SISTEMAS GENÓMICOS
A MEMBER OF SYNLAB

Somatic Oncogenetics

MainTumorSolution - GeneSGKit® CE-IVD

Sistemas Genómicos has developed **MainTumorSolution-GeneSGKit®** to provide the reliable and efficient analysis of point mutations, Indels, large Indels, ALU insertions and CNVs, as well as analysis of fusions in the *ALK*, *RET* and *ROS1* genes associated with the diagnosis of somatic mutations in 38 genes related to sporadic cancer from paraffin-embedded solid tumor samples.

MainTumorSolution-GeneSGKit® has CE-IVD marking and contains reagents **to process 12 samples** with their corresponding bioinformatic analysis through the GeneSystems© platform.

The work process is streamlined to create libraries in a maximum of 2 working days. 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

Integrated solution unique in the market:
Complete bioinformatics analysis and display of results: **MainTumorSolution-GeneSGKit®** contains reagents to process 12 samples with their corresponding bioinformatic analysis, visualization and prioritization of variants through the GeneSystems© platform. The kit, the bioinformatic computation and the visualization have CE-IVD marking.

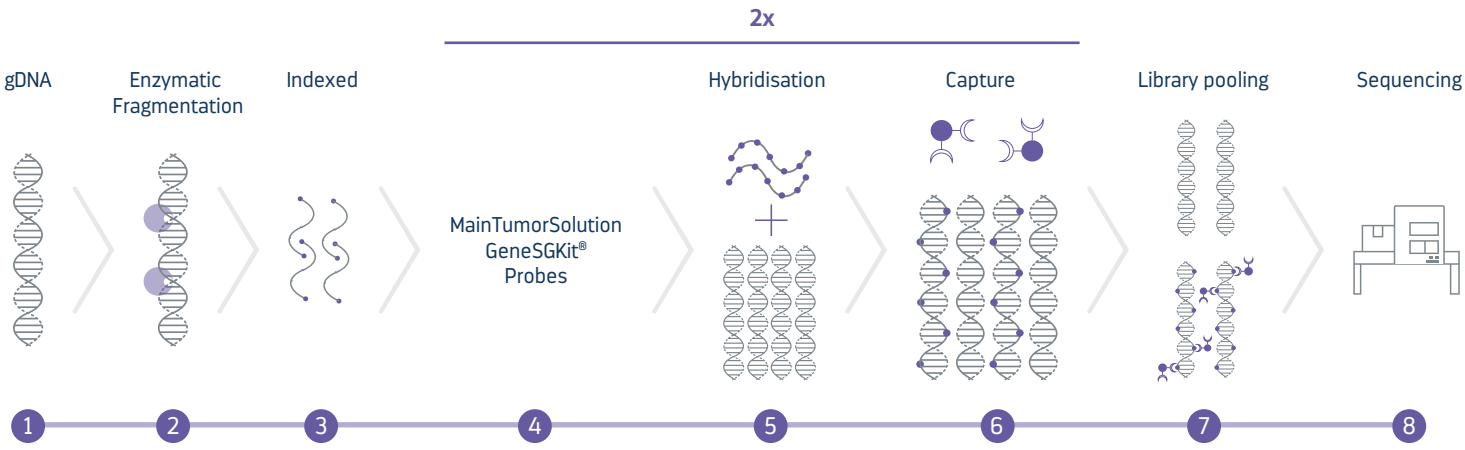
Complete bioinformatics analysis and display of results:
The data is computed in less than 48 hrs and recorded against the main open and private databases reporting the prioritised variants according to different algorithms which are easily visualised, filtered and reported through GeneSystems©.

Speed:
The libraries are ready after just 2 days of work from 10-200 ng of DNA.

High sensitivity:
Average coverage of 300x, ensuring the reliability of the data obtained without the need for alternative validations. Limit of detection up to 5% of the tumor percentage.

Complete Diagnostic Solution:
Analysis of point mutations, Indels, large Indels, MNVs, CNVs and ALU insertions, as well as analysis of fusions in the *ALK*, *RET* and *ROS1* genes.

MainTumorSolution 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©.

MainTumorSolution-GeneSGKit® provides high performance from only 10-200 ng of DNA. The design of the capture probes and bioinformatic **pipeline** guarantees high sensitivity and specificity for point mutation, Indels, large Indels, MNVs, ALUs and CNV screening detection with a recommended average coverage of 900x.

