



# Setting The Standard

## 2021 LFSA IMPACT REPORT

Anna Joy Ryan  
LFSA Canada Youth Chair

LI-FRAUMENI SYNDROME  
 **LFSA**  
ASSOCIATION  
[LFSAssociation.org](https://LFSAssociation.org)

Research • Education • Awareness • Community • Hope

# A Message from the LFSA President

Dear Friends of the Li-Fraumeni Syndrome Association,



I'm overjoyed at all that we have accomplished this year because of your support of the LFS Association. Whether you've donated money, given your time and expertise or organized your community on our behalf, you're the reason we've achieved so much this year.

This Impact Report highlights the many ways your contributions are helping the LFSA set the standard of care and offer hope to families and individuals with Li-Fraumeni syndrome. Because of you, we've done some incredible things to support the LFS patient community and drive research toward a cure for LFS.

This year, we've driven research with a global reach. Your donations have resulted in our first ever research grant presented to Professor Arnold Levine for an LFS breast cancer study. The LFSA's Scientific Advisory Board is in the process of announcing another grant currently, and more are planned for next year.

We've also added an LFS Association - Africa chapter, bringing our international chapter number to 11, covering six

continents! Being more inclusive globally benefits all of us with LFS. We're able to reach people that may otherwise have no support for LFS. And, we're able to enhance LFS research efforts by sharing the knowledge base of international scientific, medical, and genetic perspectives.

We are building on all that we've accomplished this year to expand our impact next year. The best collaboration takes place in person, so we are a full green light on more face-to-face education, including our 6th International LFS Association Symposium and International Youth Conference. These will be in-person events with improved technology to allow more people to participate from around the world.

We plan to get up close and personal through more outreach and more in-person connection, so get ready to see MORE of us! As your association, we'll continue to provide hope and resources for individuals and families with LFS and ultimately find a cure.

*Gratefully yours,*

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

## Medical Director's Message



Since the inception and formation of the LFSA, it has been our intention to form a large coalition of LFS patients, researchers, and providers worldwide to provide support, raise awareness and promote LFS-directed research. In doing so, we have linked together unlikely partners and connected disparate groups with one common goal – to improve the lives of those with LFS.

An inspiring example of these collaborative efforts is a recent publication in JAMA Oncology in October, initiated by LFSA's own Medical Advisory Board member, LIFE Consortium member and German LFS chapter co-chair, Prof. Christian Kratz, Hannover Medical School in Hannover.

Using the International Agency for Research on Cancer database, the research team analyzed data from patients with hereditary *TP53* variants and identified differences between variants in patients with severe disease compared to those with less severe courses. First presented at LFSA's 5th International Symposium in 2020, the concept of analyzing tumor patterns and genetic testing criteria can accelerate future research toward the development of more tailored approaches for cancer surveillance with the potential to better predict cancer risk.

According to Dr. Kratz, "With the increasing use of *TP53* sequencing over the last three decades, it became clear that the

disease spectrum is broader than originally described. The new classification is an important step toward defining the factors that predict the unique cancer risk in individuals with LFS."

Going further, defining these "unique factors that predict cancer risk" potentially buried in decades of accumulated data may hold the key to answering many of our longstanding questions in LFS. Using the tools that we have, such as existing databases, and modernizing data collection with strategies such as in the LiftUp study, there is the potential to further improve clinical outcomes in those with LFS-related cancer. It is the mission of the LFSA to inspire a new crop of researchers to pursue these strategies and lead us into the future.

We also honor those that have laid the groundwork for progress in Li-Fraumeni syndrome. This year, we lost Prof. Thierry Frébourg, a major contributor to cancer genetics research who was devoted to advancing the care of those with inherited predisposition to cancer. Thierry was a wonderful human being and a very dear friend of the LFSA, who, on his own, initiated the LFSA France chapter. The entire LFS community mourns his loss and yet we celebrate his amazing accomplishments. His friendship, leadership, and warm affection were deeply treasured and will be missed.

**Robert Lufkin, D.O.**  
**LFSA Scientific and Medical Director**

# LFSA Scientific Advisory Board Driving More LFS Research

In its first year, the LFSA Scientific Advisory Board has established a grant program and awarded its first grant to Professor Arnold Levine, PhD, Institute for Advanced Study, Princeton, N.J. Levine and his colleagues are collaborating with the Memorial Sloan Kettering Cancer Center in a study on breast cancer in LFS patients.

**LFSA Scientific Advisory Board goal:**  
to bring together basic scientists to collaborate with clinicians in order to initiate research with Li-Fraumeni syndrome patients in the areas of immunotherapy and other possible novel treatments.

