SISTEMAS GENÓMICOS
A MEMBER OF SYNLAB

Myeloid Line Disorders

MLD-GeneSGKit®

CE-IVD

Sistemas Genómicos has developed **Myeloid Line Disorders (MLD)-GeneSGKit®** to provide reliable and efficient analysis of point mutations and CNVs associated with the **diagnosis, prognosis, follow-up and treatment of myeloid lineage blood cancers.**

MLD-GeneSGKit® has CE-IVD marking and contains reagents to **process 12 samples** with the relevant bioinformatics analysis using the GeneSystems© platform. Developed in collaboration with experts in this type of disease, MLD-GeneSGKit® is designed for the study of 58 genes.

The work process is streamlined to create libraries in a maximum of 12 hours and the technology used allows the multiplexing of up to 6 samples prior to hybridisation and capture of the relevant probes, greatly facilitating the laboratory protocol in the wet phase. It also provides for the combination of different pools from other GeneSGKits® in a single run on Illumina® sequencing platforms, ensuring an equitable distribution of data between the different samples.

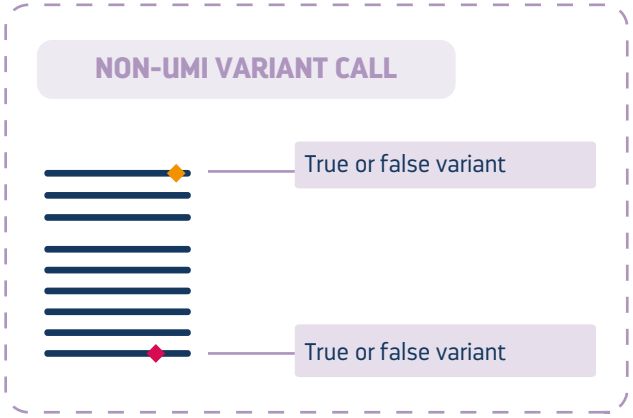
HIGHLIGHTS

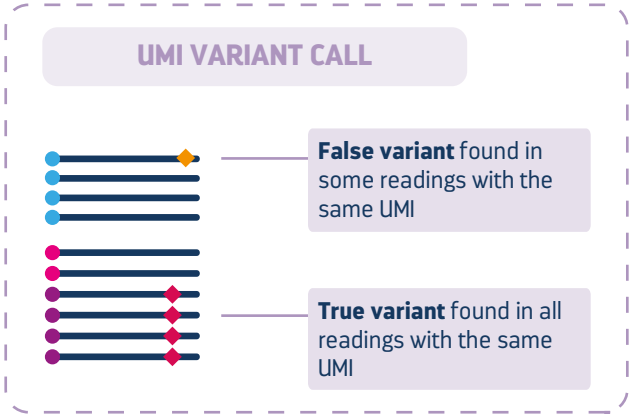
A comprehensive solution, the only one of its kind on the market:
Complete bioinformatics analysis and display of results:
The MLD-GeneSGKit® contains reagents to process 12 samples with the corresponding bioinformatic analysis through the GeneSystems©. The kit, the bioinformatic computation and the display have CE-IVD marking.

Complete bioinformatics analysis and display of results:
The data is computed in less than 48 hrs and recorded against the main open and private databases reporting the prioritised variants according to different algorithms which are easily visualised, filtered and reported through GeneSystems©.

