

PALB2 Gene Mutation

Cancer Risks

Genes are the set of instructions that tell the cells in our body how to work. Some genes, like PALB2 protect us from tumors and cancers. Everyone has two copies of this gene. When the PALB2 gene works correctly, it protects us from cancer. A genetic change (mutation) in PALB2 means that a gene has stopped working that normally helps to prevent cancer. When the gene stops working correctly, cancer risk increases

Cancer Type	General Population Cancer Risk	People with PALB2 Risk
Breast Cancer- Females	10%-12%	14%-58%- Dependent on family history
Breast Cancer- Male		Male breast cancer risk may be increased; specific risk is unknown at this time.
Secondary Breast Cancer		The chance to develop a new or second breast cancer may be increased, but the risk is unknown
Pancreatic Cancer	1%-2%	Specific risk is unknown. Further research needed.

Screening and Prevention

Screening is used to monitor people with a higher risk for cancer. The goal of screening is to detect cancer as early as possible when it may be easier to treat. Recommendations for people with PALB2 mutations include screening earlier and more frequently than the general population.

The screening guidelines below are based on age and personal and family history.

Breast Cancer

- Annual mammogram and breast magnetic resonance imaging (MRI) at age 30, or 10 years before the youngest diagnosis in the family. Alternate mammogram with breast MRI every 6 months.
- Preventative breast surgery may be considered based on family history.

Patient Education



Pancreatic Cancer

- There are no specific guidelines; however cancer screening may be considered based on family history.

It is important to discuss these options with your provider. Recommendations may change as researchers learn more about how PALB2 mutations impact cancer risk.

Family History

People who have a PALB2 mutation inherited the mutation from either their father or mother. There is a 50/50 (1 in 2) chance to pass down the mutation to each of their children. Both men and women are equally likely to inherit or pass down the mutation.

At-risk relatives may undergo predictive genetic testing for the mutation. This may help determine who needs to adjust their cancer screening and prevention options. If a person did not inherit the PALB2 mutation, screening recommendations will be made based on family history.

Individuals with a PALB2 mutation may want to talk with their partner about testing for the mutation before having children. If both parents have a PALB2 mutation, their child has a risk for inheriting two PALB2 mutations. Fanconi Anemia is caused by an individual inheriting two PALB2 gene mutations. Fanconi Anemia is a rare, childhood blood disorder that causes bone marrow failure, physical abnormalities, and increased risks for cancers.

Resources

FORCE

www.facingyourrisk.org

FORCE is a nonprofit organization with resources for families with an increased risk for developing breast and ovarian cancer. FORCE has education resources and in-person and virtual support programs.

Bright Pink

www.brightpink.org

Bright Pink is a nonprofit organization that provides support and education to women ages 18 to 45 who have a high risk for developing breast and ovarian cancer.

Patient Education



PanCAN (Pancreatic Cancer Action Network)

www.pancan.org

PanCAN is a nonprofit organization dedicated to pancreatic cancer research and education. The website provides lifestyle information, patient resources, and support groups.

The Genetic Information Nondiscrimination Act (GINA)

www.ginahelp.org

This website provides information about the laws protecting against discrimination based on genetic information. It includes information about employers and health insurance, and answers commonly asked questions about the Genetic Information Nondiscrimination Act (GINA).

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