Patient ID / MRN	Patient Name		Birth Date	Gender	Age
Accession Number	Account Name		Ordering Physician		
Sample Type	Collected	Received	Reported		
Bone marrow aspirate					
Test		Test Indication			
1725334 – Comprehensive Chromosomal Profiling by sWGS, Blood & Bone Marrow		B- Lymphoproliferative neoplasm			

Comprehensive Chromosomal Profiling by sWGS, Blood & Bone Marrow

RESULT

Abnormal

ISCN result: sseq (xx)x3[0.3]

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

- **Copy Number Gains: Trisomy xx detected,** present in approximately **30% of the sample analyzed.**
- **Copy Number Losses:** None.
- **Overall Genomic Imbalance Profile:** Intermediate-risk

INTERPRETATION

The genomic profile demonstrates a trisomy xx as a sole abnormality, detected in 30% of the sample analyzed. Trisomy xx is a recurrent abnormality in CLL and is generally considered an intermediate-risk alteration. No evidence of a complex karyotype, hyperdiploid profile, 13q deletion, or high-risk imbalances such as del(17p) or del(11q) was observed.

Results should be interpreted in the context of the patient's clinical presentation, histopathological findings, flow cytometry results, and, where applicable, in correlation with other molecular studies.
FISH for xxx rearrangement have been set up and the report will follow.

METHODOLOGY

Bone marrow aspirate was processed and DNA extracted and amplified using the Ion ReproSeq™ PGS kit. 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 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). 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 blood neoplasms. It serves as a screening tool to aid in diagnosis and risk stratification and may guide further targeted testing.

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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.

REFERENCES

1. WHO Classification of Tumours of Haematopoietic and Lymphoid Tissues. 5th ed. International Agency for Research on Cancer; 2022.

REVIEWED BY

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