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Basics of Essential Thrombocytopenia: A Teachers Note

Chidinma Nkolika Ikeani[1], Silas Anayo Ufelle[2], Alphonsus Ogbonna Ogbuabor[1,3], Dorathy Chizoba Onovo[3]

[1]Department of Medical Laboratory Science, Faculty of Health Sciences and Technology, University of Nigeria, Enugu State.

[2]Department of Medical Laboratory Science, Faculty of Health Sciences and Technology, University of Nigeria, Enugu State.

[1,3]Department of Medical Laboratory Science, Faculty of Allied Health Sciences, Enugu State University of Science and Technology, Agbani, Nigeria.

[3]Department of Medical Laboratory Science, Faculty of Allied Health Science, Enugu State University of Science and Technology, Agbani, Nigeria.

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Abstract: **Essential thrombocythemia (ET) is a chronic myeloproliferative neoplasm characterized by persistent elevation of platelet counts due to clonal proliferation of megakaryocytes. It is relatively rare, with an estimated annual incidence of 1–2 cases per 100,000 individuals, and predominantly affects middle-aged and older adults, with a slight female predominance. Clinically, ET may be asymptomatic or present with vasomotor symptoms such as headache, dizziness, erythromelalgia, and visual disturbances; thrombotic and hemorrhagic complications are major concerns. Diagnosis is based on sustained thrombocytosis, bone marrow findings, exclusion of reactive causes, and detection of driver mutations such as JAK2, CALR, or MPL. Prognosis is generally favorable, with near-normal life expectancy in low-risk patients, though complications may impact outcomes. Management focuses on risk stratification, with low-dose aspirin for most patients and cytoreductive therapy, such as hydroxyurea, for high-risk individuals to prevent thrombotic events.**

Keywords: **Essential thrombocythemia, Epidemiology, Presentations, Pathophysiology, Diagnosis, Prognosis, Management.**

1. INTRODUCTION

Essential thrombocythemia (ET) is a chronic myeloproliferative neoplasm characterized by persistent thrombocytosis resulting from clonal expansion of hematopoietic stem cells, with predominant involvement of the megakaryocytic lineage.[1] It is classified among the BCR-ABL1–negative myeloproliferative neoplasms, alongside polycythemia vera and primary myelofibrosis.[2] Recent epidemiological data indicate an annual incidence of approximately 1–3 cases per 100,000 individuals, with higher prevalence in older populations and a slight female predominance.[3] Although often indolent, ET is associated with significant risks of thrombotic and hemorrhagic complications, which remain the principal causes of morbidity and mortality.[4]

Advances in molecular biology have identified key driver mutations, including JAK2, CALR, and MPL, which are integral to disease pathogenesis and form part of current diagnostic criteria.[5,6-9] Clinical presentation varies widely, ranging from asymptomatic thrombocytosis to microvascular symptoms such as headaches, erythromelalgia, and visual disturbances.[6] Contemporary diagnostic and prognostic models now incorporate clinical and molecular factors, enabling improved risk stratification and individualized management approaches.[7] This paper explores the essential aspects of ET, focusing on its prevalence, clinical features, diagnostic evaluation, prognosis, and current therapeutic strategies.

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Epidemiology

Essential thrombocythemia (ET) is a rare myeloproliferative neoplasm with an annual incidence of approximately 1–3 cases per 100,000 individuals and a prevalence estimated at 20–40 per 100,000.[1,3] It predominantly affects adults, with a median age at diagnosis of 60 years, although younger individuals may also be affected.[3,2] A slight female predominance has been consistently reported.[8] Geographic variation is minimal, though improved diagnostic capabilities have increased reported cases globally.[9] ET is often identified incidentally during routine blood tests, contributing to underestimation in earlier studies.[10,11] Survival is generally prolonged, leading to increased disease prevalence despite low incidence.[7]

Clinical Presentations

Clinical presentation of essential thrombocythemia (ET) is highly variable, ranging from asymptomatic cases detected incidentally to symptomatic disease with significant complications.[12] Many patients are diagnosed through routine blood tests showing persistent thrombocytosis.[10] Symptomatic individuals commonly present with microvascular disturbances, including headaches, dizziness, visual disturbances, and erythromelalgia.[6,13,14] Thrombotic events, both arterial and venous, such as stroke or deep vein thrombosis, are major clinical manifestations and contribute significantly to morbidity.[4] Conversely, bleeding complications may occur, particularly in patients with extreme thrombocytosis due to acquired von Willebrand syndrome.[11,15] Splenomegaly is present in a minority of cases and is usually mild.[3] Constitutional symptoms such as fatigue, night sweats, and weight loss are less common but may occur.[8] The heterogeneity in clinical presentation underscores the importance of early recognition and risk stratification to guide management and prevent complications.[16]

