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NIH Rare Disease Day 2019

Thursday, Feb. 28, 2019  8:30 a.m. - 4:00 p.m. ET
Main Auditorium, Natcher Conference Center, Building 45
National Institutes of Health; Bethesda, Maryland

Sponsored by NCATS and the Clinical Center, this year’s event will feature interactive panel discussions on collective research models for rare diseases, patient registries, rare cancer research initiatives, and "no disease left behind, no patient left behind." Other highlights include posters and exhibits by rare disease groups and researchers as well as artwork, videos and CC tours.

Admission is free and open to the public.
*REN representatives will be attending and exhibiting a poster!

Learn more and register here.

TS Alliance Research Grants Program

Letters of intent (LOIs) are due by Monday, April 8 at 11:59 pm ET
Full proposals from invited applicants will be due in August.

Types of grants:
- Postdoctoral Fellowships
- Research Grants

Duration and budget:
Typical duration is two years, but can be shorter if appropriate for the project.
Maximum total costs of $75,000 per year, $150,000 total.
A maximum of 10% of the total costs may be applied to indirect costs
Full details are available at www.tsalliance.org/grants

4th International Symposium on Hypothalamic Hamartomas

Inviting young investigators and new faculty experienced or interested in hypothalamic hamartomas (HH) to attend the 4th International Symposium on Hypothalamic Hamartomas. Hypothalamic Hamartomas is a rare disorder associated with refractory epilepsy, epileptic encephalopathy, intellectual disability, behavioral comorbidity, and endocrinological dysfunction. The Symposium is being presented by Hope for Hypothalamic Hamartomas and Children's National Medical Center (CNMC) in Washington, DC on September 12-14, 2019.

The Symposium will analyze HH as well as other disorders/diseases entities that involve hypothalamic dysfunction and other genetic epileptic encephalopathies also with psychiatric co-morbidities to find commonalities and distinctions to inform investigations and a strategic roadmap. A limited number of scholarships are available for young investigators and new faculty.

For more information and to register, click here.
2019 International Tuberous Sclerosis Complex Research Conference: Changing the Course of TSC

June 20-22, 2019
Chelsea Hotel, Toronto, Ontario, Canada

Abstracts due March 22, 2019

Early-career researchers (including graduate students, postdoctoral fellows, and junior faculty members [non-tenure-track faculty or tenure-track for less than three years]) are eligible to receive travel awards. Abstracts of eligible authors requesting consideration for travel awards will be reviewed by the Organizing Committee, excluding any committee members who may have a conflict of interest. The Organizing Committee will select a limited number of abstracts for receipt of travel awards of USD 1,000 each, depending on available funds.

More information available at tsalliance.org/researchers/research-conferences/

29th Annual Pediatric Neurology Update Course

The Children’s National’s Center for Neuroscience and Behavioral Medicine will host the 29th Annual Pediatric Neurology Update course. This year’s course will feature on three critical areas in pediatric neuroscience and neurodevelopment: epilepsy with focuses on innovations in epilepsy surgery and new therapeutics; tuberous sclerosis including neurosurgical advances and transition to adulthood; and autism spectrum disorder with emphasis on new understandings and pre-requisites for an “Autism Friendly Hospital”.

Internationally renowned speakers will complement talks from Children’s National faculty. This year’s Richmond Paine Lecturer, Dr. Shlomo Shinnar, a renowned epileptologist, will speak on the Consequences of Prolonged Febrile Seizures followed by Dr. William D. Gaillard and others from Children’s National to discuss innovative aspects of epilepsy management. Dr. Chima Olugbo, director of functional neurosurgery at Children’s National will discuss new techniques and indications for epilepsy surgery and Dr. Howard Weiner, this year’s McCullough Lecturer, will present his groundbreaking work on surgery for children with epilepsy and tuberous sclerosis.

Children’s National expanded Tuberous Sclerosis team will then discuss new innovations, including new therapeutic initiatives aided by Dr. Martina Bebin, national principal investigator of the Prevent Trial (sponsored by the TS Alliance) and Dr. Peter Crino, Chairman at the University of Maryland discussing transition of TS care to adulthood.

