2023 ANNUAL REPORT

CURE LBSL

www.CureLBSL.org

Founders' Message

Dear Friends,

Amazing things happened this year! Not the least of which was our first-ever meeting with the U.S. Food & Drug Administration (FDA). The voice of the LBSL community was seen and heard by dozens of high-ranking government officials from both FDA and the National Institutes of Health (NIH). A huge thanks to all who participated. This was an important first step toward approval of future drug therapies.

Speaking of drug therapies, on page 6 we detail the state of research projects aimed at developing potential cures. The ASO therapies our team has developed have been shown to regulate gene expression in human cell models. Their gene therapies have been able arrest disease in both human cells and mice. Our foundation is preparing for the next round of FDA meetings, which are important to establishing clinically meaningful outcomes for human trials.

This was our second year of funding through the Chan Zuckerberg Initiative Rare As One grant. Participation in this program has been transformative for the organization and our community. Our intrepid Director of Patient Engagement, Melody Kisor, traversed the country representing LBSL at important meetings. Her interactions led to LBSL being included in new studies and featured in new public relations campaigns.

Perhaps one of her most important contributions has been the support she shows patients – hopping on trains to meet people at the airport, arranging translation services, and creating a set of invaluable patient protocols to help families communicate their unique needs to doctors and teachers. One mother expressed her gratitude through happy tears, saying, “Melody is an angel.” That sums up what she has meant to our organization over the last two years.

We would be remiss if we did not thank our donors. You made possible all the progress outlined in this report. As we have said many times, this effort is fueled by love, and we would be nowhere without your unwavering support and generosity.

Sincerely,
Beth and Michael McGinn
Beth and Mike

Our Team

2023 Board of Directors

Edward Blakey

Beth Crane
Chair

Chris Gulugian

Beth McGinn
Co-Founder

Michael McGinn
Co-Founder

Megan Mitchell

Lance Stern

Staff

Melody Kisor, MS
Director of Patient Engagement

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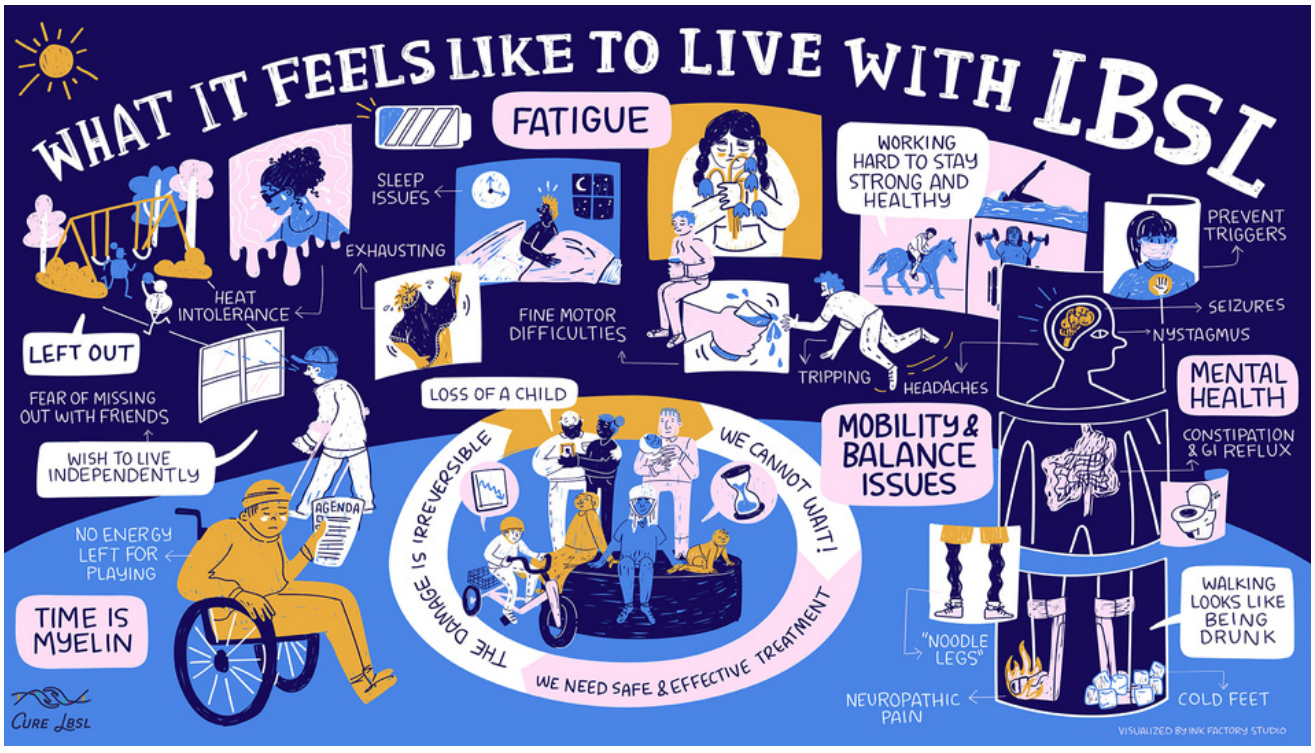
Our Work

