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SAVE THE DATES

PCDH19 Alliance Professional and Family Conference

June 29-30, 2018
Hyatt Regency Aurora-Denver Conference Center

PAME (Partners Against Mortality in Epilepsy) Conference

June 14-16, 2018
Alexandria, VA

The fourth PAME conference will again bring together healthcare providers, researchers, public health officials, patient advocates, caregivers, families, and patients living with epilepsy. This diverse group of stakeholders will focus on the shared goal of improving our understanding of mortality in epilepsy, including Sudden Unexpected Death in Epilepsy (SUDEP) Register here!

Bridge the Gap SYNGAP Annual Golf Classic

FRIDAY, MAY 11, 2018
CYPRESS LAKES GOLF CLUB
18700 CYPRESSWOOD DRIVE, CYPRESS, TX 77429
12:00 P.M. SHOTGUN START

Dinner and Silent Auction Only
4:30 - 6:00 P.M.
$10 Tickets

4th International CDKL5 Family Education & Awareness Conference

June 29-30, 2018
Hyatt Regency Aurora-Denver Conference Center
**United in Hope: Phelan-McDermid International Family Conference and “Phelan-McPosium”**

**Dallas, Texas  ♦  July 18-22, 2018**

The 11th Biennial Phelan-McDermid Syndrome Foundation International Family Conference “United in Hope” is for those of all ages and provides families with educational workshops, family support sessions and firsthand research information on Phelan-McDermid Syndrome (PMS). The “Phelan-McPosium”, which is run in conjunction with the International Conference, gives families an opportunity to interact directly with researchers and inform future areas of study.

Our International Conference is the largest support group gathering in the world for those whose lives are affected by PMS. More than 800 people are expected to attend, including families and individuals with PMS, researchers from around the globe, educational and medical professionals and caregivers of PMS individuals. Attendees come from all over the world. Our 2016 conference had attendees from the United States, Canada, Germany, France, Spain, Ireland, Italy, Brazil, Costa Rica, Mexico, the United Kingdom and Malaysia.

This conference is sure to be one of the most fulfilling experiences along a family’s journey with Phelan-McDermid Syndrome.

More information can be found at [www.pmsf.org](http://www.pmsf.org). Follow us on Facebook at [https://www.facebook.com/PMSF22q13/](https://www.facebook.com/PMSF22q13/) and Twitter at @Phelan_McDermid.

If you’d like to be a sponsor for our conference, please go to [http://www.pmsf.org/family-support/pmsf-international-conference/sponsorships/](http://www.pmsf.org/family-support/pmsf-international-conference/sponsorships/)

* - Nick Assendelft, Vice President, Phelan-McDermid Syndrome Foundation

**Dravet Syndrome Foundation:**
**Biennial Family & Professional Conference**

**Aurora, Colorado  ♦  July 19-22, 2018**

This 3-day event is unique in that it is designed to unite all groups committed to improving the lives of those with Dravet syndrome — including families, caregivers, clinicians, researchers and professionals in the pharmaceutical industry. There will be speaker presentations on the latest advances in research as well as sessions with up to date information impacting patient care. This event allows the opportunity to foster new relationships and collaborations, both for families and professionals. In collaboration with Children's Hospital Colorado.

More details at [www.dsfconference.org](http://www.dsfconference.org)

**Dup15q Alliance Opportunities**

RFA for Research Grant Program and registration for the August Research Symposium are open. Learn more [here](http://www.dup15q.org)!
**FasterCures: Data Matters - a patient-centered approach to demystify health data**

FasterCures has been awarded a $250,000 PCORI (Patient-Centered Outcomes Research Institute) grant to develop a tool to help people understand what makes up their health data, how they are collected and shared, and who uses them. Surveys, in-depth interviews and user testing with participants will inform the design and refinement of a tool that is brief, easily accessible to individuals and aligned with their educational needs.

