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 proceding with donor sperm or eggs**

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www.igenomix.com

V. 2024

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

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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 you are 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.** Genes act as instructions to make proteins and are inherited by the mother (egg source) and father (sperm source).

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

Anyone can unknowingly carry one or more mutations.
The CGT test identifies which of your genes have mutations.

Why get a CGT test?

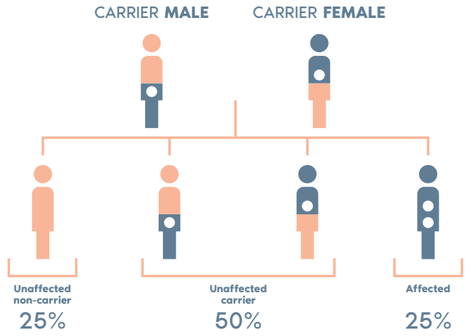
Parents only realize they are carriers* of serious genetic disorders after an affected child is born. Most genetic disorders can't be cured, but they can be prevented.

What happens if I'm a carrier?

Being a carrier means you have one normal copy of a gene and one copy with a mutation.

Most of us are carriers of 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 exome sequencing

FEATURES	CGT Plus — Expanded Panel	CGT Mirror — Flexibility to mirror any carrier screen and match a previously tested individual
Genes	Male: 470 Female: 535 (include 65 X-linked)	Variable
Numbers of diseases	>500	Variable
Estimated carrier rate (%)*	~55%	Variable
Estimated mean of mutations/individual**	1.7	Variable
Sample	Blood or saliva	
TAT	30 working days	

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

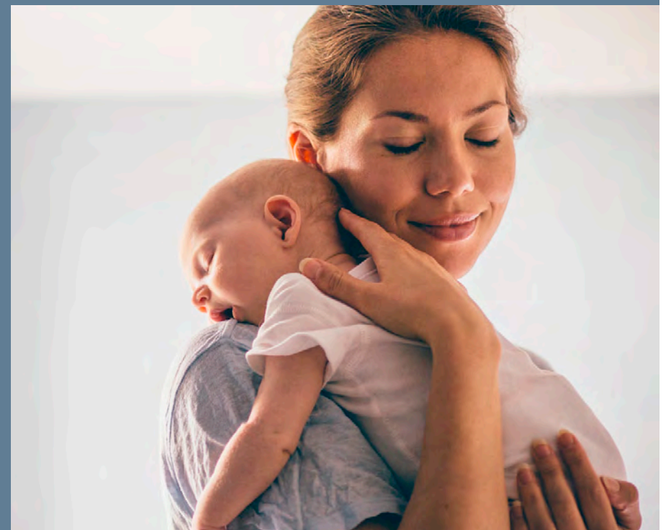
What if both reproductive partners test positive?

It is recommended to consult with your doctor about options for conceiving a healthy child. Igenomix can support you with Genetic Counseling.

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

Some patients may use egg or sperm donation, or choose a different donor, 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 1 in 100 newborn infants.

1/100

Approximately 20% of infant mortality and an estimated 18% of pediatric hospital admissions are caused by these disorders and illnesses. (**)

20% Infant mortality

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 list 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).