KCNT1 EPILEPSY
HOPE IS ON THE HORIZON

Foundation News & Updates
August 2023

Dear Families,

As summer comes to an end, we wish to provide you with some updates and remind you of our **family meeting on Saturday, September 9**. We will be making some announcements during this call, and co-founder, Dr Justin West, will provide updates on the pipeline drugs that are under various stages of development. Our special guest, Dr David Bearden, will talk about how your participation in studies can influence the drug development process.

And we couldn't get through summer without noting that at least three of our warrior moms are expecting, and we've had a vast number of birthdays -- including the surprising birthday of Nick who turned 44. Sadly, there were also four deaths that we are aware of. It certainly feels like the doors of Heaven are revolving constantly, with so many new lives entering the world and so many souls departing. These losses touch us all: those who've experienced it, and those who fear it. To those hurting, we're here. And to parents worried about their child's health, we understand. **Let's lean on one another, share stories, and remind each other of the support we have.** Together, we can foster hope, and drive change. We can resolve to work even harder, channeling our efforts into finding new treatments, honoring these children's memory with action.

With compassion and determination,

The KCNT1 Epilepsy Foundation

Mark Your Calendars

All Family Meeting: September 9th

11am Eastern

Clinical Trial Readiness: Are We Ready?

With Special Guest, Dr David Bearden

Family Meeting

Ever Need Someone to Talk to?

Attend our first **Parent Circle**

Saturday, September 30, 2:00 Eastern

This will be our first KCNT1 Support Group, drop in!

Parent Circle

Family Meeting at Epilepsy Awareness Day at Disneyland (EADDL)

October 30th 1:00pm

Let us know if you're coming! (More info below!)

RSVP EADDL

OUR FAMILIES

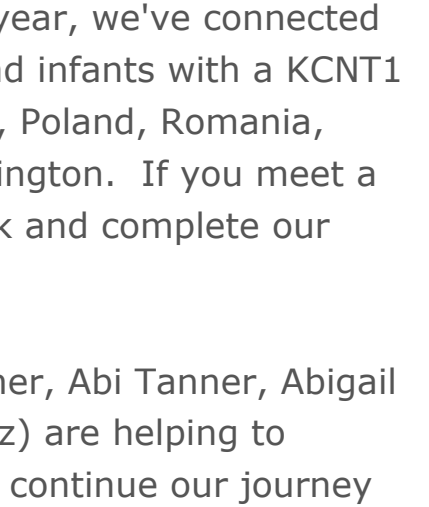
New Families Join Our Network

Exciting News: Our network is growing stronger! This year, we've connected with **15** new families, including adults, adolescents, and infants with a KCNT1 variant. These families are from India, Israel, Moldova, Poland, Romania, Scotland, Colorado, Florida, Texas, Virginia, and Washington. If you meet a new family, please encourage them to join our network and complete our family contact form.

Our parent support specialists (April Hawk, Justin Tanner, Abi Tanner, Abigail Abhul, Heather Patterson, Andy Ip and Kristy Salkewicz) are helping to welcome them with open arms, offering support as we continue our journey together.

Bilingual Volunteers Needed

We are looking for people willing to be translators. If you can help, sign up with this form. Sign up form



Navigating the Path Ahead: Introducing New Educational and Engagement Opportunities

We understand that the road you're traveling, caring for your warrior and managing your life, is far from smooth. It's more like a bumpy road, with unexpected twists and turns that demand your strength and resilience. That's why we're working on new and improved educational materials and opportunities to connect. To help with these initiatives we welcome April Hawk. April has a focus on mental health and also heads up support for another rare disease organization. Stay tuned as we unveil more details about our upcoming meetings and resource releases in the coming months. We will be asking for your input! If you'd like to volunteer to develop educational materials, email us!

SHARE YOUR STORY

As a family with a loved one with a rare disease, you hold incredible power. By lending your voice to the collective narrative, you provide invaluable insights into the challenges, triumphs, and experiences that shape your journey. These stories not only raise awareness but also foster empathy and understanding within the community and beyond. Your voice can shape the design of clinical trials and make an impression on policy makers and regulators. We invite you to make your mark by creating a personal video at the provided link, or by scheduling an interview with our foundation. Your story can inspire, educate, and make a lasting impact.
http://KCNT1.memfox.io/tjzfmh

Family Meeting at Epilepsy Awareness Day at Disneyland October 30th 1:00pm

We are excited to announce a KCNT1 regional family meeting during the Epilepsy Awareness Expo at Disneyland on October 30, 2023, 1:00. At this event, we'll have a chance to meet and say hello. Plus, we have some special guests joining us for a question and answer session. Dr Steenari, pediatric neurologist from Children's Hospital of Orange County will be there. Also, we'll have representatives from pharmaceutical companies and Invitae.

