

CancerNext-Expanded®

PATIENT
GUIDE

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Patient Guide

A Guide To Genetic Testing
For Hereditary Cancer



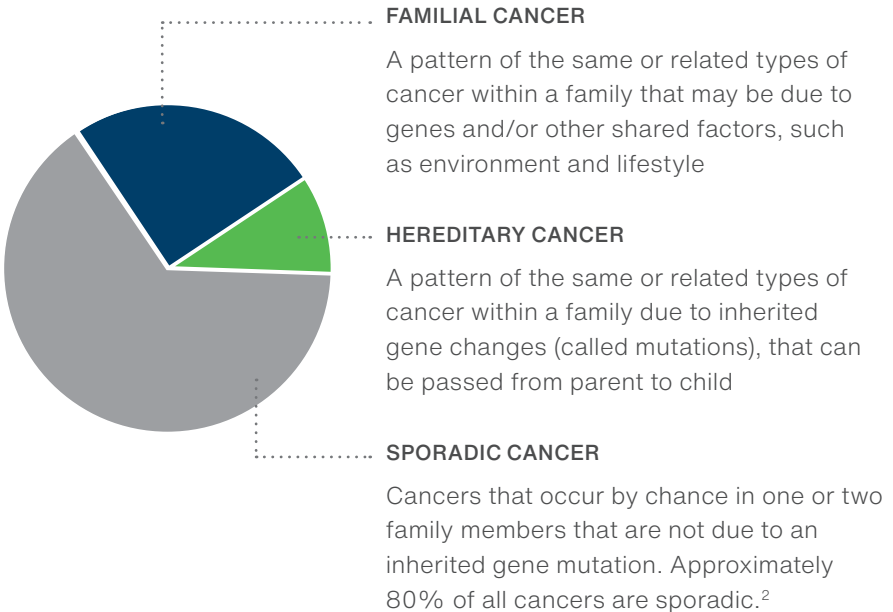
Ambry Genetics®

Understanding The Basics

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



Genes and Associated Cancers

CancerNext-Expanded is a genetic test that includes 77 genes that are linked to an increased lifetime risk for one or more cancers and/or tumor types, including breast, ovarian, uterine, colorectal, kidney, prostate, brain, and others. The chart below shows the types of cancers associated with each gene.

GENE(S)	BREAST	OVARIAN	COLORECTAL	UTERINE	PANCREATIC	PROSTATE	STOMACH	KIDNEY	ENDOCRINE*	BRAIN OR OTHER NERVOUS SYSTEM	MELANOMA**	OTHER
AIP										✓		
ALK										✓		✓
APC			✓		✓		✓		✓	✓		✓
ATM	✓				✓	✓						
AXIN2			✓									
BAP1								✓			✓	✓
BARD1	✓											
BLM	✓		✓									
BRCA1	✓	✓			✓	✓						
BRCA2	✓	✓			✓	✓					✓	
BRIP1	✓	✓										
BMPR1A, SMAD4			✓				✓					
CDC73									✓			✓
CDH1	✓						✓					
CDK4											✓	
CDKN1B									✓	✓		✓
CDKN2A					✓					✓	✓	
CHEK2	✓		✓			✓						✓
CTNNA1							✓					

* Endocrine indicates at least one of the following: paraganglioma (a rare tumor can that arise in certain nerve cells) , pheochromocytoma (a rare tumor of adrenal gland tissue that can impact heart rate, metabolism, and blood pressure), thyroid cancer, carcinoid tumors (slow-growing cancerous tumors that can arise in several places throughout the body), pancreatic neuroendocrine tumors, and/or adrenal tumors (cancerous or noncancerous growths on the adrenal glands)

**Melanoma: a form of skin cancer which can also occur in the eyes (ocular melanoma) and other parts of the body

GENE(S)	BREAST	OVARIAN	COLORECTAL	UTERINE	PANCREATIC	PROSTATE	STOMACH	KIDNEY	ENDOCRINE*	BRAIN OR OTHER NERVOUS SYSTEM	MELANOMA**	OTHER
DICER1		✓								✓		✓
EGFR												✓
EGLN1									✓			
FANCC	✓											
FH								✓	✓			✓
FLCN								✓				
GALNT12			✓									
GREM1			✓									
HOXB13						✓						
KIF1B									✓			
KIT												✓
LZTR1										✓		
MAX									✓			
MEN1									✓	✓		✓
MET										✓		
MITF										✓		
MLH1, MSH2, MSH3, MSH6, PMS2, EPCAM		✓	✓	✓	✓	✓	✓	✓		✓		✓
MUTYH	✓		✓									
NBN												✓
NF1	✓								✓	✓		✓
NF2									✓	✓		✓
NTHL1			✓									
PALB2	✓	✓			✓	✓						

