

# Li-Fraumeni Syndrome (LFS) Fact Sheet for Medical Providers

- LFS is a hereditary cancer predisposition syndrome caused by inherited or de novo pathogenic variants (also called mutations) in one copy of the **TP53** tumor suppressor gene, present in the patient from birth (i.e. germline variant).
- People with LFS only have one fully functional **TP53** gene to protect against accumulation of further cancer-causing genetic mutations.
- Risk for rare and common cancers is significantly elevated for both **children and adults** with LFS, leading to significant screening recommendations. Below: Approximate percentage of males and females with a first primary cancer by age (PMIDs: [21779515](#), [27496084](#)) and summary of recommended screening modalities by age.

	15 years	30 years	45 years	60 years
Males	15-25%	20-40%	40-60%	>70%
Females	5-15%	40-50%	75-90%	>90%

	Abdominal Ultrasound	Complete Physical +/- Bloodwork	Whole-body MRI	Brain MRI	Dermatology	Endoscopy/Colonoscopy	Breast MRI (Females)	Mammogram (Females)
Age	Pediatric q3-4 mos	All ages; peds: q3-4 mos; adults: annual	All ages annual	All ages annual	18+ years annual	25+ years q2-5 yrs	20-75 years annual	30-75 years annual

- The **most common** cancers seen in LFS are premenopausal breast cancer, soft tissue sarcoma and osteosarcoma, brain cancer, and adrenocortical carcinoma (especially pediatric). Hematological cancers are also common.
- Not everyone with LFS will develop a cancer; others will develop multiple cancers.
- **Medical Providers caring for patients with LFS should. . .**
  - Maintain a high index of **suspicion of malignancy** during office visits.
  - Be familiar with LFS cancer **screening recommendations** and coordinate with other specialists as necessary to ensure the patient has access to appropriate screening.
  - Connect the patient with an **LFS clinic** or a **local provider** who is willing to coordinate recommended screening scans, including annual whole-body MRI or a suitable alternative if whole-body MRI is unavailable.
  - **Avoid ordering scans involving radiation** except when medically necessary.
  - **Be cognizant of psychological stresses** individuals with LFS may experience both chronically and acutely, particularly surrounding periods of screening or diagnosis, and consider referral to a **mental health** provider as necessary.

## MORE INFORMATION



### SUMMARIES

GeneReviews: <https://www.ncbi.nlm.nih.gov/books/NBK1311/>  
 UpToDate: <https://www.uptodate.com/contents/li-fraumeni-syndrome>  
 PubMed PMIDs: [21779515](#), [27496084](#), [26014290](#), [29076966](#)

### QUESTIONS & SUPPORT

Your institution's cancer genetic counselor  
 A local cancer genetic counselor (<https://www.findageneticcounselor.org>)  
 Li-Fraumeni Syndrome Association (LFS) Genetic Counseling Advisory Group and Medical Advisory Board  
<https://www.lfsassociation.org/genetic-counseling-advisory-group/>  
<https://www.lfsassociation.org/medical-advisory-board/>

### SCREENING GUIDELINES

**Adult:** NCCN Guidelines: [Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic](#)  
**Pediatric:** Kratz CP, Achatz MI, Brugières L, et al. Clin Cancer Res. 2017;23(11):e38-e45. doi:10.1158/1078-0432.CCR-17-0408.  
 PMID: [28572266](#)  
**Whole Body MRI Publications:** PMIDs: [33151095](#), [28572262](#), [28772291](#)

### LFS CLINICS

<https://www.lfsassociation.org/medical-resources/>; <https://www.lfsassociation.org/medical-resources/treatment-facilities/>  
[https://www.lfsassociation.org/wp-content/uploads/2020/06/LFS\\_Genetic\\_Counselor\\_Directory\\_6320.pdf](https://www.lfsassociation.org/wp-content/uploads/2020/06/LFS_Genetic_Counselor_Directory_6320.pdf)