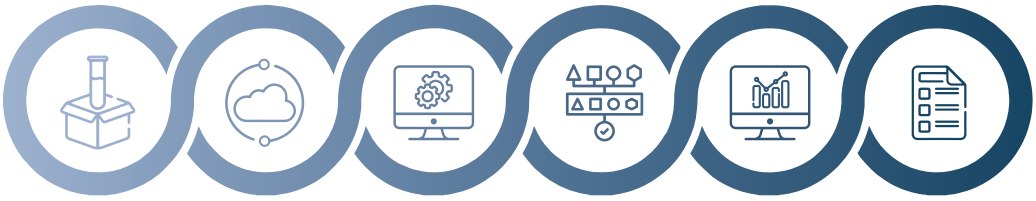
Sistemas Genómicos understands the vital importance of quality customer service and follow-up. This includes a personalised service that offers not only support over the telephone for all sorts of questions and concerns that may arise while using GeneSGKits® or the GeneSystems© analysis platforms, but 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:



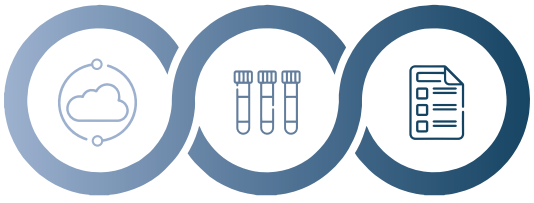
GeneSGKit® | Upload data to the private cloud | Configure your analysis | Bioinformatics pipeline | Display of results | Custom report (optional)

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.



Upload and download data | Processing the analyses performed | Upload and download data

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**.

APPLICATIONS OF GENESYSTEMS©

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 - MainTumorSolution-GeneSGKit®

Technical data

GeneSGKit®	Size (MB)
MainTumorSolution-GeneSGKit®	0,28

References
MainTumorSolution-GeneSGKit® 12 reactions Ref.: LV4582 (CE-IVD)

MainTumorSolution-GeneSGKit®
38 genes

- Coding regions of all included genes.
- CNVs screening for all genes.
- SVs (fusions) for the most common genes *ALK*, *ROS1* and *RET*.
- TERT promoter included.

AKT1	*DDR2*	*KRAS*	*PTEN*
*ALK**	*EGFR*	*MAP2K1*	*RET**
AR	*ERBB2*	*MET*	*ROS1**
ARAF	*ERBB3*	*MTOR*	*SMARCB1*
ARIAD1A	*ESR1*	*NF1*	*TERT Prom*
ATM	*FGFR3*	*NRAS*	*TP53*
BRAF	*HRAS*	*PDGFRA*	*TSC1*
BRCA1	*IDH1*	*PIK3CA*	*TSC2*
BRCA2	*IDH2*	*POLE*	
CDKN2A	*KIT*	*PTCH1*	

MILESTONES

1998
Creation of Sistemas Genómicos

2003
Establishment of the biomedical division

2005
Merger with the Institut de Genètica Mèdica i Molecular (IGEM)

2005
Participation in the largest genome sequencing project carried out in Spain 'International Tomato Genome Sequencing Project'

2006
New headquarters in Paterna (Valencia)

2006
ISO9001 certification for the pre-implantational genetic diagnosis unit

2006
IVGEN Inaguration

2006
ISO9001 Certification for the Sanger sequencing units

2011
LIFE project

2011
VENOMICS: FP7 project focused on the massive sequencing of toxins for the development of new drugs based on natural venoms

2012
ISO15189 accreditation for the PGD unit

2013
FRAILOMIC: FP8 project focused on biomarkers of fragility at advanced age

2014
GeneSystems launch, an innovative tool for NGS data analysis and interpretation

2014
License for the manufacturing of sanitary products (NGS kits) granted by AEMPS

2014
CLIA-USA certificate obtained

2014
First patent from LIFE project

2014
First product for diagnosis witch CE marking (NGS kits)

2015
Extension of the scope of license for the manufacturing of sanitary products (bioinformatic analysis software) granted by AEMPS

2015
ISO 13485 certification for in vitro diagnosis medical devices manufacturing

2015
Laser biopsy service

2015
Preconception Geneprofile

2016
GeneSystems© software registration with CE conformity marking for medical devices

2016
3 Horizon 2020 European research projects launched: GOODBERRY, DESIREE and LIQBIPSENS

2018
First university specialization course in embryo biopsy

2017
Expansion of the SG BabyTest range: Launch of the Advanced version

2019
Creation and Launch of GeneBytes

2018
Preconception Focus Geneprofile

2020
Sars-Cov-2 detection services and kits

2019
Publication of cut points for FISH automated in sperm

2020
Preconception Optimized Geneprofile

2021
Preconception Universal Geneprofile

2022
Creation and Launch of HubExome Plus Panel - GeneSGKit®

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