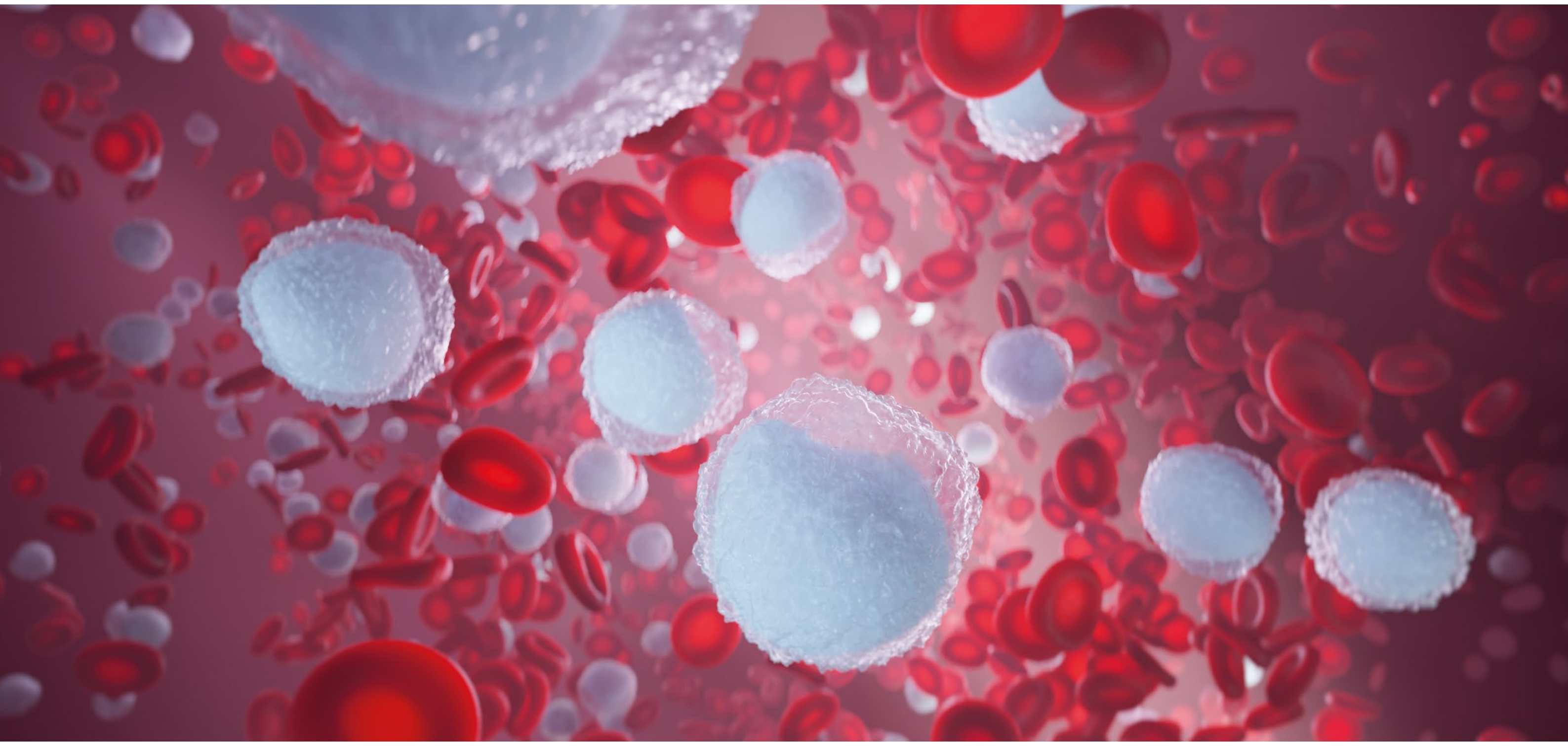
Together for a better healthcare journey

**PUBLICATION SPOTLIGHT | July 2025**

## Streamlined Molecular Profiling for Haematological Cancer Research

# SureSeq™ NGS panels



Molecular profiling is critical for haematological malignancies as it allows for the precise identification of genetic mutations and chromosomal abnormalities that play a pivotal role in disease pathogenesis and progression. By detecting genomic alterations, researchers can better understand the biological mechanisms underlying the disease, enabling more accurate characterisation and stratification of samples based on genetic risk factors.

