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HLID#: chromo **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 Microarray
CLINIC ID#:
ACCOUNT:
CPT:

Patient XY HLID#

Specimen Type: CD138+ enriched cell population from Bone Marrow Aspirate

Clinical History/Indications: A xx year old female with a clinical history of multiple myeloma. Previous MM FISH studies on CD138+ enriched cell population revealed an abnormal cell clone characterized by monosomy 13 with indeterminate prognostic significance (see separate report for details).

Plasma cell - CGH Microarray : ABNORMAL

Results: Monosomy 12
Monosomy 13
Monosomy 22
Chromothripsis* of chromosome 17

Prognostic Group: High Risk

Interpretation:

- **These findings reveal the presence of an aberrant CD138+ plasma cell population harboring a loss of chromosome 12, 13, 22 in addition to chromothripsis* of chromosome 17.**
- ***Chromothripsis has been reported as a rare event in multiple myeloma and has been linked to poor prognosis*** *(Blood. 2011 Jul 21;118(3):675-8. Epub 2011 May 31)***.**
- Clinical and histological correlation required for definitive diagnosis.

*Chromothripsis detected by microarray analysis defines genomic instability featuring a large amount of chromosomal rearrangements involving a localized genomic region. Chromothripsis has been postulated as a novel clonal evolution process occurring as a single event, with a single or a limited number of chromosomes shattering into pieces and subsequently being spliced back together producing highly complex derivative chromosomes.

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 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: CD138+ plasma cells were isolated by magnetic-activated cell sorting using anti-CD138 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 was performed using the Agilent SurePrint G3 Cancer CGH+SNP microarray with 25 KB overall median probe spacing (Design based on UCSC hg19 [NCBI Build 37, February 2009]). The array features 180,000 genomic loci, including ~20,000 cancer-associated CGH as well as ~60,000 SNP probes. The assay was performed on the extracted DNA sample and referenced to a normal female DNA sample.

PATIENT NAME:

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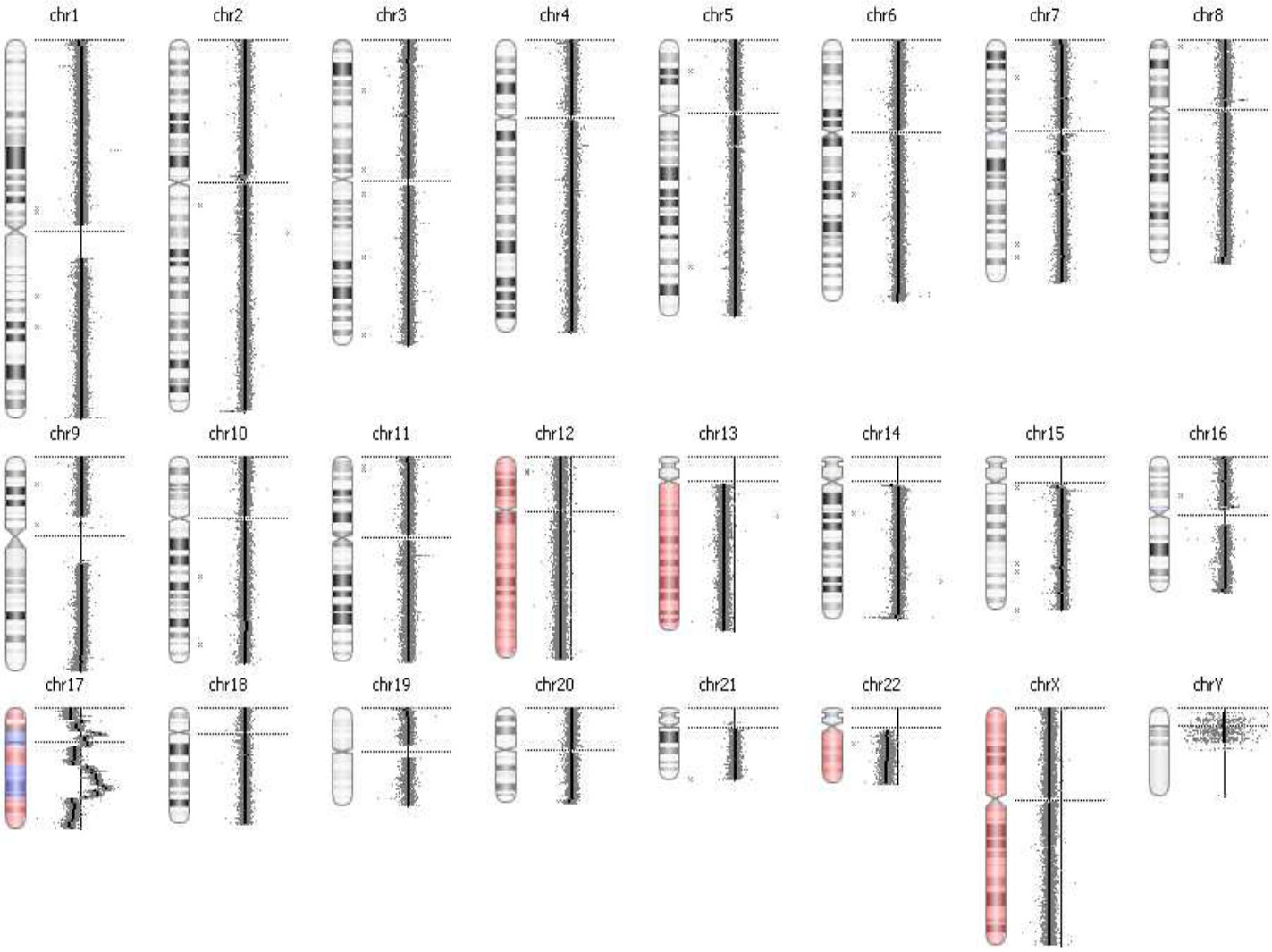

Electronically signed by: Barbara K. Zehentner, Ph.D., HCLD (ABB), Director of Molecular Analysis and Denise A. Wells, MD, Medical Director

This test was developed and its performance characteristics determined by HematoLogics, Inc. It has not been cleared or approved by the US Food and Drug Administration