

# Celebrating Resilience, Resolve & Results!

2020 LFSA IMPACT REPORT



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

# A Message from the LFSA President

Dear Friends of the Li-Fraumeni Syndrome Association,

Without a doubt, 2020 has been an extremely challenging year. A pandemic, social and political unrest, catastrophic wildfires and floods, economic hardship...even giant hornets. It was one of those “what next?” kind of years.

Yet, despite the challenges and even because of them, the LFS Association pivoted, grew, and evolved with unwavering focus on its mission to support and advocate for individuals and families with Li-Fraumeni syndrome. In this year’s Impact Report, we recognize the silver linings of 2020, and we celebrate the resilience, resolve, and results of our LFS community.

We celebrate:

- **RESILIENCE** to bounce back after setbacks
- **RESOLVE** to stay focused on our mission
- **RESULTS** of connecting our LFS medical, scientific, and patient community like never before

Yes, the COVID-19 pandemic forced us to cancel the in-person events we had planned this year. Silver lining? We moved the LFSA’s Youth Conference and the 5th International LFS Association Symposium to an online format. This move allowed us to reach more people than ever before and connected LFS doctors, scientists, genetic counselors, and patients around the world in a whole new way.

In another 2020 silver lining, the LFSA is one of 30 organizations to be awarded the Chan Zuckerberg Initiative

(CZI) grant for the association’s patient-led approach to supporting community members and financing research for rare diseases. We are so grateful not only for the timing and funding of the CZI grant, but also for the education and training they have provided to help LFSA grow as an organization and develop new initiatives to support our LFS community.

And grow we did! With the addition of our newest chapter in India, LFSA now has chapters in 10 different countries. Having an active international presence allows us to support and learn from one another to advance LFS research and treatment.

With all the craziness of 2020, we had every excuse to slow down or put a hold on starting anything new until the crazy eased. But we didn’t. In fact, the LFS Association likely had its busiest year yet with new initiatives like the Scientific Advisory Board, new grant program, expanded Youth Program, and targeted webinars to answer timely questions, ease fears and build community. We didn’t stop because Li-Fraumeni syndrome doesn’t stop.

We are resilient despite adversity. We have the resolve to make a better future for individuals and families with LFS. And, we achieve results. Join me in celebrating the silver linings of 2020.

— Jenn Perry,  
LFSA President, LFS patient



# Medical Director’s Message

As we celebrate the 10th anniversary of the founding of the LFS Association, we reflect on our accomplishments and, more importantly, look to a better future for individuals and families with LFS. I recall that first meeting organized by the National Cancer Institute ten years ago. At the time, LFS families felt isolated, unconnected, and they faced significant uncertainty regarding where to turn for help with medical information, guidance, and support.

My family was similar. As an oncologist, but also a husband and father of those with LFS, I had those same feelings.

The foresight ten years ago to bring LFS families together with LFS researchers cannot be overstated and led to the formation of the LFSA.

Today, ten years later, we find ourselves in a much better place thanks to the coalition of LFS patients, researchers, and physicians. We are so grateful to

the hundreds of volunteers who organized, fundraised, and served in leadership for the LFSA. Your efforts have helped us raise over \$2 million throughout the years to provide grants for LFS research and drive patient support initiatives, like whole body MRI screening studies and the LFS database housed at the City of Hope.

The LFS research and physician community is incredibly talented, caring, and dedicated to finding answers for our families with LFS. Over the last decade, we’ve seen dramatic changes in the therapeutics of treating cancer, breakthroughs in understanding *Tp53* and its connection to LFS, and critical support for screening and early cancer detection.

There is much more work to be done. The LFS Association is poised to lead the way in this charge, always keeping in mind the patient voice and always looking to a future without LFS.

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







## LFS Association Symposium Proves a Virtual Success

Originally planned as a live in-person event to be held in Boston, the 5th International LFS Association Symposium was transformed into a virtual event for the first time in its history. No small feat, LFSA quickly transitioned the live meeting format into a virtual platform that reached more LFS families than any other symposium and proved an unprecedented success.

The LFS Association Symposium was hosted by Dana-Farber Cancer Institute in partnership with the LiFE Consortium. Over two weekends, the symposium connected scientists, doctors, geneticists, and LFS families from around the world virtually through the REACH 20 LIVE online platform.

The coordinated effort involved developing and launching the REACH 20 LIVE online event platform and mobile app and organizing the live meeting and sessions with nearly 50 scientists, doctors, geneticists, and other presenters. A huge LFSA thank you goes out to Dr. Judy Garber and her team at Dana-Farber Cancer Institute and Dr. David Malkin of Toronto Sick Kids, who coordinated the medical/scientific portion of the meeting, creating a robust agenda.