The final session will cover multiple aspects of care of children with autism spectrum disorder, led by Dr. Lauren Kenworthy and Kevin Pelphrey and others involved in the concerted efforts of Children’s National to create an Autism Friendly Hospital Initiatives to enhance family and patient education (The Get Well Network) and translational care will be presented. A multifaceted round table, encouraging registrant participation, led by Julie Bascom and Dr. Kenworthy, an internally known advocate for children with autism, will close the Update.

Learn more and register here.
The LGS Foundation Research Award provides funding for young and established investigators, physician residents, and clinicians who seek to advance our understanding of Lennox-Gastaut Syndrome (LGS). Grants may be one-year or two-years and support research projects that answer questions related to the underlying biology, clinical aspects, therapies, and/or causes of LGS. Grant recipients are asked to give a presentation at the LGS Foundation’s annual research meeting at the American Epilepsy Society the following year to discuss their work. The LGS Foundation does not require an LOI, however, we will evaluate LOI’s from investigators prior to submission of a full application.

Areas of Interest:

- Causes of LGS and increasing our understanding of the underlying biology
- Evolution of LGS from intractable seizures to Slow Spike & Wave on EEG
- Evolution of tonic seizure (GPFA on EEG) in LGS
- Evolution of multiple seizure types in young children, which may lead to future LGS
- Understanding convulsive/non-convulsive status epilepticus and cluster seizures in LGS
- Therapies to prevent or treat Lennox-Gastaut Syndrome
- Quality of Life for individuals living with LGS and their families
- Epidemiology and better data collection/management of data from individuals of LGS

Budget: Requests may be made for up to $30,000 for one year, or $50,000 for two-year projects. Indirect costs are not supported.

<table>
<thead>
<tr>
<th>2019 FUNDING PROGRAM</th>
<th>Dates</th>
</tr>
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<tbody>
<tr>
<td>Program Opens</td>
<td>February 1, 2019</td>
</tr>
<tr>
<td>Full Applications Due</td>
<td>June 1, 2019</td>
</tr>
<tr>
<td>Award Announcement</td>
<td>July 15, 2019</td>
</tr>
<tr>
<td>Anticipated Project Start Date</td>
<td>September 15, 2019</td>
</tr>
</tbody>
</table>

Application Instructions:
Applications must be submitted by the deadline to Tracy@LGSfoundation.org and must include the following.

1) Title Page: Enter proposal title and PI information.
2) Abstract: Provide a lay summary and a scientific summary of the project.
3) Budget Period Detail: Provide a detailed budget and justification. All expenses must in U.S. dollars.
4) Proposal (NIH format is acceptable)
5) Biosketch for each PI
6) Letters of support or collaboration as relevant

Inquiries: Questions regarding these guidelines are welcome and should be directed to Tracy Dixon-Salazar at Tracy@LGSfoundation.org.
International Epilepsy Day 2019 - February 11th!

Started in 2015 and organized by the International Bureau for Epilepsy (IBE) and the International League Against Epilepsy (ILAE), the day provides a platform for people with epilepsy to share their experiences and stories with a global audience. The day also calls for all people to advocate for appropriate legislation that will guarantee human rights of people with epilepsy and encourages people with epilepsy to live to their fullest potential. The Epilepsy Foundation is a full member of IBE.

Despite being one of the world's oldest known medical conditions, public fear and misunderstanding about epilepsy persists, making many people reluctant to talk about it. That reluctance leads to lives lived in the shadows, discrimination in workplaces and communities, and a lack of funding for new therapies research. For many people living with epilepsy, the misconceptions and discrimination can be more difficult to overcome than the seizures themselves.

International Epilepsy Day seeks to raise awareness and educate the general public on the true facts about epilepsy and the urgent need for improved treatment, better care, and greater investment in research.

Read about the different ways you can take part here!

Rare Disease Day 2019 - February 28th!

Rare Disease Day takes place on the last day of February each year. The main objective of Rare Disease Day is to raise awareness amongst the general public and decision-makers about rare diseases and their impact on patients' lives.

The campaign targets primarily the general public and also seeks to raise awareness amongst policy makers, public authorities, industry representatives, researchers, health professionals and anyone who has a genuine interest in rare diseases.

Building awareness of rare diseases is so important because 1 in 20 people will live with a rare disease at some point in their life. Despite this, there is no cure for the majority of rare diseases and many go undiagnosed. Rare Disease Day improves knowledge amongst the general public of rare diseases while encouraging researchers and decision makers to address the needs of those living with rare diseases.

