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Original Research Article

Peripheral Blood Film Pattern in Thalassemia and Haemoglobinopathy

Mala Kumari[1], Puja Kumari[2], Md. Ghulam Tabraiz[3]

[1]Tutor, Department of Pathology, JNKTMCH, Madhepura, Bihar, India
[2]Tutor, Department of Pathology, JNKTMCH, Madhepura, Bihar, India
[3]Professor, Department of Pathology, JNKTMCH, Madhepura, Bihar, India

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Corresponding Author: Puja Kumari
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Abstract:

Background: Common genetic red cell abnormalities that cause hemolysis, chronic anaemia, and many systemic consequences are hemoglobinopathy and thalassemia. Peripheral blood film testing is still an easy, affordable, and useful diagnostic technique, especially in environments with low resources. Using 50 laboratory-confirmed cases over an 11-month period, this retrospective study assessed peripheral blood film patterns in individuals with hemoglobinopathy and thalassemia.

Methods: Laboratory records of 50 patients diagnosed by hematological parameters and hemoglobin analysis were reviewed. Demographic details, complete blood count findings, and peripheral smear morphology were analyzed. Smears were assessed for microcytosis, hypochromia, target cells, anisopoikilocytosis, nucleated red blood cells, basophilic stippling, and other abnormalities.

Results: The most frequent diagnosis among 50 cases was beta thalassemia trait (40%), which was followed by beta thalassemia major (24%), sickle cell disease (18%), and other hemoglobinopathies (18%). 84% of cases had microcytosis and hypochromia, 72% had target cells, 68% had anisopoikilocytosis, and the majority of cases with severe thalassemia had nucleated red blood cells ($p<0.001$). Sickle cell illness was substantially correlated with sickle forms ($p<0.001$).

Conclusion: The study demonstrates characteristic smear abnormalities that can strongly suggest thalassemia or hemoglobinopathy and guide further confirmatory testing. Peripheral blood film remains a valuable first-line screening tool in hematology practice.

Keywords: Thalassemia, Hemoglobinopathy, First-Line Screening, Microcytosis, Hemoglobin.

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Introduction

Hemoglobinopathy and thalassemia are two of the most prevalent monogenic illnesses in the world. They are very common in regions of Africa, the Middle East, South Asia, and the Mediterranean region. Chronic anemia, transfusion reliance, iron overload, development retardation, organ failure, and psychosocial strain are all significantly impacted by these illnesses. Thalassemias cause hemolysis, microcytic hypochromic anemia, and inefficient erythropoiesis due to decreased or missing alpha or beta globin chain production. Clinical severity might vary from severe transfusion-dependent illness to asymptomatic carrier states. Sickle cell disease and other aberrant hemoglobins that affect red cell deformability, survival, or oxygen affinity are examples of hemoglobinopathies, which are structural variations of hemoglobin [1].

Prenatal screening, genetic counseling, transfusion planning, patient treatment, and preventative initiatives all depend on early diagnosis. Even while molecular techniques, hemoglobin electrophoresis, and high-performance liquid chromatography (HPLC) offer a conclusive diagnosis, these tests might not always be readily available in every situation. Conventional hematological examination is therefore still very important. Examining peripheral blood films is a quick, affordable, and widely available technique that provides significant morphological indicators. Microcytosis, hypochromia, target cells, anisopoikilocytosis, tear drop cells, basophilic stippling, and nucleated red blood cells in severe cases are typical signs of thalassemia. In hyposplenic conditions, sickle hemoglobin diseases can manifest as sickle cells, target cells, polychromasia, and Howell-Jolly bodies [2].

The ability to differentiate inherited hemoglobin opathies from iron deficiency anemia and other types of microcytosis is enhanced when smear results are correlated with red cell indices. Retrospective laboratory studies offer valuable

information about common smear morphology and local illness patterns. Clinicians and pathologists can use this information to prioritize screening techniques and choose the best confirmatory tests. The purpose of this 11-month retrospective study was to assess peripheral blood film patterns in 50 instances of hemoglobinopathy and thalassemia. Finding common morphological abnormalities and correlating specific smear findings with illness categories were the goals of the investigation. Comprehending these trends helps enhance early detection and economical diagnostic procedures [3].

Materials and Methods

Study Design: Retrospective descriptive study.

