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.

*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.

¹ Mohamedali et al BLOOD 2007 110/9