



# 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.

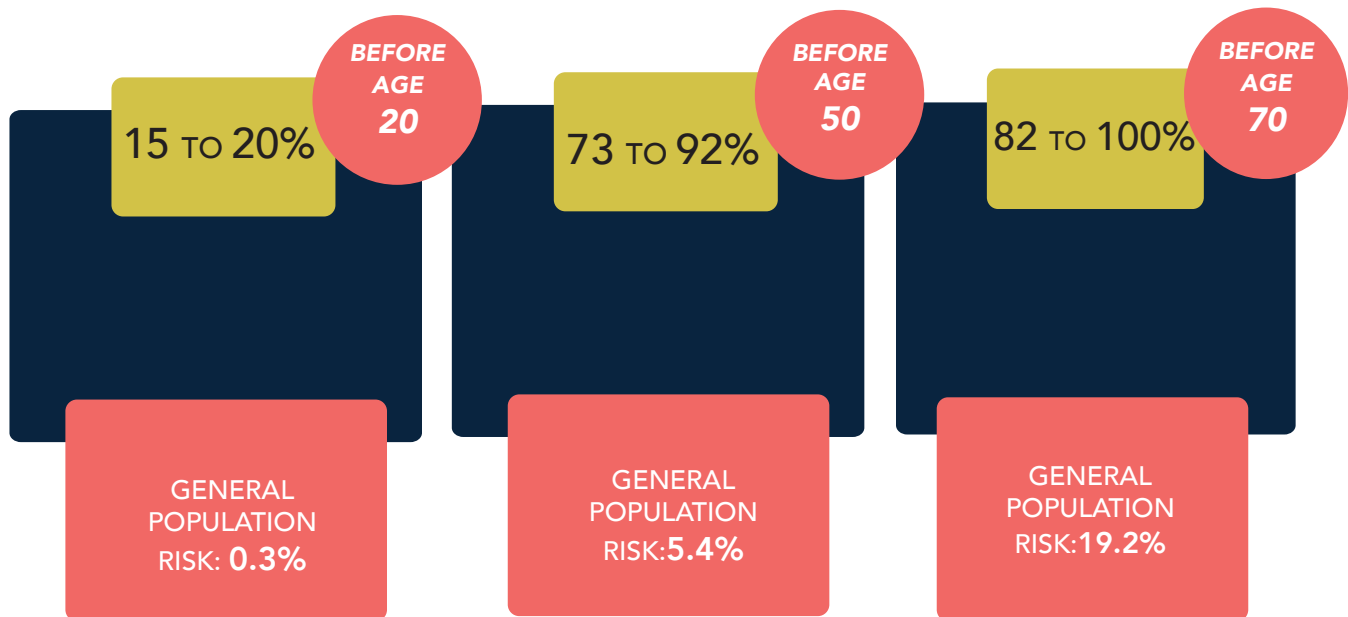
This overview is for informational purposes and does not constitute a personalised recommendation. **Recommended options may vary based on your personal and family history. Access to some options may also vary from one medical center to another.** The specific references should be consulted for more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



**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



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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

18

#### STARTING AT AGE 20

- CLINICAL BREAST EXAM BY A PHYSICIAN EVERY 6 TO 12 MONTHS
- BREAST MAGNETIC RESONANCE IMAGING (MRI) WITH CONTRAST **OR** (ONLY IF MRI IS NOT AVAILABLE), MAMMOGRAM WITH CONSIDERATION OF TOMOSYNTHESIS EVERY 12 MONTHS.

20

30

#### STARTING AT AGE 30

- BREAST MRI WITH CONTRAST **AND** MAMMOGRAM WITH CONSIDERATION OF TOMOSYNTHESIS EVERY 12 MONTHS (ALTERNATING EACH EXAM EVERY 6 MONTHS).

75

#### 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 3 TO 4 MONTHS UNTIL AGE 18
- WHOLE BODY MRI AND BRAIN MRI (EITHER AT THE SAME TIME OF WHOLE BODY MRI OR SEPARATELY) EVERY YEAR
- BLOOD TESTS (COMPLETE BLOOD COUNT, ERYTHROCYTE SEDIMENTATION RATE, LACTATE DEHYDROGENASE) EVERY 4 MONTHS

18

#### STARTING AT AGE 18

- DERMATOLOGY EXAMINATION EVERY YEAR
- COMPREHENSIVE PHYSICAL EXAM INCLUDING NEUROLOGIC EXAMINATION EVERY 6 TO 12 MONTHS

25

#### STARTING AT AGE 25

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



## BREAST CANCER

### RISK REDUCTION

#### SURGERY

PROPHYLACTIC BILATERAL MASTECTOMY (REMOVAL OF THE BREASTS BEFORE CANCER DEVELOPS), WITH OR WITHOUT RECONSTRUCTION, HAS BEEN SHOWN TO REDUCE THE RISK OF DEVELOPING BREAST CANCER BY UP TO 90%.

## 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.

#### 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>

Schneider K, Zelle K, Nichols KE, et al. Li-Fraumeni Syndrome. 1999 Jan 19 [Updated 2013 Apr 11]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2019. <https://www.ncbi.nlm.nih.gov/books/NBK1311/>