Attend lectures, visit booths at the Expo then join us at 1:00 then join other KCNT1 families the Park!

If you can stay longer, the two-day Expo is followed by Epilepsy Awareness Day at Disneyland Park on November 1st. Reach out to parent, Lorena Avonce, who is coordinating the social plans.

HOW MUCH: Registration for the Expo is free. After you register for the expo, you will receive a link to the discounted Disneyland Park tickets. (Everyone age 3 and above need a ticket to get into the Disneyland Park.) You are welcome to purchase through your own sources or come with a Magic Key Pass – just make sure to make a reservation for that day!

Are you in? Maybe? Help us plan by RSVPing here.

Take our Poll! In-person Family Conference in 2024

Help Us Plan for an In-Person Family Conference(s) in 2024. Fill out this survey to share your preferences and thoughts. So far, the top destinations are: Orlando, Philadelphia and Nashville!

Complete this poll!

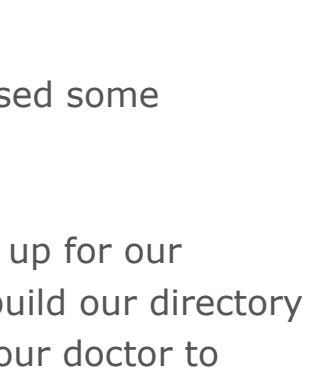
Homework Corner - Past Due!

As part of our Clinical Trial Readiness plans, we would like to ask each of our families to be begin a journey to learn about what clinical trials are, and how they work. We ask that you review these videos and visit the page on our website,>> Participate in Research. There is information on three clinical trials that may be of interest.

The ABCs of Clinical Trials Part 1 >> Video
The ABCs of Clinical Trials Part 2 >> Video

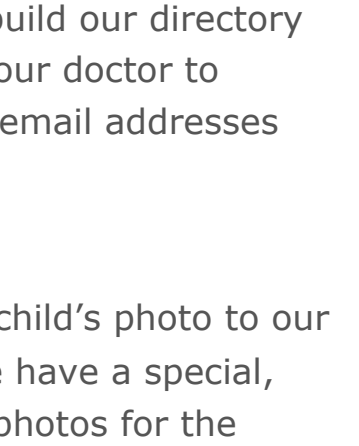
SAMi Sleep Monitor – Discount for KCNT1 Families

Our families understand the importance of monitoring their kiddo's movements during sleep hours. Our friends at SAMi are offering a discount to KCNT1 families when you use this link. http://bit.ly/KCNT1-Foundation Note, with SAMi, You can adjust the sensitivity to the alarm, a feature we know is important to you.



Shhhh...Get Ready!
Grandparents Day is September 10th

Watch for our Facebook post and be prepared to share photos of your warrior and their grandparents! Let's surprise them and use this day to show them our love and appreciation! Remember to tag them!



HAPPY GRANDPARENTS DAY
Share a picture of your KCNT1 warrior with their Grandparent!

Website Updates

If you haven't visited our website lately you may have missed some additions. We have many other changes in the works!

Doctor Directory: We began inviting your doctors to sign up for our directory. We hope to get permission from many more to build our directory of those with KCNT1-related experience. Feel free to ask your doctor to register using the form on our website, as we do not have email addresses for everyone.

Warrior Gallery with Personal Pages: You can add your child's photo to our warrior gallery and make it clickable to your own page! We have a special, dedicated section for our angel warriors. If you submitted photos for the Birthday Crew posts, we will be adding your loved one to our family gallery. Sign up here. We have a special gallery for our angel warriors, sign up here.

Participate in Research and Clinical Trials: This section outlines the important studies our families can participate in. (Some are for U.S. residents only.) Currently we have a few trials that older children may qualify for. Talk to your doctor if you are interested. One day soon we hope to have KCNT1-specific trials to share!

Symptoms and Treatments: This section was written with Dr Bearden in 2021 and will soon be going through an update. If you would like to share any content for this section please reply to this email.

Newly Diagnosed: With the help of our parent support specialists we are updating this page to improve how we support newly diagnosed families. If you would like to offer your assistance to walk alongside a newly diagnosed family, please email April.

