

Making Hope a Reality

News & Yearly Recap
December 2024

KCNT1 EPILEPSY
HOPE IS ON THE HORIZON



Dear Families, Grandparents and Supporters,

As we wrap up another remarkable year, we are filled with gratitude for your dedication and support. Together, we've achieved milestones that bring us closer to better treatments and brighter futures for our KCNT1 warriors. In this issue you will find a look back at some of the highlights that you helped make possible!

2025 is going to be a year of renewed hope, with many opportunities for you to drive research and hopefully, participate in clinical trials. Please see an important and heartfelt letter from one of our own parents, Amanda Abuhl.

Wishing you a wonderful Holiday season and a 2025 filled with hope and progress!

Warm regards,

Sarah Drislane, Justin West, Ali Rosenberg, Brad Bryan, Megan Wright, Amanda Abuhl, Lauren McCabe, our Board of Directors and all our volunteers

Number of KCNT1 Families Exceeds 500 Worldwide

The number of families we have identified has nearly doubled this year. Some of these families have newborns that were diagnosed within days of birth and one who was diagnosed before birth, while others are have been members of various Facebook groups but not engaged with the foundation until recently.

Our Family Contact database plays a vital role in accurately tracking the number of diagnosed families. Additionally, we will use your contact information to keep you informed about educational opportunities and clinical trials, with updates expected in 2025.

Thanks to all of you who have welcomed the new families with open arms and been kind and encouraging when sharing information. If any of you would like to sign up to be "parent support specialists" or foundation ambassadors for your country, we could use help in supporting new families. [Reach out](#) if interested.

Citizen Health Enrollment Grows

We're thrilled to see **96 families** enrolled in Citizen Health this year, up from 62 last year! This platform centralizes medical records and connects patients, families, and researchers. By contributing your de-identified data, you're not only aiding groundbreaking research but also setting the stage for clinical trial readiness. Let's aim for **100 families** to strengthen our community's impact. U.S. families can sign up [here](#).

What Parents Are Saying

"Keep raising awareness and being such incredible advocates! You are making a difference in countless kids' lives."



Clinical Trials Location Survey

Not sure if you'd drive, fly, or even consider a clinical trial yet? That's okay! We're here to help you make the best decision for your family when the time comes.

Wondering what clinical trials are, the difference between Phase 1 and 2, or how inclusion/exclusion criteria work? These are personal choices, and we're here to guide you.

[Watch](#) our *Pathway to Trials* series, explore resources, and get ready—several companies are nearing trial stages! Help shape trial locations by completing our anonymous poll.

Take the anonymous [poll!](#)

Donate

Science and Research News

You Are Fueling Important Research

In the past few years our collective efforts have gone from introducing KCNT1 to researchers and life sciences companies to a reality where KCNT1-specific therapeutics are on the cusp of clinical trials. This progress is nothing short of extraordinary considering the size of our rare disease community. But we are not there yet, and won't get there without all our participation.

Through our shared efforts including our hosted Research Roundtables, we've attracted researchers who have spent years studying the KCNT1 ion channel, even before KCNT1-related epilepsy was recognized as a distinct disease. These scientists have now been able to develop treatments that could alter the lives of our children. This year, many of these researchers joined our foundation as members of the Scientific Advisory Board, further strengthening our efforts at driving the science forward.

Your contributions have been critical to this success. Biosamples donated to our biobank, participating in our registry, the Biogen KITE study, involvement in the Conceptual Model study have all created essential knowledge essential for designing treatments. These efforts, alongside the work being done by Dr Jillian McKee's lab (funded through our 2024 Seed grant) will be published in 2025 and pave the way for clinical treatment guidelines, and most importantly, clinical trials.

Parent-driven Research Questions

This year, the amazing support from the parent community has made a huge difference in driving research. Together, we've started tackling important questions -- like why having a virus can stop seizures, how is ADSHE different, what causes breathing problems in our kids, and how can we better treat MAPCAs.