Study Duration: 11 months.

Study Setting: Department of Hematology/ Pathology in a tertiary care hospital.

Sample Size: 50 confirmed cases of thalassemia and hemoglobinopathy.

Inclusion Criteria

- Confirmed diagnosis by HPLC/electrophoresis or documented hematology records
- Peripheral smear available for review

Exclusion Criteria

- Inadequate smear quality
- Incomplete records
- Recently transfused samples with uninterpretable morphology

Statistical Analysis: Categorical variables expressed as number and percentage. Chi-square/Fisher exact test used. $p < 0.05$ considered significant.

Results

Table 1: Diagnostic Spectrum (n=50)

Diagnosis	n (%)	p-value
Beta thalassemia trait	20 (40%)	0.002*
Beta thalassemia major	12 (24%)	
Sickle cell disease	9 (18%)	
Other hemoglobinopathies	9 (18%)	

Table 2: Peripheral Smear Findings

Finding	Cases n (%)	p-value
Microcytosis	42 (84%)	<0.001*
Hypochromia	42 (84%)	<0.001*
Target cells	36 (72%)	0.004*
Anisopoikilocytosis	34 (68%)	0.01*

Table 3: Severe Morphological Changes by Diagnosis

Parameter	Beta Thal Major (n=12)	Others (n=38)	p-value
Nucleated RBCs	10	6	<0.001*
Marked Poikilocytosis	9	8	0.003*

Table 4: Sickle Cells Association

Parameter	SCD (n=9)	Non-SCD (n=41)	p-value
Sickle cells present	8	1	<0.001*
Target cells present	7	29	0.18

