KCNT1 EPILEPSY
HOPE IS ON THE HORIZON

Parent Newsletter
Jan-April

FUNDRAISING NEWS

ARPIN STRONG: CORPORATE CHARITABLE GIVING

We had no idea what an impact one KCNT1 grandfather could make when he suggested to one of his clients (Arpin International) that they consider the KCNT1 Epilepsy Foundation to be one of the recipients for their charitable giving campaign. For the month of March Arpin created a webpage and promoted a social media campaign to support our warriors. The company is known for their charitable giving and has over 4,000 employees, many of whom made donations to support our mission. Thanks to you who followed their page and thanked them for their support! It's not too late, you can visit the page and express your gratitude here. Arpin page.

MILLION DOLLAR BIKE RIDE (MDBR)

Research Grant Matching Program: UPenn Orphan Disease Center Million Dollar Bike Ride (MDBR)
This is a unique grant-matching program from UPenn Medicine Orphan Disease Center. They will match the funds we raise (up to $30k) and apply it towards research from the most promising KCNT1 research projects available. We have three teams traveling to Philadelphia and two virtual teams so far! Watch our Facebook page to meet them over the coming weeks!

There are many ways you can support this important opportunity:
Ride: Register to Join TEAM KCNT1 and ride in Philly
Volunteer: register as a TEAM KCNT1 volunteer (be sure to create a fundraising page or you won' show up on our team)
Virtual: Register as a virtual team and fundraise-- and host your own event
Donate to TEAM KCNT1 so we can meet our minimum of $20k in order to get the matching dollar for dollar grant!
We must raise $20,000 in order to get the matching grant for research!
Registration is open https://charity.pledgeit.org/t/eXltXfZG2a
We can't do this without you!

FAMILY FUNDRAISERS

Wow! What an amazing turn out with family fundraisers so far this year! Thanks to Charlie Heutling, Jacalyn Kerr, Alex Wild, Justin and Lisa West, Stefanie Becerril and Chloe Zapantis! Did we miss someone? Your fundraisers are an important to help us fund the work we do and the research programs that need funding. If you need ideas or help, email us!




Bonfire Store - We Need Your Photos

Thanks you to those who have purchased merchandise from our shop on Bonfire! We'd love to feature you in future social media posts with your new gear! Here is Nate's mom, Lorena, sporting her t-shirt for the Million Dollar Bike Ride she is participating in!

Rare Disease Day Update

Thanks to all of you who rallied on Rare Disease Day sharing our posts and spreading the word about KCNT1 with your social media frames. The extra effort of tagging the Foundation helped expand our reach and gain new supporters. With your help, you raised $7,800.

OUR FAMILIES

HUGE Announcement: Coming Soon!
The Australian Branch of the KCNT1 Epilepsy Foundation

We are so excited to announce that KCNT1 mom, Chloe Zapantis, has agreed to lead the first sister organization of the Foundation in Australia! The details are being worked out, but we believe this will help bring funding to the KCNT1 work being done in Australia, and bring clinical trials to Australia. Welcome Chloe and thanks for Levi for sharing his mum with us!

New! Birthday Crew

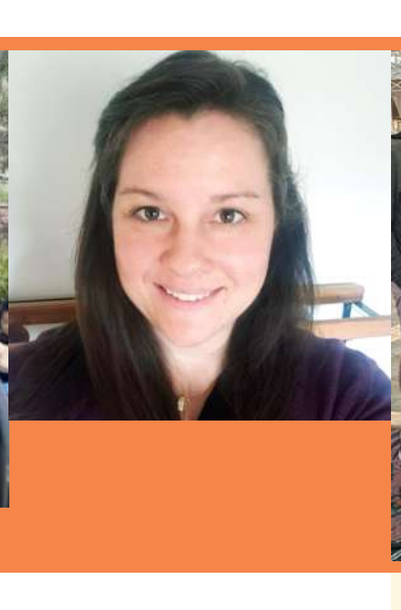
We are excited to have parent, Megan Murray, step up to manage the Birthday Crew program. It is wonderful to see all the birthday wishes being exchanged on our Facebook Page. You can still sign up your warrior or angel warrior >> here. But wait – there's more! For an added bonus, you can spread the joy and also host a birthday fundraiser for the Foundation on your own page. Let us know if you need help!