"There has been a good deal of research on LFS but much less than other cancers because it is a fairly rare syndrome," said Prof. Levine. "Now we have a group dedicated to increasing the level of fundamental research in the Li-Fraumeni syndrome area."

The LFSA Scientific Advisory Board is thrilled to be facilitating this collaborative research project, which is funded in part by the LFS Association. "This is among the first of many examples to come where donations to the LFS Association are funding active research at the interface of the clinic and basic sciences," Levine said.

## Why LFS breast cancer research?

About 70% to 90% of women with LFS and the inherited *TP53* gene alteration develop breast cancers. And, 90% of women with LFS will get cancer in their lifetime because of their propensity to develop breast cancer.

In this study, Levine and his collaborators seek to determine if the immune system is responding to breast cancer in Li-Fraumeni syndrome patients and what type of immune cells may be responding. Knowing the particulars of an immune response in *TP53*-driven LFS breast cancers may lead to a better understanding of which immunotherapies to utilize to treat patients and possibly lead to new and more effective therapies.

"In the grant process, we look for LFS research that could have a direct application in the clinic," said Sean Downing, chairman, LFSA Scientific Advisory Board. "In this case, we'll

know whether or not immunotherapy treatments are beneficial."

## Collaborative research brings more focus to LFS

Levine points out that collaborating on LFS research with more generalized cancer centers, like this one with Memorial Sloan Kettering Cancer Center, expands LFS awareness among the medical community and builds the scientific knowledge base for LFS and all cancer.

"We've brought in physicists to be able to analyze the data, clinicians, researchers, and immunologists who will be collaborating with us for this particular grant. Many of whom will be working on Li-Fraumeni syndrome for the very first time," Levine notes.

Just beginning the research, Levine and his team will be recruiting LFS patients to volunteer for the study as well as individuals with spontaneous mutations of p53, or Triple Negative Breast Cancer (TNBC), to serve as the control group. Ultimately, the study is the first step in determining whether immunology would be possible to treat breast cancer in patients with LFS.

## New research funding in the works

Levine's breast cancer study is the first of several research grants in the works for the LFSA Scientific Advisory Board. A second grant will be awarded by the end of this year, and two more grants (spring and fall) are planned for 2022.

Grants beyond next year depend on the continued generosity from friends of the LFSA. "We can't provide funding for more research unless we get funding ourselves," Downing said. "We rely on benefactors to provide the funds that drive research projects specific to LFS."

**"There has been a good deal of research on LFS but much less than other cancers because it is a fairly rare syndrome. Now we have a group dedicated to increasing the level of fundamental research in the Li-Fraumeni syndrome area."**

~Prof. Arnold Levine



## LFSA'S SCIENTIFIC ADVISORY BOARD

(Left to Right) Dr. Maria Isabel Achatz, PhD, Hospital Sirio-Libanês, Sao Paulo, Brazil • Prof. Arnie Levine, PhD, Princeton Institute for Advanced Studies, New Jersey, USA  
Prof. Pierre Hainaut, PhD, Université Grenoble Alpes, Grenoble, France • Prof. Moshe Oren, PhD, Weizmann Institute of Science, Rehovot, Israel  
Prof. Guillermina Lozano, PhD, MD Anderson Cancer Center, Texas, USA • Dr. Drew Pordall, PhD, Johns Hopkins, Maryland, USA • Dr. Sean Downing, PhD, Takeda, Boston, USA

# Genetic Counselors Bridge the Gap for LFS Families

LFSA's Genetic Counseling Advisory Group (GCAG) published a number of resources this year to help individuals with LFS empower themselves and educate their providers.

"Genetic counselors are all about education and informing people," said GCAG Co-chair Kathryn Schneider, a licensed genetic counselor in Massachusetts with more than 25 years in cancer genetic counseling. "When you have a rare genetic syndrome like LFS, the individual with LFS is often the one responsible for educating their own physicians, schools, and other providers."

The group developed an *LFS Fact Sheet for Medical Providers* and a *Quick Reference Resource for Mental Health Providers* for LFS patients to share with their providers and offer documentation that addresses special LFS needs.

