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PENNSYLVANIA CANCER CONTROL

Patient and Family Centered Genetics/Genomics Toolkit

A Resource for Patients, Families, and Care Providers

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Dear Reader:

We hope you find the Patient-Centered Cancer Genetics/Genomics Toolkit helpful.

The toolkit is intended to provide you and/or your family with information and resources to increase your understanding of hereditary cancers and hereditary risk factors. The toolkit provides information on genetic counseling and testing, and when to talk to your doctor or provider about if you or someone in your family should consider genetic counseling.

Genetics and genomic testing play a key role in cancer prevention, early detection, diagnosis and treatment, and improving outcomes and quality of life.

The toolkit was created by the Genetics/Genomics Workgroup of the Pennsylvania Cancer Coalition (PCC).

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Introduction

The Pennsylvania Cancer Coalition (PCC) is the statewide cancer coalition whose members focus efforts on reducing the burden of cancer using the **2023-2033 Pennsylvania Cancer Plan** (Cancer Plan) as the blueprint or guide for coalition activities.

Membership is open to all cancer stakeholders, including private and nonprofit organizations and individuals. *Cancer survivors and caregivers are welcomed and encouraged to join. Membership link and contact information can be found here:* **Pennsylvania Cancer Coalition** (PCC).

In 2019, the **Genetics/Genomics Workgroup** was created within the PCC to address ways to increase availability and access to genetic counseling and testing, the importance education, insurance coverage, and the promotion of policy and systemic change to advance cancer genetics/genomics to save lives and improve health and quality of life.

This **Patient and Family Centered Genetics/Genomics toolkit** is intended to provide you and your family members with information and resources to help you make informed decisions about cancer genetic counseling and testing. The toolkit will provide you with basic information about cancer genetics, testing and how to gather and share information about your family medical history with your doctor and/or genetic counselor.

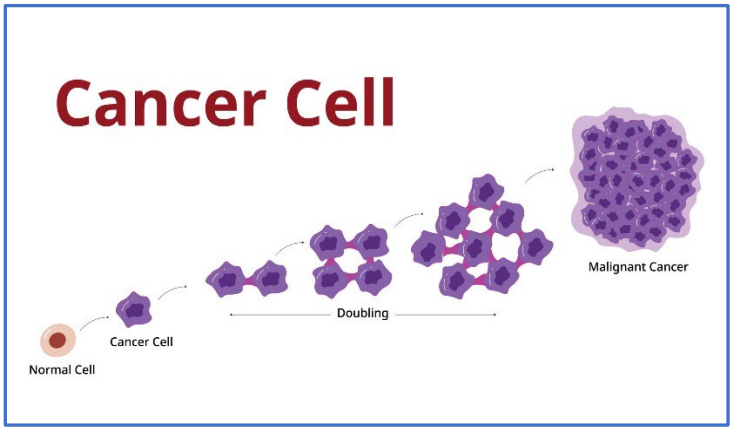
What is Cancer?

Cancer is a disease in which damaged cells in the body grow uncontrollably and, in some cases, can spread to other areas in the body. Normally, cells grow to create new cells as the body needs them, such as when cells grow old or become damaged and die.

When this process breaks down, damaged cells can make copies of themselves when they shouldn't. This is because the cells acquire a gene mutation that changes the way the cell functions. The damaged cells can grow out of control and form an abnormal mass of tissue called **tumors**.

Some tumors are benign (or non-cancerous), and others can become malignant (or cancerous). Benign tumors do not spread but can grow to be very large. Although benign tumors are not usually life-threatening, they still may need to be removed.

Cancerous tumors grow uncontrollably, invading nearby tissue and organs. Cancerous tumors can also spread to distant parts of the body. This is called **metastasis.** Cancerous tumors can be life-threatening and require medical or surgical attention.



For more Information about what cancer is and how cancer occurs click links below:

- What is Cancer?
- American Cancer Society

Video links:

- What is Cancer
- Cancer is caused by mutations
- https://www.youtube.com/watch?v=BmFEoCFDi-w

Cancer Genetics Basics

- **Genes** are the instructions inside of your cells tha**t tell your body how to work.**
- **Genes** are made up of genetic **building blocks called DNA (**deoxyribonucleic acid). For more information, see this link: Deoxyribonucleic Acid (DNA).
- Sometimes, changes occur in the DNA of genes which stops the genes from working properly. These **changes** are called and **pathogenic variants or mutations.**
  Changes or mutations in DNA can occur naturally as part of the aging process. However, some people are born with DNA mutations that cause or increase the risk of certain cancers or other diseases.

