## Sample Case 4: Need for Clinical Cell Sorting to Confirm AML MRD by NGS

**Next Generation Sequencing Results on unseparated Specimen**

**Specimen Type:** Bone Marrow Aspirate (un-separated specimen)

**Clinical History/Indications:** A xy-year-old patient with a clinical history of AML. Flow cytometry revealed acute myeloid leukemia at 1.3%.

**Extended AML (NGS) mutation panel results: SUSPICIOUS (FLT3-ITD)**

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**Analysis/Conclusions:**

* The specimen tested ***suspicious*** for the presence of an insertion mutation in Exon 14 of the **FLT3 gene** with a variant allelic frequency of 2.5% below the detection level of this analysis. Activating mutations of the fms-like tyrosine kinase 3 receptor have been described to be an important prognostic factor in AML and are associated with worse prognosis [Thiede et al. Blood 2002, 99:4326-4335] [Kottaridis et al. Blood 2001, 98:1752-1759]. Clinical and histological correlation required for comprehensive interpretation

**NGS Results on Flow Cytometric Sorted Cells**

**Specimen Type:** Flow cytometry sorted CD34+ cell fraction of Bone Marrow Aspirate

**Clinical History/Indications:** A xy-year-old fe/male with a clinical history of AML. Flow cytometry revealed acute myeloid leukemia at 1.3%. (See separate report).

**Extended AML (NGS) mutation panel results: POSITIVE (FLT3-ITD)**

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**Analysis/Conclusions:**

* The specimen tested ***positive*** for the presence of an insertion mutation in Exon 14 of the **FLT3 gene** in the CD34-positive sorted myeloblast population of this specimen. Activating mutations of the fms-like tyrosine kinase 3 receptor have been described to be an important prognostic factor in AML and are associated with worse prognosis [Thiede et al. Blood 2002, 99:4326-4335] [Kottaridis et al. Blood 2001, 98:1752-1759]. Clinical and histological correlation required for comprehensive interpretation.