Hereditary Breast Cancer

Breast cancer is the most common cancer in women. One in eight women will develop breast cancer in their lifetime. About 5-10% of these cases are inherited. Hereditary breast cancer (HBC) occurs when a person has a pathogenic (disease-casing) variant that increases the risk of cells becoming cancerous. The variant can be passed through generations in a family. Women and men with HBC have a higher risk of developing breast, and sometimes other cancers or health problems, as well.

WHAT CAUSES HBC?

HBC is most commonly inherited in an autosomal dominant way. For dominant conditions, having only one copy of a pathogenic variant is enough to cause symptoms. Less commonly, HBC can be inherited in an autosomal recessive way. For recessive conditions, both copies of a person's gene must have a variant to increase the risk for cancer. Importantly, not everyone who has a pathogenic-variant in a cancer-risk gene will develop cancer, but their chances are higher.

WHAT ARE HBC SYNDROMES?

HBC syndromes are conditions that impact other areas of an individual's body in addition to increasing their risk for breast cancer. Some examples include:

Cowden Syndrome (CS) - People with Cowden syndrome often have large heads (macrocephaly) and are at an increased risk for different types of benign and malignant tumors, including endometrial, thyroid, and breast cancer. The lifetime risk for breast cancer in women with Cowden syndrome can be as high as 85%.

Hereditary Breast and Ovarian Cancer (HBOC) - *BRCA1*- and *BRCA2*- associated hereditary breast and ovarian cancer causes an increased risk for several cancers. The highest risk elevations are for breast cancer, in both men and women, and for ovarian cancer. Additional cancer risks include melanoma, pancreatic, and prostate cancers.

WHO IS THIS TEST FOR?

These panels may be appropriate for anyone with a personal or family history of breast cancer. Testing is strongly recommended if you or a member of your family has been diagnosed with breast, ovarian, or colorectal cancer before the age of 50, or if there have been multiple cases of cancer in a single individual. It is also recommended that people of Ashkenazi Jewish ancestry, as their risk factors are higher.

4978 Santa Anita Ave, Suite 205, Temple City, CA 91780 | www.FulgentGenetics.com | info@fulgentgenetics.com | **P** +1 626.350.0537 | **F** +1 626.454.1667
 D# FLY-Hereditary Breast Cancer-2023-V1

BENEFITS OF GENETIC TESTING

- Establish or confirm a diagnosis
- Identify risk for other cancers and health conditions
- Assist in modifying lifestyle changes, including diet and exercise
- Result in more personalized treatment, which may include routine screening tests, pharmacological treatments, and prophylactic surgeries
- Inform family members of their own risk factors

TEST OPTIONS

Visit Fulgent website for most up-to-date list

Breast Cancer Focus Panel

13 genes tested: *ATM, BARD1, BRCA1, BRCA2, CDH1, PTEN, STK11, TP53, CHEK2, PALB2, NF1, RAD51C, RAD51D*

Breast Cancer Comprehensive Panel

36 genes tested: *ABRAXAS1, AKT1, ATM, BARD1, BLM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, DICER1, EPCAM, FANCC, FANCM, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PIK3CA, PMS2, PTEN, RAD50, RAD51C, RAD51D, RECQL, SDHB, SDHD, SMARCA4, STK11, TP53, XRCC2*

TEST SPECIFICATIONS

Sample Requirements (kits available upon request):

- Blood, two 4-mL EDTA tubes lavender top (preferred)
- Extracted DNA, 3 µg in TE buffer
- Buccal swab or saliva

Coverage* ~99% at 50x

Turnaround Time** 2-3 weeks

* Full gene sequencing and deletion/duplication, including detection of MSH2 inversion for select panels, BRCA2 Alu variant, and PMS2 (including exons 1-5 and 12-15), as well as sequencing of noncoding regions for selected genes. Please contact us for more details.

**Rush order available upon request

WHERE CAN I LEARN MORE?

Bright Pink - brightpink.org
FORCE (Facing Our Risk of Cancer Empowered) - facingourrisk.org/index.php
PTEN Hamartoma Tumor Syndrome Foundation (Cowden Syndrome) - ptenfoundation.org

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A Patient's Guide to Genetic Testing

What does a genetic test check for?

Genetic testing checks the order of one's DNA sequence (coded by the letters A, T, G, C) in specific genes linked to genetic conditions. Letters that were added, missing, or changed, are known as variants and can sometimes be harmful to one's health, increasing the risk for a genetic condition.

What are the potential results?

There are three possible results from genetic testing:

Positive

A pathogenic/likely pathogenic variant is detected in one's DNA. This type of variant is known to increase one's risk of a genetic condition. Identifying the specific gene involved can help confirm a diagnosis, inform screening and management, and reveal risk factors for an individual and/or their family.

Negative

No variation known to be associated with a genetic condition was detected in one's DNA. While a result may not show an increased risk for the condition(s) tested for, one can still be at risk for disease, especially if there is a family history.

Variant of Uncertain Significance (VUS)

A variant was detected in one's DNA, however, not enough information is known about this variant to determine whether or not it is known cause the condition(s) tested for. More research is needed to better understand this variant.

What about family members?

Children, siblings, and parents of individuals who have a variant(s) identified in genetic testing could carry the same variant(s) and benefit from testing. Regardless of whether or not a variant was identified, individuals can still be at an increased risk for a genetic condition, especially with a family history.

Do genetic test results affect health insurance or employment?

No, the Genetic Information Nondiscrimination Act (GINA) was signed into law in 2008.
It protects individuals from discrimination by an employer or a health insurance company based on genetic testing results and genetic information. GINA does not protect against life and disability insurance discrimination. For more information on GINA, go to www.ginahelp.org.

Where can I learn more?

Medline Plus/Genetics Home Reference - medlineplus.gov/genetics/understanding
National Society of Genetic Counselors - nsgc.org
Fulgent Genetics - fulgentgenetics.com/products/carrierscreening/learning.html

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