

# LFS Association. Putting Cancer To The Test.

## 2022 IMPACT REPORT



Research • Community • Hope

# A Message from the LFSA President

Dear Friends of LFS Association,



As I look at all that LFSA has accomplished this year as a result of your generous and essential support, I am overflowing with gratitude. You make our progress possible. Your gifts, volunteer efforts, scientific tenacity, advocacy, and passionate commitment to eradicate LFS are at the heart of our success.

You are a shining example of what is known as the **butterfly effect**.

The butterfly effect explains how the single flutter of a butterfly's wings can have a significant effect on weather on the other side of the world. It is symbolic of how a seemingly small act can result in profound change.

One sparkling example of the butterfly effect is our grant program, led by LFSA's Scientific Advisory Board. Because of your meaningful gifts, we awarded our first LFSA grant to help fund a first-of-its-kind breast cancer study in LFS patients. This first award of \$45,000 may have been small within the world of scientific research, but it was huge for the LFSA.

The gift was synonymous to a butterfly flapping its wings. As evidence, it became transformational when it served as seed money that in turn sparked interest and additional funding from several large cancer research organizations. I am thrilled to share

with you that today, the LFS breast cancer study is supported by substantial research funding and the combined scientific expertise of some of the most influential cancer researchers in the world.

Each member of the LFSA community of families, researchers, and medical experts embodies the power of a butterfly. Together, we are making a profound difference in the lives of all of us who are affected by LFS.

In acts both large and small, together, we are furthering our understanding of this syndrome and advancing scientific discovery and treatments for all who grapple with LFS. Learn more about how LFSA continues its work to support individuals and families through its consortium of fiercely dedicated researchers, medical providers, and caregivers.

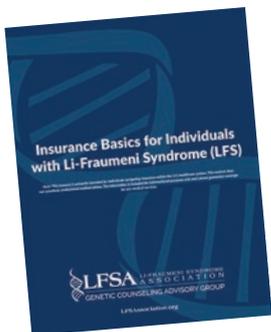
Together, with your help, we are the building blocks of a future without LFS.

***We are all interconnected. No act is too small to make a big difference.***

With my deepest gratitude,

**Jenn Perry**  
LFSA President, LFS patient and LFS mom

## Insurance Resources for LFS Families



Navigating insurance and medical bills are pressing concerns for LFS families. The good news is that LFSA's Genetic Counseling Advisory Board has released updated insurance information that will serve as a vital resource.

Available through the LFSA website as a downloadable PDF, Insurance Basics for Individuals with Li-Fraumeni Syndrome is intended for individuals within the U.S. healthcare system. It provides an

overview of insurance costs and insurance plan information. It also answers frequently asked questions by those with LFS.

### Frequently Asked Questions

- *Will my health insurance pay for my genetic testing?*
- *Can my health insurance raise my premiums or deny me coverage because of my LFS?*
- *Why is it so difficult to get coverage for my whole-body MRI?*

The LFSA's latest insurance information includes several supporting documents to help LFS families discuss their unique needs with care providers.

You can find the Health Insurance Resources, including the Pediatric and Adult Appeal Letters, Whole-Body MRI Worksheet and LFS Association's Surveillance Consensus Statement by visiting [bit.ly/3VDdcv0](https://bit.ly/3VDdcv0) or **scanning this QR code**.





# Leading the Charge



Eradicating LFS is a global imperative as demonstrated at REACH22 where experts and families representing 24 countries gathered to collaborate, share, and learn. At our first post-COVID hybrid event, more than 400 people and 237 families gathered at the National Cancer Institute (NCI) in Bethesda, Maryland, both virtually and in person.

Hosted by the NCI – part of the National Institutes of Health and the federal government’s principal agency responsible for cancer research – and in partnership with the LiFE Consortium, the 6th International LFSA Symposium attracted some of the greatest minds in cancer research, treatment, and care, along with those who have unique expertise in LFS.

