Li-Fraumeni Syndrome Screening - Adult

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.

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.

Pre-Test Considerations:
- Personal and/or family history suggestive of Li-Fraumeni syndrome or family member diagnosed with Li-Fraumeni syndrome through genetic testing

### Assessment

- Genetic testing indicates pathogenic or likely pathogenic TP53 variant?
  - Yes: Genetic test results do not clearly indicate whether the TP53 variant is germline, mosaic or somatic in nature
    - No
      - Patient willing to confirm through skin fibroblast testing?
        - Yes
          - TP53+ confirmed by fibroblast?
            - Yes: Recommendations will be specific to the individual
            - No: No
          - No: No
        - No: Yes
          - See Box A
  - No: Yes
    - Refer to Geneticist for genetic testing

### Recommendations

- Refer to appropriate oncology clinic based on clinical symptom
- Li-Fraumeni syndrome screening for other body sites based on cancer prognosis, per Oncologist

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1 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.

2 Refer to Patient Education - Li-Fraumeni Syndrome Education and Early Detection Program (LEAD) - Adult Screening Program

3 If moderate to severe anxiety related to Li-Fraumeni syndrome screening is identified, refer for psychiatric evaluation and/or counseling

4 See Physical Activity, Nutrition, and Tobacco Cessation algorithms; ongoing reassessment of lifestyle risks should be a part of routine clinical practice

5 Patient Education - Li-Fraumeni Syndrome Education and Early Detection (LEAD) - Adult Screening Guidelines

6 Skin fibroblast testing requires a skin punch biopsy

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

<table>
<thead>
<tr>
<th>Cancer</th>
<th>Exams and Tests</th>
<th>Frequency</th>
</tr>
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<tbody>
<tr>
<td>General</td>
<td>Complete physical exam</td>
<td>Every 6 months</td>
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</table>
| Adrenocortical Tumor (ACT) | • MRI¹ whole body  
      • DHEA-S, ACTH, testosterone                                                                                                     | Annually             |
| Brain                   | MRI¹,² brain                                                                                                                           | Annually             |
| Breast (begin at age 20-25 years old) | Clinical breast exam (begin at age 20 years old)  
      • Mammogram (begin at age 30 years old)  
      • MRI¹ breast (begin at age 25 years old)                                                                                         | Every 6-12 months    |
|                          | 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. | Annually (alternating every 6 months) |
| Colon (begin at age 25 years old) | • Colonoscopy  
      • Esophagogastroduodenoscopy (EGD)                                                                                              | Every 2-5 years      |
| Leukemia/Lymphoma       | CBC with differential                                                                                                                                 | Annually             |
| Melanoma                | Skin exam – see Skin Cancer Screening algorithm                                                                                         | Annually             |
| Pancreas³               | • CA 19-9  
      • HgbA1c  
      • MRI whole body                                                                                                                       | Annually             |
| Sarcoma                 | MRI whole body                                                                                                                          | Annually             |

DHEA-S = dehydroepiandrosterone sulfate  
ACTH = adrenocorticotropic hormone  
MRCP = magnetic resonance cholangiopancreatography

¹MRI of the whole body and brain 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).

²The first MRI should be performed with contrast; if normal, MRI without contrast should be performed thereafter.

³For patients with a family history of pancreatic ductal adenocarcinoma (PDAC) on affected side [1 first-degree relative (FDR) or 1 second-degree relative (SDR)]; See Pancreatic Cancer Screening algorithm.

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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 (Musculoskeletal Imaging)
- Banu Arun, MD (Breast Medical Oncology)
- Therese Bevers, MD (Cancer Prevention)
- Jessica Corredor, MS, CGC (Cancer Genetics)
- Robin Coyne, MS, RN, FNP (Cancer Prevention)
- Courtney DiNardo, MD (Leukemia)
- Wendy Garcia, BS
- Thoa Kazantsev, MSN, RN, OCN
- Julie Moskowitz, MS (Cancer Genetics)
- Louise Strong, MD (Genetics)
- Priya Thomas, MD (Cancer Prevention)
- Danielle Underferth, MS (Strategic Communications)
- Brandy Willis, MBA, BS, AAB (Diagnostic Imaging)

T Core Development Team
* Clinical Effectiveness Development Team

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