PALB2 WOMEN

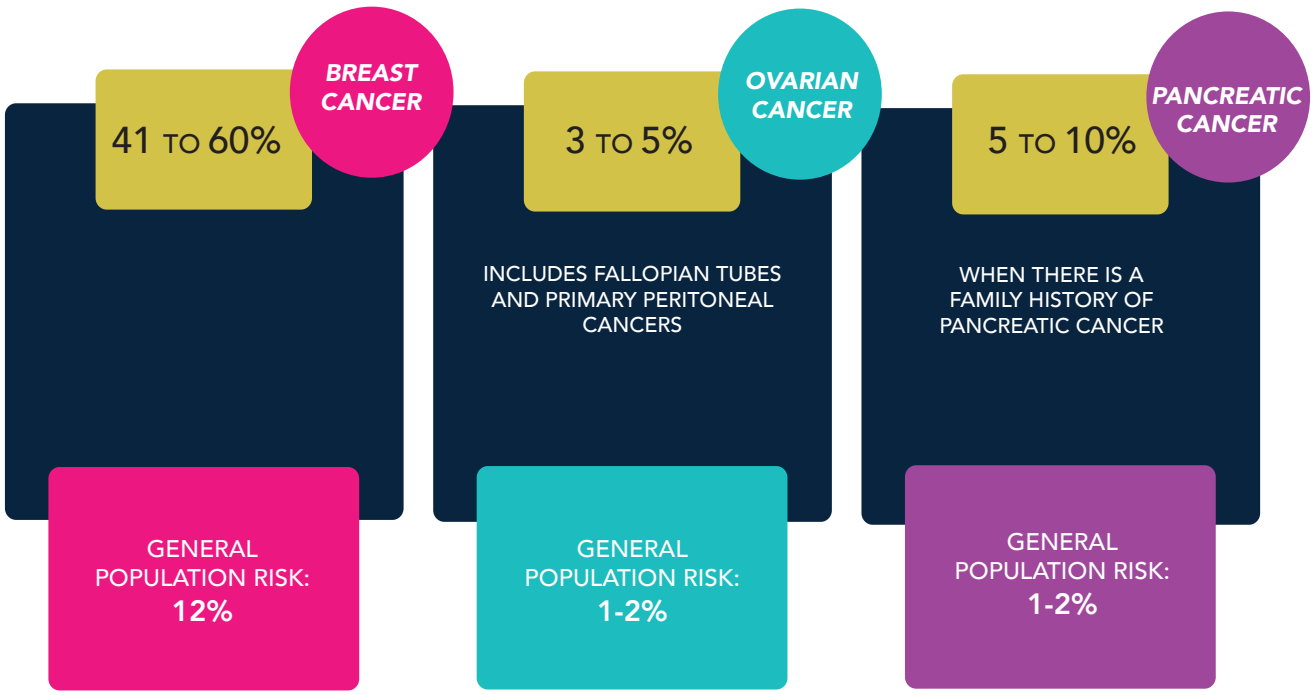
The medical management guidelines from the National Comprehensive Cancer Network (NCCN) for patients with a PALB2 pathogenic / likely pathogenic variant are listed in this document.

This overview is for informational purposes and does not constitute a personalised recommendation. Recommended options may vary based on your personal and family history. Access to some options may also vary from one medical center to another. The specific references should be consulted for more details before developing a treatment plan.

In addition, the information available on hereditary cancer susceptibility genes is constantly evolving and it is recommended to check this information annually as the management guidelines may change in the future.

LIFETIME ASSOCIATED RISKS

(UP TO AGE 75)



OTHER TYPES OF CANCER

Preliminary evidence suggests a possible increased risk for other types of cancer. However, specific risks have not been established and more research is needed to confirm these findings.

References:

Daly M et coll. NCCN Clinical Practice Guidelines in Oncology: Genetic/Familial High-Risk Assessment: Breast, Ovarian and Pancreatic. Version 2.2021. November 20, 2020. http://www.nccn.org

Antoniou AC et al. Breast-cancer risk in families with mutations in PALB2. N Engl J Med. 2014;371:497–506.

