Hereditary Cancer
PATIENT GUIDE

Hereditary Cancer Testing

Patient Guide

A Guide To Genetic Testing
For Hereditary Cancer

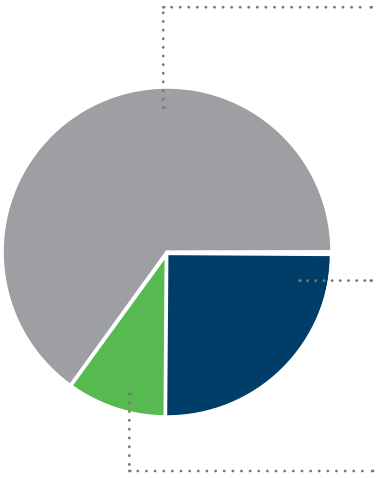
Ambry Genetics®

Understanding The Basics

Our genes are like our bodies' instructions. Some of our genes have roles in protecting against cancer. A mutation is a harmful difference in a gene that makes it not function as well. When someone is born with a mutation in a gene that should help protect against cancer, that person is at increased risk for cancer. **This is called having a hereditary risk for cancer.**

While many people have a history of cancer in their family, only **5-10%** of cancer is hereditary.[1]

CANCER FALLS INTO 1 OF 3 CATEGORIES



SPORADIC CANCER

Cancers that occur by chance or due to environmental causes, like skin cancer and sun exposure. Approximately 75-80% of cancers are sporadic.[2]

FAMILIAL CANCER

A pattern of the same or related cancers in the family that may be due to a combination of shared genetic and lifestyle factors. Approximately 20% of cancers are familial.[2]

HEREDITARY CANCER

A pattern of the same or related cancers within a family due to inherited mutations that can be passed from parent to child. Approximately 5-10% of cancers are hereditary.[2]

Should You Consider Hereditary Cancer Testing?

Talk to your healthcare provider if you answer “yes” to any of the following:

1. Have you/your close family members* been diagnosed with cancer at a young age (<50 years old)?
2. Have you/your close family members* been diagnosed with more than one cancer?
3. Have you/your close family members* been diagnosed with cancers/tumors that you have been told are usually rare, such as ovarian or pancreatic cancer?
4. Have multiple people on the same side of your family had the same or related types of cancer?
5. Have any of your close family members* been found to have a cancer gene mutation?

Your healthcare provider may identify other reasons why you could consider genetic testing.

** “Close family members” refers to blood relatives, such as brothers/sisters/parents/grandparents/aunts/uncles/cousins*

How Genetic Testing Can Impact You

Your test results may help your healthcare provider fine-tune your cancer screening plan, including the type, timing (age) of your initial screening, and its frequency.

Examples of cancer screening include mammogram, breast MRI, colonoscopy, prostate exam, dermatology (skin) exam, or other screenings appropriate for your specific cancer risk.

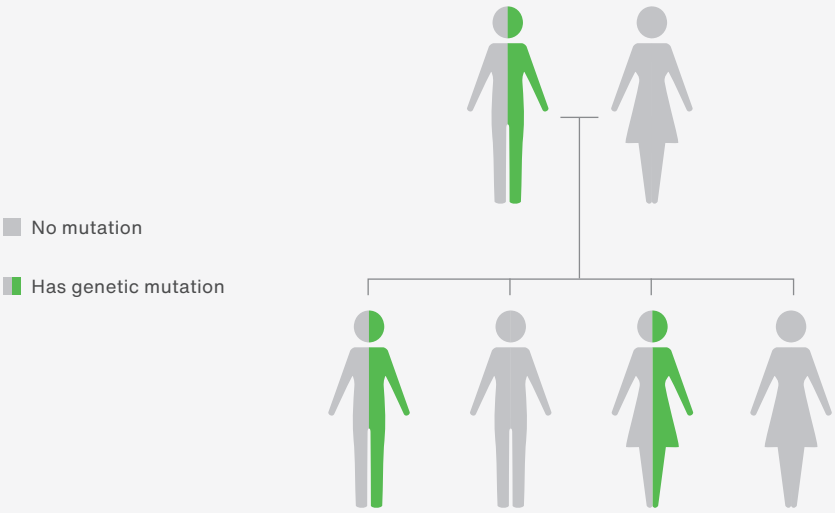
Based on your results, your healthcare team may review options to reduce cancer risk, including surgery (called “prophylactic surgery”) and the use of medications (chemoprevention).

Examples include prophylactic mastectomy (removing one or both breasts before a cancer occurs) or prophylactic salpingo-oophorectomy (removing the ovaries and fallopian tubes before a cancer occurs).