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PDGFRA												✓
PHOX2B										✓		✓
POLD1, POLE			✓									
POT1										✓	✓	
PRKAR1A									✓	✓		✓
PTCH1										✓		✓
PTEN	✓		✓	✓				✓	✓	✓	✓	✓
RAD51C	✓	✓										
RAD51D	✓	✓				✓						
RB1											✓	✓
RECQL	✓											
RET									✓			
SDHA, SDHAF2, SDHB, SDHC, SDHD								✓	✓			
SMARCA4		✓								✓		✓
SMARCB1								✓		✓		✓
SMARCE1										✓		
STK11	✓	✓	✓		✓		✓					✓
SUFU										✓		✓
TMEM127									✓			
TP53	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
TSC1, TSC2								✓		✓		✓
VHL								✓	✓	✓		✓
XRCC2	✓											

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How Genetic Testing Can Impact You and Your Family

FOR 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 indicated for your specific cancer risk.



Based on your results, your healthcare provider may review possible cancer prevention options with you, such as preventive, or prophylactic, surgery that can reduce the risk for certain cancers.

Examples include prophylactic mastectomy (removing one or both breasts before a cancer occurs) or prophylactic 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.

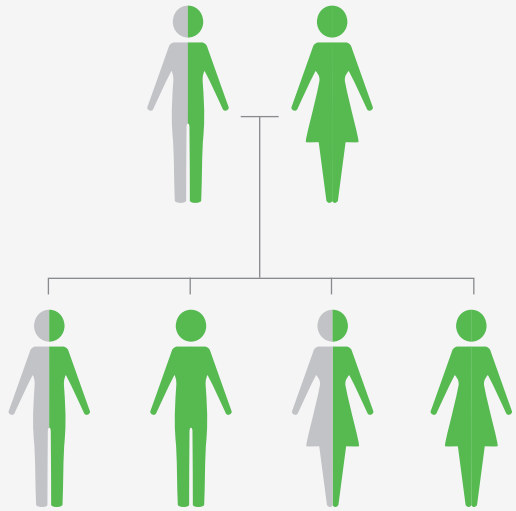
AMBRY GENETICS OFFERS NO-COST TESTING TO CLOSE RELATIVES

This is available for the specific genetic mutation identified in the first family member tested at Ambry within 90 days of the original report date.

FOR YOUR FAMILY MEMBERS:

If you test positive for a genetic mutation, your close family members (like your parents, brothers, sisters and children) have a 50/50 random chance of also having the same mutation.

-  Has genetic mutation
-  No mutation



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

Possible Genetic Test Results

POSITIVE

A mutation was found in at least one of your genes tested

Detection of a cancer-related gene mutation could explain that your cancer diagnosis is hereditary or be a warning that you are at increased risk compared to others.

Based on your results, genetic testing for certain family members may be recommended.

NEGATIVE

No genetic mutations were found in any of your genes tested

While your genetic test results were negative, personal and family history may also be a strong indicator 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.

VARIANT OF UNKNOWN SIGNIFICANCE (VUS)

At least one genetic change was found, but it is unclear if this change 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

CancerCare

cancercare.org

American Cancer Society

cancer.org

Genetic Information
Nondiscrimination Act

ginahelp.org

American Society of Clinical
Oncology

cancer.net

National Cancer Institute

cancer.gov



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 time frame 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 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 results with your family members, because they may provide additional information 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)?

Your insurance company sends you an EOB to explain any services paid on your behalf. You can contact us directly to speak with a Billing specialist with any questions or concerns about Ambry Genetics genetic testing that appears on your EOB. It is important to remember that insurance companies can take several weeks or even a couple of months to process claims.

STILL HAVE QUESTIONS?

Talk to your healthcare provider or visit our website: 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. (n.d.). Review of Cancer Genetics. Retrieved March 29, 2022, from https://www.cooperhealth.org/sites/default/files/pdfs/Review_of_Cancer_Genetics.pdf.