CLINICAL TEST RESULTS
OGM-Dx HemeOne

bionano laboratories

Patient Name: Sample Report	**Provider Name:** Dr. XXXX XXXXXXX	**Sample Type:** Whole Blood
Patient Date of Birth: XX/XX/XXXX	**Provider Phone:** XXX-XXX-XXXX	**Sample Collected:** XX/XX/XXXX
Patient Sex: XXXXX	**Provider Fax:** XXX-XXX-XXXX	**Sample Received:** XX/XX/XXXX
Accession Number: XXXXXXXXXX	**Provider email:** XXX@XXXXX.XXX	**Order Date:** XX/XX/XXXX
Testing Indication: XXXXXXXXXX	**Provider Institution:** XXXXXXXXXX	**Report Issued:** XX/XX/XXXX

Test Result Summary: Positive, Simple Genome

ISCN Nomenclature: ogm(7)x1~2

Results Summary:

Section A Guideline driven variant analysis of known Tier 1A pathogenic variants was **positive for a mosaic monosomy 7 (loss of entire chromosome 7).**

Section B Whole genome structural variant analysis was negative.

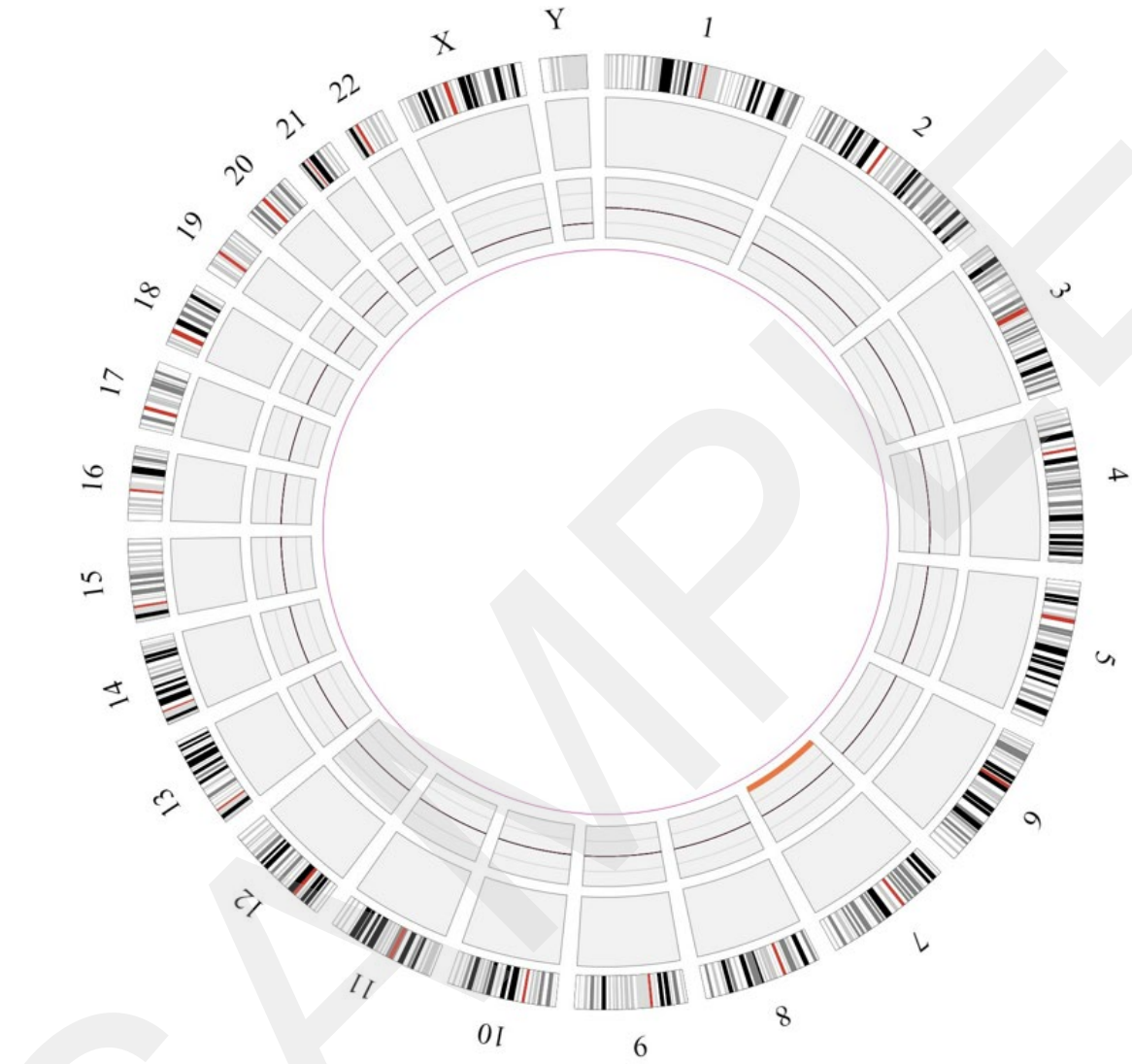
Section A: Tier 1A Guideline Driven Variant Analysis For Myelodysplastic Syndrome Results

