

DGP-GeneSGKit[®]

Sistemas Genómicos, a member of the biomedical group ASCIRES, has designed the **DGP-GeneSGKit[®]**, the most thorough and sensitive test on the market for detecting aneuploidies, chromosomal segments and unbalanced structural chromosome rearrangements and CNVs based on blastomere and trophoctoderm samples for next-generation sequencing (NGS) platforms*.

DGP-GeneSGKit[®] enables chromosome abnormalities and aneuploidies to be detected with a limit of detection of 20 Mb in human embryos prior to implantation, based on blastomere or trophoctoderm samples using low-coverage whole-genome sequencing.

The detection algorithm assigns each chromosome a low, high or indeterminate risk depending on the results obtained:

- **Low risk:** low probability of aneuploidy in the chromosome analysed.
- **High risk:** high probability of aneuploidy in the chromosome analysed.
- **Indeterminate risk:** it is not possible to completely rule out the presence of aneuploidy.

HIGHLIGHTS

A comprehensive solution that is unique on the market:

DGP-GeneSGKit[®] contains reagents for processing **48 samples** with their corresponding bioinformatics analysis, viewing and generating a personalised report using the in-cloud GeneSystems[®] platform.

Quick and easy:

The workflow comprises three simple steps based on blastomere or trophoctoderm samples, with a library production time of 3 hours.

Bioinformatics analysis:

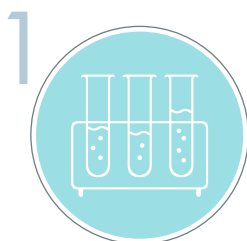
A validated complete bioinformatics analysis and personalised reports may be generated quickly and easily.

Quality:

Highly reproducible and reliable results that ensure a high-quality analysis for day-to-day practice in detecting aneuploidies, chromosomal segments and unbalanced structural chromosome rearrangements and CNV's with a limit of detection of 20 Mb.

Optimised incorporation into the laboratory:

The test may be quickly incorporated into routine practice at the user's facility and readily scaled, with no need to obtain additional resources to increase the number of samples processed.



Library preparation with
GeneSGKits[®]



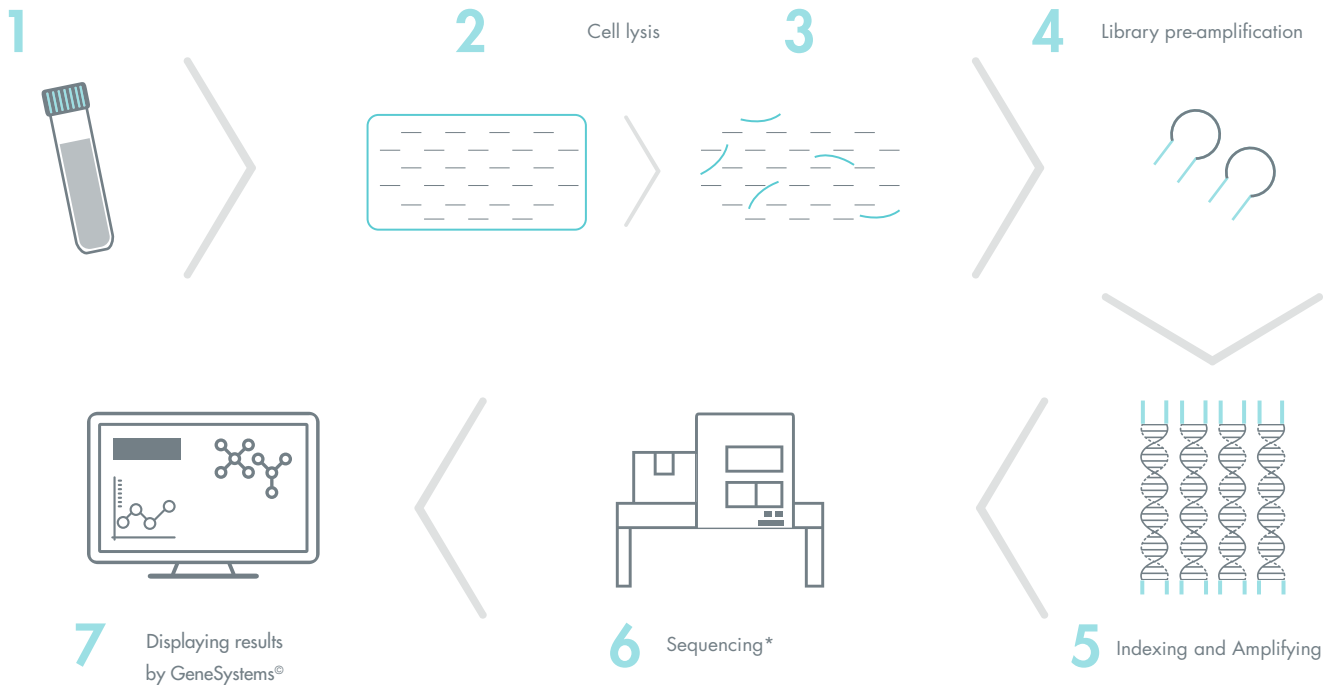
Sequencing using Illumina[®]
platform*



Analysing and interpreting data
using GeneSystems[®]