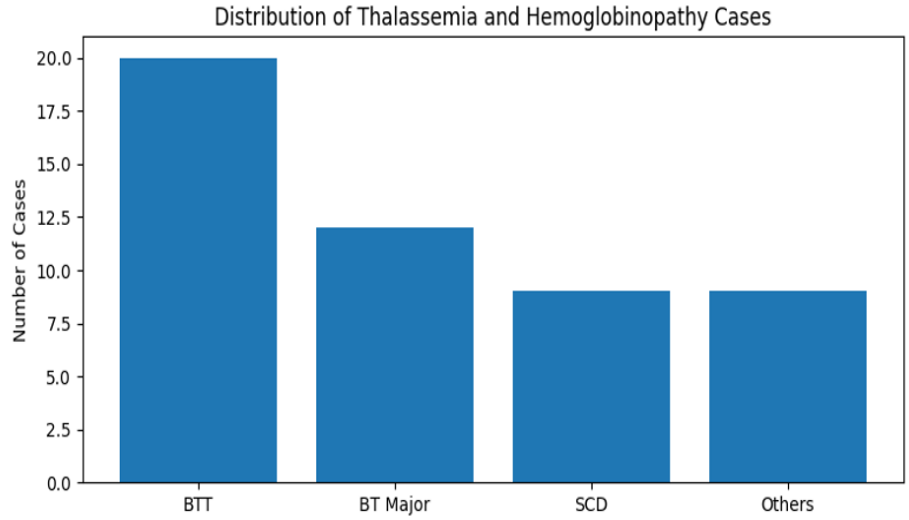

Figure 1: Distribution of thalassemia and hemoglobinopathy cases

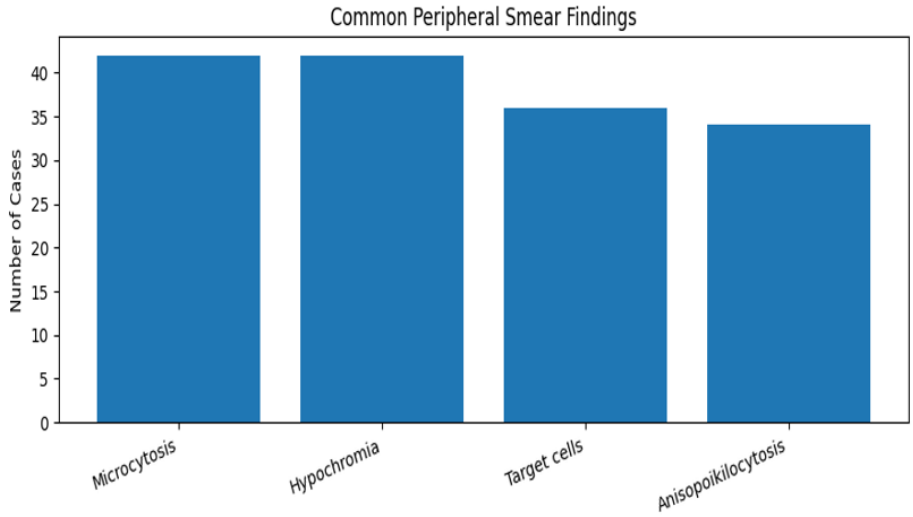

Figure 2: Common peripheral smear findings

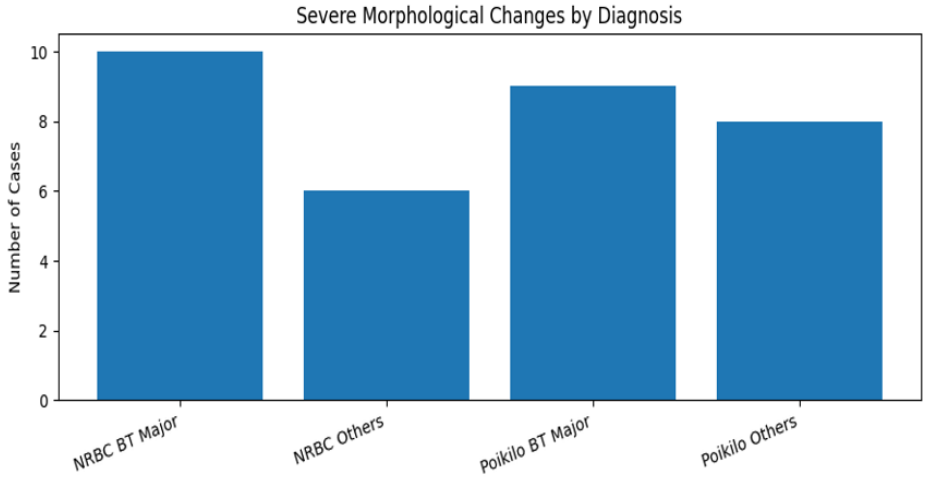

Figure 3: Severe morphological changes by diagnosis

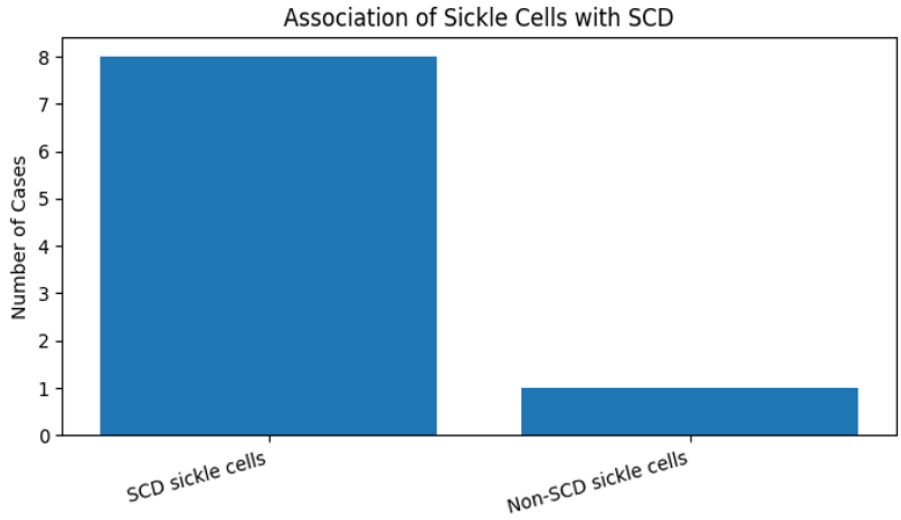

Figure 4: Association of sickle cells with SCD

Discussion

Peripheral blood film morphology was assessed in 50 patients of hemoglobinopathy and thalassemia during an 11-month period in this retrospective investigation. The results suggest that traditional smear examination is still a useful screening and supportive diagnostic tool, especially in situations where sophisticated confirmatory testing may be scarce or postponed.

The most prevalent diagnosis in this group was beta thalassemia trait. This is expected as characteristic states are more common in the population and are often found in family studies, premarital screening, prenatal screening, and anemia workup. When both parents are carriers, identification is crucial for genetic counseling and the avoidance of severe offspring sickness, even if the condition is clinically moderate [4].

