

“Identity formation in adolescents and young adults with a rare hereditary cancer syndrome: I’m scared to go have dreams.”

Wilsnack, C., Fishstein, A., Rising, C. J., Boyd, P., Sleight, A. G., Khincha, P.P., & Werner-Lin, A.
12th Annual Texas AYA Oncology Conference, 2022

Publications

Oba L, Best AF, Mai PL, Achatz MI, Albert PS, Savage SA, Khincha PP. “**Utility of interim blood tests for cancer screening in Li-Fraumeni syndrome.**” *Fam Cancer* 2021.

Siegel A, Bremer RC, Klein WMP, Savage SA, Loud JT, Khincha PP. **Uptake and timing of bilateral and contralateral risk-reducing mastectomy in women with Li-Fraumeni syndrome.** *Breast Cancer Res Treat* 2021.

Frone MN, Stewart DR, Savage SA, Khincha PP. “**Quantification of Discordant Variant Interpretations in a Large Family-Based Study of Li-Fraumeni Syndrome.**” *JCO Precis Oncol* 2021 Nov.

Andrade KC, Khincha PP, Hatton JN, Frone MN, Wegman-Ostrosky T, Mai PL, Best AF, Savage SA. “**Cancer incidence, patterns, and genotype-phenotype associations in individuals with pathogenic or likely pathogenic germline TP53**

COVID-19

COVID-19 has affected everyone across the globe, and individuals with LFS and their families are no exception. Our team has had the opportunity to apply our expertise in collaboration with other NIH investigators studying the effects of COVID-19.

Many of you filled out the COVID-19 in Rare Disease Communities surveys - thank you! The data is incoming and will soon be analyzed.

An NIH study is looking at the response to the COVID-19 vaccine in persons with immunodeficiencies. If you are interested in participating, please see <https://clinicaltrials.gov/ct2/show/NCT04852276> or email NIAIDCovidVaccineStudy@niaid.nih.gov



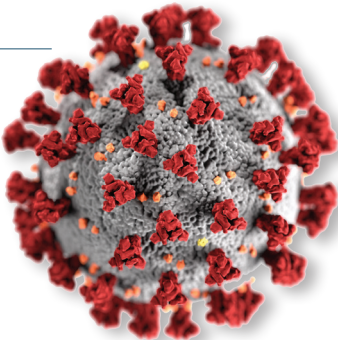
6th International LFS Symposium
Oct 14-16, 2022
Hybrid format
Hosted at the NCI

variants: an observational cohort study.” *Lancet Oncol* 2021.

Wilsnack C, Young JL, Merrill SL, Groner V, Loud JT, Bremer RC, Greene MH, Khincha PP, Werner-Lin A. “**Family Identity and Roles in the Context of Li-Fraumeni Syndrome: “No One’s Like Us Mutants”.**” *Health Soc Work* 2021.

Hatton JN, Sargen MR, Frone MN, de Andrade KC, Savage SA, Khincha PP. “**Spectrum and incidence of skin cancer among individuals with Li-Fraumeni syndrome.**” *J Invest Dermatol*. 2022.

Sleight AG, Rising CJ, Boyd P, Wilsnack C, Goodfellow M, Khincha PP, Werner-Lin A. “**I can control what I do with my daily life’: Occupational experiences of adolescents and young adults with Li-Fraumeni Syndrome.**” *J Occup Sci*. 2022.



Additional COVID-19 Resources

What people with cancer should know:
<https://www.cancer.gov/coronavirus>

Get the latest public health information from CDC:
<https://www.coronavirus.gov>

Get the latest research information from NIH:
<https://www.nih.gov/coronavirus>

SCREENING GUIDELINES FOR CANCER PREVENTION

Children

It is generally recommended that children with LFS follow screening guidelines published in the *Journal for Clinical Cancer Research* in 2017:

- Physical exam with full neurologic assessment, every 3-4 months
- Abdominal ultrasound with consideration of bloodwork, every 3-4 months

Adults

The National Comprehensive Cancer Network (NCCN) publishes consensus-based screening guidelines for adults with LFS that are reviewed about once a year. The 2022 guidelines are outlined below:

Breast Cancer Screening (Women)

- Breast awareness starting at age 18
- Clinical breast exam, every 6-12 months, starting at age 20
- Age 20-29 years, annual breast MRI (preferred) or mammogram if MRI is unavailable
- Age 30-75 years, annual breast MRI and annual mammogram
- Age >75 years, management should be considered on an individual basis
- Consider a risk-reducing mastectomy

- Annual whole-body MRI
- Annual brain MRI Note that different screening centers may slightly modify these guidelines.

Other Cancer Screening (Men and Women)

- Annual whole-body MRI, all ages
- Annual brain MRI as part of whole-body MRI or as a separate exam, all ages
- Annual dermatologic examination starting at age 18 or earlier if needed
- Colonoscopy and upper endoscopy every 2-5 years starting at age 25 or 5 years before the earliest known colon or gastric cancer in the family, respectively
- Comprehensive physical exam, including neurologic examination every 6-12 months, all ages
- New: Consider pancreatic cancer screening at age 50 if there is a close relative (first or second degree) with an exocrine (not endocrine) pancreatic cancer on the same side of the family as the *TP53* variant

Thank you for participating in our LFS study!
The strength of our study is in our participants.



