PALB2 WOMEN

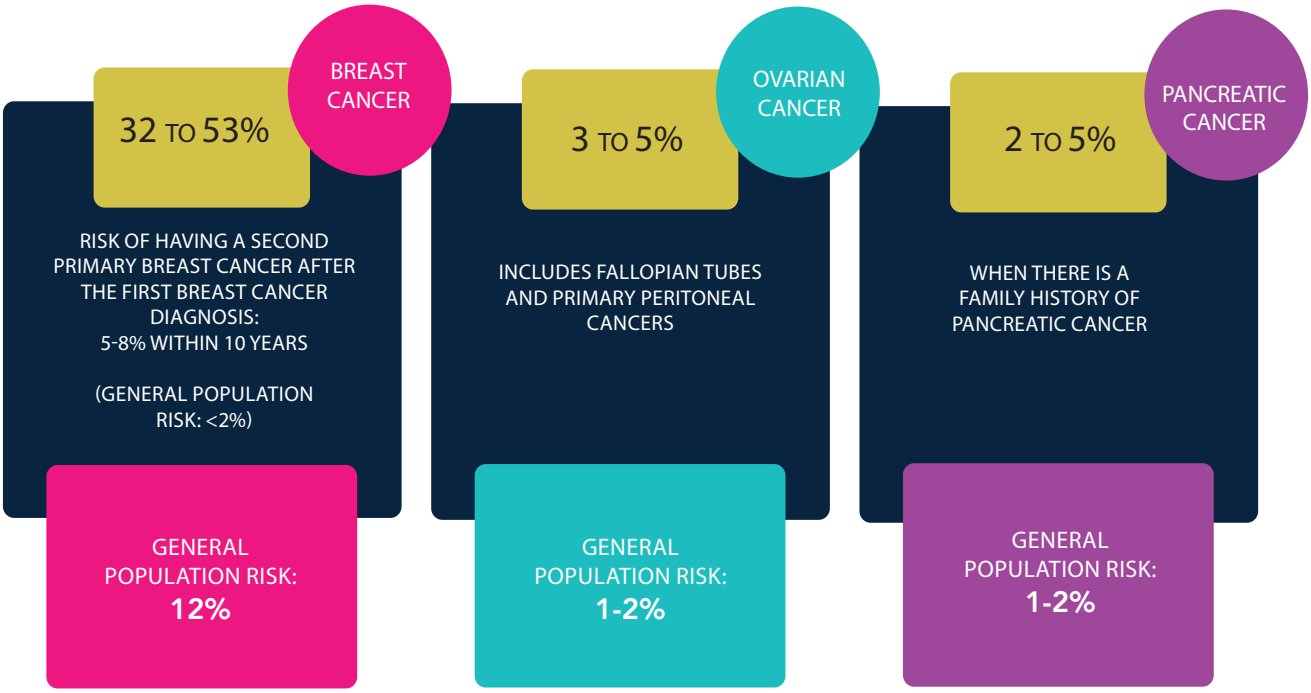
The medical management guidelines from the National Comprehensive Cancer Network (NCCN) for patients with a PALB2 pathogenic / likely pathogenic variant are listed in this document.

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)



References:

Daly M et coll. NCCN Clinical Practice Guidelines in Oncology: Genetic/Familial High-Risk Assessment: Breast, Ovarian and Pancreatic. Version 3.2025-March 6, 2025. http://www.nccn.org

Quebec Breast Cancer Foundation
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** Genetic testing for the PALB2 gene may be considered 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 AND BREAST MAGNETIC RESONANCE IMAGING (MRI) WITH AND WITHOUT CONTRAST EVERY 12 MONTHS

30

75

STARTING AT AGE 75
- MANAGEMENT SHOULD BE DETERMINED ON AN INDIVIDUAL BASIS

BREAST CANCER RISK REDUCTION

SURGERY

DISCUSS THE OPTION OF PROPHYLACTIC BILATERAL MASTECTOMY (REMOVAL OF THE BREASTS BEFORE CANCER DEVELOPS), WITH OR WITHOUT RECONSTRUCTION. THIS OPTION HAS BEEN SHOWN TO SIGNIFICANTLY REDUCE THE RISK OF DEVELOPING BREAST CANCER.

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PALB2 WOMEN

OVARIAN CANCER

RISK REDUCTION

SURGERY

- CONSIDER PROPHYLACTIC BILATERAL SALPINGO-OOPHORECTOMY, (REMOVAL OF THE FALLOPIAN TUBES AND OVARIES BEFORE CANCER DEVELOPS), STARTING AT 45-50 YEARS OLD.
- IF DONE BEFORE NATURAL MENOPAUSE, THIS SURGERY LIKELY REDUCES THE RISK OF BREAST CANCER.

IMPORTANT : THE HISTOLOGY ANALYSIS OF THE FALLOPIAN TUBES AND OVARIES SHOULD BE THOROUGH TO RULE OUT OCCULT CANCER WHICH MAY BE PRESENT IN SOME CASES.

HORMONAL REPLACEMENT THERAPY RECOMMENDATIONS SHOULD BE TAILORED DEPENDING ON PERSONAL HISTORY OF BREAST CANCER AND/OR BREAST CANCER RISK REDUCTION STRATEGIES.
HORMONAL REPLACEMENT THERAPY IS A CONSIDERATION FOR PREMENOPAUSAL WOMEN WHO DID NOT RECEIVE A BREAST CANCER DIAGNOSIS OR HAVE OTHER CONTRAINDICATIONS FOR HORMONAL REPLACEMENT THERAPY.

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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