SISTEMAS GENÓMICOS
ΔSCIRES

MYELOIDNEOPLASM

MyeloidNeoplasm-GeneSGKit®

Sistemas Genómicos has developed MyeloidNeoplasm-GeneSGKit® which enables the **study of abnormalities associated with the diagnosis and treatment of various onco-haematological diseases**. MyeloidNeoplasm-GeneSGKit® is optimised to reliably and efficiently analyse point mutations, CNVs and rearrangements associated with myelodysplastic syndromes, chronic myelomonocytic leukaemia, Ph-negative myeloproliferative neoplasms, myeloproliferative neoplasms, gliomas, etc.

MyeloidNeoplasm-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, MyeloidNeoplasm-GeneSGKit® is designed to study **37 genes, 13 translocations, 7 deletions, 2 aneusomies and a deletion/amplification in chromosome 11, associated with various onco-haematological abnormalities.**

The work process is optimised to complete the libraries in 9 hours and the technology used makes it possible to combine multiple samples and several GeneSGKits® in the same run using Illumina® sequencing platforms, thereby ensuring an equal distribution of data between the different samples.

HIGHLIGHTS

A comprehensive solution that is the only one of its kind on the market:
MyeloidNeoplasm-GeneSGKit® has CE-IVD marking and contains reagents to process 12 samples with the relevant bioinformatics analysis and visualisation using the GeneSystems© platform, the only one with CE marking for IVD.

Speed:
The libraries are ready after just 9 hours of work from 50 ng of DNA.

Complete bioinformatics analysis and visualisation of results:
Data are computed in less than 48 h, and recorded against the main open and private databases reporting SNPs and INDELs, which are visualised and easily filtered using GeneSystems©, a platform registered as a medical device with CE marking for diagnostic use *in vitro*.

High sensitivity:
Limit of detection of up to 10% of the tumour percentage.

1

Library preparation with GeneSGKits®

2

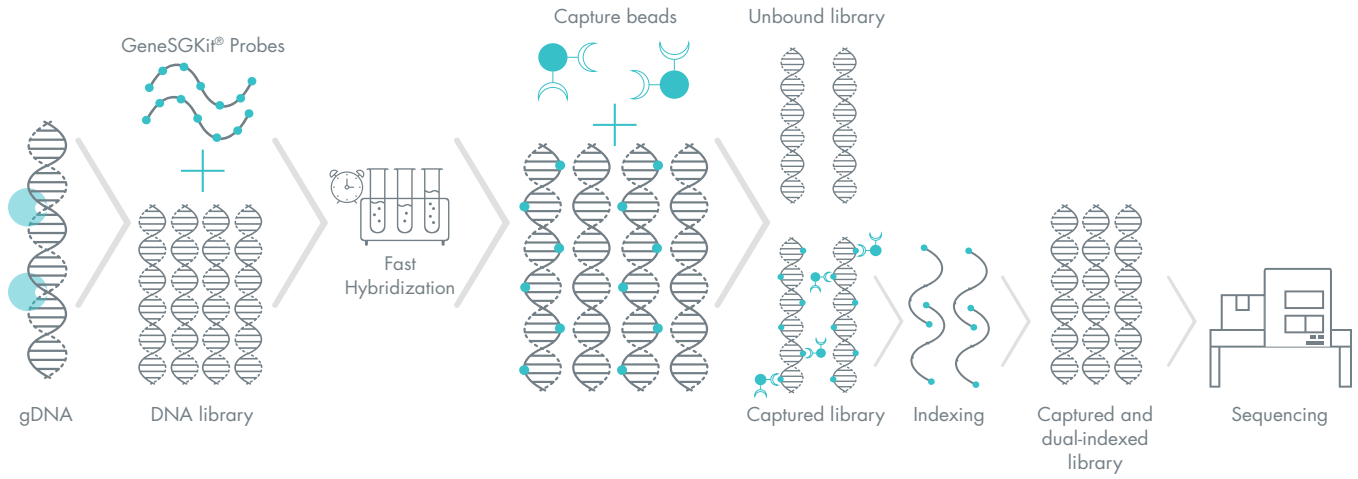
Sequencing using Illumina®* platforms

3

Analysis and interpretation of data using GeneSystems©

*HiSeq and MiSeq. Other Illumina® platforms currently being validated

WORKFLOW de GeneSGKit®



ABOUT US

Sistemas Genómicos is a pioneering company in the use of next-generation sequencing (NGS) 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), the Spanish Association for Standardisation and Certification (AENOR) and the Clinical Laboratory Improvement Amendments (CLIA).