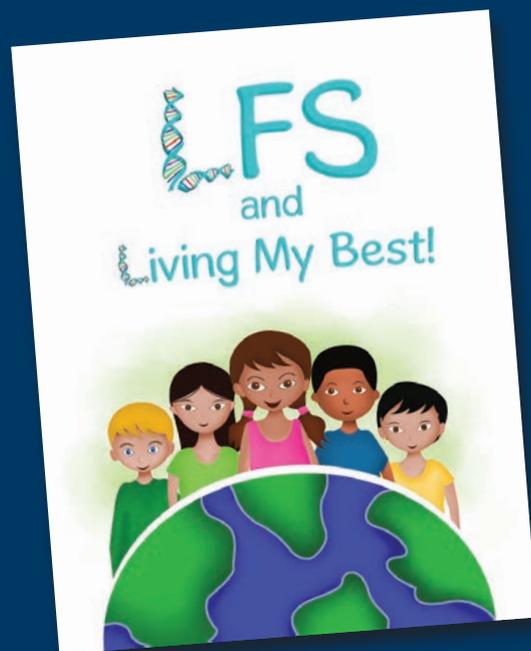
GCAG also created a brochure and a book tailored to the various life stages of those living with LFS, including the *LFS Awareness Brochure for Teens* and "*LFS and Living My Best!*" children's book. Schneider is particularly excited about the far-reaching impact of the children's book, thanks to contributions from more than 25 genetic counselors from around the world who make up the GCAG. The book is currently being translated to different languages and will be distributed internationally.

"LFSA's Genetic Counseling Advisory Group has been instrumental in supporting LFS patients all over the world, even countries who don't have genetic counselors" said Jenn Perry, LFSA president. "Addressing the psycho-social side of having LFS is critical, and genetic counselors are key for supporting patients through the full spectrum of patient life with LFS that goes well beyond genetic testing."

Schneider calls it the genetic counselors' "superpower" – their ability to talk about medical concepts in a way that's understandable. "We simplify it and interpret it for whatever age or stage of knowledge, whether a young child or older person."

A cancer patient with LFS, for example, is going to have different kinds of questions than a relative just learning about LFS and its genetic implications. To address the broadest range of questions, the GCAG collaborated on the Frequently Asked Questions document housed on the LFSA website. This living document is regularly updated with the latest research and new concerns as they arise.

"So much information is out there on other cancers, we want to provide resources specific to LFS. We want people to be able to advocate for themselves, but also share the word about LFS," said Schneider.



## LFS Resources Available Now!

Please visit LFSA's website [lfsassociation.org](http://lfsassociation.org) to access these practical resources developed by LFS Association's Genetic Counseling Advisory Group this year:

### Frequently Asked Questions

This list of frequently asked questions about LFS offers a place to start for help getting answers. Included is a glossary of terms to help you navigate an LFS diagnosis.

### Quick Reference Resource for Mental Health Providers

This printable page, designed for mental health professionals, delineates challenges that families with LFS face.

### LFS Awareness Brochure for Teens

Teens and young adults with LFS have a different set of challenges. This brochure addresses topics like LFS and school, friends and genetic counseling.

### LFS Fact Sheet for Medical Providers

Sometimes LFS patients have to educate their own medical providers about LFS. This quick fact sheet for healthcare teams is a great place to start.

### "LFS and Living My Best!" Children's Book

Designed for children ages 8 to 13, this illustrated children's book can be downloaded to share with your child.

# LFS Youth Empowered By Friendship

The LFS Youth Program continued to inform and connect LFS teens and young adults this year through quarterly virtual workshops and digital newsletters. Designed to help young people with LFS live their lives to the fullest, LFS's Youth Program debuted its Youth Connect virtual workshops, which invite medical professionals to speak on topics specific to youth concerns. The online workshops also set aside time for attendees to ask questions and simply get to know one another.

"When a teen gets diagnosed with cancer, like I did when I was 14, you don't meet a lot of other people like you," said Cameron Block, LFS Youth Program Chair, USA Chapter. Having a rare syndrome like LFS can compound those feelings of loneliness and isolation.

"We want to provide LFS youth and young adults with a positive environment where they can learn more about LFS, empower them with information and even make some good friends," Block said.



Block got involved with the LFS Youth Program at the first International Youth Workshop held in Salt Lake City in 2017. There, she connected with LFS youth from around the world and has remained in contact with many of them through a group chat on WhatsApp. "Being able to meet those kids at the youth workshop was incredible! Seeing them and not feeling alone actually brought me to tears."

Today, Block works closely with fellow youth chairs Anna Joy Ryan, Canada, and Isabel Costa and Olivia Naves de Andrade, Brazil, to plan Youth Connect meetings. Isabel and Olivia even translate the newsletters into Portuguese and Spanish.

Block says making friends living with LFS in other countries has opened her eyes to some of the different challenges they face, like accessing full-body MRIs. "It is our goal to have at least one youth chair in each of our chapters, so that every young adult with LFS has a resource in their country where they can feel encouraged and supported," she said.



