

# INTERNATIONAL LI-FRAUMENI SYNDROME AWARENESS DAY

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## Genetics matter: know your family, know your genes.

### What is Li-Fraumeni Syndrome?

- LFS is an inherited cancer predisposition disorder associated with early age at onset cancers.
- 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 – one of its functions is to guard against cancer.
- LFS predisposes carriers to a diverse range of childhood and adult-onset cancers - some rare, often early onset.
- LFS carriers have an approximately 50% chance of developing cancer by age 40, and up to a 90% 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 associated with many other cancers, as well.

### Why is Genetic Testing Important?

- Classically, LFS is diagnosed based on family history but up to 20% of all LFS patients are the first in their families to discover they have LFS (“de novo” cases).
- Participation in early-detection screening has proven to increase the overall survival for those with LFS.
- Radiation therapy should be considered in people with LFS only if its use is essential to the control or cure of the cancer because of its link to increased risk of a new cancer in the field of radiation.

### Unique Risk to Women

- Females 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, even in their 20's and 30's.
- It may be important to know if a breast cancer patient has LFS because the information can influence treatment decision, like avoiding radiation..

### 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.

### How Li-Fraumeni Syndrome and *TP53* Impact 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 cancer in people with LFS.
- Malfunctioning *TP53* are involved with most all cancers in the general population. LFS cancer research strongly contributes to all cancer research.
- Increased genetic testing of people with cancer has identified more carriers of pathogenic *TP53* variants than expected based on their family history.
- A positive genetic test for LFS could affect a cancer patient's treatment plan.
- Carriers of pathogenic *TP53* variants can take proactive measures such as lifestyle changes to reduce the risk of developing cancers and participate in early detection screening to best improve treatment outcomes.



Research • Education • Advancement • Collaboration • Hope • Support

## **Dedicated to ending early cancer deaths and improving lives for those living with LFS.**

### **What is LFSA?**

LFS Association (LFSA) was first founded in 2010 by patients and their families to help promote research, create awareness, and support patient families. We are comprised of an all-volunteer board, supported by our Medical Advisory Board, our Scientific Advisory Board, and our Genetic Counselor Advisory Group. We are global, with international chapters consisting of medical professionals, patient family advocates, and youth coordinators. We are members of the National Organization of Rare Diseases (NORD) and their Rare Cancer Coalition and have been recognized and awarded by the Chan Zuckerberg Rare as One foundation.

### **What are We Doing?**

The LFS Association mission includes worldwide public awareness and education, patient support services, and cancer research funding.

LFSA launched the premier model for international chapters comprised of a medical professional with a patient family volunteer as co-chairs, along with a youth program coordinator. Our international chapters include Australia/New Zealand, Canada, France, Germany, India, Japan, Latin America, Netherlands, Saudi Arabia, and Singapore.

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

We have established a Youth Program and a Young Adult Program, providing opportunities for teens and young adults living with LFS from around the world to connect and help each other navigate through living with LFS.

We host and sponsor international symposiums where providers, researchers, medical students, and people with LFS from around the world have a unique opportunity to collaborate in furtherance of advanced science and a deeper understanding of the needs of LFS families.

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

We promote awareness on the critical determination of whether or not to use radiation in the treatment plan for people with LFS.

**Visit us:**

**[www.LFSAssociation.org](http://www.LFSAssociation.org)**

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