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Prevalence and Hematological Profile of Beta Thalassemia Trait in an Urban Indian Population: A Cross-Sectional Study from Chandigarh

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Abstract

Background: Beta Thalassemia Trait (BTT) is a prevalent inherited hemoglobin disorder in India, often undetected due to its asymptomatic presentation. Understanding its distribution and hematological profile in urban populations is essential for effective screening and prevention strategies.

Objective: This study aimed to determine the prevalence and hematological characteristics of Beta Thalassemia Trait among patients in Chandigarh, a North Indian urban center, using a comprehensive diagnostic protocol.

Methods: A cross-sectional study was conducted among 500 patients referred for anemia evaluation. Complete blood count (CBC), peripheral blood smear (PBS), serum ferritin, and High-Performance Liquid Chromatography (HPLC) were used for diagnosis. Individuals with elevated HbA2 levels (>3.5%) and normal ferritin were identified as BTT carriers. Descriptive statistics were used to analyze demographic and hematological data.

Results: Out of 500 screened individuals, 50 (10%) were diagnosed with BTT. The majority were females (70%), particularly in the 26–34 age group. Most were asymptomatic, though some reported fatigue or reproductive challenges. Hematologically, carriers showed reduced hemoglobin (mean: 9.5 g/dL), low MCV and MCH, normal ferritin, and PBS findings of microcytosis, hypochromia, and target cells. HPLC confirmed elevated HbA2 (mean: 5.5%) and mild HbF elevation in select cases.

Conclusion: The study reveals a significant prevalence of Beta Thalassemia Trait in Chandigarh, particularly among women of reproductive age. Routine screening, especially during antenatal care, and integration of genetic counseling and partner testing are crucial for preventing the transmission of Beta Thalassemia Major. Findings support the expansion of urban thalassemia screening programs in India.

Keywords: Beta Thalassemia Trait, Hemoglobinopathy, Prevalence, High-Performance Liquid Chromatography, Genetic Counseling

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1. Introduction

Beta Thalassemia is one of the most common inherited hemoglobin disorders globally, with a particularly high burden in South Asia, including India. It results from mutations in the beta-globin (HBB) gene that reduce or eliminate beta-globin chain synthesis, leading to imbalanced hemoglobin production, microcytic hypochromic anemia, and varying clinical severity depending on zygosity (Weatherall & Clegg, 2001; Modell & Darlison, 2008). The heterozygous form, known as Beta Thalassemia Trait (BTT), is usually asymptomatic or associated with mild anemia but poses significant public health challenges due to the risk of transmitting the gene to offspring when both partners are carriers (ICMR, 2018; Kumar et al., 2018).

In India, the prevalence of Beta Thalassemia Trait varies between 3% and 17%, depending on geographic and ethnic factors (Colah et al., 2010; Balgir, 2019). Northern states like Punjab, Haryana, and union territories such as Chandigarh report relatively higher carrier rates, likely due to socio-cultural practices including consanguineous and endogamous marriages (Jain & Bagul, 2018; Suthar & Patel, 2014). However, despite advancements in diagnostics, routine screening for BTT in urban populations remains limited, and many individuals remain unaware of their carrier status until incidentally detected during antenatal visits or health check-ups (Sinha et al., 2021; Narayan, 2006).

High-Performance Liquid Chromatography (HPLC) is considered the gold standard for diagnosing BTT, characterized by elevated levels of HbA_2 (>3.5%) (Bain, 2006; Verma et al., 2016). Yet, given the high prevalence of nutritional anemia in India, particularly iron deficiency anemia (IDA), differential diagnosis through peripheral blood smear (PBS), complete blood count (CBC), and serum ferritin remains essential for accurate classification and treatment (Kumar et al., 2018; Ghosh et al., 2009).

Chandigarh, a union territory and capital city for both Punjab and Haryana, represents a dynamic urban population with high levels of in-migration and diverse socioeconomic backgrounds. These factors can impact awareness, access to screening, and healthcare-seeking behaviors. Therefore, understanding the prevalence and hematological characteristics of Beta Thalassemia Trait in such a setting is crucial for guiding targeted interventions, antenatal screening strategies, and genetic counseling programs.

The present study aims to evaluate the prevalence and hematological profile of Beta Thalassemia Trait among patients presenting with anemia and suspected hemoglobinopathies in Chandigarh. By using a combination of CBC, PBS, serum ferritin, and HPLC, this study seeks to strengthen the case for systematic urban screening and inform region-specific public health strategies.

2. Methodology

2.1 Study Design and Setting

This research was a **cross-sectional observational study** conducted

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between January and June 2025 in Chandigarh, India. Patients were recruited from various clinical and diagnostic centers within the city, encompassing both private and public facilities. Chandigarh, being an urban hub with a multicultural population, was selected to represent a diverse sample relevant to the northern Indian demographic.