SHARE YOUR STORY

Even though our children have a KCNT1 gene variant, each child is unique. And each family has a unique story. If you are willing to share your story on video, here is >> a link and a photo consent. It is helpful to have your stories when we talk to pharmaceutical companies, regulators, donors and the public when trying to build awareness!

Peer Support Specialists Are Here for You

With the support of our donors, we were able to fund peer-support training for five parents hosted by the Child Neurology Foundation. These parents will serve as a point of contact for recently diagnosed families, or anyone wanting to to talk to someone. They are also helping to develop new resource materials. A form has been added to website for parents to request a meeting with any of them. Thanks to Andy Ip, Amanda Abhul, Abby and Justin Tanner, and Heather Patterson. Ambassadors and parents Angie Utting and Michele Martel are also joining in! If any of you are interested in volunteering or becoming a Peer Support Specialist, a new class will begin in the fall. Email us.

Family Meet-ups Coming Soon

We recently organized family meet-ups for Dads, and another for grandparents. If you missed it don't worry, we will have more sessions in the future.

Parents & researcher at Leeds April 26

Dad's meetup is S**aturday, May 6th 8pm** Eastern, sign up >> here >>

Grandparents can sign up >>here

ANDFLE (SHE) interest form >> here

Brazil Webinar May 6 >>

Homework Corner

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 Clincal Trials Part 2 >> Video

Welcome our Ambassador to Japan

We'd like to welcome our newest parent ambassador, Dr Hiroharu Yamashita. A father of a KCNT1 warrior, a surgeon and a professor, Dr Yamashita and his wife have been busy doing outreach in Japan locating other KCNT1 families and have connected with ten so far! Thanks for all your hard work! We also thank Catherine Holliday (UK), Ana Serena (Brazil) and Angie Utting!




RESEARCH AND DRUG DEVELOPMENT

Participate in Research and Trials

There are several ways you can do your part to contribute to the knowledge about KCNT1, including participating in observational studies. See the page on our website for more information about observational studies and clinical trials.

Our New Longitudinal Study is Open on Luna

The Foundation has a new longitudinal study being hosted on the LunaDNA platform. This is our international research registry. Throughout the year we will add new surveys to this study and get a clearer understanding of how each family is impacted, and, hopefully capture the unusual signs and symptoms each of you describe in the Facebook groups. There are two surveys in the study now: the KCNT1 demographics and genetics (which is in 7 languages: German, French, Portuguese, Italian, English, Spanish and Swedish) and a Family Impact Scale which you will take twice per year. Angel parents are encouraged to take the KCNT1 Specific Demographics survey from their child's account.

We developed a guide to ensure you are in the correct account type before taking the surveys. (Please take surveys from within a child account.) Here is a link to get started or see our >>guides. We can also schedule a time to help you create a child account, enroll and start the surveys. Email info@kcnt1epilepsy.org

Biobank Launched

Working with our partner, COMBINEDBrain, we have launched our biorepository. Thank you to the parents who have signed up and submitted samples! Having this asset will attract more pharma interest so the more samples we can get the better! When we get more samples we will issue press release and posts in an effort to reach pharma and researchers. We've had one inquiry so far.

In some cases we can have a phlebotomist come to your home. There will be blood collection locations in the following cities:

Camden, NJ - April 30
Westmister, CO – July 30
Queens, NY - Aug 3-6
San Diego, CA – Sept 19
Denver, CO Oct 5-7
Los Angeles, CA – Oct 30
Orlando, FL – Nov and Dec

If you are able to participate, you can fill out this interest form >>here. It gives the Foundation permission to share your information with COMBINEDbrain to help coordinate your appointment.

Invitae/Ciitizen Study Recruitment & Insights

Thanks to all who signed up for the digital natural history study on the Invitae/Ciitizen platform. We are excited to share that we will be partnering with Children's Hospital of Philadelphia to analyze the data. We will share insights as they are provided!

52 patients are now enrolled in the Invitae study and have received e-gift cards for participation. Some of you have started the process but were unable to move forward. (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 by clicking on the green box on our webpage

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

New Interest from Pharmaceutical Companies

The more data we collect + the more specimens we have + and the more patients identified = the more interest we garner. We are excited to share that in the past three months we have been approached by two additional pharma companies. If you hadn't read the president's end of the year letter in December, you can find it >> here.