*MiSeq, NextSeq

DGP GeneSGKit® WORKFLOW



This technology generates libraries with a high capacity for multiplexing and low ranges of duplication:

- ✓ Higher yield compared to other technologies
- ✓ Quick and easy workflow
- ✓ Uniform coverage regardless of GC content
- ✓ Required materials commonly used at laboratories

*Enables sequencing of multiple samples by run or lane

WHO WE ARE

Sistemas Genómicos (ASCIREs Group) is a leader in the **use of next-generation sequencing (NGS)** for genetic diagnosis. We have extensive experience in reproductive genetics using different techniques specifically developed for pre-implantation genetic diagnosis, tests for carriers of recessive mutations, genetic diagnosis of infertility and more.

Accredited and certified under the most stringent **quality controls (ISO 9001, ISO 13485, ISO 17025, ISO 15189 and CLIA)**, Sistemas Genómicos (ASCIREs Group) offers solutions for implementing and standardising NGS-based genetic diagnosis. We give professionals the tools they need to complete the diagnostic process quickly and efficiently.

OUR GOAL

Sistemas Genómicos (ASCIREs Group) is working to **provide optimised solutions**, based on the most suitable technology, so that our customers may capitalise on our experience and knowledge for the benefit of patients. DGP-GeneSGKit®, a pre-implantation genetic analysis kit from Sistemas Genómicos (ASCIREs Group), allows for incorporation of this service into laboratories with no need to outsource samples and yields highly reliable results, thereby reducing waiting times for obtaining a results report.

GeneSGKit[®]

DESIGN AND WORKFLOW

The DGP-GeneSGKit[®] work protocol is optimised for preparing libraries in 3 simple steps, in a single tube, thereby reducing the risk of errors during the process:

- cell lysis.
- library pre-amplification.
- amplification.



SUPPORT

High-quality customer service, support and follow-up are of the essence to Sistemas Genómicos (ASCIREs Group). We provide a personalised service enabling efficient implementation of NGS-based solutions using our **adapted training programmes**. These programmes make it easy to quickly learn the skills needed to successfully and efficiently use the **latest advances in sequencing technology, and to quickly incorporate GeneSGKits[®]** into routine diagnostic practice at any laboratory.



Sensitivity and specificity by chromosome

To obtain sensitivity and specificity values, samples have been processed with the DGP-GeneSGKit[®] and sequenced using next-generation sequencing platforms from Illumina[®]. Samples have been analysed using a bioinformatics algorithm developed and validated by Sistemas Genómicos (ASCIREs Group).

SENSITIVITY	SPECIFICITY
> 99%	> 99%



GeneSystems®

SISTEMAS GENÓMICOS

GeneSystems® boasts a number of versatile, easy-to-use tools to analyse NGS sequencing data as well as view and interpret results. Thus, it offers a complete, personalised service capable of being adapted to customer needs.



**Uploading and downloading
data**



**Processing analyses
performed**



**Viewing and prioritising
results**

Comprehensively developed in a cloud-computing setting with advanced bioinformatics algorithms created by Sistemas Genómicos (ASCIREs Group), GeneSystems® offers high availability and scalability, thereby minimising use of the user's own resources. The platform is adapted to meet the different needs that arise during an analytical process, from analysis of sequencing results to preparation of the final report using an intuitive interface optimised for DGP data that requires no prior specialised computer knowledge.

ANALYSING SEQUENCING DATA

In a few hours, GeneSystems® is capable of detecting aneuploidies, chromosomal segments and unbalanced structural chromosome rearrangements with a limit of detection of 20 Mb in the samples analysed. Following low-coverage whole-genome sequencing (~0.1x), GeneSystems® aligns the sequencing data for the samples with the reference genome. Using an algorithm developed and validated by Sistemas Genómicos (ASCIREs Group), abnormalities are detected in the chromosome set based on variations in the embryo chromosome segment count versus a control sample standardisation.

References:

DGP-GeneSGKit® Ref.: LV3764 (Kit RUO)

Reference:

Vendrell X, Fernandez Pedrosa V, Triviño JC, Bautista-Llacer R, Collado C, Rodríguez O, García-Mengual E, Ferrer E, Calatayud C, Ruiz-Jorro M. *New protocol based on massive parallel sequencing for aneuploidy screening of preimplantation human embryos*. Syst Biol Reprod Med. 2017, 63(3):162.178. doi: 10.1080/19396368.2017.1312633



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