



RESEARCHING TO CURE EPILEPSY

FALL 2022



Collaboration in epilepsy research accelerates progress. CURE Epilepsy is at the forefront of creating partnerships to drive advances.

Every day, I'm inspired by the scientific advancements that our investigators have made and how they have improved the outcomes for people with epilepsy.

In past reports, we have shared the impact of the work that we initiated in infantile spasms, epilepsy genetics, Sudden Unexpected Death in Epilepsy (SUDEP), and post-traumatic epilepsy (PTE). In this report, you will read about some of the new initiatives that we have undertaken to drive progress in areas of need, such as Jeavons syndrome and rare epilepsies.

The key to our success has been the commitment and tenacity of our scientists. They have taken on epilepsy's most difficult puzzles, motivated by the ability to help the 65 million people worldwide who have epilepsy. It has been an honor to accelerate the careers of young epilepsy researchers through our *Taking Flight Award*, which has led to discoveries and potential therapies that bring us one step closer to a cure. And we are grateful to our more seasoned researchers who use our *CURE Epilepsy* and *Catalyst* grant programs to look for ways to prevent epilepsy, modify the disorder, and develop new therapies.

Just as we rely upon a broad and diverse community of researchers to advance our understanding of epilepsy and the brain, CURE Epilepsy also depends upon a network of people with epilepsy, their families, and the larger body of champions committed to funding research that will bring us one step closer to a cure. CURE Epilepsy has funded over 280 grants, and we are incredibly appreciative of those individuals who give their time

JOIN US IN CHICAGO FOR OUR

25TH ANNUAL BENEFIT

SATURDAY, MAY 6, 2023

to ensure that the point of view of a person with epilepsy is represented in determining what grants we select for funding.

Our ability to fund the amazing research that we do is made possible by all of you who give generously to CURE Epilepsy. One of the primary vehicles for raising funds is our annual benefit held in Chicago. For the first time since the pandemic began, we gathered in person on June 1 for our 2022 benefit and raised over \$1.9 million. Whether you have recently begun contributing to CURE Epilepsy or you've been with us since our founding in 1998, I want to express my sincerest thanks and hope that you will help us continue the fight against epilepsy in 2023. Through research, there is hope.

With continued gratitude,

Beth Lewin Dean
Chief Executive Officer, CURE Epilepsy

PATIENTS FUEL OUR RESEARCH



Since our founding, we have developed a strategic scientific approach to ensure that the research projects we fund match our priority areas.

An important factor in our decision to award a grant is the project's potential to make a difference for real people who are living with epilepsy or at risk of developing epilepsy.

Several of our new projects going into 2023 were driven by families and patients, as they seek freedom from the physical and mental health effects of unpredictable seizures. In the following pages, read the stories of patients like Lincoln, who is inspiring us to pursue research into a rare genetic epilepsy, and Jack, who is benefiting from our investigations into post-traumatic epilepsy, which is a risk for military veterans and anyone who has had a head injury.

THE CURE EPILEPSY GRANT REVIEW PROCESS: FUNDING THE VERY BEST SCIENCE

CURE Epilepsy has funded over 280 grants for projects that will bring us one step closer to a cure.

CURE Epilepsy remains committed to funding patient-focused research by integrating the patient voice into the research we fund. In addition to our scientific reviewers, CURE Epilepsy's lived-experience reviewers also participate in the grant review process by reading research proposals from a patient or caregiver perspective. This ensures that the stakeholder point of view is critically represented during the review process.



**OUR PROCESS ENSURES
THAT ONLY THE VERY
BEST SCIENCE IS
SELECTED TO RECEIVE
CURE EPILEPSY GRANTS.**

* Letters of intent are reviewed by scientific reviewers. Full proposals are reviewed by scientific and lived-experience reviewers.

** Grants are selected by the CURE Epilepsy Research Team and Research Committee and approved by the Board of Directors.

PARTNERSHIP PROPELS RESEARCH TO CURE RARE GENETIC EPILEPSIES

With the increase in genetic research and the advancement of genetic testing in the past few decades, scientists have discovered over 1,000 gene mutations known to contribute to the development of epilepsy. Although passionate and driven communities of patients, families, and supporters have arisen around many of these individual gene mutations, they often lack the resources and infrastructure to support research.

CURE Epilepsy is committed to developing cures for everyone living with epilepsy, even when the epilepsy syndrome affects only a handful of people. To continue making a difference for children and adults living with rare epilepsies, we initiated discussions with leaders from several advocacy groups, which highlighted the need to develop research tools to drive research and understanding of these epilepsies.

Now, we are leveraging our expertise and leadership in the epilepsy community to invest in a collaborative effort to drive research in the rare epilepsies. In 2022, we created the **Rare Epilepsy Partnership Awards** and selected a handful of organizations to partner with. Together, we will co-fund one-year, \$100,000 grants that will help researchers develop:

- Rare epilepsy-specific cellular models and appropriate genetic animal models.
- Novel research tools and techniques to study rare epilepsies and identify new therapeutic strategies.
- Research tools and techniques demonstrating the biology that leads to a rare epilepsy.
- Patient registries to understand the natural history and disease progression.
- Technologies that will accelerate accurate diagnoses for rare epilepsies.

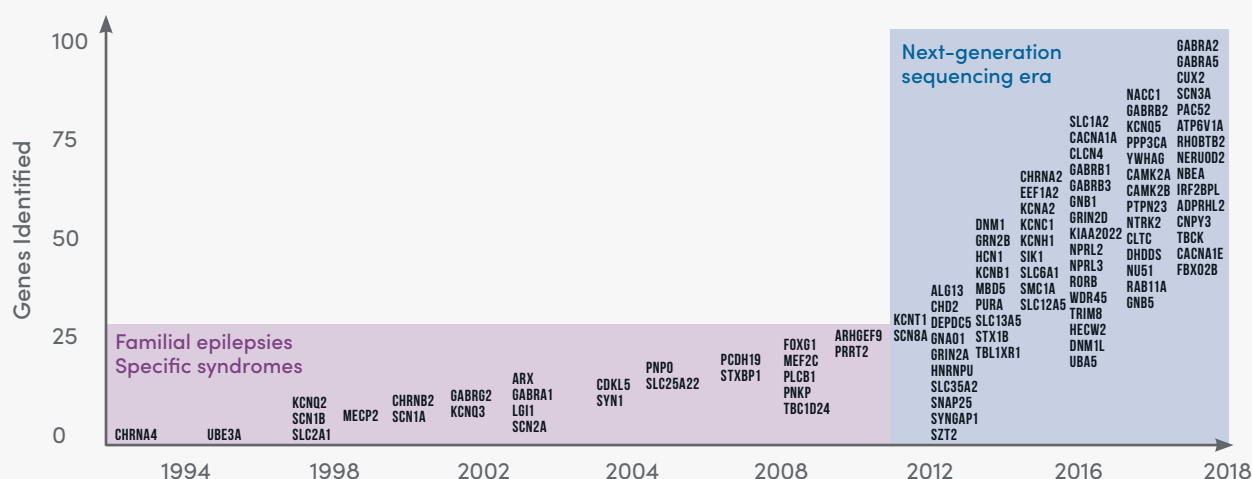
RARE EPILEPSIES

ARE THOSE THAT IMPACT LESS THAN 200,000 PEOPLE IN THE U.S.

