What Is Cancer?

Cancer is class of disease where there is uncontrolled growth of abnormal cells in the body. Cancer develops when the body's normal process of cell growth goes wrong and new cells form when the body doesn't need them and old or damaged cells do not die as they should. Buildups of these abnormal cells often invade healthy cells and may form a mass of tissue, called a lump, growth or tumor.

There are some cancers, like leukemia, cancer of the blood that does not form tumors. They grow in the blood cells or other cells of the body.

Cancer Is Not Just One Disease

There are many types of cancer. It's not just one disease. Cancer can start in the lungs, breast, colon, or even in blood. Cancer cells can also spread to other parts of the body. For instance, cancer cells in the lung can travel to the bones and grow there. When cancer cells spread, it's called metastasis. When lung cancer spreads to the bones, it's still called lung cancer. It's not called bone cancer unless it started in the bones.

Cancer And Genetics

Cancers that run in families can be caused by an abnormal gene that is passed from generation to generation. Although this is often referred to as inherited cancer, what is inherited is the abnormal gene that can lead to cancer, not the cancer itself. Only about 5 to 10% of all cancers are thought to result directly from gene defects or mutations inherited from a parent.

Gene Mutations Can Be Inherited Or Acquired

An **inherited gene mutation** is present in the egg or sperm cell that formed the child and is passed down through generations.

An **acquired gene mutation** does not come from a parent, but is acquired some time later. It starts in one cell, and then is passed on to any new cells that are created from that cell. This kind of mutation is not present in egg or sperm cells, so it is not passed on to the next generation. Acquired mutations are much more common than inherited mutations. Most cancers are caused by acquired mutations.

Risk Factors

Certain things make it more likely that cancers in a family are caused by a family cancer syndrome, such as:

- Many cases of the same type of cancer, especially if it is an uncommon or rare type of cancer.
- Cancers occurring at younger ages than usual such as colon cancer in a 20-year-old.
- More than one type of cancer in a single person such as a woman with both breast and ovarian cancer.
- Cancers occurring in both of a pair of organs like both eyes, both kidneys, or both breasts.
- More than one childhood cancer in siblings for example sarcoma in both a brother and a sister.
- Cancer occurring in the gender not usually affected such as breast cancer in a man.
- Cancer occurring in many generations, like in a grandfather, father, and son.

When trying to determine if cancer might run in your family, first gather some information. Look at:

- Who has the cancer? How are you related? Which side of the family are they on (mother's or father's)?
- What type of cancer is it? Is it rare?
- How old was this relative when they were diagnosed?
- Did this person get more than one type of cancer?
- Did they have any known risk factors for their type of cancer (such as smoking for lung cancer)?

Otogenetics Cancer Screening Panels

- **BRCA1/2 – 2 genes**
- **Breast and Ovarian Cancer – 15 genes**
  ATM, BRCA1, BRCA2, BRIP1, CDH1, MLH1, MSH2, MSH6, PALB2, PTEN, RAD51C, RET, STK11, TP53, VHL
- **Colorectal, Endometrial and Ovarian Cancer Panel - 12 genes** APC, BMPR1A, CDH1, EPCAM, MLH1, MSH2, MSH6, MUTYH, PMS2, PTEN, SMAD4, STK11
- **Comprehensive Inherited Cancer Panel – 39 genes linked to Breast, Ovarian, Endometrial, Colorectal, Lynch Syndrome, Gastric, Melanoma, Pancreatic, Polyposis, Prostate, Renal, Thyroid/Parathyroid, Uterine and other major cancers** APC, ATM, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKn2A, CHEK2, ELAC2, EPCAM, FANCC, HRAS1, MEN1, MET, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, NF1, NTRK1, PALB2, PALLD, PMS2, PTCH, PTEN, RAD50, RAD51, RAD51C, RAD51D, RET, SMAD4, STK11, TP53, VHL
- **Lynch Syndrome - 5 genes**
  EPCAM, MLH1, MSH2, MSH6, PMS2

Mutations in each gene may be rare when evaluated individually, however they collectively account for a significant amount of hereditary cancer susceptibility. This panel may be appropriate in a number of scenarios, particularly if the family history shares features of several different hereditary cancer syndromes with multiple cancer types.

