Case Report

# Unmasking Essential Thrombocythemia: Myocardial Infarction as a Rare Complication in a Young Patient


## Abstract:

Essential thrombocythemia (ET) is a rare myeloproliferative disorder characterized by elevated platelet counts and seldom presents with myocardial infarction (MI), particularly in younger individuals without traditional cardiovascular risk factors. We report the case of a 41-year-old woman with no significant medical history who presented with acute chest pain and elevated troponin levels. Her laboratory results indicated a markedly high platelet count, and coronary angiography revealed thrombotic stenosis in the proximal left anterior descending artery. Diagnosis of ET was confirmed by JAK2-V617F mutation and bone marrow biopsy. The patient was initially treated with hydroxyurea to reduce platelet levels, allowing for successful angioplasty three weeks later. This case highlights the importance of considering ET in the differential diagnosis of MI, especially in younger patients without other risk factors, and underscores the need for tailored management and close monitoring in such rare presentations.



**Keywords:** Essential thrombocythemia- Myocardial Infarction- JAK2-V617F mutation


## Introduction:

Essential thrombocythemia (ET) is a myeloproliferative disorder that rarely complicates into myocardial infarction., especially among young individuals. ETis characterized by an abnormal increase in platelet production, seldom manifests through acute coronary events, making such cases a medical rarity.therapeutic management remains a dilemma, given the high thrombotic risk. The implications of this unusual presentation highlight the complexity and diagnostic challenges in identifying ET in younger patients who present with myocardial infarction symptoms. Understanding these rare occurrences is crucial for timely diagnosis and management in clinical practice.

## Clinical case:

We report the case of a 41-year-old woman, with no cardiovascular risk factors or significant medical history, who presented to the emergency department with recurrent retrosternal constrictive chest pain radiating to the left arm, occurring abruptly 24 hours prior to admission. Physical examination revealed no abnormalities. The ECG did not reveal any significant abnormalities.(Figure1). Initial high-sensitivity troponin I was elevated at 4 ng/mL (normal range <0.04 ng/mL). Laboratory investigations revealed a platelet count of 824,000/mm$^3$, hemoglobin level of 15 g/dL, and white blood cell count of 9,000/mm$^3$, with normal

red blood cell counts. Inflammatory markers were negative.the lipid profile was as follows: LDL cholesterol at 0.8 g/l, Triglyceride at 0.72 g/l, HDL at 0.32g/l.HBA1C was 5.1% with correct fasting plasma glucose. The echocardiogram showed predominant segmental hypokinesia in the anterior region at the basal and mid levels witha left ventricular ejection fraction of 66%. The patient received a loading dose of aspirin and 2.5 mg fondaparinux and was referred to the coronary angiography suite within 24 hours. The coronarographyrevealed a long and significant stenosis involving the proximal left anterior descending artery (LAD), encompassing the origin of the first diagonal branch (bifurcation lesion) of thrombotic appearance, the rest of the coronary arteries were lesion-free with smooth arteries(Figure2).After discussion with the catheterizes and given the high risk of stent thrombosis, angioplasty was postponed after investigation and control of platelet levels. Essential thrombocythemia (ET) was confirmed by the presence of the JAK2-V617F mutation, the absence of BCR-ABL1 translocation and compatible bone marrow biopsy results,for which the patient was put on Hydroxyurea with good progression. Percutaneous coronary intervention on the LAD/diagonal branch was successfully performed after achieving a platelet count of less than 500,000/mm³ with hydroxyurea therapy three weeks later. Two drug-eluting stents were placed, and the patient was subsequently put on dual antiplatelet therapy (DAPT).In consultation with hematologists, no JAK2 mutation inhibitor therapy was initiated, and close monitoring was scheduled."