Learn more at [https://www.healthdatabasics.org](https://www.healthdatabasics.org)

**Hypothalamic Hamartoma & Gelastic seizure storyline featured on ABC television show Grey’s Anatomy**

The Grey's anatomy Twitter account (with 4.7 million followers!) shared information and gave a shout out to Hope for HH.

**Dreams for Danny: Surgical evaluation travel scholarship from The Brain Recovery Project, Childhood Epilepsy Surgery Foundation**

Many families struggling with financial issues find it difficult to obtain a surgical evaluation at a hospital far from home. Airfare, ground transportation, and lodging costs can add up when your child needs several days of video EEG monitoring, imaging, and other tests.

This is why Dreams for Danny was created – to help families afford travel and lodging associated with an epilepsy surgery consultation. Funded by a very generous donation from the Abel family in honor of their son, Danny, this new program provides up to $1,000 in approved travel funds for scholarship awardees.

F.D.A. Peripheral and Central Nervous System Drug Advisory Committee hearing on Epidiolex

On April 19, 2018, Epilepsy Foundation President & CEO Phil Gattone testified before the U.S. Food & Drug Administration (FDA) Peripheral and Central Nervous System Drug (PCNS) Advisory Committee in support of the approval of Epidiolex for the treatment of Dravet and Lennox-Gastaut syndromes. Phil was joined by Polly VanderWoude, the mother of Olivia, who has been part of the Epidiolex clinical trials under the care of Dr. Orrin Devinski. Polly shared with the committee Olivia’s journey before and after Epidiolex and how the medication has allowed her daughter to experience seizure free days and an improved quality of life. Many other families shared stories similar to Olivia’s during the open public hearing portion of the meeting. Also in attendance was EF Professional Advisory Board member, Dr. Barbara Kroner with her daughter Ellie, who has also been part of the clinical trial.

The Advisory Committee voted unanimously (13 -0) in support of approval of Epidiolex. This follows the recommendation by staff that Epidiolex be approved. A decision by FDA is expected by June 27. We issued a statement applauding the Advisory Committee’s action and several news outlets picked up the story (MedPage Today, CNN, NBC News and Washington Post).

Epidiolex represents hope for the many individuals living with intractable seizures and rare epilepsies. The Epilepsy Foundation will continue to advocate for changes in state laws required to ensure a state pathway for timely access to Epidiolex once approved by FDA. Many have joined us on this effort which has been underway since 2016.

(Epictured L-R: Phil Gattone, Orrin Devinsky, Polly VanderWoude, Barbara Kroner)

- Beatriz Duque Long, Senior Director Government Relations, Epilepsy Foundation

Epilepsy Foundation (EF) News: The National Walk, Public Policy Institute and Teens Speak Up Conference 2018

On Saturday, April 14, more than 2,400 walkers, including people living with epilepsy and their family and friends, took part in the 12th Annual National Walk for Epilepsy on the National Mall in Washington, D.C. On a beautiful spring day, participants celebrated epilepsy awareness, connected with new friends and old, and learned about how to get involved with the Epilepsy Foundation. Virtual walkers across the country also joined by walking and spreading awareness in their local communities.

A reoccurring feature at the Epilepsy Walk is the Remembrance Wall, where participants decorate flags in memory of loved ones who lost their life due to epilepsy-related causes.

Elizabeth Dewey, a freshman at the George Washington University, attended the walk for the first time as a volunteer. Elizabeth’s mother Eva passed away from sudden unexpected death in epilepsy (SUDEP) when Elizabeth was only eight years old. She took time to honor her mother at the Remembrance Wall with her best friend Surabhi, another volunteer.
“Putting the flag on the Remembrance Wall was a lot more emotional than I thought,” Elizabeth said. “When I went over to the wall and put the flag up, I started crying. It wasn’t necessarily out of sadness. It just meant so much to me that I was able to be there and honor her in a really special way with other people that share similar experiences.”

REN Co-PI Barbara Kroner and her daughter Ellie (pictured, right) enjoyed the day!

The 2018 Walk has raised $1,112,028 to date.

Fundraising will continue until May 14th - there is still time to donate!