NATIONAL CANCER INSTITUTE



Li-Fraumeni Syndrome (LFS) Study
Newsletter
Summer 2022

Division of Cancer Epidemiology and Genetics • Clinical Genetics Branch

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RETIREMENTS

Jennifer Loud, D.N.P., C.R.N.P.



A board-certified adult nurse practitioner, Dr. Loud served as a study investigator on the LFS study until her retirement in 2020, concluding 27 years of service at the National Institutes of Health (NIH). She was involved in multiple clinical and epidemiologic studies to learn about hereditary cancer and the psychosocial impact of individuals with high genetic risk of cancer. In addition to her responsibilities on the LFS study, Dr. Loud spent her career mentoring the next generation of researchers and clinicians across the Clinical Genetics Branch (CGB) in the Division of Cancer Epidemiology and Genetics (DCEG) at the National Cancer Institute (NCI) and at NIH.

Janet Bracci, R.N., B.S.N.



Ms. Bracci is a registered nurse with decades of experience in nursing-related administrative, consulting, educator, and practitioner roles. An experienced oncology and bone marrow transplant nurse, Ms. Bracci served as the lead research nurse on the LFS study for 10 years until her retirement in 2021. Ms. Bracci was one of the first and most consistent contacts that LFS participants had with the study.

STUDY TEAM MEMBERS

Principal Investigator

Payal Khincha, M.B.B.S., M.S.H.S., a board-certified pediatric hematologist/oncologist, is the principal investigator of the LFS study

Lead Medical Advisor

Sharon Savage, M.D., is the lead medical advisor for the LFS study and focuses particularly on the molecular and genetic aspects of LFS

Clinical Team

Margarita Aryavand, CAPT., U.S.P.H.S., M.S.N., C.R.N.P., is a board-certified nurse practitioner and a Commissioned Officer and Captain in the U.S. Public Health Service. She performs clinical evaluations for LFS study participants who come to the NIH for screening

Megan Frone, M.S., C.G.C., is the lead board-certified genetic counselor for the LFS study

Jessica Hatton, M.S., C.G.C., is a board-certified genetic counselor for the LFS study

Fellows

Kelvin de Andrade, M.Sc., Ph.D., is a research fellow in CGB who is involved in a series of genetic, clinical, and epidemiological studies

***Ixta Obregon** is a postbaccalaureate fellow working with the LFS study team on evaluating the immunologic status of individuals with germline *TP53* pathogenic variants and its association with cancer

***Mone’t Thompson** is a postbaccalaureate fellow working with the LFS study team on evaluating rare sarcomas and their diagnostic challenges

Study Managers

Renee Bremer, M.S., is the epidemiology program analyst in CGB. She is currently involved in several of the psychosocial analyses and follow-up data collection within the LFS study

***Cecilia Higgs, M.H.S.**, is a program manager in CGB. She leads the regulatory matters and manages the study files, including the protocol, consent documents, and submissions to the Institutional Review Board

Psychosocial and Behavioral Science Research Team

Camella Rising, Ph.D., M.S., R.D.N., is a research fellow investigating health behaviors, communication behaviors, and psychosocial needs of individuals and families living with LFS as a researcher on the adolescent and young adulthood LFS study

***Rowan Forbes Shepherd, Ph.D.**, is a postdoctoral fellow who is developing psychosocial interventions for young people with high genetic risk of cancer

***Ashley Thompson** is a genetic counseling graduate student working on the psychosocial component of the LFS study

* Indicates new team members since our last newsletter

UPDATES, COLLABORATIONS, AND NEW PROTOCOL DEVELOPMENTS

New Study on Screening Experiences

We have launched a new study to understand the experiences of individuals undergoing cancer screening for LFS. If you have received cancer screening with us at the NIH Clinical Center, you are eligible to participate. The study involves a ~45-minute telephone interview with a member of our team and is aimed at understanding how individuals prepare for and deal with screening tests and results. If you are interested, please contact Dr. Khincha at 240-276-7267.

Patient Communication Update

Our research team is working to create an online communications **hub and research management system** (CHARMS). This system will replace our paper questionnaires and study consents and will allow new participants to enroll online. Eventually, the system will also support secure messaging. CHARMS is already up and running for other studies at the NCI, and we expect it will be available for LFS study enrollment later this year.