The first Youth Connect topic focused on stress and coping with LFS and another addressed nutrition. Each Youth Newsletter features an interview with a medical expert and LFS youth. Visit the LFS website to view the latest Youth Connect sessions and Youth Newsletters:

- **"Stress and Coping with LFS"**  
Panelists Rowan Forbes-Shepherd, PhD, Peter MacCallum Cancer Center; Allison Werner-Lin, PhD, LCSW, Senior Advisor, National Cancer Institute; Camella Rising, PhD, MS, RDN, National Cancer Institute; and Pat Boyd, PhD, National Cancer Institute
- **"Nutrition: A Pillar of Health"**  
Camella Rising, NCI Researcher and Registered Dietitian
- **Winter 2021 Issue**  
Meet five members of Dr. Schiffman's lab
- **Spring 2021 Issue**  
Meet Lamia Alsubie, LFS Saudi Arabia Chair
- **Fall 2021 Issue**  
Meet Camella Rising and Anna Joy Ryan, LFS Canada Youth Chair

Scan this QR code to learn more about the LFS Youth Program.



# Making Connections During Pandemic



## LFS and COVID-19 Webinar Answers Questions

The LFSA and Drs. Joshua Schiffman and Kara Maxwell hosted a special webinar for LFS families to share insights on the COVID-19 vaccines and concerns within the LFS community. Guest panelists addressed audience questions about vaccine reactions for those with LFS and studies supporting whether it is safe for LFS patients and cancer patients undergoing treatment. A recording of the webinar is available on [lfsassociation.org](http://lfsassociation.org).

## LFSA Announces New Chapter in Africa!

The LFS Association launched its newest chapter in Africa, which is chaired by Samuel Oliech Omolo. Now with 11 international chapters, LFSA has a presence on six continents. Other chapters include Latin America, Canada, France, Germany, India, Japan, Netherlands, Australia/New Zealand, Saudi Arabia and Singapore.

"We want to support people with LFS and reach them no matter where they are," said Jenn Perry. "It is especially difficult when you have a rare disease, like LFS. We need people fighting for LFS research, treatment and care in every corner of the world."

Each international chapter is chaired by a medical professional who is based in that region, so they can connect LFS patients with the support they need locally and share LFS resource materials in their native languages. They also manage cultural demands and medical systems unique to each country.

## LFS Awareness Saves Lives



5/3 p53

The LFSA is committed to raising awareness of Li-Fraumeni syndrome because it literally saves lives. An improved understanding and recognition of LFS increases the likelihood of diagnosis, early cancer detection, and better-informed cancer treatment decisions. To encourage LFS awareness, LFSA kicked

off its inaugural **International LFS Awareness Day** on May 3. The day, 5/3, was chosen for its connection to p53, the cancer-fighting protein that is damaged in LFS patients.

Throughout the month of May, the LFSA community promoted a better understanding of LFS, worldwide. LFS families shared their stories and LFSA informational products through their personal and professional networks. LFSA President Jenn Perry appeared in a video explaining the background of Li-Fraumeni syndrome and talked about the founders of the syndrome, Drs. Frederick Li and Joseph Fraumeni, Jr.

## Community Fundraisers Get Creative

Friends of LFSA found creative ways to fundraise for the association this year. Thousands of dollars were raised to help us continue critical patient programs and boost LFS awareness. Here are a few ways LFSA supporters rallied their communities.

### Rollerblading Event

LFSA Australia & New Zealand held a rollerblading event last spring to raise funds for the association and build awareness about LFS and p53 in their region. The group also designed an awesome p53 sweatshirt!

### LFSA Face Masks

A generous supporter donated LFSA face masks to be given as gifts to donors during LFSA's "Share the Love" campaign in February.

### Winning Essay

Addy Black wrote an essay about LFS for her school, Georgia-Cumberland Academy, and was awarded \$1,000 for LFSA, the non-profit she selected.

### Colorado Mountain Hike

Noelle Johnson and her team hiked Horsetooth Mountain in Fort Collins, CO, in their annual LFS Fight Club event. The 6-mile hike featured a 1,600-ft. elevation gain and a challenging scramble to the summit.

One week of the month was dedicated to recognizing the LFSA International Chapters and all they do for the global LFS community. Another week focused on the LFSA Youth Program for LFS children and young adults, and the importance of empowering young people with LFS to drive key aspects of their healthcare and decision making.

Intended to be an annual event, the first International LFS Awareness Day proved a big success and answered a much-needed call for LFS awareness. Look to May 3rd, 2022, for more opportunities to share your LFS stories and open more eyes to p53, LFS and the cancer connection that can impact all.

The LFSA would like to express its deep appreciation to the National Cancer Institute's Division of Cancer Epidemiology and Genetics for their unwavering support and dedication to the LFS Association, our mission, and our inaugural International LFS Awareness events!