MEMORANDUM

To: Nova Scotia Health Authority Hematologists and Hematopathologists
Provincial Laboratories

From: Dr. J.G. Heathcote, Department of Pathology and Laboratory Medicine
Dr. Wenda Greer, Director of Molecular Diagnostics
QEII Health Sciences Centre

Date: August 20, 2015

Subject: Revised-Calreticulin (CALR) mutation analysis

The Molecular Diagnostics Laboratory is now offering calreticulin (CALR) exon 9 mutation analysis to aid in the diagnosis and prognostication of patients with a myeloproliferative neoplasm.

Test utility

- Mutations in JAK2, MPL and CALR genes are commonly associated with myeloproliferative syndromes and can be useful in distinguishing between the diagnoses of PV, ET and PMF
- CALR mutations are typically variable insertions or deletions in exon 9 and are rarely seen together with mutations in JAK2 or MPL.
- CALR mutations are not associated with PV but have been identified in 67% and 88% of JAK2 and MPL negative cases of ET and PMF respectively (25% and 35% of all ET and PMF cases).
- They have been shown to predict a favorable prognosis in some studies.
- CALR mutations should be considered in light of clinical and pathological findings since they have been observed in rare cases of RARS-T, MDS, CMML and atypical CML.

Specimen/information required

- A peripheral blood (4ml) or bone marrow sample collected on EDTA.
- A copy of CBC result must accompany referred in specimens.

If you have any questions please contact the Molecular Diagnostic Lab at 902-473-6665, Dr Wenda Greer at 902-473-6691 (wenda.greer@nshealth.ca) or Faye Lively at 902-473-6387 (faye.lively@nshealth.ca)

CC. Dr. Irene Sadek, Ms Shauna Thompson, Mr. Randy Veinotte, Ms Faye Lively.