**Li-Fraumeni Syndrome Screening - Adult**

**PRESENTATION**

- Personal and/or family history indicative of Li-Fraumeni syndrome

**ASSESSMENT**

- Genetic testing indicates germline p53 mutation?
  - Yes
    - Assessment/evaluation
      - Complete physical exam
      - Symptom assessment; review findings
      - Psychiatric evaluation and counseling evaluation
      - Lifestyle risk assessment
    - Positive clinical symptom?
      - Yes
        - Refer to appropriate oncology clinic based on clinical symptom
        - Li-Fraumeni syndrome screening for other body sites based on cancer prognosis per oncologist
      - No
        - See Page 2 for screening guidelines
    - No
      - Cancer screening may be tailored based on personal and family history, per discussion with Genetic Counselor
  - No

**RECOMMENDATION**

- **Positive clinical symptom?**
  - Yes
  - No

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1. Patient Education - Li-Fraumeni Syndrome Education and Early Detection (LEAD) Adult Screening Program
2. See Physical Activity, Nutrition, and Tobacco Cessation algorithms; ongoing reassessment of lifestyle risks should be a part of routine clinical practice

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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.
# Li-Fraumeni Syndrome Education and Early Detection (LEAD) - Adult Screening Guidelines

<table>
<thead>
<tr>
<th>Cancer</th>
<th>Exams and Tests</th>
<th>How Often</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>General</strong></td>
<td>A complete physical exam and check of these body systems</td>
<td>Every 6 months</td>
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<tr>
<td></td>
<td>• Brain</td>
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<td></td>
<td>• Thyroid</td>
<td></td>
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<tr>
<td><strong>Adrenocortical Tumor (ACT)</strong></td>
<td>• Whole body MRI&lt;sup&gt;1&lt;/sup&gt;</td>
<td>Annually</td>
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<tr>
<td></td>
<td>• Blood tests: DHEA-S, ACTH, testosterone</td>
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<tr>
<td><strong>Breast (begin at age 20-25 years old)</strong></td>
<td>Clinical breast exam by doctor</td>
<td>Every 6 months</td>
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<tr>
<td></td>
<td>Mammogram and MRI&lt;sup&gt;1&lt;/sup&gt;</td>
<td>Annually</td>
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<tr>
<td></td>
<td>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.</td>
<td>Age and patient appropriate</td>
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<tr>
<td><strong>Brain</strong></td>
<td>Brain MRI&lt;sup&gt;1&lt;/sup&gt;</td>
<td>Annually</td>
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<tr>
<td><strong>Colon (begin at age 25 years old)</strong></td>
<td>• Colonoscopy</td>
<td>Every 2-5 years</td>
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<td></td>
<td>• Esophagogastrroduodenoscopy (EGD)</td>
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<tr>
<td><strong>Leukemia/Lymphoma</strong></td>
<td>Blood tests: CBC with differential</td>
<td>Annually</td>
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<td><strong>Melanoma</strong></td>
<td>Skin exam – See Skin Cancer Screening algorithm</td>
<td>Annually</td>
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<tr>
<td><strong>Ovarian (females begin at age 35 years old)</strong></td>
<td>Refer to a doctor who specializes in high risk ovarian cancer screening – See Ovarian Cancer Screening algorithm</td>
<td>See Ovarian Cancer Screening algorithm</td>
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<tr>
<td><strong>Pancreas</strong></td>
<td>Refer to a doctor who specializes in high risk pancreatic cancer screening – See Pancreatic Cancer Screening algorithm</td>
<td>See Pancreatic Cancer Screening algorithm</td>
</tr>
<tr>
<td><strong>Sarcoma</strong></td>
<td>Whole body MRI</td>
<td>Annually</td>
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</tbody>
</table>

<sup>1</sup>The whole body MRI and brain MRI are both performed on an annual basis, staggered with a six month interval in between. The breast MRI should be performed at the same time as the brain MRI (but on different days due to the contrast dose).
SUGGESTED READINGS


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

Behrang Amini, MD, PHD (Diagnostic Radiology)
Therese Bevers, MD, BS (Clinical Cancer Prevention)†
Jasmina Bojadzieva, MS, BS (Cancer Genetics)†
Robin Coyne, MS, FNP, RN (Clinical Cancer Prevention)
Suzanne Day, MS, FNP, RN (Clinical Cancer Prevention)†
Tiffiny Jackson, MS, FNP, RN (Clinical Cancer Prevention)†
Louise Strong, MD, BA (Genetics)†
Priya Thomas, MD (Clinical Cancer Prevention)
Anita M. Williams, BS* (Clinical Effectiveness)
Brandy Willis, MBA, BS, AAB (Imaging Physics Operations)
Sonal Yang, PharmD* (Clinical Effectiveness)

†Core Development Team
*Clinical Effectiveness Development Team

Disclaimer: This algorithm has been developed for MD Anderson using a multidisciplinary approach considering circumstances particular to MD Anderson’s specific patient population, services and structure, and clinical information. This is not intended to replace the independent medical or professional judgment of physicians or other health care providers in the context of individual clinical circumstances to determine a patient’s care.