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Case Report

A Case of Acute ST-Segment Elevation Myocardial Infarction as an Initial Presentation of Essential Thrombocythemia in a Young Healthy Male

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Abstract

In this report, we present the case of a 34-year-old young male from the Middle East who was admitted to the emergency department with ST-elevation myocardial infarction (STEMI) as the first manifestation without known cardiovascular risk factors (CVRFs). On examination, he showed signs of tachypnea and bilateral basal inspiratory crackles. The patient was treated with a loading dose of aspirin, clopidogrel, and anticoagulants, and underwent emergency coronary angiography, which revealed thrombotic occlusion of the proximal left anterior descending artery (LAD). His laboratory investigations revealed increased platelet count, and genetic studies showed a positive JAK2 V617F gene mutation. Upon discharge, he was regularly followed up and remained stable without any further thrombotic or hemorrhagic complications.

Keywords: Essential Thrombocytosis; JAK 2 Mutation; Platelets; ST-Segment Elevation Myocardial Infarction

Introduction

Essential Thrombocythemia (ET), previously known as haemorrhagic thrombocythemia, is a chronic myeloproliferative malignancy characterized by an elevated platelet count and an increased predisposition to thrombotic complications [1-3]. ET patients with high platelet count can experience various clinical manifestations, including both bleeding events and thrombotic complications. Previous evidence has shown that thrombotic complications, particularly arterial thrombosis in the cerebral, coronary and peripheral arteries are more frequent and have a greater impact on morbidity and mortality compared to hemorrhages in ET patients [2]. Approximately 60% of patients with ET harbor a V617F mutation in the Janus Kinase 2 (JAK2) gene, which is responsible for encoding JAK signal transducers in hematopoietic progenitor cells [3]. These JAK signal transducers play a crucial role in initiating signal transduction from receptors of hematopoietic growth factor. Coronary artery thrombosis in ET patients is rare, especially in the absence of traditional risk factors. Additionally, events such as acute coronary syndrome are less likely to occur in patients under the age of forty [4]. Despite its rarity, we present a case of 34-year-old male patient who was previously healthy and devoid of conventional cardiovascular risk factors, who presented with an ST-segment elevation myocardial infarction (STEMI) as the initial clinical manifestation with positive Janus Kinase (JAK2) V617F gene mutation, ultimately leading to the diagnosis of ET.

Case Presentation

A 34-year-old male with no significant past medical or surgical history presented to the hospital with sudden onset chest pain, which was progressive in nature, accompanied by a feeling of impending doom, chest tightness, nausea, and excessive sweating. There was no history suggestive of tobacco or alcohol consumption, and no significant family history. On examination, he was conscious, oriented, moderately built, and adequately

nourished. Physical examination revealed tachypnea, with blood pressure of 135/95 mm Hg, heart rate of 90 beats per minute, respiratory rate of 22 breaths per minute, and oxygen saturation of 98% on room air. Respiratory system examination revealed bilateral basal inspiratory crackles, while there were no abnormal heart sounds or murmurs. Electrocardiography showed sinus rhythm with significant ST-Segment elevation in leads I, aVL, and V1 through V5, consistent with acute anterolateral myocardial infarction (Figure 1).

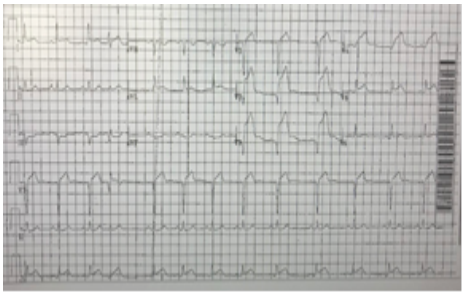

Figure 1: 12 lead ECG shows sinus rhythm with significant ST-Segment elevation in leads I, aVL and V1 through V5 consistent with acute anterolateral MI.

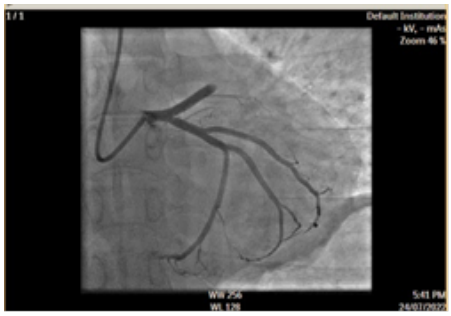

Figure 2: Coronary angiography showing thrombotic occlusion of the proximal left anterior descending artery (LAD).