The money raised from the National Walk for Epilepsy helps the Foundation provide services for people living with epilepsy, provide awareness programs for proper seizure recognition and first aid, provide a voice to make sure health care options for people living with seizures remain strong, and provide much needed research funding toward better treatment options and ultimately cures.

2018 Public Policy Institute and Teens Speak Up! Conference

The Epilepsy Foundation welcomed members of the epilepsy community to Washington, D.C. for its annual signature advocacy event, the Public Policy Institute and Teens Speak Up! conference on April 15-17, 2018.

This year brought together more than 175 teens, parents, local office staff, members of the rare epilepsy community, providers, policy experts, and volunteers. A day and a half of learning and advocacy training culminated with a successful day on Capitol Hill, where families and state advocacy leaders shared their personal stories to raise awareness and advocate in support of federal funding for epilepsy research and programs, in more than 200 Congressional meetings.

Representatives from 32 chapters and affiliates across 32 states participated, and this was the first year that members from the rare epilepsy community participated. The Foundation was thrilled to have Bridge the Gap – SYNGAP, Mickie’s Miracles, The Brain Recovery Project, and the Tuberous Sclerosis Alliance join voices on Capitol Hill. The Advocacy team look forward to having even more members of the epilepsy community join us next year. To learn more, please visit advocacy.epilepsy.com/TSU2018.

- Britt Dorfman, Government Relations & Grassroots Associate, Epilepsy Foundation
CURRENT EPILEPSY STUDIES ON THE EPILEPSY FOUNDATION CLINICAL TRIALS PORTAL

Preventing Epilepsy Using Vigabatrin In Infants With Tuberous Sclerosis Complex

This Phase IIb trial will test whether earlier treatment versus standard treatment with vigabatrin in infants with Tuberous Sclerosis Complex (TSC) will have a positive impact on developmental outcomes at 24 months of age. It also tests whether early treatment prevents or lowers the risk of developing infantile spasms and refractory seizures. It is a randomized, double-blind, placebo-controlled clinical trial design. Infants under the age of 6 months diagnosed with TSC but without history of seizures or infantile spasms may be eligible.

ClinicalTrials.gov Identifier: NCT02849457

If interested, contact:

Jessica Krefting, RN  
(205) 975-2890  
jessicakrefting@uabmc.edu

Regina Ryan, MS  
(205) 975-2890  
reginaryan@uabmc.edu

200 people with treatment-resistant focal epilepsy from around the United States have the opportunity to change the lives of millions by joining in the newest Human Epilepsy Project study on focal seizures. Could you be one of them?

THE STUDY
UNDERSTANDING THE CHALLENGES
This project will look at changes in seizure frequency, treatments used, comorbidities like depression, healthcare costs, and quality of life over time.

NATIONAL EFFORT
Enrolling 200 participants from across the U.S. who have frequent focal seizures that do not respond to medications

ANNUAL CHECK-UPS
Participants complete seizure diaries and other online surveys, along with annual in-person visits to collect biosamples over a two-year period.

Interested? Enroll at info@humanepilepsypoint.org

Learn more at: www.HEPStudy.org
What is This Study About?

This three-month study will look at people between 18-65 years old, taking their own anti-seizure medications in addition to either the study drug (eslicarbazepine acetate or placebo). The aim is to look for changes in seizure, visual fields (the area you can use with each eye) and visually (how clear your vision is) will be measured in people before, during, and after the study.

Who Can Participate?

The study is looking at how effective and safe eslicarbazepine acetate is in people ≥18 years old with focal or partial-onset seizures, when it’s added to their current anti-seizure medication(s).

- ≥18 years old
- Has NEVER taken ESL before
- The study is 31 weeks in duration
- Up to 10 site visits and 3 telephone visits

How Can I Participate?