ULTRA RARE EPILEPSIES

AFFECT APPROXIMATELY LESS THAN 6,500 PEOPLE IN THE U.S.¹

EPILEPSY GENE IDENTIFICATION HAS INCREASED RAPIDLY SINCE 1994 SINGLE-GENE MUTATIONS KNOWN TO CAUSE EPILEPSY



Content collected from the lab of former CURE Epilepsy Taking Flight Awardee Gemma Carvill, PhD, Assistant Professor of Neurology (Epilepsy/Clinical Neurophysiology), Pediatrics, and Pharmacology at the Northwestern University Feinberg School of Medicine.

¹ Harari S. Why we should care about ultra-rare disease. Eur Respir Rev 2016; 25: 101-103. Statistic based on current U.S. census population of ~332.5 million at an incidence of one patient per 50,000 people.



CURE EPILEPSY IS RESEARCHING FOR...

Lincoln

PENDLETON, SOUTH CAROLINA

Lincoln Tanner is an amazing little boy. He loves bubbles, trampolines, and cuddles, and he's one of only 200 people worldwide with a condition known as KCNT1-related epilepsy.

A few days after his birth, Lincoln began having strange episodes that were later determined to be seizures. When Lincoln turned purple at four weeks old, his mom, Abigail, and his dad, Justin, rushed him to the hospital. Lincoln soon began having about 200 seizures daily, at one point seizing every five minutes. He was diagnosed with malignant migrating partial seizures of infancy (MMPSI).

Lincoln's doctors prescribed medication that helped reduce his seizures tenfold, and the family joined an epilepsy support group to connect with others affected by the disease. When Lincoln was three, one of the group members, a little girl named Emma, tragically passed away from SUDEP. This alerted the Tanners to the risk Lincoln also faced. Now, Lincoln's parents put him on a hospital-grade monitor every night.

Today, at age five, Lincoln enjoys spending time with his older brother, Cash; older sister, Chloe; and younger sister, Shiloh. He is nonverbal and mostly immobile and enjoys communicating with music. In recent months, Lincoln has experienced the happiest and most comfortable time of his life, with an explosion of smiles and increased alertness.

Lincoln's family is grateful for the research that CURE Epilepsy is funding into rare epilepsies as well as the wealth of information available on SUDEP, which they believe can help save their son's life.

SNEAK PEEK: 2022 *Taking Flight* Grant Funded to Study KCNT1-Related Epilepsy

William Tobin, PhD, at the University of Vermont and State Agricultural College, has been awarded a one-year, \$100,000 *Taking Flight* grant to optimize cutting-edge gene and drug therapies for KCNT1-related epilepsy. Each treatment will be evaluated in terms of seizure control and ability to normalize brain activity, recognizing our goal to holistically restore normal function in the epileptic brain. **This project is being funded in partnership with the KCNT1 Epilepsy Foundation.**

Stayed tuned for the 2022 *Taking Flight* and *CURE Epilepsy* Award grantees announcement later this year.



Lincoln is an absolute joy. He's one of the best things to happen to our family. We're so glad we now know the true risk he faces from SUDEP – and ways to help prevent it – thanks to CURE Epilepsy's unrelenting research."

ABIGAIL TANNER, LINCOLN'S MOM AND EPILEPSY ADVOCATE



IMPROVING DIAGNOSIS AND TREATMENT FOR JEAVONS SYNDROME

Jeavons syndrome (JS) is an underreported epilepsy syndrome characterized by eyelid myoclonia with or without absences, eyelid closure-induced seizures or electroencephalogram (EEG) paroxysms (epileptiform discharges and/or seizures), and photosensitivity.² JS has a distinctive EEG pattern and a hallmark symptom characterized by marked, brief, repetitive, often rhythmic, fast jerking of the eyelids followed by an upward rolling of the eyeballs and a slow eyelid closure.

JS takes an average of 10 years to be correctly diagnosed.³ Doctors often mistake it for facial tics, Dravet syndrome, or random eye blinking. Patients experience brief seizures – usually six seconds or less – occurring multiple times per day. Unfortunately, JS can be difficult to treat, with seizures typically resistant to antiseizure medications.⁴ People with JS are at risk for developing cognitive and learning challenges if the condition goes untreated.

PATIENT-INSPIRED RESEARCH INTO JS

When a concerned parent approached CURE Epilepsy about his daughter's JS journey, we recognized a need to establish standards and guidelines for diagnosis and treatment as well as increase awareness among physicians and parents. We are leading this effort by convening and facilitating international pediatric epilepsy experts to review the available data and establish a consensus, where possible, on diagnosis and treatment.

“Many people I speak with, including fellow neurologists, have never heard of epilepsy with eyelid myoclonia, also known as Jeavons syndrome. This is an understudied and often underdiagnosed syndrome where the average diagnosis is 10 years. As researchers, we hope to shorten this diagnostic journey and eventually improve treatment outcomes for those living with epilepsy.”

ELAINE WIRRELL, MD
DIRECTOR OF PEDIATRIC EPILEPSY
MAYO CLINIC

QUICK FACTS ABOUT JEAVONS SYNDROME

- JS is formally known as epilepsy with eyelid myoclonia.
- The average age at onset is six to eight years.
- JS is thought to account for 0.56% to 2.7% of all epilepsies.
- JS occurs approximately twice as often in females as in males.
- Nearly 50% of people with JS have a family member with a history of epilepsy.

