UPGRADED

CGT

Carrier Genetic Test

by Igenomix®

A simple DNA test that helps you understand inherited risks before pregnancy

Practical insight to guide your family-planning journey

Igenomix®

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What is the Carrier Genetic Test (CGT)?

CGT is an important test when planning a family because **it helps identify whether you or your partner carry a genetic change** that could be passed on to a child.

The upgraded CGT uses Clinical Exome Sequencing (CES), a focused approach that looks only at genes known to be linked with medical conditions. This makes the test clearer, faster, and more precise

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**

Helping you plan for a healthy start to your family

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?

Our cells contain genetic information or DNA. **Genes are small sections of the DNA** that act as instructions for how our bodies grow and function. Genes are inherited from the mother (egg) and father (sperm).

Genes can acquire changes or mutations in their sequence, which may impact how the gene functions and in some instances, can be harmful. Inherited harmful mutations (variants) are responsible for genetic conditions.

Anyone can unknowingly carry one or more variants.
The Upgraded CGT test uses Clinical Exome Sequencing (CES) to identify important disease related variants that could be passed on to a child.

Why get a CGT test?

Many people don't know they are carriers because they are completely healthy, and most carriers never have symptoms. Most only learn they are carriers after having a child affected by a genetic condition. While most of these conditions cannot be cured, they can be prevented.

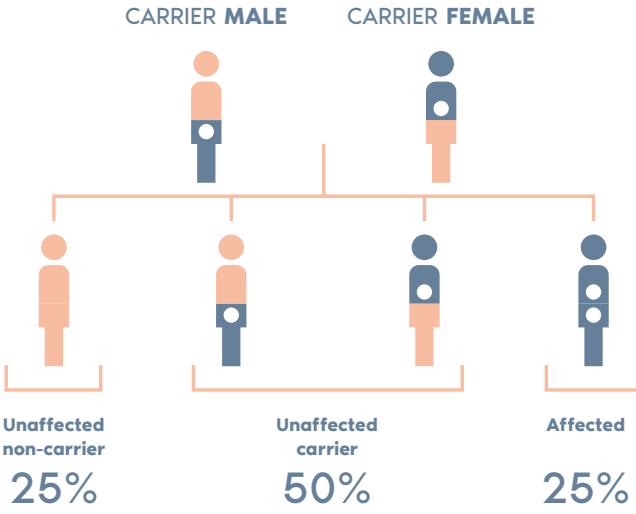
The upgraded CGT offers a clearer focus on genes that matter clinically for reproductive health.

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 Clinical Exome Sequencing

FEATURES
Genes
Numbers of diseases
Estimated carrier rate (%)*
Estimated mean of mutations/individual**
Sample
TAT

AR: autosomal recessive; XL: X-linked. X-linked genes are reported for female samples only.
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 testing (PGT-M) can show which embryos have not inherited 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

CGT Plus	CGT Exome
Expanded Panel	Premium Expanded panel
463 AR 65 XL*	1648 AR 65 XL*
>500	>1,700
~55%	~67%
1.7	2.7
Blood or saliva	
15 working days	

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. (**)

The upgraded CGT covers a wide range of serious genetic conditions recommended by professional organisations, and analyses only the genes known to be linked to health. This CES approach ensures clearer results and a more focused assessment of reproductive risk (***).

See the complete list of genes included in the Upgraded CGT test at igenomix.eu

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)
(****) Carriers identified in the general Caucasian population.

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