CGT | Carrier Genetic Test

igenomix
PIONEERS IN **REPRODUCTIVE GENETICS**

Study on more than 6,000 mutations associated with over 600 genetic disorders

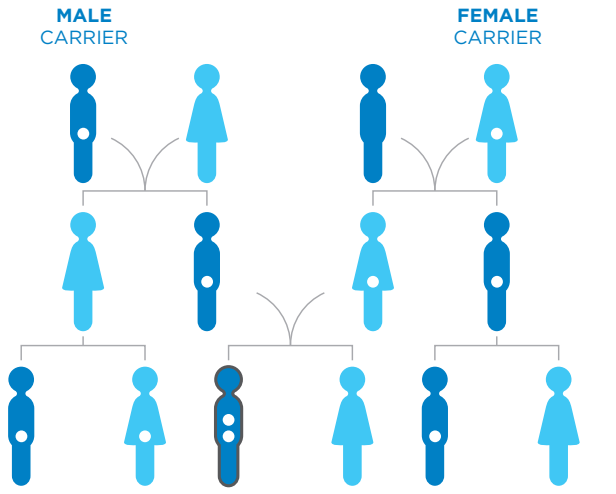
Before pregnancy, this genetic test can determine if a person is a carrier of genetic mutations that could be transmitted to their children.

If both partners in a couple carry a mutation in the same gene, there is an increased risk of having an affected child.

It evaluates disorders such as Cystic Fibrosis, Spinal Muscular Atrophy, Fragile-X Syndrome and Beta Thalassemia, amongst others.

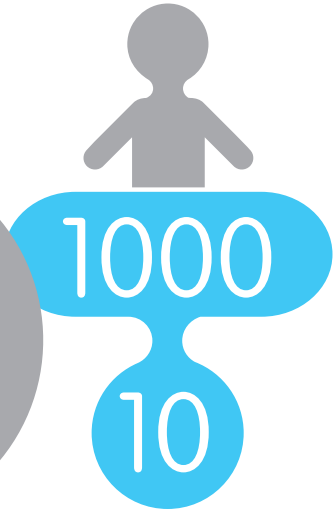
Carrier Genetic Test for single gene disorders by **next generation sequencing (NGS)**

Most carriers of genetic mutations don't have a family history of these disorders



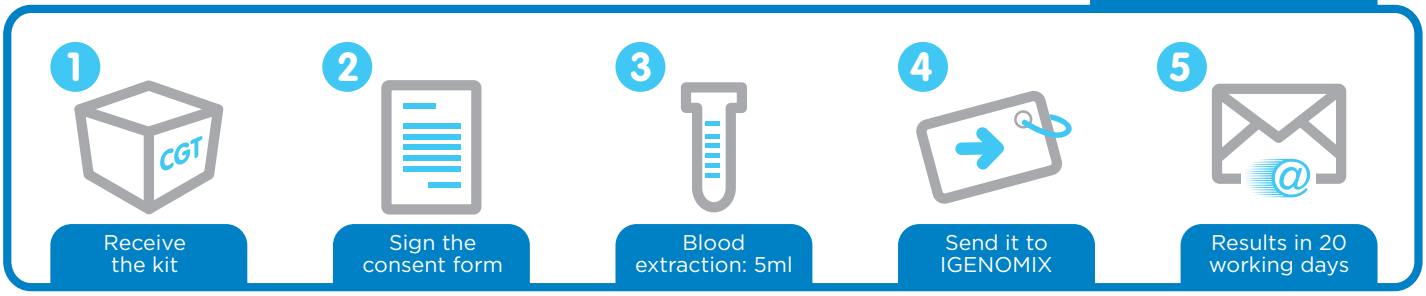
In recessive disorders, mutations are passed unnoticed through generations until two carriers have an affected child.

The estimated global prevalence of these disorders is 10/1000 live births
(WHO data)*



(*) According to the World Health Organization (WHO)
http://www.who.int/genomics/public/geneticdiseases/en/index2.html

SIMPLE AND EASY



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PIONEERS IN **REPRODUCTIVE GENETICS**

STUDY ON MORE THAN 6,000 MUTATIONS ASSOCIATED WITH OVER 600 GENETIC DISORDERS

1 What is a single gene disorder?

- It's a hereditary disorder caused by the presence of a mutation, or mutations, in a particular gene.

Single gene disorders can't be cured, but they can be prevented.

2 What is the test for?

- It identifies couples in which both partners are carriers. In this way it prevents serious genetic disorders in their offspring.

- If both partners carry a mutation in the same gene, they will be at high risk of having an affected child. In these cases, there are options for conceiving healthy children, like Preimplantation Genetic Diagnosis or gamete donation. It's also possible to use prenatal diagnosis after conceiving naturally.

- A **negative result indicates that the person does not carry** any of the mutations studied.

3 What is the turnaround time for the results?

20 working days

4 Why choose Igenomix's CGT?

- It is a clinically validated genetic screening test based on Next-Generation Sequencing (NGS).

  Our different CGT options, tailored to each patient's needs, make it the most complete test available.

5 Application of the test to donor banks

- At Igenomix we offer the test to infertility clinic patients receiving sperm or egg donation. The joint study of both donor and recipient significantly reduces the possibility of having a baby affected by the disorders studied.

METHOD MAIN STAGES OF THE TEST

Exon trapping, library construction, and quality control	Obtaining the complete sequencing data	Filtering and biological evaluation of the variants obtained	Clinical interpretation and results report
EXON & LIBRARY Construction	NEXT GENERATION SEQUENCING	BIO INFORMATICS	CLINICAL INTERPRETATION

Test limitations

- The sensitivity of the test is 98%.
- We only analyze genes on the list available at www.cgt.igenomix.com, reporting results based on knowledge current at the time of testing. Therefore, only genes included in this list will be detected.
- The method is based on analysis by massive parallel sequencing and bioinformatics, studying all the exons of the genes included on our gene list. This includes adjacent intronic regions before or after position +5 and -5. Variants located outside the gene regions studied, such as gene-expression regulatory regions or intronic regions before or after position +5 and -5 are not detected.
- No inversions, deletions, or duplications over 20 nucleotides long that are not referred to in the list available at www.cgt.igenomix.com are studied.
- Germ mosaics (mutations only present in gametes) are not detected by this analysis because the DNA material studied is obtained from a blood sample.
- A negative result for the genes indicated does not exclude the possibility that a de novo mutation may appear in the offspring.