Almost 25% of cases had beta thalassemia significant. This group usually exhibits severe anaemia, splenomegaly, marrow enlargement, and transfusion dependency in childhood. Due to compensatory marrow stress and poor erythropoiesis, their peripheral smears frequently exhibit noticeable abnormalities. This study's high correlation between thalassemia major and nucleated red blood cells and marked poikilocytosis is consistent with severe marrow stimulation and early release of erythroid precursors. Overall, the most common results were hypochromia and microcytosis [5]. These alterations are typical of thalassemia syndromes and show decreased hemoglobinization of red blood cells. They may, however, overlap with iron deficiency anaemia and are not specific. Red cell indices, RBC count, red cell distribution width, clinical history, and confirmatory haemoglobin analysis must all be included in the interpretation. Microcytosis may be out of proportion to the severity of anaemia in the thalassemia trait, although the red blood cell count may be mostly unaffected [6].

In many of the cases, target cells were seen. Target cells are frequently observed in thalassemia, liver illness, post-splenectomy conditions, and hemoglobinopathies. They are caused by an elevated surface area-to-volume ratio. When combined with microcytosis, they should raise the possibility of hereditary haemoglobin problems. Anisopoikilocytosis, which indicates variance in cell size and shape due to membrane damage and inefficient erythropoiesis, was also prevalent. Strange poikilocytes, teardrop cells, fragments, and polychromasia may be seen in cases of acute illness. These results could be used to identify patients who need immediate additional assessment [7].

As anticipated, there was a high correlation between sickle cell illness and sickle forms on smear. But not all sickle cell disease smears show a lot of sickled cells, particularly after transfusion or outside of crises. On the other hand, sporadic sickle-like shapes might not always be artifactual. Therefore, morphology should be carefully analysed and verified using electrophoresis or HPLC. Low cost, quick turnaround, and the capacity to concurrently identify alternative diagnoses including hemolysis, infection-related alterations, megaloblastic characteristics, or platelet abnormalities are some of the practical benefits of peripheral smear screening. It is still particularly useful in peripheral labs and bulk screening.

There are restrictions on the study. It had a small sample size, was retrospective, and probably had referral bias. Molecular genotyping, transfusion history, ferritin status, splenic status, and quantitative haematological indices were not consistently accessible. Patients who have recently

received transfusions may have changed morphology, which could obscure diagnostic characteristics. Another factor to take into account when interpreting smears is interobserver variability [8].

The study emphasises the ongoing significance of morphology in contemporary haematology despite these drawbacks. Early detection of inherited anaemia syndromes can be enhanced by standardised smear review procedures, red cell morphology education, and integration with CBC parameters. Smear results should be compared with RBC indices, HPLC fractions, iron status, and genotype correlations in bigger cohorts in future research. Artificial intelligence and digital image analysis may also improve the identification of minute morphological patterns. Peripheral blood film is still an essential first-line test for hemoglobinopathy and thalassemia, directing prompt confirmation testing and patient counselling [9].

Conclusion

Peripheral blood film evaluation offers useful morphological clues for early identification, as this retrospective investigation of 50 cases of hemoglobinopathy and thalassemia showed. The most common disorder was beta thalassemia trait, which was followed by sickle cell disease and beta thalassemia major. The most common smear results included microcytosis, hypochromia, target cells, and anisopoikilocytosis. Sickle cells showed a substantial correlation with sickle cell disease, whereas nucleated red blood cells and pronounced poikilocytosis were significantly connected with severe thalassemia. These distinctive patterns can be used to prioritise additional research, including molecular testing, electrophoresis, and HPLC.

Due to its affordability, speed, and accessibility, peripheral smears continue to be very helpful in environments with minimal resources. It can facilitate prompt care by distinguishing genetic haemoglobin opathies from other causes of anaemia when evaluated with full blood count data and clinical history. The study highlights the necessity of integrating morphology into standard haematology practice and providing ongoing training in smear interpretation. It is advised to do larger prospective studies that correlate smear results with genotype and disease severity. Peripheral blood film is still crucial for the screening, diagnosis, and monitoring of individuals with hemoglobinopathy and thalassemia.

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