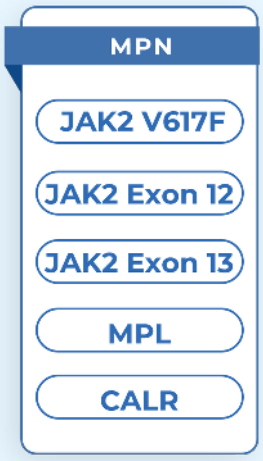
Precipio Spotlight:

MPN Panel that includes JAK2 Exon 13 coverage reveals a rare mutation and co-expression with CALR mutation to support an accurate diagnosis and thus a more effective treatment.

Case Background

- 78 YO | Male
- Unspecified Thrombocytosis
- Suspicious for MPN

Technology Advantage



HemeScreen MPN Panel

More Case Studies

Abstract

This Precipio Spotlight presents a 78-year-old male with unspecified thrombocytosis, whose molecular testing using Precipio's HemeScreen MPN panel revealed a rare JAK2 Exon 13 mutation with a co-expression of a CALR mutation. Most laboratories do not test for JAK2 Exon 13 mutations, nor do they employ a panel approach to detect MPN-related mutations simultaneously. The presence of both mutations alters the prognosis and treatment strategy. While a single CALR mutation typically correlates with fibrosis mutation, its expression alongside JAK2 Exon 13 has prognostic implications. Consequently, the patient is expected to respond favorably to hydroxyurea treatment. Failure to employ a panel approach and include JAK2 Exon 13 in MPN testing might lead to incorrect treatment decisions in this case, underlining the necessity for comprehensive molecular testing.

Case Work Up and Findings

- HemeScreen MPN Panel testing
- Dual JAK2 Exon 13 AND CALR mutations detected

The Precipio Difference:

Precipio's advanced molecular technologies, such as the HemeScreen MPN panel, are critical in diagnosing and managing myeloproliferative neoplasms (MPNs). The identification of a rare JAK2 Exon 13 mutation, in conjunction with a CALR mutation, alters the prognosis and guides treatment decisions. Without this, clinicians may arrive at incorrect treatment conclusions. In this instance, the patient's optimal treatment with hydroxyurea highlights the potential clinical impact of overlooking less common mutations. Integrating advanced molecular testing methodologies into routine clinical practice can significantly enhance diagnostic accuracy and therapeutic efficacy in managing MPNs.

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MOLECULAR

Patient: John Doe
DOB/Gender: XX/XX/19XX (78 yrs) - Male
Patient ID/MRN: 12345
Date Collected: XX/XX/20XX XX:XX

Case#: PXX-XXXXX
Status: Final
Report Category:
Detected

Provider: Jane Smith, MD
XYZ Hematology/Oncology
Tel: + 800-123-4567
fax: +800-765-4321

DIAGNOSIS:

Peripheral blood:
Bloodhound™ MPN results reveal:
- Negative for JAK2 V617F point mutation
- Negative for JAK2 exon 12 mutations
- Positive for JAK2 exon 13 (G571S) mutation at 20.40%
- Negative for MPL W515L/K point mutations
- Positive for CALR exon 9 insertion/deletion mutations at 4.89%

INTERPRETATION

Myeloproliferative neoplasms (MPNs) arise from issues stemming from the bone marrow which lead to abnormally high numbers of certain blood cell types in the blood. Classic MPNs include disorders such as polycythemia vera (PV; Red blood cells), essential thrombocythemia (ET; Platelets), and primary myelofibrosis (PMF; Fibers and blasts).1 Specific variants in the JAK2, CALR, and MPL genes (see above) are useful biomarkers for these diseases as they can play a role in either disease diagnosis or provide information regarding disease prognosis.2-5 These essentially mutually exclusive variants occur in a relatively high frequency, as 98% of sample with PV and 50-65% of samples with ET or PMF exhibit mutations in JAK2. Variants in CALR and MPL are observed in approximately 20-25% or 5-7% of both ET and PMF samples respectively, with only 10-15% of these samples exhibiting triple-negative morphology.1

The mutations covered:
MPL Exon 10: c.1544G>T; p.W515L ; c.1543_1544TG>AA; p.W515K
CALR Exon 9: c.1099_1150del; p.L367fs*46
JAK2 Exon 12: c.1611_1616delTCACAA; p.F537_K539delinsL ; c.1624_1629delAATGAA; p. N542_E543del ; c.1615_1616delAAinsTT; p.K539L
JAK2 Exon 13: c.1711G>A; p.G571S
JAK2 Exon 14: c.1849G>T; p.V617F

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