SISTEMAS GENÓMICOS

ΔSCIRES

MITOCONDRIAL

# Mitocondrial-GeneSGKit®

Sistemas Genómicos has developed Mitocondrial-GeneSGKit® for the study of **207 genes relevant to the diagnosis of mitochondrial diseases due to mutations in nuclear genes.** Mitocondrial-GeneSGKit® allows the diagnosis of different pathologies such as nuclear genetic disorders of the mitochondrial respiratory chain, nuclear genetic disorders of the mitochondrial respiratory chain, nuclear genetic disorders of the mitochondrial respiratory chain, nuclear genetic disorders associated with mitochondrial depletion, CoQ10 coenzyme primary deficiency, Barth syndrome, glutaric acidemia, etc.

The workflow is optimised to prepare the libraries in nine hours and the technology used allows the combination of multiple samples and different GeneSGKits® in a single run on Illumina® sequencing platforms, ensuring an equitable balance of data from the different samples.

## HIGHLIGHTS

### The most complete solution on the market

Mitocondrial-GeneSGKit® is cleared for CE-IVD and contains reagents to process twelve samples with the corresponding bioinformatics analysis and visualization by the GeneSystems© platform.

### Fast workflow and low input requirements

The libraries are ready within nine hours of work starting with 50 ng of DNA.

### Complete bioinformatic analysis and data visualization

The bioinformatics pipeline is executed in less than 48 hours, providing the user with a rich variant annotation including public, private and Sistemas Genómicos' reference databases. This comprehensive annotation reports SNPs and indels, which are visualized and easily filtered on GeneSystems©.

### CNVs detection

Sensitive CNVs detection validated in prestigious public and private organisations.

1

**GeneSGKits® library preparation**

2

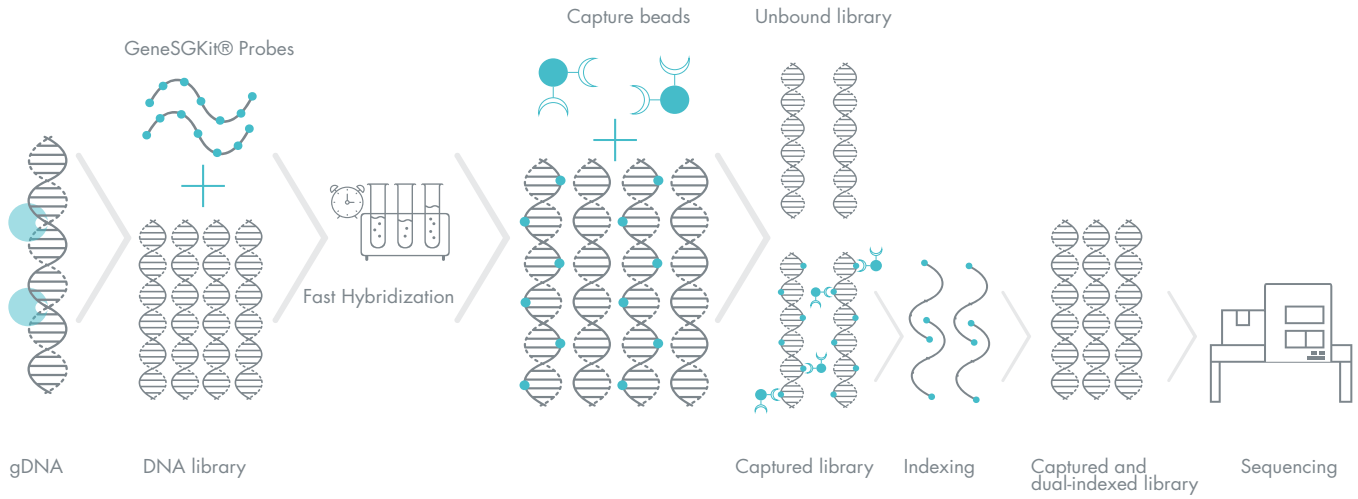
**Sample sequencing on Illumina®* platforms**

3

**Analysis and data interpretation on GeneSystems©**

*HiSeq and MiSeq. Other sequencing platforms in validation

## GeneSGKit® WORKFLOW




## ABOUT US

Sistemas Genómicos is a pioneering company in the application of Next Generation Sequencing technology for genetic diagnosis. Accredited and certified by the major entities in quality assurance (ENAC, AENOR and CLIA), we offer solutions to accelerate and standardize clinical genetic testing, enabling clinicians to provide accurate diagnostics to their patients.