Routine laboratory evaluation revealed a hemoglobin level of 18.4 g/dL and a platelet count of 1 million, while other laboratory studies were within normal limits. The patient was treated with a loading dose of aspirin, clopidogrel, and anticoagulants. Emergent coronary angiography revealed 95% narrowing with thrombotic occlusion of the proximal left anterior descending artery (LAD) before the first diagonal branch. Other coronary arteries were normal (Figure 2). After manual aspiration thrombectomy, a drug-eluting stent (DES) was successfully placed in the proximal LAD, and thrombolysis in myocardial infarction (TIMI) grade 3 flow was achieved. Subsequently, on day one post percutaneous coronary intervention, an echocardiography revealed a depressed left ventricular ejection fraction (LVEF) of 30% with extended areas of akinesia in the LAD territory. Given the increased platelet count, appropriate genetic studies were performed, which revealed a positive JAK2 V617F gene mutation. Additionally, an ultrasound of the abdomen showed mild fatty liver with mild splenomegaly (13.1cm). Following consultation with a hematologist, the patient was treated with cytoreductive therapy using hydroxyurea along with dual antiplatelet therapy (DAPT). Post-hospitalization, the patient was regularly followed up, and there were no further thrombotic or hemorrhagic complications. The patient remained stable. The thrombectomy was performed by manual aspiration which was followed by stenting. Drug-eluting stent (DES) was placed successfully in the proximal LAD and thrombolysis in myocardial infarction (TIMI) grade 3 flow was obtained. Post Percutaneous coronary intervention, an echocardiography was performed on day one which revealed a depressed Left ventricular ejection fraction (LVEF) of 30 % with extended areas of akinesia in the LAD territory. As there was increased platelet count, appropriate genetic studies were done which revealed positive JAK2 V617F gene mutation. Ultrasound of abdomen showed mild fatty liver with mild splenomegaly (13.1cm). After discussing with hematologist, patient was treated with cytoreductive therapy using hydroxyurea along with dual antiplatelet therapy (DAPT). Post hospitalization patient was followed up regularly and he was stable without further thrombotic or hemorrhagic complications.

Discussion

The low incidence and atypical symptoms of ET often contribute to the misdiagnosis of acute myocardial infarction. The reported incidence rate of MI in patients with ET ranges from 0.38 to 1.7 cases per 100000 individuals per year, with a higher prevalence in females and older age groups [6]. Among these cases, the majority of patients who presented with both MI and ET had occlusion of the LAD artery [4-9]. While several theories have been proposed to explain the occurrence of MI in ET patients, the underlying mechanism is unclear. Previous research has indicated that factors such as cigarette smoking, and hypertension increase the risk of thrombosis development in individuals with ET [2]. However, in our case, the patient was a non-smoker, and did not exhibit any other traditional risk factor for MI including dyslipidemia, diabetes mellitus or hypertension. ET is characterized by a significantly elevated platelet count [7]. This condition can lead to the occlusion of the venous system, arterial system, or microcirculation [8]. In our case, the patient presented with a platelet count exceeding 1 million/μL at admission, indicating a high risk for both thrombosis and hemorrhage. Previous studies investigating the pathogenesis of acute myocardial infarction have emphasized the role of platelet thrombus formation [11], which occurs due to vascular endothelial damage resulting from plaque rupture [10]. To address the heightened thrombotic risk in our patient, platelet-lowering therapy, including cytoreductive therapy with hydroxyurea, was initiated in conjunction with dual antiplatelet therapy (DAPT). This combination was deemed appropriate given his classification

as high risk for thrombotic complications. Hydroxyurea is considered the first-line drug for cytoreductive therapy in ET [11-26]. Its inclusion in the treatment regimen aims to reduce the risk of thrombotic events associated with ET while managing platelet count and minimizing the potential for hemorrhagic complications. Janus kinase (JAK) is a specific type of tyrosine kinase that plays a crucial role in cellular signalling through membrane receptors [9,10]. In the context of essential thrombocytosis (ET), it has been observed that approximately 50% of patients harbor a specific mutation known as JAK2V617F [8]. In our case, we encountered a rare scenario involving a 34-year-old male who had no significant medical or surgical history. He presented with ST-elevation myocardial infarction (STEMI), which was later attributed to the formation of an intracoronary thrombus. Further investigation led to the diagnosis of essential thrombocytosis with the presence of the JAK2V617F mutation. This case serves as a notable example highlighting the association between ET, the JAK2 mutation, and the development of acute STEMI. It underscores the importance of recognizing and investigating the underlying etiology of thrombotic events in individuals with essential thrombocytosis, particularly those who present with acute myocardial infarction.

Conclusions

The absence of preceding cardiovascular risk factors in a patient presenting with acute ST-elevation myocardial infarction (STEMI) should raise suspicion for essential thrombocytosis (ET). This serves as a potential red flag, highlighting the importance of conducting a secondary workup for STEMI cases with isolated intracoronary thrombus formation but otherwise normal coronary vasculature without focal atherosclerosis. Although a rare cause, ET should be included as a differential diagnosis for patients who present with persistently elevated platelet counts and the presence of a thrombus within a patent coronary artery. It emphasizes the significance of considering ET as an underlying etiology in such cases, necessitating further investigation to establish an accurate diagnosis and appropriate management.

Declaration of interest: There are no potential conflicts of interest to be disclosure

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