Precipio Spotlight:

Omnia™ Methodology Enables Rapid and Cost Effective Diagnosis of MDS With Peripheral Blood Instead of Bone Marrow Biopsy

Case Background

- 82 YO | Female
- Transfusion-dependent anemia and pancytopenia
- Progressive weakness

Technology Advantage

HemeScreen Cytopenia Panel

More Case Studies

Abstract

Myelodysplastic syndrome (MDS) is a clonal disorder of hematopoietic stem cells that is characterized by morphological dysplasia in one or more major hematopoietic cell lines. The clinical presentation of MDS can range from asymptomatic to life-threatening, depending on the severity of cytopenias, with some transforming to acute myeloid leukemia.

Recent advances in molecular technologies, the 2023 NCCN guidelines allow for diagnosis of MDS on peripheral blood alone. This enables diagnosis of MDS without the need for painful and costly bone marrow biopsy. Here we present a case where using the Omnia™ methodology, supported by advanced molecular techniques enabled the Precipio lab to diagnose a patient with MDS using only a peripheral blood sample.

Case Work Up and Findings

- The flow cytometry indicated granulocytes with a left-shift, aberrant CD56 expression, and abnormal CD13/CD16 maturation pattern suggestive of dysmyelopoiesis. Monocytes decreased from previous results. The lymphocytes (24%) included 3% polyclonal B-cells, 84% mature T-cells with a normal CD4/CD8 ratio, and 12% natural killer (NK) cells.
- Molecular analyses were performed using the HemeScreen® Cytopenia panel revealing positive ASXL1 (additional sex combs like 1) and WT1 (Wilms' Tumor 1) mutations.
- The findings point to an underlying myeloid stem cell disorder, including a myelodysplastic syndrome and possibly a higher grade myeloid neoplasm.

Clinical Implications

ASXL1 mutations have been associated with an adverse prognostic impact on overall survival in patients with MDS and an increased likelihood of transformation to AML.

Studies, of large MDS cohorts have established WT1 mutations as an independent predictor of poor prognosis. These studies have also demonstrated a correlation between WT1 mutations and lower hemoglobin levels, as well as a higher percentage of bone marrow blasts.

The Precipio Difference:
Obtaining accurate diagnoses through minimally invasive methods enables less painful and more efficient patient work-ups. Keeping current with the latest guidelines to support triage and testing advancements provided for more comprehensive care for this patient without prolonged, invasive sample collection visits.

For more information please contact:

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COMPREHENSIVE REPORT

Patient: Jane Doe
DOB/Gender: XX/XX/19XX (82 yrs) - Female
Patient ID/MRN: 12345
Date Collected: XX/XX/20XX XX:XX

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

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

DIAGNOSIS:

Myeloid neoplasm with dysplastic features, see comment.

COMMENT

The findings of marked anemia, minimal thrombocytopenia, circulating blasts (<5%), aberrancies by flow cytometry, and an ASXL1 mutation are diagnostic for an underlying myeloid stem cell disorder, most likely a myelodysplastic syndrome. However, correlation with the bone marrow biopsy findings, especially the blast count, as well as more extensive genetic studies including karyotype, is needed for final interpretation.

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: Blasts present, 3%.
Left-shifted neutrophilic granulocytes.
Marked anemia.

Flow Cytometry: Myeloid aberrancies and expanded population of myeloblasts (3.6%), see comment.

Molecular: HemeScreen™ Anemia results reveal:
- Positive for ASXL1 mutations* see comments
- Positive for WT1 mutations* see comments
- Negative for DNMT3A mutation
- Negative for RUNX1 mutations
- Negative for SF3B1 mutations

CLINICAL DATA

ICD-10: D61.818, D64.9. Other pancytopenia. Anemia, unspecified.

Received CBC, reported on 07/18/2022: WBC 4.73; RBC 2.17; HGB 6.9; HCT 21.3; MCV 98.2; MCH 31.8; MCHC 32.4; RDW 15.9%; PLT 143; MPV 10.4; LYM 30.0%; GRAN NP; MID NP; MON 7.0%; NEU 48%; EOS 2.0%; BAS 0.0%; (NP = not provided)

Electronically Signed By: Gail Bentley, MD (08/18/2022 13:50)