About Mutations in the BRCA1 Gene

This information explains how having a mutation in the BRCA1 gene may affect you and your family.

In this resource, the word “family” means family members related to you by blood. They are not related to you through marriage or adoption.

Your BRCA1 gene normally helps prevent cancers. A mutation in this gene causes it to stop working like it should. This increases your risk for certain types of cancers.

What is my cancer risk if I have a BRCA1 mutation?

If you have a mutation in the BRCA1 gene, this means you have a condition called Hereditary Breast and Ovarian Cancer (HBOC) syndrome. HBOC syndrome increases your risk for certain types of cancers, including:

- Breast cancer
- Ovarian cancer
- Prostate cancer

HBOC syndrome may also increase your risk for other cancers, but this is less common. Examples of less common cancers linked to HBOC syndrome are:

- Male breast cancer (breast cancer in males)
- Pancreatic cancer
As we learn more about these mutations, we may learn they increase the risk for other types of cancers. Your genetic counselor will give you more information about your cancer risk if you have a mutation.

For more information, read *BRCA1 and BRCA2 Gene* - www.mskcc.org/genetics/brca1-brca2-genes

**What can I do about my cancer risk if I have a **BRCA1** mutation?**

If you have a mutation, your genetic counselor will review your results and your personal and family history of cancer and give you cancer screening recommendations.

They may recommend you start having cancer screenings at a younger age, have them more often than most people, or get specialized screenings to help find cancer as early as possible.

Some examples of these cancer screenings include:

- Having breast magnetic resonance imaging (MRI) scans and mammograms (x-rays of your breast) starting at an earlier age.

- Getting prostate exams and prostate specific antigen (PSA) blood tests (a blood test to check for prostate cancer) starting at an earlier age.

They may also discuss having surgery to remove your ovaries to prevent ovarian cancer. If you decide to have surgery, talk with your genetic counselor about the right time to have it. Surgery to remove the ovaries affects fertility (ability to have biological children). If you plan to have biological children, your genetic counselor can talk with you about your options.

Your genetic counselor will also talk with you about whether there are any other screening or prevention options that may be right for you.
What happens if I don’t have a BRCA1 mutation?

If you don’t have a mutation, your genetic counselor will review your personal and family history and talk with you about the general cancer screening guidelines you should follow.

What does a BRCA1 mutation mean for my blood relatives?

If you have a mutation, your biological parents, siblings, and children each have a 50% chance of having the same mutation. This means there’s an equal chance they will or won’t have the mutation. Your distant family members may also be at risk for having the same mutation.

Males and females have an equal chance of passing down a mutation in their family. You only need to inherit a mutation from one parent to have an increased risk for cancer.

Your genetic counselor will review your family history and talk with you about whether they recommend genetic testing for your blood relatives.

What does this mean for family planning?

If you have a BRCA1 mutation and plan to have children, there are options to prevent your children from inheriting the mutation. You may want to consider discussing these options especially if both you and your partner have a BRCA1 mutation.

If you both have a mutation in the BRCA1 gene, which is rare, there’s a chance your child could be born with a serious condition called Fanconi Anemia (FA). FA is a genetic disorder that can cause birth defects, bone marrow failure, and a risk of cancer. If you already have children, it’s unlikely they have FA since this is usually diagnosed early in life. For more information about genetic testing and family planning, talk with your genetic counselor.
Contact Information

If you have any questions or concerns, talk with a genetic counselor in the Clinical Genetics Service. You can reach them Monday through Friday from 9:00 a.m. to 5:00 pm at 646-888-4050.

For more resources, visit www.mskcc.org/pe to search our virtual library.