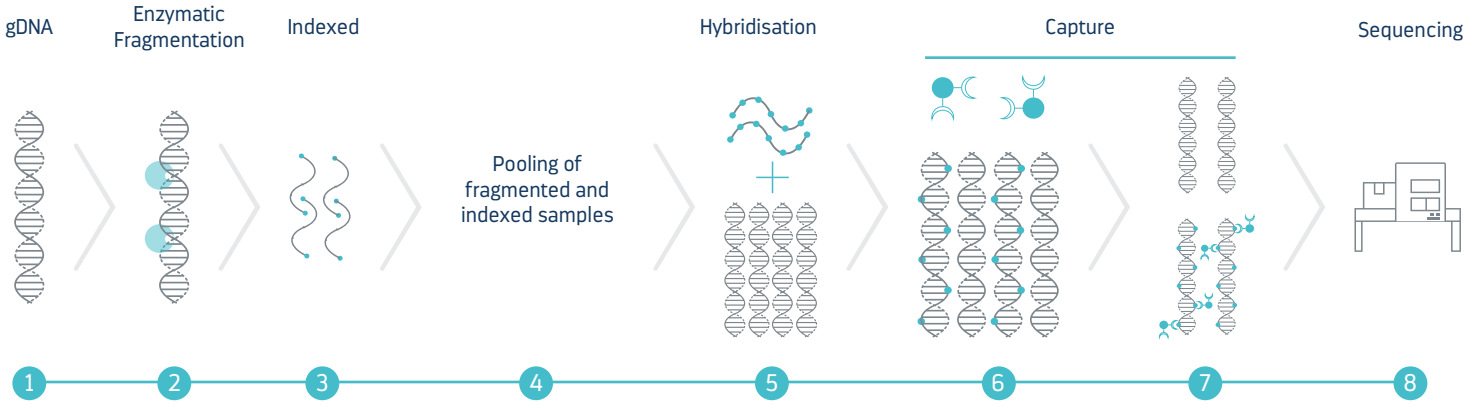
Speed:
The libraries are ready after just 12 hours of work from 100 ng of DNA.

High sensitivity:
It incorporates the innovative individual molecule tagging technology through **UMIs**, ensuring a thorough bioinformatic processing of each reading. The optimisation of this process provides an **average coverage of 3000x**, reaching a **diagnostic limit of variants of up to 0.5% of tumour percentage.** The panel's extreme sensitivity makes it an ideal solution for the **diagnosis and monitoring** of both acute and chronic blood cancer or during the treatment phase.





GeneSGKit® WORKFLOW



ABOUT US

Sistemas Genómicos is a pioneering company in the use of next-generation sequencing for genetic diagnosis. With more than 6,000 analyses per year, we are leaders in the sector, supported by years of experience and accredited and certified by the National Accreditation Body of Spain (ENAC) and the Spanish Association for Standardisation and Certification (AENOR).

OUR OBJECTIVE

To capitalise on all our experience in analysis and diagnosis by using NGS to develop a series of tools that enable genetic diagnosis in situ in hospitals and research centres. We have achieved this by developing a comprehensive solution, the only one of its kind on the market, based on more than 35 GeneSGKits® for various diseases, all with CE-IVD marking and the GeneSystems©.

MLD-GeneSGKit® provides a high performance from only 100 ng of DNA. The design of the capture probes and bioinformatic pipeline guarantees high sensitivity and specificity for detecting point mutations and CNVs with a recommended average coverage of 3000x.

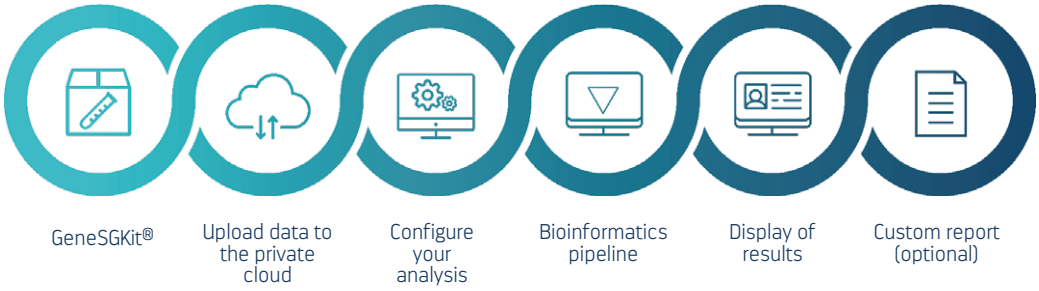
Sistemas Genómicos understands the vital importance of quality customer service and follow-up. This includes a personalised service that offers not only support over the telephone for all sorts of questions and concerns that may arise while using GeneSGKits® or the GeneSystems© analysis platforms, but also personalised training for the user who purchases the product. These training sessions will facilitate the set-up and roll-out of a genetic diagnostic service in situ at hospitals and laboratories using GeneSGKits®.

GeneSGKit®

GeneSystems©
SISTEMAS GENÓMICOS

GeneSystems© is our bioinformatics tool for genetic diagnosis and research. It is a platform registered as a medical device with CE marking for diagnostic use *in vitro*, to process information from massive DNA sequencing, detect variants and help genetic diagnosis. Developed in a Cloud Computing environment, it can provide access your data from any computer device, at any time and place, without consuming the user's own resources.

