Patient-Focused Drug Development Meeting
Wednesday, June 21, 2017
8:30 am to 4:30 pm
Hyatt Regency Washington on Capitol Hill
400 New Jersey Avenue, NW
Washington, DC 20001

Morning Session (8:30 am – 12:10pm): Treatments to Prevent the Development of Epilepsy in Infants with TSC

Afternoon Session (1:00 pm – 4:30 pm): Treatments to Prevent Development of Angiomyolipomas and LAM in Young Adults with TSC and/or LAM

The purpose of this meeting is for individuals affected by tuberous sclerosis complex (TSC) and caregivers of dependent adults or children to communicate your perspectives on living with TSC directly to the FDA. You will be able to share the impacts of TSC on individuals’ daily lives, and individuals’ and caregivers’ perspectives on how well available therapies are working.

This meeting is free and open to the public and it will be webcast live online.

For more information and to register, visit www.tsalliance.org/PFDD today.

The 2017 International Research Conference on TSC and LAM: Innovation Through Partnerships
June 22-24, 2017
Hyatt Regency Washington on Capitol Hill
Washington, DC

The conference will be co-hosted by the Tuberous Sclerosis Alliance (TS Alliance) and The LAM Foundation. Through communication and collaboration of participating senior and junior researchers and trainees, the conference goal is to stimulate and accelerate basic and clinical research to improve health care and quality of life for individuals with TSC, LAM, and potentially many other related disorders including epilepsy, autism, cancer, and rare diseases with overlapping phenotypes.
Upcoming Events

**Dup15q Alliance 2017 Science Symposium**
*July 27-28, 2017*
Luskin Conference Center
UCLA, Los Angeles, California
For more information and to register visit [http://bit.ly/2pFa1F9](http://bit.ly/2pFa1F9)

2017 Family Forum
*December 2-3, 2017*
Washington, DC

**Hope for Hypothalamic Hamartomas** (501c3 non profit foundation) is planning a 2017 Family Forum in Washington, DC December 2-3 for patients, caregivers, and families touched by HH. The Forum will begin with up-to-date information about treatment and research. Interactive discussions will focus on the challenges of managing an HH diagnosis including cognition, behavior, endocrine, transitions, alternative therapies, family dynamics and more. For more information, click [here](http://bit.ly/2pFa1F9). Discounted hotel rooms are available but will book fast as Washington DC is a popular destination in the winter. Please register and reserve your hotel rooms ASAP.

SCN8A Epilepsy Research Grant

2017 Request for Applications by: The Cute Syndrome Foundation and Ajudé o Rafa

Together the Cute Syndrome Foundation and Ajudé o Rafa are thrilled to announce our 2017 SCN8A Epilepsy Research Grant Request for Applications. This Request for Applications is an open call for proposals. It is intended both to stimulate new research ideas and approaches and to support existing research that will advance our understanding of SCN8A epilepsy and open new avenues for treatment. We solicit research applications for up to $40,000 that are collaborative in nature in (but not necessarily restricted to) the following major areas:

- Cellular, molecular, and systems-level understanding of underlying pathogenic mechanisms.

- Novel treatments that may impact progression of the disease, with epilepsy as a primary target, and motor skills, behavior, and/or cognitive capacity as secondary targets.

- Effects of SCN8A mutations on cardiac myocytes and cardiac arrhythmias.

- Causation and prevention of SUDEP in those with SCN8A mutations.

**June 1, 2017: Full Proposals Due**
**August 15, 2017: Awards Announced**

For more information, please visit [http://www.thecutesyndrome.com/scn8a-grant-call-for-proposals.html](http://www.thecutesyndrome.com/scn8a-grant-call-for-proposals.html)
The Epilepsy Foundation and Hope for Hypothalamic Hamartomas hosted a special Facebook Live, "When Laughing or Crying is a Seizure." on March 6

Special guests included Lisa Soeby, co-founder and vice president of Hope for Hypothalamic Hamartomas and Dr. Nathan Fountain professor of neurology and director of the F.E. Dreifuss Comprehensive Epilepsy Center at the University of Virginia School of Medicine.

Experts spoke about what it’s like to have HH and what new approaches are available and are on the horizon. The event was very well attended.

The NORSE Institute sponsored the first international symposium on NORSE and FIRES on April 5, 2017, in Salzburg, Austria. Co-chairs were Drs. Rima Nabbout, France and Nicolas Gaspard, Belgium. The purpose of the symposium was two-fold: to present newly-proposed consensus definitions of new onset refractory status epilepticus (NORSE) and febrile illness-related epilepsy syndrome (FIRES) and to bring together experts of both pediatric and adult epilepsy populations, critical care, immunology and genetics to discuss these new consensus definitions, current and future research and NORSE/FIRES registries.

