LFS. EVOLUTION. REVOLUTION.

OUR 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.

2018 IMPACT REPORT

LI-FRAUMENI SYNDROME
LFSA
ASSOCIATION

History of Li-Fraumeni syndrome (LFS) and the LFS Association

1969

Two NIH scientists, **Drs. Frederick Li and Joseph Fraumeni**, report a rare familial syndrome of multiple cancers in children and young adults, including sarcomas, breast cancer, and other tumors. This discovery of childhood tumors and cancer-prone families occurs at a time when little attention was given to the role of genetic susceptibility in cancer.

1979

The TP53 tumor suppressor gene is co-discovered by Drs. David Lane and Arnold Levine, and over time, is recognized to be the cause of a wide range of cancers. TP53 is currently one of the most studied genes in the world.

1982

Researchers in the United Kingdom are the first to coin the name **"Li-Fraumeni syndrome."**

1988

Drs. Judy Garber, Li, Fraumeni and colleagues document the elevated risk of subsequent cancers in 24 families with LFS, note the especially high risk for breast cancer in young women, and propose the first "classical" definition of LFS based on clinical and familial criteria. Drs. Louise Strong in Houston, Jillian Birch in Manchester, and Ros Eeles in London, provide important insights into the LFS component tumors and mode of inheritance.

PRESIDENT'S LETTER

Dear Friends of the Li-Fraumeni Syndrome Association,

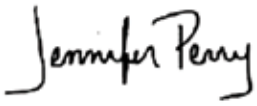
In almost 8 years of supporting Li-Fraumeni syndrome (LFS) patients and their families, furthering LFS research, and raising awareness of LFS, we at the LFS Association (LFSA) have seen first-hand how critical awareness, education, and research are to ensuring a better tomorrow for those with LFS. However, we realized from the beginning that it's not only important to provide resources to the LFS community, but to also build hope, inspiration, and strength. It's about creating a movement where we in the LFS community lift each other up, lift each other's spirits, and lift a helping hand, so that together, we can build a brighter future for LFS families.

In this annual impact report, you will learn about many ways that the LFSA has built hope, inspiration, and strength as we worked to raise awareness of LFS, educate the global LFS community, and provide critical advocacy and support services in 2018. To give you a quick preview, I've highlighted a couple of our accomplishments here:

- The LFSA held our 4th International LFS Association Symposium which brought together over 300 LFS patients and families, medical professionals, researchers, and scientists from across the globe to advance LFS and cancer genetics research, screening, awareness, support, and treatment.
- The LFSA launched the "LiFT for LFS" social media campaign to raise awareness for Li-Fraumeni syndrome and help strengthen the LFS community. The notion of raising something, anything of inspiration up in the air is meant to demonstrate support of the cause. We've had friends get creative by lifting a pen to write a letter of encouragement or lifting their arms to hug a loved one.

The LFSA knows that we alone can't address all of the issues facing the LFS community, and the families and medical professionals with whom we partner will still have challenges to address in 2019. But we can provide hope, and we can create opportunities so that patients with LFS feel like they have a chance for their lives to be better and their futures to be brighter – and this is only because of contributions of time, efforts and donations by dedicated supporters like you. On behalf of the LFSA, I want to say thank you and leave you with one question: In 2019, what will YOU lift for LFS?

Jenn Perry



LFSA President

MEDICAL DIRECTOR'S MESSAGE

In 2018, the LFSA has successfully continued our efforts to foster scientific advancement that will improve the lives of those with LFS. The 4th Annual LFSA Symposium was held in Toronto in April 2018 and hosted by The Hospital for Sick Children and the LiFE Consortium. The symposium was a tremendous success highlighting the unprecedented global interest in understanding the complexities of Li-Fraumeni syndrome – both from medical professionals and researchers but also affected families. From novel LFS modeling strategies, to efforts to improve LFS screening protocols, to potential future therapies for LFS related cancer, there has been remarkable progress since our last symposium. Despite this we are aware that there is more work that needs to be done. With your help in 2019 it is our goal to further expand LFSA's worldwide reach in the medical and research community supporting novel scientific endeavors that will truly change the future of those affected with Li-Fraumeni syndrome.

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

1990

A multi-institutional team led by **Drs. David Malkin and Stephen Friend in Boston discovers that inherited ("germline") mutations of TP53 are the primary cause of LFS.** This opens the door for predictive and diagnostic genetic testing.

