Calreticulin (CALR) Mutation Detection

Indication: Somatic calreticulin (CALR) mutations have been identified in approximately 20-35% of patients with essential thrombocythemia (ET) and myelofibrosis (PMF). This represents approximately 80-90% of ET and PMF patients who lack somatic JAK2 and MPL mutations. Acquired CALR mutations have also been seen at low levels in patients with RARS-T, MDS, CMML and atypical CML.

Testing Method: Amplicon based targeted next generation sequencing. CALR mutation detection can be ordered as an individual test or as part of Myeloproliferative Neoplasm panel. Both Type 1 (deletion) and Type 2 (insertion) mutations are detected.

Test Parameters: The limit of detection of this assay has been determined to be approximately 5 clonal cells in 100 normal cells.

Turnaround Time: 5-7 business days

Sample Requirements:

Blood - Specimen stability: Ambient - 72 hours; Refrigerated - 1 week

- 3 ml peripheral blood in lavender top tube (EDTA)

  Note: One lavender tube of blood is sufficient for multiple DNA based tests

Bone marrow aspirates (anticoagulated with either heparin or EDTA and, if possible, placed into tissue culture medium) **- Specimen stability: Refrigerated - 1 week** (ship cold)

CPT Code(s): 81219