This information is essential for researching prognostic factors, as certain mutations or chromosomal aberrations are associated with more aggressive forms of the disease or a higher likelihood of relapse.

Additionally, molecular profiling informs treatment decisions by identifying targetable mutations or cell signalling pathways that can be addressed with specific therapies – tyrosine kinase inhibitors, monoclonal antibodies, or emerging immunotherapies. Effective application of precision medicine increases the likelihood of positive treatment responses and minimises unnecessary exposure to less effective or toxic treatments.

**The unique designs of SureSeq™ panels offer unparalleled coverage completeness and uniformity, allowing for the accurate detection of low-frequency SNVs, indels and structural aberrations such as ITDs, PTDs, CNVs, LOH and translocations.** Combined with our SureSeq Universal NGS Workflow Solution V2, SureSeq alleviates the burden of running multiple assays and streamlines research, delivering comprehensive results using a single NGS workflow.

## References


**[1]** **Wojtaszewska M *et al.* (2024):** Precise and Accurate Detection of Mutations and Copy Number Alterations in CLL Using Enrichment-Based Targeted NGS. The Journal of Molecular Diagnostics, Volume 26, Issue 11, S46.

**Key message:** The SureSeq CLL + CNV V3 Panel is an effective method for detecting both small variants and copy number alterations in clinical research CLL samples. It provides higher resolution than other methods, allowing for the reliable identification of CNAs smaller than 1Mbp, which are often missed by FISH.

**[2]** **Poloni G *et al.* (2023):** Using a Targeted NGS Approach to Detect Copy Number Alterations and Gene Variants in Chronic Lymphocytic Leukemia. The Journal of Molecular Diagnostics, Volume 25, Issue 11, S35.

**Key message:** The SureSeq CLL + CNV V3 Panel effectively detects common CNAs, SNVs and indels in CLL research samples, even at a tumour content as low as 20%. The panel offers high concordance with FISH results and enables the simultaneous detection of multiple aberrations, providing a cost-effective and comprehensive genomic profiling solution for CLL research.

**[3]** **Semenuk L *et al.* (2024):** Integrated Analysis of Diverse Genomic Variants for the Profiling of Myeloid and Lymphoid Neoplasms by Targeted Hybrid Capture Next-Generation Sequencing. The Journal of Molecular Diagnostics, Volume 2, Issue 11, S44.

**Key message:** NGS can effectively detect a wide range of genomic variants, including SNVs, MNVs and CNVs, across various myeloid and lymphoid neoplasms, offering diagnostic, prognostic and predictive insights, providing a comprehensive molecular analysis in one test. The study demonstrates high sensitivity and specificity for variant detection, with the potential to improve efficiency, reduce costs and streamline reporting in clinical settings by replacing the need for multiple assays.

**[4]** **Akabari R *et al.* (2022):** Technological Advances: CEBPA and FLT3 Internal Tandem Duplication Mutations Can be Reliably Detected by Next Generation Sequencing. Genes (Basel). 1;13(4):630.

**Key message:** SureSeq offers a reliable solution for detecting clinically important mutations in myeloid malignancies, such as CEBPA and FLT3, with enhanced coverage uniformity and accuracy compared to other NGS panels. SureSeq achieves more homogeneous coverage of critical regions, particularly in genes with difficult-to-sequence areas, such as CEBPA. Its hybrid capture-based approach minimises dropouts and ensures more reliable mutation detection, addressing limitations seen with amplicon-based assays.


## Glossary

**SNVs:** Single Nucleotide Variants; **MNVs:** Multiple Nucleotide Variants; **Indels:** Insertions and Deletions; **ITDs:** Internal Tandem Duplications; **PTDs:** Partial Tandem Duplications; **CNVs:** Copy Number Variations; **CNA:** Copy Number Alteration; **LOH:** Loss of Heterozygosity; **CLL:** Chronic Lymphocytic Leukaemia; **NGS:** Next-Generation Sequencing

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