Some of the most promising research was discussed in sessions that included understanding the LFS spectrum of *TP53* variants, emerging research on *TP53* biology and the connection between elephants, evolutionary medicine and Li-Fraumeni syndrome. Other sessions focused on a range of issues including the

implications of testing children for LFS and implementing support group programs for individuals and families with LFS.

Connecting LFS families with scientists and clinicians whose life’s work is preventing and treating cancer provides vibrant sharing of knowledge, hope, and support among all participants. The symposium sessions were further enhanced by breakout sessions where families engaged in issues ranging from how to navigate a new LFS diagnosis to understanding the impact of health insurance on LFS.

One of the most inspirational sessions was a discussion among the youngest attendees, all of whom are grappling with an LFS diagnosis. To a rapt audience, they shared their experiences, hope for the future, and a sense of urgency to address the full gamut of issues, from early diagnosis, to improving treatments, and putting an end to LFS.



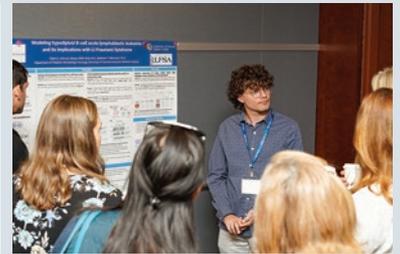
LFSA volunteers come together at the NCI in preparation for REACH22



The LFSA youth and young adults discuss navigating life with LFS and tips for self advocacy



Elaine Shiang reflects on the life and work of her late husband Dr. Frederick Li



One of LFSA’s original youth program members, Elijah Johnson, presents during the poster session



Following his presentation on the early days at the NCI, Dr. Fraumeni reconnects with the team



LFS families and genetic counselors connect during a breakout session



Dr. Josh Schiffman shares the latest breakthroughs on p53 elephant research



Guest panelists answer questions on the last day of the symposium with an open-panel session

# PUT CANCER TO THE TEST

Learn how genetic testing saves lives through early detection.

[LFSAssociation.org/get-tested](https://LFSAssociation.org/get-tested)

**Genetics matter.**  
Share this information with members of your family.

This year, LFS Association launched, *Put Cancer To The Test*, an awareness campaign that is educating the public about Li-Fraumeni syndrome and the importance of genetic testing in families where there has been a prevalence of cancer diagnoses. Determining whether LFS is a familial concern is a first step to early diagnosis and access to vital resources. Early intervention can lead to early detection, a surefire path to saving lives. Learn more on our website and help us spread the word.

Learning your family history is your best resource in addressing LFS. LFSA wants the public to know that many cancers are associated with LFS with these being most prevalent:

- Soft tissue sarcoma (such as anaplastic rhabdomyosarcoma)
- Osteosarcoma
- Breast cancer (premenopausal)
- Brain and central nervous system (CNS) tumors (such as glioma, choroid plexus carcinoma, SHH subtype medulloblastoma, neuroblastoma)
- Adrenocortical carcinoma
- Acute leukemia
- Lung adenocarcinoma
- Melanoma
- Gastrointestinal tumors (such as colon, pancreas)
- Kidney
- Thyroid
- Gonadal germ cells (such as ovarian, testicular, and prostate)

## Coming Soon! The LFSA Youth Workshop 2023

Deep friendships have been forged among LFS teens and among LFS parents since the LFSA held its first Youth Workshop in 2017 at the Huntsman Cancer Institute. The relationships, support, deep connections, and affection that evolved from the first Workshop continue to enrich families to this day.

Our Youth Program helps young people with LFS live their lives to the fullest and make connections with others who keenly understand the experience of an LFS diagnosis. The Workshops ensure our young people never feel alone. Open to those ages 13 to 26, the program hosts quarterly Youth Connect webinars and publishes an LFSA youth newsletter composed of articles written by LFSA youth from around the world, all coordinated under the creative eye of our Youth Program Coordinator, Cameron Block. In the most recent webinar, the Hirsch Wellness Network took participants through a Zentangle exercise, a meditative form of art.



