers (Mean ± SD)

Parameter	Mean ± SD	Reference Range
Hemoglobin (g/dL)	9.5 ± 0.6	12.0–15.5
MCV (fL)	68.2 ± 4.7	80–100
MCH (pg)	21.5 ± 2.3	27–32
RBC Count (×10⁶/µL)	5.2 ± 0.4	4.0–5.2
Serum Ferritin (ng/mL)	52.7 ± 13.5	15–150
HbA_2 (%) (HPLC)	5.5 ± 0.4	<3.5 (non-BTT range)

All women showed elevated HbA_2 levels (>3.5%) on HPLC, confirming Beta Thalassemia Trait. The normal serum ferritin excluded iron deficiency anemia.

3.4 Reproductive History and Outcomes

Among the 35 women, 24 (68.6%) had a history of pregnancy. Of these, 7 (29.2%) reported one or more miscarriages, while 5 (20.8%) had experienced complications such as intrauterine growth restriction or anemia-related fatigue during pregnancy. Four participants had undergone genetic counseling, while none reported prior partner screening.

Table 4: Reproductive History and Outcomes (n = 24)

Reproductive Outcome	Frequency (n)	Percentage (%)
History of miscarriage	7	29.2
Pregnancy-related fatigue	5	20.8
Intrauterine growth restriction	3	12.5

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Uneventful pregnancies	9	37.5
Received genetic counseling	4	16.7
Partner screened for BTT	0	0.0

The findings indicate a potential reproductive burden among women with BTT, particularly in the form of recurrent pregnancy loss and fatigue. Although causality cannot be inferred from this cross-sectional data, these results suggest that BTT may be an underrecognized factor in poor obstetric outcomes. The lack of partner testing further underscores the gap in preventive genetic counseling services in the studied population.

3.5 Summary of Key Findings

- Majority of female BTT carriers were of reproductive age (26–34 years) and largely asymptomatic.
- Common clinical complaints included fatigue and pallor, but were generally mild.
- Hematological findings showed microcytic hypochromic anemia with elevated HbA_2 and normal ferritin.
- Nearly one-third of women reported reproductive complications, particularly miscarriages.
- Very few women received genetic counseling, and none had undergone partner screening.

These findings demonstrate that while BTT may be clinically mild, it has meaningful implications for women's reproductive health. The absence of structured genetic counseling and partner screening may increase the risk of Beta Thalassemia Major births and pregnancy complications.

Discussion

This study explored the clinical presentation and reproductive experiences of women diagnosed with Beta Thalassemia Trait (BTT) during a diagnostic screening program in Chandigarh, India. The findings highlight that while most female carriers of BTT were asymptomatic, a significant proportion presented with nonspecific symptoms such as fatigue and pallor, and a notable number had a history of adverse reproductive outcomes including miscarriage. These results reaffirm the silent but impactful burden of BTT among women of reproductive age in urban India.

The majority of women identified with BTT in this study fell within the 26 to 34-year age group, consistent with findings from previous studies which show that thalassemia carriers are most often diagnosed during reproductive health screenings or antenatal check-ups (Sinha et al., 2021; Kaur & Kaur, 2023). This demographic trend highlights the

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importance of integrating thalassemia screening into routine reproductive healthcare, especially in urban regions where women may otherwise go undiagnosed.

Although 65.7% of the women were asymptomatic, about one-third reported symptoms such as fatigue and pallor. These symptoms, while mild, can mimic iron deficiency anemia and often result in unnecessary iron supplementation. Such inappropriate treatment not only fails to address the underlying issue but may also lead to iron overload, especially when supplementation is prolonged (Rund & Rachmilewitz, 2005; Mittal & Agarwal, 2011). The distinction between BTT and nutritional anemia is therefore essential in settings like India, where anemia prevalence is already high among women.