Visit https://www.rarediseaseday.org to learn more!
Epilepsy can affect anyone with a brain. And anyone with a brain can affect epilepsy.

We’re using our brains to End Epilepsy through Action Together. These are just two examples of how you can join us:

**Create And Share Your Own Brain Art**

Help us spread awareness and understanding of epilepsy by creating your own brain art to End Epilepsy. It’s easy to create and easy to share.

Start >

Learn more about Brain Art and other awareness activities at https://endepilepsy.org

**2019 Walk to End Epilepsy**

The Walk to End Epilepsy is a community celebration of music, entertainment and camaraderie among friends, families, corporations, and organizations gathering to celebrate their fundraising success and demonstrate their support in the fight against epilepsy.

Walkers raise funds for research, programs, services and education.

Take steps to save lives. Register now

Learn more about the Walk and how you can register, volunteer or participate virtually here.
For more information, visit
WHAT
Safety and efficacy of cannabidiol (CBD)

WHO
People aged 1 year to 65 years with Tuberous Sclerosis Complex (TSC)

WHERE
Epilepsy Centers throughout the United States

HOW
16 weeks of treatment with either CBD or a placebo
Followed by the opportunity for all participants to receive CBD for an initial period of 1 year

IMPORTANT CONSIDERATIONS
You must be willing for your primary care doctor and other responsible authorities to be notified of your participation in this study if it is required by law in your state. Your liver function will be monitored throughout the study. If you are pregnant, taking an mTOR inhibitor, or have liver impairment, you are not eligible to participate due to increased risk with CBD.

READ MORE
Cannabidiol study: https://www.epilepsy.com/clinical_trials/cannabidiol-add-therapy-tuberous-sclerosis-complex
Perampanel study: https://www.epilepsy.com/clinical_trials/perampanel-study-infants-epilepsy
REN Future Plans

In mid-2019, REN will transition under umbrella of the Epilepsy Foundation Research program. Next phase plans are in development, with the following pillars:

**Rare Research**
- Disseminate data
- Streamline REN survey modules and re-launch

**Rare Care**
- Connection with the Epilepsy Learning Healthcare System (ELHS) – ensuring all patients are referred appropriately for comprehensive epilepsy care; *See below for more information
- Developing recommendations for rare epilepsy centers of excellence

**Rare Connect**
- In development: A secure online environment for physicians to connect with professional peers and rare epilepsy experts, to democratize knowledge of best practices in diagnosis, treatment and standards of care for patients with a rare epilepsy

*REN as a partner of Epilepsy Learning Healthcare System (ELHS):*

With the support of the Epilepsy Foundation, the National Association for Epilepsy Centers, the Patient Centered Outcomes Research Institute and the Anderson Center for Health Systems Excellence, the ELHS is building a quality improvement and research network dedicated to improving outcomes for children and adults with epilepsy.

By connecting epilepsy centers and including people with epilepsy and their families, the ELHS will empower all people with epilepsy to live their highest quality of life, striving for freedom from seizures and side effects.

The REN is part of the ELHS Community Engagement Core, which at both a center-level and network-level help to determine clinical priorities for pursuit. Representatives from the REN helped to form the ELHS aim above and to design the improvement strategies that will be used to achieve the goal.

ELHS centers learn from every patient at every visit. Data is gathered from ELHS clinics across the country into a registry. The data is analyzed centrally to find best practices that will lead to better outcomes for patients and their families.

*The ELHS Key Driver Diagram (KDD) can be found on the next page.*

A driver diagram is a visual display of a our team’s theory of what “drives,” or contributes to, the achievement of a project aim. This clear picture of a our shared view is a useful tool for communicatoing to a range of stakeholders where a team is testing and working. It shows the relationship between the overall aim of the project, the primary drivers (sometimes called “key drivers”) that contribute directly to achieving the aim, the secondary drivers that are components of the primary drivers, and specific change ideas to test for each secondary driver.

*More to come in the following months!*
Epilepsy Learning Health System: System Level Key Driver Diagram (KDD)

Vision/Global Aim

Our vision is for all people with epilepsy to live their highest quality of life, striving for freedom from seizures and side effects, and we won’t stop until we get there.

