Hematologics, Inc.

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RECEIPT DATE: UNITS: 1 Microarray

Patient Name HLID#

Specimen Type: Bone Marrow Aspirate

Clinical History/Indications: A xx year old male with a listed clinical history of myelodysplastic syndrome (MDS). Previous flow cytometry results revealed abnormal myeloid antigen expression and abnormal myeloblasts in 1.2% of the bone marrow, **FISH and cytogenetic studies were negative** (see separate reports).

CLINIC ID#:

ACCOUNT: CPT:

aCGH/SNP Microarray : <u>ABNORMAL</u>

Positive for gain of 8q24.13-q24.21 (4 Mb) [including c-MYC]

Interpretation:

- These findings reveal the presence of an aberrant cell population harboring a 4 Mb gain at 8q24.
- Amplification of the c-MYC gene (located at 8q24) has been reported in MDS and AML [Storlazzi et al. 2006 Hum Mole Gene 15/6].
- Clinical and histological correlation required for definitive diagnosis.

The presence of chromosome aberrations and LOH*/UPD** below the detection sensitivity of this analysis (20% for gains and losses; 1 Mb in length for gains or losses and 20 Mb for LOH/UPD) cannot be completely ruled out.

*Loss of heterozygosity (LOH) is defined as consecutive homozygous SNP tracts exceeding 1Mb in length. LOH can result from deletion of all copies of one homolog or from uniparental disomy** (UPD) in which both copies, or region, of a chromosome pair have the same parental origin. Regions of UPD are continuous stretches of homozygous SNP calls (LOH) *without copy number loss*. Both LOH and UPD play known roles in tumorigenesis.

Method: Genomic DNA was isolated from the whole bone marrow aspirate. Oligonucleotide array-based comparative genomic hybridization (array CGH) and SNP (single nucleotide polymorphism) analysis of 180,000 genomic loci, including ~20,000 cancer-associated CGH as well as ~60,000 SNP probes, was performed on the extracted DNA sample and referenced to a normal male DNA sample.



