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LETTER TO THE EDITOR

Molecular screening for an underlying myeloproliferative neoplasm in rheumatology patients

Stephen Langabeer

St. James's Hospital, Cancer Molecular Diagnostics, Dublin, Ireland

The hematological manifestations of rheumatic diseases are varied with therapeutic agents also capable of inducing hematological abnormalities such as cytopenias.[1] Some of these hematological features can mimic those of the myeloproliferative neoplasms (MPNs) of primary myelofibrosis (PMF) and essential thrombocythemia (ET) such as anemia, neutropenia, thrombocytopenia or thrombocytosis. The most commonly acquired "driver" mutation in PMF and ET is the JAK2 V617F[2] with requests to detect this mutation from rheumatology clinics an infrequent, but recurrent event. An audit was, therefore, performed to address the clinical value of screening for the MPN-associated JAK2 V617F mutation in patients from rheumatology clinics.

Between January 2007 and December 2020, a total of 20,139 requests for JAK2 V617F mutation analysis were received at a center for hematological malignancy molecular diagnostics. The JAK2 V617F mutation detection methodology was unchanged throughout the audit period[3] and of these requests, 3,523 (17.5%) were positive. A total of 89 referrals were identified from rheumatology clinics (48 males, 41 females). The JAK2 V617F was detected in 8/89 (9.0%) patients with hematological details including anemia, pancytopenia, thrombocytopenia, neutrophilia, basophilia, thrombocytosis, or a raised lactate dehydrogenase level. In this rheumatology cohort, the age of the JAK2 V617F-positive patients (median 81 years; range 51-84 years) was higher than those JAK2 V617F-negative patients (median 56 years; range 23-91 years). The final MPN diagnosis was not available in these eight patients.

This brief audit reveals several issues worthy of comment. Firstly, the volume of requests from rheumatology clinics did not impact on laboratory workload. Secondly, although the detection rate of the JAK2 V617F mutation was half that of the non-rheumatology-derived requests, screening did detect molecular evidence of an underlying MPN: it is acknowledged that rheumatic disease co-existing with an MPN is a rare, but reported phenomenon.[4,5] Lastly, the age of JAK2 V617F-positive cases was conspicuously higher than their JAK2 V617F-negative counterparts, suggesting that it is in these more elderly patients in the rheumatology clinic with persistent hematologic abnormalities, that molecular screening for an MPN remains a worthwhile endeavor.

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Correspondence: Stephen Langabeer, MD. St. James's Hospital, Cancer Molecular Diagnostics, D08 W9RT Dublin, Ireland.
Tel: 353 14162413 e-mail: slangabeer@stjames.ie

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