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Hemoglobinopathies Amongst Antenatal Women in a Tertiary Care Centre, IGMC Shimla – A Cross Sectional Study

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Abstract:

Background: Hemoglobinopathies are among the most common inherited disorders globally. In India, thalassemia and sickle cell disorders are prevalent and pose serious maternal and fetal health risks during pregnancy.

Objective: To determine the prevalence and hematological profiles of hemoglobinopathies among antenatal women attending a tertiary care hospital in Himachal Pradesh.

Methods: A hospital-based cross-sectional study was conducted from August 2024 to April 2025 at IGMC Shimla, including 1085 antenatal women. Hematological indices were evaluated using a 5-part hematology analyzer. Women with anemia or microcytic indices were further assessed using HPLC. CBC, reticulocyte count, iron profile, Vitamin B12, and folic acid levels were evaluated.

Results: Hemoglobinopathies were diagnosed in 24 women (2.2%): 22 had β-thalassemia trait (2.1%), one had sickle cell trait (0.1%), and one had a suspected β-chain variant (0.1%). Mean hemoglobin in BTT cases was 10.0 g/dL. All BTT cases showed low MCV and MCH. Mentzer's Index showed a sensitivity of 77.2% and specificity of 98.3% for detecting BTT. Coexistent iron deficiency was seen in two BTT cases. No cases of HbE, HbD-Punjab, or other variants were found.

Conclusion: Hemoglobinopathies, especially β-thalassemia trait, are prevalent among antenatal women in Himachal Pradesh. Early antenatal screening using HPLC is crucial for timely diagnosis, genetic counseling, and prevention of severe hemoglobinopathies.

Keywords: Hemoglobinopathies, Antenatal Women, β-Thalassemia Trait, HPLC, Himachal Pradesh

1. Introduction:

Hemoglobinopathies, including thalassemias and structural variants like sickle cell disease, arise due to mutations in globin genes. These disorders are particularly concerning in pregnancy due to increased risks of anemia, miscarriage, low birth weight, and fetal loss. Despite national screening guidelines, many cases remain undiagnosed due to lack of resources and awareness. This study aimed to assess the prevalence and hematological characteristics of hemoglobinopathies in antenatal women at a tertiary care center in Himachal Pradesh.

2. Materials and Methods:

- **Design:** Cross-sectional hospital-based study
- **Duration:** August 2024 to April 2025
- **Setting:** IGMC Shimla, Himachal Pradesh
- **Sample Size:** 1085 antenatal women
- **Inclusion:** All consenting antenatal women
- **Exclusion:** Recent blood transfusion within 4 months
- **Investigations:** CBC, reticulocyte count, serum iron, vitamin B12, folic acid, and HPLC (Bio-Rad Variant II)
- **Statistical Analysis:** Data analyzed using SPSS v25; Chi-square and t-tests used where appropriate

3. Results:

- **Demographics:** Mean age = 26.67 ± 4.9 years; 84.6% from rural areas
- **Prevalence of Hemoglobinopathies:**
  - β-thalassemia trait (BTT): 22/1085 (2.1%)
  - Sickle cell trait (SCT): 1/1085 (0.1%)
  - Suspected β-chain variant: 1/1085 (0.1%)
- **CBC in BTT cases:**
  - Mean Hb = 10.09 ± 1.48 g/dL
  - MCV = 63.83 fL; MCH = 20.1 pg; RBC count = 5.1 million/mm^3
  - RDW = 13.1%; Reticulocytes = 2.2%
- **Mentzer's Index:** Sensitivity 77.2%; Specificity 98.3%
- **Nutritional Co-deficiencies:**
  - Iron deficiency: 2 BTT cases (HbA2: 4.3% and 4.8%)
  - No B12/folate deficiency in BTT group
- **Sickle Cell Trait:** One case, a migrant from Jharkhand, confirmed via sickling test
- **Family Screening:** All husbands (n=24) tested negative for hemoglobinopathies

4. Discussion:

The prevalence of BTT (2.1%) in our study aligns with regional data from North India. HPLC proved to be a valuable tool for accurate detection. Mentzer's Index demonstrated high specificity but moderate sensitivity, reduced by the presence of iron deficiency in a few cases. SCT and other variants were rare, reflecting the ethnic and geographic profile of the population. Early identification and partner screening are vital to reduce disease burden through counseling and prenatal diagnosis.

5. Conclusion:

Hemoglobinopathies, especially β-thalassemia trait, are not uncommon in antenatal women in Himachal Pradesh. Incorporation of routine antenatal screening using HPLC can facilitate early detection and genetic counseling, thereby reducing the incidence of severe hemoglobin disorders in offspring.

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