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.

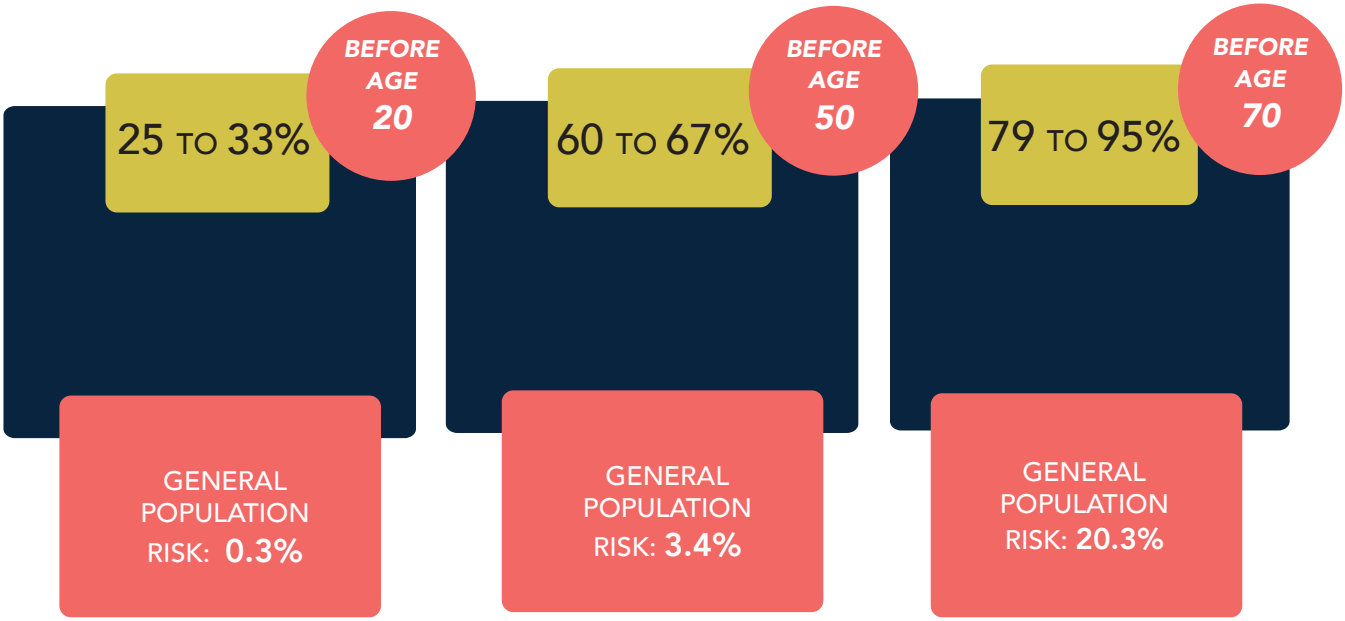
This overview is for informational purposes and does not constitute a personalised recommendation. Recommended options may vary based on your personal and family history. Access to some options may also vary from one medical center to another. The specific references should be consulted for more details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



RISQUE D'ÊTRE ATTEINT D'UN 2ÈME CANCER AU COURS DES 10 ANNÉES SUIVANT LE PREMIER DIAGNOSTIC : 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 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, LACTASE DESHYDROGENASE) 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

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

Schneider K, Zelley 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

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