

Essential Thrombocythemia: A Unique Case

Navyamani V D Kagita MD¹, Keerthy Joseph DO¹, Aisha Sultana Shaik MD¹, Rajesh Thirumaran MD²
Mercy Catholic Medical Center Department of Medicine¹, Department of Hematology & Oncology²

INTRODUCTION:

Essential thrombocythemia (ET) is a chronic myeloproliferative neoplasm, marked by excessive and clonal platelet production, leading to risk of thrombotic and hemorrhagic manifestations. It is a very rare entity with only 1 to 2.5 new cases per 100,000 individuals annually. Here, we present the case of a patient with ET, whose genetic testing resulted negative for all classic mutations usually linked with ET.

CASE:

A 68-year-old African American woman with no significant past medical history was brought to the hospital with complaints of right-sided weakness, facial droop and lethargy of unknown duration. Initial vitals were stable. Labs were notable for platelet count of 1008 Thou/uL, hemoglobin of 8.2 g/dl, WBC count of 15.4 Thou/uL, and INR of 1.3. Computed tomography (CT) brain showed subacute infarct in the left posterior ganglio-capsular region. MRI brain revealed multiple bilateral acute lacunar infarcts, highly suspicious of embolic disease. Aspirin, statin and clopidogrel were started. Initial EKG and telemetry monitoring did not show any arrhythmias. Trans-thoracic echo (TTE) with bubble study was negative for intracardiac thrombi or septal defects. Infectious workup was initiated to further evaluate leukocytosis. Urinalysis, CT chest, abdomen, and pelvis were normal; blood cultures remained negative. Given no clinical signs or symptoms of infections, antibiotics were deferred. Thrombocytosis persisted and peaked to 1318 Thou/uL. Further work-up with flow cytometry was negative for JAK2 V617F, Calreticulin (CAL-R), and myeloproliferative leukemia virus (MPL) mutations. The patient was diagnosed with a stroke secondary to essential thrombocythemia (ET). She was initiated on hydroxyurea, which reduced her platelets to a count of 557 thou/ul. Her neurological status remained the same throughout the hospitalization and was discharged home with recommendations to follow-up with hematologist/oncologist and physical therapist in the outpatient setting.

DISCUSSION:

ET is usually diagnosed in the 60s and is two times more common in women. It manifests with symptoms from infarcts or hemorrhages to vessels. JAK2, CAL-R, MPL are the mutations seen in 90% of the patients. Treatment is directed to prevent future thrombosis and is tailored based on the presence of risk factors, which include history of thrombosis, JAK2/MPL mutation, and advanced age. Our case is rare and unique as it is a case of ET with negative testing for the major mutations. This patient came under intermediate risk with no history of thrombosis, age more than 60 years, unmutated JAK2/MPL. Prognosis is good with a 15 year survival rate of 75%.

REFERENCES:

Sarah Chuzi & Brady L. Stein (2017) Essential thrombocythemia: a review of the clinical features, diagnostic challenges, and treatment modalities in the era of molecular discovery, *Leukemia & Lymphoma*, 58:12, 2786-2798, DOI: [10.1080/10428194.2017.1312371](https://doi.org/10.1080/10428194.2017.1312371)

Tefferi, A., Vannucchi, A. M., & Barbui, T. (2018). Essential thrombocythemia treatment algorithm 2018. *Blood cancer journal*, 8(1), 2. <https://doi.org/10.1038/s41408-017-0041-8>