Your doctor can also identify and discuss other personalized medical management options that might be appropriate based on your genetic test results.

For Your Family

If you test positive for a genetic mutation, each of your first-degree relatives (parents, brothers, sisters, children) has a 1 in 2 or 50/50 chance of also having the same mutation. Genetic counseling and/or genetic testing may be beneficial for your family members to discuss their risks and if they should make changes to medical management.



- Men and women have the same chance of inheriting a mutation, but their chance of developing cancer may be different.

Testing With Ambry Genetics

As the first lab to offer clinical hereditary cancer panel testing in 2012, Ambry Genetics has robust experience in hereditary cancer testing. We are also committed to using best-in-class technology so that you and your healthcare team can confidently make decisions about your health.

Ambry Genetics offers a menu of several different hereditary cancer tests. Each test includes analysis of different genes. Your healthcare provider can discuss the available options and recommend the best genetic test for you. Their recommendation may be based on factors like your medical history, family history, and personal preferences.

The genetic testing recommended for you today includes one or more of the tests below.

- ☐ BrainTumorNext®
- ☐ *BRCA*1/*BRCA*2
- ☐ BRCANext®
- ☐ BRCANext-*Expanded*®
- ☐ BRCAplus®
- ☐ CancerNext®
- ☐ CancerNext-*Expanded*®
- ☐ ColoNext®
- ☐ CustomNext-*Cancer*®
- ☐ Familial Adenomatous Polyposis
- ☐ Lynch Syndrome
- ☐ MelanomaNext®
- ☐ *MUTYH*-associated Polyposis
- ☐ PancNext
- ☐ PancNext plus Pancreatitis
- ☐ PGLNext®
- ☐ ProstateNext®
- ☐ RenalNext®
- ☐ Single Gene Test: _____
- ☐ Other: _____

VISIT OUR WEBSITE

See updated information on which genes are included on the test your healthcare provider selected above: ambrygen.com/patients/cancer

How Does Genetic Testing Work?

1. Talk to your healthcare provider about your personal and family history of cancer and options for genetic testing.

2. Submit your blood or saliva samples through your healthcare provider, who will ship it to our laboratory for analysis.

3. Your doctor will notify you once your test results are available.

4. Discuss your test results with your healthcare provider, and plan your health management. We can also connect you with a genetic counselor at no additional cost if your results indicate a positive (called pathogenic or likely pathogenic) or an inconclusive (called VUS) result.

Affordable Testing, Personalized Support

At Ambry, we believe that each person should be treated as an individual when it comes to genetic testing. This remains true as it pertains to the cost associated with testing. We understand that results are used to make life-saving medical decisions, and we are committed to reducing financial barriers to care.

- Many insurance plans cover genetic testing, and Ambry is contracted with the majority of U.S. health plans.
- Your out-of-pocket cost may vary based on your individual health insurance plan.
- We offer competitive cash pricing when insurance is not an option.
- Financial assistance is available through Ambry's Patient Assistance Program.
- A team of dedicated specialists is available to answer any questions you may have. Call or email our Billing Support team at +1 949-900-5795 or billing@ambrygen.com with any questions.

Accessible Counseling

For patients with a positive or VUS result, complimentary genetic counseling sessions are available via a third-party organization with board certified genetic counselors. They provide counseling services via telehealth.

No-Cost Family Testing

The Ambry Genetics Familial Single Site Analysis (SSA) program provides free testing to all close family members of a patient who has received a positive result after completing full gene or panel testing at Ambry. The family members must be tested within 90 days of the original Ambry report date.

Possible Genetic Test Results

+ POSITIVE

A harmful variant (genetic difference also known as mutation or pathogenic variant) was found in at least one of your genes tested.

Detection of a cancer-related gene mutation means that you are at increased risk for cancer compared to the general population, meaning those at average risk. If you have already developed cancer, this mutation may explain your history. You may be at increased risk for other cancers, too.

Based on your results, genetic testing for certain family members is often recommended.

− NEGATIVE

No genetic variants or differences were found in any of your genes tested.

While your genetic test results were negative, personal and family history may also be a strong sign of cancer risk(s) and may inform your medical management.

Talk to your healthcare provider to find out if genetic testing should be considered for your family members.

C CARRIER: PATHOGENIC OR LIKELY PATHOGENIC MUTATION DETECTED

A harmful variant (or mutation) in ONE copy of a gene was found, and you are a 'carrier' of a hereditary condition. Our genes come in pairs. Some hereditary cancer syndromes are caused by having harmful variants in BOTH copies of the same gene. Carriers usually do not have the condition because symptoms typically only occur when mutations occur in BOTH gene copies.