² Smith KM et al. Jeavons Syndrome: Clinical Features and Response to Treatment. *Pediatr Neurol*. 2018 Sep;86:46–51. doi: 10.1016/j.pediatrneurol.2018.06.001. Epub 2018 Jul 10. PMID: 30082241.

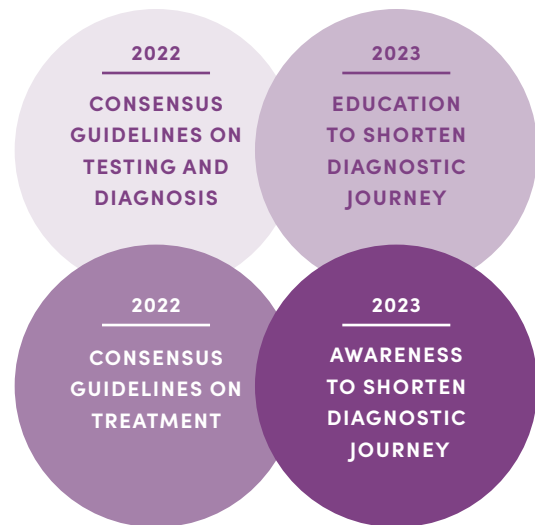
³ Ibid.

⁴ Zawar I, Knight EP. Epilepsy With Eyelid Myoclonia (Jeavons Syndrome). *Pediatr Neurol*. 2021 Aug;121:75–80. doi: 10.1016/j.pediatrneurol.2020.11.018. Epub 2020 Dec 1. PMID: 34167046.

JEAVONS SYNDROME PROJECT VISION

We are seeking to shorten the diagnostic and treatment journey and develop better diagnostic tools that may lead to an increase in JS clinical trials and potential therapies.

The project is comprised of a steering committee of patients with JS and family members as well as physicians with expertise in JS from around the world. By the end of 2023, specific goals include defining diagnostic criteria indicative of JS and publishing an educational video about the disease to YouTube to increase general awareness in an effort to shorten the diagnostic and treatment journey. This project is consistent with our work on other rare epilepsies and can serve as a template for collaboration across lesser-known epilepsy syndromes to advance understanding and refine diagnoses.



A NEW TOOL IN OUR FIGHT AGAINST SUDEP

CURE Epilepsy has been a pioneer in SUDEP research since 2002, when we awarded our first grant to investigate this devastating outcome. In the Spring 2022 issue of *ReSearching to CURE Epilepsy*, we highlighted stories of families who lost a loved one to SUDEP and how our two decades of research have led to breakthroughs in prevention.

Now, we're continuing our pioneering efforts in SUDEP research with a new project: the creation of tools for the standardization of preclinical SUDEP research. The **SUDEP Data Standardization Project** will standardize data collection and reporting around animal models of SUDEP in a clinically meaningful way, creating a common language for information such as the animal's respiratory or cardiac function. Made possible by the BAND Foundation, this project will advance the rigor and transparency of research, increase the potential of combining smaller datasets into larger ones for analysis, and ultimately speed the translation of preclinical research to a clinical setting, accelerating the pace of discovery.



3,000 +

CHILDREN AND ADULTS WITH EPILEPSY DIE DUE TO SUDEP ANNUALLY.⁵ CURE EPILEPSY WON'T STOP RESEARCHING UNTIL THAT NUMBER IS ZERO.

⁵ Thurman DJ, Hesdorffer DC, French JA. Sudden unexpected death in epilepsy: assessing the public health burden. *Epilepsia*. 2014;55(10):1479–1485. doi:10.1111/epi.12666.

THE WIDE-RANGING IMPACT OF POST-TRAUMATIC EPILEPSY

A motorcycle accident survivor. A former NFL player. An Iraq War veteran. They may not appear to have much in common, but they are all at risk of developing seizures as a result of traumatic brain injury (TBI).



Left: Dr. Ramon Diaz-Arrastia. Right: Friend of CURE Epilepsy, former U.S. Army Captain Patrick Horan, who lives with PTE.

Dr. Ramon Diaz-Arrastia, Professor of Neurology at the University of Pennsylvania, explains what we know about post-traumatic epilepsy (PTE) and TBI:

When someone experiences a mild, moderate, or severe TBI, some of their neurons are damaged and other neurons are lost. The remaining neurons attempt to rewire to repair the circuit – but that rewiring is sometimes imperfect. That's how an epileptic circuit arises.

When we talk to patients who have developed epilepsy after a brain injury, we often realize that they had subtle behavioral problems or subtle memory problems that preceded the development of clinically apparent seizures. In retrospect, those were probably very small focal seizures. We need research to identify changes that take place in the brain before PTE develops so that we can begin working to prevent it in people with TBI.

OVER **40%**
OF COMBAT TROOPS WHO SUFFER
SEVERE TBI WILL SUBSEQUENTLY
DEVELOP PTE.⁶



Your donations are funding research into TBI and PTE.
Help us continue the fight – make a donation at
[CUREepilepsy.org/2022fall](https://www.cureepilepsy.org/2022fall).

⁶ <https://www.cureepilepsy.org/seizing-life/post-traumatic-epilepsy-a-lasting-impact-on-our-veterans/>

THE PTE INITIATIVE: STOPPING EPILEPSY BEFORE IT STARTS

CURE Epilepsy is committed to preventing people who experience TBI from developing PTE and finding a cure for people who have already acquired PTE.

In 2015, CURE Epilepsy received a \$10 million grant from the U.S. Department of Defense to establish our PTE Initiative. This research, which commenced in 2017, takes a team-science approach, assembling multidisciplinary experts into investigative teams whose work is overseen by an external advisory council. So far, the Initiative has improved the way we are studying PTE in a laboratory setting and is shedding light on PTE in people who are impacted by TBI. It is also allowing us to identify biomarkers that could reveal who will develop PTE after TBI.

THIS PROGRAM IS LAYING THE GROUNDWORK FOR THE CREATION OF NOVEL THERAPIES TO PREVENT THE DEVELOPMENT OF PTE.

WORLDWIDE INVESTIGATIONS INTO TREATMENT AND PREVENTION

Teams of researchers based in the U.S. and Europe have contributed findings to the Initiative. Several teams have used animal models to investigate potential biomarkers of PTE following TBI. The team at Massachusetts General Hospital (see page 9 for details) tracked the development of PTE in a large animal model with a system that can be used to better understand changes that contribute to PTE. Another team is following human subjects for up to two years following a TBI in search of biomarkers, such as changes in protein levels that can be measured in the blood, that may predict the development of PTE.

