Letter to the editor:

THE JAK2 V617F MUTATION IN RETINAL VEIN OR ARTERY OCCLUSION

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Dear Editor,

The JAK2 V617F mutation is the most commonly acquired driver mutation of the myeloproliferative neoplasms (MPN), detected in more than 95% of patients with polycythaemia vera and in 50-60% of patients with essential thrombocythaemia and primary myelofibrosis (Ferreira Cristina et al., 2018). Patients with these MPN have a considerably increased risk of thrombosis, particularly at uncommon sites (Ball et al., 2018). Risk factors for retinal vein/artery occlusion include atherosclerosis, inflammation and compression, with thrombosis possible due to a malignancy-associated thrombophilic state (Ip and Hendrick, 2018). Retinal vein/artery occlusion is recognised as a rare but recurrent presenting feature of MPN (Tache et al., 2005; Arikan et al., 2011; Dhrami-Gavazi et al., 2015; Rao et al., 2016).

In order to address the clinical value and laboratory impact of requesting JAK2 V617F mutation status in patients with retinal vein/artery occlusion, a retrospective audit was performed on JAK2 V617F requests received at a molecular diagnostics centre for haematological malignancies. From January 2006 to September 2018 inclusive, 17332 diagnostic requests for JAK2 V617F mutation analysis were received. Of these, 29 requests (0.2%) were identified that included clinical details provided of either retinal vein/artery occlusion (n=11) or thrombosis (n=18). The median age was 49 years and comprised 12 males and 17 females. MPN-associated haematological abnormalities were noted as either erythrocytosis (n=4), raised haemoglobin and/or haematocrit (n=6), thrombocytosis (n=9) or not provided (n=10). Using a standardised screening assay unchanged throughout the audit period, the JAK2 V617F mutation was detected in five patients (17.2%) with either raised haemoglobin and/or haematocrit (n=3) or thrombocytosis (n=2).

While the number of requests in patients with a retinal vein/artery occlusion does not appreciably impact on overall laboratory workload, reflexive screening for the JAK2 V617F, particularly in those patients with the aforementioned haematological abnormalities, is justified in order to identify an underlying MPN.
Conflict of interest

The author declares no conflict of interest.

REFERENCES


