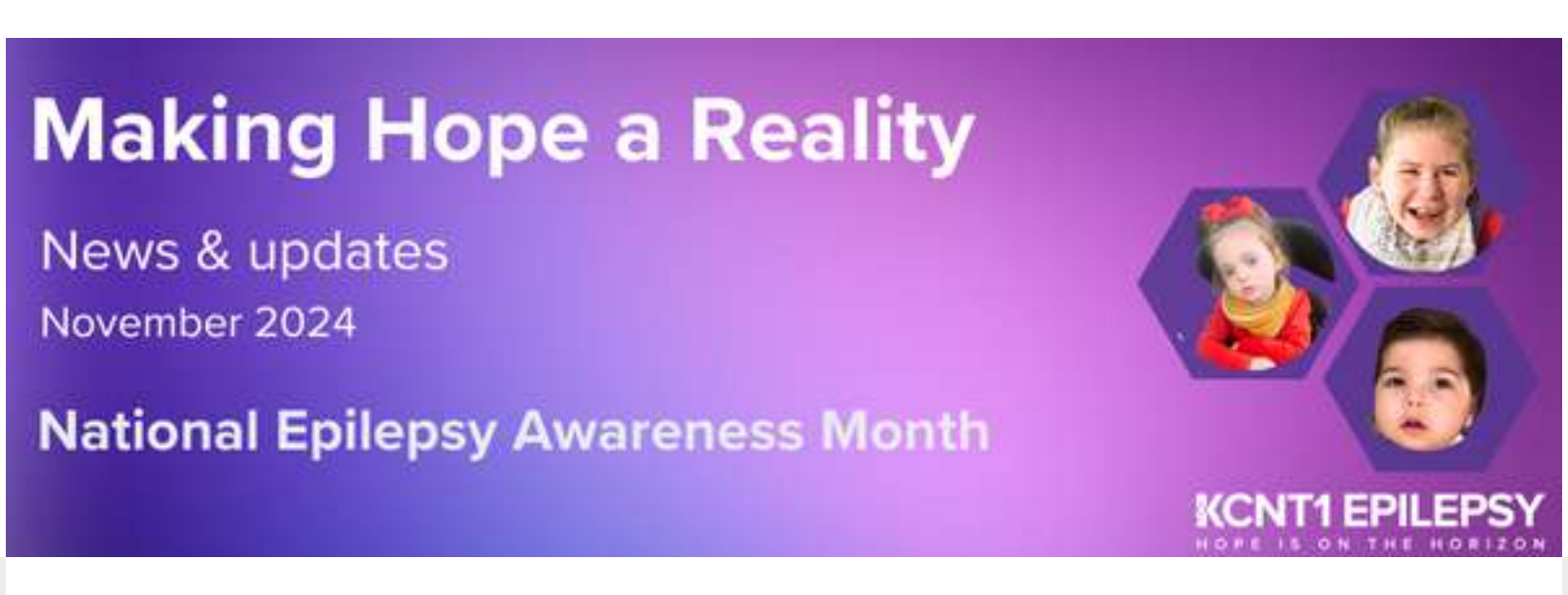

Dear Friends and Families,

As November unfolds, we are thrilled to kick off Epilepsy Awareness Month with a full calendar of meaningful ways for you to engage, support, and connect. This month, we are focusing on raising awareness for KCNT1-related epilepsy, while also offering many opportunities for families to participate in research, fundraise, share experiences, and help us drive progress.

Whether you're a parent, family member, or supporter, there's a place for you in this journey. We encourage you to get involved in whatever way feels right for you—whether that's participating in research, sharing our Giving Tuesday campaign, volunteering or attending our upcoming events.

Thank you for standing with us as we work together to create a future full of hope, progress, and possibility for everyone affected by KCNT1-related epilepsy. We can't wait to see the impact we'll make this month—together.

Warmly,

Sarah Drislane
Executive Director

IMPORTANT: We Need 20 More Families by November 30th!

We are 20 families short of meeting our goal of 100. If you are living in the U.S., please enroll in Citizen Health today and build our database of medical records! Enroll by Nov 30 and be entered into a drawing. If you need help, contact Amanda Abuhl!

Medical records and genetic data can provide valuable insights into how KCNT1-related conditions manifest and progress, showing the "natural history" of the disease. Analyzing patterns can help us identify effective treatments and improve quality of life for our kids.

Enroll in Citizen Health, here.

Get instructions how to **refresh your account** here.

What We Are Learning from Citizen and Registry Data

By Brad, Bryan, PhD, MBA, Director of Operations

Every day, our community learns and grows from the experiences of individuals affected by KCNT1 variants, and the challenges our families face inspire us to keep moving forward. The KCNT1 Epilepsy Foundation wants to thank all the families who have taken the time to fill out the family contact form on our website, shared their medical records through Citizen Health, and completed the various surveys we've conducted. Your contributions have a real and lasting impact on our understanding of KCNT1-related conditions and the journeys within our community, and they're crucial for helping us find new therapies for those affected.