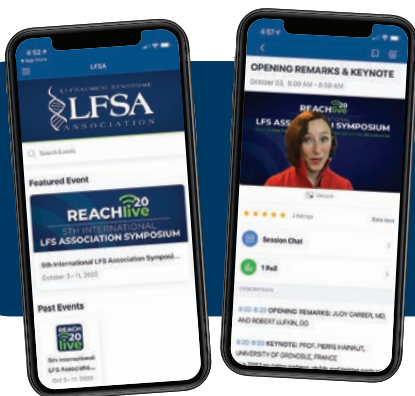
Sessions covered a wide range of LFS scientific topics, such as the latest research on early tumor detection, surveillance and interpreting *Tp53* variants. Other event highlights included the Ask a Genetic Counselor session, which featured small group breakout rooms where panels of genetic counselors answered

questions for LFS families. Another session flipped the script and allowed doctors, scientists, and genetic counselors to pose questions to LFS Youth about life with LFS. The online platform gave event attendees from all over the world the opportunity to participate in the meeting, allowing them to watch the recorded sessions on-demand if they were hours off from our time zone.

More than double the attendance of previous meetings, 470 people joined the event, with over 330 LFS family members who participated. The meeting platform provided them the opportunity to connect with other families (and presenters) through the social platform, which included a social wall, and group and private chat features.

Because the sessions have been recorded and are still available in the event platform and app, the information and materials have become a valuable educational resource for LFS families, physicians, scientists, and students who may have missed the live meeting. At present, total watch time of the symposium streaming video was more than 20 days from 17 countries!

The impact of the 5th International LFS Association Symposium will continue into 2021 and beyond as more people watch the recorded sessions and learn from the top Li-Fraumeni syndrome experts in the world.



Download the new LFS mobile app! Get access to all REACH 20 sessions at your fingertips. Connect with hundreds of LFS families, doctors, geneticists and researchers today!



# New Scientific Advisory Board to Drive LFS Research



The LFS Association is thrilled to announce its new Scientific Advisory Board. In addition to the association's Medical Advisory Board and Genetic Counseling Advisory Board, the newest board brings together top scientists in the field of Li-Fraumeni syndrome.

Co-chaired by Dr. Maria Isabel Achatz and Prof. Arnie Levine, LFSA's Scientific Advisory Board will help develop and guide new and ongoing research around LFS and *Tp53*.

"Our goal is to stimulate original Li-Fraumeni syndrome research," Achatz said. "We've only begun to organize, but we have the top international experts in molecular biology, immunology, clinical research and *Tp53*."

"Our primary goal is to identify research problems that are right for action, put a team together, and find funding to pursue research that will help patients with LFS," Levine said. "We'd also like to meet with LFSA's Medical Advisory Board to look for joint projects that we can work on together. This synergy of medical doctors and research scientists will benefit LFS patients around the world. It's wonderful to have an association like LFSA that connects patients with researchers and scientists in the ways that they do."

## LFSA'S SCIENTIFIC ADVISORY BOARD

**Dr. Maria Isabel Achatz, PhD**

*Hospital Sirio-Libanes, Sao Paulo, Brazil*

**Prof. Arnie Levine, PhD**

*Princeton Institute for Advanced Studies, New Jersey, USA*

**Prof. Pierre Hainaut, PhD**

*Universite 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*

**Sean Downing, PhD**

*Takeda, Boston, USA*

## LFSA Volunteers Take Fundraising to a New Level

The mandate to social distance put a damper on in-person events for fundraisers after the first quarter this year. However, friends of the LFSA stepped up with some creative ways to raise awareness about LFS and encourage donations.

### **Philpott Memorial Bowling Tournament, Sacramento, CA**

Michele Williams coordinated the 3rd Annual David Philpott Memorial Bowling Tournament, inviting people to bowl to help fight LFS and childhood cancer. The event also featured live music and raffle prizes, and raised more than \$1,600!

### **Higgins Family Sock it to Cancer for LFSA, Sayre, PA**

Kathy and Jamie Higgins held several fundraisers this year, selling sock cupcakes and crafts in memory of their two daughters, Maureen and Kerry. Kathy spoke to people about LFS at various craft shows, passed out LFS awareness brochures, educating the public about LFS. Despite having to postpone their annual Mo Songs For Kerry music event to 2021, the Higgins raised over \$1,300 in cupcake sales for the LFSA Youth Program!