2.2 Study Population

The study population included individuals referred for evaluation of anemia or suspected hemoglobinopathies. Participants were screened based on clinical presentations (e.g., pallor, fatigue) and/or laboratory indicators of microcytic hypochromic anemia.

Inclusion Criteria:

- Patients of all ages and genders.
- Individuals presenting with anemia and/or red cell indices suggestive of hemoglobinopathy.
- Individuals providing informed consent to participate.

Exclusion Criteria:

- Patients with diagnosed iron deficiency anemia (confirmed via low serum ferritin).
- Known cases of other hemoglobinopathies (e.g., sickle cell disease, alpha-thalassemia).
- Patients unwilling to participate in the study.

2.3 Sample Size and Sampling Technique

A total of 500 patients were screened during the study period. Of these, 50 individuals were diagnosed with Beta Thalassemia Trait based on HPLC findings and included for detailed hematological analysis. A convenience sampling method was used due to practical and logistical considerations in diagnostic settings.

2.4 Data Collection Tools and Diagnostic Criteria

The following investigations were conducted for each patient:

- **Complete Blood Count (CBC):**
  Performed using the Yumizen H1500 hematology analyzer. CBC measured hemoglobin levels, mean corpuscular volume (MCV), mean corpuscular hemoglobin (MCH), and red cell distribution width (RDW) (Bain, 2006; Kumar et al., 2018).
- **Peripheral Blood Smear (PBS):**
  Stained with Leishman stain and analyzed microscopically for morphological abnormalities such as microcytosis, hypochromia, anisopoikilocytosis, and target cells (Verma et al., 2016).
- **Serum Ferritin:**
  Estimated using standard ELISA kits to exclude iron deficiency anemia—a necessary step in

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India's context of high IDA prevalence (ICMR, 2018).

- **High-Performance Liquid Chromatography (HPLC):** Conducted using the Bio-Rad D10 system. Elevated HbA_2 levels (>3.5%) were used to confirm Beta Thalassemia Trait, while HbF values were noted for additional characterization (Pradhan et al., 2014).

2.5 Ethical Considerations

Ethical approval was obtained from the Institutional Ethics Committee of Desh Bhagat University, Mandi Gobindgarh. Written informed consent was obtained from all participants, or from guardians in the case of minors. Confidentiality of data and patient privacy were maintained throughout the research.

2.6 Data Analysis

Collected data were entered in Microsoft Excel and analyzed using basic descriptive statistics. Mean and standard deviation were calculated for continuous variables (e.g., Hb, MCV, MCH), while categorical data (e.g., gender, prevalence rate) were expressed as frequencies and percentages. The diagnostic yield of CBC and PBS was correlated with confirmatory HPLC findings.

3. Results

A total of 500 patients were screened for anemia or suspected hemoglobinopathies at various diagnostic centers in Chandigarh from January to June 2025. Based on elevated HbA_2 levels (>3.5%) via HPLC, 50 individuals (10%) were confirmed to have Beta Thalassemia Trait (BTT). The results are organized below according to the study objectives.

3.1 Prevalence of Beta Thalassemia Trait

Out of 500 screened individuals, 50 cases tested positive for BTT, indicating a prevalence rate of 10%.

3.2 Demographic Characteristics of BTT-Positive Cases

Table 1: Age and Gender Distribution of BTT Patients (N = 50)

Age Group (Years)	Male (n)	Female (n)	Total (n)	Percentage (%)
18–25	4	6	10	20.0%
26–34	8	27	35	70.0%
35–45	3	2	5	10.0%
Total	**15**	**35**	**50**	**100.0%**

The majority (70%) of BTT carriers were in the 26–34-year age group, a critical reproductive window. A higher proportion of cases were female (70%), which reflects

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increased screening during antenatal visits. This gender difference is consistent with patterns observed in earlier Indian studies (Kaur & Kaur, 2023; Sinha et al., 2021).

3.3 Clinical Presentation

Table 2: Presenting Complaints among BTT Patients

Clinical Presentation	Frequency (n)	Percentage (%)
Asymptomatic (Detected incidentally)	32	64.0%
Fatigue	10	20.0%
Pallor	6	12.0%
Recurrent miscarriages	2	4.0%

A large proportion (64%) were asymptomatic carriers, discovered through routine or antenatal screening. Mild fatigue and pallor were reported by a smaller group. Notably, 4% (n=2) of female carriers had recurrent miscarriages, suggesting potential reproductive implications even in trait conditions.