Mission/SMART Aim

Design and implement a system of co-production that will improve outcomes for people with epilepsy and their families/caregivers in the following specific ways:

* Improve quality of life from X to Y
* Improve seizure control from X to Y
* Improve seizure freedom from X to Y

* separated by seizure type and epilepsy syndrome

Population

People of all ages diagnosed with epilepsy by a practitioner and in an established case relationship, and their families and caregivers

Key Drivers: “WHAT”

1. **Infrastructure and Culture**
   - Establish a culture of continuous quality improvement
   - Provide a shared platform for knowledge and best practices
   - Establish ongoing engagement and feedback mechanisms

2. **Care Delivery and Improvement**
   - Implement standardized processes for epilepsy care
   - Establish and monitor quality metrics for epilepsy care
   - Provide ongoing education and training for care providers

3. **Collaboration**
   - Engage communities, organizations, and stakeholders in the design and implementation of solutions
   - Establish partnerships for knowledge sharing and collaboration
   - Establish ongoing engagement and feedback mechanisms

4. **Sustainability**
   - Establish a sustainability plan for the KDD
   - Establish a sustainability plan for the KDD
   - Establish a sustainability plan for the KDD

Key Drivers: “HOW”

1. **Patient/Caregiver Activation**
   - Increase the proportion of highly activated patient/caregiver from X to Y by Z

2. **Seizure Frequency/Reduction**
   - Increase the percent of epilepsy visits in which seizure frequency is documented from X to Y by Z

3. **Quality of Life**
   - Increase the percent of patient/caregiver that have met their quality of life goals from X to Y by Z

4. **Referral & Conversion to Comprehensive Epilepsy Care**
   - Increase the number of referrals to comprehensive epilepsy care from X to Y by Z

5. **Epilepsy Program (Quality & Cost-Effectiveness)**
   - Increase the number of epilepsy programs that meet quality and cost-effectiveness criteria from X to Y by Z

6. **Transitions of Care (Quality Care Measures)**
   - Increase the number of patients that transition to epilepsy programs from X to Y by Z

7. **Strategic Plan (Quality Measures)**
   - Increase the proportion of patients that achieve seizure freedom from X to Y by Z

8. **Systemic Changes**
   - Increase the number of systemic changes in epilepsy care from X to Y by Z

9. **Implementation**
   - Increase the number of organizations that implement the KDD from X to Y by Z

10. **Outcomes**
    - Increase the number of outcomes that are achieved from X to Y by Z

11. **Robust and efficient research**
    - Establish a robust research agenda
    - Establish a robust research agenda
    - Establish a robust research agenda
**REN Enrollment is currently closed. The survey will reopen in spring/summer 2019!**

## Enrollment Update

<table>
<thead>
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<th>Syndrome</th>
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<th>Syndrome</th>
<th>No.</th>
</tr>
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<td>Aicardi Syndrome</td>
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<td>Ohtahara Syndrome</td>
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<tr>
<td>Angelman 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>CDKL5 Disorder</td>
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<td>Progressive Myoclonic Epilepsy (PME)</td>
<td>8</td>
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<td>Congenital Bilateral Perisylvian Syndrome</td>
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<td>RAS Pathway Disorders</td>
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<td>CSWS/ESES</td>
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<td>Rasmussen’s Encephalopathy</td>
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<td>Severe Myoclonic Epilepsy of Infancy</td>
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<td>Glut1 Deficiency</td>
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<td>SCN2A Mutation</td>
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<td>Hypothalamic Hamartomas</td>
<td>91</td>
<td>SCN8A Mutation</td>
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<tr>
<td>Infantile spasms/West Syndrome</td>
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<td>SLC13A5 Mutation</td>
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<td>Jeavon’s Syndrome</td>
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<td>Sturge Weber Syndrome</td>
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<tr>
<td>KCNQ2 Mutation</td>
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<td>SYNGAP1 Mutation</td>
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<td>Landau-Kleffner Syndrome</td>
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<td><em>Encephalopathy with seizures and major delay in development</em></td>
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<td>MERFF</td>
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<td><em>Other mutation associated with seizures</em></td>
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<td>Mitochondrial Disorders (Alper’s Disease, Leigh’s Disease)</td>
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<td><strong>TOTAL</strong></td>
<td><strong>1459</strong></td>
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</tbody>
</table>
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

By phone: (888) 886-3745
By email: ren@efa.org
Visit us on the web at: http://www.epilepsy.com/ren