### **LFSA Fall Gymnastics Invitational, Boston, MA**

The third annual LFSA Fall Gymnastics Invitational was dedicated to Henning Sablowski, LFSA's first LFSA Youth Chair in Germany. After much work to ensure COVID-19 health and safety precautions, the gymnastics competition and fundraiser brought gymnasts together to compete after months away from the gym and raised more than \$8,500 for the LFSA Youth Program!

### **Facebook Fundraisers & Company Matching Gift Programs**

What fun and such great support shown in the face of adversity this year when more than 20 members of our community held Facebook fundraisers to benefit the LFSA this year! LFSA received more than \$5,000 in 2020 through multiple Facebook fundraisers. The social media network has made fundraising for your favorite nonprofit an easy process, so be sure to consider LFSA to fundraise for a birthday, special occasion or just because!

# Celebrating Collaboration & The Power of Us



In 2020, we celebrated two major milestones in LFS history. This year is the 30th anniversary of the discovery of *Tp53* and its involvement with Li-Fraumeni syndrome, and it marks the 10th anniversary of the first Li-Fraumeni Syndrome Association meeting at the National Cancer Institute.

The first family pedigree with LFS was discovered in 1969 by Drs. Frederick Li and Joseph Fraumeni. But it wasn't until 1990 that the LFS connection to *Tp53* was realized. Dr. David Malkin, LFSA Medical Advisory Board member, who co-discovered this connection with Dr. Stephen Friend, reflected on the significance of their discovery during a session at the 5th International LFS Association Symposium in October.

It started with a question: "What causes Li-Fraumeni syndrome?" Malkin said. "There was no rational reason to come up with a plan other than we made the decision to try," he said. The road to answering this question wasn't smooth. In fact, their first theory proved altogether wrong. But, such is the beauty of science. The missteps, trials, errors and flat out wrongs lead us to discover the right.

Malkin and Friend pivoted to the *Tp53* gene. The *Tp53* protein was initially thought to drive cancer, but later research proved it actually suppressed the growth of tumor cells.

In 1990, Dr. David Kim and a team of scientists at the Massachusetts General Hospital Cancer Center sequenced the genes of five families with LFS. Malkin recalls Kim reading the family sequence out loud, while Malkin read the normal *Tp53* sequence. "We realized there was a mutation in Family 1 that was carried in two members of the family at the time," Malkin said.

"Steve (Friend) came in the next morning. We showed him the data, and for the next three weeks all we did was repeat the sequencing over and over again to ensure it was correct," Malkin said.

This discovery that inherited (germline) mutations of *Tp53* are the primary cause of LFS opened the door for predictive and diagnostic testing. It also opened up a host of new questions that still need to be answered.

## The priorities of the LFS Association are patient-centered:

- How do we prevent cancers in LFS patients?
- What are the best approaches to early tumor detection?
- How do we manage the psychosocial consequences of LFS?



The LFS Association was formed in 2010 with the LFS patient community at its heart. A meeting at the National Cancer Institute brought together LFS researchers and, for the first time, LFS patients and family members. There, the Li-Fraumeni Syndrome Association was formed to partner with the Li-Fraumeni Exploratory (LiFE) Consortium, an international and multidisciplinary alliance of scientists, clinicians, psychologists and genetic counselors.

The LFS Association brought the patient forward, bringing real people with LFS into the forefront as the reason behind the research. As we celebrate our 10th Anniversary of this coalition between science and patient, we recognize our progress and are grateful for the medical, scientific, and psychosocial advances.

As we carry out our mission to support and advocate for individuals and families with LFS, we also look to our future. We look to a future of hope, a future with answers to looming questions about LFS and, ultimately, a future without LFS.

## LFSA'S MISSION

**The LFS Association provides a wide range of information, advocacy and support services for individuals and families with Li-Fraumeni syndrome. We support a consortium of researchers, medical providers and caregivers to further research and promote optimal care for the LFS community.**

## 2020 LFS Association Global Pioneer Award

Dr. David Malkin was recognized by the LFSA at the 5th International LFSA Symposium with the 2020 Global Pioneer Award for his contributions to the LFS community for research, clinical care, education, mentorship and patient advocacy. As the co-discoverer of the relationship between *Tp53* and LFS, and leading the research and development of early screening and treatment guidelines, known as the "Toronto Protocol," Dr. Malkin is reshaping the impact of LFS for families all over the world. Dr. Malkin is a dedicated advocate who serves on the LFSA Medical Advisory Board, is co-chair for the LFSA Canadian Chapter, and is part of the LiFE Consortium.