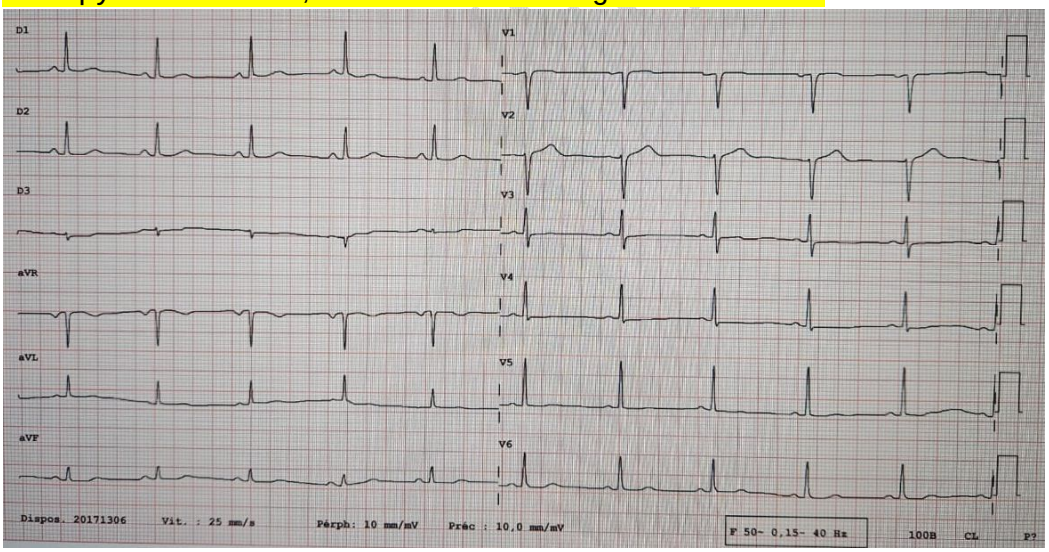


*Figure 1 :The patient's ECG*

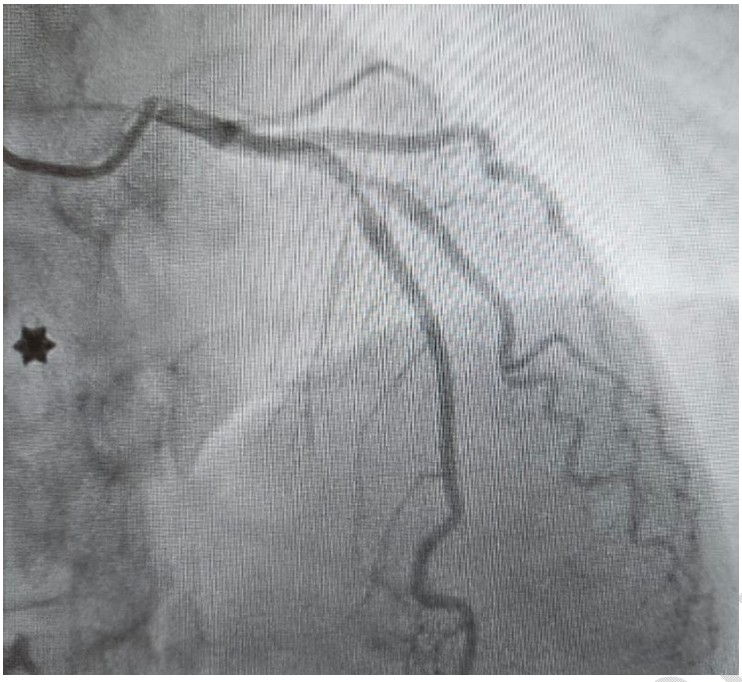

*Figure 2:CoronaryAngiographyShowingSignificantStenosis in Proximal LAD withThrombotic Bifurcation Lesion*

**Discussion :**

Essential thrombocythemia is a chronic myeloproliferative syndrome characterized by a sustained elevation in platelet count exceeding 450,000/mm$^3$. (1)

The diagnosis is made after excluding reactive thrombocytoses (such as inflammatory syndrome, iron deficiency, or asplenia) and other myeloproliferative syndromes.
The primary molecular marker of myeloproliferative syndromes in the absence of the Philadelphia chromosome is the JAK2 V617F mutation. This mutation is present in most cases of polycythemias and in half of essential thrombocythemia cases. (2)
Common clinical manifestations of essential thrombocythemia (ET) arise from thromboembolic events or directly from thrombocytosis itself. (3)
Most individuals with ET experience at least one thrombotic or hemorrhagic event at some point during the course of their disease. (4)

The occurrence of acute coronary syndrome remains rare in cases of essential thrombocythemia. Particularly among young patients, it has been reported that hypertension and cigarette smoking are risk factors for the development of thrombosis in patients with essential thrombocythemia (ET). (5)

Rossi et al. followed 170 patients with essential thrombocythemia (ET) for 10 years. During the follow-up period, only 9.4% of these patients experienced a

myocardial infarction (MI), and among those who had an MI, 75% also had additional cardiovascular risk factors. (6)
Several theories have been proposed to explain the mechanism of coronary thrombosis associated with thrombocytosis. These include abnormalities in the activation of the fibrinolytic system, enhanced procoagulant activity of platelets, and increased blood viscosity.
Inhibiting platelet aggregation and reducing platelet production play a crucial role in the treatment of essential thrombocythemia complicated by coronary thrombosis. (7)

coronary involvement seems to preferentially involve the left anterior descending artery (LAD), as observed in our patient. This may be explained by its sensitivity to endothelial damage, which is a consequence of greater stress related to blood flow. (8)

In the event of acute coronary syndrome occurring in essential thrombocythemia, there are no specific guidelines, and the standard treatment involves dual antiplatelet therapy.
Treatment typically includes cytoreduction, antithrombotic therapy, aspiration thrombectomy, and revascularization with distal protection to prevent distal embolization.
Cytoreduction is recommended before revascularization to mitigate platelet activation and reduce the risk of recurrent thrombosis. (9)

