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

People who have Li-Fraumeni syndrome (LFS) (confirmed by genetic test report where a germline TP53 mutation was found) are at increased risk of developing several types of cancers. Because of this increased cancer risk, it is important for people with LFS to follow a comprehensive screening plan to detect cancer as early as possible.

Hospitals may have different screening programs. This document explains MD Anderson’s LFS screening program for adults age 21 and over. This is called the LEAD program.

The LEAD program is for people who have LFS and fall into any of these categories:

- People with no personal history of cancer
- Cancer survivors at least 1 year after active treatment is finished (such as chemotherapy and/or radiation)
- Cancer survivors at least 6 months after cancer surgery (if surgery was the only treatment for their cancer)

Individuals not eligible for the LEAD program include people:

- Who do not have a confirmed diagnosis of LFS by genetic testing
- Who are actively being treated for cancer
- Whose cancer has spread to other areas of the body

Screening Exams and Frequency

Every 6 Months

- Comprehensive physical exam, including brain and thyroid gland assessments
- Clinical breast exam (females only)

Words to Know

A genetic counselor will talk to you about these terms and what they might mean for you.

Mutation – a change in a person’s genetic information that results from damage to a cell or cells. Mutations can be somatic or germline.

Somatic TP53 gene mutation – changes in a person’s genetic information that can cause a tumor or cancer to develop. These mutations can develop in various cells as a person ages, has excessive sun or radiation exposure or long-term exposure to toxic chemicals, etc.

Germline TP53 gene mutation – changes in a person’s genetic information that can cause an increased risk to develop multiple types of cancer. The person is born with a germline mutation and that mutation is present in every cell of their body.
Once Per Year (Yearly)
- A blood draw that checks your adrenal gland function and tumor markers. Blood tests can be done at an outside facility if the results are sent to your MD Anderson doctor.
- Skin check by a dermatologist

Annually (Alternating Every 6 Months)
Each of the screening tests below should be completed once per year. You will have screening every 6 months to rotate the different types of screening. For example, if you have a brain MRI scan in January, then you should have a whole body MRI scan in June.

Screening tests include:
- Mammogram and breast MRI scan (females only, starting at age 20, or 5 to 10 years before the youngest age of breast cancer in your family).
- Whole body MRI and brain MRI scan.

The screening exam reports may take from a few days up to 1 week to be available. This is because your doctor and the doctor who performed the exam will need to discuss the findings. Please note that the exams may detect some changes in your images. The majority of these will not be cancer.

Every 2 to 5 Years (As Recommended by Your Health Care Provider)
- Colonoscopy – exam of the colon (starting at age 25 or 5 years before the youngest age of colon cancer diagnosis in your family)
- EGD – exam of the esophagus (starting at age 25)

Additional Recommendations
- Ask for a referral to a doctor who specializes in high-risk ovarian cancer screening.
- Ask for a referral to a doctor who specializes in high-risk pancreatic cancer screening.
- Women may want to consider having preventive surgical removal of the breasts, called prophylactic bilateral mastectomy. Ask your doctor if this surgery is an option for you.
- Learn about the signs and symptoms of leukemia and lymphoma. These may include, but are not limited to:
  - Always feeling weak and tired
  - Losing weight without trying
  - Enlarged lymph nodes
  - Excessive sweating
  - Frequent infections, easy bleeding or bruising, etc.
- Learn about the signs and symptoms of adrenocortical tumors (ACT). These may include, but are not limited to:
  - High blood pressure (hypertension)
  - A condition caused by prolonged hormone changes in the body that may cause symptoms such as progressive weight gain and skin changes (Cushing syndrome)
- Learn about the signs and symptoms of brain tumors. These may include, but are not limited to:
  - Headaches, vomiting or visual changes
  - Weakness or sensory changes
- Increased sleeping and fatigue
- Seizures or abnormal movements
- Cognitive decline or personality change

- Learn about the signs and symptoms of thyroid cancer. These may include, but are not limited to:
  - A lump that can be felt on the front of your neck
  - Changes to your voice, including hoarseness
  - Problems swallowing
  - Pain in your neck and throat
  - Swollen lymph nodes in your neck

- Learn about the signs and symptoms of melanoma. These may include, but are not limited to:
  - A change in an existing mole (itching, oozing or bleeding)
  - The development of a new pigmented or unusual looking skin growth. To help identify unusual looking growths think of the letters ABCDE:
    - **A** is for asymmetrical shape. Look for moles with irregular shapes, such as two very different-looking halves of a mole.
    - **B** is for irregular border. Look for moles with irregular, notched or scalloped borders. These are signs of melanoma.
    - **C** is for changes in color. Look for growths that have many colors or an uneven color.
    - **D** is for diameter. Look for new growth in a mole larger than 1/4 inch (about 6 millimeters).
    - **E** is for evolving. Any change in color, size or shape. This may include bleeding and itching.

**Resources**

**LFS Association**
[https://www.lfsassociation.org/](https://www.lfsassociation.org/)
This association provides information, advocacy and support services for people and families with LFS. They also support researchers, medical providers and caregivers to further research and promote care for the LFS community.

**LivingLFS**
LivingLFS encourages, educates and empowers individuals living with Li-Fraumeni Syndrome by connecting them with care, resources and other families living with LFS.

**LFS Research Study at MD Anderson**
[https://www.mdaanderson.org/lfsstudy](https://www.mdaanderson.org/lfsstudy)
This research study is led by Dr. Louise Strong. It comprises one of the largest collections of families with LFS in the world. The data and observations collected from the participants have contributed much knowledge about this rare syndrome.