SISTEMAS GENÓMICOS
A MEMBER OF SYNLAB

HubExome Plus Panel

HubExome Plus Panel - GeneSGKit®

CE-IVD

Single kit for the study of all human pathologies from whole exome capture and mitochondrial DNA with CE-IVD marking.

Single integrated solution on the market:
HubExome Plus Panel® includes reagents to process 16-48 samples with bioinformatic computation (secondary analysis) and visualization, priorization and interpretation system included (tertiary analysis). The kit, secondary and tertiary analysis have CE-IVD marking.

Maximum coverage, sensitivity and specificity:
Captures all exonic regions of the genome (›19000 genes), flanking regtions (+/- 20 bp) and mtDNA with a compact size of ~ 40 Mb. Its high specificity and sensitivity in the capture process ensure ›98% horizontal coverage at 20x, with an average coverage of 100x for the nuclear target, and 100% at 250x with an average coverage of 5000x for the mitochondrial genome.

Comprehensive study of all types of mutations:
Validated capture for the detection of SNVs, INDELs, large INDELs, MNVs, ALUs and CNVs.

Absolute traceability of samples:
Sample tracking system included in order to ensure a correct association between patient and result.

Speed:
Libraries are ready in just 12 hours from 100 ng of DNA.

Automation:
Possibility of automating the wet process with different commercial robots for generating libraries.

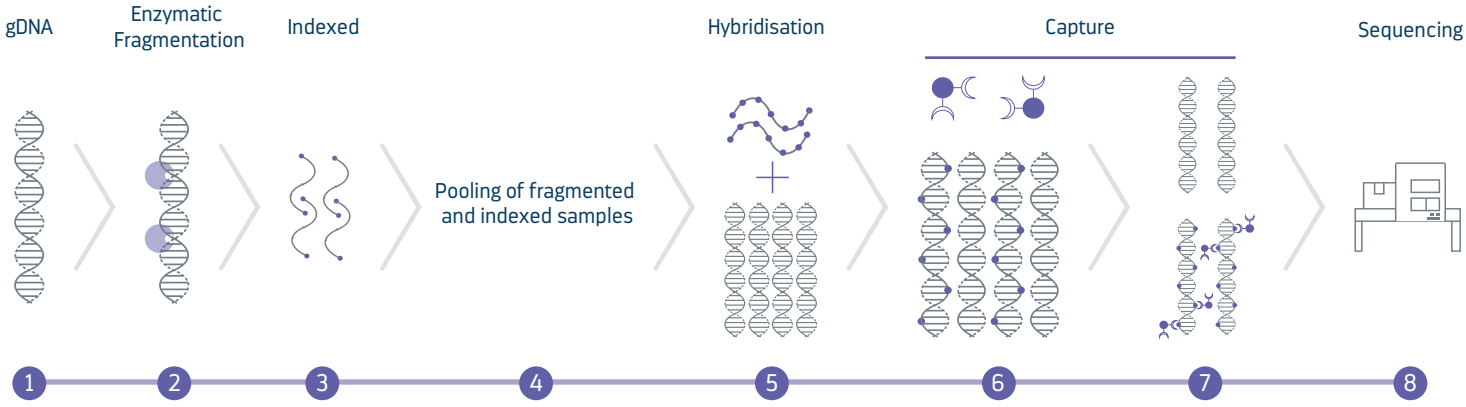
Versatility:
Compatibility with Illumina® or MGI sequencing platforms, allowing for interoperabillity and flexibility in the use of different sequencing equipment.

Complete bioinformatic analysis and results visualization with GeneSystems®:
The data is computed in less than 24 hours, scoring against the latest versions of more than 100 databases, both public and private.

HubExome Plus Panel® includes within the CE-IVD scope the analysis of the following pathologies:

- Bone Dysplasia and Collagen Disorders
- Cardiology
- Ectodermal Diseases
- Endocrinology
- Epilepsy
- Gastroenterological Diseases
- Hearing Loss
- Hematology
- Immunology
- Intellectual Disability and Austism Spectrum Disorder
- Metabolopathy
- Mitochondrial Diseases
- Neurology
- Ophtalmology
- Renal Diseases

WORKFLOW de GeneSGKit®



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©.

HubExome Plus Panel® provides high performance from only 100 ng of DNA. The design of the capture probes and bioinformatic pipeline guarantees high sensitivity and specificity for the detection of SNVs, indels, large indels, MNVs, ALUs and CNV screening with a recommended average coverage of 100x.

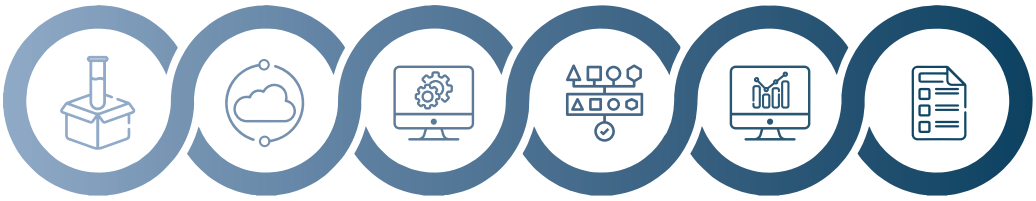
Sistemas Genómicos understands the vital importance of quality customer service and follow-up. This includes a personalised service that offers not only support over the telephone for all sorts of questions and concerns that may arise while using GeneSGKits® or the GeneSystems© analysis platforms, but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