All these patients are classified as high-risk ET. Previous research indicates that a history of thrombosis at diagnosis correlates significantly with recurrent thrombosis. In some instances, despite treatment with antiplatelet agents and hydroxyurea, another thrombotic event may occur. (10)

The follow-up of patients with essential thrombocythemia who have suffered a myocardial infarction lacks specific recommendations.
Hematologically, these patients are considered high-risk and should undergo regular consultations and close monitoring until achieving a complete response, defined by a platelet count below 400 × 10^9/L, white blood cell count below 10 × 10^9/L, and absence of clinical signs of the disease.
Cardiologically, follow-up is similar to other myocardial infarction cases, involving close initial consultations followed by annual visits once the patient is stable and asymptomatic. (11)

Special attention should nevertheless be given to these patients, as the risk of experiencing a first myocardial infarction is 10% in those with essential thrombocythemia, which is twice that of the general population. Moreover, the risk of recurrence also appears to be higher. (12)

**Conclusion :**

While atherosclerosis remains the most common cause of acute coronary syndrome (ACS), resulting from the association of one or more cardiovascular risk factors, there are rarer etiologies that include myeloproliferative syndromes, notably essential thrombocythemia.

This clinical case serves as a reminder that essential thrombocythemia should be considered in the diagnosis when acute coronary syndrome occurs, especially in patients without other cardiovascular risk factors. This remains a rare situation, necessitating a multidisciplinary approach for management.

## Disclaimer (Artificial intelligence)

Author(s) herebydeclarethat NO generative AI technologies such as Large LanguageModels (ChatGPT, COPILOT, etc) and text-to-image generators have been usedduringwriting or editing of manuscripts.

**EthicalApproval:**

**As per international standards or university standards writtenethicalapproval has been collected and preserved by the author(s).**

**Consent**

**As per international standards or university standards, patient(s) written consent has been collected and preserved by the author(s).**

**Refernces** :


1. **Giraudet G, Wibaut B, Ducloy A-S et al.** Thrombocytémieessentielle et grossesse. GynecolObstetFertil. 2011 ; 39(4):205-210.
2. **Tefferi A, Thiele J, Orazi A, Kvasnicka HM, Barbui T, HansonCA, et al.**Proposals and rationale for revision of the WorldHealth Organization diagnostic criteria for polycythemia vera,essential thrombocythemia, and primary myelofibrosis:recommendations from an ad hoc international expert panel.Blood. 2007 ; 110(1): 1092-1097.
3. **Wolanskyj AP, Lasho TL, Schwager SM, et al.** JAK2 mutation in essential thrombocythaemia: clinical associations and long-term prognostic relevance. Br J Haematol. 2005;131(2):208–213.
4. **Vianello F, Cella G, Osto E, et al.**Coronary microvascular dysfunction due to essential thrombocythemia and policythemiavera: the missing piece in the puzzle of their increased cardiovascular risk? Am J Hematol. 2015;90:109–113. doi:10.1002/ajh.23881
5. **Bhat T, Ahmed M, Baydoun H, Ghandour Z, Bhat A, McCord D.** Acute STsegment elevation myocardial infarction in a young patient with essential thrombocythemia: a case with long-term follow-up report. J Blood Med 2014 ;5:123–7.
6. **Rossi C, Randi ML, Zerbinati P et al.**Coronary disease in essential thrombocythemia and polycythemia vera. J Intern Med. 1998 ; 244(1):49-53.

7. **Michaels AD, Whisenant B, MacGregor JS.** Multivessel coronarythrombosis treated with abciximab (ReoPro) in a patient withessential thrombocythemia. Clin Cardiol. 1998 ; 21(2):134-138.
8. **Gao W, Shen W, Luo X, Shi H, Jiang X, Pan J.** ST-segment elevation myocardial infarction in patient with essential thrombocythemia without associated risk. Int J Cardiol 2015;180:223–5
9. **Pósfai É, Marton I, Borbényi Z, Nemes** A: Myocardial infarction as a thrombotic complication of essential thrombocythemia and polycythemia vera. Anatol J Cardiol. 2016, 16:397-402.
10. **De Stefano V, Za T, Rossi E, et al.:** Recurrent thrombosis in patients with polycythemia vera and essentialthrombocythemia: incidence, risk factors, and effect of treatments. Haematologica. 2008, 93:372-80.
11. **Tefferi A, Barbui T.** Polycythemia vera and essential thrombocythemia: 2015 update on diagnosis, risk-stratification and management. Am J Hematol 2015;90(2):162–73.
12. **Tefferi A, Fonseca R, Pereira DL, Hoagland HC**. A long-term retrospective study of young women with essential thrombocythemia. Mayo Clin Proc 2000;76(1):22–8