



# About PALB2 Gene Mutations

### **About Genes**

Genes are in every cell in our bodies. Genes are made of DNA, which gives instructions to cells about how to grow and work together. We have two copies of each gene in each cell—one from our mother and one from our father. When genes work properly, they help stop cancer from developing.

When it works right, the *PALB2* gene works together with the *BRCA1* and *BRCA2* genes to help prevent cancer. Sometimes changes to the *PALB2* gene happen. These changes are called mutations. Mutations can make the *PALB2* gene stop working and raise the risk for certain types of cancer.

Having a mutation in the *PALB2* gene makes your risk of getting breast and pancreatic cancers higher than average.

### **PALB2** Mutations and Cancer Risk

#### **Breast Cancer**

About 1 in 10 women get breast cancer during their lifetime. The low range of lifetime breast cancer risk for women with a *PALB2* mutation is about 3 in 10. The risk for women with an *PALB2* mutation and a family history of breast cancer can be up to 6 in 10. Women with an *PALB2* mutation who already had breast cancer have a higher risk to get a new breast cancer.

### **Pancreatic Cancer**

Pancreatic cancer is rare. About 1 in 100 men and women get this type of cancer in their lifetime. The risk is somewhat higher for people with a *PALB2* mutation. The risk is even higher for people with a family history of pancreatic cancer and a *PALB2* mutation.

### **Other Cancers**

The risks for other cancers may also go up with *PALB2* mutations. Currently, there are no changes to cancer screening recommendations for these other cancers. Researchers are studying the *PALB2* gene to understand more.

## Recommendations

#### WOMEN

Starting at age 30: Mammogram and breast MRI every year (scheduled 6 months apart).

Some medicines can lower the risk of getting breast cancer. Surgery to remove both breasts may be an option for some women who have a strong family history of breast cancer.

### **WOMEN AND MEN**

Screening for pancreatic cancer has benefits and limitations. We do not recommend this screening for most people with *PALB2* mutations.

People who have a *PALB2* mutation and a family history of pancreatic cancer should be seen in a high risk clinic to discuss pancreatic cancer screening options. Screening may be considered at age 50 or 10 years prior to the youngest age a relative received a pancreatic cancer diagnosis (whichever comes first).

## **KIDS AND SIBLINGS**

Children and siblings of people with a *PALB2* mutation have a 1 in 2 chance of also having the mutation. We recommend genetic testing and counseling for them after age 18.

If two people with *PALB2* mutations have a child together, there is a 1 in 4 chance the child will have a condition called Fanconi anemia (FA). Signs of FA include anemia, bone marrow failure, and birth defects such as malformed thumbs or forearms. If only one parent has a *PALB2* mutation, their children are not at risk to have FA.

# **FAMILY MEMBERS WHO TEST NEGATIVE**

Family members without the *PALB2* mutation probably do not have a higher risk of getting cancer. The family history of cancer and other risk factors may raise their risk somewhat. Family members who test negative for the *PALB2* mutation should talk with their doctor or genetic counselor about the right cancer screenings.

Do you have questions about your risk for cancer? Our doctors and genetic counselors can help find the cancer screening plan you need. Call Huntsman Cancer Institute's Family Cancer Assessment Clinic to learn more: 801-587-9555.