imegen
a part of healthincode

Haematology
OncoKitDX

Study of syndromes associated with hematologic neoplasms.

For diagnostic use.
Format: 24rx / Technology: NGS / Reference: IMG-363

> Characteristics

- Panel for the study of the different syndromes associated with hematologic neoplasms, such as acute myeloid leukemia (AML), chronic myeloid leukemia (CML), myeloproliferative neoplasms (MPN), and acute lymphoid leukemia (ALL).
- It allows detecting SNVs, indels, genetic fusions, CNVs, and pharmacogenetic variants from a single DNA sample.
- Bioinformatics analysis using the software Datagenomics.
- Automated generation of a report including functional and clinical classification of the variants.
- STIDs can be included: Integrated system for the identification of samples for their traceability.
- Based on high sensitivity RNA-probe technology with **UMI** barcoding (unique molecular identifiers).
- Mean coverage: 1400X Mean coverage after UMI analysis: 1000X. Coverage: 99.3% of the bases covered at a depth of 100X.
- Uniformity: 98.4% of bases covered at >20% mean coverage.
- Specificity: >99%. Sensitivity: >99%. Repeatability: >99.99%. Reproducibility: >99.9%

> Specifications

- **Compatible sequencers:** Illumina NextSeq
- **Number of reactions:** 24.
- **Number of samples per run:** 8 samples in a cartridge MID 150 cycles on NextSeq.
- **Sequencing:** Paired-end (2 x 75 cycles).
- **Type of sample:** DNA from peripheral blood and bone marrow.
- **Amount of input DNA:** 50-100 ng.
- **Limit of detection:** SNVs and Indels: 2%, CNVs: 20%, and copy losses or gains: 10%
- Fully automated panel for Magnis NGS Prep System Dx equipment.
- CE-IVD and analysis software labeling.

imegen
a part of healthincode

For more information, please visit our website

> Composition of the Haematology OncoKitDx

- **Sequencing of full exonic region for 76 genes:**

ARID5B	*CDKN2B*	*ETNK1*	*IKZF1*	*NFE2*	*PTK2B*	*SRSF2*
ASXL1	*CEBPA*	*ETV6*	*IL7R*	*NOTCH1*	*PTPN11*	*STAG1*
ASXL2	*CHIC2*	*EZH2*	*JAK1*	*NPM1*	*RAD21*	*STAG2*
ATRX	*CREBBP*	*FBXW7*	*JAK2*	*NR3C1*	*RB1*	*STAT5B*
BCOR	*CSF3R*	*FLT3*	*JAK3*	*NRAS*	*RUNX1*	*TET2*
BCORL1	*CSNK1A1*	*GATA1*	*KIT*	*P2RY8*	*SETBP1*	*TP53*
BLNK	*CUX1*	*GATA2 (+I4)*	*KMT2A*	*PAX5*	*SF3B1*	*TYK2*
BRAF	*DDX3X*	*GATA3*	*KMT2C*	*PHF6*	*SH2B3*	*U2AF1*
CALR	*DDX41*	*HAVCR2*	*KRAS*	*PIGA*	*SMC1A*	*WT1*
CBL	*DNMT3A*	*IDH1*	*MPL*	*PPM1D*	*SMC3*	*ZRSR2*
CDKN2A	*EP300*	*IDH2*	*NF1*	*PTEN*	*SRP72*	

- **Capture of 27 fusion genes in any of their potential rearrangements:**
  (inclusion of intronic regions described as breakage points in the literature)

ABL1	*CBFB*	*FGFR1*	*MEF2D*	*NUP214*	*RARA*	*STIL*
ABL2	*CSF1R*	*FUS*	*MNX1*	*NUP98*	*RBM15*	*TAL1*
BCR	*EPOR*	*JAK2*	*MYH11*	*PDGFRA*	*RUNX1*	*TCF3*
CBFA2T3	*ETV6*	*KMT2A*	*NPM1*	*PDGFRB*	*SET*	

- **Detection of CNVs** throughout the whole genome (detection of hypo- and hyperploids) or in a whole gene included in the panel. Validation of results and detection of copy-neutral LOH through a low-density SNP array throughout the genome.

- **Detection of pharmacogenetics-related variants:**

ABCB1	*CYP2C9*	*MTHFR*	*NUTD15*	*SLCO1B1*
CEP72	*ITPA*	*MTRR*	*PNPLA3*	*TPMT*

> Added value

- Technical support from first protocol implementation, both online and on-site. All protocols will be fine-tuned in collaboration with laboratory personnel.
- Clinical support or genetic counseling, with clinical geneticists available to answer any clinical questions.
- Possibility of externalizing 10% of the samples to our laboratory at the same price during periods of excess laboratory workload or urgent deadlines.

Contact our oncology team for clinical counseling on this product:
oncologia@healthincode.com | (+34) 963 212 340