## Sample Case 2: AML MRD confirmed by FISH

## "Difference from Normal" Multidimensional Flow Cytometry Results

Specimen Type: Bone Marrow Aspirate

**Clinical History/Indications:** A xy year old patient with a clinical history of acute myeloid leukemia with t(9;11). Previous flow cytometric findings revealed no evidence of aberrant myeloid antigen expression or abnormal myeloblasts.

Analysis/Conclusions: The flow cytometric findings reveal an aberrant myeloblast population present at 0.3% of total non-erythroid cells, consistent with reemergence of AML (see comment). Histopathologic, cytogenetic, and clinical data are required for complete interpretation. Comment: The abnormal myeloid progenitor cell population must be confirmed by FISH and/or PCR studies for t(9:11). Please contact the laboratory for additional testing.

**Flow Cytometric SSC/CD45 Differential:** 31% lymphocytes, 2.6% monocytes, 53% myeloid forms (all stages of maturation), 3.9% lymphoblasts, and 0.3% abnormal myeloid progenitor cells

**Immunophenotypic Findings:** Independent immunophenotypic analysis of the myeloblast population reveals abnormal surface antigen expression consisting of HLA-DR, heterogeneous CD34, CD13, CD123, CD33, dim CD38 and increased CD117. The myeloblasts lack expression of CD11b, CD14, CD16, CD36, and all additional lymphoid antigens tested including CD19. The blasts are intermediate in size by light scatter. Total non-erythroid cells expressing CD34 are present at 3.2%

## Flow Cytometric Sorted FISH Results

Specimen Type: CD117+/CD38 dim flow cytometry sorted cell population from Bone Marrow Aspirate

Clinical History/Indications: A xy year old patient with a clinical history of acute myeloid leukemia with t(9;11). Current flow cytometric findings reveal an aberrant myeloblast population present at 0.3% of total non-erythroid cells, consistent with reemergence of AML.

FISH (fluorescence in situ hybridization) Result: <u>ABNORMAL</u> Positive for t(9;11) nuc ish(MLLT3,MLL)x3,(MLLT3 con MLL x2)[66/201]

## **Interpretation:**

- These findings are consistent with the presence of an aberrant myeloblast clone characterized by t(9:11).
- Consistent with acute myeloid leukemia reoccurrence.
- Clinical and hematopathology correlation is recommended.

Fluorescence-activated cell sorting was utilized to isolate the CD117+/CD38 dim myeloblast cell population. FISH (fluorescence in situ hybridization) was performed on the isolated cells using the MLL(11q23)(/MLLT3(9p21;AF9) dual fusion translocation probe, specific for the t(9;11) associated with adult and childhood ALL as well as AML. This probe set allows detection of the MLL/MLLT3 gene fusion on both the derivative 9 and the derivative 11 chromosome and allows detection of a translocation and/or a deletion at one of the breakpoints. Of a total of 200 interphase nuclei examined, 33% showed an abnormal positive double fusion signal, indicative for t(9;11).

These findings cannot be used for quantification purposes, since only CD117+/CD38+ cells were analyzed.

FISH Analysis Summary: Number of Cells Analyzed: 200 Cells Analyzed: Interphase Probes Utilized: MLL/MLLT3

Source and Lot Number: Kreatech, 43950

Control Probe Utilized: database

