CinCSeq Heme Cancer Panel

The CinCSeq Heme Cancer Panel is designed to interrogate for single nucleotide variants (SNV), multi-nucleotide variants (MNV), insertion and deletion (Indel) variants, and copy number variants (CNV) across 73 hematologic cancer-related genes (Table 1) from genomic DNA. The DNA portion covers approximately 95% of coding exons of sequenced genes. The RNA portion is designed to identify gene fusions and select structural variants (SV) involving 47 hematopoietic cancer-related genes (Table 2). It can identify known fusion partners as well as ~98% of novel fusion partners of driver oncogenes.

Sequencing is performed using the GOAL consortium (https://www.goalabs.org/) hybrid capture probe set with custom modifications for pediatric and young adult cancer on high-throughput Illumina sequencing platforms. Analytic sensitivity is 3-5% variant allele fraction (VAF) with at least 300x sequencing depth for detection of SNV, MNV, and small Indels (<10bp) and 5-10% VAF with a minimum of 200x sequencing depth for larger Indels (>10bp) and SV, including internal tandem duplication (ITD) variants. Genomic regions with lower coverage are specified in the report. A minimum of 5 unique spanning reads supporting a gene fusion call is required. Orthogonal confirmation for one or more genomic alterations may be performed, if appropriate. For more information, call the lab at 513-636-9820.

Reporting Units:

CinCSeq is a qualitative NGS assay that reports clinically actionable and potentially clinically actionable sequence variants with interpretation text. Variants of uncertain significance (VUS) are listed in the report without any associated interpretation text. Benign or likely benign variants are not included in the report.

Acceptable Specimens:

- 3-5 mL whole blood in EDTA tube
- 3-5 mL bone marrow in EDTA tube
- FFPE tissue: 1 H&E slide and 6 unstained slides at 5 microns each; fix in 10% neutral buffered formalin
- Paraffin block (block will be returned upon completion of test)
- A minimum of 20% viable tumor cells is required for DNA mutation and gene fusion identification. For reliable copy number variation (CNV) detection, at least 50% viable tumor cells are required.

Unacceptable Specimens:

- Decalcified tissue
- Frozen whole blood or bone marrow
- Sample collected in heparin

Shipping Conditions:

- Blood and bone marrow samples collected within the institution must be delivered to the lab within 6 hours of collection. Do NOT freeze.
- Blood and bone marrow samples collected outside the institution must be shipped at 4°C in an insulated container by **overnight** courier. Do NOT freeze.
- Tissue, slides, or blocks at ambient temperature.
- **For all samples, ship for delivery on Monday through Friday. Receiving docks are CLOSED on weekends and holidays; do NOT ship if delivery will occur on those days.**

Testing Schedule:

CinCSeq testing is performed Mon-Fri. For testing outside of this schedule, call the lab at 513-636-9820.
TAT: 7-10 business days (maximum 14 business days)

CPT Codes:

88363 – Tissue review & selection
81455 – Targeted genome analysis
G0452 – Physician interpretation

Clinical Lab Index:

CinCSeq Heme Panel:
https://www.testmenu.com/cincinnatichildrens/Tests/1169863

Contact Information:

Cincinnati Children's Division of Pathology
Molecular and Genomic Pathology Services
Phone: 513-636-9820
Fax: 513-517-7099
Email: pathology@cchmc.org
Website: cincinnatichildrens.org/pathology

For pricing or billing questions, call 513-636-4261.

Shipping Address:

Cincinnati Children's Hospital Medical Center
Attn: Molecular and Genomic Pathology Services (MGPS)
240 Albert Sabin Way, R2.001
Cincinnati, OH 45229

References:
1. Cancer Genome Atlas Research Network. *Cell*. 2017; 171:950-965.e28.
2. Li M, et al. *J Mol Diagn.* 2017; 19:4-23.

Table 1: DNA gene list (SNV, MNV, Indels, CNV - 73 genes)

^ - Only Internal Tandem Duplication (ITD) interrogated

ABL1	ABL2	ASXL1	ATM	BCL6	BCOR^	BRAF	BTK	CALR	CBL	CDKN2A	CDKN2B	CEBPA
CRLF2	CSF1R	CSF3R	DDX41	DNMT3A	ERG	ETV6	EZH2	FLT3^	GATA1	GATA2	GATA3	HRAS
IDH1	IDH2	IKZF1	IKZF2	IKZF3	IL7R	JAK1	JAK2	JAK3	KIT	KMT2A	KRAS	MAP2K1
MPL	MSH2	MSH6	MYC	MYD88	NF1	NPM1	NRAS	PDGFRA	PDGFRB	PHF6	PIK3R1	PPM1D
PRPF8	PTEN	PTPN11	RAD21	RB1	RUNX1	SETBP1	SETD2	SF3B1	SH2B3	SMC1A	SMC3	SRSF2
STAG2	STAT3	TCF3	TET2	TP53	U2AF1	WT1	ZRSR					

Table 2: RNA gene list (47 genes)

ABL1	ABL2	ALK	BCL2	BCL6	BRAF	CBFB	CCND1	CREBBP	CRLF2	CSF1R	DEK	EGFR
EPOR	ERG	ETV6	FGFR1	FGFR2	FGFR3	FUS	GLIS2	JAK2	KAT6A	KMT2A	MECOM	MET
MKL1	MLLT10	MLLT4	MYBL1	MYC	MYH11	NTRK3	NUP214	NUP98	PDGFRA	PDGFRB	PML	RARA
RUNX1	RUNX1T1	TCF3	TFE3	TYK2	IGH	DUX4	CIC					