We'd Love More of Your Warrior Photos

We need your photos for Epilepsy Awareness Month (November) and the holidays! (Yes..we said, it...) Fill out our photo release form and attach photos! Thanks!

Happy Holidays!
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FUNDRAISING NEWS

MILLION DOLLAR BIKE RIDE (MDBR) - You Brought Hope

Thanks to you, and your friends and families who came out to support our participation in the Penn Medicine Orphan Disease Center Million Dollar Bike Ride (MDBR) we raised $39,274 and unlocked the matching $30,000 grant for KCNT1 related research. Foundation executive director, Sarah Drislane rode alongside parents Lorena Avonce, Michelle Hunt and her team, as well as Jeremy and Kristy Salkewicz. A good time was had by all, and it was wonderful to get warrior families together!

Full Court Press!
Leading up to the event we were able to collaborate with our parents to spread awareness for KCNT1 with two television spots in Pennsylvania and Southern California, and an online article in Jersey! Head on over to our website to check them out! We showed the world that research for KCNT1 matters! Sharing your stories is an important way for us to spread awareness about KCNT1, raise funds, and attracts the interest of researchers and drug developers! We are so grateful for our family participation in the interviews! Look out Hollywood!

What's Next?
We personally invited 70 researchers across the world specializing in *KCNT1 and related epilepsies, interneurons, GABAergic neurons, glutamatergic neurons, ion channels, rare genetic epilepsies, gene editing, and clinical trials* to apply for our $70,000 research grant that will be managed by Penn Medicine and the Orphan Disease Center. Penn also invited THEIR network of scientists. **Stay tuned!** We will award one exceptional researcher who successfully passes our rigorous review process this fall.

If you're interested in participating in next year's MDBR, planning or organizing next year's event, please simply reply to this email! **We can't wait to hear from you.**

Sign up here if you want to be on our mailing list to volunteer or participate in 2024! https://forms.gle/r44AGw9sydGKGbiT9

FAMILY FUNDRAISERS

Thanks to our families who continue to find creative ways to fundraise. Recently Flourish Crystals of Australia held a campaign for Jacalyn and warrior Ember and donated a portion of the proceeds from their purple night! Co-founder, Seth and Susan are hosting an annual birthday fundraiser for Lucy with a BBQ and concert. There are additional fundraisers described below!

The simplest way is to create a "Facebook fundraiser" and use the 'invite' button to share it with your FB friends. Learn how at this link! There are three other family fundraisers planned this fall!

Your fundraisers are an important way to help us fund the work we do and the research programs that need funding. We hope to see many creative fundraisers for Giving Tuesday -- which we are naming **GRATITUDE TUESDAY** this year! (That is the last Tuesday of November.) If you need ideas or help, email us!

Arpin Strong/Arpin International Charity Golf Tournament Sept 29, Bellingham, MA

This year, the KCNT1 Foundation was graciously chosen to be one of the beneficiaries of the 9th Annual Arpin Strong Golf Tournament, Bob Sullivan & Mark Dearborn Memorial in Bellingham, MA. (Other beneficiaries include GIFT, Marguerite's Place, and The Coalition Against Pediatric Pain)

How can you help? Even if you don't live near Bellingham, Massachusetts, you can help!

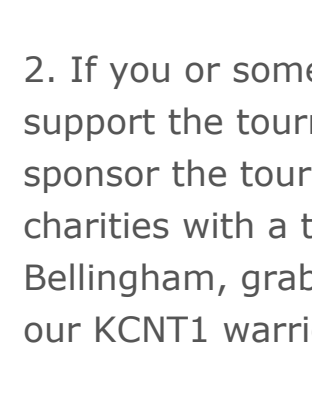
1. **Follow Arpin Strong on social media and thank them in their posts.** Include your photo with your warrior so they can see the people they are helping. Checkout their Facebook page!

2. If you or someone you know owns a business, we encourage them to support the tournament with donations to the raffle, or the business can sponsor the tournament and get great publicity while supporting four charities with a tax-deductible donation! And of course, if you live near Bellingham, grab your sticks and register to play! Bonus: Come meet two of our KCNT1 warriors who will make a special appearance!

3.**Donate an item or service to be raffled** - Arpin Strong is seeking your support. Please consider donating an item or service to be raffled.

This is an amazing opportunity for us to participate in an event that will impact the trajectory of our mission to find a cure for KCNT1 epilepsy. Hope is on the horizon - and that's only possible because of you!