In 2024 we kicked off big efforts, including funding biomarker studies and an early gene editing feasibility studies. These studies are already showing interesting results, and we'll keep building on them as we move into 2025. We've also started looking closely at our cell lines and animal models to make sure researchers have the best tools to get reliable results.

As we move into the new year, this work will remain a top priority. Contributions to our Giving Tuesday campaign will help keep these efforts growing strong. Every gift will support these projects, pushing us closer to answers and new treatments for our kids.

Thank you for being such an important part of this journey. Together we are making strides toward a brighter future for our children.

Donate

Pathway to Trials: Clinical Trial Readiness Education

We are entering the most hopeful and exciting phase yet—the era of clinical trials for KCNT1-specific therapeutics. These trials carry the potential to transform hope into reality, giving our children a renewed chance at health, development, and joy.

To prepare, we launched a **Clinical Trial Readiness Campaign** to educate and empower you about clinical trials, from what to expect to how to participate. We want to ensure we are organized, informed, and ready to take the next step together. Our **Pathway to Trials** webinar series, began with presentations by Brad Bryan and Ali Rosenberg, to educate families on clinical trial participation and the importance of data sharing. If you missed them, catch the recordings on our YouTube channel.

Watch on [YouTube](#)

Community Fundraising

In 2024, families have shown the power of community through amazing fundraisers on Facebook and Instagram. These platforms have made it easier to share our mission, reach people around the world, and raise much-needed funds. Events like the 5th Annual Bike the Pike and the Arpin Strong Charity Golf Tournament show how family-led efforts can make a big difference for KCNT1 epilepsy research and other causes.

Families have gotten creative too, with school bake sales and even a 12-hour game streaming fundraiser on Tiltify led by one dad, who plans to do it again in February.

On Giving Tuesday, families raised money for research tools through a fun and engaging campaign featuring Fil the fruit fly, who helped spread awareness about research readiness, raising almost \$15,000 for research. From heartfelt birthday fundraisers to exciting events like the Penn Million Dollar Bike Ride, which raised \$34,000 (and was matched by Penn for research), every effort has made a huge impact.



As we look to the new year, these fundraisers remind us that every contribution counts. The hard work and dedication of families give us hope for more progress and support in the future. If you'd like to participate in a fundraising committee, please [reach out!](#)

Continue Your Support

Community News

There were many ways to connect with other KCNT1 families and get involved in supporting the foundation. We will be offering many more social opportunities and volunteer roles in 2025!

Family Conference in Philly: Families from as far as Canada and Portugal gathered to meet each other, hear from clinicians at CHOP and meet face to face with drug developers to share top concerns and need for treatments.



June 8, 2024

Jessica Bush Photography

Thank You!

Sibling Circle: Siblings of KCNT1 warriors met online throughout the year to play games and bond over shared experiences with a trained moderator who plans to continue in 2025.

Bereavement Support: Brad Thompson from the Child Neurology Foundation moderated several sessions to help grieving families navigate their journeys with compassion and support.

Family Meetups for Parents of Teen and Young Adults: Parents of our older KCNT1 warriors connected for the first time and shared experiences.

Advocacy and Awareness: Thanks to co-founder Dr. Justin West's relentless advocacy, the KCNT1 community's voice is reaching federal regulators, drug developers, and global experts. His mantra is we want **"better treatments tomorrow, not perfect treatments in a decade."**

FDA Patient Listening Session

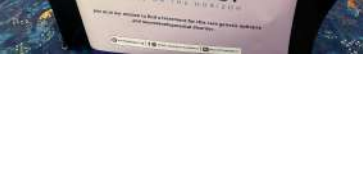
FDA Patient Listening Session: Over 20 FDA representatives heard firsthand from families about the urgent need for KCNT1 treatments. This milestone ensures that patient voices are at the forefront of regulatory considerations as we anticipate companies applying for trials next year. Read our [summary](#).

Chan Zuckerman Initiative

Chosen for the Chan Zuckerman Rare As One Network: Out of hundreds of applicants, we were selected to join this network and receive a multi-year grant to strengthen our infrastructure, enabling us to support families and advance our mission more effectively.



