Genetic Testing

for Identifying Hereditary Cancer Risks



Inherited Mutations

Most cancers occur by chance, but sometimes a person carries a change in a gene (an inherited mutation) that increases their chances of developing cancer. One example of a genetic mutation is BRCA 1 or BRCA 2.

It is estimated that up to 25% of ovarian cancers and up to 10% of uterine cancers are hereditary and related to an inherited mutation. In gynecologic cancer, genetic testing results can help personalize your cancer treatment.



What Is Genetic Testing?

- A Blood TestOR
- A Saliva Test
- Genetic testing checks if you carry a genetic mutation passed down to you from a parent, which is associated
- **Test** with increased risk of ovarian and other cancers.

VS

- Genetic Testing
- Tests the cells you were born with
- Looks for genetic changes that are present in every cell in the body

Biomarker Testing

- Tests your cancer
- Looks for changes that are only present in the cancer

Why is it important?

For Your Cancer Treatment

- In ovarian cancer, genetic testing results may determine if your clinician recommends maintenance therapy after chemotherapy.
- In endometrial or uterine cancer, your genetic testing results may change what treatment your clinician recommends for advanced-stage cancer (when cancer has spread beyond the uterus) or recurrent cancer (when cancer comes back).

For Your Health And Family

- If your testing identifies an inherited mutation that is a genetic cause of your cancer, it is recommended that your firstdegree relatives (parents, children, siblings) also be tested for the same gene.
- Knowing if you have an inherited mutation is important because your family members will have options to prevent cancer. There are preventative options like enhanced screening or a risk-reducing surgery for both male and female family members who have the same inherited mutation.

What Is Genetic Counseling?

- Genetic counseling is a process during which a genetic counselor or other appropriate medical professional will ask a series of questions about you and your family's cancer history as well as your heritage. This will help tailor what genetic tests they recommend for you and your family.
- Genetic counseling may happen before or after genetic testing.



If you have concerns that you may have an inherited risk for developing the disease, OCRA offers free genetic testing to eligible candidates. If you have questions about genetic testing or gynecologic cancer, call OCRA's patient support line at 212-268-1002 or visit www.OCRAhope.org.

Types Of Genetic Tests

 You can test for a single gene or a broader panel of genes (up to 40) associated with hereditary cancer risk.

Consider Retesting If:

- You were tested just for BRCA 1 or 2.
 Consider getting tested with a broader panel of genes.
- You had genetic testing prior to 2010 because new genes have been identified and will continue to be.

Types Of Test Results

- Negative or Normal: no inherited mutation.
- Positive: You have an inherited mutation.
- Likely Pathogenic Variant: You likely have an inherited cancer mutation.
- Variant of Uncertain Significance (VUS):
 Your clinician can help you figure out how to interpret and follow a VUS result. Usually, VUS are reclassified as normal after further study.
- ? Insufficient sample: This means the test will need to be redone.

Genetic Testing Q&As

Where Can I Get Tested?

 You can obtain genetic testing through your medical or gynecologic oncologist, a genetic counselor or hereditary cancer clinic, directto-consumer testing, or OCRA Genetic Testing Program.

What Is The Process Like?

- Genetic testing can be obtained with a blood draw, saliva sample, or cheek swab. Your clinical team will talk to you about the best way for you to be tested.
- The testing company will analyze your genes and send you and your clinician a report, which can take 4-8 weeks. You and your clinical team can then review how the findings impact your care.

Financial Implications

 Most of the time, your genetic testing will be covered by your insurance. You may or may not have a co-pay associated with the testing. If testing is not covered by your insurance or you struggle with the cost, please let your care team know. There are resources to assist with high co-pays or the cost of genetic testing.

When Should I Get Tested?

- Following an ovarian cancer diagnosis, genetic testing is recommended in the first 1 to 4 months.
- Following a uterine or endometrial cancer diagnosis, genetic testing is recommended for patients based on their tumor staging, tumor testing, or family history.

Who Has Access To My Results?

- Only you and your clinical team have access to your genetic testing results.
- Federal Laws: Health Insurance Portability and Accountability Act (HIPAA) of 1996 protects the privacy and security of individual's health information and ensures the right to access one's own health information. Genetic Information Nondiscrimination Act (GINA) of 2008 prohibits employers and health insurers from using genetic information, including your family history and genetic test results, to discriminate against you. However, current federal laws don't prohibit the use of family history or genetic test results by life, long-term care, or disability insurers.



OCRA strongly believes that genetic testing is an important first step in understanding and managing your risk for ovarian cancer. If you have concerns that you may have an inherited predisposition to developing the disease, OCRA's **patient support line** is open at **212-268-1002** to answer questions about genetic testing and counseling and to help guide you toward next steps.

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Biomarker Testing

for Better Understanding Cancer Behavior



A biomarker is a substance in your body that can be measured and give information about your cancer.

"Biomarker" is a term combining 'biological' and 'marker.' You may hear other words to describe biomarker testing like molecular, genomic, tumor or somatic testing, or precision oncology. These all refer to getting information about your cancer to better understand its behavior and tailor your cancer treatment.

What Is Biomarker Testing?

Biomarker testing is a way for your doctors to gather as much information as possible about your cancer. Biomarkers may analyze your blood or tissue for tumor mutations or activity. Biomarkers may change over time, and tests may need to be repeated.

Some examples of testing include CA-125, a blood test for ovarian cancer surveillance; and mismatch repair deficiency testing, a protein test performed on uterine cancer biopsies.

Biomarker testing may involve:



Blood or saliva test



Testing of tumor from surgery or biopsy

Biomarker Testing Q&As

When Should I Get Tested?

 Consider biomarker testing at the time of your cancer diagnosis and re-testing in the event of a recurrence.

What Is The Process Like?

- Your clinician may send a tumor sample that has already been removed and stored (typically from a prior surgery), schedule a biopsy for new tumor tissue, and/or collect blood or saliva samples.
- The samples will be sent to a lab that will look for a variety of biomarkers. Most oncologists work with national companies on biomarker testing. You may get a call from the company prior to them running the tests to discuss costs.
- It can take up to 6 weeks to get biomarker testing results.

Why Is The Testing Important?

 The biomarker test results may help tailor your treatment to address your specific cancer. These results may lead to being able to use targeted treatments (precision oncology) which can offer a greater chance of improved outcomes in your cancer care.

Financial Implications

- Most of the time, your biomarker testing will be covered by your insurance. You may or may not have a co-pay associated with the testing. If testing is not covered by your insurance or you struggle with the cost, please let your care team know.
- There are resources to assist with high copays or the cost of biomarker testing.

Questions To Ask Your Doctor:

- Have I had genetic testing?
- Have I had biomarker testing?
- Are there biomarkers that could provide information about chemotherapy, targeted therapy, or clinical trial options for me?



- Who will perform the testing?
- How will test results influence my treatment options?
- How long will it take to get my results back?
- Can I get a copy of my results?

For assistance with questions or concerns about genetic testing in gynecologic cancer, contact Ovarian Cancer Research Alliance (OCRA)'s Patient Support Line at 212-268-1002. For information and support, visit www.OCRAhope.org Made possible with support from our partners: AstraZeneca, GSK, Merck, Genentech, Mersana Therapeutics.