Capturing the Patient Voice

Staff conducted nearly two dozen patient voice sessions as well as private interviews with patients and caregivers from across the globe. Through this careful listening, we discovered insights never before published, including a large percentage of people with disturbed sleep. These insights were shared with the FDA and will help drive research priorities.

As part of this effort we commissioned an emotional three-minute video about the real-world impact of this disease on patients and their families. The video has been shown to doctors, researchers, and drug regulators to help bring a human face to their work. (For privacy reasons, the video has not been made public.)

Cure LBSL also commissioned an illustration from Ink Factory Studios entitled "**What It's Feels Like to Live with LBSL.**" The art was shared with FDA regulators in May, and featured at NORD, Global Genes, and CZI Science in Society meeting last fall.



Our Work Cont...

Spreading Awareness

- September 20 was officially declared "**LBSL Awareness Day**." All month long we shared facts about LBSL on our social media accounts.

- We joined **EURORDIS** - the largest rare disease organization in Europe.

Supporting Patients

- We consulted experts to create **emergency protocols** for LBSL patients. Our website now offers resources such as patient care guides for doctors, **school safety and learning plans** for students, information about **fitness trackers and wellness apps**, and **travel resources**. This includes background information for teachers; a list of school needs and accommodations for administrators and special education staff; a sample email to new teachers; and a video presentation by a special education expert to help parents and students prepare for 504 and IEP meetings.

- We were busy spreading the good news: effective Oct. 1, 2023, diseases caused by "ARS" gene mutations, including DARS2 (LBSL) have their own ICD-10 code: **E88.43 Disorders of Mitochondrial tRNA Synthetase**. This internationally recognized code is used by doctors and health insurance companies for billing and tracking. Having this code could help ensure patients receive coverage of payment for services, and may also help researchers understand medical utilization and unmet needs.

- Launched an **international contact registry** to connect patients and families with resources and research opportunities. We have over 150 contacts from 27 countries and five continents in our registry, which is now available in 22 languages.

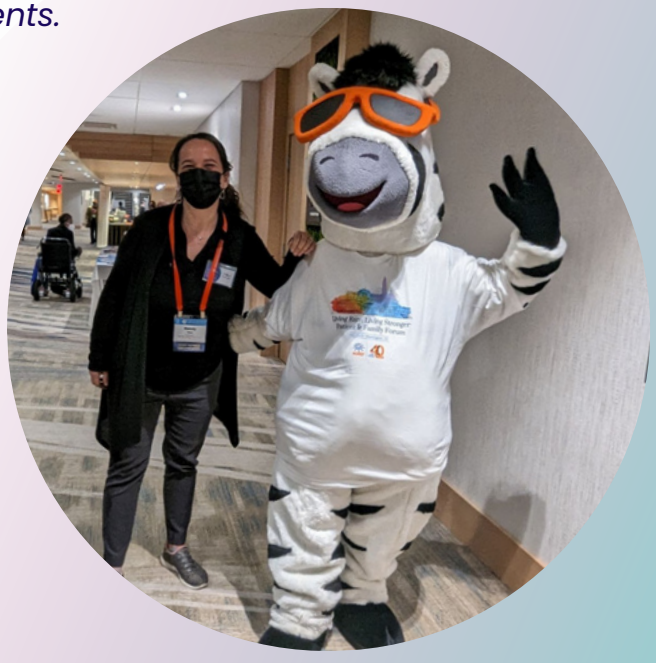
Conferences

Connections made at patient and scientific gatherings lead to new ideas and collaborations that advance research and improve lives. Here are some of the conferences and meetings we attended in 2023:

