What is the Igenomix Carrier Genetic Test?

The CGT is an important genetic test when planning a family, because **it helps to determine the risk of having a child with a genetic disease.** The test tells us whether the parents carry one or more recessive genetic mutations.

Carriers are usually healthy but when two parents carry a mutation in the same gene they might produce an affected child.

Who is the CGT for?

The test is recommended in the following cases:

- **Before attempting a pregnancy by natural means**
- **Before an assisted reproduction treatment**
- **Before treatment with donor sperm or eggs**

Igenomix®
WITH SCIENCE ON YOUR SIDE

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CGT

Carrier Genetic Test

by Igenomix®

A simple DNA test prior to pregnancy to prevent genetic disorders in the baby

The most advanced way to plan your family

Igenomix®
WITH SCIENCE ON YOUR SIDE

At Igenomix we care about the health of your future baby

Every year many parents are caught off guard by the birth of a baby with some form of genetic disease.

Igenomix has developed an advanced carrier genetic test, prior to pregnancy, that can reveal if a couple is at risk of having a baby with one of these serious illnesses. If the results of the test come back positive, the necessary steps can be taken to favor the birth of a healthy baby.

What are genes?

Each one of our cells contains genetic information, or DNA, organized into basic units, genes. Those that don't function correctly are the ones responsible for genetic disorders.

Anyone can unknowingly carry one or more mutations.
The CGT test allows us to know which genes are altered in each person.

Why do a CGT test?

Generally, parents only realize they are carriers of serious genetic disorders after an affected child is born. Genetic disorders can't be cured, but they can be prevented.

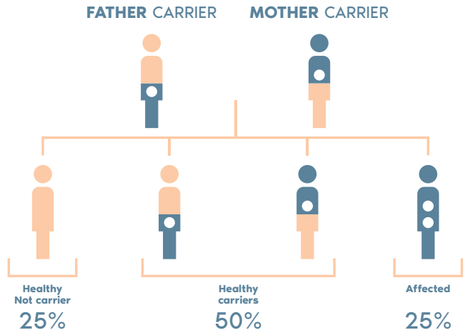
What happens if I'm a carrier?

NOTHING

Being a carrier of a mutation doesn't mean you will develop the illness*.

We are all carriers of certain genetic mutations. Although carriers are healthy people, if both parents have a mutation in the same gene the probability of having a sick child is 25 %.

*autosomal recessive or X-linked disorders (women)



3 CGT testing options

	CGT Basic Carrier Genetic Test	CGT Plus Carrier Genetic Test	CGT Exome Carrier Genetic Test
Type of panel	Recommended by the medical societies	Extended panel (includes CGT Basic)	Whole exome compatible with most carrier platforms in the market
Genes	8	306	1590
Numbers of diseases	8	352	+1600
Estimated carrier rate (%)*	10,6%	54,8%	62,7%
Estimated mean of mutations/individual**	1,06	1,46	2,28
Sample	Blood or saliva		
TAT	20 days		

* In-house data base of 30,000 tests
**Estimated mean of positive individuals

What if both parents test positive?

The recommendation is to consult with a specialist about the options for conceiving a healthy child.

PGT-M can prevent those couples from having a baby with a disease.

Other parents may turn to egg or sperm donation to prevent these illnesses.

The parents can also think about adoption to avoid having a sick child.

(1) Martin et at. Fertil Steril. 2015

What disorders are included?

According to data from the World Health Organization (WHO)(*), the global prevalence of these illnesses is 10 in 1000 newborn infants.

10/1000

There are estimates that, taken together, indicate that these illnesses represent 20% of the causes of infant mortality in developed countries and that they are behind 18% of the interventions in pediatric hospitals (**)

20%

The test covers a wide range of mutations that result in serious genetic illnesses. It includes screening of all the mutations recommended by professional gynecology and genetic associations (***).

See the complete panel of mutations included in the CGT test at www.igenomix.com

THE MOST COMMON MONOGENIC DISORDERS DETECTED WITH THE CGT TEST ARE:	PROPORTION OF CARRIERS
Cystic fibrosis	1 in 25
Spinal Muscular atrophy	1 in 50
Autosomal recessive polycystic kidney disease	1 in 70
Non-syndromic hereditary sensorineural hearing loss	1 in 80
Mucopolysaccharidosis	1 in 80
Sickle-cell anemia	1 in 150
Gaucher's disease	1 in 200
Fragile-X syndrome	1 in 250
Beta thalassemia	1 in 300

(*) According to data from the World Health Organization (WHO) http://www.who.int/genomics/public/geneticdiseases/en/index2.html
(**) Kingsmore S. PLOS Currents Evidence on Genomic Tests. 2012 May 2. Edition 1. doi: 10.1371/4f9877ab8ffa9.
(***)The American College of Medical Genetics (ACMG) and The American Congress of Obstetricians and Gynecologists (ACOG).