Acute Promyelocytic Leukemia: Guideline to Diagnosis and Follow-up

Clinical or morphologic suspicion for acute promyelocytic leukemia (APL)

Testing begins with:
- LCMS / Leukemia/Lymphoma Immunophenotyping, Flow Cytometry, Varies
- Cytochemical stains as determined by reviewing pathologist

Bone marrow morphology and/or flow cytometric immunophenotyping: acute leukemia, possible APL?

On bone marrow specimen, order:
- AMLMF / Acute Myeloid Leukemia (AML), Specified FISH, Varies (PML/RARA only)
- CHRB M / Chromosome Analysis, Hematologic Disorders, Bone Marrow
- PMLR / PML/RARA Quantitative, PCR, Varies

Positive for t(15;17)(q24;q21) and PML/RARA

Diagnostic for APL

APL follow-up

Chromosome and genetic studies for PML/RARA are not recommended at end of induction.
Monitoring after consolidation therapy should be performed by quantitative reverse transcriptase-polymerase chain reaction (RT-PCR) for PML/RARA; FISH is not recommended for residual disease evaluation.
Peripheral blood studies for residual disease in APL are less optimal; bone marrow studies are generally more informative.

Negative PML/RARA but possible alternate RARA translocation*

Alternate RARA locus translocations present by cytogenetics/flourescence in situ hybridization (FISH)

APL with a variant RARA translocation

Follow Acute Myeloid Leukemia: Testing Algorithm

Not diagnostic of APL (APL with PML/RARA)

*Note: In some cases of AML with APL-like features PML-RARA is not observed and alternate translocations involving the RARA locus can occur.

Examples:
ZBTB16/RARA; NUMA1/RARA; NPM1/RARA; STAT5B/RARA

RT-PCR for PML-RARA will not detect rare alternate RARA fusions in such cases.