*Hirsch Wellness Network's Instructor, Michele Rieder, demonstrates Zentangle during an LFSA Youth Connect Workshop*



**LFSA has announced that its 4th LFSA Youth Workshop will be held in 2023.**

For the latest details and for access to past webinars, visit the Youth Program page here. Sign up for regular updates on youth news and please help share the information.

# LFSA Medical and Scientific Advisory Boards Join to Accelerate Progress



(L-R) Rob Lufkin, Josh Schiffman, Christian Kratz, Sharon Savage, Phuong Mai, Jenn Perry, David Malkin, Kara Maxwell, Pierre Hainaut, Arnold Levine, Sean Downing, Maria Isabel Achatz, Judy Garber

In a milestone meeting, the LFSA's Medical and Scientific Advisory Boards joined together for the first time at the 6th International LFS Association Symposium to collaborate, strategize, and create synergies that will benefit treatment of LFS. That these two powerful teams of experts convened at the National Cancer Institute in Bethesda, Maryland, was especially meaningful because it was here that the LFS Association was launched 12 years ago.

Our vision for the LFSA was to forge connections among LFS families, the research community, and medical experts. That vision has been realized. Today, the LFSA is a global coalition providing patient support, advocacy, and research funding – all in a coordinated effort to address the unique challenges of LFS.

Members of the LFSA Medical and Scientific Advisory Boards came together to probe the deeper questions about Li-Fraumeni syndrome and to discuss how the unique medical and scientific perspectives could shed new light on fresh approaches to combat the syndrome. This meeting was the beginning of a bold initiative that will lead to a better understanding of p53 and LFS-related cancers.

## Among the issues discussed:

**Surveillance strategies.** The increased availability and prevalence of genetic testing has forced us to address what it really means to have LFS. A new LFS classification system is being created to better encapsulate the risks of the various p53 variants. Our goal is to implement more effective, risk adapted surveillance strategies.

**Early detection and intervention.** Peripheral blood diagnostics have greatly improved in recent years, and the LFSA is eager to participate in advanced cancer screening and monitoring programs as they continue to evolve. This intense focus on early diagnosis and cancer prevention WILL continue. There is consensus that the best defense against LFS remains early detection and intervention.

**Novel therapies.** We are entering into an era of astounding new scientific discoveries. Novel immunotherapies and immunotherapy combinations are being used for an increasing number of cancers. We now have drugs that target deficits associated with certain germline mutations to reactivate the protein. The LFSA is funding research grants in both areas. In addition, there has been an explosion of newer antibody drug conjugates that promise more precise, effective targeted therapies that will promise to impact LFS patient care. We must explore how to use these therapies to treat LFS-related cancer.

By working together, our LFS research and clinical experts are tapping into a vast network of knowledge, creativity, and insights. The LFSA is fiercely committed to continuing to drive LFS research and medical knowledge through this type of global connectivity and by creating partnerships that will prompt innovative approaches to the unique challenges of LFS.

**Robert Lufkin, D.O. – LFSA Scientific and Medical Director**  
**Sean Downing, PhD – LFSA Scientific Advisor**

## Boosting LFS Awareness Among Medical Professionals

As COVID restrictions eased during 2022 in many parts of the world, LFSA was eager to participate in events to promote engagement with scientists, physicians, and genetic counselors, as a path to forge vital collaborations and partnerships.

For the first time, LFSA participated in the largest cancer conference in the world, the American Society of Clinical Oncology, ASCO. This annual meeting held in Chicago each year attracts world experts in oncology. Through a prominent presence in the ASCO exhibition hall, LFSA leadership Kathy Higgins, Debbie Soprano, and Gina Vild, met with many of the 40,000 professionals who attended the five-day event. They came away with deeper knowledge of how physicians from around the world view and manage LFS patients and with robust contacts that we are hopeful will spur further sponsorship.



This year, LFSA leadership also attended the Oncology Nursing Society in Anaheim, California, and the American Academy of Pediatrics meeting in Gettysburg, Pennsylvania. LFSA also continues important collaborations with the Rare Cancer Coalition and the National Organization for Rare Disorders, groups united in their shared goal to eradicate cancer.