Epilepsy Awareness Day at Disneyland: Our booth at this incredible event brought families and experts together to share knowledge and hope. Two KCNT1 families came to help with the booth and we are grateful for Laura and Rebekkah!



American Epilepsy Society Meeting: Our booth at the largest epilepsy event in the U.S.A brought clinicians, industry and experts together to discuss plans for clinical trials. There were four posters about KCNT1, two presented by us from our registry and Citizen Health data.

A Special KC Call to Action from Parent, Amanda

To My Fellow KCNT1 Parents,
My daughter will turn 8 in 2025. When she was diagnosed with KCNT1 epilepsy in 2017 we were told to take her home and enjoy her but there was nothing else to be done. I asked about gene therapy and her epileptologist responded, "Not in her lifetime."
As a family, we cared for her one day and another. Soon the days turned to weeks and months. Little did I know that as I slogged through diaper changes, seizures, rescue meds and hospitalizations a foundation was being built. This organization started with parents like me who wanted to fight back against "not in her lifetime."
Now we are standing on the precipice of 2025. Multiple pharmaceutical companies have made KCNT1 their focus for drug development. In the next year, we anticipate clinical trials, multiple research studies with incredibly valuable data, and an even bigger community for support. What was not in my daughter's lifetime, is suddenly right in front of her.
Here is the thing I want every parent in this community to know: Hope for helping our children is within our reach but only if everyone participates. Decide now that 2025 will be the year you respond. The KCNT1 Foundation will be increasing engagement, and we need you to be an active participant. By simply responding to emails, surveys, or attending a virtual meeting you too will be fighting back against, "not in her lifetime."
Merry Christmas, Amanda Abuhl, Parent Liaison

Looking Ahead

We have big plans for 2025, including the hope of clinical trial announcements, more family-focused events and in-person meetups, and innovative research initiatives. Your continued involvement fuels our progress and inspires hope for the entire KCNT1 community.

How You Can Help

Your involvement remains critical to our mission. Here's how you can contribute:

Sign Up: Enroll in the Family Contact Form and Citizen Health. (Our new patient registry selection will be announced soon!)

Contribute: Provide biosamples to the Biobank to support ongoing research. Take our surveys this year.

Engage: Attend parent meetings, conferences, and Clinical Trial Readiness webinars.

Share: Tell your story to raise awareness and inspire others.

Volunteer and Fundraise: Help us sustain and expand the initiatives that directly benefit our children.

"As a parent of a child with a rare disease, we face two choices: accept what is or fight for better. Together, we've chosen to fight, and we are closer than ever to a brighter future."

~ Justin West, MD, Founder and Andrew's Dad



Celebrating Our Fifth Year

This year, we celebrated the **5th Anniversary of the KCNT1 Epilepsy Foundation**, a milestone made possible through the passion and dedication of our incredible founders—Seth, Samantha, and Justin—and the leadership of our Executive Director, Sarah. Together, they have transformed personal experiences with KCNT1 into a powerful force for change, uniting families, researchers, and clinicians worldwide in the fight for better treatments and brighter futures. We also want to extend our heartfelt gratitude to our donors, whose belief in our mission fuels every step forward. Starting as far back as 2019, your generosity has enabled us to fund groundbreaking research, support families, and bring hope to our community. To honor this milestone, we've shared a special write-up about each of our founders. Learn more about [Seth's story](#), [Samantha's journey](#), [Justin's advocacy](#), and [Sarah's leadership](#), and join us in celebrating their unwavering commitment to our community. Our team has grown to include Ali Rosenberg, PhD, Brad Bryan MBA, PhD, Megan Wright and Amanda Abuhl. Here's to many more years of progress, leadership, partnerships, and hope!

Thank you for being a part of this journey. Together, we're making a difference.

With gratitude,

The KCNT1 Foundation Team

P.S. Don't forget to connect with us on social media and add our email to your contacts to stay updated!

Donate

Together we can do this!

Our Contact Information
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{{(Organization Address)}}
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