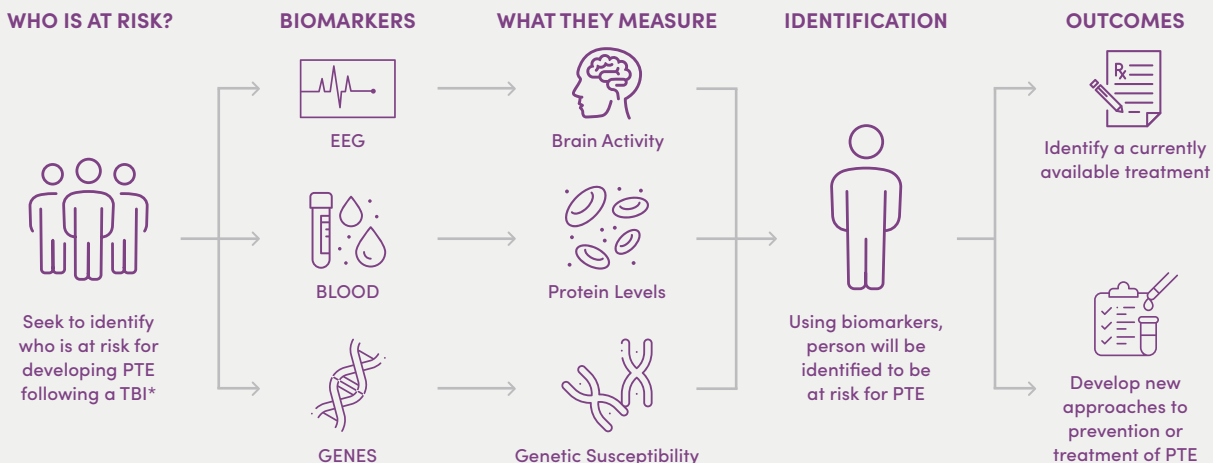
WANT TO LEARN MORE?

Discover each current project on the PTE Initiative webpage:
CUREepilepsy.org/post-traumatic-epilepsy/.



A **biomarker** is a measurable characteristic of the human body, such as heart rate or blood pressure. **Biomarkers are critical** to finding a cure because they help predict who is at risk for developing epilepsy and by measuring the effects of investigational drugs during clinical trials.

THE PROMISING ROLE OF BIOMARKERS IN PTE RESEARCH



* The use of biomarkers in epilepsy research applies to other types of epilepsy as well. Additional outcomes may exist for other epilepsy types.



CURE EPILEPSY IS RESEARCHING FOR...

Jack

TROUT VALLEY, ILLINOIS

At 25 years old, Jack Somers had already been to seven foreign countries, serving military deployments as far away as Afghanistan. Yet while participating in a turkey trot in his hometown, he suddenly had no idea where he was or why he was running. This was Jack's first absence seizure, with several more to follow in the next few months.

In 2012, Jack began experiencing tonic-clonic seizures that knocked him to the ground – forcing him to medically retire from his beloved career as an infantry officer in the U.S. Marine Corps. He struggled to embrace his new identity, passing through various careers before opening up about his struggles.

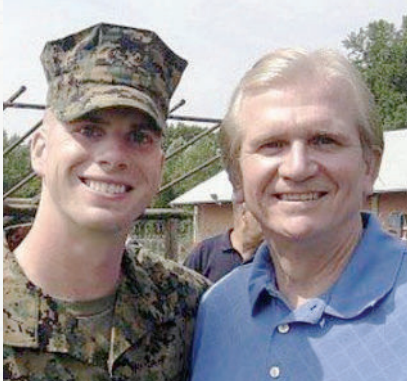
Jack's doctors weren't sure what caused his epilepsy, but they put him on medication to control his seizures. As his symptoms continued, however, doctors began increasing his medications. He knew something wasn't right when he began having trouble focusing and feeling lethargic every day.

After completing a seizure study in the hospital, Jack was taken off some of his medications and has been on a normal dose of his remaining prescriptions for several years. He has had a few minor absence seizures since but is doing better and feels more in control. Jack is grateful for the work CURE Epilepsy is doing to find a cure for PTE.

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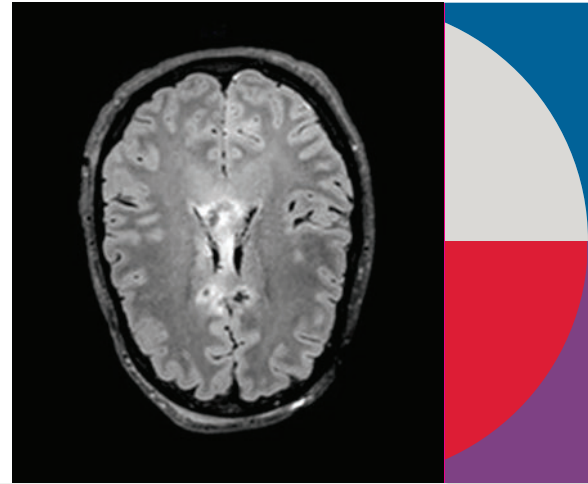
My generation of warriors has so many risk factors for developing seizures – from exposure to burn pit fumes to mild TBI from blasts. I'm so glad CURE Epilepsy is relentlessly pursuing a cure for seizures triggered by TBI.”

JACK SOMERS, EPILEPSY PATIENT AND ADVOCATE



BREAKTHROUGHS IN LARGE ANIMAL MODELS OF PTE

ROBUST, LONG-TERM VIDEO EEG MONITORING IN POST-TRAUMATIC EPILEPSY



Left to right: Keiran Normoyle, MD; Kyle Lillis, PhD; Beth Bartell, PhD; and Kevin Staley, MD

MASSACHUSETTS GENERAL HOSPITAL TEAM

Researchers have found animal models to be helpful when investigating the underlying mechanisms of TBI and PTE. Most PTE animal models have used rodents; however, the rodent brain is very different from the human brain, making translation of findings from the rodent to the human more difficult.

To more closely approximate how PTE develops in humans, a research team at Massachusetts General Hospital studied a large animal model, whose brain is similar in size and structure to ours. Also like humans, this large animal model has a long latent period, or the time between the initial TBI and onset of PTE.

The research team developed a subcutaneous (under the skin) EEG implant with synchronized video to record freely moving large animals for up to 13 months following TBI. While this invention is not applicable for use in humans, it shows promise in helping us understand more about changes in brain activity that occur during the period between brain injury and PTE onset. Some of the advantages of the technology are that it is commercially available and simple to install, with a low failure rate after implementation.

