

# Hereditary Ovarian Cancer

Ovarian cancer occurs when the cells in the ovaries grow abnormally into a malignant tumor. Approximately 24% of ovarian cancer cases are inherited while most are not. Hereditary ovarian cancer (HOC) occurs when a person has a pathogenic (disease-causing) variant that increases the risk of cells turning cancerous. Women with the variant have a higher risk of developing ovarian cancer, and often



## WHAT CAUSES HOC?

HOC is most commonly inherited in an autosomal dominant way. For dominant conditions, having only one copy of a variant is enough to cause symptoms. Less commonly, HOC 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 HOC SYNDROMES?

Hereditary ovarian cancer syndromes are conditions that impact other areas of an individual's body in addition to increasing their risk for ovarian cancer. Two examples of dominant HOC syndromes are listed

**Lynch Syndrome** - Also known as Hereditary Non-Polyposis Colorectal Cancer (HNPCC), Lynch syndrome accounts for up to 15% of HOC cases. The condition is caused by a variant in a gene whose job is to repair the DNA damage that normally occurs as cells grow and divide. Because of the high risk for cancer, people with Lynch syndrome require increased screening earlier than those without the condition.

**Li-Fraumeni syndrome (LFS)** - People with LFS have up to a 78% lifetime risk for cancer. Many different types of cancer can occur in LFS, and more than one kind can arise in a single person. It is important to diagnose LFS, because exposure to radiation therapy raises the cancer risk even further.

## WHO IS THIS TEST FOR?

These panels may be appropriate for anyone with a personal or family history of ovarian cancer. Testing is strongly recommended if you or a member of your family has been diagnosed with colorectal or endometrial, breast, 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 for people of Ashkenazi Jewish ancestry.

## 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

### Ovarian Cancer Focus Panel

14 genes tested: *BRCA1, BRCA2, BRIP1, EPCAM, MLH1, MSH2, MSH6, MUTYH, PALB2, PMS2, RAD51C, RAD51D, STK11, TP53*

### Ovarian Cancer Comprehensive Panel

20 genes tested: *ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, EPCAM, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, RAD51C, RAD51D, SMARCA4, STK11, TP53*

## 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](http://brightpink.org)

**FORCE (Facing Our Risk of Cancer Empowered)** - [facingourrisk.org/index.php](http://facingourrisk.org/index.php)

**CCARE Lynch Syndrome** - [fightlynch.org](http://fightlynch.org)

**Li-Fraumeni Syndrome Association** - [lfsassociation.org](http://lfsassociation.org)

# 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](http://www.ginahelp.org).

## Where can I learn more?

**Medline Plus/Genetics Home Reference** - [medlineplus.gov/genetics/understanding](http://medlineplus.gov/genetics/understanding)

**National Society of Genetic Counselors** - [nsgc.org](http://nsgc.org)

**Fulgent Genetics** - [fulgentgenetics.com/products/carrierscreening/learning.html](http://fulgentgenetics.com/products/carrierscreening/learning.html)