1992

A team led by Drs. Alan Balmain and Larry Donehower in Houston creates the first p53-deficient mouse. It has a very high incidence of cancer that is subsequently shown to occur earlier when the mice are exposed to radiation.

2001

A collaboration of investigators in Brazil and Memphis **describe a unique germline TP53 mutation in children with adrenal cortical cancer in southeastern Brazil.**

1992

Recommendations that address clinical, psychosocial, ethical, economic and legal ramifications of genetic testing in LFS with applications to other genetic disorders, particularly in children, are published.

1998

A team led by Drs. Li and Fraumeni documents the elevated risk of subsequent cancers in LFS patients, even outside the radiation field of a primary malignancy.

2004

Teams led by Drs. Gigi Lozano in Houston and Tyler Jacks in Boston describe the first TP53 mutant mouse models of LFS, which are subsequently used to better understand how cancers develop and progress.

4TH INTERNATIONAL LFS ASSOCIATION SYMPOSIUM

The 4th International LFS Association Symposium entailed a combination of plenary sessions, workshops, poster sessions, panel discussions, evening events and a welcome reception. We consider ourselves extremely fortunate to have hosted 45 guest speakers and over 300 LFS patients and family members, researchers, scientists, and medical professionals from across the globe!

Our hearts were especially warmed by the attendance of **Dr. Joseph Fraumeni and his lovely wife, Tricia, as well as the late Dr. Frederick Li's wife, Dr. Elaine Shiang.** During our celebration at the Hockey Hall of Fame, they were presented LFS Association Global Pioneer Awards for the identification of the familial cancer syndrome.

The LFSA would like to thank **Dr. David Malkin** for his hard work organizing a rigorous scientific agenda, The Hospital for Sick Children staff members **Gaetano, Arlene & Jodi** for their helpful contributions that went above and beyond, and The Hospital for Sick Children in Toronto, Ontario, for graciously hosting the Symposium.

"*The 4th International LFS Association Symposium was an unforgettable experience for me. It was such a unique opportunity to be able to meet and talk with such a wide range of people – researchers, doctors, genetic counselors, students, and fellow LFS patients and families – all in one place! I came away from the conference with many of my questions answered, a wealth of information on promising research, and a strong sense of support from all of the new friends and connections I had made. It was a tremendous learning experience and has positively affected the choices I make with my LFS-related healthcare.*"
- **Cameron Block**, LFS Patient

"*The 4th International LFS Association Symposium was simply amazing. It brought together families living with LFS and researchers in the field to discuss topics spanning from the clinical management of LFS to the basic biology of p53. The mixing of families and researchers (some of whom had never met a person with LFS before) created an environment that was engaging and energizing. It was also so inspiring to meet Dr. Joseph Fraumeni and remember the life and great accomplishments of Dr. Frederick Li. It is thanks to them that we are where we are today in terms of our recognition and understanding of LFS. Kudos also to Jenn Perry, the LFSA and Dr. David Malkin and his team for organizing such a monumental and groundbreaking Symposium. It has set the bar high and served to motivate all of us who work in the field to continue our efforts to find a cure for this challenging condition.*"
- **Kim E. Nichols, MD**, St. Jude Children's Research Hospital

"*The LFSA symposium in Toronto was a wonderful conference, from the cutting-edge research results being presented by world renowned medical providers to the special connections made with families who live with LFS. As a genetic counselor, I appreciated the high level of science and medicine presented at this meeting and also enjoyed the many opportunities to speak with the inspiring individuals who have LFS from active Youth Group members to older adults. The special sense of energy and hope conveyed throughout this symposium will help carry our collective efforts forward throughout the year!*" - **Katherine A. Schneider, M.P.H., L.G.C.**, Dana-Farber Cancer Institute

"*Attending the LFSA Symposium was an invaluable experience. The impressive collection of presenters shed light on the latest medical research and scientific advancements. I feel lucky to have been at the center of what's happening with our disease and to finally interact with fellow patients. We shared stories, hope, and an unspoken bond.*" - **Patrick Moscatiello**, LFS Patient

2007

Dr. Maria Isabel Achatz provides evidence that the 'Brazilian' TP53 mutation is a "founder mutation" derived from a common ancestor migrating long ago from Portugal. The spectrum of cancers in these families resembles those with "classic" LFS.