MEET OUR 2022 *CATALYST* AWARD WINNERS

The *Catalyst* Award is a two-year, \$250,000 grant to fund research supporting the nimble development of new transformative therapies for epilepsy, bringing discoveries one step closer to a cure.



JOHN GLEDHILL, PHD

COGNIZANCE BIOMARKERS, LLC

Confirmation of Plasma Biomarkers That Predict Seizure Control in People With Newly Diagnosed Focal Epilepsy

People newly diagnosed with focal epilepsy may face months to years of uncertainty as they try to control their seizures with different medications or other options, such as surgery. This project aims to boost quality of life for newly diagnosed patients through the development of a blood test that will predict whether typical drug treatments will work to control their seizures. This effort will build upon preliminary research showing that people with treatment-resistant epilepsy have differences in inflammation-associated proteins in the blood compared with patients who do respond to treatment.



**JAMES PAULY, PHD, MATTHEW GENTRY, PHD,
AND GREG GERHARDT, PHD**

UNIVERSITY OF KENTUCKY

Safety and Biodistribution of a Novel Enzyme-Antibody Fusion in a Canine Model

Left to right: Pauly, Gentry, Gerhardt

Lafora disease (LD) is a devastating and fatal childhood epilepsy for which there is no cure. LD is characterized by aberrant sugar-like aggregates that form in brain cells. Mouse models of LD have shown that removing these sugar aggregates may be a potential therapy. In collaboration with Enable Therapeutics, Drs. Pauly, Gerhardt, and Gentry developed a drug called VAL-1221 that can penetrate cells and degrade the aberrant sugar aggregates. This project will test the safety and brain distribution of this drug in a canine model, which will help move the therapy closer to clinical application.

THIS PROJECT IS CURE EPILEPSY'S SECOND GRANT AWARDED FOR LD RESEARCH – AND ANOTHER EXAMPLE OF OUR ONGOING SUPPORT FOR INVESTIGATION INTO THE RARE EPILEPSIES.

LAFORA DISEASE

IS A SEVERE, AUTOSOMAL RECESSIVE, PROGRESSIVE MYOCLONUS EPILEPSY. THE DISEASE USUALLY MANIFESTS IN PREVIOUSLY HEALTHY ADOLESCENTS, AND DEATH COMMONLY OCCURS WITHIN 10 YEARS OF SYMPTOM ONSET.⁷

⁷ Nitschke F et al. Lafora disease – from pathogenesis to treatment strategies. Nat Rev Neurol. 2018 Oct;14(10):606–617. doi: 10.1038/s41582-018-0057-0. PMID: 30143794; PMCID: PMC6317072.

BACK IN PERSON: RESEARCH SEMINAR SERIES



Left: Dr. Peter Crino speaks at a seminar hosted by Dr. Howard Goodkin. Right: Dr. Kevin Wang hosts Dr. Elisa Zanier with a team of his young investigators.

CURE Epilepsy is passionate about kickstarting the careers of young researchers and fostering their innovative ideas. Our **Frontiers in Epilepsy Research Seminar Series**, generously supported by the Nussenbaum-Vogelstein family, exposes young investigators to leading epilepsy findings as presented by distinguished scientists.

This summer, the series returned in person for the first time since the pandemic.



JUNE 24, 2022
London Health Sciences Centre,
London, Ontario

SPEAKER:

BIRGIT FRAUSCHER, MD, PHD
*Montreal Neurological Institute and Hospital,
McGill University*

HOST:

ANA SULLER MARTI, MD
Western University



JULY 1, 2022
University of Florida, Gainesville

SPEAKER:

ELISA ZANIER, MD
*Istituto di Ricerche Farmacologiche Mario Negri
IRCCS, Italy*

HOST:

KEVIN WANG, PHD
University of Florida



JUNE 29, 2022
University of Salamanca, Spain

SPEAKER:

NORBERTO GARCIA CAIRASCO, PHD
University of São Paulo

HOST:

MARIA DOLORES LOPEZ GARCIA, PHD
University of Salamanca



JULY 15, 2022
The University of Virginia, Charlottesville

SPEAKER:

PETER CRINO, MD, PHD
University of Maryland

HOST:

HOWARD GOODKIN, MD, PHD
University of Virginia

The Isaiah Stone Foundation

THE ISAIAH STONE FOUNDATION, FOUNDED BY RENZI AND LEE ANNE STONE IN 2011, WAS CREATED IN HONOR OF THEIR SON, ISAIAH. HE PASSED AWAY FROM SUDEP JUST BEFORE HIS FIRST BIRTHDAY AND WAS POSTHUMOUSLY DIAGNOSED WITH DRAVET SYNDROME.

Renzi and Lee Anne Stone work tirelessly to fund research in the rare pediatric epilepsies and SUDEP. We sat down with Renzi to learn more about how he and Lee Anne discovered CURE Epilepsy and why they believe so strongly in our mission.

Your son Isaiah's bright future was tragically cut short due to SUDEP, a common cause of death for people with Dravet syndrome. What would you like others to know about his life and legacy?

Isaiah joined our family on June 15, 2009. He was the second child born to me and my wife, Lee Anne, and the new little brother to our first child, Jackson. With his arrival, our family of four was complete. We spent 11 amazing months with Isaiah. He was a wonderful little boy. Even when struggling with discomfort, he was always a joy. After he passed, Lee Anne and I asked ourselves what his short time on earth might contribute. We grieved by deciding to be active. The idea of setting up a foundation came pretty quickly.

Tell us about Isaiah's first seizure. What was the prognosis?

It was a typical evening in late October when Isaiah suddenly began shaking for 10 straight minutes.



HELP US CURE RARE GENETIC EPILEPSIES IN CHILDREN

This fall, donate to support research that will help babies and kids enjoy a childhood free of seizures. To make a gift in memory or honor of a loved one, please scan the QR code to the left.



Renzi and Lee Anne Stone, with sons Jackson and Isaiah

We rushed him to the emergency room and learned he had experienced a tonic-clonic seizure. Lee Anne and I knew nothing about epilepsy, so it was terrifying. Isaiah tried and failed several medications, but the overwhelming consensus from multiple doctors was that we had to wait and see. Most were optimistic that Isaiah would outgrow his seizures. We were shocked when Isaiah passed from SUDEP on May 17, 2010 – his 333rd day of life. We should have known it wasn't good – he often had seizures lasting 20 minutes. There just wasn't a lot known about epilepsy at the time. Losing Isaiah left a gigantic hole, but his life motivates us every day to pursue a cure for other babies and children.

