What is the Igenomix Carrier Genetic Test?

CGT is a type of genetic test that involves analysing your DNA to identify whether you are a carrier of a genetic condition.

Being a carrier does not generally have any implications for your own health, but you may have an increased chance of having an affected child.

For some conditions, this may be particularly relevant if the child's other biological parent is also a carrier of the same condition.

Is a CGT for you?

The test is suitable for anyone who wants to learn more about their risk of transmitting an inherited condition:

- **Couples who are planning to conceive naturally**
- **Individuals/couples undergoing assisted reproduction**
- **Intended recipients of donor eggs/sperm**
- **Those with a higher risk for recessive conditions**

Igenomix®
WITH SCIENCE ON YOUR SIDE

www.igenomix.co.uk
info.uk@igenomix.com
+44 (0)20 8068 8176

V. 2022

CGT

Carrier Genetic Test

by Igenomix®

A simple DNA test prior to pregnancy to provide information about genetic conditions

Let science help plan your healthy family

Igenomix®
WITH SCIENCE ON YOUR SIDE

At Igenomix, we know how much care about the health of your future child.

Every year many healthy parents give birth to a baby with a genetic condition.

Igenomix has developed an advanced carrier genetic test, for use prior to pregnancy, that can reveal if a couple has an increased chance of having a child with a serious genetic condition.

What are genes?

Each of our cells contain genetic information known as DNA. Genes are short sections of DNA which contain the instructions for our bodies to develop and function. We have two copies of every gene: one copy is inherited from each of our biological parents.

A 'spelling mistake' or **variant** in a gene may affect its function, resulting in a faulty gene. If an individual has one faulty copy of a gene and one working copy, they are known as a **'carrier'** of the genetic condition(s) associated with that particular gene.

More than **80% of people** are estimated to be carriers of **at least one genetic condition** without knowing it.

Why should you have a CGT?

Many parents only realise they are carriers* of serious genetic conditions after an affected child is born. Most genetic conditions can't be cured, but they can be prevented.

2-4% of reproductive couples are at risk of conceiving a child with an autosomal recessive or X-linked genetic condition.[1]

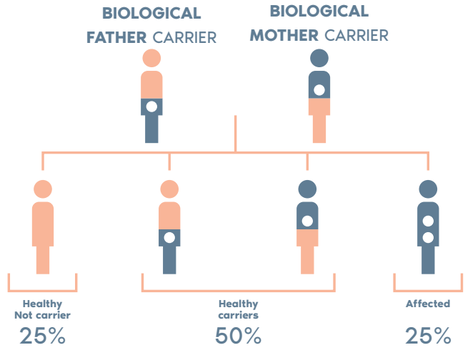
88% of carriers have no known family history of a condition.[2]

What happens if I'm a carrier?

Being a carrier of a genetic condition doesn't mean you will develop the condition.*

Most of us are carriers of one or more genetic conditions. Although carriers are generally healthy, if both parents have a disease-causing variant in the same gene, the probability of having an affected child is 1 in 4 or 25%.

*autosomal recessive or X-linked conditions (biological females)



1 Capalbo et al 2021
2 Archibald et al 2018

Our advanced approach to carrier screening using whole exome sequencing

FEATURES	CGT Plus Expanded Panel	CGT Exome Premium Expanded panel
Genes	Male: 454; Female: 518 (includes 66 X-linked conditions)	Male: 1,993; Female: 2,057 (includes 66 X-linked conditions)
Number of diseases	>500	>2,200
Estimated carrier rate (%)[3]	~55%	~67%
Average number of variants detected per individual[4]	1.7	2.7
Sample type	Blood or saliva	
Turnaround time	20 working days	

3 In-house database of 30,000 tests
4 Estimated average of positive individuals

What if both biological parents are carriers?

We recommend genetic counselling to understand your options:

- Prenatal testing
- Using a donor gamete (sperm or egg)
- Adoption
- Continue with your reproductive plans, knowing your risk

What conditions are included?

The test covers a wide range of genes that are associated with serious genetic conditions. It includes screening of all the conditions recommended by professional gynaecology and genetic organisations.[5]

The complete list of genes included in our CGT panels can be found on our website.

SOME OF THE MOST COMMON MONOGENIC CONDITIONS DETECTED WITH A CGT ARE:	EXAMPLE CARRIERS RISK
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
Sickle cell disease	1 in 150
Gaucher disease	1 in 200
Fragile X syndrome	1 in 250
Beta thalassaemia	1 in 300

5 The American College of Medical Genetics and Genomics (ACMG) and The American College of Obstetricians and Gynecologists (ACOG).