Detection of ONE gene mutation may not increase your personal risk of cancer or other disease, but being a 'carrier' linked with a hereditary cancer condition could be important for family planning reasons. If two carriers of the same condition have children together, there is a ¼ or 25% chance in each pregnancy to have a child with that condition. Your genetic test results are also still important for family members.

Talk to your healthcare provider to find out if genetic testing should be considered for your family members.

? VARIANT OF UNKNOWN SIGNIFICANCE (VUS)

At least one genetic variant or difference was found, but it is unclear if this difference causes an increased risk for cancer.

While your genetic test results were inconclusive and do not change your medical management, personal and family history may also be a strong indicator of cancer risk(s) and may inform your care.

Talk to your healthcare provider to find out if genetic testing should be considered for your family members.

It is possible to have a combination of positive and VUS results, since multiple genes are tested.

Resources For You

Ambry Genetics' Patient Education Website

ambrygen.com/patient

American Cancer Society

cancer.org

FORCE: Facing Hereditary Cancer Empowered

facingourrisk.org

Genetic Information Nondiscrimination Act

ginahelp.org

National Society of Genetic Counselors

aboutgeneticcounselors.org

National Cancer Institute

cancer.gov

CancerCare

cancercare.org

American Society of Clinical Oncology

cancer.net

FIND A GENETIC COUNSELOR

National Society of Genetic Counselors

nsgc.org

Canadian Association of Genetic Counsellors

cagc-accg.ca

Frequently Asked Questions

1 HOW IS GENETIC TESTING PERFORMED AND HOW LONG DOES IT TAKE?

Genetic testing requires a blood or saliva sample, which is collected using a special kit that is shipped overnight to Ambry Genetics by your healthcare provider. The testing, which looks for mutations that cause an increased risk for cancer, takes less than three weeks to complete, and results are sent to your healthcare provider.

2 WHAT WILL HAPPEN WHEN MY RESULTS ARE READY?

Your healthcare provider will receive your results; they will not be sent directly to you. Every healthcare provider may have a different method and timeframe for reviewing your results with you, so it is important to discuss this process with them when your test is performed. Your healthcare provider will discuss recommended next steps based on your test results.

3 WILL MY GENETIC TEST RESULTS AFFECT MY INSURANCE COVERAGE?

In the U.S., the Genetic Information Nondiscrimination Act (2008) prohibits discrimination by most health insurance companies and employers, based on genetic information. Depending on where you live in the world, you may have different (or fewer) laws in this area. Visit ginahelp.org to learn more.

4 HOW WILL MY TEST RESULTS BE PROTECTED?

We are required by law to maintain the confidentiality of your protected health information in accordance with the Health Insurance Portability and Accountability Act (HIPAA). Visit HHS.gov to learn more.

5 SHOULD I TELL MY FAMILY MEMBERS ABOUT MY GENETIC TEST RESULTS?

It is important to share your genetic test results with your family members because the results may provide additional information for your family about their own cancer risks and management options. If you feel unsure about how to approach the subject, your healthcare provider may be able to offer some advice.

6 WILL GENETIC TESTING BE COVERED BY MY INSURANCE?

Many insurance plans cover genetic testing, and Ambry Genetics is contracted with the majority of U.S. health plans. Your out-of-pocket cost may vary based on your individual plan. A team of dedicated specialists is available to help you get access to the genetic testing you need, and provide further details about our payment options. Please call or email our Billing department at +1.949.900.5795 or billing@ ambrygen.com with any questions. Visit ambrygen.com/patientbilling for more information.

7 WHAT IS AN EXPLANATION OF BENEFITS (EOB)?

An EOB is a statement of benefits provided by your insurance company. This is not a bill, but it will outline the expenses your insurance company will cover for the medical care you received. If you have any questions regarding your EOB, you can contact your insurance company directly or connect with a Billing specialist from Ambry. Please note that the processing of claims by insurance companies may take a few weeks or even a couple of months.

STILL HAVE QUESTIONS?

Talk to your healthcare provider or visit our website: ambrygen.com

One Enterprise, Aliso Viejo, CA 92656 USA Toll Free +1.866.262.7943 Fax +1.949.900.5501 **ambrygen.com**

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References

1. Anon, Family Cancer Syndromes. American Cancer Society. Available at: https://www.cancer.org/cancer/cancer-causes/genetics/family-cancer-syndromes.html [Accessed March 22, 2022]

2. Review of Cancer Genetics. Retrieved March 29, 2022, from https://www.cooperhealth.org/sites/default/files/pdfs/Review_of_Cancer_Genetics.pdf.