You didn't find out that Isaiah had a rare genetic epilepsy until after his death. How did that turn out to be a blessing for your family?

Lee Anne and I were interested in having another child but wanted to make sure they wouldn't be at risk for genetic epilepsy. After a posthumous genetic test of Isaiah's blood came back negative, we proceeded to welcome our son Walker. Then around 6 p.m. on New Year's Eve of 2015 – five-and-a-half years after losing Isaiah – I received an unexpected call from his neurologist. The lab had run Isaiah's blood again, and a new test revealed that he had a mutation in the SCN1A gene, the primary cause of Dravet syndrome, which is a severe and catastrophic form of pediatric epilepsy. Our first thought was, "What about Walker?" We quickly

learned that Dravet syndrome appears by age two, and Walker was already three. We're so glad we discovered Isaiah's diagnosis after Walker was born, or we may not have expanded our family.

Tell us about the remarkable way you first heard of CURE Epilepsy.

A few hours before Isaiah had his first seizure, 60 Minutes aired its new episode featuring CURE Epilepsy Founder Susan Axelrod and her husband, David. I happened to catch the program. Months after Isaiah was diagnosed with epilepsy, I remembered I saw the episode the same day he went to the ER. I got together with Susan and was very impressed with the mission of her organization. A few months later, I joined the CURE Epilepsy board.

CURE Epilepsy is the primary beneficiary of the Isaiah Stone Foundation. Tell us about your foundation's mission and the opportunities you've had to advance it through CURE Epilepsy.

The Isaiah Stone Foundation funds primary research that helps young scientists advance knowledge around epilepsy, with a special focus on children. We also offer one-time grants to help families living below the federal poverty level.

In 2016, CURE Epilepsy provided a funding opportunity that helped heal our family's greatest source of pain: SUDEP. Dr. Poduri and her team from Harvard Medical School discovered a potential link between the SCN1A gene and sudden death in pediatrics, laying the foundation for preventing further tragedies. It makes us want to invest more. We believe that CURE Epilepsy will find a cure, and we look forward to shutting down the Isaiah Stone Foundation because it's no longer needed.

How has your family generated donations for a cure? What has been your most meaningful fundraising experience?

Since 2010, we've raised \$1 million through individual donations and events. CURE Epilepsy is the biggest recipient of our funds. My sons have actively participated – Jackson raised \$5,000 by hosting a basketball clinic for his friends. Our most meaningful fundraiser was working with Edmond Memorial High School in Edmond, Oklahoma, to be its annual fundraising cause. This fundraising program is a big deal. We applied two years in a row and were selected by the student council in 2017. It was amazing to see the students rally around our cause without ever knowing Isaiah. I have immense appreciation for those kids. They raised almost \$400,000 for our foundation.

In what ways does Isaiah's life continue to provide a guiding light to your family, particularly your children?

Isaiah's name comes up every day, as he is constantly in our prayers. He has an enduring positive influence on Jackson and Walker. Losing their brother has added a bit of compassion that our sons otherwise wouldn't have had, and it shows in their interactions with others. Our sons understand the obligation to pay Isaiah's life forward.



Isaiah Stone at age 9 months

“CURE Epilepsy provided a funding opportunity that helped heal our family's greatest source of pain. It makes a donor want to give more when they see that their investment did something collectively and personally.”

RENZI STONE



Renzi and Lee Anne Stone, with sons Jackson and Walker

ACCESS THE LATEST EPILEPSY RESEARCH

WATCH OUR WEBINARS



IDENTIFICATION AND TREATMENT OF AUTOIMMUNE EPILEPSY AND TREATMENT CONSIDERATIONS

Stephen VanHaerents, MD
Northwestern University

When the immune system mistakenly attacks healthy brain cells, it can lead to brain inflammation (encephalopathy), resulting in memory loss, cognition problems, impaired speech, and seizures. These two webinars explain how doctors recognize autoimmune encephalitis, what makes them suspect autoimmune-related seizures and epilepsy, and which techniques and considerations they apply around treatment.



SPEAKING ABOUT SUDEP: ARMING THE RARE EPILEPSY COMMUNITY WITH THE LATEST RESEARCH

Samden Lhatoo, MD, FRCP
The University of Texas Health Science Center
at Houston

Parents of children with rare epilepsy need cutting-edge prevention techniques and empathetic support to prevent the tragedy of SUDEP. This webinar provides both a physician and familial perspective on the risk of SUDEP, with research insights presented by Dr. Lhatoo and tips for talking to doctors from Abigail Tanner, mother to Lincoln, who has a rare form of epilepsy (see his patient story on page 3).



MENTAL HEALTH AND EPILEPSY: IMPROVING QUALITY OF LIFE

Heidi Munger Clary, MD, MPH
Wake Forest School of Medicine

For people with epilepsy who experience anxiety, depression, or mood disorders, treating these conditions can lead to greater quality of life than reducing seizures alone. This webinar discusses how anxiety and depression impact people with epilepsy in different ways, what neurologists can do to alleviate the symptoms, and ways that treating these symptoms can influence treatment of the seizures themselves.



WANT MORE WEBINARS?

Visit CUREepilepsy.org/webinars
to view recordings or register
to attend upcoming events.

“ Dr. Clary’s talk was filled with facts and resources I wasn’t aware of. My son battles with epilepsy and depression. I was relieved to finally have a forum where I could get answers to MY questions.”

CARMEN S.
CURE EPILEPSY WEBINAR ATTENDEE

CHECK OUT OUR INAUGURAL TREATMENT TALK

New in 2022, this social media broadcast highlights conditions related to epilepsy and options available for treatment of related seizures and their side effects.



LENNOX-GASTAUT SYNDROME (LGS) AND FENFLURAMINE

Michael Chez, MD
Sutter Institute for Medical Research

JUNE 25, 2022

LGS is a severe form of childhood epilepsy that arises during infancy or early childhood. In this talk, pediatric neurologist Dr. Michael Chez sits down with Heather Bushey, the mother of a boy with LGS, to discuss how LGS is diagnosed, common seizure types and comorbidities associated with LGS, how fenfluramine works to reduce some of these seizure types, and any risks and benefits found in recent clinical trials featuring fenfluramine.