Dr. Lawrence Hirsch (USA) began the process of developing consensus definitions first within the members of the NORSE Institute Medical Advisory board and then sought the opinions of experts across the US and abroad. The objective was to standardize terms for these uncommon severe and vexing presentations of seizures and status epilepticus. These agreed upon terms would facilitate communication, research efforts and web searches for families and caregivers. They form the basis on which to begin integrated research on NORSE.

The proposed definition of NORSE is: New-Onset Refractory Status Epilepticus: A clinical presentation, in a patient without active epilepsy, with new onset of refractory status epilepticus without a clear acute or active structural, toxic or metabolic cause.

- Most of the common acute or active structural, toxic or metabolic etiologies can be identified in the first few hours, but it may take up to 72h to rule out acute strokes, brain masses, drug overdoses, etc.
- Includes viral infections; autoimmune syndromes of new onset, even if clear in first 72h, e.g. classic anti-NMDA encephalitis; and allows remote brain injuries or resolved epilepsy.
- Requires imaging, CSF analysis, toxicology and blood tests, as recommended for evaluation of SE in other guidelines (e.g., Neurocritical Care Society)
- Typically presents as super-refractory status epilepticus (SRSE), but this is not required for the diagnosis of NORSE
- Subgroup: Cryptogenic after extensive workup. Known as “cryptogenic NORSE”.
- Allows prior but resolved epilepsy due to possible coincidence of the two disorders.

The proposed definition of FIRES is: Febrile illness-related epilepsy syndrome: a subcategory of NORSE that requires a prior febrile illness with fever starting between 2 weeks and 24h prior to onset of refractory status epilepticus (RSE).

- Note: has to be refractory SE; no age cutoff; can be infant, child or adult.
- Note: can be with or without fever at the time of onset of SE (about 50% have fever per prior literature)
- Excludes most or all cases of febrile SE as fever in febrile SE is usually acute onset (few hours or less).

Complete definitions of NORSE, FIRES, related syndromes, and status epilepticus of different severities are posted on www.norseinstitute.org. Symposium abstracts will be posted when available.
Brandy Fureman, PhD attended and exhibited at the University of Minnesota sponsored Rare Disease Day on February 24, 2017.

Brandy also attended and exhibited at NIH Rare Disease Day on February 28, 2017.

Cincinnati Children’s: Learning Networks Program

On April 6-7, 2017, the Learning Networks Program (an initiative originating out of Cincinnati Children’s Hospital Medical Center in Ohio) held an Introduction to Design Day and Learning Network Community Conference. Dr. Kathleen Farrell, Senior Program Manager for Clinical Research at the Epilepsy Foundation was in attendance to learn how the Rare Epilepsy Network might benefit from partnering with this community.

The first day of the meeting saw a team led by Dr. Peter Margolis, MD, PhD (Professor of Pediatrics and Co-Director of the Anderson Center for Health Systems Excellence) and Dr. Carole Lannon, MD, MPH (Director, Learning Networks Program, Anderson Center for Health Systems Excellence) welcome nine patient groups from across the medical spectrum to share the mission and method of the Learning Network. The Learning Networks (LN) program focuses on data utilization for improved clinical care and successful research, with an emphasis on quality improvement (QI) and all-stakeholder benefit (patients, families, clinicians and researchers). The advice for groups looking to incorporate as a Learning Network was to be action-oriented and outcomes-focused. Talks from Dr. David Hooper, MD (Division of Nephrology, Cincinnati Children’s) and Dr. Margolis covered the experiences of building IROC (Improving Renal Outcomes Collaborative) and ICN (Improve Care Now, for pediatric inflammatory bowel disease) networks, respectively. The LN utilizes tools including the maturity model and driver diagram to elucidate a groups’ mission, current status, and goal outcomes. Mr. Lloyd Provost, a statistician and author, provided valuable insight into network development strategies and QI processes.

Day two of the meeting gathered several existing LN groups for updates and continued education on how to best run a network. There are approximately 425 teams and 270 sites involved in the Learning Network program, across 43 states and four countries.

Dr. Farrell returned to the Rare Epilepsy Network team enthusiastic to continue to learn more about this program. There is potential for the LN to elevate REN to become a more visible initiative, to help increase participation the current study, and raise awareness among patients and their families, clinicians and researchers, and potential donors for research in rare epilepsies.

A follow up meeting between the REN Primary Investigators and the LN team is scheduled for May 2017.
## Enrollment Update

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To enroll, please visit the [Rare Epilepsy Network](#)
ABOUT US

To expedite research into the rare epilepsies, 26 rare epilepsy foundations have joined forces with the Epilepsy Foundation, Research Triangle Institute, Columbia University and New York 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.