Update on the Cancer Screening Arm of the LFS Study

The COVID-19 pandemic forced us to pause the

Study Support

Stephanie Steinbart, R.N., M.P.H., is a research nurse who serves as CGB’s referral nurse

Kathryn Nichols, R.N., B.S.N., is the lead research nurse on the LFS study

Nicole Dupree, B.B.A., M.P.H., is a research assistant on the LFS study

Katharine Beebe, B.S., M.P.H., is a data manager on the LFS study

Please visit the NCI LFS website <https://lfs.cancer.gov> to find out more about the key staff members and their roles on the study.

cancer screening component of the LFS study at the NIH Clinical Center for some time, especially for participants traveling long distances. During this time, many of our screening participants were successful in receiving screening, including whole body MRI, at hospitals near them. This experience, which came after nearly a decade of cancer screening at the NIH, coincided with the planned timing for a comprehensive evaluation of our data to determine and further quantify the extent to which screening identifies cancers early and improves outcomes for people with LFS. With this in mind, we plan to close the screening arm of our study and fully transition our screening participants to local care by the end of 2022. This marks a momentous milestone in LFS cancer screening research, and we hope our data will further encourage screening availability and insurance coverage in years to come. This step will also increase our bandwidth to begin our next study.

Read an article highlighting some of our recent publications:

“Making Strides Toward Precision Medicine for Individuals with Li-Fraumeni Syndrome”
https://dceg.cancer.gov/news-events/news/2022/lfs-precision-medicine?cid=eb_govdel

RECENT PRESENTATIONS AND PAPERS

Presentations

“An Extended Case Series of Gastrointestinal Neoplasms in Li-Fraumeni Syndrome”

Hatton, J., Frone, M., Savage, S., Khincha, P.P. National Society of Genetic Counselors 39th Annual Conference, 2020

“Supporting reproductive decision making for adolescents and young adults with Li-Fraumeni Syndrome (LFS): Interprofessional perspectives from a U.S. National Cancer Institute (NCI) study”

Wilsnack, C., Werner-Lin, A., Rising, C. J., Boyd, P., Sleight, A. G., Hutson, S. P., Khincha, P. P. European Society of Human Genetics (ESHG) Annual Conference, 2020

“Cancer Incidence, Patterns, and Genotype-Phenotype Associations in an Observational Cohort Study of Individuals with Pathogenic or Likely Pathogenic Germline *TP53* Variants”

de Andrade, K.C., Khincha, P.P., Hatton, J.N., Savage, S.A. American Society of Human Genetics Annual Meeting, 2021

“Lung Cancer Case Series in National Cancer Institute Li-Fraumeni Syndrome Cohort”

Hatton, J.N., Seastedt, K.P., Oba, L., Savage, S.A., Hoang, C.D., Khincha, P.P. National Society of Genetic Counselors 40th Annual Conference, 2021

“Characterization of Gastrointestinal Neoplasms in the NCI Li-Fraumeni Syndrome Study”

Hatton, J.N., de Andrade, K.C., Frone, M.N., Savage, S.A., Khincha, P.P. 2021 Collaborative Group of the Americas on Inherited GI Cancers Annual Meeting

“Prevalence of autoimmune diseases in Li-Fraumeni Syndrome”

Obregon, I., de Andrade, K.C., Bremer, R., Khincha, P.P., Savage, S.A. American Society of Human Genetics Annual Meeting, 2021

“Family communication about Li-Fraumeni syndrome: Perspectives of adolescents and young adults”

Rising, C. J., Wilsnack, C., Boyd, P., Sleight, A. G., Huston, S. P., Khincha, P. P., Werner-Lin, A. Society of Behavioral Medicine’s (SBM) 2021 Annual Meeting and Scientific Sessions, 2021

“Challenging Interactions with Family about Li-Fraumeni Syndrome Experience by Adolescents and Young Adults”

Rising, C. J., Wilsnack, C., Boyd, P., Sleight, A. G., Hutson, S. P., Khincha, P. P., Werner-Lin, A. 71st Annual International Communication Association (ICA) Conference, 2021

“Breaking through barriers: Identifying healthcare challenges for adolescents and young adults living with Li-Fraumeni Syndrome”

Wilsnack, C., Rising, C. J., Sleight, A. G., Boyd, P., Khincha, P. P., Werner-Lin, A. 25th Annual Conference of the Society for Social Work and Research (SSWR), 2021

“Coping with healthcare needs for adolescents and young adults with Li-Fraumeni Syndrome (LFS): You’re having to put up a fight to take care of your health.”

Wilsnack, C., Rising, C. J., Boyd, P., Sleight, A. G., Hutson, S. P., Khincha, P. P., Werner-Lin, A. European Society of Human Genetics (ESHG) Annual Conference, 2021

“Cancer Risk Perceptions of Adolescents and Young Adults with Li-Fraumeni Syndrome: You Do Not Know If You Have Lived Half Your Life Already.”

Werner-Lin, A., Rising, C. J., Wilsnack, C., Boyd, P., Sleight, A., Hutson, S. P., Khincha, P. P. Society for Social Work and Research (SSWR), 2022



LFS Study Team having a 100% vaccinated lunch, November 2021

Pictured from left: Payal Khincha, Sharon Savage, Megan Frone, Rowan Forbes Shepherd, Ixta Obregon, Renee Bremer, Margarita Aryavand, Jessica Hatton, Camella Rising, Kelvin de Andrade