Pathophysiology

The pathophysiology of essential thrombocythemia (ET) is driven by clonal proliferation of hematopoietic stem cells, resulting in excessive production of platelets due to abnormal megakaryocyte expansion.[1,17,18] Central to this process are somatic mutations in key driver genes, most commonly JAK2, CALR, and MPL, which lead to constitutive activation of the JAK-STAT signaling pathway.[5,19] This persistent signaling promotes uncontrolled cell proliferation, survival, and differentiation of megakaryocytes. Mutant megakaryocytes exhibit abnormal morphology and increased sensitivity to growth factors such as thrombopoietin, further enhancing platelet production.[20]

In addition to quantitative abnormalities, platelets in ET are often functionally altered, contributing to both thrombotic and hemorrhagic complications.[6,21] Increased interaction between activated platelets, leukocytes, and endothelial cells promotes a prothrombotic state through inflammatory and coagulation pathways.[4,22] Furthermore, elevated platelet counts may lead to acquired von Willebrand syndrome, predisposing patients to bleeding.[11,23] Bone marrow microenvironmental changes and cytokine dysregulation also play a role in disease progression. Overall, ET represents a complex interplay of genetic mutations, abnormal signaling pathways, and cellular dysfunction.

Diagnosis

The diagnosis of essential thrombocythemia (ET) is based on a combination of clinical, hematological, molecular, and bone marrow findings, as outlined by contemporary WHO criteria.[2] A key feature is sustained thrombocytosis, defined as a platelet count $\geq 450 \times 10^9$/L, in the absence of reactive causes such as infection, inflammation, or iron deficiency.[1,24-27] Bone marrow examination typically reveals increased numbers of enlarged, mature megakaryocytes with hyperlobulated nuclei.[1,28] Molecular testing plays a central role, with detection of driver mutations in JAK2, CALR, or MPL supporting the diagnosis.[5,29] Exclusion of other myeloproliferative neoplasms, particularly prefibrotic primary myelofibrosis and polycythemia vera, is essential. Additional laboratory tests, including inflammatory markers and iron studies, help rule out secondary thrombocytosis.[10,30-33] Risk stratification models incorporating age, mutation status, and history of thrombosis further guide clinical decision-making.[7,34] Accurate diagnosis is critical to ensure appropriate management and prevention of complications.

Prognosis

The prognosis of essential thrombocythemia (ET) is generally favorable, with many patients experiencing near-normal life expectancy, particularly those classified as low-risk. Prognostic outcomes are largely influenced by age, history of thrombosis, and molecular profile, including the presence of JAK2 mutations, which are associated with increased thrombotic risk. Risk stratification models, such as the revised International Prognostic Score for ET (IPSET), are widely used to categorize patients and guide management.[7,35] Despite its indolent course, ET carries a risk of complications, including arterial and venous thrombosis, hemorrhage, and, less commonly, progression to myelofibrosis or acute myeloid

leukemia.[4,36] Transformation rates are relatively low but increase with disease duration and certain high-risk features. Overall survival is reduced in high-risk patients compared to the general population.[3,37] Early diagnosis, appropriate risk assessment, and tailored therapeutic interventions are essential to improving long-term outcomes and minimizing disease-related complications.

Management

Management of essential thrombocythemia (ET) is primarily guided by risk stratification aimed at preventing thrombotic and hemorrhagic complications.[1,38] Patients are categorized into low-, intermediate-, and high-risk groups based on age, history of thrombosis, and molecular features such as JAK2 mutation status.[7] Low-risk patients are typically managed with low-dose aspirin to reduce microvascular symptoms and thrombotic risk, provided there are no contraindications.[6]

High-risk patients, particularly those over 60 years or with prior thrombotic events, require cytoreductive therapy in addition to antiplatelet treatment.[4] Hydroxyurea remains the first-line cytoreductive agent due to its efficacy and safety profile.[10,39] Alternative therapies, including interferon-alpha, are preferred in younger patients or during pregnancy. Anagrelide may be used as a second-line agent in cases of intolerance or resistance to first-line treatments. Management also includes control of cardiovascular risk factors and regular monitoring for disease progression or complications. In cases of extreme thrombocytosis with bleeding risk, treatment adjustments are necessary to address acquired von Willebrand syndrome.[11,40] Overall, individualized treatment strategies based on patient risk profiles are essential for optimizing outcomes and minimizing adverse events.

2. CONCLUSION

Essential thrombocythemia is a chronic myeloproliferative neoplasm with variable presentation but generally favorable prognosis. Early diagnosis, appropriate risk stratification, and individualized management are essential to prevent complications. Ongoing advances in molecular understanding continue to improve diagnostic accuracy and therapeutic strategies, ultimately enhancing patient outcomes and long-term disease control.

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