Repurposing Drug Studies

We are working with researchers and doctors to finalize a treatment and trial protocol for a drug identified in the repurposing screening process. We must get approval to test it for seizures because it's not approved for seizures. We will also fund the testing of this drug on KCNT1 mice at Virginia Tech. We expect future repurposing trials to take place in the UK and Australia.

FUNDED RESEARCH

Note From Researcher, Will Tobin, University of Vermont

The discovery of epilepsy-causing gene mutations has allowed the development of precision medicines that target the root cause of disease. Yet, to realize the full, side effect-free, therapeutic potential of these tools we will need to treat the right cells, in the right part of the brain. This year, with the support of a Taking Flight Award funded jointly by the KCNT1 Epilepsy Foundation and CURE Epilepsy I am studying whether cutting-edge gene and drug therapies can be improved by targeting only the most severely affected cells and brain networks in mice with KCNT1-related epilepsy. This work is designed not only to optimize seizure control but also the correction of abnormal brain activity outside of seizures in epileptic mice, recognizing that our true goal is to holistically restore normal function in the epileptic brain.

Variants in the KCNT1 gene cause severe childhood epilepsies. At the molecular level, they cause the potassium channel encoded by the KCNT1 gene to be overactive. New drugs and gene therapies have been developed to inhibit the channel and show promise in mouse models, but not all KCNT1-expressing cells contribute to disease. Identifying which cells to treat for maximum benefit is critical because treating the wrong cells can needlessly disrupt brain activity, lead to side effects, and undermine treatment goals. By studying individual brain cells from mice with KCNT1-related epilepsy, we've learned that the variant most severely impacts a group of cells called inhibitory interneurons. Currently, we are testing whether treating only these cells improves outcomes by suppressing seizures and other abnormal activity without otherwise disrupting brain function. Additionally, by imaging activity in the intact brains of these mice, we discovered that a few specific areas in a structure called the cortex predictably generate epileptic activity. Following a similar logic, we are also investigating whether restricting treatment to these *regions* can improve outcomes in the same way. To assess these treatment strategies, we are both measuring how they affect seizure burden and also how well they "normalize" pathological brain activity outside of seizures themselves. We accomplish this by comparing brain activity imaged in the cortex of epileptic mice to that seen in healthy individuals, a process that reveals where, how, and to what degree activity deviates from the normal range.

This study builds on our progress in understanding basic disease biology to address a critical next step in improving therapies: targeting them to the right place. Moving forward, we hope to extend this work to include optimizing cell type and brain area targeting *in combination*, studying how age at the time of treatment affects outcomes, and how to use mouse behavior to better evaluate outcomes. These efforts are ultimately driven by our vision of a future in which patients and families that receive a KCNT1 epilepsy diagnosis have more hope because effective therapeutics developed using sound science are available. And without support from the KCNT1 Epilepsy Foundation, none of this would be possible.

About Will Tobin: Will pursued his undergraduate degree in Neuroscience at the University of California Santa Cruz, where he developed a strong foundation in the biological basis of behavior. He then went on to complete his PhD in Neurobiology at Harvard Medical School, where he honed his skills in genetics and molecular biology. After completing his training, Will decided to return to his roots in Vermont and joined the Weston lab in 2017. Since then, he has been working tirelessly to uncover the genetic mechanisms underlying epilepsy, with the goal of developing more effective treatments for this debilitating condition.
Outside of the lab, Willie is a devoted family man. He is married to a talented chef and restaurateur, who co-owns two popular restaurants in Burlington. Together, they have two young sons, who keep them busy and entertained.
Despite his busy schedule, Will always makes time to pursue his passions outside of work. He is an avid outdoorsman, and loves to explore the forest, bird watch, and stargaze. He is also an amateur mycologist, and enjoys cultivating gourmet mushrooms in his spare time.

We Can Do This Together

Thanks for all you do! There are many ways you can help support KCNT1 research, outreach and community! Participate in research. Volunteer. Consider a donation today. We are so grateful! Email us if you have questions!

Donate

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

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