Examples Of Hereditary Cancer Syndromes

Breast Cancer is the most common cancer in women in developed countries, affecting about 1 in 8 (12.5%) women in their lifetime. The National Cancer Institute (NCI) estimates that approximately 231,840 new cases of female breast cancer and 2,350 new cases of male breast cancer will be diagnosed in the U.S. in 2017.

The majority of breast cancers are sporadic, but 5-10% are due to inherited causes. Hereditary breast cancer tends to occur earlier in life than non-familial cases and is more likely to occur in both breasts. The BRCA1 and BRCA2 genes appear to be responsible for ~half of hereditary breast cancer. However, additional genes are associated with increased breast cancer risk as well. Mutations in the genes included in the Comprehensive Panel can confer an estimated 20–87% lifetime risk for breast cancer. Some of these genes are associated with increased risks for other cancers, such as pancreatic cancer with PALB2, ovarian cancer with BRCA1, BRCA2, RAD51C, and sarcoma with TP53.

Ovarian Cancer is the fifth most common cancer among women in developed countries, affecting approximately 1 in 71 (1.4%) women in their lifetime. The NCI estimates that approximately 21,290 new cases of ovarian cancer will be diagnosed and 14,180 ovarian cancer deaths will occur in the U.S. in 2017. BRCA1 and BRCA2 are the most common causes of hereditary ovarian cancer, but several other genes are associated with increased ovarian cancer risk as well.

Colorectal Cancer (CRC) affects about 1 in 20 (5%) men and women in their lifetime. The NCI estimates that ~132,700 new cases will be diagnosed and ~49,700 CRC deaths will occur in the U.S. in 2017. Lynch syndrome is the most common form of hereditary CRC, but other genes on the Panel are also associated with increased CRC risk.

Uterine Cancer affects about 1 in 38 (2.6%) women in their lifetime. The NCI estimates that approximately 54,870 new cases of uterine cancer will be diagnosed and ~10,170 uterine cancer deaths will occur in the U.S. in 2017. Increased risk for uterine cancer has been identified in a number of hereditary cancer syndromes, including Lynch syndrome and Cowden syndrome.

Prostate Cancer is the second most common cancer in men in the United States, after skin cancer. The NCI estimates that approximately 180,890 new cases of prostate cancer will be diagnosed in the U.S. in 2017. Hereditary prostate cancer may be diagnosed at younger ages and may also be more aggressive. For example, BRCA1 and BRCA2 gene mutations have been shown to be associated with more aggressive prostate cancer.

What Do My Test Results Mean?

Testing does not tell you if you currently have cancer. Your test results tell you about your inherited ***risk*** of having breast and ovarian cancer, or other cancers.

A negative test result means that you do not have a gene mutation in what were evaluated in the Gene Panel Testing. Although the gene test can detect the majority of mutations:

- You could have a gene mutation or type of mutations that the test wasn't targeted to detect.
- Or if your family carries a high-risk gene mutation that researchers haven't yet identified then the test would not be able to detect this gene mutation and you could be a carrier.

A negative test result does not eliminate the chance of developing non-hereditary cancer. You may have the same cancer risk as that of the general population.

A positive test result means that you have a gene mutation that is associated with cancer. A positive test result **does not** mean that you have cancer or that you will certainly have cancer. A positive test result tells you that you have a higher risk of developing cancer compared with someone who does not have the mutation.

Hereditary Cancer Mutations Can Be Passed On In A Family

- If you have a mutation in one of these genes, your parents, your children, and your brothers and sisters have a chance that they have the same mutation.
- Other relatives may be at risk to carry the same mutation. Testing is the only way to accurately identify mutation carriers.
- It is important to share test results with family members.
- Individuals may differ in their viewpoints and reactions to genetic testing.

Cancer Risk Management

Options for reducing cancer risk are available whether or not you have already had a diagnosis of cancer and/or polyps (adenomas). Your healthcare providers will discuss your testing options and medical management options for reducing risk such as **Increased Surveillance, Surgical Management, and Chemoprevention.**

For more information and patient advocacy groups, please visit our Patient Portal at
www.otogenetics/patientportal.com

Otogenetics Corporation
4553 Winters Chapel Road, Suite 100, Atlanta GA 30360

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Understanding Your Hereditary Cancer Screening Options

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