- **These pathogenic variants (also called mutations or pathogenic variants) can be passed on in families.** Some family members may have the pathogenic variant and others do not. Family members who have the pathogenic variant may be at increased risk for cancer, and family members who do not have the variant may have average risk for cancer.
- **Not all family members with a pathogenic variant will get cancer** and sometimes family members who do not have the pathogenic variant will get cancer anyway.
- **Informed decision-making with your physician, primary care provider, or genetic counselor are important to determine if you have a risk for cancer.**

For more information about cancer genetics and how genetic mutations can influence cancer risks:

- The Genetics of Cancer
- Navigating Cancer Care

What are Hereditary Cancer Syndromes?

Hereditary cancer syndromes are when someone is born with a genetic change in a gene that increases a person's chances of getting cancer. These genetic changes can be passed down through families.

Only 10% of cancers are due to hereditary cancer syndromes. Most people do not know they have a hereditary cancer syndrome. They are often undiagnosed.

While there are many known hereditary cancer syndromes, the **two most common are Hereditary Breast and Ovarian Cancer (HBOC) and Lynch syndrome (LS)**.

How are Hereditary Cancer Syndromes Inherited?

People inherit two copies of each gene, one copy from each of their parents. Hereditary cancer syndromes are typically inherited in an autosomal dominant pattern -- only one copy of a mutated gene is needed to cause a disorder. If a parent has an autosomal dominant hereditary cancer syndrome, there is a 50% chance that each of their children could inherit the same mutated gene.

Common types of Familial or Hereditary Cancer Syndromes	
Syndrome	*Associated Cancer Risks*
Hereditary Breast and Ovarian Cancer- *BRCA1* and *BRCA2* genes	breast, ovarian/fallopian tubes, prostate, and pancreatic cancers
Li-Fraumeni Syndrome	breast, soft tissue sarcomas, and brain tumors
Lynch Syndrome	colorectal, endometrial (uterine), ovarian, and other cancers
Familial Adenomatous Polyposis (FAP)	colorectal cancer

For more information about types of hereditary cancer syndromes, see these sources:

- Hereditary Cancer Syndromes > Fact Sheets > Yale Medicine
- Hereditary Breast and Ovarian Cancer Syndrome (HBOC) > Fact Sheets > Yale Medicine
- BRCA Gene Changes: Cancer Risk and Genetic Testing Fact Sheet - NCI
- Lynch syndrome > Fact Sheets > Yale Medicine
- https://www.cancer.org/cancer/risk-prevention/genetics/family-cancer-syndromes.html

How do I know if I have a Hereditary Cancer Syndrome?

You **may** have a hereditary cancer syndrome **if you or your family members have certain patterns of cancer. It is important to know your family medical history and to share this information with your doctor.**

You and your family members may share genes, behaviors, and environments that can affect your risk of getting cancer. **Talk to your doctor if you are worried about cancer in your family.**

Your doctor or other healthcare provider can help determine if you or other members of your family should be tested for a hereditary cancer syndrome.

The following can be "red flags" for a hereditary cancer syndrome:

- Two or more relatives on the same side of the family diagnosed with the same types of cancer.
- A cancer diagnosis under the age of 50

- A diagnosis of a rare cancer like ovarian cancer or male breast cancer
- A person in your family diagnosed with more than 1 type of cancer.
- Bilateral cancers or a cancer occurring in a "pair of organs" such as both breasts and both kidneys
- Ashkenazi (Eastern European) Jewish ancestry

You will want to document your family health and cancer history to discuss with your provider. Based on the information you provide, genetic counseling may be recommended.

Family History Tools and Resources

For more Information on how to collect and share information about your family medical history, go to:

- **CDC's Family Health History Tools and Resources**
- **Mapping Your Family's Cancer History**
- **My Family Health Portrait**
- **Family Health History for Breast Cancer**
- **The Geisinger Guide to Family Health History**
- **It Runs in My Family: Health History Tool**

What is Genetic Counseling?

Genetic counseling seeks to assist individuals and families with:

- Understanding they have a hereditary cancer syndrome.
- Informed consent for genetic testing when appropriate.
- Discussing options for screening, prevention and family planning based on genetic test results.
- Providing support to patients and families.

