KCNT1 EPILEPSY
HOPE IS ON THE HORIZON

Sharing Milestone Moments

June 2024

Summer has arrived and we hope you will find some time to relax and enjoy your families. We know how much our warriors love to float in the pool and we look forward to your photos in FB and IG! We will keep working hard at the Foundation because don't want to lose our momentum! Grab your coffee because this newsletter is chock full of highlights from our KCNT1 community. We will also ask for a few things from you, because it takes a village, and we need everyone's involvement and support to find new treatments and a better quality of life for our loved ones with KCNT1. **We are proud of this community, its strength, its participation and commitment**. Together, we can do this.

Warmly,

The KCNT1 Epilepsy Foundation

✅ Mark Your Calendars ✅

Sibling Circle
Sunday, June 23 2:00 EST
Siblings ages 5 and older

Sign up for Sibling Circle

Dad's Meetup
Sunday, June 29, 2:00 Eastern
Join us for a virtual gathering of dad and granddads

Dad's Meetup

News & Updates

FDA Patient Listening Session

On April 22, the Foundation, Dr. Bearden, and seven KCNT1 families met with the FDA to share the message that we need new treatments. The private 90-minute virtual session allowed families to discuss caring for their loved ones with KCNT1-related epilepsy and neurological disorders.

We want to thank Julie and Jared Schiller, Erna Glisan, Samantha MacMechan, Preston and Jennifer McIntosh, Lorena and Alvaro Avonce, Heather and Brian Atkinson, and Jacalyn Kerr for sharing their stories and representing our community.

For those unfamiliar, FDA listening sessions let caregivers and foundations directly engage with regulators, offering insights crucial for future decisions on clinical trials.

Though not everyone could attend, your voices matter. Your fellow parents bravely spoke up, laying the groundwork for future discussions with regulators and the development of new treatments.

This meeting was pivotal. Our families' firsthand experiences are driving progress in therapy development.

Thank you to all who participated. Your dedication and advocacy are invaluable to our community.

A Successful Scientific Research Roundtable

We hosted another well-attended research roundtable with 91 participants, including scientists, researchers, industry representatives, students, and parents. The agenda prominently featured discussions on molecular genetics and ongoing research focused on KCNT1. This forum provides an excellent opportunity to share KCNT1 research findings and foster collaborations, bringing hope for new treatments in the future. If you'd like to learn more, contact our Scientific Outreach Officer, Ali Rosenberg.

Speaker	Topic
Helen Willsey, Assistant Professor; UCSF	KCNT1 at the cilium
Amy McTague, Principal Research Fellow and Honorary Consultant Paediatric Neurologist Developmental Neurosciences; Zayed Centre for Research into Rare Disease in Children, UCL Great Ormond Street Institute of Child Health	Patient-derived 2D and 3D model of KCNT1-related epilepsy
Bing Zhang, Professor; Division of Biological Sciences, University of Missouri	Allostatic Compensatory Mechanisms Enable the Mutant KCNT1 Channels to Induce Seizures and Hyperexcitability
Dirk Isbrandt, Senior research group leader; German Center for Neurodegenerative Diseases (DZNE) & Director; Institute for Molecular and Behavioral Neuroscience, University of Cologne	Deciphering the Pathophysiological Mechanisms of Rare Diseases: Uncovering the Complexity of Ion Channelopathies
Len Kaczmarek, Professor of Pharmacology and of Cellular and Molecular Physiology; Yale University	How do KCNT1 mutations result in intellectual disability?
Rajvinder Karda, Associate Professor in Gene Therapy/ **Ellie Chilcott**, Post-doctoral Research Fellow; EGA Institute for Women's Health, University College London	Novel proof of concept therapy for KCNT1 epilepsy
Mattie Monroe/ Carolyn Bell; MS candidates in Human Genetics & Genetic Counseling Candidate, Stanford University	Pharmacogenomic Resources for Prescribing ASMs
David Bearden, Department of Neurology and Pediatrics, University of Rochester Medical Center	Title TBD
Sarah Poliquin, Postdoctoral Fellow; COMBINEDBrain Consortium for Outcome Measures and Biomarkers for Neurodevelopmental Disorders	Title TBD
Sumaiya Islam, Director of Translational Research; Tempus AI, Inc	Proposal to develop a KCNT1-related immune signature
Dr. Bearden as Moderator, all participate.	Open Q and A to all speakers Open questions/guided discussion

Our Family Conference Brings Hope

With the help of our friends at the **Children's Hospital of Philadelphia**, we hosted a successful conference where 12 families from as far as Portugal, Canada, and California came together. Many families brought their KCNT1 warriors, and we were also joined by two bereaved families—Monica and Peter Ahn, and Andy Ip—who shared their passion to support our cause. Families met with doctors and therapists from the CHOP team to learn about respiratory health, the ketogenic diet, and effective physical therapy methods. (CHOP presentations can be found here.)