The hematological findings in this study align with the classic features of BTT. The women exhibited microcytic, hypochromic anemia with normal or slightly elevated red cell counts, low MCV and MCH values, and elevated HbA_2 levels as confirmed by HPLC. Importantly, their serum ferritin levels were within the normal range, excluding iron deficiency. These observations underscore the value of comprehensive screening that includes not only complete blood count and peripheral smear but also serum ferritin and HPLC to establish an accurate diagnosis (Pradhan et al., 2014; Hoffbrand et al., 2016).

One of the most compelling findings from this study is the high incidence of adverse reproductive outcomes among BTT carriers. Among the women with pregnancy histories, 29.2% reported miscarriages and 20.8% experienced complications such as pregnancy-related fatigue or intrauterine growth restriction. Although Beta Thalassemia Trait is not a direct cause of miscarriage, its presence may exacerbate gestational anemia or oxidative stress, which can contribute to unfavorable pregnancy outcomes (De Sanctis et al., 2013; Kattamis et al., 2013). These findings suggest a need for closer hematological monitoring of pregnant women with BTT, particularly in the second and third trimesters.

Moreover, the study highlights significant gaps in reproductive counseling and partner screening. Only four women had received genetic counseling, and none reported that their partners had been screened. This is particularly concerning in a population where the marriage of two carriers can result in a 25% chance of having a child with Beta Thalassemia Major—a condition associated with lifelong transfusions, iron chelation, and significant psychosocial and economic burden (Cappellini et al., 2014; Narayan, 2006). Similar studies from Pakistan and Iran have shown that integrating premarital or antenatal partner screening programs with community education has led to substantial reductions in the incidence of thalassemia major (Ansari & Shamsi, 2010; Gharaibeh et al., 2011).

The low rates of counseling and absence of partner testing in this study point to

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the urgent need for strengthening public health policy and practice around hemoglobinopathy management in India. While national guidelines exist, their implementation remains inconsistent, particularly in private or semi-urban healthcare settings (ICMR, 2018). To prevent the intergenerational transmission of severe thalassemia syndromes, public health systems must adopt a proactive approach by promoting universal carrier screening for women of reproductive age, mandatory partner testing during antenatal care, and widespread availability of genetic counseling services.

In addition, public awareness campaigns must address the stigma and misinformation surrounding genetic conditions like thalassemia. Women in this study, though medically stable, faced reproductive challenges without the benefit of informed support or care pathways. Targeted educational interventions in schools, colleges, and maternal health clinics could help address these gaps and empower couples to make informed reproductive decisions (Suthar & Patel, 2014; Sharma & Patel, 2024).

This study adds valuable insights into the intersection of hemoglobinopathies and women's reproductive health in urban India. However, it also has limitations. The study was conducted at a single center using a cross-sectional design, which limits causal inferences. Additionally, genetic mutation analysis was not performed, which could have provided further clarity on genotype-phenotype correlations.

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Despite these limitations, the findings strongly support the argument that Beta Thalassemia Trait is more than a benign genetic condition—it has significant implications for women's clinical and reproductive health. Early diagnosis, proper differentiation from iron deficiency anemia, and integration of genetic counseling into women's health services are essential steps toward improving outcomes.

5. Conclusion

This study highlights that Beta Thalassemia Trait, though often asymptomatic, can have significant clinical and reproductive implications for women. A considerable number of female carriers experienced symptoms such as fatigue and pallor, which may be misdiagnosed as nutritional anemia. More notably, nearly one-third of women with BTT reported a history of miscarriage or pregnancy complications, suggesting a potential reproductive burden associated with the trait.

Despite these implications, access to genetic counseling and partner screening was minimal among participants, underscoring a critical gap in preventive reproductive healthcare. The findings stress the need to incorporate routine thalassemia screening into antenatal care and reproductive health services. Early identification, differentiation from iron deficiency anemia, and provision of genetic counseling are essential for informed reproductive decision-making and for preventing the birth of children with Beta Thalassemia Major.

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Public health initiatives must prioritize awareness, education, and screening among women of reproductive age to mitigate the intergenerational impact of this genetic disorder. Strengthening diagnostic pathways and ensuring equitable access to counseling services can transform Beta Thalassemia Trait from a silent burden into a manageable public health condition.

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