## OUR MISSION

We strive to provide the best diagnostic solutions with the best technology and knowledge available to maximise patient benefit. Our integrated systems have helped laboratories for the last two decades to improve their diagnostics and research work, by developing aunique range of CE-IVD solutions.

## DESIGN

The GeneSGKits® offer excellent performance from only 50 ng of DNA. The capture probes are based on ultra-long RNA baits (120 nucleotides) with high sensitivity and specificity for the detection of SNPs, indels and CNVs. Our probe design captures all the coding regions of each targeted gene, including splicing sequences, with a recommended average coverage of 200x.

## PERFORMANCE

With GeneSGKits® clinicians and researchers can be confident in the accuracy and reproducibility of the sequencing data. Each sample is sequenced with a minimum coverage of 20x in over 98% of the regions of interest for relevant pathologies, guaranteed by strict validation protocols.

## SUPPORT

For Sistemas Genómicos, customer is at the center of our work. This includes a personalised service to enable a seamless implementation of diagnostic next generation sequencing. We offer personalised training programs to ensure a quick acquisition of the skill set required to successfully and reliably harness the latest advances in sequencing technology to maximize patient benefit.

GeneSGKit®

# GeneSystems©
## SISTEMAS GENÓMICOS

GeneSystems© is a leadeing platform for the analysys of NGS data for diagnosis and genetic research which is registered as a health product with a CE Marking for diagnostic use in vitro for processing information from massive DNA sequencing to detect genetic variants and help to diagnose at the molecular level heterogeneous genetic diseases by means of directed resequencing.
Developed in a private, secure and reliable Cloud Computing environment, it includes:

1 

**Uploading and downloading data**

2 

**Complete acces to all the data, from raw data to annotated variants**

3 


**Visualization and prioritization of results**

It is developed by a multidisciplinary team of geneticits, bioinformatic and engineers, and validated by more than 200 companies, including hospitals, laboratories, research centers, universities, pharmaceutical companies. The platform has been audited independently and allows the detection of SNP and INDELS.

## DATA ANALYSIS

GeneSystems© is designed to solve the different stages of the analytical data cycle of massive sequencing and provides solutions from the FASTQ files to the clinically-relevant data.

GeneSystems© captures annotations at gene, variant and transcript level and incorporates the most clinically relevant databases for the interpretation of variants, such as Ensemble, 1000Genomes, HGMD, dbSNP, RefSeq, Interpro, Prosite, Cosmic, HapMap, OMIM, ClinVar, ExAC, etc. Furthermore, GeneSystems© users have access to the proprietary variant database of Sistemas Genómicos. Within the platform there is also information on conservation scores, pathogenicity in silico predictors, flanking regions, among others. A one stop solution to find clinical meaning in your sequencing data.

GeneSystems© provides a highly sensitive CNVs detection validated in prestigious public and private organisations, which optimizes the MLPA experiments thus reducing costs and diagnostic turnaround times.

### Hight data quality.

With the GeneSGKits® technology, clinicians and researchers will have the best tools for genetic diagnosis, thanks to the excellent quality of the sequencing data of the samples prepared with GeneSGKit®. Each sample will be sequenced with low variance in the coverage values with 98% of the regions sequenced at a minimum depth of 20x.