GeneSystems© is designed to provide a solution to the different stages of the NGS data analysis step:

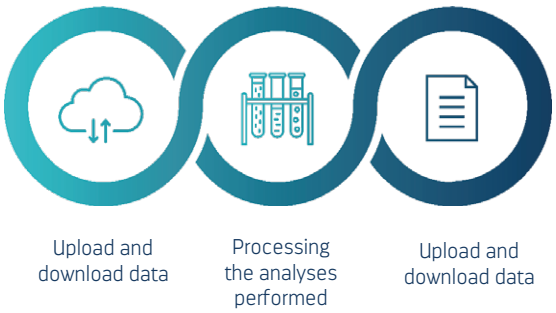


FEATURES OF GENESYSTEMS©

Integral infrastructure **bioinformatic analysis** for the data analysis from NGS platforms.

Automation of the entire data analysis process.

Cloud-Computing platform: access from any device with an internet connection.



It was developed by a multidisciplinary team of geneticists, bioinformaticians and engineers, and has been validated by over 200 companies, including hospitals, laboratories, research centres, universities and pharmaceutical companies. The platform has been independently audited and detects **SNPs, INDELs, large INDELs, MNVs, ALUs, CNVs and SVs.**

ANALYSIS OF SEQUENCING DATA WITH GENEBYTES

In the laboratory routine

- Cloud-based platform: no software installation required.
- Upload and download results quickly and easily.
- Bioinformatic analysis of results from raw data, BAM and VCF.
- Exhaustive report of the computation process.
- Resource optimisation: support for preparing pools of non-equimolar libraries prior to sequencing.
- View the results of computations in the corresponding application.

In research

- Gene, exome and genome panels: view and interact with the results obtained through NGS.
- Annotation of variants compared to the major public and private databases.
- Flexible filtering.
- Simultaneous analysis of samples.

In diagnosis

- Flexible viewing and prioritisation of the variants obtained.
- Identification of point variants, Indels, large Indels and structural variants (balanced and unbalanced).
- Simultaneous analysis of samples: optimised diagnostic analysis time.
- Full annotation for variant classification:
  - DBNLVar (Sistemas Genómicos' own database).
  - Public and private reference databases.
  - In silico predictors.
- Integrated IGV for a graphic analysis of the genomic environment.
- Customisable reports of genetic results.
- Generation of virtual panels to prioritise variants according to HPO terminology.
- Automatic classification of variants based on ACMG guidelines.
- Handles the major databases in the sector (DBNLVar, Ensembl, 1000Genomes, HGMD-Pro, ClinVar, etc.)
  - Pathogenicity indicators with links to scientific articles.

CATALOGUE - MLD-GeneSGKit®

Technical data

GeneSGKit®	Size (Mb)
MLD-GeneSGKit®	0.7

References
MLD-GeneSGKit® X (NextSeq) 12 reactions Ref.: LV4485 (CE-IVD)

MLD-GeneSGKit®
58 Genes

*ANKRD26**	*CSF3R**	*GATA2**	*MYC**	*RAD21*	*SRP72*
*APC**	*CYP11B2**	*GRIN2B**	*NBN**	*RUNX1*	*SRSF2**
*ASXL1**	*CYP3A5**	*HNF4A*	*NF1*	*SAMD9*	*STAG2*
*ATOX1**	*DDX41*	*IDH1**	*NIPBL**	*SAMD9L*	*TCOF1**
BCOR	*DNMT3A**	*IDH2**	*NPM1**	*SETBP1**	*TET2*
*BCORL1**	*EGR1**	*IKZF1**	*NRAS**	*SF3B1**	*TP53**
CALR#*	*ETNK1*	*JAK2**	*NRG1**	*SH2B3*	*WT1**
*CAV1**	*ETV6*	*KIT**	*PIGA**	*SMC1A*	*ZRSR2*
*CBL**	*EZH2**	*KRAS**	*PRDM14**	*SMC3*	
*CEBPA**	*FLT3*#*	*MPL**	*PTPN11*	*SRC**	

***** Genes that exclusively include specific regions where clinically relevant mutations have been reported.
This panel has been validated for the detection of indels in exon 14-15 of the FLT3 gene (whose tandem deletions are present in 20-27% of acute myeloid leukaemias) and in exon 9 of CALR.