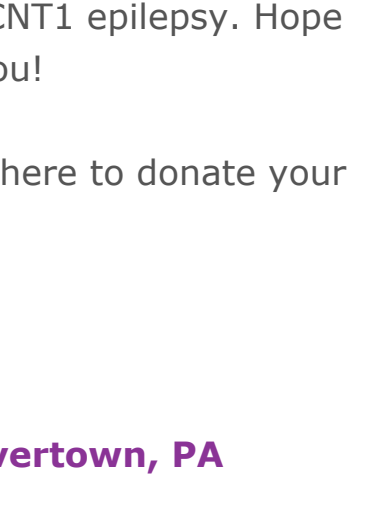
To learn more about ways to support, get involved, or where to donate your raffle item, please visit their page.



Bike the Pike 2023 – Sept 16, Havertown, PA

Another ambitious and generous family member is hosting a bike ride in honor of his second cousin, KCNT1 warrior, Leighanne. For several years, Tyler has organized this ride. Each year this event gets larger, and this year is sure to be a real success! To learn more, visit the event page.

Georgia Power Corporate Golf Tournament Atlanta, GA



Meet Bailee and Emerson!

What are the odds that two families who work for the same company, have a child with a KCNT1 diagnosis? Well, it has occurred. Bailee and Emerson's parents both work for Georgia Power, a company with over 8,000 employees! We are so grateful to have been selected as a beneficiary of their corporate golf tournament!

This event is on October 13th
Canongate 1 | The Clubs of Peachtree City & Newnan | Peachtree City, GA (invitedclubs.com) 924 Shaw Road, Sharpsburg, GA 30277

RESEARCH AND DRUG DEVELOPMENT

You can Help Attract the Interest of Researchers and Pharmaceutical Companies

Our team is working every day to "pitch" our disease to potential partners to increase the odds that someone will develop a drug for us. Some of these drugs will fail. Others will go unfunded. This is why we need to continue to get new drugs into "the pipeline." One important way to attract drug developers and researchers is to have **health data** on all the signs and symptoms associated with KCNT1 gene mutations. Another is to have **blood samples and cell lines**. And importantly, having you share your **stories**.

Right now there are several studies we are asking you to participate in, note some are for U.S. families only:

See the page on our website for more information about observational studies and clinical trials.

Longitudinal Study on Luna

Our longitudinal study being hosted on the LunaDNA platform. This is our international research registry. Throughout the year we will add new surveys. Angel parents are encouraged to take the KCNT1 Specific Demographics survey from their child's account.

Here is a link to get started >>guides. We can schedule a time to help you create a child account, enroll and start the surveys.

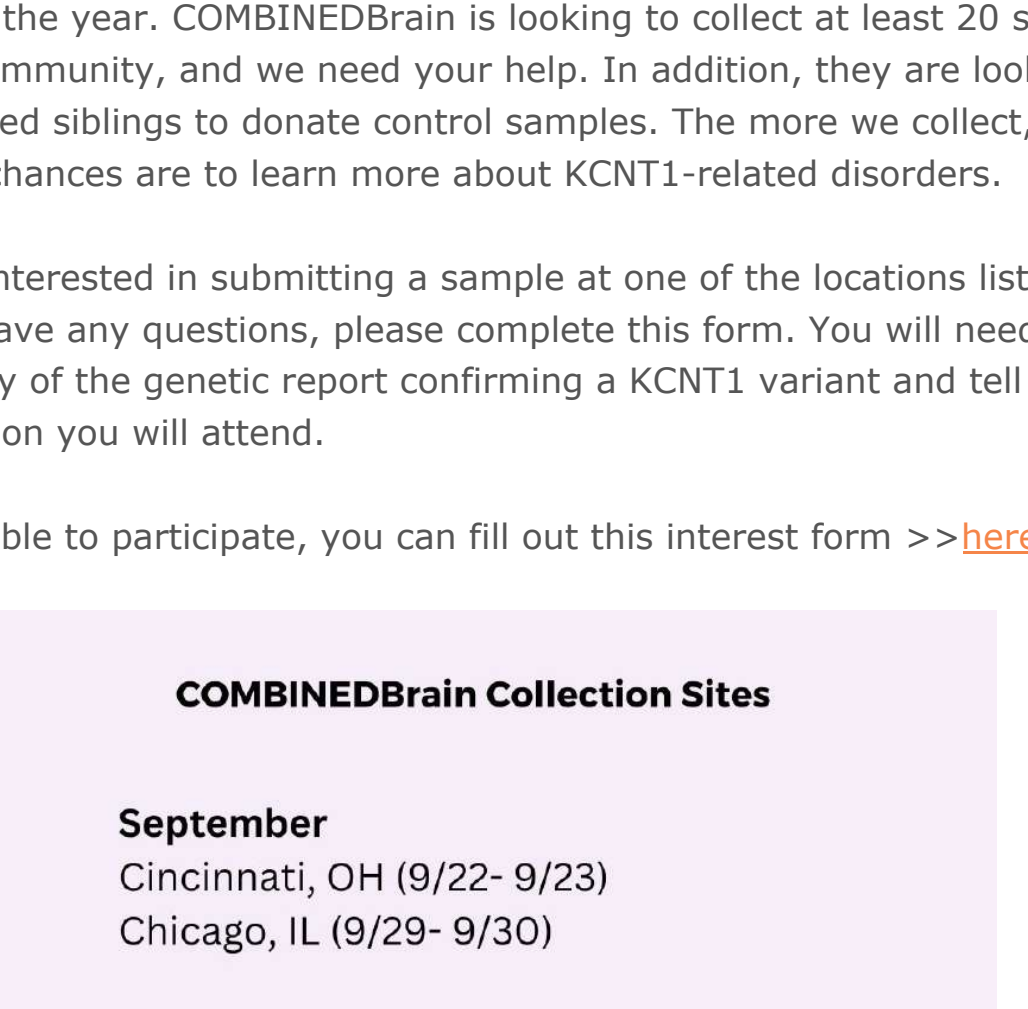
Are you participating in the Biomarker and Biobank Project?