2010

NIH convenes a meeting of LFS researchers and, for the first time, LFS patients and family members, to generate plans for an international and multidisciplinary alliance of scientists, clinicians, psychologists and genetic counselors – the Li-Fraumeni Exploratory (LiFE) Consortium. At this meeting, families form the LFS Association (LFSA) to partner with LiFE and best meet the needs of the LFS patient community.

BY THE NUMBERS

LFSA SYMPOSIUM FACTS

325 PATIENTS
FROM
7 COUNTRIES
ATTENDED THE 4TH INTERNATIONAL LFS ASSOCIATION SYMPOSIUM

49 OF THE WORLD'S MAJOR LEADERS IN THE STUDY OF P53, LFS, AND CANCER GENETICS
PRESENTED

37 POSTERS WERE PRESENTED
BY STUDENTS AND RESEARCHERS

LFSA FEATURED
13 SPONSORS & EXHIBITORS

LFSA GLOBAL OUTREACH FACTS

280,000+
UNIQUE VISITORS TO THE LFSA WEBSITE

172 COUNTRIES
HAVE INQUIRED ON LFSASSOCIATION.COM

1130
FACEBOOK FOLLOWERS

LFSA ATTENDED
9 CONFERENCES
IN BRAZIL, CANADA, GERMANY, AUSTRALIA, AND THE UNITED STATES TO RAISE AWARENESS AND ADVOCATE FOR LFS PATIENTS AND FAMILIES

LFSA HELD
7 COMMUNITY EVENTS
IN GERMANY, ITALY, AND THE UNITED STATES TO RAISE FUNDS AND AWARENESS FOR LFS

LFSA LAUNCHED
5 NEW CHAPTERS
IN AUSTRALIA, FRANCE, GERMANY, SAUDI ARABIA, AND SINGAPORE

LFSA TEAM FACTS

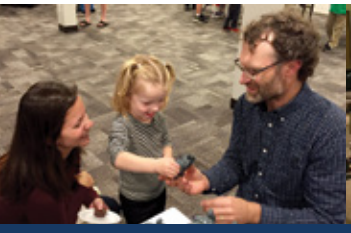
LFSA'S MEDICAL ADVISORY GROUPS ARE ASSOCIATED WITH
18 DIFFERENT
HOSPITALS AND MEDICAL INSTITUTIONS

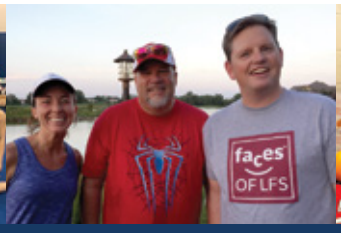
LFSA'S GENETIC COUNSELOR ADVISORY GROUP FEATURES
25 MEMBERS
REPRESENTING 7 COUNTRIES

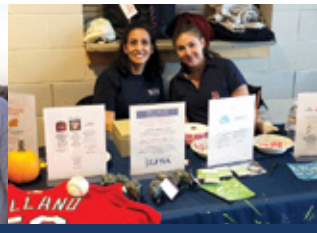
LFSA BOARD OF DIRECTORS IS COMPRISED OF
6 VOLUNTEERS
WHO ARE LIVING WITH LFS OR ARE A FAMILY MEMBER OF AN LFS PATIENT

COMMUNITY EVENTS

The LFSA has a powerful presence in local communities, thanks to an international group of dedicated volunteers that hold community events to raise funds, awareness, and support for the LFSA mission. This section highlights the efforts of real people making a difference with—and through—the LFSA.







Mo Songs for Kerry 5th Annual Cancer Fundraiser
Kathy and Jamie Higgins held the 5th Annual Mo Songs for Kerry Fundraiser, a celebration of life in memory of their two daughters, Maureen and Kerry. The event was unprecedented in size and success, featuring a large craft show, music festival, BBQ, crafts and games for the kids.

Elijah Johnson LFSA Benefit Concert
Elijah Johnson, young LFSA champion and talented flute player, raised awareness of LFS and funds for the LFSA by holding a classical musical concert at his church where he performed among other top Colorado high school musicians.

Noelle's LFS Fight Club Triathlon 2018
This was year number 4 for Noelle's LFS Fight Club! Noelle Johnson and her team once again completed a triathlon to raise money for the Li-Fraumeni Syndrome Association.

3v3 Memorial David Pagani Basketball Tournament
The inaugural 3v3 Memorial David Pagani basketball tournament entailed a 3-day event of fun, music, basketball, and LFS awareness. Thank you to David's ever-dedicated family and friends for organizing a spectacular and loving tribute to his memory. Funds raised at this event will help finance future LFSA programs and research initiatives.