We'd like to share some insights we've gained from your feedback. Thanks to your efforts, we've identified KCNT1 families across 42 U.S. states and in several countries, including sizable groups in Brazil, China, the UK, France, and Germany. It's inspiring to note that our oldest KCNT1 member has reached 50 years old, which reminds us of the unique and meaningful paths each journey with KCNT1 can take.

We've also learned that symptoms of KCNT1-related conditions usually appear within the first three months of life, with a median diagnosis age of just 103 days. Through your input, we've identified five genetic variants that account for the majority of KCNT1 cases, along with many other variants that contribute to the rest. So far, all these KCNT1 variants lead to an overactive version of the protein, which results in neurological dysfunction like seizures, as well as issues affecting the respiratory system, gastrointestinal tract, muscles, and other body systems. This understanding helps us focus on the treatments that caregivers find most effective for controlling seizures, such as the Keto Diet and medications like Clonazepam, Clobazam, Phenobarbital, and Levetiracetam.

Our registry data (which is comprised mainly of people with EIMFS) revealed some important trends among individuals with KCNT1 variants. For instance, 84% of these individuals use feeding tubes to manage their nutritional needs, and nearly 40% have undergone nerve or muscle block procedures. Seizures and respiratory complications are the leading causes of hospital visits, with almost everyone with KCNT1 variants admitted for seizures and 86% admitted for respiratory issues.

Every new finding from our community, whether through our contact list, the Citizen Health Database, or our Foundation's surveys, brings us closer to better care and support for KCNT1 everywhere. We encourage everyone who has previously shared their information in these surveys to continue doing so. If you haven't contributed yet, please consider adding your information—it benefits not just your loved ones, but also everyone around the world affected by KCNT1.

News & Events

Have a KCNT1 warrior who is age 16 or older?

Tuesday, November 12, 8pm Eastern

Join us for a special family meet-up! This is a space for families with teens and young adults transitioning from pediatric to adult care. Connect with other KCNT1 families, share experiences, and discuss the unique challenges of this phase.

To help us get to know whose coming! Please take a moment to RSVP with this survey and we will send you a link

Sibling Circle

Sunday, December 1, 2:00 EST

Sign up for the last session of the year. Join moderator, Jessica Kruger, and meet other siblings 5-17 for fun and friendship.

Register Today

Epilepsy Awareness Day at Disneyland, Anaheim, California

November 18 & 19

Thanks to Laura and Rebekah for volunteering to come help at our booth! We look forward to meeting more KCNT1 Families in person at the largest gathering of epilepsy physicians and patients on earth! Learn more!

Keto Diet Discussion

December 12, 2:00 EST

Sign up for to hear from parent, Karen Philips and clinician from Cincinnati Children's Hospital all about the keto diet.

Register Today

Game Night Anyone?

We're excited to organize an **online game night** to bring families together for fun and to connect! Whether you're a gaming pro or a complete beginner, this is a chance for parents and kids to enjoy games like Minecraft.

We need your input to get started! Please take a moment to fill out our quick survey to express your interest in organizing or participating, and let us know what games you'd like to play.

We look forward to gaming with you! Let us know if you are interested here!

SHE/ADNFLE Family Research Study Coming Soon

In our recent family meeting of parents of those with ADSHE/ANDFLE/SHE or similar phenotypes, we all agreed that a study was needed to help understand the non-seizure symptoms of the children and their carrier parents. Thanks to those who attended the meeting and are sharing research for KCNT1!

Science and Research News

KCNT1 Research Roundtable

By Ali Rosenberg, PhD, Scientific Outreach Officer

This week we hosted our second "research roundtable" of 2024, where we bring together scientists and clinicians from around the world to update us on their KCNT1 research. This is one of the ways that we build community-- by introducing researchers and their work to each other, and listening to how we can link them to each other to share resources and ideas and help their research get to the next stage. At this meeting we heard from 8 scientists from the US, France, Italy, and China.

Two talks were relevant to aspects of future clinicial trials for KCNT1. One scientist is working on a massive data set that uses biospecimens from our own KCNT1 children (thank you to the families who contributed samples!), in combination with biosamples from individuals with 28 other neurodevelopmental and seizure disorders, along with seizure-free individuals. This project is looking to find something called a biomarker, which is an objective measure that can show, for example, disease progression or improvement with treatment. This is something the FDA would like to see in most clinical trials. Another project that is important for clinicial trials is that out of Dr. McKee's lab at CHOP, who won our Seed Grant. One of her students gave an overview on their project to develop an understanding of the natural history of KCNT1 by analyzing Citizen electronic medical records. This study can help with the design of trials based on how KCNT1 epilepsy progresses and help clinicians best direct treatments.