We are a member of COMBINEDBrain, a nonprofit consortium of over 60 rare disease patient advocacy groups. Recognizing the need to collect patient samples for researchers to identify biomarkers to be used to treat/develop treatments for our community, COMBINEDBrain is collecting bio-fluid samples (blood and urine) from member organizations through the end of the year. Samples collected from our community will be stored and available to researchers worldwide.

They are holding collection sites at partner conferences across the country throughout the year. COMBINEDBrain is looking to collect at least 20 samples from our community, and we need your help. In addition, they are looking for unaffected siblings to donate control samples. The more we collect, the better our chances are to learn more about KCNT1-related disorders.

If you are interested in submitting a sample at one of the locations listed below, or have any questions, please complete this form. You will need to email a copy of the genetic report confirming a KCNT1 variant and tell us which location you will attend.

If you are able to participate, you can fill out this interest form >>here.



COMBINEDBrain Collection Sites

September
Cincinnati, OH (9/22- 9/23)
Chicago, IL (9/29- 9/30)

October & November
Rogers, MN (10/1)
Denver, CO. (10/5 – 10/7)
San Antonio, TX (10/12 – 10/15)
Washington, DC (10/15-10/16)
Mercer Island, WA (10/29)
Los Angeles, CA. (10/30-11/1)

December
Orlando, FL. (12/1 – 12/3)

Invitae/Ciitizen Study Recruitment & Insights

Thanks to all who signed up for the digital natural history study on the Invitae/Ciitizen platform. We need 5 more participants to get us to 60. Below you can see a sample of the type of insights we can get from combining all the warrior health records.

55 patients are now enrolled and some of you have received e-gift cards for participation. Some will receive W-2 forms.

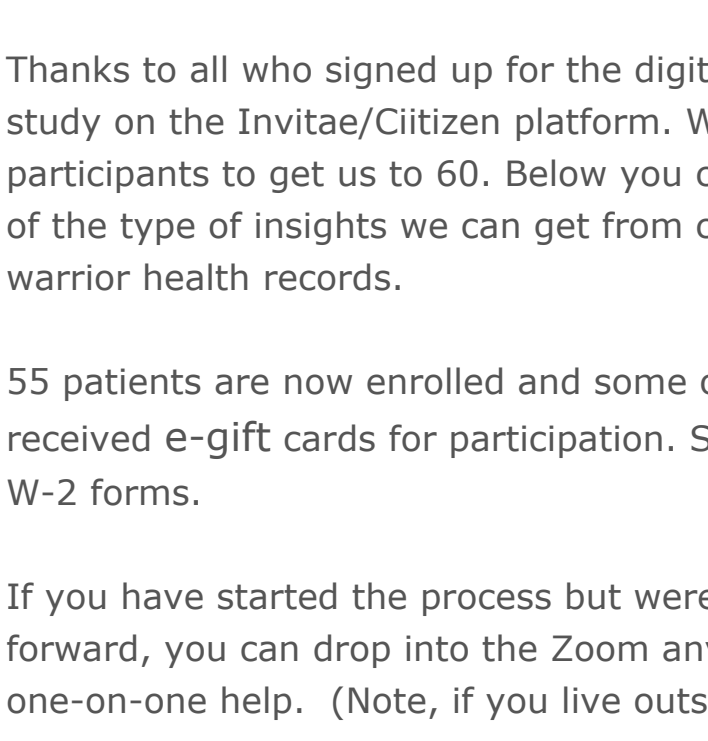
If you have started the process but were unable to move forward, you can drop into the Zoom any Wednesday for one-on-one help. (Note, if you live outside the U.S. and tried to create an account, you will not be able to move forward in the system.)