- Mitochondrial ARS (Mt-aaRS) Genes Scientific Symposium 2023 (virtual)
- Broad Institute of MIT & Harvard Rare Disease Day Event (virtual)
- Global Genes Rare Drug Development Symposium (Philadelphia, PA)
- NORD Living Rare, Living Stronger Patient and Family Forum (Washington, DC)
- World Orphan Drug Conference (National Harbor, MD)
- Global Leukodystrophy Initiative Meeting at ULF (Itasca, IL)
- United Leukodystrophy Foundation Scientific Symposium (Itasca, IL)
- United Leukodystrophy Foundation Family Conference (Itasca, IL)
- LBSL Breakout Session at ULF Conference (Itasca, IL)*
- C-Path Rare and Orphan Disease Conference (Washington, DC)
- Global Genes RARE Advocacy Summit (San Diego, CA)*
- Chan Zuckerberg Initiative–Science in Society 2023 (Newport Beach, CA)*
- n-Lorem Nano-rare Patient Colloquium (Cambridge, MA)
- NORD Breakthrough Summit (Washington, DC)*
- Rare Disease Clinical Research Network Meeting (Arlington, VA)

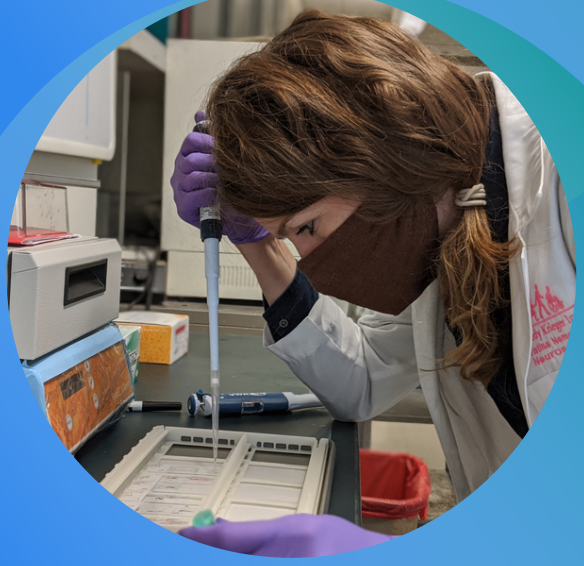
**Cure LBSL presented posters and/or conducted sessions at these events.*

Additionally, we attended monthly events with other patient advocacy groups as part of the **Chan Zuckerberg Initiative Rare As One Network** and the **Global Leukodystrophy Initiative** Patient Advocacy Committee.



SAVE THE DATE:
2024 International LBSL Conference
July 26 - 29, 2024
Baltimore, Maryland (USA) & Zoom

Research



Cure LBSL funds both clinical and preclinical studies at five hospitals and universities around the globe. This year, we provided nearly $886, 500 in grants and open source scientific tools to help advance cures.

Kennedy Krieger Institute, Baltimore, MD, USA
$518,000 in Funding

- Gene Therpay
- ASO Therapy
- Natural History Study

Children's Hospital of Orange County, CA, USA
$318,000 in Funding

- Metabolic Studies
- Drug Repurposing

Children's Hospital of Philadelphia, PA, USA
$30,000 in Funding

- Drug Repurposing

Hospital Pequeno Príncipe, Curitiba, Brazil
$20,000 in Material Support

- Natural History Study

University of Helsinki, Finland
$500 in Material Support

- Natural History Study

Fundraising

A special thank you to all who supported Cure LBSL in 2023. Your donations, big and small, kept hope alive for families living with LBSL.

$636,000 Angel Donors
$200,000 CZI Grant
$40,302 General Giving
$1,378 Facebook
$1,700 Giving Tuesday
$328 Amazon Smile

$879,708 Raised

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