# The Global Reach of LFSA

LFSA now has 12 chapters around the world as Italy joins Africa, Latin America, Canada, France, Germany, India, Japan, Netherlands, Australia/New Zealand, Saudi Arabia, and Singapore to support families with LFS around the world.

Each international chapter is chaired by a local medical professional who connects LFS families with vital local resources. The newly formed Italian chapter is led by Dr. Valentina Arcangeli and sisters Debora Weiss and Marcia Peterson Leite.

“No matter where we are in the world, we can collaborate and challenge each other to solve this puzzle that is LFS,” says Jenn Perry, LFSA President. “We are profoundly grateful for this vast network that shares information and supports our LFS families.”



## Here's just a sampling of our international efforts:

**Africa:** The recently formed Africa chapter is led by Samuel Oliech Omolo, a biotechnologist from Kenya, who traveled from Kenya to attend LFSA's Symposium. LFSA - Africa recently hosted a booth at the Africities Summit, a Pan Africa conference convened to improve the living standards of the citizens, where LFSA was able to increase awareness of LFS to more than the 11,000 people who attended the event. Samuel now has family support and a youth program volunteer.

**Australia/New Zealand:** This LFSA chapter was one of several organizations to support Omico's (the Australian Genomic Cancer Medicine Centre) application for annual whole-body MRI (wbMRIs) for *TP53* variant carriers to the Australian Medical Services Advisory Committee. The impressive result is that wbMRIs will now be funded under the Medicare Benefits Scheme starting March 1, 2023. Dr. Laura Forrest spoke at the LFS UK meeting organized by the George Pantziaka Trust in September, and Samantha Arthur continues to support families with LFS throughout Australia and New Zealand.

**Canada:** LFSA - Canada is co-chaired by Chiquita Hessels and Dr. David Malkin, along with Anna Joy Ryan, all passionate and active LFS family advocates. A highlight this year for our May 3rd International Awareness Day was a spectacular lighting of the Newfoundland State House in the theme colors of the LFSA during the whole month of May.

**France:** Carrying on the legacy of the late Prof. Thierry Frébourg, founder of LFSA - France, Drs. Pierre Hainaut and Gaëlle Bougeard-Denoyelle are building community and enhancing care for LFS families in France.

**Germany:** Youth involvement is a pillar of Germany's robust efforts. The chapter, co-chaired by Reinhard and Claudia Sablowski, under the guardianship of Dr. Christian Kratz, held many family meetings and youth outings this year.

**India:** LFSA - India is co-chaired by Dr. Sameer Rastogi and the National Cancer Institute's Dr. Payal Khincha, who are publishing *Lessons Learned from Clinic-genomic Profiling of Families with Li-Fraumeni Syndrome*. This is recognized for being the largest case series in India.

**Japan:** LFSA - Japan chair Chieko Tamura also serves on LFSA's Genetic Counseling Advisory Group. Certified as a genetic counselor in the U.S. and Japan, Tamura brings to her work a unique perspective that integrates American and Japanese cultures and is actively using it to serve and support LFS patients and their families.

**Latin America:** LFSA - Latin America has grown tremendously, expanding its reach far beyond Brazil. Led by Dr. Maria Isabel Waddington Achatz, LFSA - Latin America hosted several family meetings this year and has very active youth coordinators assisting in many arenas.

**Netherlands:** Chapter Chair Leendert Zaaijer, who has LFS, is connecting Dutch LFS families with the care, community and information they need.

**Saudi Arabia:** Under the leadership of genetic counselor Lamia Fahad Alsubaie, LFSA - Saudi Arabia, who also participates in the LFSA - GCAG, is a shining example of how cultural challenges combined with determination can drive results. The chapter represented LFSA at the Saudi Cancer Society Survivor Day and hosted webinars on LFS.

**Singapore:** LFSA - Singapore, under the leadership of Dr. Joanne Ngeow, held an event to strengthen the connection between LFS families and the medical community.