JAK2 QUALITATIVE

CERNER ORDERABLE
JAK2 QL PCR ; JAK2

CPT CODE
81270

CLINICAL UTILITY
The JAK2 V617F mutation results in unregulated JAK2 tyrosine kinase activity leading to myeloproliferative disorders—polycythemia vera (65-97%), essential thrombocytopenia (23-57%), and idiopathic myelofibrosis (35-57%). It can also be infrequently (3-5%) noted in myelodysplastic syndrome and chronic myelomonocytic leukemia. Studies have also indicated that the presence of the JAK2 V617F mutation is associated with a response to hydroxyurea in patients with essential thrombocythemia and with poorer survival in patients with idiopathic myelofibrosis.

Qualitative JAK2 mutation detection aids in confirmation of diagnosis of polycythemia vera, essential thrombocytopenia, and idiopathic myelofibrosis. Detection aids in confirmation of a clonal hematopoietic stem cell disorder; and/or to confirm myeloproliferative diseases in cases with high erythrocyte, leukocyte, and platelet counts[1].

METHODOLOGY
Qualitative PCR

SPECIMENS
Peripheral blood drawn in a 3 mL or 6 mL EDTA tube or bone marrow drawn in a 3 mL or 6 mL EDTA tube. Minimum acceptable volume is 2 mL. For pediatric patients, an EDTA micro container may be used. Minimum acceptable pediatric volume is 500 µL. Do not spin.

SPECIMEN STABILITY and SHIPPING
Store and ship whole blood or bone marrow specimen refrigerated. Do not spin.

CAUSES FOR REJECTION
Clotted or contaminated sample; if collected in any other anticoagulant.

SPECIFICITY
Validated clinical specificity is 100%. Primers and probes specific for JAK2 V617F mutation. This test does not detect codon 12 mutations or any other mutations.

ASSAY RANGE
Qualitative results (Detected/Not detected)

TURNAROUND TIME
Tuesday, 7 days

1. Reference information can be found in the Indiana University Health Molecular Assay Procedures.