# Making Connections During Pandemic

In a year that kept many people apart because of the COVID-19 pandemic, LFSA was extremely active bringing the LFS community together. Through webinars and virtual meetings, LFSA connected LFS patients with one another and with medical, scientific, genetic, and even mental health experts. These are some of the events and initiatives LFSA put forth this year to connect our communities. Recordings of the webinars can be viewed on our website, [www.lfsassociation.org](http://www.lfsassociation.org).

## LFSA Youth Workshop Virtual Meeting



LFSA Youth Workshop 2020 was held virtually for the first time since its inception four years ago. A huge success, the virtual meeting brought together LFS youth from all over the world to connect with one another and learn from top experts in the field, including Dr. Kara Maxwell and

Jacquelyn Powers, LCGC, of Penn Medicine, Philadelphia; Dr. Josh Schiffman of the Huntsman Cancer Institute, Salt Lake City; and Dr. Maria Isabel Achatz of Unidade de Oncogene ca do Hospital Sirio-Libanês, Sao Paulo, Brazil.

## Expansion of Youth Program to Young Adults

The LFSA Youth Program was designed to help young people with LFS live their lives to the fullest and stay connected with one another. This year, LFSA expanded this program to include young adults ages 18 to 25 to address their specific questions and concerns as they transition from pediatric to adult medical care.

"We saw a real need in this age group as they faced transitioning to young adults, and we wanted to offer our support," Perry stated. The Youth Workshop featured sessions for young adults with LFS that focused on topics like new cancer risks in adulthood, the differences between adult and pediatric care, and how to self-manage the medical care and screening so critical to LFS patients throughout their lifetime.

## Li-Fraumeni Syndrome & COVID-19 Webinar



The LFS Association, together with the Huntsman Cancer Institute, hosted a special webinar for families with Li-Fraumeni syndrome to share insights on the COVID-19 virus and how it affects the LFS community. Five panelists with specialties in pediatrics, general

medicine and genetics answered attendee questions live during the webinar and addressed questions submitted in advance. Panelists spoke on COVID-19 risks for people with LFS, special precautions related to COVID-19 and cancer in general, and concerns over postponing scans, treatments, and elective surgeries. Panelists included: Joshua Schiffman, MD, Professor of Pediatrics, University of Utah and CEO of PEEL Therapeutics; Luke Maese, DO, Assistant Professor of Pediatrics, University of Utah; Wendy Kohlmann, MS, CGC, Genetic Counselor, Huntsman Cancer Institute, University of Utah; Jennie Vagher, CGC, Genetic Counselor, Huntsman Cancer Institute, University of Utah; and Kara Maxwell, MD, PhD, Assistant Professor of Medicine, UPenn.

## Cultivating Greater Happiness Webinar with Author Gina Vild



LFS patients are no strangers to uncertainty, but the COVID-19 pandemic added another potential layer of fear and isolation. To help navigate these times, the LFS Association invited Gina Vild, who co-authored *The Two Most Important Days – How to Find Your Purpose*

and *Live a Happier, Healthier Life*, to speak on resilience. Vild discussed the characteristics of happy, resilient people and offered techniques and practices to choose happiness, increase gratitude and, in turn, live a healthier, intentional, more purposeful life.

# LFSA Announces New Grant Program

The LFS Association has established a new grant program to help fund more scientific research focusing on Li-Fraumeni syndrome. The inaugural grant of up to \$45,000 is on track to be awarded in the first quarter next year.

LFSA Scientific Advisory Board Member Sean Downing, PhD, Takeda, Boston, will oversee the new grant process. "Because LFS is so rare, it doesn't get a lot of attention," Downing said. "By providing this grant, LFSA wants to raise awareness about the syndrome and drive more research to get a better understanding of LFS and provide better treatments for people living with the syndrome."

The first step in the process is to submit a letter of intent through a form available on the LFSA website,

[lfsassociation.org](http://lfsassociation.org). Letters of intent include basic information and a 500-word synopsis of the proposed LFS research.

LFSA's Scientific Advisory Board (SAB) will review the letters of intent to narrow down and invite the best candidates to submit full grant applications for further review. Once a grant is awarded, the board intends to stay actively engaged with the grantee to help guide the research, assist with getting it published and seek out larger grants to further the research.

"Our hope is to continue offering grants for LFS research on a consistent, regular basis, but that is highly dependent on continued funding to the LFSA," Downing added.