CURE EPILEPSY IS
RESEARCHING FOR...

Nathan

GARDNERVILLE, NEVADA

Four-year-old Nathan Bushey and his family were staying with relatives following the birth of his sister when Nathan's cousin made a terrible discovery: Nathan was making disturbing noises in his sleep. This turned out to be Nathan's first documented seizure, and it landed him in the ER later that night.

The hospital neurologist evaluated Nathan's EEG and believed that he would grow out of his seizures. Unfortunately, Nathan had another seizure on the school bus shortly after his initial hospital visit. At age six, he was diagnosed with Lennox-Gastaut syndrome and started on what his mom, Heather, calls "medication roulette."

Now 13, Nathan struggles most to control his tonic-clonic seizures, which often keep him – and his family – up most of the night. In 2020, Dr. Chez prescribed fenfluramine (*Fintepla*®), an FDA-approved medication that has reduced Nathan's tonic-clonic seizures from three to four per week to one every two to three weeks.

“

By reducing his seizures with fenfluramine, Nathan has been able to make cognitive and behavioral strides. I'm so glad CURE Epilepsy is funding research into medications that more effectively target the specific EEGs of individuals with epilepsy.”

HEATHER BUSHEY, NATHAN'S MOM AND EPILEPSY ADVOCATE



LISTEN TO THE SEIZING LIFE® PODCAST

The *Seizing Life*® podcast series is one of our primary ways of educating patients, families, and the public. Join us to learn about the daily challenges and triumphs of living with epilepsy as well as the latest information on key topics, such as SUDEP.



RARE EPILEPSY: COLLABORATING FOR ADVOCACY, RESEARCH, AND COMMUNITY

Yssa DeWoody, PhD
Co-founder and Research Director,
Ring14 USA – a member of the
Rare Epilepsy Network (REN)

After Yssa DeWoody's youngest daughter, Marie, was diagnosed with a rare epilepsy called Ring14 syndrome, she joined four other mothers to found Ring14 USA. This organization provides a supportive community for families impacted by rare epilepsy and helps people with this diagnosis find targeted treatments and therapies. Yssa provides a primer on rare epilepsies, discussing diagnosis, treatment, challenges, and resources, including clinical trials.



A REMARKABLE JOURNEY TO SEIZURE FREEDOM THROUGH THE KETOGENIC DIET

Jim Abrahams
Producer, Writer, and Director

Jim Abrahams was enjoying success from a string of Hollywood hits (*Airplane*, *Top Secret*, *The Naked Gun*) when his infant son, Charlie, was diagnosed with epilepsy. Jim recounts how Charlie's doctors gave him a devastating prognosis and how he and his wife, Nancy, struck out on their own to discover the healing effects of the ketogenic diet, which cured Charlie of his disease.



A POST-TRAUMATIC EPILEPSY JOURNEY: DIAGNOSIS ACCEPTANCE LEADS TO CAREER HELPING OTHERS

Mark DeFee
Mental Health Educator, Coach,
and Speaker

During Mark DeFee's high school football career, he sustained multiple concussions that led to a PTE diagnosis. Mark shares highlights from his 26-year journey living with epilepsy, including the epiphany that led him to adopt a positive outlook and create a productive, meaningful lifestyle. Now a mental health counselor, Mark offers guidance and hope, including advice that can help people accept their epilepsy diagnosis.

100

EPISODES OF
SEIZING LIFE®

Check out host Kelly Cervantes's interview with David Axelrod, husband of CURE Epilepsy Founder Susan Axelrod, which premiered October 5.



WANT TO LEARN MORE?

Check out the full series at
CUREepilepsy.org/seizinglife.

UNDERSTANDING HOW EPILEPSY IS DIAGNOSED

Receiving an epilepsy diagnosis can be difficult for patients and families to process – but what goes into the diagnosis itself? Can epilepsy be diagnosed from one seizure? What kinds of doctors can diagnose epilepsy? What are the different potential diagnoses?

To help patients navigate this experience, CURE Epilepsy has put together **Understanding Epilepsy**, an online resource offering information on:

- The diagnostic process, tests, and tools, such as neurological exams and genetic tests.
- EEG types, how EEGs work, and when one test might be prescribed over another.
- Magnetic resonance imaging (MRI), including how it can identify where seizures start.
- Other diagnostic tests, including neuroimaging scans that construct images of the brain.
- Epilepsy syndromes, including West syndrome and Dravet syndrome.
- The adult vs. pediatric journey: how these patients may have different experiences.

Find the **Understanding Epilepsy** section under the **For Patients** tab at **CUREepilepsy.org**. This section is made possible through the generous support of SK life science.



To access the full epilepsy diagnosis resource, scan the QR code above.



A PARENT'S GUIDE TO YOUR CHILD'S EPILEPSY DIAGNOSIS AND TREATMENT

Douglas Nordli, MD
UChicago Medicine

While every child's epilepsy journey is different, there are procedures, information, and choices that all parents should know about. This episode is the must-have guide to help parents access the best care for their child, including drugs and alternatives. Dr. Nordli, a pediatric neurologist and Co-director at UChicago Medicine's Comprehensive Epilepsy Center, offers advice for seeking a second opinion, finding an epileptologist, and accessing quality care, as well as tips for families and physicians to collaborate on treatment.

“

When we suspect a child has had a seizure, waiting several months to see a pediatric neurologist is not acceptable.

Parents should have their pediatrician call the neurologist directly to explain that this is a new onset. Telemedicine can also be a way to get the ball rolling.”

DR. DOUGLAS NORDLI

BACK IN PERSON: ANNUAL CHICAGO BENEFIT

JUNE 1, 2022

For the 24th consecutive year – and the first time in person since the pandemic – our Chicago benefit brought together a dedicated community of people committed to ending epilepsy. Over 400 attendees gathered at the beautiful Navy Pier and spent the evening sharing inspirational stories, learning about CURE Epilepsy's latest research, and enjoying great food and music courtesy of Barenaked Ladies.