3.4 Hematological Findings

Table 3: Summary of CBC Findings in BTT Patients (N = 50)

Parameter	Minimum	Maximum	Mean ± SD	Reference Range
Hemoglobin (g/dL)	8.8	10.5	9.5 ± 0.45	12–15 g/dL
MCV (fL)	67.0	70.0	68.5 ± 0.62	80–100 fL
MCH (pg)	22.0	24.0	23.1 ± 0.43	27–32 pg
RBC Count (million/µL)	4.8	6.2	5.4 ± 0.38	4.2–5.9

All patients exhibited microcytic hypochromic anemia, as indicated by low MCV and MCH values. The mean hemoglobin was 9.5 g/dL, indicating mild anemia, which is typical for Beta Thalassemia Trait (Bain, 2006; Kumar et al., 2018). Interestingly, RBC counts were within or slightly above normal range, consistent with BTT profiles.

3.5 Peripheral Blood Smear Findings

Table 4: Morphological Abnormalities in Peripheral Smear (N = 50)

Morphological Feature	Frequency (n)	Percentage (%)

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Microcytosis	50	100.0%
Hypochromia	50	100.0%
Target cells	46	92.0%
Anisopoikilocytosis	40	80.0%
Basophilic stippling	8	16.0%

Microcytosis and hypochromia were universal findings. Target cells and anisopoikilocytosis were also common, which are hallmark features of BTT on smear examination (Bain, 2006). These features help distinguish BTT from IDA, especially when supported by normal ferritin and elevated HbA2.

3.6 HPLC and Ferritin Results

Table 5: HPLC and Serum Ferritin in BTT Patients (N = 50)

Parameter	Minimum	Maximum	Mean ± SD	Diagnostic Threshold
HbA2 (%)	3.8	6.1	5.5 ± 0.40	>3.5% (BTT)
HbF (%)	0.8	4.5	1.5 ± 0.72	<2% (normal adults)
Serum Ferritin (ng/mL)	28.0	110.0	65.7 ± 18.3	>30 normal; <15 = IDA

All patients had HbA2 > 3.5%, confirming the diagnosis of BTT. Mean HbA2 was 5.5%, consistent with published diagnostic criteria (ICMR, 2018; Verma et al., 2016). HbF was mildly elevated in some cases but remained within acceptable ranges for carriers. Serum ferritin levels were normal, ruling out concurrent iron deficiency anemia and highlighting the importance of ferritin testing in distinguishing causes of microcytic anemia in India (Mittal & Agarwal, 2011).

3.7 Summary of Representative Case Profiles

Table 6: Selected Case Summaries of BTT Carriers

Case ID	Age/Gender	Hb (g/dL)	HbA2 (%)	HbF (%)	Ferritin (ng/mL)	Clinical Note

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C001	26/M	10.2	5.1	4.5	65	Detected via workplace wellness camp
C047	32/F	9.8	5.9	1.5	72	Recurrent miscarriage, antenatal screen
C065	28/F	10.0	5.6	1.3	60	Both partners are carriers; PGT advised
C073	30/F	9.5	6.0	1.8	78	History of abortion; partner screened
C090	34/M	9.0	5.8	1.4	69	Incidentally found during pre-employment

These representative cases reflect the clinical diversity of BTT. While most cases were asymptomatic, some presented during antenatal screening or due to reproductive challenges. One male case (C001) showed a markedly high HbF (4.5%) and was identified through a corporate wellness program, emphasizing the potential of non-antenatal detection platforms.

3.8 Key Findings Summary

- **Prevalence** of BTT was **10%**, consistent with regional averages in Northern India.
- **Females aged 26–34 years** were the most affected demographic group.
- **64% were asymptomatic**, underscoring the silent nature of BTT.
- **All patients had elevated HbA2 levels**, with consistent CBC and PBS findings of **microcytic hypochromic anemia**.
- **Normal serum ferritin** levels ruled out iron deficiency in all cases.
- **Reproductive implications** observed in a few cases reinforce the importance of genetic counseling and partner screening.

4. Discussion

This study examined the prevalence and hematological profile of Beta Thalassemia Trait among individuals in Chandigarh, an urban setting in North India. The observed prevalence of 10 percent is consistent with rates reported in Northern Indian regions such as Punjab, Haryana, and parts of Uttar

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Pradesh. Several studies have indicated that prevalence in these areas ranges between 8 and 12 percent, and this is largely attributed to endogamous marriage practices and lack of routine genetic screening (Colah et al., 2010; Balgir, 2019; Jain and Bagul, 2018). The prevalence noted in this study confirms that Beta Thalassemia Trait continues to be a significant silent burden in the Indian population, often going undiagnosed until incidental screening or antenatal care uncovers it.

The demographic characteristics of the diagnosed cases in this study revealed that the majority were female and within the reproductive age group of 26 to 34 years. This is in line with findings from other Indian studies where antenatal screening remains the most common route to diagnosis (Sinha et al., 2021; Kaur and Kaur, 2023). The gender difference in our study does not reflect a biological predilection but rather the higher likelihood of women being screened during pregnancy. This further supports the need to embed thalassemia screening within routine antenatal programs. National and international recommendations already advocate for this, emphasizing that early diagnosis allows timely counseling and reproductive decision-making (ICMR, 2018; WHO, 2006).

