

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

People who have Li-Fraumeni syndrome (LFS) (confirmed by a genetic test report where germline p53 mutation was found) are at an 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.

Different hospitals may have different screening programs. This document explains MD Anderson's LFS screening program for children and adolescents (under age 20). This is called the LEAD program.

Individuals with a confirmed diagnosis of LFS who should follow the LEAD program include:

- People with no personal history of cancer
- Cancer survivors who are at least 1 year past completing active treatment (such as chemotherapy and/or radiation)
- Cancer survivors who are at least 6 months past completion of surgery (if surgery was the only treatment for their cancer).

Individuals who **may not** be 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

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 p53 gene mutation – changes in a person's genetic information that can cause a tumor or cancer to develop. These mutations are acquired in various cells as a person ages, has excessive sun or radiation exposure or long-term exposure to toxic chemicals, etc.

Germline p53 gene mutation - changes in a person's genetic information that can cause an increased risk to develop multiple types of cancer. Individuals are born with a germline mutation and that mutation is present in every cell of their body.

Screening Exams and Frequency by Age

Patients 0-1 Year Old

Every 6 Months

- Complete physical and brain function exam
- Ultrasound (US) of abdomen and pelvis
- Referral for brain ultrasound at pediatric hospital
- A blood draw that will check adrenal gland function and tumor markers. Blood tests can be completed at an outside facility, if the results are sent to your MD Anderson doctor.

Patients 1-10 Years Old

Every 6 Months

- Complete physical and brain function exam
- Ultrasound (US) of abdomen and pelvis
- A blood draw that will check adrenal gland function and tumor markers. Blood tests can be completed at an outside facility if the results are sent to your MD Anderson doctor.

Once Per Year (Alternating Every 6 Months)

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

- Brain MRI
- Whole body MRI (beginning at 2-3 years of age or based on family history or if recommended by your doctor)

Patients 10-20 Years Old

Every 6 Months: Exams and Review of Body Systems

- Complete physical
- Brain function
- Thyroid gland
- Skin

Once Per Year

A blood draw to check adrenal gland function and tumor markers. Blood tests can be completed at an outside facility if the results are sent to your MD Anderson doctor.

Once Per Year (Alternating Every 6 Months)

The screening tests below should be done once per year, but you will have screening every 6 months to rotate the different types of screening. For example, if you have a whole body MRI in January, then you should have a brain MRI in June.

- Whole body MRI
- Brain MRI

The screening exam reports may take from a few days up to 1 week to be available. This is because frequently your doctor and the doctor who performed the exams will need to discuss the findings. Please note that the exams most likely will detect some variants in your images, but the majority of these will not be cancer or something that we need to investigate further.

Additional Recommendations for all Children and Adolescents

- 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:
 - Hypertension (high blood pressure)
 - Cushing syndrome (caused by prolonged hormone exposure in the body and may cause symptoms such as progressive weight gain and skin changes)
 - Virilization (typical effects seen in children are growth of pubic hair, accelerated growth and bone maturation, increased muscle strength and adult body odor before the age puberty should normally begin)
- 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 through the skin on your neck
 - Changes to your voice, including increasing hoarseness
 - Difficulty 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 growth on your skin. 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.
 - **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, shape; can include bleeding, and itching

Li-Fraumeni Syndrome Resources

LFS Association

www.lfsassociation.org

LFS Association provides information, advocacy and support services for individuals and families with LFS. They also support researchers, medical providers and caregivers to further research and promote care for the LFS community.

LivingLFS

<http://www.livinglfs.org/>

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

LFS research study at MD Anderson

www.mdanderson.org/lfsstudy

The Li-Fraumeni Syndrome 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 research participants have contributed much knowledge about this rare syndrome.

More Resources

National Cancer Institute

<http://www.cancer.gov>

This site has valuable cancer related health information on more than 200 cancer types, clinical trials, cancer statistics, prevention, screening, risk factors, genetics and support resources.

American Cancer Society (ACS)

<http://www.cancer.org>

The ACS is a voluntary national health organization that supports research, provides information about cancer and offers many programs and services to patients and their families.