Dear Patient,

Knowing you have a genetic mutation helps you get good care. This handout answers common questions and concerns that may arise.

**How does knowing you have this gene help you?**

This information can be lifesaving because now...

- You can take advantage of available risk-reducing measures.
- You can help family members minimize their risk.
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**What is a deleterious mutation?**

It's a gene (a tiny piece of DNA in all your cells) associated with an increased risk of certain types of cancer. Think of your genes like phrases in an instruction manual for keeping your cells healthy. Genetic mutations are like typos. Most are insignificant, without any ill effects. Deleterious mutations are like typos that change the instructions, making it easier for some cells to become cancerous.

**Does this gene put you at increased risk for all types of cancer?**

No. It increases your risk for only one or, at most, a few types of cancer, depending on which mutation you have. In most of your cells, the mutated gene is there but of no consequence (like a stain on the underside of a chair).

**Are you destined to develop the types of cancers associated with this gene?**

No, for three reasons: First, even if the risk is greatly increased compared to people without the mutation, many people with the gene do not develop cancer. Second, a variety of other factors help determine your overall risk. All other things being equal, you may have less risk of developing the associated cancer than someone with the same gene but a much stronger family history of that cancer. Third, you can take advantage of available measures to decrease your risk.

**How do we know what to do to decrease your risk?**

The mutation changes the equation for when and how we use available screening and prevention. Compared to the general population, you may benefit from...

- Starting standard screening tests at an earlier age.
- Doing screening tests more frequently.
- Using more sensitive screening tests (e.g., 3D mammography or MRIs of the breast instead of standard mammograms).
- Taking medication that helps lower the risk.
- Undergoing preventive (prophylactic) removal of the organ(s) at risk.

**Do my children have this mutation?**

Each child has a 50-50 chance of also having the deleterious gene. That was determined when he or she was conceived. Nothing you did or didn’t do affected whether a child inherited the gene.

**Do I need to test my children?**

In general, we delay genetic testing of children until they are at least 18 years old so they can make their own decisions about genetic testing. Letting children decide as adults is paramount over other concerns because, even in the presence of a mutation, most risk-reducing measures begin after age 35 at the earliest.

If your children are young adults, determining the best time to be tested is a personal decision. Some want to know as soon as possible. Others want to delay until they are closer to the age when risk-reducing measures become an option for them. Consulting with a genetic counselor can help them determine the best time for them.

**What if you feel guilty that your children may (or do) have the gene?**

You did nothing wrong and have no reason to feel guilty. What feels like guilt may be your instinct to protect your children, along with any sadness, anger, and/or anxiety about your children possibly having the gene. If anything, you were taking good care of your children by finding out about your mutation. Now your children can benefit from genetic testing that may help them prevent cancer—and even save their life. And it comforts your children to know that now you can receive more personalized care.

**What are your family obligations?**

We ordered the genetic test to take the best care of you. By its nature, a positive result may impact some of your family members. A genetic counselor can tell you which family members may also have the mutation and can guide you on how best to fulfill your obligation to inform relatives. A Cancer in the Family by Theodora Ross, MD, PhD, also provides useful insights and advice.

**Will you lose your insurance?**

The Genetic Information Nondiscrimination Act (GINA) prohibits the use of genetic information in insurance or employment. Unfortunately, GINA does not apply if your employer has fewer than 15 employees, if you are in the U.S. military, or if you are receiving health benefits through the Veterans Health Administration or Indian Health Service. GINA also doesn’t protect against genetic discrimination in obtaining life, disability, or long-term care insurance.

**What if you feel anxious?**

Anxiety may rise while you are adjusting to the news, making medical decisions, and dealing with family members. Notify us if anxiety interferes with your ability to eat, sleep, make decisions or fulfill your responsibilities. Even if you are functioning fine, please notify us if anxiety persists.

**What if you feel ashamed? Embarrassed?**

In life, your thoughts, feelings and actions define you—not your genes. In every way, you are as healthy and whole as you were before the test. And now you are less vulnerable because you can take advantage of risk-reducing measures. From our perspective, you are stronger and smarter than those people who choose to not to learn about their genetic health.

**What now?**

Make an appointment with a genetic counselor to review the findings and discuss the next steps. Keep us informed of how you are doing and any questions or concerns. Remember, knowing you have this mutation does not change what is. It changes what you can do about what is.