Dr. Bearden explained different treatments like small molecules and ASOs, talked about clinical trials, and emphasized how we can help speed up the process. Representatives from five drug companies explained the long process of testing new medicines. We were hopeful to hear they are all committed to developing treatments for KCNT1, which was encouraging for our growing community.

The conference highlighted the importance of surveys, registries, and initiatives like Ciitizen to gather data. We also got updates on support programs and research funded by the Foundation. Emotional stories from parents moved everyone, including the pharmaceutical representatives from Biogen, Servier, Actio, Atalanta and UCB:

> **"We were incredibly grateful to the families and all that they shared of themselves. This sort of venue gives us reason to press on!"**
>
> **"It was informative, impactful and showed the courage and resolve of the families. It left a lasting impression that I will carry with me while we continue to work in this space."**
>
> **"I can't thank you enough for letting me come down and join you today...I think I have met over 15 families due to your invitations. It really puts so much in perspective for me and further reinforces the importance of moving things forward."**

Parents found the conference informative and hopeful, as they shared in their testimonials. We ended the day with a friendly social event and an Italian dinner from a local restaurant called Ambrosia. It was a time for families to connect and support each other, with lots of hugs and smiles. We also celebrated Aksel Ahn's 6th birthday with cake and a birthday song.

Watch some of our parent testimonials here.

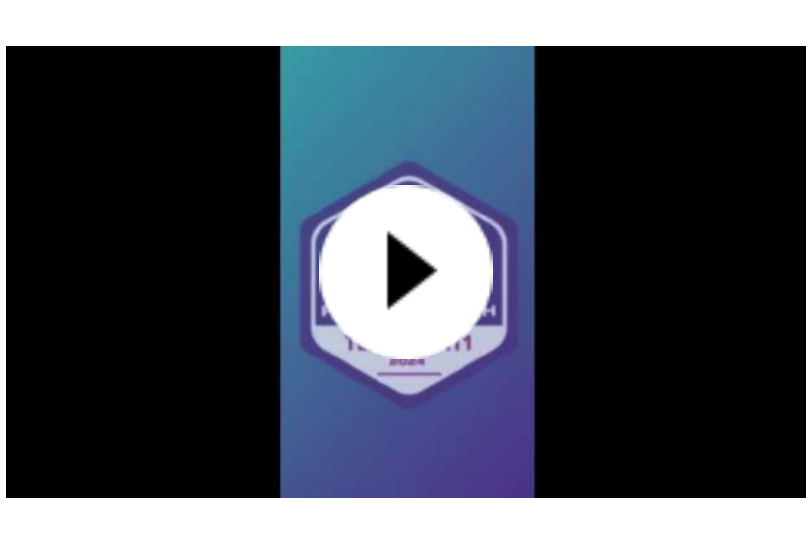
We are grateful for our conference sponsors!

We Reached our Goal for the Million Dollar Bike Ride

As the morning sun dried the dew on Highline Field, we gathered with our bikes and helmets, ready to commemorate our warriors. We started with a touching moment, releasing butterflies in memory of those we've lost too soon. Parents and children joined together to launch these butterflies into flight.

Then, our activities kicked off: some joined dozens of rare families on a one-mile walk, while others embarked on cycling routes through Philadelphia, spanning either 10 or 32 miles! Jeremy Salkewicz, James' father, proudly led the way as the first cyclist to complete the challenging 32-mile route. Bringing up the rear were President Justin West, along with friend, Dr. Andy Burchard, and Executive Director Sarah Drislane, who faced unexpected challenges, including Sarah's two flat tires within just 10 miles!

Your support and sharing of our posts and MDBR campaign made a significant impact —we raised $36,000, unlocking an additional $30,000 matching grant from Penn Medicine Orphan Disease Center for KCNT1 research! Team Grace in Sydney, Australia rode 77 miles in support of the event, and Team Emerson and the L5 Cycling team rode 75 miles in Atlanta raising nearly $5,000! Team Garlewicz was atop the leader board raising almost $14,000 for KCNT1 research! And the momentum continues: our fundraising efforts extend until June 30, with an opportunity to secure not one, but two research grants if we reach an additional $14,000. In July, Penn will announce a Request for Research Proposals (RFPs), to be evaluated by a dedicated grant review team, with awards slated for December or January. This is real progress for KCNT1!



Meet some of our families and hear about why they rode in the Million Dollar Bike Ride come back to read more and see our group photo and event **photos here!**

Your Voice Matters

Coming to a Town Near You!
Donate Blood for our Biobank

Biobank Interest Form

Researchers need blood and cells from real people with KCNT1 variants to conduct their research and test new drugs. You can help by donating blood, tissues and urine to our biorepository from your KCNT1 warrior and an unaffected sibling. We have a partnership with COMBINEDBrain who manages the process and helps distribute samples to qualified researchers. COMBINEDBrain sets up their collection sites at various rare disease conferences and opens it up to all partner groups for their families who live in the city. Below is a schedule of cities where there will be collection sites. If you are interested, complete our form and we will be in touch with next steps! Thank you to the 13 families who already contributed!

