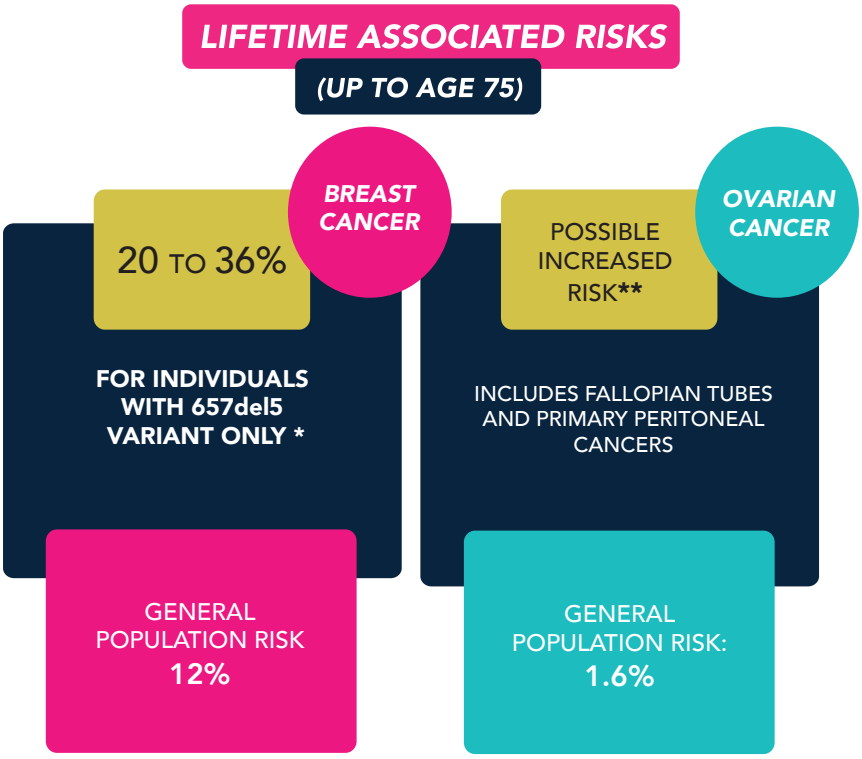
NBN WOMEN

The medical management guidelines from the National Comprehensive Cancer Network (NCCN) for patients with a NBN 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.



*Current data suggest that breast cancer risks are **NOT** increased for other pathogenic / likely pathogenic variants.

OTHER TYPES OF CANCER

****Preliminary evidence suggests a possible increased risk for other types of cancer, including ovarian 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 and Ovarian. Version 1.2020. December 4, 2019. http://www.nccn.org

Quebec Breast Cancer Foundation
Phone : 514 871-1717
Toll Free Number : 1 877 990-7171
Mail : info@rubanrose.org

NBN WOMEN

**** Genetic testing for the NBN gene is recommended for the spouse of an individual with a NBN pathogenic/likely pathogenic variant to assess the risk of Nijmegen breakage syndrome (NBS) in this couple's children.**

When both parents carry a NBN pathogenic/likely pathogenic variant, their children have a 25% risk of inheriting both NBN pathogenic/likely pathogenic variants, which is associated with a rare genetic disease called Nijmegen breakage syndrome (NBS). This condition is associated with physical anomalies, immunodeficiency, increased cancer risk and cognitive impairment.

BREAST CANCER SCREENING

INDIVIDUALIZED
- BREAST AWARENESS
  - ◊ PROMPTLY REPORT ANY CHANGES TO YOUR HEALTHCARE PROVIDER

STARTING AT AGE 40
- MAMMOGRAM WITH CONSIDERATION OF TOMOSYNTHESIS EVERY 12 MONTHS
- CONSIDER BREAST MAGNETIC RESONANCE IMAGING (MRI) WITH CONTRAST EVERY 12 MONTHS

40

75

WHEN GENETIC RISK IS IDENTIFIED
- CLINICAL BREAST EXAM BY A PHYSICIAN EVERY 6 TO 12 MONTHS

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

BREAST CANCER RISK REDUCTION

SURGERY

REGARDING THE NBN GENE, THERE IS CURRENTLY INSUFFICIENT EVIDENCE TO RECOMMEND A PROPHYLACTIC BILATERAL MASTECTOMY (REMOVAL OF THE BREASTS BEFORE CANCER DEVELOPS).

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

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OVARIAN CANCER

SCREENING

CURRENT KNOWLEDGE IS INSUFFICIENT TO ESTABLISH SPECIFIC GUIDELINES FOR SCREENING FOR THE RISK OF OVARIAN CANCER ASSOCIATED WITH THE NBN GENE.

- IT IS SUGGESTED TO FOLLOW THE RECOMMENDATIONS FOR PREVENTION AND SCREENING FOR THE GENERAL POPULATION.
- **FOR INDIVIDUALS WITH A FAMILY HISTORY OF OVARIAN CANCER**, SCREENING RECOMMENDATIONS SHOULD BE INITIATED AT A YOUNGER AGE.
  - ◊ CONSIDER TRANSVAGINAL ULTRASOUND AND CA-125 MEASUREMENT EVERY 6 MONTHS.

RISK REDUCTION

REGARDING THE NBN 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.

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