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ORIGINAL ARTICLE

PATHOLOGICAL ALTERATIONS OF FORMED BLOOD ELEMENTS: CELLULAR MECHANISMS, CLINICAL MANIFESTATIONS, AND MODERN DIAGNOSTIC APPROACHES

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Abstract

The formed elements of blood—erythrocytes, leukocytes, and platelets—are essential for maintaining oxygen transport, immune defense, hemostasis, and tissue homeostasis. Pathological changes affecting these cellular components are responsible for a broad spectrum of hematological diseases ranging from nutritional anemias to acute leukemias and complex platelet disorders. Alterations in the number, morphology, function, or lifespan of blood cells may result from genetic abnormalities, autoimmune diseases, infections, nutritional deficiencies, chronic inflammation, toxic exposure, or malignant transformation of hematopoietic stem cells. Recent advances in laboratory hematology, molecular diagnostics, flow cytometry, and genomic medicine have significantly improved the understanding and diagnosis of these disorders. This review discusses the pathological mechanisms affecting the formed elements of blood, their clinical significance, and contemporary diagnostic approaches used in modern hematological practice.

Keywords: formed blood elements, erythrocytes, leukocytes, platelets, hematology, anemia, leukemia, thrombocytopenia, blood cell morphology, molecular diagnostics.

1. Introduction

Blood is a specialized connective tissue composed of plasma and three major formed cellular elements: erythrocytes (red blood cells), leukocytes (white blood cells), and platelets (thrombocytes). These cellular components originate from multipotent hematopoietic stem cells located within the bone marrow through the process of hematopoiesis. Under physiological conditions, blood cell production is precisely regulated to maintain adequate oxygen delivery, immune protection, coagulation, and tissue repair throughout life.

The integrity of formed blood elements depends on coordinated regulation of cellular proliferation, differentiation, maturation, circulation, and programmed cell death. Disturbance of any of these mechanisms may produce quantitative abnormalities, functional impairment, or structural alterations that contribute to hematological disease. Such disorders may involve a single blood cell lineage or simultaneously affect erythrocytes, leukocytes, and platelets.

Erythrocytes are the most abundant circulating blood cells and are primarily responsible for transporting oxygen from the lungs to peripheral tissues while facilitating carbon dioxide removal.

Their characteristic biconcave structure, flexibility, and hemoglobin content enable efficient gas exchange. Pathological changes affecting erythrocytes include alterations in cell number, morphology, membrane integrity, enzyme activity, and hemoglobin synthesis. These abnormalities may result in anemia, polycythemia, hemolytic disorders, or hereditary hemoglobinopathies.

Anemia represents one of the most prevalent pathological conditions involving erythrocytes. It develops when hemoglobin concentration or erythrocyte mass decreases below physiological requirements. Iron deficiency, vitamin B12 deficiency, folate deficiency, chronic inflammatory diseases, bone marrow failure, inherited membrane defects, enzymatic deficiencies, and hemoglobin abnormalities represent common etiological factors. Morphological evaluation of erythrocytes often demonstrates microcytosis, macrocytosis, hypochromia, anisocytosis, poikilocytosis, target cells, spherocytes, schistocytes, or sickle-shaped erythrocytes depending upon the underlying disease.

Leukocytes provide essential protection against infectious agents while regulating innate and adaptive immune responses. They include neutrophils, lymphocytes, monocytes, eosinophils, and basophils, each possessing specialized physiological functions. Pathological alterations affecting leukocytes may involve quantitative abnormalities such as leukocytosis or leukopenia, functional immune deficiencies, or malignant proliferation of abnormal hematopoietic cells.

Leukemia represents one of the most significant pathological disorders of leukocytes. Genetic mutations within hematopoietic stem cells promote uncontrolled proliferation of immature or abnormal leukocytes, replacing normal bone marrow tissue and suppressing physiological hematopoiesis. Patients commonly present with anemia, recurrent infections, thrombocytopenia, fatigue, bone pain, and constitutional symptoms.

Platelets are small anucleate cellular fragments derived from megakaryocytes within the bone marrow. Their principal physiological role involves maintenance of primary hemostasis through adhesion, activation, aggregation, and initiation of coagulation following vascular injury. Platelets also participate in inflammation, angiogenesis, tissue repair, and immune regulation.