Several researchers spoke about projects with goals to develop new therapeutics for KCNT1. Dr. Rima Nabbout, who is on our Scientific Advisory Board, spoke about a new initiative in France supported both by European epilepsy groups, private companies, various healthcare and research institutions, and national groups. This initiative's goals are to develop precision medicines for KCNT1. Two researchers described their work developing brand new small molecules to block KCNT1, one of whom also showed the effect of carvedilol on improving KCNT1 function. If your family is interested in trying carvedilol as a part of our observational study, please get in touch with Sarah, who will be happy to help you get started. One researcher spoke about a new KCNT1 mouse model that appears to have seizures much like those seen in people, and they are working to understand the characteristics of this mouse, including looking at seizures, sleep, and sleep apnea. This new mouse model, when fully characterized, will be very valuable for testing new potential therapeutics. Our own Scientific Advisory Board member, Dr. Weston, spoke about their success testing a small molecule developed by the biotech company Actio in a KCNT1 mouse, and how it drastically reduced seizures, which is extremely exciting news!

Over 80 individuals from around the world signed in to hear these talks and offer their questions, ideas, and support to our speakers. From these talks and all the groups we collaborate with, we see a lot of progress in the different areas that bring KCNT1 closer to a working therapeutic, and we are filled with hope by all of their efforts.

Community News

Pathway to Trials: Clinical Trial Readiness Education

If you missed our most recent family meetings about clinical trials, you can view them at the links below. Ali Rosenberg and Brad Bryan provided an overview describing how the KCNT1 community can prepare for upcoming clinical trials and what insights we are gleaning from the data you have shared. They cover key areas such as:

Clinical Trial Readiness: Learn why it's crucial to have detailed medical records, complete genetic testing, and register in patient databases for quick enrollment.

Patient Data : Understand the importance of accurate patient numbers and contact information to support clinical trials and research, encouraging more families to participate in patient registries.

Reach out if you have any questions.

Pathway to Trials Link

What Your Data Tells us Link

Your Voice Matters

Important Poll: Where Would You Travel for a Clinical Trial?

Whether you'd drive, fly, or are still unsure if you'd be interested in a clincial trial—that's totally fine. You could influence where trials could be located by completing our anonymous poll!

Take the anonymous poll!

Fundraising News

Family Fundraiser: Giving Tuesday on Dec 3rd

Fundraise

Do you have friends and co-workers who ask how they can help? As we enter the season of gratitude, you can offer them the opportunity to support us on Giving Tuesday -- and beyond! How about $5 per month? In 2022 we had 45 supporters on Giving Tuesday. In 2023, thanks to YOU, we had 90 supporters! What can we do in 2024?

Last year our winning fundraising teams included Alex Wild, Samantha MacMechan, Laura Aguilar, Lisa West, Michele Martel McKendall, Shifra Wagner, Lorena Avonce, Rotem Miller, Krystle Stanley, Kady Lucetti and Tammy Williams! Can we count on you this year?

Scroll to the +Fundraise button and there you can sign up to be a Fundraising Team Member and begin your own fundraising team for Giving Tuesday, where we *Rally for Research Readiness*! Our goal is to fund essential research assets, such as the collection of blood samples to create specialized cell lines and advanced animal models that help test potential therapies. Before new drugs can be tested on humans, they must first be proven effective on models genetically edited to reflect the same gene variants as our kids. These models include **mice**, **zebrafish**, and yes, even **fruit flies**!

Fruit flies may seem small, but they play a huge role in epilepsy research. Scientists can use them to mimic seizure activity, allowing for quick and affordable testing of new treatments—getting us closer to breakthroughs that could make a real difference.

You can volunteer to donate blood and other samples from your child and yourself, and you can also give your family and friends the opportunity to get involved on this one giving day, and help us fund the tools researchers need.

By supporting our campaign this Giving Tuesday, you'll help provide researchers with the tools they need to find life-changing treatments for our children.

Our top 3 fundraisers will receive a free t-shirt! Or you can purchase a limited edition t-shirt here!

As we reflect on the world today, we recognize that every family faces its own unique challenges, and we're deeply aware of the struggles taking place both near and far. It is our hope that, in some small way, we can provide you with a sense of comfort—knowing that you are part of a community that truly understands you -- and stands by your side. Together, we share the journey of parenthood and the determination to find better treatments for our children.

We are profoundly grateful for the moments of hope, the progress being made, and for each of you—your resilience, your commitment, and your belief in this mission. Thank you for being an essential part of this pathway to brighter possibilities and life-changing breakthroughs.

Together, we will keep moving forward, with gratitude and hope lighting the way.

The KCNT1 Epilepsy Foundation Team

Donate

Our Contact Information
{{Organization Name}}
{{Organization Address}}
{{Organization Phone}}
{{Organization Website}}

KCNT1 EPILEPSY
HOPE IS ON THE HORIZON