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 27 GeneSGKits® for various diseases, all with CE-IVD marking and the GeneSystems© bioinformatics computation and results analysis platform.

DESIGN

MyeloidNeoplasm-GeneSGKit® offers a high yield from just 50 ng of DNA. The capture probes are based on Ultra-Long RNA Baits (120 nucleotides), and their design ensures high sensitivity and specificity in the detection of point mutations, CNVs and rearrangements, including translocations, major deletions and other structural abnormalities, with a recommended average coverage of 300x.

SUPPORT

Sistemas Genómicos understands the essential 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, specifically to process information from massive DNA sequencing, detect genetic variants and help to diagnose heterogeneous genetic diseases on a molecular level through targeted sequencing. Having been developed in a cloud environment, it features a number of technological services, including:

1 

Uploading and downloading of data

2 

Processing of analyses performed

3 

Visualisation and prioritisation of results

It was developed by a multidisciplinary team of geneticists, bioinformaticians and engineers, and has been validated by more than 200 companies, including hospitals, laboratories, research centres, universities and pharmaceutical companies. The platform has been independently audited and detects SNPs and INDELs.

ANALYSIS OF SEQUENCING DATA

GeneSystems© allows access to your data from any electronic device, anytime, anywhere, through a dedicated website with a username and password, thanks to having been developed in a cloud-computing environment. The platform is designed to resolve the different steps of the data analysis cycle of next-generation sequencing, from the uploading of the files to the final visualisation of the results obtained.

GeneSystems© incorporates the most important databases for interpreting variants, selected for their data quality and importance in clinical practice, such as Ensemble, 1000Genomes, HGMD, dbSNP, RefSeq, Interpro, Prosite, Cosmic, HapMap, OMIM, ClinVar and ExAC. In addition, GeneSystems© users have access to DBNLVar, Sistemas Genómicos's own variant database, with information on how variants have been reported, historical variants, validations, etc. The platform also features information on conservation indicators, pathogenicity predictors, variant flanking regions, etc.

Along with the results for visualisation, the user receives a report on sequencing quality, useful readings and regions not covered at 20x, as well as a report on CNV detection thanks to pipelines developed at Sistemas Genómicos that drastically reduce MLPA experiments, thereby reducing costs and wait times.

High quality of data analysed

Thanks to GeneSGKit® technology, clinics and researchers will have the best tools on the market for genetic diagnosis and analysis, thanks to the extremely high quality of the sequencing data for samples prepared using the GeneSGKit®.

CATÁLOGUE MyeloidNeoplasm-GeneSGKit®

Technical data

GeneSGKit®	Size (MB)
MyeloidNeoplasm-GeneSGKit®	0.49

References

MyeloidNeoplasm-GeneSGKit® ref.: LV3832 (CE-IVD Kit)/LV3831 (RUO Kit)

MyeloidNeoplasm-GeneSGKit®

Genes

NPM1	*KIT*	*KMT2A-PTD*	*CALR*	*PTPN11*	*ANKRD26*
FLT3	*TET2*	*NRAS*	*CSF3R*	*ABL1*	*ETV6*
DNMT3A	*RUNX1*	*KRAS*	*SETBP1*	*ETNK1*	*GATA2*
IDH1	*ASXL1*	*SRSF2*	*ZRSR2*	*CBL*	*VHL*
IDH2	*TP53*	*JAK2*	*SF3B1*	*NF1*	*EPOR*
CEBPa	*WT1*	*MPL*	*EZH2*	*DDX41*	*IKZF1*
HNF4A					

Translocations

t (9;22)(q34;q11.2)	*t (16;16)(p13.1;q22)//inv(16)(p13;q22)*	*t (6;9)(p23;q34)*
t (5;var)(q31-q33,var)	*t (16;16)(p13.1;q22)//inv(16)(p13;q22)*	*t (3;3)(q21;q26.2)//inv(3)(q21;q26.2)*
t (8;var) (p11,var)	*t (15;17)(q22;q12)*	*t (1;22)(p13;q13)*
t (8;21)(q22;q22)	*t (17;var) (q12)*	*t (8;9)(p22;p24)*
t (11;var)(q23;var)		

Deletions

del4q12 (CHIC2)	*del(12p)*
del(5q)	*del 17p13.1 (TP53)*
del(7q)	*del(20q)*
del(11q)/ amplifications	

Aneusomies

+8	*+19*

Other structural abnormalities

i(17q)

atencion-cliente@sistemasgenomicos.com
www.sistemasgenomicos.com

Parque Tecnológico de Valencia - Ronda G. Marconi, 6
46980 Paterna, Valencia (España)

Tel. +34 961 366 150
Fax +34 961 366 151