KCNT1 EPILEPSY

RESEARCH NETWORK

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 joining the Epilepsy Foundation for their Walk to Defeat Epilepsy in Miami, and provide updates on the positive drugs that are under various stages of development. Our special guest, Dr David Bearden, will talk about his current participation in studies with the Children's Hospital Development process.

And we couldn't get through summer without noting that at least three of our warrior moms are expecting, and we had a last number of birthdays - including the surprising birthday of Nick who turned 44. Sadly, there were also many families affected by the Maui fires and Hurricane Idalia, and we remain in our thoughts and prayers, with so many new lives entering the world and so many souls departing. These losses such as all those who've experienced the loss of Ella, Lily, Ryan, Jackson, Ethan, Ava, Ella, and parents worried about their child's health, we collectively **call to have no one another, share stories, and remind each other of the support we have.** Together, we can foster hope, and share the stories that make our world even kinder, channeling our efforts into finding new treatments, honoring those 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

Even Need Someone to Talk to?
Attend our first **Parent Circle**
Saturday, September 30, 2:00 Eastern
This will be our first KCNT1 Support Group on Zoom!

Parent Circle

Family Meeting at Epilepsy Awareness Day at Disneyland (SADEL)
October 20th 1:00pm
Let us know if you're coming! (More info below!)

SIGN UP SADEL

OUR FAMILIES

New Families Join Our Network

Exciting News: Our network is growing strong!! This year, we've connected with 88 new families, including adults, adolescents, and infants with a KCNT1 variant. These families are from India, Israel, Malaysia, Poland, Romania, Russia, Slovenia, Spain, Taiwan, Turkey, and United Arab Emirates. To our new family, please encourage them to join our network and connect with other family contact forms.

Our parent support specialists (April Hunt, Sarah Tarrier, Mary Tanner, Abigail Allhut, Heather Patterson, Andy Ip and Kristy Salkeros) 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 here

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, demanding your unwavering attention. That's why we're working on new and improved educational resources and opportunities to connect. To help with these initiatives we welcome April Hunt to our team. April will be joining us as our new parent support and another new disease organizer. Stay tuned as we unveil more details about our upcoming workshops 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 living with epilepsy, you can help transform your journey into a collaborative adventure. Join potential therapeutic insights and methodologies, triumphs, and experiences shared within our journey. Share stories not only raise awareness but also foster empathy and understanding within the community. These narratives can significantly influence the design of clinical trials and wield an impression on policy-making and regulators. We invite you to make your mark by creating a personal video at your convenience. Share your story, contribute to our collective foundation. Your story can inspire, educate, and make a lasting impact.
https://docs.google.com/forms/d/e/1FAIpQLSfqNzUrgJ0Zqzzzzzzzzzzzzzzzzzzzzzzzzzzzzzzzzzzzzzzzzzz/viewform?usp=sf_link

Family Meeting at Epilepsy Awareness Day at Disneyland October 20th 1:00pm

We are excited to announce a KCNT1 regional family meeting during the Epilepsy Awareness Expo at Disneyland on October 20, 2023, 1:00- 3:00 PM. Our family meeting is open to all ages and family. Plus, we have some special guests joining us. Dr. Bearden and other epilepsy specialists from Children's Hospital Los Angeles (Children's Hospital of Orange County will be there. Also, we have representatives from pharmaceutical companies and clinical trials.

Attend lectures, visit booths at the Expo (you can join us at 1:00 then join other KCNT1 families the rest)

If you can drive longer, the two-day Expo is followed by Epilepsy Research Day at Disneyland's Grand Californian Hotel. Dr. Bearden, Andrew Escayg, 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 come to the Expo without purchasing a ticket to visit a Magic Kingdom.) Just make sure to make a reservation for that day.

Are you a* 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 in 2024. Fill out this survey to share your preferences for a family meeting the top destinations are: Orlando, Philadelphia and Nashville!

Complete this poll!

Homework Corner - Post Due!

As part of our Clinical Trial Readiness plans, we would like to ask all our families to be begin a journey to learn about the clinical trials process. We have created a new page on our website >> Keeping Research There is information on these critical tasks that may be of interest:

The ABCs of Clinical Trials Part 1 >> Video

The ABCs of Clinical Trials Part 2 >> Video

SAIH Sleep Monitor - Discount for KCNT1 Families

Our families understand the importance of monitoring their kid's movements during sleep hours. Our friends at SAIH are offering a discount for our families. Contact them at info@saih.com or http+1(415)377-0400x2 Note with SAIH: This will relate the sensitivity to the alarm, a feature we know is important to us.

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

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

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 let our directory of those with KCNT1 related experience. Feel free to ask your doctor to sign up. If they are not on our website, we do not have their permission 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 sample dedicated section for our angel warriors. If you submitted photos for the Warrior Wall, we have added your child's photo to our 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 5-15 needed only.) Currently we have a few trials that older children may qualify for. Talk to your doctor if you are interested. One day we hope to have our 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 developing a section for our newly diagnosed families. Let us know whether you would like to offer your experience to seek 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!