U.S. families can enroll here.

If you would like help completing your enrollment, Invitae has live support on Wednesdays, just click on the Zoom link:

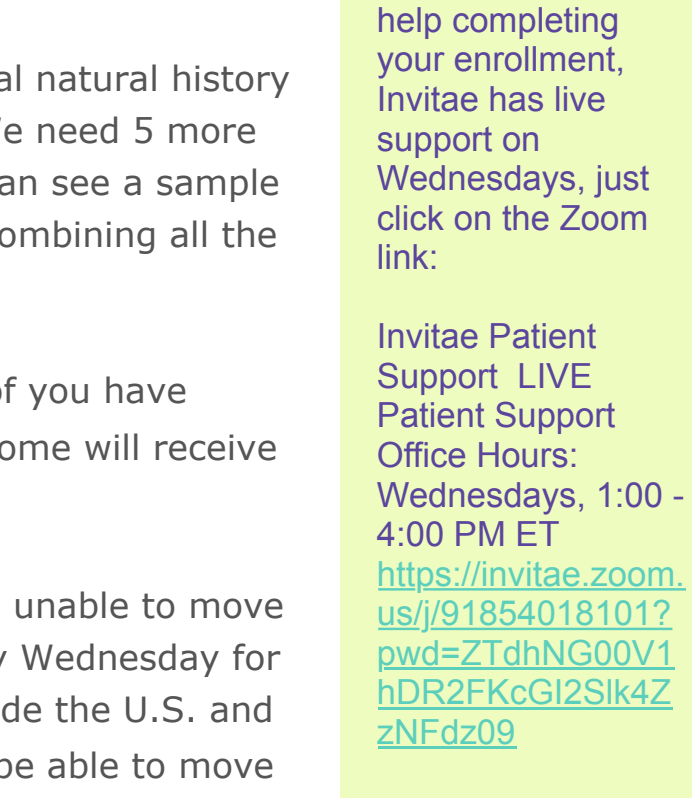
Invitae Patient Support LIVE Patient Support Office Hours: Wednesdays, 1:00 - 4:00 PM ET
https://invitae.zoom.us/j/91854018101?pwd=ZTdhNG00V1hDR2FKcGI2Slk4ZzNFdz09

ID: 91854018101
Passcode: 278849



What is our Invitae/Ciitizen study finding?

For those with ongoing seizures, a majority of our kiddos are on 5 or more anti-seizure medications.



U.S. Families
Are you in our Invitae/Ciitizen study ?
Diagnosis: These are the diagnosis that are being given to our kiddos

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FUNDED RESEARCH

University of South Australia: Through the connection of one of our Australian families, the Barns of Freeling in Australia, an event space (on a crop farm turned sunflower field) hosted a fundraiser to benefit the Foundation. We are using the funds to support the testing of an existing drug (developed for something other than seizures) at the University of South Australia! Professor Leanne Dibbens from the Epilepsy and Neurogenetics Group at the University of South Australia will lead the research which aims to identify new treatments for KCNT1-related epilepsy by testing the effectiveness of existing drugs in blocking the overactivity of KCNT1 channels using cell models. We do not know the name of the drug yet but will share it with the community when this information becomes available.

We are grateful to the Schuster family, the Barns of Freeling, and the generous donors who continue to support our mission to find new treatments.

Other Pending Research Collaborations

We have several potential research collaborations that will be announced soon. These may include: **Virginia Tech, University of California Irvine, Northwestern**, and **Nationwide Children's Hospital**. Our supporters, your friends and family, make this possible!

We Can Do This Together

Your dedication makes all the difference. Let's work together to create a brighter future for those affected by KCNT1. We are truly grateful for your unwavering support! We are so grateful! Email us if you have questions!

Donate

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

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Looking for something?
Check out our Linktr.ee list of links!
https://linktr.ee/kcnt1