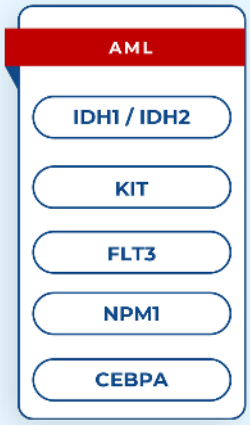
Precipio Spotlight:

Omnia Methodology Specifies Rare Hairy Cell Leukemia Variant and Uncovers Unsuspected AML

Case Background

- 91 YO | Male
- Suspicious for Hairy Cell Leukemia
- Initial labs show pancytopenia
- No previous labs or suspicion for AML

Technology Advantage



HemeScreen AML Panel

More Case Studies

Abstract

Acute myeloid leukemia, myelodysplasia-related (AML-MR) is a type of acute myeloid leukemia (AML) that is characterized by the presence of genetic mutations that are also found in myelodysplastic syndromes (MDS). By definition, AML presents as an acute condition that requires immediate intervention. If AML is not clinically suspected, required testing to confirm AML will not be ordered; or more comprehensive testing that may help with AML diagnosis will be reported at a surface level such as a reporting of blasts on a flow cytometry work up. In this case, using Precipio's Omnia approach, additional clinical information was used to identify a rare variant of HCL (BRAF mutation negative), and early AML, a clinical scenario that may warrant faster, more aggressive intervention.

Case Work Up and Findings

- Omnia Work-Up
- Cytogenetics shows complex karyotype
- BRAF negative HCL identified
- Emerging AML uncovered
- AML-MR with bi-allelic TP53 mutations.
- Low level involvement by B cell lymphoma, hairy cell leukemia variant (10%)
- Mild kappa light chain predominant plasmacytosis (5%).

Clinical Implications

- Bi-allelic TP53 and RB1 mutations suggests a poor prognosis.
- SF3B1 mutation is associated with a favorable prognosis.
- Prognostic implications of CRLF2 and ERG mutations in AML are unknown.
- Therapeutic options for TP53-mutated AML include Aurora kinase A inhibitors, Wee1 inhibitors, Chk1 inhibitors, kevetrin, APR-246, nutlins, and potentially gene therapy approaches.
- Potential therapeutic strategies of RB1 mutations include Aurora A kinase inhibitors or BCL2 inhibitors in combination with cisplatin-based therapy.

The Precipio Difference:

As part of the Omnia process, appropriate testing and communication allowed for proper classification of the variant form of hairy cell leukemia, and more importantly, a diagnosis of emerging AML. While prognosis in this case is not favorable, by working with Precipio, the treating physician was able to make better informed treatment decisions. Prior lab work-ups from other reference labs failed to uncover this HCL variant and emerging AML condition.

For more information please contact:

contact@precipiodx.com | www.precipiodx.com | Tel: +1.203.787.7888

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COMPREHENSIVE REPORT

Patient: John Doe
DOB/Gender: XX/XX/19XX (91 yrs) - Male
Patient ID/MRN: 12345
Date Collected: XX/XX/20XX XX:XX

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

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

DIAGNOSIS:

Relative and absolute granulocytosis with absolute monocytosis with no detected genetic abnormalities (see comment).
Minute kappa monoclonal B-cell population, 7% of lymphocytes, <1% of total leukocytes, consistent with low level monoclonal B cell lymphocytosis (MBL).

COMMENT

Neutrophilia and monocytosis may be associated with infectious disease, effects of some drugs or hormones (lithium corticosteroids), inflammatory (autoimmune) disorders, certain metabolic states (e.g. ketoacidosis), inflammatory bowel disease, celiac sprue, cytokine therapy, acute injury/stress, smoking, post-splenectomy status, and in myeloid neoplasms. FISH and molecular studies are negative, however this does not completely exclude the possibility of a myeloid neoplasm. If clinical concerns persist, a bone marrow biopsy may be helpful. See below for summary of testing.

Flow cytometry detected a minute monoclonal B-cell population (<1% of total leukocytes). The current WHO/IWCLL criteria designate such small populations (<0.5 x 10e9/L absolute count) as "low count" monoclonal B-cell lymphocytosis (MBL), unless the patient has evidence of extramedullary disease. Low count MBL has an extremely limited chance of progression to CLL.

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.

COMPONENT DIAGNOSES

Peripheral Smear: Relative and absolute granulocytosis with absolute monocytosis.

Flow Cytometry: Minute kappa monoclonal B-cell population, 7% of lymphocytes, <1% of total leukocytes, consistent with low level monoclonal B cell lymphocytosis (MBL)
There is no phenotypic evidence of a B cell neoplasm, dysmyelopoiesis or increased (circulating) blasts.

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

Molecular: HemeScreen™ 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
- Negative for CALR exon 9 insertion/deletion mutations