Denver: June 21, 22
Philadelphia: July 19 – 21 and July 27-28
Seattle: July 29 – 30
Atlanta: September 27-28
Los Angeles: December 4-5

Friendly Faces You'll Be Seeing

Megan Wright – Megan, the mom of Lexi, came forward to offer her time and talents to the Foundation and newly diagnosed families as well as to take on the responsibility of Administrator for the Facebook KCNT1 Gene Mutation Support Group. As part of this role, Megan issued a survey to collect your input and ideas about the group and we still need more responses. Your participation is important in shaping the future direction of the group. Please take a moment to share your thoughts through the survey. We look forward to sharing the survey findings with the group soon. Click here to participate: Survey Link

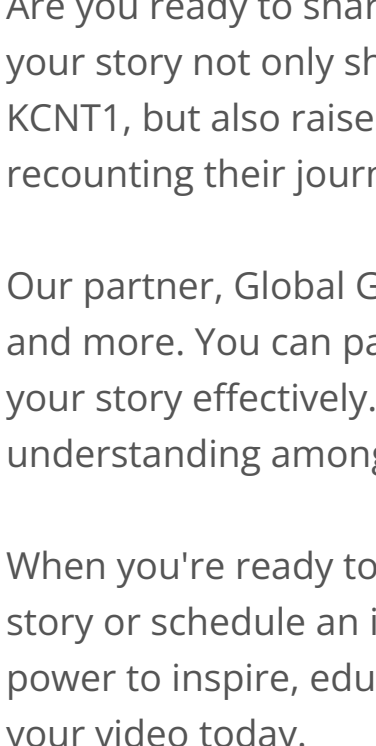
Heather Patterson – Heather is the mom of Emma, and with six years of experience with caring for her warrior, Heather decided to offer her experience to the Foundation and the community and signed up for the parent support training and other special projects. Heather's years of experience in the hospitality industry was very helpful when we needed to plan the conference!

Brad Bryan, PhD – Dr. Brad Bryan joins the KCNT1 Epilepsy Foundation as our new Data Strategist Lead, bringing extensive healthcare experience, including running clinical trials and building patient registries. With a PhD in Medical Sciences from Texas A&M University and an MBA from the University of Massachusetts, Dr. Bryan's strategic insights and background in rare disease advocacy will be instrumental in advancing our mission. Outside of work, Dr. Bryan is a dedicated veteran and volunteer instructor in youth sports and enjoys hiking, traveling, and playing musical instruments. We are excited to welcome him and look forward to his innovative contributions.

Kaitlyn Esposito – Kaitlyn is our clinical coordinator for the observational repurposing study of carvedilol, bringing ten years of experience in patient-centered research and health education. She has experience with other patient advocay groups and has directed research programs, managing multiple studies and overseeing a large-scale study across national sites. She holds a Masters of Public Health and a Bachelor of Science in Biological Sciences. Kaitlyn's dedication to research and supporting patients makes her a valuable addition to our team.

Community Spotlight: Lucy's Legacy

Each newsletter we will share the story of someone from our community -- it may be a donor, a family, a researcher, clinician, caregiver, grandparent, Board member or Foundation volunteer or staff person.



In recognition of our first anniversary, we feature the founders, **Seth and Susan Greenblott** who launched the foundation when their youngest daughter, Lucy was diagnosed. You can read about Lucy's Legacy here.

Other Happenings

How to Share Your Zebra Story – Workshop July 16 12ET

Are you ready to share your rare disease journey but unsure where to begin? Sharing your story not only sheds light on the challenges and triumphs you've experienced with KCNT1, but also raises awareness and offers support to others. Many find that recounting their journey is empowering.

Our partner, Global Genes, offers valuable resources to guide you through this process and more. You can participate in webinars specifically designed to help you articulate your story effectively. These narratives serve to increase awareness and foster understanding among others.

When you're ready to create a brief video, you can use the provided link to record your story or schedule an interview with our foundation. Your unique experience has the power to inspire, educate, and create a lasting impact. Click here to begin recording your video today.

Sign up here for the story telling workshop Tuesday, July 16 12:00 ET

Last Chance - Join the Others Who Have Completed Our Report Card

Please take our survey to provided us some feedback on our efforts. As a nonprofit we have a duty to use our time and donations effectively to ensure we are marching towards our mission and your feedback can help guide us. Click here!

FUNDRAISING NEWS

They're Back! The Haverford, PA biking team is back again for their 7th year! The cousin of a KCNT1 warrior is the expert organizer bringing together the entire town to support our cause on September 14th! #BikethePike

Some things are too good to be true! But yes, the KCNT1 Epilepsy Foundation has been selected as one of the beneficiaries of an Arpin Strong Golf Fundraiser in Rhode Island on September 27th! We look forward to our community rallying behind this event to find auction items, sponsors and players to make this event a huge success! Contact us if you'd like to help!

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! Email us if you have questions!

Donate