

## How Invitae carrier screening works



### 1. Select your tests

You and your healthcare provider will select the disorders that you should be tested for.



### 2. Provide a sample

Your healthcare provider's office will draw a blood sample or collect a saliva sample and ship it to our laboratory for analysis.



### 3. Get your results

Your results will be ready 10–21 calendar days after Invitae receives your sample (on average); they can be easily accessed through the online portal.



### 4. Make your plan

Discuss your results with one of our genetic counselors, your healthcare provider, or both. Schedule an appointment to speak with a board-certified genetic counselor through our online portal.

## Reproductive options for at-risk couples

If your results show that you are at increased risk of having a child with one of the disorders on the carrier screen, it is important to note that there are reproductive options available to you depending on where you are in your reproductive journey.

These options may include:

- continuing with family planning and pregnancy without any changes
- undergoing in vitro fertilization (IVF) and preimplantation genetic testing (PGT)
- conception through egg, sperm, or embryo donation
- undergoing prenatal testing during pregnancy
- adoption is another great way to build a family

To learn more about carrier screening, visit [www.invitae.com/patients/reproductive-health](http://www.invitae.com/patients/reproductive-health).

### Questions?

Visit [www.invitae.com/contact-us](http://www.invitae.com/contact-us) for a complete list of contact information or call on 1800 961 370 9:00am to 5:00pm Monday to Friday (AEST).



## About us

Invitae is a genetics company whose mission is to bring comprehensive genetic information into mainstream medicine to improve healthcare for billions of people. Invitae testing provides answers to essential health questions—understanding disease risk, guiding a healthy pregnancy, or finding a diagnosis—at high quality, fast turnaround, and low prices.

We strive to make testing affordable and accessible.

## Understanding carrier screening



# What is carrier screening?

When you are pregnant or planning to become pregnant, you want everything to go right. While most babies are born healthy, with every pregnancy there is a small chance of having a baby with a genetic disorder.<sup>1</sup> With Invitae carrier screening you can learn your risk for passing on an inherited genetic disorder to your child.

Carrier screening is a type of genetic test that analyzes your DNA to provide specific information about your child's risk for certain genetic disorders. This information allows you to make informed reproductive choices.

## What does it mean to be a "carrier" of a genetic disorder?

If you are a carrier, it means you have a change (called a variant) in one copy of a gene that increases the risk of your child inheriting the associated disorder. For most tested disorders, both reproductive partners have to be carriers (of the same disorder) for their children to be at increased risk. However, some disorders are X-linked, meaning they're typically passed on from carrier mothers and more commonly affect boys.

- We are all carriers of one or more genetic disorders.
- Carriers are typically healthy and do not have symptoms.
- Carriers often do not have a family history of the disorder.
- Reproductive options are available to patients who learn they are at increased risk of having a child with a genetic disorder.

1. Stevenson RE, et al. Human malformations and related anomalies. 3rd ed. Oxford University Press; 2015.

## Who should consider carrier screening?

Carrier screening can provide important information for people who:

- are currently pregnant or planning a pregnancy
- are at increased risk for a specific disorder based on their ethnicity
- have a family history of a genetic disorder
- plan to donate eggs, sperm, or embryos
- would like additional information about reproductive risks of having a child with a genetic disorder

## What disorders should I be screened for?

You and your healthcare provider can select the tests that are most appropriate for you based on your medical and family history, ethnicity, personal preference, and the guidelines of leading medical organizations like the Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG). Every patient is different, and our panels are customizable to meet your unique needs.

## Invitae carrier screening panel options

- **Core carrier screen:** Includes screening for cystic fibrosis (CFTR), fragile X syndrome (FMR1) and spinal muscular atrophy (SMN1). This panel is appropriate for patients who are interested in the most commonly screened disorders.

Cystic fibrosis (CF)	Spinal muscular atrophy (SMA)
1 in 31 people are carriers	1 in 50 people are carriers
<ul style="list-style-type: none"><li>• CF is characterized by chronic respiratory and digestive problems.</li><li>• With treatment, people with severe CF can live into their 30s and sometimes beyond.</li></ul>	<ul style="list-style-type: none"><li>• SMA is characterized by severe muscle weakness and progressive loss of voluntary muscle control.</li><li>• In severe cases, SMA results in death before 2 years of age.</li></ul>

## Fragile X syndrome

- 1 in 178 women are carriers
- Fragile X is the most common cause of inherited intellectual disability.
  - Life expectancy for people with fragile X is generally normal.

- **Broad carrier screen (more than 100 genes):** This panel is appropriate for patients who want to be screened for disorders with severe presentation that are prevalent across different ethnicities.
- **Comprehensive carrier screen (more than 550 genes):** This panel is appropriate for patients of all ethnicities who want a more comprehensive carrier screen.

## What do my results mean?

### POSITIVE

A positive result means that a disease-causing variant was found, and you are a carrier of one or more of the disorders tested. Being a carrier typically does not affect your own health; however, it does mean that there is an increased risk of having a child with that disorder. The next step is usually to test your partner. Our genetic counselors are available to discuss your specific risks and concerns.

### NEGATIVE

A negative result means that no disease-causing variants were identified for any of the disorders tested. A negative test provides reassurance because the chance of you having a child with any of the tested disorders is reduced. However, no test can detect all carriers, so there is still a small chance, called a residual risk, of being a carrier.

## Ready to help

Planning to start a family involves many decisions. Have questions? Genetic counselors are available to help you understand your results and discuss your next steps. Genetic counselors are healthcare providers specifically trained in medical genetics; they are experts at helping people understand what their genetic test results mean.

Scan here to learn more  
about carrier screening  
[invitae.com/carrier-screening](https://invitae.com/carrier-screening)