The first step is to see if the study is right for you at a screening visit with a healthcare provider. A study team member will explain the details of the study with you, if you qualify and are willing, you can give your consent to participate. You can always choose to withdraw from the study at any time. Some or all of the following tests will be done:

- medical history review
- medication review
- physical examination
- neurological examination
- vital signs and weight
- electrocardiogram (heart rhythm test)
- blood tests
- electroencephalography (EEG or brain wave test)
- Computed Tomography (CT SCANS) or Magnetic Resonance Imaging (MRI) or computerized pictures of your brain (if necessary)

Your eyes may also be examined at this time, including dilating your pupils to see the back of the eye.

If you are eligible for this study, you will be randomly assigned by chance (like the flip of a coin) to receive either eslicarbazepine acetate or placebo starting 2 weeks before screening. Blood tests and a physical exam will be done at the screening visit. After screening, you will take the study drug or the placebo, whichever you were assigned, in addition to your usual anti-seizure medications.

During the course of the study, you will have several checkups with your healthcare provider. Some or all of the following will be done at these visits: physical exam, external or internal eye exam, blood and urine sample collection, review of medications, and discussion of seizure frequency.

If you are found to be having any visual effects, you will also have visual testing done at the follow-up visit. Assessment of any visual effects requiring follow-up will be supported by Pfizer.

Who is Eligible to Join the Study?

- People between the ages of 18-65
- Diagnosed with epilepsy with at least one of the following seizure types:
  - simple partial-onset seizures with motor symptoms
  - complex partial-onset seizures with impaired awareness
  - partial-onset seizures that go into generalized seizures
- Has had at least 3 focal-onset seizures in the last 6 months
- Taking a stable dose of levetiracetam or lamotrigine for at least one month before screening, with no history of adjunctive (or add-on) treatment
- Taking a stable dose of 1 or 2 anti-seizure drugs (except oxcarbazepine)

Diagnosis of epilepsy with one or more of the following seizure types:
- simple partial-onset (focal) seizures with motor symptoms
- complex partial-onset (focal) seizures with impaired awareness
- partial-onset (focal) seizures that go into generalized seizures

Are There Risks?

People with Asian ancestry may carry a genetic marker (called allele HLA-B*1502), which may put them at increased risk of serious dermatologic reactions (e.g., Stevens-Johnson Syndrome).

The treatment may not improve a person’s seizures.

The dosing plans and safety checks that are designed to protect participants are detailed in a protocol that was designed by researchers and doctors and reviewed by an Institutional Review Board (IRB). If the participant is unable to tolerate treatment with ESL, the study team will help them to taper off the medication slowly, where possible, unless abrupt discontinuation is necessary.

For more information, visit https://clinicalconnection.com/clinical-trials-from-other-databases/study-location-selection/431534

Learn more & Participate HERE now!
# Enrollment Update

Please feel free to share this newsletter to others with rare epilepsies!

They can [enroll HERE now!](#)

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<th>Syndrome</th>
<th>No.</th>
<th>Syndrome</th>
<th>No.</th>
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<td>Ohtahara Syndrome</td>
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<td>PCDH19 Female Epilepsy</td>
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<td>Batten Disease</td>
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<td>Phelan-McDermid Syndrome</td>
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<td>Progressive Myoclonic Epilepsy (PME)</td>
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<td>SCN2A Mutation</td>
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<td>SCN8A Mutation</td>
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<td>SYNGAP1 Mutation</td>
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<td>Tuberous Sclerosis Complex</td>
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<td>Other mutation associated with seizures</td>
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<td>Mitochondrial Disorders (Alper’s Disease, Leigh’s Disease)</td>
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ABOUT US

To expedite research into the rare epilepsies, 31 rare epilepsy groups have joined forces with the Epilepsy Foundation, Research Triangle Institute and Columbia University to create the first ever Rare Epilepsy Network (REN). With seed funding from the Patient-Centered Outcomes Research Institute (PCORI), the REN is building a patient registry to collect information about rare epilepsy patients to better understand these conditions, improve treatments, and improve the lives and quality of care of patients living with them.

CONTACT US

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By email: ren@efa.org
Visit us on the web at: http://www.epilepsy.com/ren