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 Peter Pedrizzetti Cancer Foundation Center Million Dollar Bike Ride, we raised $1,000 for the Epilepsy Foundation! Our team was the only KCNT1-related research. Foundation executive director Sarah Grosser rode alongside parents Lorena Arocena, Michelle Hunt and her team, as well as Jeremy and Emily Sallstrom. It could have been sad 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 partners to promote participation. We had a presentation at the Epilepsy Foundation of Southern California, and an online article in Senior Health ran in our favor! Kudos!! We thought that we should the world that research for KCNT1 epilepsy is underway! We hope that this will lead to more conversations about KCNT1, raise funds, and attract the interest of researchers and drug developers! We are so grateful for our family participation in the interviews! Love ya! Hollywood!

What's Next?
We have started KCNT1 research across the world specializing in KCNT1 and related epilepsies, intersecting, Gabbergic receptor pathologies neurons, ion channels, rare genetic epilepsies, gene editing, and clinical trials to apply to KCNT1 research grants. We are also working on a research grant application with our partners at Children's Hospital Los Angeles and Children's Hospital of Orange County. **Stay tuned!** We need to award one research fellowship 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 receive updates in 2024! https://forms.gle/H4KEwXcwWGbCf9 ...

FAMILY FUNDRAISERS

Thanks to our families who continue to find creative ways to fundraise. Recently Pharoah Cordalis of Australia held a campaign for Jacob's birthday and raised $1,000 for the Epilepsy Foundation! Jessica Wright (Jo-Ruoxan, Seth and Susan are holding 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' function to invite your friends to donate to our cause at this time. 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 calling **GRATITUDE TUESDAY** this year! This is the last Tuesday of November, the day before Black Friday!

Arpin Strong/Arpin International Charity Golf Tournament
Sept 25, Bethlehem, NJ.

This year, the KCNT1 Foundation was previously chosen to be one of the beneficiaries of the 9th Annual Arpin Strong Golf Tournament, Rob Sullivan & Mark Dearborn Memorial in Bethlehem, NJ. Other beneficiaries include GIFT, Marquette's Place, and The Cauliflower Against Pediatric Pain.

How can you help? Even if you don't live near Bethlehem, Pennsylvania, you can help!

1. **Follow Arpin Strong on social media and thank them** 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 contact sponsorship@arpinstrong.com to see if they can sponsor or sponsor the tournament and get great publicity while supporting four charities with a tax-deductible donation! And of course, if you feel ready and willing to share your child's story with a sponsor, please let us know! All our KCNT1 warriors who will make a special appearance!

3 Donate **or 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 send them email.

Bike the Pike 2023 - Sept 16, Moorestown, PA

Another ambitious and generous family member is holding a bike ride in honor of his second cousin, KCNT1 warrior, Ella. Ella's family and friends will be joining the Bike the Pike ride. Each year the event gets larger, and this year is sure to be a real success! To learn more, visit the event page.

Georgia Power Corporate Surf Tournament
Atlanta, GA

What are the odds that two families who work for the same company have a child with a KCNT1 variant? Well, Georgia Power and Emerson's parents both work for Georgia Power, a company with over 8,000 employees! We are so grateful that Emerson's parents are a beneficiary of their corporate surf tournament!

This event is on October 13th.
Campaign 1 / The Cause of Peachtree City & Newnan / Peachtree City, GA
Jordyblackburn.com 707 Grove Road, Fayetteville, 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 treatment for our children. We need your help! We need to show that our community is ready and able to get new drugs into "the pipeline". One important way to show drug developers and researchers is to have **health data** on all the signs and symptoms of our children. We need **parents to fill out the KCNT1 Clinical Trial Readiness Survey.** And importantly, having parents 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 LunaBrid 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: >>Update. 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 CONNECtRDiam, a nonprofit consortium of over 60 rare disease patient advocacy groups. Recognizing the need to collect patient samples and data to speed the development of diagnostics and treatments for our community, CONNECtRDiam is collecting the first special (blood and urine) from member organizations through the end of the year. Samples and data from our community will be stored and available to researchers worldwide.

They are looking collection sites at partner conferences across the country throughout the year. CONNECtRDiam is seeing to collect at least 20 samples from our community. We need to show that our community is ready and able to unaffiliated 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 provide your child's name (to start 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.

CONNECtRDiam Collection Sites

September
Cincinnati, OH (8/29- 9/23)
Chicago, IL (8/29- 9/30)

October & November
Rogers, MN (10/2)
Denver, CO. (10/5 - 10/7)
San Antonio, TX (10/12 - 10/13)
Nashville, TN (10/19-10/20)
Mercer Island, WA (10/28)
Los Angeles, CA. (10/30-10/31)

December
Orlando, FL. (12/1 - 12/3)

Invitae/Citizen Study Recruitment & Insights

Thanks to all who signed up for the digital natural history study on the Invitae/Citizen platform. We need 5 more participants to get up to 60. Below you can see a sample of the kind of insights we can generate using the data from our warrior health records.

If you would like to join our Invitae/Citizen study please contact Kristy Salkeros or Parent Support Specialist April Hunt on Wednesday, 1st Sept 2023 at 1:00 Eastern
Join Zoom Meeting
Parent Support Specialists Wednesday 1:00 - 2:00 Eastern
https://zoom.us/j/93884495811?pwd=Z3Rpb0ZNMFFzZ0ZNMFFzZ0ZNMFFzZ