Silvestri V, et al. Whole-exome sequencing and targeted gene sequencing provide insights into the role of PALB2 as a male breast cancer susceptibility gene. Cancer. 2017 Jan 1;123(2):210-218.

Quebec Breast Cancer Foundation

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PALB2 WOMEN

**** Genetic testing for the PALB2 gene is recommended for the spouse of an individual with a PALB2 pathogenic/likely pathogenic variant to assess the risk of Fanconi Anemia in this couple's children.**

When both parents carry a PALB2 pathogenic/likely pathogenic variant, their children have a 25% risk of inheriting both PALB2 pathogenic/likely pathogenic variants, which is associated with a rare genetic disease called Fanconi Anemia. This condition is associated with an increased risk of childhood cancers as well as physical anomalies.

BREAST CANCER SCREENING

STARTING AT AGE 30

- MAMMOGRAM WITH CONSIDERATION OF TOMOSYNTHESIS EVERY 12 MONTHS
- CONSIDER BREAST MAGNETIC RESONANCE IMAGING (MRI) WITH CONTRAST EVERY 12 MONTHS

30

75

STARTING AT AGE 75

- MANAGEMENT SHOULD BE DETERMINED ON AN INDIVIDUAL BASIS

BREAST CANCER RISK REDUCTION

SURGERY

PROPHYLACTIC BILATERAL MASTECTOMY (REMOVAL OF THE BREASTS BEFORE CANCER DEVELOPS), WITH OR WITHOUT RECONSTRUCTION, HAS BEEN SHOWN TO REDUCE THE RISK OF DEVELOPING BREAST CANCER BY UP TO 90%.

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OVARIAN CANCER

SCREENING

THERE ARE NO SPECIFIC GUIDELINES AVAILABLE FOR SCREENING FOR OVARIAN CANCER.

FOR INDIVIDUALS WITH FAMILY HISTORY OF OVARIAN CANCER, SCREENING RECOMMENDATIONS MAY BE CONSIDERED:

- CONSIDER TRANSVAGINAL ULTRASOUND AND CA-125 MEASUREMENT EVERY 6 MONTHS.

RISK REDUCTION

REGARDING THE PALB2 GENE, THERE IS CURRENTLY INSUFFICIENT EVIDENCE TO RECOMMEND A PROPHYLACTIC BILATERAL SALPINGO-OOPHORECTOMY, (REMOVAL OF THE FALLOPIAN TUBES AND OVARIES BEFORE CANCER DEVELOPS).

THIS OPTION MAY BE CONSIDERED BASED ON THE FAMILY HISTORY OF OVARIAN CANCER.

PANCREATIC CANCER

SCREENING

CURRENTLY, SCREENING FOR PANCREATIC CANCER IS NOT RECOMMENDED IN ABSENCE OF A FAMILY HISTORY OF PANCREATIC CANCER.

FOR PATHOGENIC/LIKELY PATHOGENIC VARIANT CARRIERS **WITH ≥1** FIRST-DEGREE RELATIVE (PARENT, CHILD OR SIBLING) OR SECOND-DEGREE RELATIVE (GRAND-PARENT, AUNT OR UNCLE, NIECE OR NEPHEW) (ON THE SAME SIDE OF THE FAMILY) DIAGNOSED WITH PANCREATIC CANCER:

STARTING AT AGE 50
OR 10 YEARS BEFORE THE EARLIEST PANCREATIC CANCER IN THE FAMILY

- CONSIDER SCREENING WITH MRI/MAGNETIC RESONANCE CHOLANGIOPANCREATOGRAPHY (MRCP) AND/OR ENDOSCOPIC ULTRASONOGRAPHY (EUS), IN AN EXPERIENCED CENTER, IDEALLY UNDER RESEARCH PROTOCOL.

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