SISTEMAS GENÓMICOS
ASCIRES

CFTR

CFTR-GeneSGKit®

Sistemas Genómicos has developed the CFTR-GeneSGKit® for the **most complete, reliable and efficient study of the CFTR gene** and detection of variants involved in Cystic Fibrosis diagnosis.

CFTR-GeneSGKit® contains reagents to process **12 samples with the corresponding bioinformatic analysis** through the GeneSystems©platform. It is designed to capture all the *CFTR* adjacent exons and intronic regions for Cystic Fibrosis diagnosis.

The workflow is optimised to create the **libraries in just 6 hours** and the technology used allows the **combination of multiple samples and different GeneSGKit® in a single run** on Illumina® sequencing platforms, ensuring an equitable distribution of data between the different samples.

HIGHLIGHTS

Only integrated solution of its kind on the market:
CFTR-GeneSGKit® contains reagents to process 12 samples with the corresponding bioinformatic analysis and visualisation using the GeneSystems© platform.

Fast:
The libraries are ready after just 6 hours of work.

Complete bioinformatic analysis and visualisation of results:
The data is computed and recorded against the main open and private databases reporting SNP, Indels and CNVs which are visualised and easily filtered through GeneSystems©.

1

Preparation of the library with GeneSGKits®

2

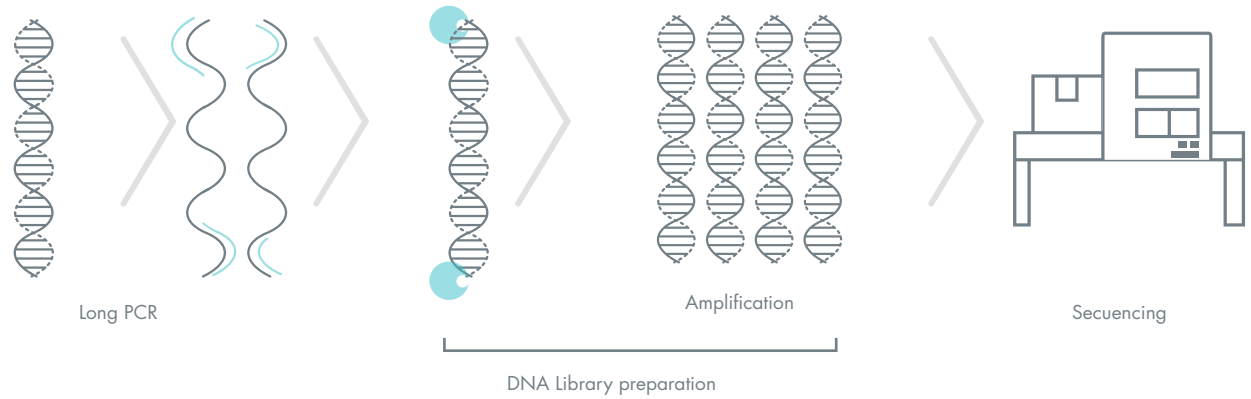
Sequencing on Illumina® platforms*

3

Analysis and interpretation of data through GeneSystems©

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

CFTR-GeneSGKit® WORKFLOW



WHO WE ARE

Systems 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 approved by ENAC (Entidad Nacional de Acreditación, [National Accreditation Body of Spain]), AENOR (Asociación Española de Normalización y Certificación [Spanish Association for Standardization and Certification]) and CLIA (Clinical Laboratory Improvement Amendments).

OUR GOAL

To use all our experience in analysis and diagnosis by applying NGS to develop a series of tools that allow genetic diagnosis in situ in hospitals and research centres. For this, we have developed an integrated solution that is the only one of its kind on the market: GeneSGKit®, with more than 27 references with CE-IVD marking and the GeneSystems© bioinformatic computing platform and analysis of results.

DESIGN

CFTR-GeneSGKit® provides a high performance from only 1 µg of DNA. The workflow includes a first long PCR step for the amplification of the regions of interest, followed by the addition of adaptors and indexing. Its design guarantees **high sensitivity and specificity for detecting point mutations, indels and CNVs with a recommended average coverage of 300x**.

PERFORMANCE

GeneSGKit® a minimum coverage of 20x for 100% of *CFTR* regions included in the panel design according to the results obtained on Illumina®, sequencing platforms, supported 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 leading 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, DBNLVar, 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 100% of the regions sequenced at a minimum depth of 20x.

CFTR-GeneSGKit® CATALOGUE

Technical data

GeneSGKit®	Genes	Size (MB)	% COVERED 12x	% COVERED 20x
CFTR-GeneSGKit®	1	0,01	100	100

700x recommended coverage

References

CFTR-GeneSGKit® Ref.: LV3835 (Kit IVD-CE) / LV3834 (Kit RUO)

Description of genes

CFTR-GeneSGKit®

CFTR

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