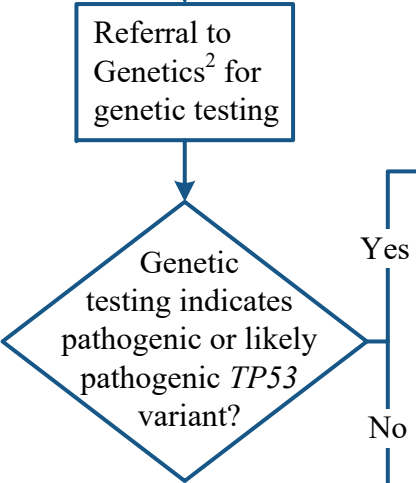


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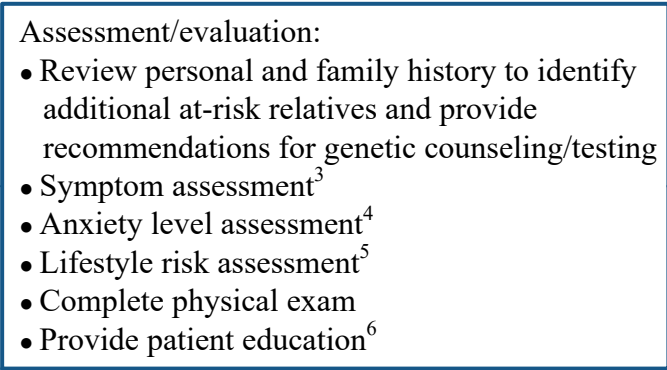
Note: Screening is only intended for asymptomatic individuals. Individuals undergoing Li-Fraumeni Syndrome screening should have a 10-year life expectancy and no co-morbidities that would limit the diagnostic evaluation or treatment of any identified problem. The screening technique should be performed with a consistent technique and process.

PRESENTATION

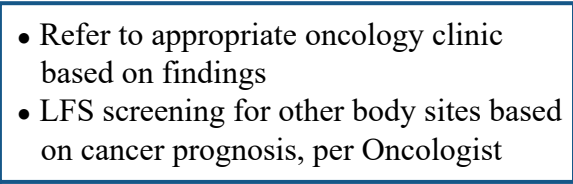
Personal and/or family history suggestive of Li-Fraumeni Syndrome (LFS) or family member diagnosed with LFS through genetic testing¹



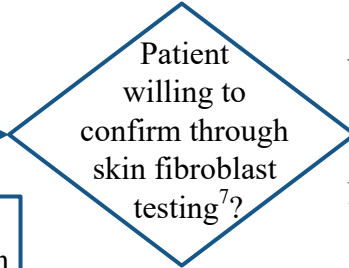
ASSESSMENT



RECOMMENDATION



Genetic test results do not clearly indicate whether the *TP53* variant is germline, mosaic or somatic in nature



Recommendations will be specific to the individual

¹ Personal and/or family history of LFS-associated cancers including adrenocortical carcinomas, breast cancer, central nervous system tumors, osteosarcomas, and soft-tissue sarcomas. Additional LFS-associated cancers include leukemia, lymphoma, gastrointestinal cancers, cancers of head and neck, kidney, larynx, lung, skin (e.g., melanoma), ovary, pancreas, prostate, testis, and thyroid. See CRIT-7 LFS testing criteria within the NCCN guidelines.

² Patients will be referred to Genetics within their home center. If the patient is new or does not have a genetics counselor assigned to their home center, they can be referred through any home center.

³ Refer to Patient Education: [Li-Fraumeni Syndrome Screening Program - Adult Screening Program](#)

⁴ If moderate to severe anxiety related to LFS screening is identified, refer for psychiatric evaluation and/or counseling

⁵ See [Physical Activity](#), [Nutrition](#), [Obesity Screening and Management](#), and [Tobacco Cessation Treatment](#) algorithms; ongoing reassessment of lifestyle risks should be a part of routine clinical practice

⁶ Refer to Patient Education: [Li-Fraumeni Syndrome Screening Program - Adult Screening Guidelines](#)

⁷ Skin fibroblast testing requires a skin punch biopsy

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Li-Fraumeni Syndrome Screening Program - Adult Screening Guidelines

