Impact of JAK2V617F mutational on haematologic features in Sudanese patients with essential thrombocythemia and thrombotic risk assessment

ABSTRACT

Objective: We correlated selected haematological parameters in Sudanese essential thrombocythemia (ET) patients based on their homozygous/heterozygous JAK2V617F genotype, as well as the application of thrombotic risk assessment using different thrombotic risk scoring models. Methods: In this single-center study, we evaluated 60 patients with ET at the time of the diagnosis without any prior treatment. Amplification refractory mutation system-polymerase chain reaction (ARMS-PCR) technique was used to determine JAK2V617F mutation status. Complete blood count was evaluated using the Sysmex analyzer. Furthermore, the thrombotic risk assessment of ET patients using different thrombotic risk scoring models was applied. Results: The JAK2V617F mutation was detected in 29/60 patients (48.3%), of whom 23 (38.3% of total) were heterozygous and 6 (10.0%) were homozygous. Compeered to JAK2 wild-type or JAK2 heterozygous patients, JAK2 homozygous patients for JAK2V617F mutation were associated with older age($p < 0.05$), significantly higher mean leukocytes count ($P = 0.001$), significantly lower Hb concentration ($p < 0.05$), and splenomegaly ($p < 0.05$), while the mean of the platelet counts was slightly higher, although not reached a significant level. We also found two patients who developed thrombotic events throughout follow-up and were initially classified as a low-risk category in the traditional classification. One of them with age < 60 years, hypertension, and JAK2 homozygosity but without thrombosis history, was allocated in a high-risk category by IPSET-t and r- IPSET-t scores. The second patient was stratified in a low-risk category by all scoring models with age < 60 years, hypertension, leukocytosis, unmutated JAK2, and without a history of thrombosis. Conclusions: The JAK2 V617F homozygosity correlated with older age, higher leukocyte count, lower Hb concentration, and a higher risk of thrombosis in Sudanese ET patients. Evaluation of hypertension and identification of JAK2 V617F homozygosity at diagnosis of ET might give the clinician more meaningful prognostic information and so improve the therapeutic management.

Keyword: Essential thrombocythemia; JAK2 mutation; JAK2 V617F homozygosity; Thrombotic risk