



## Genetics and Your Risk for Cancer

### What are genes? How do they relate to whether or not I will get cancer?

First, it is important to understand what is meant when something is "inherited". Inherited traits are passed from one generation of a family to the next. These traits, like eye color or hair texture, are passed on in the form of **genes**.

- We have about 30,000 different genes in each cell of our body.
- We have two copies of each of these genes. We inherit one copy from our mother and one copy from our father.
- Some genes code for particular traits, like eye color or hair color, while other genes keep the cells in our body healthy.
- Each gene is made up of **DNA**. The order of DNA is very specific so that the gene can perform a specific function.
- If there is a **mutation** (or change) in the DNA order in any gene, this may cause the gene to not function properly.
- Mutations in some genes can lead to a higher chance of getting certain diseases. A gene that could lead to a higher chance of getting cancer is called a **cancer susceptibility gene**.

### What are some examples of cancer susceptibility genes?

We know of several cancer susceptibility genes. Some of the genes that we frequently talk about in genetic counseling are listed below.

- Next to the name of the gene(s) is the name of the genetic syndrome associated with the gene(s).
- A syndrome is a cluster of symptoms or diseases that happen in an individual or family because of a mutation in a specific gene. If your family cancer history suggests a syndrome, your genetic counselor will provide you with more detailed information about the particular syndrome.

Hereditary Breast and Ovarian Cancer syndrome (HBOCS)	<i>BRCA1, BRCA2</i>
Cowden syndrome (CS)	<i>PTEN</i>
Hereditary Non-Polyposis Colorectal Cancer (HNPCC)	<i>MLH1, MSH2, MSH6</i>
Familial Adenomatous Polyposis (FAP)	<i>APC</i>
MYH- Associated Polyposis(MAP)	<i>MYH</i>

### **How common is to get cancer because of gene mutations?**

Most cancers are considered **sporadic** and are not caused by an inherited factor. Individuals can develop sporadic cancer due to chance, environmental exposures or certain lifestyle factors.

Hereditary factors (For example, inherited mutations in cancer susceptibility genes) are responsible for about 5-10% of cancer cases.

There are four features that we typically see in a family when there is an inherited mutation in a cancer susceptibility gene. These features include:

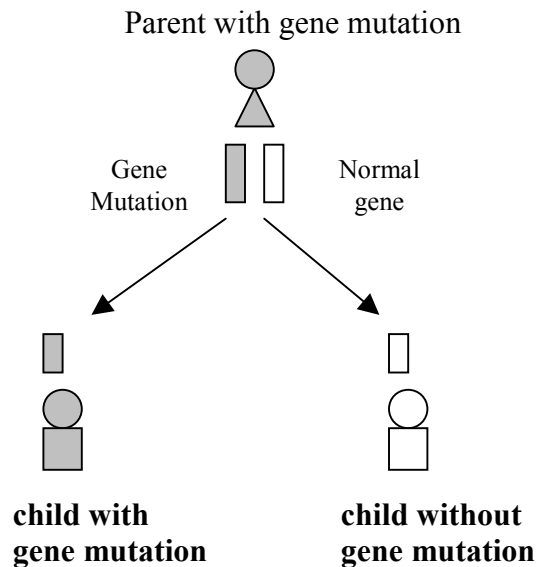
- 1) Multiple family members that have had cancer of the same tissue/organ or other possibly related organs (such as breast cancer and ovarian cancer on the same side of the family, or colorectal and uterine cancer on the same side of the family).
- 2) Young ages of getting cancers (such as cancer before age 50).
- 3) Individuals in the family may have multiple cancers such as two separate, primary colorectal cancers or colorectal and uterine cancer in the same person.
- 4) Cancer occurring in multiple generations (for example, both grandmother and mother had breast cancer).

### **What is the chance that a person who has a mutation in a cancer susceptibility gene will pass it on to his/her children?**

- We all have two copies of every gene.
- If a person has a mutation in a cancer susceptibility gene, the other copy of the gene is probably normal. It is extremely rare for a person to have mutations in both copies of a cancer susceptibility gene.
- We pass on only one copy of each gene to each of our children.
- With each child, there is a 50% chance (or 1 in 2 chance) that the child would inherit the copy with the mutation and a 50% chance that the child would inherit the normal gene.

The figure below illustrates this concept.

**Figure 1: Inheritance**



**Conclusion: 1 in 2 (or 50%) chance for each child to inherit mutated gene.**

### **How does a lab test for mutations in a cancer susceptibility gene?**

- First, a sample of blood is taken from the person who has chosen to have genetic testing.
- Then DNA is isolated from the white blood cells. It is analyzed to look for mutations in the order of DNA.
- Think of the DNA order of a gene as a book of instructions spelled out by a specific sequence of letters.
- The lab “reads” along the particular gene and compares it with a copy of the correct sequence of DNA. By comparing a test sequence with the normal DNA sequence, the lab can look for differences in “spelling.”
- Sometimes, a sequence difference is benign, which means it does not cause problems in the function of the gene.
- Other times, it can hurt the function of a gene, and is then called a gene mutation.

### **What do I need to think about when considering genetic testing?**

Your genetic counselor will talk with you about different issues so you can make an informed decision about genetic testing. Issues that you should think about include:

- What would I do with the information if I decide to have testing?
- How will the result affect me emotionally?
- How/when would I share this information with my family?
- Will my insurance cover the test and the genetic counseling visit?
- What are the laws that protect me against genetic discrimination?

There are laws that forbid health insurances and businesses from discriminating based on genetic information. We feel that the risk of genetic discrimination in these areas is very low. Your genetic counselor will discuss this with you in more detail.

## **What happens in a genetic counseling visit?**

### **Preparing for the visit**

- Ask relatives for clear information about your family history ahead of time. We need to know:
  - The type of cancer
  - The age when the cancer was found
  - The type of treatment the person received.
- Pathology records on the cancers are also helpful to the genetic counselor.
- You may find it helpful to bring someone with you to your genetic counseling visit. The genetic counselor may go over a lot of information.

### **During the Visit**

- Your genetic counselor will ask questions about your medical history and the medical history of your relatives (i.e. siblings, parents, aunts, uncles, and grandparents). They will be especially interested if anyone has had cancer or pre-cancerous lesions.
- Your risk for a hereditary factor will be based on the information you provide. If more information is needed, we may ask you to follow up or help you get records.
- If your family history suggests a hereditary syndrome, we will offer genetic testing. Again, your genetic counselor will discuss with you the different issues surrounding genetic testing.

### **Results of the Visit**

- Your genetic counselor may find that your risk of having a mutation in a cancer susceptibility gene is low.
  - Therefore, genetic testing is not needed for you or your family.
  - Your genetic counseling team will provide you and your other health care providers with recommendations for cancer screening according to the family history, if appropriate.
- In other cases, your genetic counselor may recommend that another member of your family consider getting testing first. More helpful information might be found that way.
- Regardless, you and your genetic counselor will come up with a plan for genetic testing.
- Your genetic counselor will summarize the visit in a letter to you as well. This way you will have a review of the information for your personal records.

**If you have any questions regarding this information or if you would like to schedule an appointment for genetic counseling and testing, please call us at 617-724-1971.**