Cancer	Exams and Tests	Frequency
General	Complete physical exam including neurological examination	Every 6-12 months
Adrenocortical Tumor (ACT)	<div><div>• MRI whole body¹ Assess for clinical signs/symptoms: Females only: • Hirsutism • New changes in menstrual cycle regularity If patient reports any of the above order the following ACT screening labs: DHEA-S, ACTH, total testosterone, total cortisol, BMP and referral/discussion with Endocrinology, as needed.</div><div>Males and females: • New onset or newly uncontrolled hypertension, new onset or newly uncontrolled diabetes mellitus, unexplained weight gain, new lower extremity edema, new hypokalemia, new acne</div></div>	Annually
Brain	MRI ^{1,2} brain	Annually
Breast (begin at age 20-25 years old)	Clinical breast exam (begin at age 20 years old)	Every 6-12 months
	<div>• Breast awareness (begin at age 18 years old) • Mammogram (begin at age 30 years old) • MRI¹ breast with and without contrast (begin at age 20 years old)</div>	Annually (alternating every 6 months)
	<div>• Consider surgical removal of both breasts to prevent cancer (bilateral prophylactic mastectomy) • For women treated for breast cancer, screening of remaining breast tissue should continue</div>	Age and patient appropriate
Colon (begin at age 25 years old or 5 years before earliest known colon/gastric cancer diagnosis in family history, whichever comes first)	<div>• Colonoscopy • Esophagogastroduodenoscopy (EGD)</div>	Every 2-5 years
Leukemia/Lymphoma	CBC with differential	Annually
Melanoma	Skin exam – see Skin Cancer Screening algorithm	Annually
Pancreas ³	<div>• CA 19-9 • Amylase • MRI whole body¹ • HgbA1c • Lipase • Fasting glucose</div>	Annually
Prostate	PSA (begin at age 40 years old)	Annually
Sarcoma	MRI whole body ¹	Annually

DHEA-S = dehydroepiandrosterone-sulfate ACTH = adrenocorticotrop hormone BMP = basic metabolic panel PDAC = pancreatic ductal adenocarcinoma PSA = prostate specific antigen

¹ MRI of the whole body and brain are both performed on an annual basis, staggered with a six month interval in between. MRI breast with and without contrast should be performed at the same time as the MRI brain (but on different days due to the contrast dose).

² The first MRI brain should be performed with and without contrast using gadopdeclinol; if normal, MRI brain without contrast should be performed thereafter. If patient has a history of malignancy, all brain MRIs should be performed with and without contrast.

³ For patients with a family history of PDAC on affected side [1 first-degree relative (FDR) or 1 second-degree relative (SDR)], see [Pancreatic Cancer Screening algorithm](#)

Department of Clinical Effectiveness V4

Approved by the Executive Committee of the Medical Staff on 12/17/2024

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SUGGESTED READINGS

- Kratz, C. P., Achatz, M. I., Brugières, L., Frebourg, T., Garber, J. E., Greer, M. L. C., . . . Malkin, D. (2017). Cancer screening recommendations for individuals with Li-Fraumeni Syndrome. *Clinical Cancer Research*, 23(11), e38-e45. <https://doi.org/10.1158/1078-0432.CCR-17-0408>
- National Comprehensive Cancer Network. (2024). *Genetic/familial high-risk assessment: Breast, ovarian and pancreatic* (NCCN Guideline Version 1.2025). Retrieved from https://www.nccn.org/professionals/physician_gls/pdf/genetics_bop.pdf
- Saya, S., Killick, E., Thomas, S., Taylor, N., Bancroft, E. K., Rothwell, J., . . . Eeles, R. A. (2017). Baseline results from the UK SIGNIFY study: A whole-body MRI screening study in TP53 mutation carriers and matched controls. *Familial Cancer*, 16(3), 433-440. <https://doi.org/10.1007/s10689-017-9965-1>
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DEVELOPMENT CREDITS

This screening algorithm is based on majority expert opinion of the Li-Fraumeni Syndrome workgroup at the University of Texas MD Anderson Cancer Center. It was developed using a multidisciplinary approach that included input from the following:

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