Memorial Sloan Kettering Cancer Center

PATIENT & CAREGIVER EDUCATION

About Mutations in the PALB2 Gene

This information explains how having a mutation in the *PALB2* 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 *PALB2* 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 *PALB2* mutation?

A *PALB2* mutation increases your risk for breast cancer. It can also increase your risk for pancreatic cancer, but this is less common.

A *PALB2* mutation may also increase your risk for ovarian cancer, but more research is needed for us to better understand this risk. Your genetic counselor will give you more information

about what we know so far and what it means for you.

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 *Hereditary Breast Cancer* - www.mskcc.org/genetics/breast-cancer and *Hereditary Pancreatic Cancer* - www.mskcc.org/genetics/pancreatic-cancer

What can I do about my cancer risk if I have a *PALB2* 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.

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 *PALB2* 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 *PALB2* 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 *PALB2* 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 *PALB2* mutation.

If you both have a mutation in the *PALB2* 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 AM to 5:00 PM at 646-888-4050.

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