OVER \$1.9 MILLION RAISED TO FIND
A CURE FOR EPILEPSY

MARK YOUR CALENDAR FOR OUR
25TH ANNIVERSARY CHICAGO BENEFIT
ON MAY 6, 2023

Clockwise from top left to right: CURE Epilepsy Board Member Tina Sacks and Nora Hennessy; CURE Epilepsy Board Chair, Kelly Cervantes; CURE Epilepsy Board Member Blake Cunneen with mom, Pat; CURE Epilepsy Board Member Lisa Cotton and daughter, Caroline; Renzi Stone, son Walker, and Chelsea Watkins, with CURE Epilepsy CSO Dr. Laura Lubbers and CEO Beth Dean; CURE Epilepsy Board Member Hannah Whitten and family; Performance by Barenaked Ladies; Supporters of Fund-a-Cure



SPREADING EPILEPSY AWARENESS AT CISCO

JUNE 21, 2022

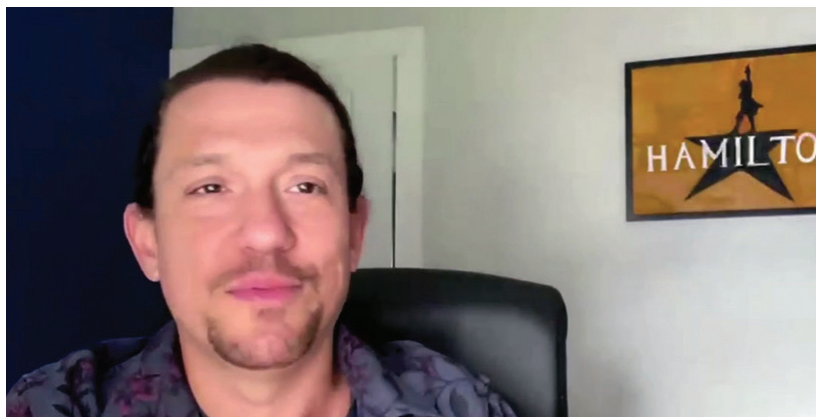
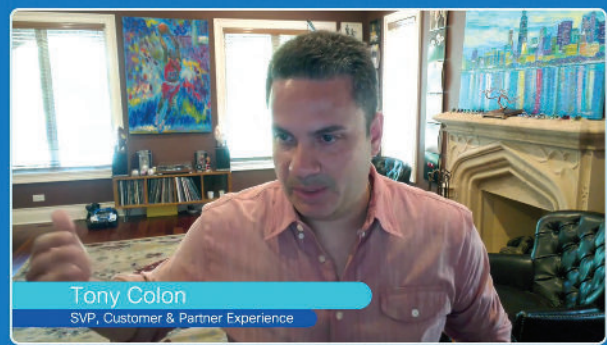
When Cisco senior vice president Tony Colon was looking for a cause to rally his team around, he immediately thought of epilepsy research. Tony's daughter, Ellie, has an epilepsy diagnosis, and he is a dedicated supporter of CURE Epilepsy. CURE Epilepsy worked closely with Tony to deliver a custom virtual event that informed and inspired Cisco employees while generating donations for new investigations. Cisco employees heard from parents who lost their child to epilepsy, a young professional who battles epilepsy daily, and a scientist who taught them some of the latest epilepsy facts.

OVER 300 CISCO EMPLOYEES ATTENDED FROM ACROSS THE U.S.

CISCO EMPLOYEES LEARNED THAT...

- 3.4 million Americans live with epilepsy, and an estimated one in 26 will develop epilepsy in their lifetime.
- Epilepsy affects more people than multiple sclerosis, cerebral palsy, Parkinson's disease, and ALS combined.
- To be diagnosed with epilepsy, a person must have had two or more seizures more than 24 hours apart that were not provoked by a specific event, such as stroke or an infection.

Clockwise from top left to right: Jackson Cervantes performs a song in memory of his sister Adelaide; Britton Greene shares his daily struggles with epilepsy; Miguel Cervantes performs hits from Hamilton



BRING CURE EPILEPSY TO YOUR WORKPLACE

Enhance your company's commitment to making a difference and help raise money for epilepsy research with a custom online event delivered by CURE Epilepsy. For more information, contact Brandon Laughlin, CURE Epilepsy Senior Manager of Partnerships & Programs, at brandon.laughlin@CUREepilepsy.org.



RACING OUR WAY TO A CURE: CELEBRATING CURE EPILEPSY CHAMPIONS

UMES 5K STRIDES FOR EPILEPSY

9TH ANNUAL EVENT HELD APRIL 23, 2022 |
NEARLY \$2,000 RAISED

Hosted by the University of Maryland Eastern Shore (UMES) School of Pharmacy, this race gives college students an active way to raise critical funds for epilepsy research.

ELLA'S RACE

7TH ANNUAL EVENT HELD AUGUST 21, 2022 |
NEARLY \$82,000 RAISED

Ella Cunneen has epileptic spasms – a rare form of epilepsy that doctors don't know much about. This year, Ella's family partnered with CURE Epilepsy to put the proceeds from her annual event toward a special \$500,000 grant to fund research into epileptic spasms.

MANSFIELD WALK TO CURE EPILEPSY

INAUGURAL EVENT HELD MAY 14, 2022 |
NEARLY \$7,000 RAISED

Twenty-two-year-old Anthony Maffie was working toward becoming a nurse when his life was cut short from SUDEP on January 4, 2019. This year, his family hosted a race in their hometown to support research into SUDEP prevention.

REAGAN'S RUN

INAUGURAL EVENT HELD SEPTEMBER 11, 2022 |
NEARLY \$30,000 RAISED

Reagan Monast was diagnosed with epilepsy and cerebral palsy just before age two. Nine years later, Reagan is challenging people in her community to raise money for epilepsy research by running, jogging, or walking.



GIVINGTUESDAY

NOVEMBER 29

**HELP US RAISE \$100,000
ON GIVING TUESDAY!**

Use our new fundraising platform to kick off your campaign and rally your friends and family to help us find a cure.



**CHECK OUT THE NEW
CURE EPILEPSY TIKTOK CHANNEL**

Follow us for fun, informative videos offering bite-sized facts and insights into epilepsy research.

@CUREepilepsy



**EXPLORE EPILEPSY
CLINICAL TRIAL OPPORTUNITIES**

If you're living with epilepsy, consider volunteering to test new medications that could lead to breakthroughs in treatment.

**CUREepilepsy.org/for-patients/
understanding/clinical-trials**



**GET TICKETS TO THE JOEY'S SONG
BENEFIT CONCERT ON JANUARY 7**

Promote epilepsy research and awareness with a night of star-studded entertainment. Attend in person at the Sylvee Theater in Madison, Wisconsin, or bid on one-of-a-kind music packages in the online auction.





[CUREEPILEPSY.ORG/2022FALL](https://cureepilepsy.org/2022fall)

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