# Mitocondrial-GeneSGKit® CATALOG

## Technical data

| GeneSGKit® | Genes | Size (MB) | % COVERAGE 12x | % COVERAGE 20x |
|---|---|---|---|---|
| Mitocondrial-GeneSGKit® | 207 | 0.48 | 99.78 | 99.77 |

## References

Mitocondrial-GeneSGKit® Ref.: LV3482 (Kit CE-IVD) / LV3483 (Kit RUO)

## Genes description

### Mitocondrial-GeneSGKit®

| | | | | | | |
|---|---|---|---|---|---|---|
| *AARS2* | *CISD2* | *ETFDH* | *MARS* | *NDUFAF1* | *PDHX* | *SPG7* |
| *ABCB7* | *COA5* | *ETHE1* | *MARS2* | *NDUFAF2* | *PDP1* | *SUCLA2* |
| *ABCD4* | *COA6* | *FARS2* | *MCCC1* | *NDUFAF3* | *PDSS1* | *SUCLG1* |
| *ACAD9* | *COQ2* | *FASTKD2* | *MCCC2* | *NDUFAF4* | *PDSS2* | *SURF1* |
| *ACADM* | *COQ4* | *FBP1* | *MCEE* | *NDUFAF5* | *PET100* | *TACO1* |
| *ACADS* | *COQ6* | *FBXL4* | *MFN2* | *NDUFAF6* | *PNPT1* | *TARS2* |
| *ACADSB* | *COQ9* | *FECH* | *MGME1* | *NDUFAF7* | *POLG* | *TAZ* |
| *ACADVL* | *COX10* | *FH* | *MICU1* | *NDUFB3* | *POLG2* | *TIMM44* |
| *ACAT1* | *COX14* | *FOXRED1* | *MMAA* | *NDUFB7* | *PPM1K* | *TIMM8A* |
| *ACO2* | *COX15* | *FTMT* | *MMAB* | *NDUFB9* | *PUS1* | *TK2* |
| *ADCK3* | *COX20* | *G6PC* | *MMACHC* | *NDUFS1* | *RARS2* | *TMEM126A* |
| *AFG3L2* | *COX6B1* | *GFER* | *MMADHC* | *NDUFS2* | *REEP1* | *TMEM70* |
| *AGK* | *COX7B* | *GFM1* | *MPC1* | *NDUFS3* | *RMND1* | *TPK1* |
| *AIFM1* | *CPT2* | *GFM2* | *MPC2* | *NDUFS4* | *RRM2B* | *TRMU* |
| *ALAS2* | *CYC1* | *GYS1* | *MPV17* | *NDUFS6* | *SARS2* | *TSFM* |
| *ALDH2* | *D2HGDH* | *GYS2* | *MRPL3* | *NDUFS7* | *SCO1* | *TTC19* |
| *AMACR* | *DARS2* | *HADHA* | *MRPL44* | *NDUFS8* | *SCO2* | *TUFM* |
| *ATP5A1* | *DBT* | *HADHB* | *MRPS16* | *NDUFV1* | *SDHAF1* | *TYMP* |
| *ATP5E* | *DECR1* | *HLCS* | *MRPS22* | *NDUFV2* | *SERAC1* | *UNG* |
| *ATPAF2* | *DGUOK* | *HMGCS2* | *MTFMT* | *NFU1* | *SFXN4* | *UQCC2* |
| *AUH* | *DLAT* | *HSD17B10* | *MTO1* | *NUBPL* | *SLC19A2* | *UQCRB* |
| *BCKDHA* | *DLD* | *IBA57* | *MTPAP* | *NUP62* | *SLC19A3* | *UQCRC2* |
| *BCKDHB* | *DNA2* | *IDH2* | *MUT* | *OPA1* | *SLC25A1* | *UQCRQ* |
| *BCS1L* | *DNAJC19* | *ISCU* | *NDUFA1* | *OPA3* | *SLC25A12* | *VARS2* |
| *BOLA3* | *DNM1L* | *LARS* | *NDUFA10* | *PC* | *SLC25A13* | *VTI1A* |
| *C10orf2* | *EARS2* | *LIAS* | *NDUFA11* | *PCCA* | *SLC25A15* | *WFS1* |
| *C12orf65* | *ECSIT* | *LMBRD1* | *NDUFA12* | *PCCB* | *SLC25A19* | *YARS2* |
| *CA5A* | *ELAC2* | *LRPPRC* | *NDUFA2* | *PCK2* | *SLC25A3* | |
| *CD320* | *ETFA* | *LYRM4* | *NDUFA8* | *PDHA1* | *SLC25A4* | |
| *CHKB* | *ETFB* | *LYRM7* | *NDUFA9* | *PDHB* | *SLC37A4* | |

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

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

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