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HLID#: **PATIENT NAME:**
PATIENT ID#: **DOB :** **SEX:** F
NPI: **ORDERING PHYSICIAN:**
SPECIMEN TYPE: Bone Marrow Aspirate
COLLECTION DATE: **RECEIPT DATE:**
REPORT DATE: **ICD-9:** **UNITS:** 1 CGH/SNP Microarray
CLINIC ID#:
ACCOUNT:
CPT:

Patient XY HLID#

Specimen Type: CD34+ enriched cell population from Bone Marrow Aspirate

Clinical History/Indications: A xx year old female with a clinical history of *myelodysplastic syndrome*. Previous cytogenetic and FISH studies were *negative* (see X-xxxx).

CD34+ cell - aCGH/SNP Microarray: ABNORMAL

- **Loss of heterozygosity* (LOH) of 7q**
- No other chromosome aberrations were detected

Interpretation:

- **These findings reveal the presence of an aberrant CD34+ cell population harboring a loss of heterozygosity* (LOH) of the long arm of chromosome 7.**
- ***Uniparental disomy** (UPD) affecting 7q has been associated with MDS.***
- ***7q UPD confers an intermediate/poor prognostic association.*[1]**
- Clinical and histologic correlation is requested.

Method: CD34+ progenitor cells were isolated by magnetic-activated cell sorting using anti-CD34 immunobeads and a magnetic-activated cell sorter (MACS, Miltenyi Biotec) separation system. 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 female DNA sample. *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.*

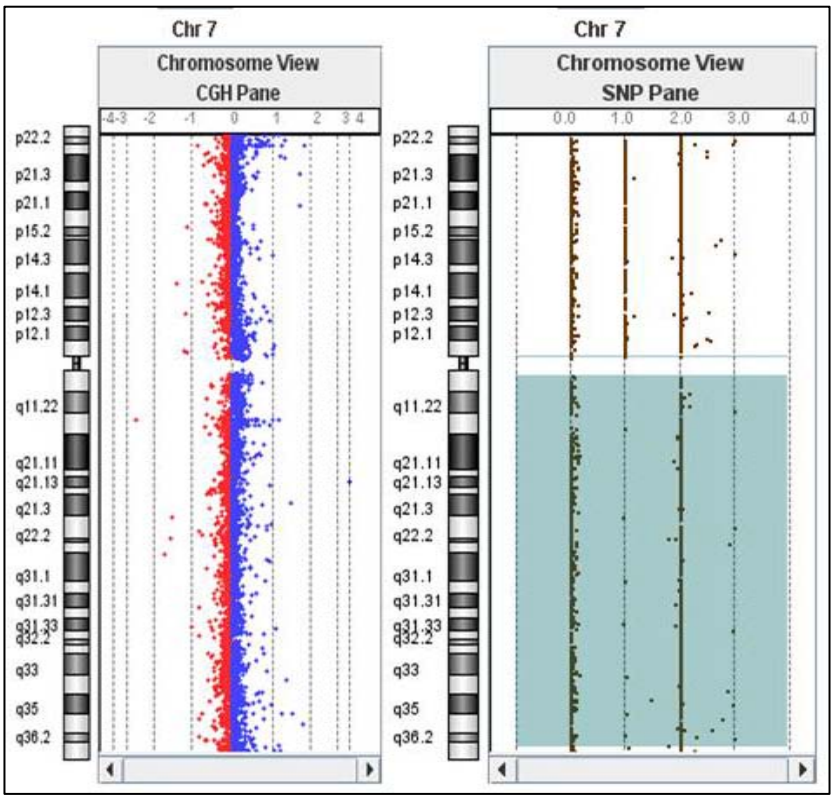

Image: Demonstrates 7q LOH.

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*__Loss of heterozygosity__ 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.

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[1] Mohamedali et al BLOOD 2007 110/9

Electronically signed by: Barbara K. Zehentner, Ph.D., HCLD (ABB), Director of Molecular Analysis and Denise A. Wells, MD, Medical Director