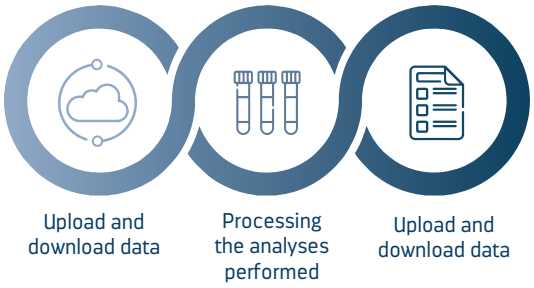


FEATURES OF GENESYSTEMS©

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 **SNVs, 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 FASTQ, 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.
- Prioritisation of results using combined algorithms:
  - Phenotype (HPO).
  - Target tissue expression (GTEX).
  - Pathogenicity (ACMG guidelines).
- Manages the primary databases in the sector (DBNLVar, Ensembl, GnomAD, HGMD-Pro, ClinVar, etc.)
  - Pathogenicity indicators with links to scientific papers.

CATALOGUE - HubExome Plus Panel - GeneSGKit®

Technical data

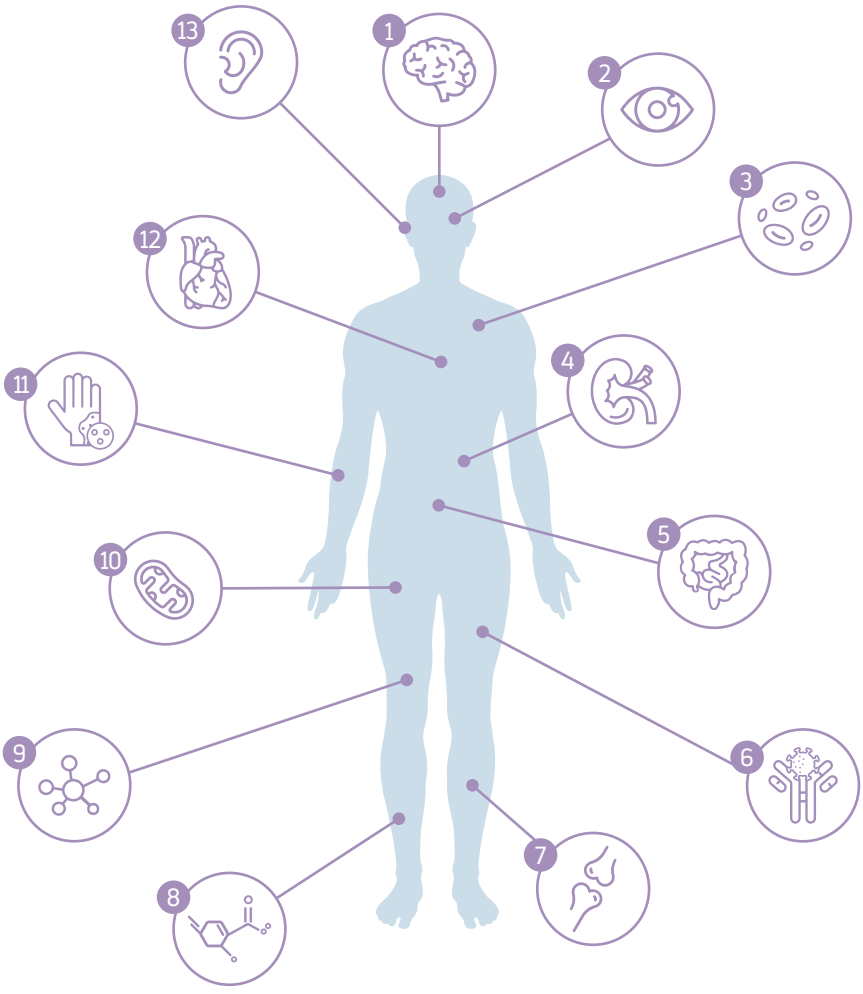
GeneSGKit®	Size (MB)
HubExome Plus Panel - GeneSGKit®	40

References

HubExome Plus Panel® 16 reactions Ref.: LV4612 (CE-IVD) (Illumina® NextSeq, Illumina® NovaSeq, MGI DNBSeq-G400)

HubExome Plus Panel® 48 reactions Ref.: LV4613 (CE-IVD) (Illumina® NextSeq, Illumina® NovaSeq, MGI DNBSeq-G400)

Virtual gene panels design has been carried out following the most up-to-date international guidelines and scientific recommendations. In addition, **full coverage** of exons and +/-20 bp of intronic and UTR regions is ensured for all of included genes.



1. Neurology | 806 genes
   Epilepsy | 523 genes
   Intellectual Disability and Austism Spectrum Disorder | 1564 genes
2. Ophtalmology | 508 genes
3. Hematology | 153 genes
4. Renal Diseases | 339 genes
5. Gastroenterological Diseases| 153 genes
6. Immunology | 458 genes
7. Bone Dysplasia and Collagen Disorders | 608 genes
8. Metabolopathy | 400 genes
9. Endocrinology | 253 genes
10. Mitochondrial Diseases | 384 genes
11. Ectodermal Diseases | 323 genes
12. Cardiology | 608 genes
13. Hearing Loss | 179 genes

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 Panel GeneSGKit

21082023V02

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