

DATE:	2021 September 7
TO:	All clinicians in North Sector
FROM:	Molecular Pathology Laboratory North
RE:	JAK2 Exon 12 Mutation Analysis Ordering Limitations

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Key Message

- Mutations in exon 12 of JAK2 are very rare and should only be tested for in patients with a high suspicion of polycythemia vera (PV) and a negative JAK2 V617F point mutation result. As such these tests should only be ordered in limited circumstances, usually by a treating hematologist.

Background

- Testing for JAK2 mutations is standard in the workup of elevated hemoglobin with normal or depressed erythropoietin levels. The most common mutation, V617F, accounts for >90% of patients with PV, and testing for this specific mutation should be the first screen for PV in patients with elevated hemoglobin. Most patients who screen negative for V617F have a secondary polycythemia and do not need further evaluation for genetic causes of PV. For those rare patients for whom the clinical scenario suggests PV and the JAK2 V617F test is negative, evaluation of JAK2 exon 12 should be undertaken. This test should only be ordered in very limited circumstances, usually by an expert in myeloproliferative neoplasms. For this reason, ordering of JAK2 exon 12 mutational analysis will heretofore be limited to the following circumstances:
 - Patients with hemoglobin greater than 160 g/L and a *previously reported* negative JAK2 V617F result
 - Orders from a hematologist or hematopathologist
- Orders outside of these two circumstances will be cancelled. If JAK2 exon 12 testing is requested and JAK2 V617F test has never been performed, the V617F test will be performed and the exon 12 test will be cancelled.

How this will impact you

- Orders for JAK2 exon 12 received without a prior V617F result will reflex to JAK2 V617F testing with cancellation of the exon 12 test, unless ordered by a hematologist or hematopathologist. Orders for JAK2 exon 12 made concomitantly with the JAK2 V617F request will be cancelled.

Action Required None

Effective September 1, 2021

Questions/Concerns

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Approved by

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