The majority of individuals in this study were asymptomatic, confirming what has been previously observed—that Beta Thalassemia Trait typically remains clinically silent. Some patients did report nonspecific complaints such as pallor and fatigue, which are commonly misattributed to iron deficiency anemia, especially in the Indian context where nutritional deficiencies are prevalent (Mittal and Agarwal, 2011). Notably, two female participants had a history of recurrent miscarriages. While Beta Thalassemia Trait is not directly linked to miscarriage, its associated anemia and oxidative stress during pregnancy may contribute to adverse outcomes (De Sanctis et al., 2013; Kattamis et al., 2013). Such reproductive complications emphasize the importance of genetic counseling and partner screening, particularly for couples planning pregnancies. Studies from other South Asian contexts such as Iran and Pakistan have demonstrated that partner screening, when implemented early, significantly reduces the incidence of Beta Thalassemia Major (Ansari and Shamsi, 2010; Narayan, 2006).

The hematological parameters observed in this study were consistent with the diagnostic profile of Beta Thalassemia Trait. Individuals showed low hemoglobin levels with reduced MCV and MCH values, yet often had relatively normal or elevated red blood cell counts. This hematological pattern is typical for BTT and helps differentiate it from iron deficiency anemia, a distinction that is critical in the Indian context (Kumar et al., 2018; Bain, 2006). Peripheral smear findings such as microcytosis, hypochromia, anisopoikilocytosis, and target cells were commonly observed. These features have been previously described as classical for thalassemic red cell morphology (Verma et al., 2016; Ghosh et al., 2009). All individuals in this study had normal serum ferritin levels, ruling out concurrent iron deficiency

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anemia and underscoring the need to assess iron stores before initiating unnecessary iron therapy.

High-Performance Liquid Chromatography was used as the confirmatory test for Beta Thalassemia Trait and proved highly reliable, with all cases exhibiting elevated HbA2 levels above 3.5 percent. The mean HbA2 value in this study was 5.5 percent, which conforms to established diagnostic benchmarks (Pradhan et al., 2014; Patrinos and Kollia, 2000). In a few cases, HbF levels were mildly elevated, which may occur in certain genetic variants of Beta Thalassemia or due to co-inheritance of other modifiers (Galanello and Origa, 2010; Weatherall and Clegg, 2001).

The findings of this study have several important public health implications. Urban regions like Chandigarh, with diverse populations and rapid social transitions, are particularly suitable for integrating community-based screening programs. Experiences from Gujarat and Maharashtra have demonstrated the effectiveness of youth-targeted screening campaigns in colleges and premarital counseling programs in reducing the birth of affected children (Sharma and Patel, 2024; Singh et al., 2025). Chandigarh could adopt similar models by incorporating routine screening into school and workplace health programs. In addition, policy efforts should aim to normalize genetic screening through educational interventions and reduce stigma by promoting community awareness. Healthcare providers should also be trained to recognize suggestive hematological patterns and refer suspected cases for HPLC and counseling.

While the study adds valuable regional data on Beta Thalassemia Trait, certain limitations must be noted. The sample was drawn from a single center, and the study duration was limited. Molecular genotyping was not performed, which could have enriched our understanding of the genetic variations responsible for the observed hematological profiles. Nonetheless, the study offers a comprehensive diagnostic snapshot using commonly available hematological tools, emphasizing the practicality of implementing such screening at the primary and secondary healthcare levels.

This research reaffirms that Beta Thalassemia Trait is prevalent, underdiagnosed, and often unrecognized in urban populations. With appropriate integration of diagnostic methods, routine antenatal and community screening, and partner testing, the burden of Beta Thalassemia Major can be significantly reduced. Strengthening public health infrastructure and aligning national policy with ground-level implementation remain crucial steps in achieving this goal.

5. Conclusion

The study confirms that Beta Thalassemia Trait affects a significant proportion of the population in Chandigarh, with a 10 percent prevalence rate identified through clinical and laboratory screening. Most individuals were asymptomatic and belonged to the reproductive age group, highlighting the

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silent yet potentially consequential nature of the condition. Hematological parameters and HPLC findings consistently supported the diagnosis, while normal serum ferritin levels helped exclude iron deficiency anemia.

These findings emphasize the urgent need for structured screening programs, particularly within antenatal services and among young adults. Genetic counseling and partner testing should be made standard practice to prevent the inheritance of Beta Thalassemia Major. Public health policies must promote awareness, accessibility of diagnostics, and integration of preventive strategies into mainstream health services.

With timely detection and counseling, the burden of this hereditary disorder can be reduced, improving health outcomes and supporting India's broader goals in non-communicable disease prevention and genetic disorder management.

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