



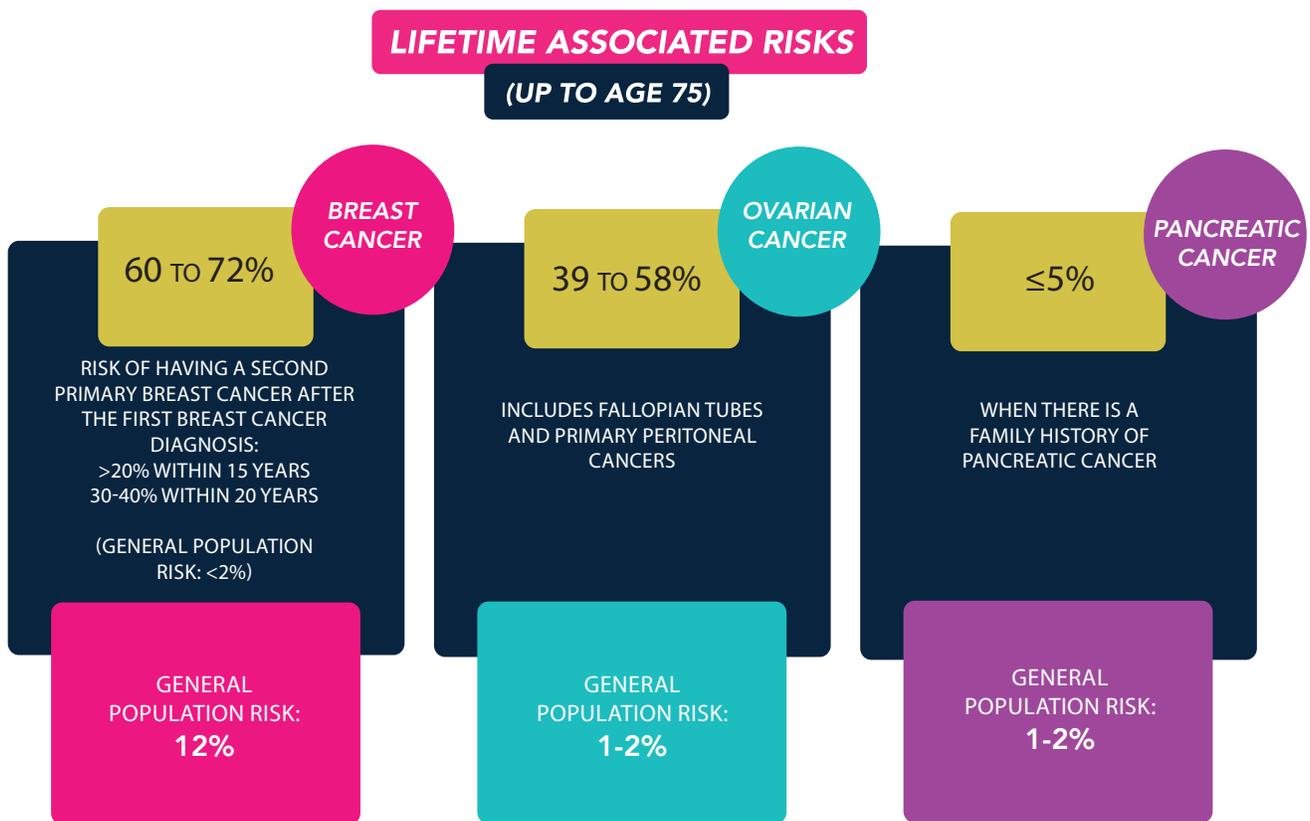
# BRCA1 WOMEN



The medical management guidelines from the National Comprehensive Cancer Network (NCCN) for patients with a BRCA1 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.**



#### 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>

Petrucelli N, Daly MB, Pal T. BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer. 1998 Sep 4 [Updated 2023 September 21]. In : Adam MP, Everman DB, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA) : University of Washington, Seattle; 1993-2022. <https://www.ncbi.nlm.nih.gov/books/NBK1247>



# BRCA1 WOMEN



\*\* Genetic testing for the BRCA1 gene may be considered for the spouse of an individual with a BRCA1 pathogenic/likely pathogenic variant to assess the risk of Fanconi Anemia in this couple's children.

When both parents carry a BRCA1 pathogenic/likely pathogenic variant, their children have a 25% risk of inheriting both BRCA1 pathogenic/likely pathogenic variants, which is associated with a rare genetic disease called Fanconi Anemia complementation group S (FANCS). This condition is associated with an increased risk of childhood cancers as well as physical anomalies.

