TP53 WOMEN

The medical management guidelines from the National Comprehensive Cancer Network (NCCN) for patients with a TP53 pathogenic / likely pathogenic variant are listed in this document.

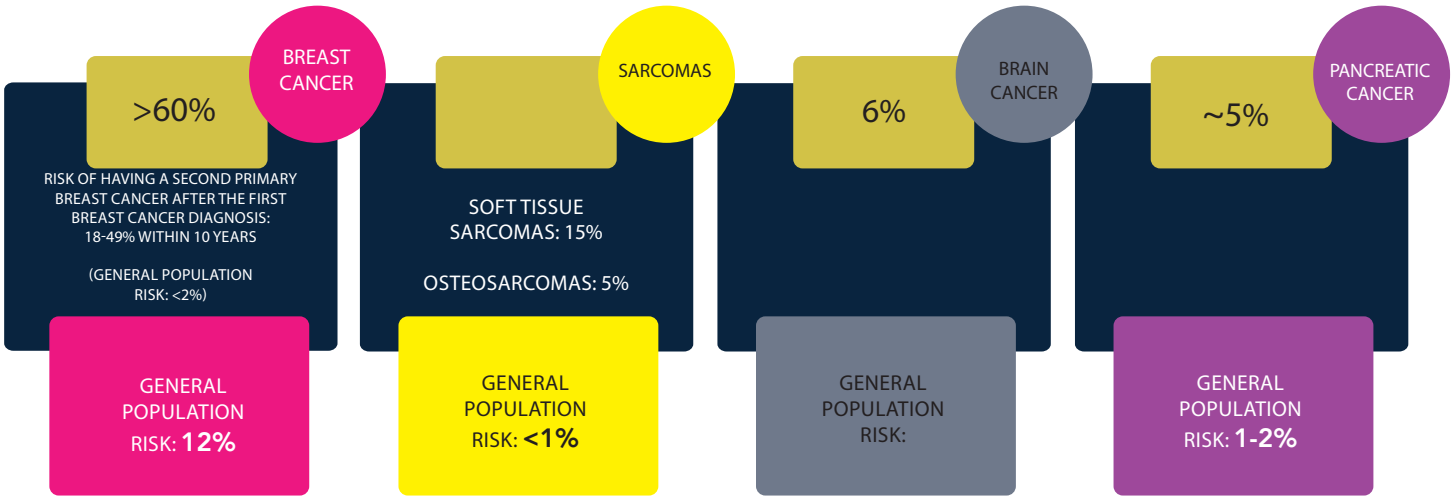
This overview is for informational purposes and does not constitute a personalised recommendation. Recommended options may vary based on your personal and family history. Access to some options may also vary from one medical center to another. The specific references should be consulted for more details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

Li-Fraumeni syndrome is rare. An individual who is a carrier of a TP53 pathogenic / likely pathogenic variant has an elevated risk of developing a cancer during their lifetime, including pediatric cancer (in children).



LIFETIME ASSOCIATED RISK OF DEVELOPING CANCER : >90%

RISK OF HAVING A SECOND PRIMARY CANCER WITHIN 10 YEARS FOLLOWING THE FIRST DIAGNOSIS: 50%.

MOST FREQUENT CANCERS:

- BREAST CANCER
- BRAIN CANCER
- SARCOMAS (SOFT TISSUE AND BONE)
- ADRENOCORTICAL CARCINOMA (ADRENAL GLANDS)

OTHER ASSOCIATED CANCERS:

- OVARIAN CANCER
- UTERINE CANCER
- COLORECTAL CANCER
- STOMACH CANCER (GASTRIC)
- PANCREATIC CANCER
- LUNG CANCER (BRONCHIOALVEOLAR)
- LEUKEMIA

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

TP53 WOMEN

BECAUSE THE MANAGEMENT OF INDIVIDUALS WITH LI-FRAUMENI SYNDROME IS COMPLEX, IT IS PREFERRED THAT THEY BE FOLLOWED AT A CENTRE WITH EXPERTISE WITH THIS SYNDROME.

THERAPEUTIC RADIOTHERAPY SHOULD BE AVOIDED WHEN POSSIBLE. DIAGNOSTIC RADIATION SHOULD BE MINIMISED.

BREAST CANCER

SCREENING

STARTING AT AGE 18
- BREAST AWARENESS
  - ◊ PROMPTLY REPORT ANY CHANGES TO YOUR HEALTHCARE PROVIDER

STARTING AT AGE 20
OR AT THE AGE OF THE EARLIEST DIAGNOSIS OF BREAST CANCER IN THE FAMILY
- CLINICAL BREAST EXAM BY A PHYSICIAN EVERY 6 TO 12 MONTHS
- BREAST MAGNETIC RESONANCE IMAGING (MRI) WITH AND WITHOUT CONTRAST EVERY 12 MONTHS.

18 20 30 75

STARTING AT AGE 30
- BREAST MRI WITH CONTRAST AND MAMMOGRAM EVERY 12 MONTHS (ALTERNATING EACH EXAM EVERY 6 MONTHS).

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

OTHER TYPES OF CANCER

SCREENING

STARTING AFTER BIRTH
- COMPREHENSIVE PHYSICAL EXAM INCLUDING NEUROLOGIC EXAMINATION EVERY 6 TO 12 MONTHS UNTIL AGE 18
- WHOLE BODY MRI AND BRAIN MRI (EITHER AT THE SAME TIME OF WHOLE BODY MRI OR SEPARATELY) EVERY YEAR
- ABDOMINAL AND PELVIC ULTRASOUND EVERY 3 TO 4 MONTHS UNTIL AGE 18
- BLOOD TESTS (COMPLETE BLOOD COUNT, ERYTHROCYTE SEDIMENTATION RATE, LACTASE DESHYDROGENASE) EVERY 4 MONTHS

18 25

STARTING AT AGE 18
- DERMATOLOGY EXAMINATION EVERY YEAR
- COMPREHENSIVE PHYSICAL EXAM EVERY 6 TO 12 MONTHS
- NEUROLOGIC EXAMINATION EVERY 12 MONTHS
- ABDOMINAL AND PELVIC ULTRASOUND EVERY YEAR

STARTING AT AGE 25
OR 5 YEARS EARLIER THAN THE YOUNGEST COLORECTAL CANCER OR GASTRIC CANCER DIAGNOSIS IN THE FAMILY
- COLONOSCOPY AND UPPER ENDOSCOPY EVERY 2 TO 5 YEARS

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

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.

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.

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

Schneider K, Zelley K, Nichols KE, et al. Li-Fraumeni Syndrome. 1999 Jan 19 [Updated 2024 Sep 5]. In: Adam MP, Feldman J, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2025. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1311/

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