

Date: May 19, 2016

To: All Zones
Physicians, Nurses, Laboratory Directors & Managers, Oncologists and Pathologists

From: Genetic Laboratory Services (GLS)

Re: Calreticulin mutation assay

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Key Messages:

As of Monday, April 4, 2016, the GLS Molecular Pathology Laboratory located at the University of Alberta Hospital has begun to offer a Calreticulin mutation assay.

Why this is important:

Mutations in the calcium-binding endoplasmic reticulum chaperone calreticulin (*CALR*) have recently been shown to be very common in patients with myeloproliferative neoplasms (MPN), affecting ~70% of patients with *JAK2* negative essential thrombocythemia and 60-90% of patients with *JAK2* negative primary myelofibrosis. In the appropriate clinical-pathological context, the presence of the *CALR* gene mutation is consistent with the diagnosis of a clonal myeloproliferative neoplasm. Patients with mutated *CALR* have a lower risk of thrombosis and longer overall survival than patients with mutated *JAK2*. Thus, the identification of the *CALR* mutations in primary myelofibrosis and essential thrombocythemia will help with the diagnosis and predict the prognosis of MPN patients.

Action Required:

Please refer to the AHS Central and Edmonton Zone Test Directory for additional test information at: <http://www.albertahealthservices.ca/3217.asp>

Inquiries and feedback may be directed to:

Dr. Iyare Izevbaye, Lab Head, Molecular Pathology Laboratory, Genetic Laboratory Services, (780) 407-8025

This bulletin has been reviewed and approved by:

Dr. Martin Somerville, Medical/Scientific Director, Genetic Laboratory Services
Dr. Carolyn O'Hara, Interim Provincial Medical Director, Laboratory Services