## BREAST CANCER

### SCREENING

#### STARTING AT AGE 18

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

18

#### STARTING AT AGE 30

- BREAST MRI WITH AND WITHOUT CONTRAST **AND** MAMMOGRAM EVERY 12 MONTHS (ALTERNATING EACH EXAM EVERY 6 MONTHS)

30

25

#### STARTING AT AGE 25

OR EARLIER IF PRESENCE OF A FAMILY HISTORY OF BREAST CANCER BEFORE AGE 30

- CLINICAL BREAST EXAM BY A PHYSICIAN EVERY 6 TO 12 MONTHS
- BREAST MAGNETIC RESONANCE IMAGING (MRI) WITH AND WITHOUT CONTRAST **OR** (ONLY IF MRI IS NOT AVAILABLE), MAMMOGRAM EVERY 12 MONTHS

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.

WOMEN TREATED FOR BREAST CANCER AND WHO HAVE NOT HAD A BILATERAL MASTECTOMY, SCREENING ANNUALLY WITH MAMMOGRAM AND BREAST MAGNETIC RESONANCE IMAGING (MRI) WITH AND WITHOUT CONTRAST IS RECOMMENDED.

#### CHEMOPREVENTION

TAMOXIFEN AND RALOXIFEN ARE MEDICATIONS USED TO TREAT BREAST CANCER. THEY ALSO HAVE BEEN SHOWN TO REDUCE THE RISK FOR CERTAIN TYPES OF BREAST CANCER.



# BRCA1 WOMEN



## OVARIAN CANCER

### RISK REDUCTION

#### SURGERY

- PROPHYLACTIC BILATERAL SALPINGO-OOPHORECTOMY, (REMOVAL OF THE FALLOPIAN TUBES AND OVARIES BEFORE CANCER DEVELOPS), IS RECOMMENDED BETWEEN THE AGES OF 35 AND 40.
- IF DONE BEFORE NATURAL MENOPAUSE, THIS SURGERY LIKELY REDUCES THE RISK OF BREAST CANCER.
- HYSTERECTOMY (REMOVAL OF THE UTERUS) MAY ALSO BE CONSIDERED. RECENT DATA SUGGESTS A SLIGHTLY INCREASED RISK OF SEROUS UTERINE CANCER.
- CA-125 AND PELVIC ULTRASOUND ARE RECOMMENDED FOR PREOPERATIVE PLANNING.

**NOTE :** SALPINGECTOMY ALONE (REMOVAL OF THE FALLOPIAN TUBES WHILE KEEPING THE OVARIES) IS AN OPTION FOR PREMENOPAUSAL WOMEN WHO ARE NOT READY FOR OOPHORECTOMY. COMPLETION OOPHORECTOMY IS RECOMMENDED ACCORDING AS LISTED ABOVE.

**IMPORTANT :** THE HISTOLOGY ANALYSIS OF THE FALLOPIAN TUBES AND OVARIES SHOULD BE THOROUGH TO RULE OUT OCCULT CANCER WHICH MAY BE PRESENT IN ABOUT 5% OF 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.

- WOMEN WHO OPT FOR HYSTERECTOMY ARE CANDIDATES FOR HORMONAL REPLACEMENT THERAPY WITH ESTROGEN ALONE, WHICH IS ASSOCIATED WITH A REDUCED RISK OF BREAST CANCER.
- WHEN UTERUS IS PRESENT, CONSIDER OPTIONS FOR HORMONAL REPLACEMENT:
  - LEVONORGESTREL INTRAUTERINE DEVICE WITH ORAL OR TRANSDERMAL ESTROGENE.
  - COMBINATION OF ESTROGEN AND PROGESTERONE WITH AWARENESS OF ENDOMETRIAL CANCER RISK.
  - COMBINATION ESTROGEN WITH SELECTIVE ESTROGEN RECEPTOR MODULATOR
  - BIRTH CONTROL PILLS TAKEN CONTINUOUSLY WITHOUT PLACEBO WEEK.

#### CHEMOPREVENTION

BIRTH CONTROL PILLS MAY ALSO BE CONSIDERED TO REDUCE SIGNIFICANTLY THE RISK OF OVARIAN CANCER. LEVONORGESTREL INTRAUTERINE DEVICE HAS BEEN SHOWN TO REDUCE THE RISK 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  $\geq 1$  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.