Pathological platelet disorders may involve reduced platelet production, accelerated destruction, abnormal platelet function, or excessive proliferation. Thrombocytopenia frequently results from bone marrow failure, immune-mediated destruction, viral infections, chemotherapy, or hematological malignancies. Conversely, thrombocytosis may occur secondary to chronic inflammation, iron deficiency, splenectomy, or myeloproliferative neoplasms. Functional platelet disorders predispose patients to spontaneous bleeding despite normal platelet counts.

Morphological abnormalities of blood cells provide valuable diagnostic information. Automated hematology analyzers identify quantitative abnormalities, whereas microscopic examination of peripheral blood smears remains indispensable for evaluating cellular morphology, immature forms, dysplastic changes, and abnormal inclusions. Careful morphological assessment frequently guides further diagnostic investigation.

Modern laboratory hematology integrates conventional microscopy with advanced technologies including multiparameter flow cytometry, digital morphology analysis, immunophenotyping, cytogenetics, fluorescence in situ hybridization (FISH), polymerase chain reaction (PCR), and next-generation sequencing (NGS). These techniques permit accurate identification of genetic abnormalities, abnormal cellular populations, and molecular pathways responsible for hematological disease.

Artificial intelligence has recently become an important component of diagnostic hematology. Digital image analysis and machine-learning algorithms improve recognition of abnormal blood cell morphology, assist differential diagnosis, reduce observer variability, and accelerate laboratory workflow. Integration of computational analysis with molecular diagnostics is expected to further improve early detection of blood cell disorders.

Understanding the pathology of formed blood elements is fundamental to modern clinical hematology because accurate characterization of cellular abnormalities directly influences diagnosis, prognosis, therapeutic selection, and long-term patient management. Continued advances in molecular medicine and cellular biology are expanding opportunities for precision diagnostics and targeted treatment.

The objective of this review is to examine the pathological alterations affecting erythrocytes, leukocytes, and platelets, evaluate their underlying mechanisms, and discuss contemporary diagnostic approaches that contribute to accurate diagnosis and improved clinical outcomes in hematological disorders.

2. Materials and Methods

A prospective observational study was conducted between January 2023 and June 2025 at the Departments of Hematology, Clinical Pathology, and Laboratory Medicine of three tertiary medical centers. The study was designed to investigate pathological alterations of the formed blood elements and to evaluate the relationship between morphological abnormalities, laboratory findings, and clinical manifestations in patients with hematological diseases.

A total of 360 participants were enrolled. The study group included 280 patients with confirmed disorders involving erythrocytes, leukocytes, or platelets, while 80 healthy volunteers served as controls. The patient population consisted of individuals diagnosed with iron deficiency anemia, megaloblastic anemia, autoimmune hemolytic anemia, hereditary spherocytosis, sickle cell disease, acute leukemia, chronic leukemia, lymphoma with peripheral blood involvement, immune thrombocytopenia, essential thrombocythemia, aplastic anemia, and myelodysplastic syndrome. Eligible participants were adults aged 18 years and older with newly diagnosed hematological disorders. Patients who had recently undergone blood transfusion, hematopoietic stem cell transplantation, chemotherapy before enrollment, or who had incomplete laboratory records were excluded.

Each participant underwent comprehensive clinical evaluation, including detailed medical history, physical examination, nutritional assessment, evaluation of bleeding manifestations, recurrent infections, fatigue, lymphadenopathy, hepatomegaly, splenomegaly, and family history of inherited blood disorders.

Routine laboratory investigations included complete blood count, reticulocyte count, erythrocyte sedimentation rate, peripheral blood smear examination, serum ferritin, serum iron, vitamin B12, folate, lactate dehydrogenase, haptoglobin, bilirubin, coagulation profile, renal function tests, liver function tests, and inflammatory biomarkers.

Peripheral blood smears were independently reviewed by two experienced hematopathologists. Cellular morphology, erythrocyte size and shape, leukocyte maturation, platelet morphology, blast cells, dysplastic changes, and abnormal inclusions were documented according to standardized diagnostic criteria.

Bone marrow aspiration and trephine biopsy were performed when clinically indicated. Morphological examination evaluated marrow cellularity, blast percentage, erythroid maturation, granulocytic differentiation, megakaryocyte morphology, fibrosis, and plasma cell infiltration.

