Precipio Spotlight:

Panel-Based Molecular Profiling Finds CALR Mutation in Patient with Persistent MPN and History of JAK2 Mutation

Case Background

- 76 YO | Female
- Hypertensive
- Previously diagnosed with ET
- Previously JAK2 v617f positive
- Recently negative for JAK2 v617f

Technology Advantage

HemeScreen MPN Panel

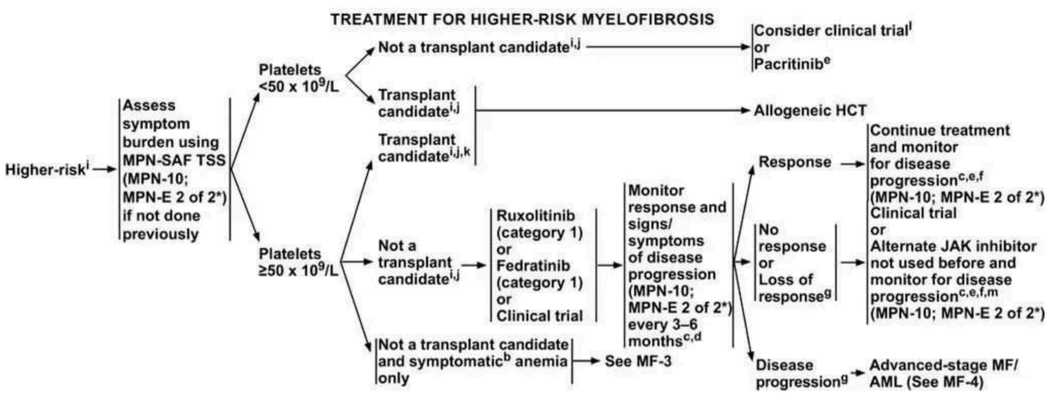
More Case Studies

Abstract

JAK2, CALR, and MPL genes are useful biomarkers for assessing Myeloproliferative neoplasms (MPN) as they can play a role in either disease diagnosis or provide information regarding disease prognosis.(1,2,3,4) Historically, testing for these MPN related mutations has been conducted in a stage & reflex approach, whereby once a single mutation is found, labs halt further testing. Recent studies indicate that dual mutations exist in a subset of the MPN patient population.(5) Therefore, using a stage and reflex approach can lead to missed identification of additional mutations, which can adversely impact care. This case demonstrates the shortcomings of the clinically-outdated stage & reflex approach commonly utilized by other laboratories, and the benefits of Precipio's HemeScreen panel approach.

Case Work Up and Findings

- Omnia ordered by clinician
- HemeScreen MPN Panel testing
- CALR mutation identified



1 Myeloproliferative Neoplasms, Version 3.2022, NCCN Clinical Practice Guidelines in Oncology

The Precipio Difference:
The hematopathology team utilized a molecular panel-based approach to go beyond reflexive JAK2 mutation testing and monitoring, thereby identifying a CALR mutation that may otherwise go undetected. The identification of CALR Exon 9 insertion/ deletion mutations in myeloproliferative neoplasms (MPN) may be useful to assist diagnosis, classification, and monitoring. The presence of CALR mutations have been associated with more benign clinical courses in comparison to corresponding disorders associated with JAK2 or MPL mutations.

For more information please contact:
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Patient: Jane Doe
DOB/Gender: XX/XX/19XX (76 yrs) - Female
Patient ID/MRN: 12345
Date Collected: XX/XX/20XX XX:XX

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

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

DIAGNOSIS:

Clinical history of thrombocytosis with treatment for (JAK2-) myeloproliferative neoplasm.
CALR exon 9 mutation detected by HemeScreen^{TM} MPN. See Comment.

COMMENT

In the context of this patient's long standing clinical history of thrombocytosis and current peripheral blood picture (platelet count> 1 milliion), The findings in this study are consistent with a CALR+ myeloproliferative neoplasm/essential thrombocytosis (CALR+ MPN/ET). Clinical correlation is recommended. If clinical concerns persist, bone marrow biopsy/comprehensive study (Omnia) may be helpful.

All myeloid and lymphoid neoplasms are now classified and named in accordance with the newly revised 2017 version of the WHO Classification of Tumours of Haematopoietic and Lymphoid Tissues.

See below for summary of testing.

COMPONENT DIAGNOSES

Peripheral Smear: Marked thrombocytosis.
Mild anemia.
No evidence of circulating (increased) blasts.

Flow Cytometry: There is no diagnostic flow cytometric evidence of T or B cell neoplasia, dysmyelopoiesis or increased blasts.

FISH: No evidence of an acquired clonal abnormality, see findings below.

Molecular: HemeScreen^{TM} MPN results reveal:
- Negative for JAK2 V617F point mutation
- Negative for JAK2 exon 12 mutations
- Negative for JAK2 exon 13 (G571S) mutation
- Negative for MPL W515L/K point mutations
- Positive for CALR exon 9 insertion/deletion mutations* see comments

CLINICAL DATA

ICD-10: D47.3, D64.9. Thrombocytosis. Anemia. Under Therapy.

Received CBC, reported on 10/04/2022: WBC 4.58; RBC 3.37; HGB 10.72; HCT 30.5; MCV 90.6; MCH 31.8; MCHC 35.1; RDW 14.1%; PLT 1438.3; MPV 7.8; LYM 22.02%; GRAN NP; MID NP; MON 14.61%; NEU 61.26%; EOS 1.66%; BAS 0.44%; (NP = not provided)