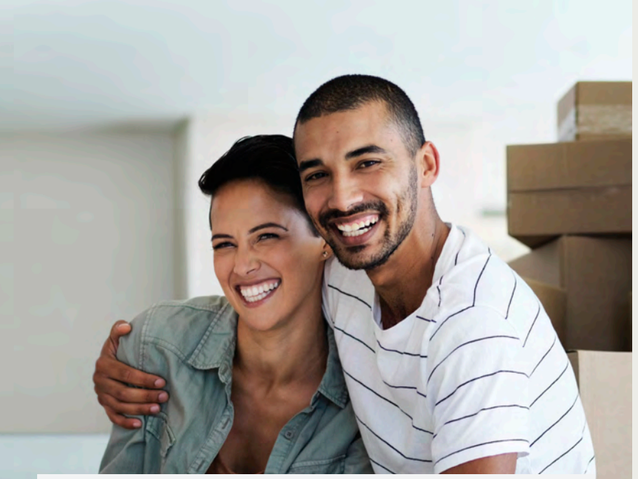


What is the Igenomix Carrier Genetic Test?

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 have 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

www.igenomix.eu

V. 2022

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 children

Every year many healthy parents are touched by the birth of a baby with some type 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 conditions.

What are genes?

Each of our cells contains genetic information or DNA. **Genes are made up of DNA.** A gene is the basic physical and functional unit of **heredity.** Genes analysed by the CGT act as instructions to make molecules called proteins.

Genes can acquire changes or mutations in their sequence, and this may affect the coded protein. Inherited harmful mutations are responsible for genetic disorders.

Anyone can unknowingly carry one or more mutations.
The CGT test lets us know which genes have mutations, in each person.

Why get a CGT test?

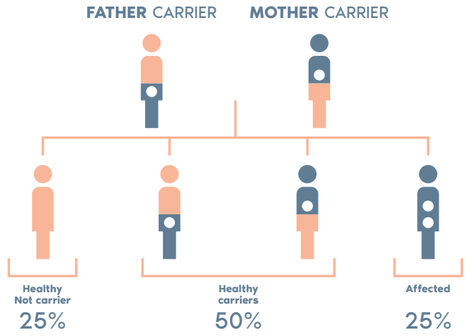
Parents only realize they are carriers* of serious genetic disorders after an affected child is born. Most genetic disorders can't be cured, but they can be prevented.

What happens if I'm a carrier?

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

Most of us are 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)



Our new universal approach to expanded carrier screening using whole exome sequencing

FEATURES	CGT Plus Expanded Panel	CGT Exome Premium Expanded panel
Genes	Male: 470; Female: 536 (include 66 X-linked)	Male 1.989; Female: 2.055 (include 66 X-linked)
Numbers of diseases	>500	>2,200
Estimated carrier rate (%)*	~55%	~67%
Estimated mean of mutations/individual**	1.7	2.7
Sample	Blood or saliva	
TAT	20 working days	

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

What if both parents test positive?

It is recommended to consult with your doctor about options for conceiving a healthy child.

Embryo diagnosis (PGT-M) can prevent from having a baby with a disease.

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

(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 conditions recommended by professional gynecology and genetic Organizations (***).

See the complete panel of mutations included in the CGT test at cgt.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 and Genomics (ACMG) and The American College of Obstetricians and Gynecologists (ACOG).