SISTEMAS GENÓMICOS
ASCIRES

PRECON

Precon-GeneSGKit®

Sistemas Genómicos has designed the **Precon-GeneSGKit® for the purpose of detecting and determining the carrier or non-carrier status** of future parents for recessive, X-linked hereditary diseases.

THE MOST COMPLETE **SOLUTION**

Only integrated solution of its kind on the market.
Precon-GeneSGKit® contains reagents to process 12 samples with the corresponding bioinformatic analysis and visualisation of results.

Complete.
Includes the analysis of 345 diseases, examining 32749 causal mutations in 320 genes.

Fast.
The libraries are ready after just 9 hours of work from 50 ng of DNA.

Complete bioinformatic analysis and visualisation of results.
The data are computed in less than 48 hours, detecting specific mutations described in regulatory, exon, intron, exon-intron intermediate and flanking regions.

WHO **WE ARE**

Sistemas Genómicos is a pioneering company in the use of Next Generation Sequencing (NGS) for genetic diagnosis. It has a long history in reproductive genetics through different techniques developed specifically for pre-implantation genetic diagnosis, infertility and analysis to detect carrier status.

Accredited and certified by the strictest quality controls (ISO9001, ISO13485, ISO17025, ISO15189 and CLIA), Sistemas Genómicos offers solutions for implementing and standardising genetic diagnosis based on NGS, giving professionals the necessary tools to complete the diagnostic process quickly and efficiently.

OUR **GOAL**

Sistemas Genómicos works to provide optimised **diagnostic solutions** with the aim of providing our customers with experience and knowledge for the benefit of patients.

Our Precon-GeneSGKit® allows this service to be carried out in laboratories without the need to send samples for external analysis and with highly reliable results, reducing the time taken to obtain the results report.

GeneSGKits® library preparation

Sample sequencing on **Illumina®** platforms

Analysis and data interpretation on **GeneSystems©**

Precon-GeneSGKit ®

Precon-GeneSGKit® is a key tool in establishing the carrier status of future parents for certain diseases of known genetic basis.

The kit makes it possible to ascertain the real genetic risk of having affected offspring. If the future biological parents are identified as being asymptomatic carriers of any of the diseases studied, adequate genetic counselling is recommended to determine the possible reproductive options in each case, on a case-by-case basis.

The diseases analysed with the **Precon-GeneSGKit®** have been selected based on clinical criteria and the recommendations of international scientific societies according to:

- **Prevalence in the general population (> 1/100,000 births).**
- **Prevalence of cases in paediatric emergency departments.**
- **Diseases causing intrauterine and neonatal death.**
- **Diseases included in extensive neonatal studies (genetic and neonatal screening, "heel prick" test).**

The detection algorithm associates each parent with a high risk, low risk or residual risk according to the results obtained:

LOW GENETIC RISK

The future parents have a low risk of transmitting one of the diseases included in the panel or the woman is not a carrier of the variant in an X-linked gene.

HIGH GENETIC RISK

The future parents have a high risk of transmitting one of the diseases included in the panel or the woman is a carrier of a variant in an X-linked gene.

RESIDUAL RISK

The risk remains even in the event that the test is negative. This risk is due to other variants that are undetectable through the study technique and/or the personal biological events of each individual, which cannot be detected by this analysis (genetic recombination in gametes, germinal mosaicism, gene conversions and de novo variants in offspring).

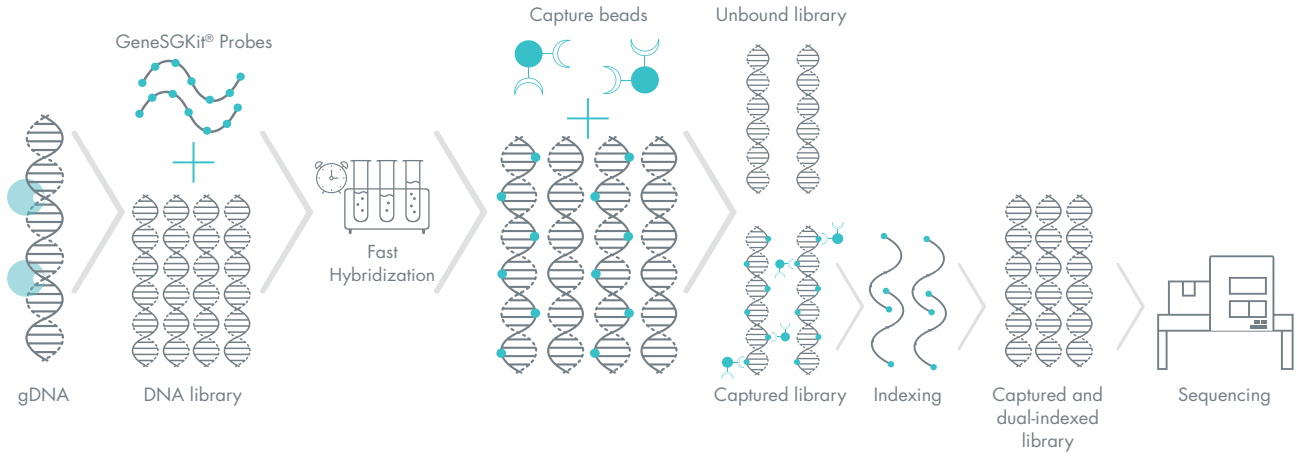
DESIGN AND WORKING PROTOCOL

GeneSGKit® provides a high performance from only 50 µg of DNA.

The capture probes are based on Ultra-Long RNA Baits (120 nucleotides) that guarantee high sensitivity and specificity for the detection of SNPs and indels.

This probe design makes it possible to capture 32749 causal mutations in 320 genes linked to 345 hereditary diseases.

The design includes regulatory, exon, intron, exon-intron intermediate and flanking regions, according to the location of the variant of interest.



PERFORMANCE

GeneSGKit® allows a minimum coverage of 20x for over 98% of the regions of interest for the specific diseases according to the results obtained on Illumina® sequencing platforms, supported by strict validation protocols.

HIGH QUALITY OF DATA ANALYSED

Thanks to the **GeneSGKit®** ® technology, clinics and researchers will have the best tools on the market for genetic diagnosis and analysis, thanks to the high quality of the sequencing data of samples prepared using the **GeneSGKit®**.

Each sample will be sequenced with uniform coverage values for the regions of interest, with 98% of the regions covered at a minimum coverage of 20x.

REFERENCES

Precon-GeneSGKit® (HiSeq, MiSeq) Ref:: LV3765 (Kit RUO)

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