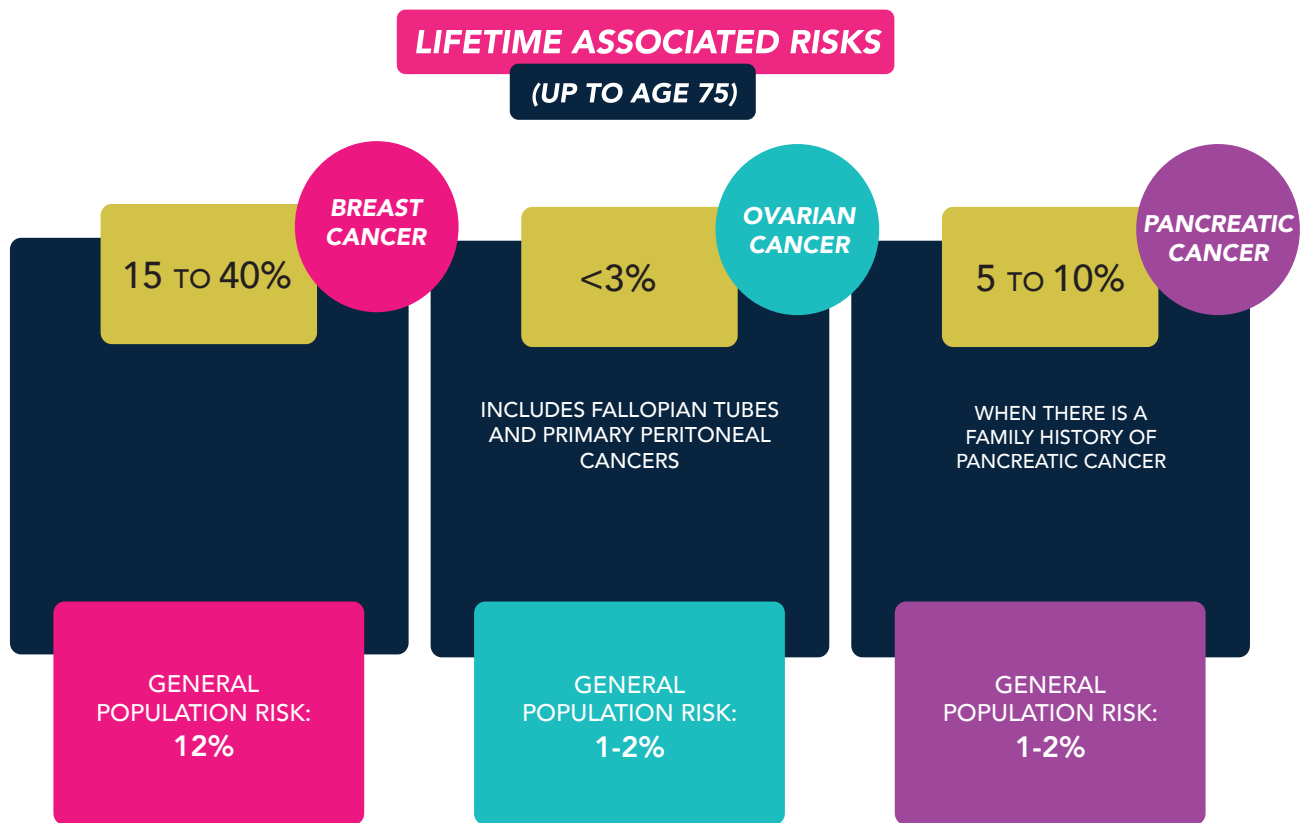




The medical management guidelines from the National Comprehensive Cancer Network (NCCN) for patients with an ATM pathogenic / likely pathogenic variant are listed in this document.

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 2.2021. November 20, 2020. <http://www.nccn.org>

Gatti R, Perlman S. Ataxia Telangiectasia. 1999 Mar 19 [Updated 2016 Oct 27]. In : Pagon RA, Adam MP, Ardinger HH, et al., editors. GeneReviews® [Internet]. Seattle (WA) : University of Washington, Seattle; 1993-2017. <https://www.ncbi.nlm.nih.gov/books/NBK26468>



**\*\* Genetic testing for the ATM gene is recommended for the spouse of an individual with an ATM pathogenic/likely pathogenic variant to assess the risk of ataxia-telangiectasia in this couple's children.**

When both parents carry an ATM pathogenic/likely pathogenic variant, their children have a 25% risk of inheriting both ATM pathogenic/likely pathogenic variants, which is associated with a rare genetic disease called ataxia-telangiectasia. This condition is associated with an increased risk of childhood cancers as well as disorder of the nervous and immune systems.

## BREAST CANCER

### SCREENING

#### STARTING AT AGE 40

OR EARLIER ACCORDING TO THE FAMILY HISTORY OF BREAST CANCER.

- CLINICAL BREAST EXAM BY A PHYSICIAN EVERY 6 TO 12 MONTHS.
- MAMMOGRAM WITH CONSIDERATION OF TOMOSYNTHESIS EVERY 12 MONTHS.
- CONSIDER BREAST MAGNETIC RESONANCE IMAGING (MRI) WITH CONTRAST EVERY 12 MONTHS.

40

75

#### STARTING AT AGE 75

- MANAGEMENT SHOULD BE DETERMINED ON AN INDIVIDUAL BASIS

## BREAST CANCER

### RISK REDUCTION

#### SURGERY

REGARDING THE ATM 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.



## OVARIAN CANCER

### SCREENING

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