SISTEMAS GENÓMICOS

ASCIRES

NIPT

NIPT-GeneSGKit®

Sistemas Genómicos has developed **NIPT-GeneSGKit®, the most complete test available for noninvasive prenatal screening for chromosomal aneuploidies** present in the fetus, as well as establishing gender.

THE MOST COMPLETE SOLUTION ON THE MARKET

NIPT-GeneSGKit® is **CE-IVD marked*** and contains **reagents** to process 12-48 samples with the corresponding bioinformatics analysis, visualization and personalized report by the **GeneSystems© platform**.

The work process involves a "hands-out" manipulation time, completed in 3 hours, once the circulating free DNA (cfDNA) has been obtained (5ng).

Validated complete bioinformatic analysis.

Simple and immediate generation of individualized reports.

Integration in user's facilities.

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 (ISO9001, ISO13485, ISO17025 ISO15189 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 maximize patient benefit.

Our NIPT allows the implementation of this service in hospitals and other centers' facilities without externalizing any sample, with highly reliable results, reducing times for obtaining the report.

GeneSGKits® library preparation

Sample sequencing on **Illumina®** platforms

Analysis and data interpretation on **GeneSystems©**

*Chromosome 21 excluded

NIPT-GeneSGKit ®

NIPT-GeneSGKit® is an integrated solution that includes bioinformatics analysis, visualization and personalized report by **GeneSystems©** platform.

This system, optimized and friendly-use, allows a **fast implementation in lab routines**, without needs of extra resources to maximize the number of processed samples.

NIPT-GeneSGKit ® allows detection of chromosomal abnormalities in the fetus at an early stage of the pregnancy, as well as the gender of the fetus using a sample of maternal blood. It examines all the chromosomes.

- **Trisomy 21* associated with Down's syndrome.**
- **Trisomy 18 associated with Edwards' syndrome.**
- **Trisomy 13 associated with Patau syndrome.**
- **Sex chromosome aneuploidies:**
  - **Turner syndrome (presence of a single X sex chromosome)**
  - **Klinefelter syndrome (47, XXY)**
  - **Triple X syndrome**
  - **Polysomy X**
  - **Karyotype XYY**
- **Mosaicisms (according to cfDNA)**

The algorithm of detection associates each chromosome to a low, high or undetermined risk depending on the obtained signal:

LOW RISK

Very low probability of fetal chromosomal aneuploidy in any of the analysed chromosomes.

HIGH RISK

High probability of fetal chromosomal aneuploidy in the indicated chromosome. The result should be confirmed by performing a definitive diagnostic test such as invasive prenatal chorionic villus sampling or amniotic fluid testing.

UNDETERMINED RISK

The risk of aneuploidy in the indicated chromosome cannot be determined by the values obtained from the test, because although the result may not be normal, it is not sufficiently abnormal to be high risk.

NIPT-GeneSGKit® has a high precision, supported by strict validation processes. Its combination with other prenatal studies (fetal ultrasound or biochemical screening performed in the first trimester), **allows the reduction of false positives to less than 1%**.

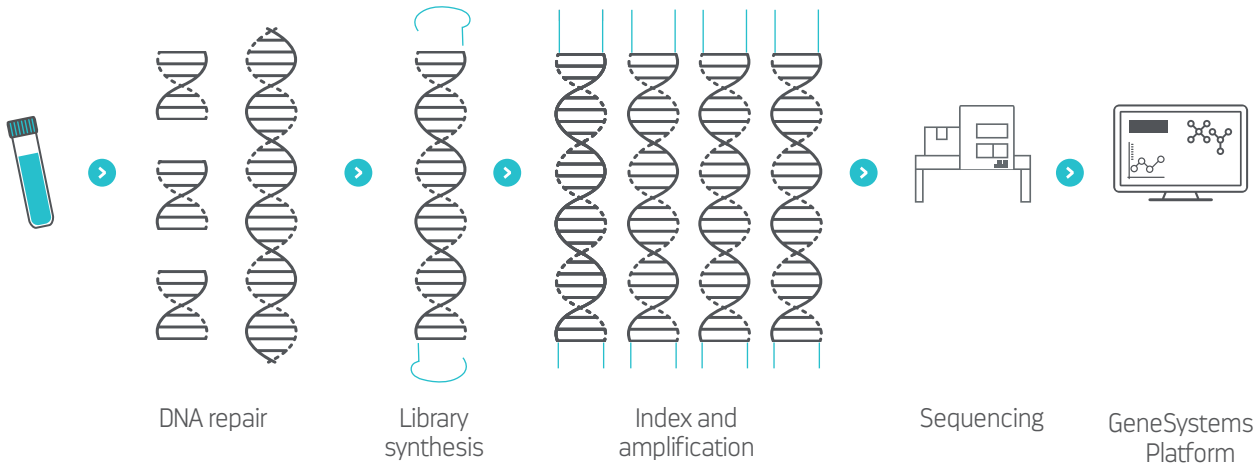
NIPT-GeneSGKit® can be performed **from week 9 of pregnancy** (with a minimum fetal fraction of 3.5%). It is particularly indicated in women who want to rule out chromosomal aneuploidies regardless of their genetic condition or family history, it can be used for all pregnant women, including pregnancies achieved through assisted reproductive techniques and oocyte donation and twins genetically identical, or twins not genetically identical it fetal fraction is suitable for the test allowing aneuploidies detection in both fetus.