Variant	Detected	Not Detected	Variant	Detected	Not Detected
3q (rearrangements)	☐	☒	Chr13 monosomy	☐	☒
Chr3 *MECOM* (rearrangements)	☐	☒	17q (duplication)	☐	☒
Chr5 monosomy	☐	☒	17p (deletion)	☐	☒
5q (deletion)	☐	☒	17p (translocation)	☐	☒
7q (deletion)	☐	☒	Chr17 monosomy	☐	☒
Chr7 whole chromosome (rearrangements)	☐	☒	Chr17 *TP53* (deletion)	☐	☒
Chr7 monosomy	☒	☐	Chr19 trisomy	☐	☒
Chr8 trisomy	☐	☒	20q (deletion)	☐	☒
9q (deletion)	☐	☒	Xp (duplication)	☐	☒
11q (deletion)	☐	☒	Xq (deletion)	☐	☒
12p (translocation)	☐	☒	ChrY monosomy	☐	☒
12p (deletion)	☐	☒			

Section B: Whole Genome Results – Tier 1 – Tier 3

Chromosome	Finding	Tier
Chr1	None	
Chr2	None	
Chr3	None	
Chr4	None	
Chr5	None	
Chr6	None	
Chr7	**ogm(7)x1~2**	**1A**
Chr8	None	
Chr9	None	
Chr10	None	
Chr11	None	
Chr12	None	
Chr13	None	
Chr14	None	
Chr15	None	
Chr16	None	
Chr17	None	
Chr18	None	
Chr19	None	
Chr20	None	
Chr21	None	
Chr22	None	
X	None	
Y	None	

Circos Plot



Interpretation

Optical Genome Mapping was performed on this specimen and showed **loss of chromosome 7 (monosomy 7)** in approximately 14% of the cells. Monosomy 7 is generally associated with a poor prognosis in myelodysplastic syndrome. **(PMID: 17074595)**

Clinical correlation is recommended.

Methodology, References and Limitations

The OGM-Dx HemeOne test is a laboratory developed test (LDT) performed using the optical genome mapping assay on the Saphyr® system at Bionano Laboratories (6777 Nancy Ridge Drive, San Diego, CA 92121). The OGM technology is based on specific labeling and mapping of ultra-high molecular weight DNA in nanochannel arrays and enables the detection of all classes of structural variations (SVs) at a very high resolution. The genome-wide structural variation analysis includes the detection of insertions, deletions, duplications, aneuploidies, balanced and unbalanced translocations, inversions, and large copy number changes. The data analysis is performed using the proprietary Bionano Access software (Ver 1.7.1), a graphical user interface tool for visualization, review and curation of findings. The subsequent interpretation and reporting of the SVs, both genome-wide and hematological malignancy subtype specific, is performed on samples meeting validated QC thresholds, by trained and competent CLS at Bionano Labs.

Genome Complexity Definition for Acute Lymphoblastic Leukemias:
A complex genome is defined as having ≥5 clonal aberrations each ≥5 Mb.
A simple genome is defined as having <5 clonal aberrations each ≥5 Mb.
Genome Complexity Definition for All Other Hematologic Malignancies:
A complex genome is defined as having ≥3 clonal aberrations each ≥5 Mb.
A simple genome is defined as having <3 clonal aberrations each ≥5 Mb.

Identified variants are classified per recommendations provided by ACMG (PMID: 35064925, 34237281, 34503197):

Tier 1	Tier 1A acquired variants that are included in the guidelines for a specific hematological disease Tier 1B acquired variants that are related to a specific hematological disease
Tier 2	Recurrent acquired variants observed in different neoplasms but not specific to a particular disease
Tier 3*	Acquired variants with no documented neoplastic association and all variants that do not meet the criteria for Tier 1 and Tier 2 and cannot be classified as constitutional benign and likely benign
Tier 4 (Not reported)	Constitutional benign and likely benign variants

*Tier 3 variants are not reported (exceptions allowed).

Performance: Analytical validation of the OGM-Dx HemeOne assay was performed at Bionano Laboratories and included a comprehensive assessment of performance characteristics. Overall accuracy, sensitivity, specificity, positive predictive value, and negative predictive value are >99%. Repeatability and reproducibility for the assay are 100% and 96%, respectively. Please note that OGM technology has different limits of resolution for different types of structural variants (>500kb for large segmental losses and gains; >70kb for translocations and inversions; 5-50kb for insertions; >7kb and >150kb for deletions and duplications, respectively).

Limitations: All laboratory tests have limitations. These results assume that the specimen received in the laboratory belongs to the named individual. Optical Genome Mapping cannot detect single-nucleotide variants and does not make any claims related to sequence variants or variants not meeting the thresholds of the test that may have potential functional impacts. This method also cannot detect balanced Robertsonian translocations, triploidy or regions with loss of heterozygosity (LOH). For triploidy and LOH determination, a request can be made for additional analysis. This method is also not validated to detect minimal residual disease (which suggests an incomplete cure or relapse of disease after treatment). Possible sources of testing error include rare and novel genetic variants that interfere with analysis, sample misidentification, and other sources. NOTE: The interpretation of OGM data is based on our current understanding of the genome. These interpretations may change over time as more information about hematological malignancies becomes available in the future. Genomic assessment is based on the human genome assembly version GRCh38.

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This test was developed and performed by Bionano Laboratories (6777 Nancy Ridge Drive, San Diego, CA 92121). CA License Number: CDF-90004882, CLIA ID: 05D2235036, CAP ID: 9336423
Pursuant to the requirements of CLIA ’88, this technical component and the professional component was performed by Bionano Laboratories. This test is used for clinical purposes. This test has not been cleared or approved by the US Food and Drug Administration (FDA).

Report reviewed by: XXXXXXXXXXXXXXXXXX
Report reviewed by: XXXXXXXXXXXXXXXXXX
Report Electronically Signed By: XXXXXXXXXXXXXXXXXX