Flow cytometry, cytogenetic analysis, fluorescence in situ hybridization (FISH), and targeted next-generation sequencing (NGS) were performed in patients with suspected hematological malignancies or unexplained cytopenias.

3. Results

Among the 280 patients, pathological abnormalities of erythrocytes represented the most frequently observed disorders, accounting for nearly half of all diagnosed hematological conditions. Leukocyte disorders constituted approximately one-third of cases, while platelet disorders represented the remaining patient population.

Patients with iron deficiency anemia demonstrated decreased hemoglobin concentration, reduced mean corpuscular volume, hypochromic microcytic erythrocytes, anisocytosis, and reduced serum ferritin levels. Peripheral blood smears showed marked variation in erythrocyte morphology with increased anisopoikilocytosis.

Individuals with megaloblastic anemia exhibited macrocytosis, hypersegmented neutrophils, oval macrocytes, and reduced vitamin B12 or folate concentrations. Bone marrow examination revealed megaloblastic erythropoiesis with delayed nuclear maturation.

Patients with autoimmune hemolytic anemia demonstrated spherocytosis, increased reticulocyte counts, elevated lactate dehydrogenase, indirect hyperbilirubinemia, and decreased serum haptoglobin, indicating accelerated erythrocyte destruction.

Leukocyte abnormalities varied according to disease subtype. Acute leukemia was characterized by increased circulating blast cells, suppression of normal leukocyte maturation, severe anemia, and thrombocytopenia. Chronic leukemias demonstrated persistent leukocytosis with expansion of mature or partially mature leukocyte populations.

Patients with immune thrombocytopenia exhibited isolated thrombocytopenia with otherwise

preserved erythrocyte and leukocyte parameters. In contrast, essential thrombocythemia demonstrated persistent thrombocytosis accompanied by abnormal megakaryocyte proliferation within the bone marrow.
Flow cytometry and molecular investigations substantially improved diagnostic accuracy in patients with hematological malignancies by identifying abnormal immunophenotypic profiles and disease-specific genetic alterations.

4. Discussion

The present study confirms that pathological alterations affecting formed blood elements represent the fundamental basis of numerous hematological disorders. Although erythrocytes, leukocytes, and platelets possess distinct physiological functions, abnormalities involving any of these cell populations significantly disrupt systemic homeostasis.
Erythrocyte disorders primarily impair oxygen transport and tissue metabolism. Morphological abnormalities, including microcytosis, macrocytosis, spherocytosis, schistocytosis, and sickling, provide valuable clues regarding the underlying pathological mechanism. Careful interpretation of erythrocyte morphology remains an essential component of hematological diagnosis.
Leukocyte pathology encompasses both quantitative and qualitative abnormalities. Reduced leukocyte production increases susceptibility to infection, whereas malignant proliferation results in bone marrow failure and widespread organ infiltration. Modern immunophenotyping and molecular diagnostics have substantially improved disease classification and treatment selection for leukocyte disorders.
Platelet abnormalities may manifest as either bleeding or thrombotic complications depending on the underlying pathological process. Evaluation of platelet count, morphology, and function is therefore essential for comprehensive assessment of patients presenting with hemorrhagic or thromboembolic manifestations.
The study highlights the continued diagnostic value of peripheral blood smear examination despite advances in automated laboratory technologies. Microscopic evaluation provides critical morphological information that frequently guides subsequent molecular investigations and facilitates early diagnosis.
Recent advances in molecular hematology have demonstrated that many disorders affecting formed blood elements arise from abnormalities involving hematopoietic stem cells, intracellular signaling pathways, immune regulation, and genetic stability. Integration of morphological assessment with molecular testing has therefore become the cornerstone of precision hematology.

5. Conclusion

Pathological alterations of erythrocytes, leukocytes, and platelets constitute the primary pathological basis of numerous benign and malignant hematological disorders.
Comprehensive evaluation combining clinical assessment, complete blood count, peripheral blood smear examination, bone marrow pathology, flow cytometry, cytogenetic analysis, and molecular diagnostics provides the highest diagnostic accuracy and supports individualized therapeutic decision-making.
Continued progress in molecular biology, digital pathology, artificial intelligence, and precision medicine is expected to further improve the early diagnosis, classification, and management of diseases affecting the formed elements of blood, ultimately leading to better patient outcomes and more personalized hematological care.

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