*CE-IVD marked excluded

DESIGN AND PROTOCOL

The working protocol is optimized to make the libraries in 3 simple steps and in the same tube, reducing the risk of errors during the process::

1. **Sample preparation:** repair of cfDNA.
2. **Library synthesis.**
3. **Library amplification:** obtaining a large number of copies of DNA fragments by PCR introducing the compatible index with NGS of Illumina® that also allows sequencing of multiple samples by run or lane.



This technology allows to generate libraries with a high capacity of multiplexing and low ranges of duplication providing the following advantages:

- It performances from only 5ng of double-stranded cfDNA obtained from maternal plasma without need of fragmentation.
- Higher performance compared to other technologies.
- Fast and simple process.
- Uniform coverage regardless of content in GC.
- The required materials are commonly used in the laboratory.

SENSIBILITY AND SPECIFICITY

To obtain the values of sensitivity and specificity, the samples were treated following the preanalytical recommendations for obtaining DNA. The libraries were processed with the kit and sequences on Illumina's NGS platforms. Finally, we proceeded to the analysis of the reads obtained by an informatic algorithm, developed entirely by Sistemas Genómicos and validated by independent centers.

CONDITION	SPECIFICITY	SENSITIVITY	VPP	VPN
Trisomy 21*	>99%	>99%	>99%	>99%
Trisomy 18	>99%	>99%	>99%	>99%
Trisomy 13	>99%	>99%	>99%	>99%
Trisomies 9, 16, 22	>99%	-	-	>99%
Trisomy autosomes (Except Other Trisomies cr.19)	>99%	-	-	>99%
Aneuploidies of sexual chromosomes (XO, XXY, XXX, XYY)	>97%	-	-	>99%

CONDITION	SENSITIVITY
Fetal Gender	>99%

*CE-IVD marked excluded

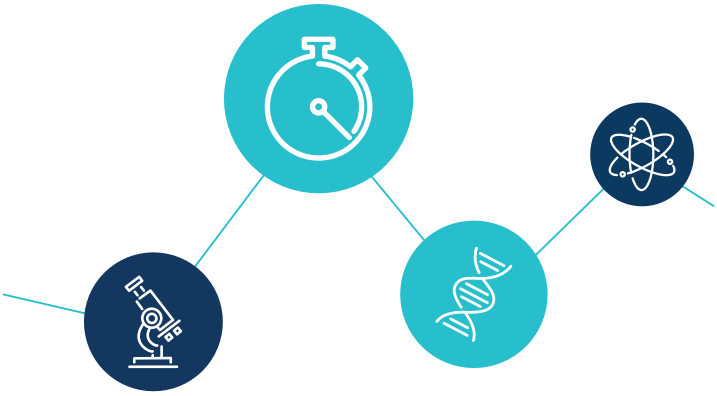
GeneSystems ©

GeneSystems© hosts a number of **versatile and easy-to-use tools for analyzing data from massive sequencing, visualization and interpretation of results**. It offers a complete and personalized service, adaptable to needs. This platform, developed entirely in a cloud computing environment with advanced bioinformatic algorithms of Sistemas Genómicos, allows high availability and scalability of data, minimizing the consumption of the user's own resources.

The platform is adapted to solve the needs during the analysis process, from the analysis of the results from sequencing to the preparation of the final report. Its intuitive and optimized interface for the data generated from the sequencing of libraries processed with **NIPT-GeneSGKit®** is designed to be **managed without bioinformatics knowledge.**

HIGH QUALITY DATA

The platform is able to analyze the data of **GeneSGKit®** and to detect aneuploidies in any fetal chromosome. After low coverage sequencing of the complete genome (0.1%), **GeneSystems©** aligns the sequencing data of the samples against the reference genome. Using an algorithm developed and validated by Sistemas Genómicos, **the platform detects alterations in the chromosomes**, based on variations in the counting of segments of the fetal chromosomes, as opposed to a normalization of control samples.



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.

CERTIFICATIONS

The manufacturing of the kits and bioinformatics analysis is certified by **ISO13485.**

REFERENCES

NIPT-GeneSGKit® 12 reactions Ref.: LV3736 (Kit CE-IVD) / LV3188 (Kit RUO)

NIPT-GeneSGKit® 48 reactions Ref.: LV3737 (Kit CE-IVD) / LV3667 (Kit RUO)

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