What is Igenomix's 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 condition.** The test tells us whether the parents carry one or more recessive genetic variants.

Carriers are usually healthy but when two individuals carry a variant in the same gene they could have an affected child.

Who is CGT for?

The test is recommended in the following cases:

- **Before becoming pregnant**
- **Before beginning assisted reproduction treatment**
- **Before selecting an egg or sperm donor**

Igenomix®
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www.igenomix.ca

V. 2024

CGT

Carrier Genetic Test

by Igenomix®

A simple DNA test prior to pregnancy to prevent genetic conditions in babies

The most advanced way to plan your family

Igenomix®
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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 condition.

Igenomix has developed an advanced carrier screen that can reveal if you are at risk of having a baby with one of these serious conditions, prior to pregnancy.

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 from 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 variants are responsible for genetic disorders.

Anyone can unknowingly carry one or more variants.
The CGT test identifies any genetic variants in your genes that could be passed on to a child.

Why get a CGT test?

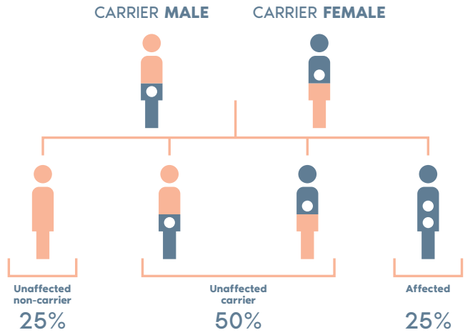
Patients sometimes only realize they are carriers* of a serious genetic condition after an affected child is born. Most genetic disorders can't be cured, but they can be prevented.

What happens if I'm a carrier?

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

Most of us are carriers of several **genetic variants.** Although carriers are usually healthy, if both individuals have a variant in the same gene, the probability of having an affected child is 25%.

*Autosomal recessive or X-linked conditions (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	CGT Sequential Test just the genes your reproductive partner carries
Genes	Female: 539 Male: 474 (include 65 X-linked)	Variable	1-10 single genes (+65 X-linked in female)
Numbers of diseases	>570	Variable	Variable
Estimated carrier rate (%)*	~55%	Variable	Variable
Average # of variants/individual**	1.7	Variable	Variable
Sample	Blood or saliva		
TAT	20 business days - 30 calendar days		

* In-house data base of 30,000 tests
**Estimated mean of positive individuals
***For Patients Using Donor Sperm or Eggs

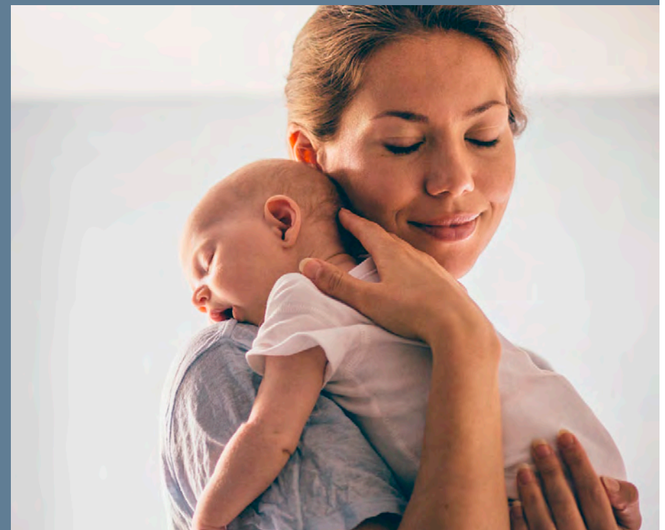
What if both reproductive partners test positive?

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

Embryo testing (PGT-M) can identify which ones did not inherit the condition.

Some patients may use egg or sperm donation, or select a different donor, to reduce their reproductive risk.

(1) Martin et at. Fertil Steril. 2015



What conditions are included?

According to data from the World Health Organization (WHO)(*), the global prevalence of these conditions is 1 in 100 newborn infants.

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

1/100

20% Infant mortality

The test covers a wide range of variants that result in serious genetic conditions. It includes screening of the conditions recommended by professional gynecology and genetic organizations (***).

See the complete list of variants included in the CGT test at cgt-panels.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 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).