Patient ID / MRN	Patient Name		Birth Date	Gender	Age
	LAST, FIRST		dd-mm-yyyy	Female	
Accession Number	Account Name		Ordering Physician		
	Clinic / Hospital		LAST, FIRST		
Sample Type	Collected	Received	Reported		
Bone marrow aspirate	dd-mm-yyyy	dd-mm-yyyy	dd-mm-yyyy		
Test		Test Indication			
1725332– Chromosomal Gains and Losses -NGS- Myeloma		Text			

Chromosomal Gains and Losses – NGS - Myeloma

RESULT

sseq (3,5,7,9,11,15,19)x3 Or sseq[GRCh37] 4p16.3p15.32(10000_15459989)x1

Shallow Whole Genome Sequencing (sWGS) was performed on DNA extracted from enriched plasma cells, and the analysis revealed the following findings:

- **Copy Number Gains:** [e.g., Gain of chromosome 1q], [e.g., Trisomy 11], or None.
- **Copy Number Losses:** [e.g., Deletion of 13q14], [e.g., Deletion of 17p (TP53 region)], or None.
- **Overall Genomic Imbalance Profile:** [e.g., Hyperdiploid profile with gains of odd-numbered chromosomes] and [e.g., High-risk profile with del(17p) and gain(1q)], or Stable.

INTERPRETATION

The copy number profile is consistent with [hyperdiploid / non-hyperdiploid / high-risk] multiple myeloma. The presence of [del(17p), gain(1q), or other high-risk lesions] is associated with adverse prognosis. The findings [are / are not] consistent with typical cytogenetic profiles observed in multiple myeloma. Results should be interpreted in conjunction with clinical presentation, histopathology, FISH, and molecular studies where applicable.

OR

sWGS reveals no copy number gains or losses in the plasma cells analyzed.

METHODOLOGY

Bone marrow samples were processed to isolate the plasma cells using the EasySep™ Human CD138 Positive Selection Kit II (STEMCELL Technologies). DNA was extracted and amplified using the Ion ReproSeq™ PGS kits. The whole genome amplified product was then barcoded, pooled, and subjected to automated template preparation using the Ion Chef™ System (Thermo Fisher Scientific). Chromosomal copy number analysis was performed on CD138+ sorted plasma cells using next-generation sequencing (NGS) on the Ion GeneStudio™ S5 System (Thermo Fisher Scientific). Sequencing data were analyzed using Ion Reporter™ Software, employing the ReproSeq Mosaic PGS w1.1 r.0 workflow (version 5.18.2.0) with alignment to the GRCh37/hg19 human genome reference build.

LIMITATIONS

This assay does not detect balanced chromosomal translocations, such as IGH rearrangements. Subclonal abnormalities present at very low levels may go undetected. Sequence-level mutations, including point mutations in genes like *TP53*, are not assessed by this method.

DISCLAIMER

This test identifies chromosomal copy number gains and losses across the genome using shallow whole genome sequencing (sWGS) of CD138-enriched plasma cells. sWGS is a cost-effective technique that sequences the genome at low coverage (~0.1x to 1x), enabling detection of large-scale chromosomal abnormalities (e.g., 1q gain, 13q deletion) commonly associated with multiple myeloma and other plasma cell neoplasms. It serves as a screening tool to aid in diagnosis and risk stratification and may guide further targeted testing.

Patient ID / MRN	Patient Name		Birth Date	Gender	Age
	LAST, FIRST		dd-mm-yyyy	Female	
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	Clinic / Hospital		LAST, FIRST		
Sample Type	Collected	Received	Reported		
Bone marrow aspirate	dd-mm-yyyy	dd-mm-yyyy	dd-mm-yyyy		
Test		Test Indication			
1725332– Chromosomal Gains and Losses -NGS- Myeloma		Text			

Interpretation is based on current scientific knowledge and the clinical information available at the time of reporting. Results should be considered in conjunction with clinical context and other laboratory findings. The test's sensitivity is influenced by tumor cell content and sample quality. A normal result does not exclude chromosomal abnormalities below the resolution limit of the assay.

This test was developed and validated by NRL Genomics Laboratory and is intended for clinical diagnostic use.

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