



TP53 MEN



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.

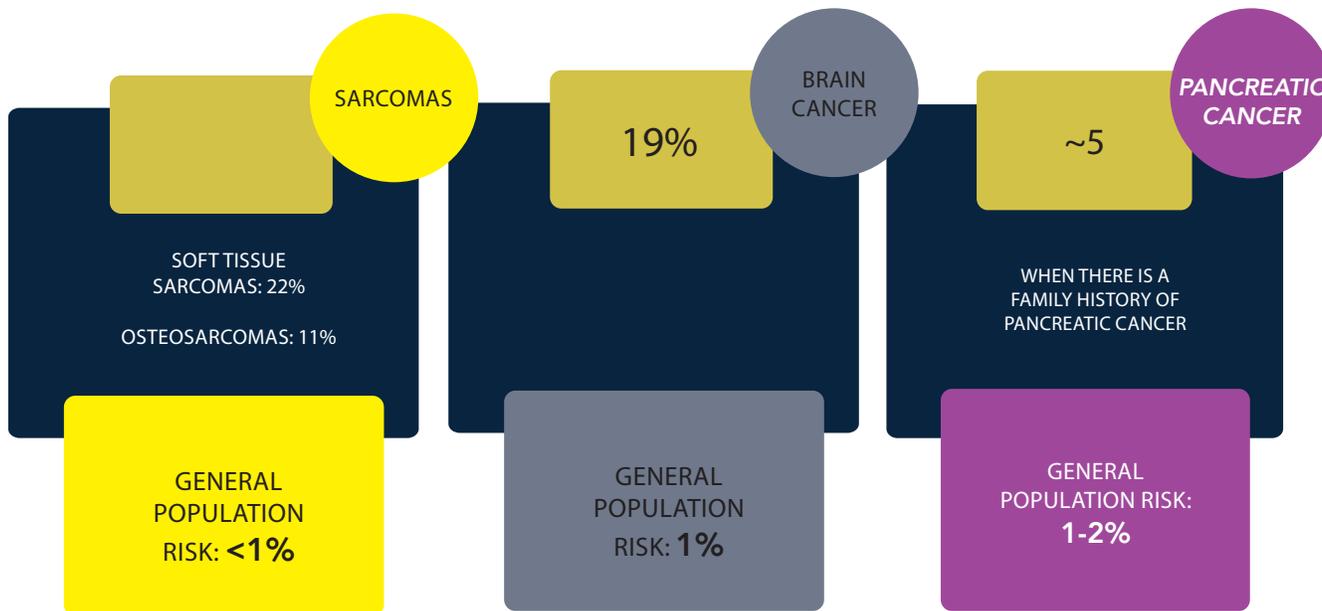
This overview is for informational purposes and does not constitute a personalised recommendation. **Recommended options may vary based on your personal and family history. Access to some options may also vary from one medical center to another.** The specific references should be consulted for more 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 : >70%

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

MOST FREQUENT CANCERS:

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

OTHER ASSOCIATED CANCERS:

- 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.

OTHER TYPES OF CANCER

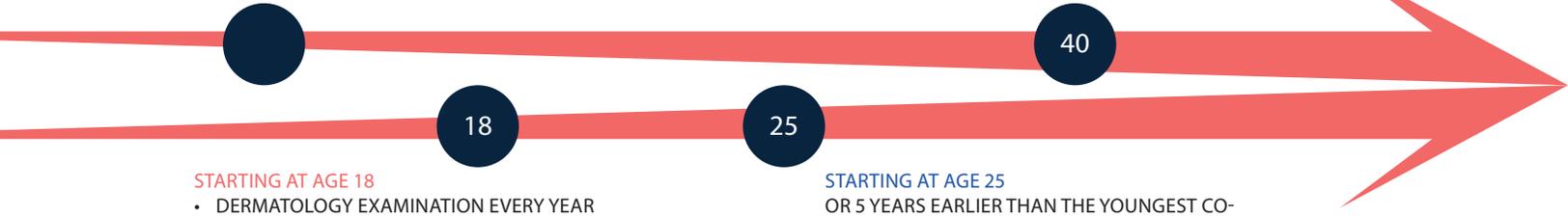
SCREENING

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, LACTATE DEHYDROGENASE) EVERY 4 MONTHS

STARTING AT AGE 40

- PSA BLOOD TEST) EVERY 12 MONTHS



18

25

40

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

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