

LI-FRAUMENI SYNDROME (LFS)

Genetics matter: know your family, know your genes.

What is Li-Fraumeni Syndrome (LFS)?

- LFS is an inherited, genetic predisposition to many types of cancers that affect both children and adults.
- It was discovered by Drs. Li and Fraumeni in 1969 after studying childhood cancer and cancer-prone families.
- Most families with LFS have an inherited pathogenic variant in the *TP53* gene – a tumor suppressor gene.
- *TP53* is the most studied human gene in the world – its primary function is to guard against cancer.
- LFS carriers have a 50% chance of developing cancer by age 40, and up to a 90% chance by age 60.
- The most commonly occurring cancers among those with LFS include breast cancer, brain tumors, adrenocortical carcinomas, osteosarcomas, and soft-tissue sarcomas. LFS is also associated with many other cancers.

Why is Getting a Diagnosis of LFS Important?

- About 80% of individuals with LFS inherit it from an affected parent.
- About 7-20% of individuals with LFS have the syndrome as the result of a new genetic variant. This is called “de novo” LFS.
- Any person with LFS has a 50% chance to pass the diagnosis of LFS on to every child they have.
- Participation in early-detection screening has been proven to increase the overall survival for those with LFS.

Unique Risk to Women

- Women with LFS have as high as a 90% risk of developing cancer in their lifetime due to their markedly increased risk of breast cancer.
- Breast cancers can occur at unusually young ages in women with LFS, as early as their 20s and 30s.
- It is critical to know if a breast cancer patient has LFS because the information can influence treatment decisions, like opting for risk-reducing mastectomy surgery and/or avoiding radiation, when possible.

Unique Risk to Children

- Each child born to a parent with LFS has a 50% chance of inheriting the mutation.
- 40% of children with LFS will develop at least one cancer by the age of eighteen.
- Almost 50% of children diagnosed with adrenocortical and/or choroid plexus carcinomas have LFS.
- Those who survive childhood cancer are at an increased risk for developing multiple primary cancers.

LFS and Public Health

- Cancer touches everyone. If you have not had a cancer diagnosis personally, you know someone who has.
- Childhood, early onset, rare, as well as common cancers (such as breast cancer) may be linked to LFS.
- Radiation treatments may increase the risk of developing additional cancers in those with LFS.
- Somatic *TP53* genetic variants are found in nearly all cancers within the general population. The work of LFS scientists and physicians is contributing to our understanding of all forms of cancer.
- Increased genetic testing of people with cancer has identified more carriers of pathogenic *TP53* variants than expected.
- A positive genetic test for LFS could affect a cancer patient’s treatment plan.
- Carriers of *TP53* variants can take proactive measures such as making important lifestyle changes to reduce their risk, and they can participate in early detection screening to best improve treatment outcomes. Importantly, they can also ensure that family members are tested.
- About 1 in 5,000 to 1 in 10,000 people in the world are thought to have LFS. In some cancer databases, the number is even higher.



Research • Education • Advancement • Community • Hope

Devoted to ending early cancer deaths and improving lives for those living with LFS.

What is LFSA?

The LFS Association (LFSA) was founded in 2010 at the National Cancer Institute's Division of Cancer Epidemiology and Genetics Branch by individuals with LFS and their families to help promote research, awareness, and support for patient families. We are managed by an all-volunteer board and supported by a Medical & Scientific Advisory Board, and Genetic Counselor Advisory Group, who are international leaders in LFS care and research. LFSA is a global enterprise with 15 international chapters composed of medical professionals, patient family advocates, and youth coordinators. LFSA is a member of the National Organization of Rare Diseases (NORD) and the Rare Cancer Coalition. It has been recognized by the Chan Zuckerberg Rare as One Foundation.

Our Work

The LFS Association mission is to advance education, patient support, cancer research funding, and worldwide public awareness. We also foster collaboration among researchers, medical providers, and caregivers to further research and promote optimal care for the LFS community.

LFSA launched the premier model for international chapters consisting of a medical professional paired with a patient family volunteer as co-chairs and a youth program coordinator. Our international chapters include Africa, Canada, China, France, Germany, India, Italy, Japan, Latin America, Netherlands, Portugal, Saudi Arabia, Singapore, Sweden, and UAE.

We actively provide support and resources for the LiFE (Li-Fraumeni Exploration) consortium – the leading international LFS researchers.

We have established a Youth and Young Adult program that provides opportunities for teens and young adults with LFS around the world to connect and help each other navigate the challenges of LFS.

We host and sponsor international symposia where providers, researchers, medical students, and those with LFS from around the world have a unique opportunity to collaborate with the goals of advancing science and creating a deeper understanding of the unique needs of LFS families.

LFSA facilitates cutting-edge research focused on early-detection screening (such as rapid whole-body MRIs and circulating tumor DNA) as well as potential treatments for LFS.

LFSA promotes awareness of the need to assess whether to use radiation in the treatment plan for those with LFS.

Visit us:

LFSAssociation.org

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