Preconception GeneProfile® is a genetic test aimed at prospective parents to determine if they are carriers or not for certain hereditary diseases.

Preconception GeneProfile® allows establishing the genetic risk of having affected offspring and, thanks to adequate genetic counselling, offering to the prospective parents the different reproductive options available according to their situation, in a personalised manner.

Who is it for?

- Any couple starting their reproductive project and wishing to know the genetic risk of transmitting one of the hereditary diseases of the panel
- Infertile couples requiring gamete donation. In this case, the donor is the one studied. In the case of couples or single prospective parents, who need egg or sperm donation, Preconception GeneProfile® allows identifying the ideally-suited donor, crossing donor and receptor genetic data through the Match-Gene system, a personalised system that most reduces the reproductive genetic risk.

Which diseases are included?

- 345 hereditary diseases, 318 autosomal recessive and 27 X chromosome-linked, including heart, skin, developmental, endocrinial, gastroenterological, haematological, hepatic, immunological, metabolic, neurological, eye, kidney, respiratory and skeletal disorders, selected according to clinical criteria and recommendations of scientific society based on the following:
  - Prevalence in general population
  - Incidence in paediatric emergency cases
  - Diseases that most frequently cause neonatal death
  - Diseases that most frequently cause intrauterine death
  - Diseases included in extended neonatal studies

- 320 genes
- 32.749 causal mutations; Preconception GeneProfile® is the panel with the highest number of mutations studied.
How are the genes studied?

Most of the genes are studied by Next-Generation Sequencing (NGS). Some diseases are concurrently studied with another technology, because they cannot be covered with NGS such as Fragile X syndrome (*FMR1* gene with TP-PCR technique), Friedrich ataxia (*FXN* gene with TP-PCR), Duchenne muscular dystrophy (*DMD* gene with MLPA) and Spinal muscular atrophy (*SMN1* and *SMN2* genes with MLPA).

Preconception GeneProfile® is a targeted study that analyses mutations known as pathological based on scientific publications. Not only mutations in coding DNA (exons) are studied but also mutations that have been described in other part of the gene: regulatory regions, introns, exon/intron intermediate regions, flanking regions. This allows the widest coverage possible and ensures maximum capacity of detection.

How to order Preconception GeneProfile®?

Preconception GeneProfile® may be ordered through our online platform. To request your access information, please contact directly with your sales representative or at +34 961 366 150 or comercial@sistemasgenomicos.com.

- Fill up the online request form.
- Send 10-15 ml of blood in EDTA at room temperature.
- Package the sample in order to avoid damages and abrupt temperature changes and send it to the following address:

  **Europe and rest of the world**
  SISTEMAS GENÓMICOS, S.L.
  Ronda G. Marconi, 6
  Parque Tecnológico de Valencia
  46980 Paterna (Valencia) Spain

  **Canada and USA**
  SISTEMAS GENÓMICOS,
  4936 Yonge Street
  Suite #603 (inside UPS facility)
  Toronto, ON M2N 6S3
  Canada

- If you have any questions, do not hesitate in contacting us at +34 961 366 150 or preconception@sistemasgenomicos.com.

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