Tee'd Off at LFS
This golf tournament was held to benefit LFSA and Kortne Waginger. Featured was a Captain's Choice Golf Tournament, and a lunch and banquet dinner with live music entertainment.


LFSA *SALUTE!* GALA AND FUNDRAISERS

***Salute!* Hartford 2018 Gala**
The LFSA held our 4th *Salute!* Hartford Gala at the Society Room in Hartford, CT to raise funds for LFS research initiatives. The event was our largest fundraiser yet, featuring cuisine stations, wine tastings, fabulous prizes, live music by the award-winning Shaded Soul Band, local beer from the City Steam Brewery, and liqueur from the Hartford Flavor Company.

2018 LFSA Fall Invitational
The LFSA held our 1st Fall Invitational, a fun-filled gymnastics competition and fundraiser for LFS youth. The event entailed a USA Gymnastics sanctioned competition, a special fundraising drawing, and prizes for attendees.

2011

Dr. Malkin and colleagues at the Hospital for Sick Children in Toronto develop screening recommendations for early cancer detection in carriers of the defective TP53. The **"Toronto protocol"** provides a comprehensive program of clinical, biochemical and imaging, including whole-body MRIs.

2015

Researchers in France, led by Drs. Thierry Frebourg and Laurence Brugieres, update the "Chompret criteria" further refining the clinical and familial characteristics widely used to help identify potential carriers and facilitate the diagnosis of LFS.

2016

The LFSA Medical Advisory Board is formed, followed shortly thereafter with the formation of the LFSA Genetic Counseling Advisory Group.

EDUCATING AND CONNECTING LFS PATIENTS

LFSA Youth Newsletter
April 2018: The LFSA launched our first LFS Youth Newsletter designed to help young people with LFS live their lives to the fullest and stay connected with one another. The first issue featured a Q&A with a Pediatric Oncologist, an interview with a Genetic Counselor, and more! Stay tuned for upcoming issues in 2019!

LFSA Germany Family Conference
July 2018: The LFSA held our first LFS family conference in Germany. Researchers from around the country presented updates on LFS and how to actively take steps to improve your health while lowering your risk factors for developing cancer. Special thank yous are in order for Dr. Christian Kratz, LFSA Medical Advisory Board Member, and Claudia Sablowski, Co-chair of the LFS Association's Germany Chapter, for their leadership organizing this conference.

Ask A Genetic Counselor Webinar Series
October 2018: The LFSA held our first "Ask a Genetic Counselor" webinar.

2018 GOING FROM RARE TO AWARE!

LiFT 4 LFS Movement #Lift4LFS
In 2018, we launched the "LiFT for LFS" campaign to raise awareness for Li-Fraumeni syndrome and help strengthen the LFS community. The "LiFT for LFS" campaign asks our friends to raise something, anything of inspiration, up in the air to demonstrate your support of the LFS community.

What will YOU lift for LFS?
It's easy. Just capture a photo or video of yourself lifting something of inspiration, whatever resonates with you, and then share on social media using #Lift4LFS.

The LFSA attends, presents, and exhibits at industry annual conferences to educate and spread awareness of LFS and the services of the LFS Association. In 2018, we participated in the following events:

- Rare Disease Week and Rare Disease Day at the National Cancer Institute in Washington D.C.
- American Society of Preventive Oncology Annual Conference in New York City
- American Association of Cancer Research Annual Meeting in Chicago, Illinois
- American Society of Clinical Oncology Annual Meeting in Chicago, Illinois
- The Cancer Predisposition Family Conference for LFS at St. Jude Children's Research Hospital in Memphis, Tennessee
- Star Island Corporation DIY Experience Conference in Rye, New Hampshire
- KConFab in Kingscliff, Australia
- Global Genes' 2018 RARE Patient Advocacy Summit in Washington D.C.
- NSGC Annual Conference in Atlanta, Georgia

2017

The LFS Association pilots its first Youth Workshop with teenage participants from around the world, and launches international chapters in Germany, Saudi Arabia, and the Netherlands, in addition to Canada, Australia/New Zealand, and Brazil.

2017

New screening recommendations are published based on the modification of the "Toronto protocol." Comprehensive consideration is given to the impact on patients to maximize participation in early tumor detection screening.