

**CALR Mutation Analysis, Myeloproliferative Neoplasm (MPN)**  
Test ID: CALR

**USEFUL FOR:**

- Rapid and sensitive detection of insertion and deletion-type mutations in exon 9 of *CALR*.
- Aids in distinction between reactive thrombocytosis and/or leukocytosis versus a MPN, especially essential thrombocythemia (ET) and primary myelofibrosis (PMF), and is highly informative in cases in which JAK2 and MPL testing are negative.
- Especially helpful to the pathologist in those bone marrow cases with ambiguous etiology of thrombocytosis, equivocal bone marrow morphologic findings of MPN, and/or unexplained reticulin fibrosis.
- Aids in prognostication of PMF and thrombosis risk assessment in ET.

**METHODOLOGY:** PCR and Fragment Analysis

**REFERENCE VALUES:** An interpretive report will be provided

**NOTE:** A supplemental report will be provided

**SPECIMEN REQUIREMENTS:**

**The following information is required:**

1. Pertinent clinical history
2. Clinical or morphologic suspicion
3. Date of collection
4. Specimen source

**Forms:** Hematopathology Patient Information Sheet (Supply T676)

**Submit only 1 of the following specimens:**

**Specimen Type:** Peripheral blood

**Container/Tube:** Lavender top (EDTA) or yellow top (ACD solution B)

**Specimen Volume:** 3 mL

**Collection Instructions:**

1. Invert several times to mix blood.
2. Send specimen in original tube.
3. Label specimen as blood.

**Minimum Volume:** 1mL

**Specimen Type:** Bone marrow

**Container/Tube:** Lavender top (EDTA) or yellow top (ACD solution B)

**Specimen Volume:** 2 mL

**Collection Instructions:**

1. Invert several times to mix bone marrow.
2. Send specimen in original tube.
3. Label specimen as bone marrow.

**Minimum Volume:** 1mL

**Specimen Type:** Extracted DNA from blood or bone marrow

**Container/Tube:** 1.5- to 2-mL tube with indication of volume and concentration of the DNA

**Specimen Volume:** Entire specimen

**Collection Instructions:** Label specimen as extracted DNA from blood or bone marrow

**SPECIMEN STABILITY INFORMATION:**

Specimen Type	Temperature	Time
Varies	Ambient (preferred)	7 days
	Refrigerate	7 days

**CAUTIONS:**

- A positive result is not specific for a particular myeloproliferative neoplasm (MPN) diagnosis and clinicopathologic correlation is necessary in all cases.
- A negative result does not exclude the presence of a MPN or other neoplastic process.
- This test is a fragment analysis assay, and only detects insertions and deletions (indels). It will not detect point mutations. However, all reported pathologic mutations in MPN described to date are insertions and/or deletions.
- This test may not differentiate between out-of-frame and in-frame indels in rare cases. However, in-frame indel mutations are very rare (<0.5%), and have only been reported in few healthy individuals and myeloproliferative neoplasm patients with JAK2V617F mutation or out-of-frame CALR mutation. Most of the in-frame indels are considered germline mutations and represent non-pathogenic polymorphisms.
- Infrequently, amplification failure can be encountered in a given sample, due to inadequate DNA, poor DNA quality, or a PCR inhibitor. In these circumstances, the assay will be reattempted and if persistently unsuccessful, the report will be issued with an "Invalid" result.

**CPT CODE:** 81479-Unlisted molecular pathology procedure

**DAY(S) SET UP:** Monday through Friday      **ANALYTIC TIME:** 3 days

**NOTE:** The following referral test code(s) will become obsolete effective May 6, 2014.

Test Name	Test ID	Referral Lab Code	Referral Lab
CALRETICULIN (CALR) Gene Mutations in Myeloproliferative Neoplasms	ZW241	4140	Knight Diagnostic Laboratories

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or Julie Breider, MML Laboratory Technologist Resource Coordinator  
Telephone: 800-533-1710