It is important that you and your family members are aware about the option of genetic counseling and testing so you can make **informed decisions about your health, such as lifestyle changes, cancer screenings, and preventative care.**

Genetic counselors are trained healthcare professionals who specialize in assessing family history and identifying genetic conditions. You might consider asking your healthcare provider to refer you to **talk to a genetic counselor if you are concerned about family cancer.**

During a genetic counseling consult or session, a genetic counselor will discuss your family tree in detail, including:

- Who specifically had cancer in the family (and on what side of the family)

- The types of cancer your relatives had
- What age were they diagnosed with cancer

Based on your family tree assessment, you may learn that you have high chances of having a hereditary cancer syndrome. Genetic testing to better understand your cancer risks might be offered. Genetic testing is done using a small amount of blood or saliva.

To learn more:

- Frequently Asked Questions + Resources
- What Is Genetic Testing? Understanding Genetic Testing for Cancer | American Cancer Society
- Genetic Testing Fact Sheet - NCI

Additional Information:

- About Genetic Counselors
- What is a Genetic Counselor?
- Find A Genetic Counselor
  Find A Genetic Clinic
- Genetic Testing for Inherited Risk for Cancer
- NCCN Genetic Testing for Hereditary Breast Ovarian Pancreatic and Prostate Cancers

Video Resources:

- Genetic Testing for Inherited Risk for Cancer

Why Have Genetic Testing for Cancer Risk?

- Genetic test results can help you better understand your chances of getting cancer.
- In some people, a normal or negative result can provide peace of mind if they did not inherit a gene mutation that was previously identified in the family (an informative or true negative result).
- Genetic test results that find a gene mutation (an abnormal or positive test result) provide an opportunity to understand and, in some cases, potentially **prevent cancer or reduce the chances of getting cancer.**
- For people who are already diagnosed with a cancer, results of genetic testing may help suggest the most appropriate and targeted treatments and provide information about their risk of getting other cancers.

What is Cancer Biomarker and Testing *after* a Cancer Diagnosis?

Cancer biomarkers are genes, proteins, or other substances that can reveal how aggressive a cancer is and how it can best be treated. Depending on why and how it's done, biomarker testing for cancer treatment can have different names.

The following list is examples of biomarker testing:

- Tumor testing, tumor genetic testing, tumor marker testing, or tumor subtyping
- Genomic testing, genomic profiling, or genome sequencing
- Molecular testing or molecular profiling
- Somatic testing
- Next generation sequencing

For more information:

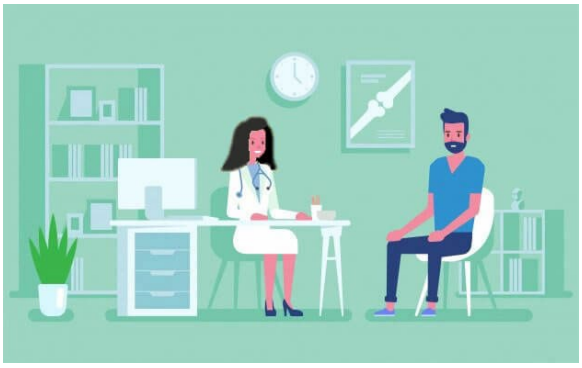
- **Biomarker Testing for Cancer Treatment - NCI**
- **Biomarker Tests and Cancer Treatment | American Cancer Society**
- **Biomarkers | Cancer Treatment**

Video:

- **What is Biomarker Testing**

Final Considerations:

- Gather your family health history by talking with relatives. Ask your medical provider if talking with a genetic counselor would be beneficial for you.
- A genetic counselor can review your family tree help you understand if you have a high chance of getting cancer. They can discuss with you if genetic testing is right for you.
- Having a hereditary cancer syndrome does not mean that you have a guarantee to get cancer. Similarly, normal genetic testing does not mean that you will never get cancer.
- **There are screenings and preventative steps that we can take to reduce the chances of getting cancer**, even when genetic test results show that you have high risk for cancer.



Additional Resources:

Find Financial Assistance:

- Support - Find Financial Assistance
- Financial & Co-Payment Assistance for Cancer Patients

Video Resources:

- Cancer Stories of Help and Hope | Video Library | Cancer Care

Podcasts:

- Podcast Resources
- American Society of Human Genetics: Podcasts
- Personal Genomics Podcast - NCI

Acknowledgment:

Genetics/Genomics Workgroup

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