REN Organizations

Aaron’s Ohtahara Foundation
Aicardi Syndrome Foundation
Alternating Hemiplegia of Childhood Foundation
Bridge the Gap SYNGAP
Carson Harris Foundation
International Foundation for CDKL5 Research
Dravet Syndrome Foundation
Dup15q Alliance
Hope for Hypothalamic Hamartomas
Infantile Spasms Community
International Rett Syndrome Foundation
The Jack Pribaz Foundation
KCNQ2 Cure Alliance
Lennox-Gastaut Syndrome Foundation
NORSE Institute
PCDH19 Alliance
Phelan-McDermid Syndrome Foundation
Pitt-Hopkins Research Foundation
RASopathies Network
Ring 14 USA Outreach
Ring 20 Chromosome Alliance
The Cute Syndrome Foundation
TESS Foundation
Tuberous Sclerosis Alliance
Wishes for Elliott

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SAVE THE DATE

71st Annual American Epilepsy Society (AES) meeting

December 1-5, 2017
Washington, DC

(See below for REN Workshop* details!)

2018 Epilepsy Foundation Pipeline Conference and Community Day

Pipeline Conference: 2/22-23/18
Community Day: February 24th
San Francisco, CA

12th National Walk for Epilepsy

April 14, 2018, Washington, DC

2017 RARE Patient Advocacy Summit

September 14-15, 2017 - Irvine, CA

The 2017 RARE Patient Advocacy Summit through Global Genes brings rare advocates from across the globe together to Connect, Collaborate and Activate. This educational conference creates a space for members of the rare disease community to share best practices, create important introductions, and help catalyze powerful collaborations. Attendees share their experience, knowledge, and expertise as conference presenters and/or participants.

Learn more and Register here.

Hope for Hypothalamic Hamartomas 2017 Family Forum

December 2-3, 2017 - Washington, DC

This forum will gather patients, caregivers, and families touched by HH.

There will be 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 and to Register, click here.

REN Workshop* @ AES 2017

The Rare Epilepsy Network (REN) Executive Committee is in the process of framing an agenda for a workshop at the 2017 American Epilepsy Society meeting in Washington, DC, with a tentative date of Thursday, November 30th from 2pm-8pm. We will focus on the topic of Outcome Measures, to incorporate non-seizure outcomes as well as patient-reported outcomes (PRO’s).

Watch this space!

Kathleen Farrell will reach out to all Steering Committee groups as more details become available.
The REN Dashboard is now LIVE!

To maximize the value of the REN homepage for all users, the Research Triangle Institute has launched the Dashboard. This new tool enables users to view data collected via the REN survey and learn about our cohort and their caregivers in variables such as demographics, diagnoses, comorbidities, etc.
NF1 Symposium leaves attendees hopeful

(The Neurofibromatosis Network is a part of the RASopathies Network within REN!)

Author: Sarah Wise

More than 80 people attended the University of Wisconsin's inaugural NF1 Symposium May 3.

"It was a truly fitting way to kick off NF Awareness Month," said Ryan Geier, a member of the NF Board of Directors. The UW Neurofibromatosis Clinic and the patient population it serves has grown considerably over the past five years. In light of that growth, it became clear that a forum was needed in order to provide accurate medical information. "Unfortunately, the NF community often at best gets incorrect information and at worst receives no information at all," Geier said.

The UW NF Clinic and the newly formed NF North Central organization worked together to host this event featuring Dr. David Gutmann from Washington University NF Center in St. Louis, Missouri.

"The NF Network could not be more proud to have participated in UW's inaugural NF Symposium," Geier said. "Our commitment to advocate on behalf of the NF community continues to drive partnerships with the medical community and it is with great pride that our partnership with the University of Wisconsin NF Clinic contributed to serving those who are forever a part of my NF family."

Dr. Neha Patel, director of the University of Wisconsin NF Clinic, worked with Dr. Gutmann and members of NF North Central to pull together a panel of both medical experts and community resources so those affected by NF could actively engage in their own education.

"The number of questions that our NF families asked during the Q&A portion of the event clearly indicate just how much our NF community craves accurate information," Geier said. "Dr. Gutmann's keynote address coupled with a presentation that outlined the advancements of cutting edge research lead by Dr. Dhanansayan Shanmuganaayam at UW left those in attendance with a feeling of hope and excitement for a future which is currently unpredictable for those with NF."

Attendees included physicians with backgrounds in genetics, oncology, and pediatrics as well as representatives from local and national community and charitable organizations, including a representative from Children's Tumor Foundation and Kim Bischoff, executive director of the NF Network.

NF North Central, the newest member organization of the NF Network, is dedicated to serving those in the Dakotas, Minnesota, and Wisconsin. "It was important to NF North Central president Gregg Erickson that this type of event be accessible to anyone and everyone who wanted to attend, so NF North Central agreed to fully fund the event not only this year, but in 2018 as well," Geier said.
TESS Research Foundation Announces 2017 Grant Recipients

TESS Research Foundation is excited to announce the recipients of the foundation’s 2017 research grants.

“TESS received a dozen grant applications from leading researchers in citrate transport, seizure disorders and neuronal development,” said Matthew Bainbridge, PhD, Associate Director of Clinical Genomics Research at Rady Children's Hospital and TESS Research Foundation Scientific Advisory Board Member.

“Applicants hailed from around the world and included world class institutions and drug development start-up companies. Selecting the three awardees from such a high caliber pool of talent was arduous,” continued Bainbridge. “Receiving such a large number of grants in the first ever granting cycle is an outstanding achievement for the foundation.”

Grants in total amount of $215,000 USD were awarded to three specific research projects. TESS Research Foundation is grateful to our donors who helped to make these grants possible, moving critical research about SLC13A5 Deficiency forward. With your continued support, we are working towards a healthier future for children suffering from SLC13A5 Deficiency. Click here to learn more about the recipients.

Infantile Spasms Action Network (ISAN)

Authors: Elizabeth Brasington, Sonya Dumanis & Stephanie Mucha

Infantile spasms (IS) is a rare, but serious form of epilepsy occurring in children, usually under one year of age.

In 2016, the Child Neurology Foundation (CNF) convened two forums that brought together epilepsy advocates and leaders. The group discussed a comprehensive strategy to raise awareness for infantile spasms (IS). In 2017, the group has grown to include over 20 national and international entities and has been dubbed the Infantile Spasms Action Network (ISAN).

On August 8, 2017, the CNF brought together a diverse group of stakeholders including medical professions, industry representatives and patient advocates to discuss the future of infantile spasms advocacy. REN members at the table included the Duq15 Alliance, the Epilepsy Foundation, the LGS Foundation and the Tuberous Sclerosis Alliance. The meeting discussions centered around planning for the 2017 Infantile Spasm Awareness Week as well as laid the groundwork for a more unified approach to infantile spasms advocacy for ISAN and its member organizations.

Stay tuned for an executive summary of the note proceedings, which will be published by CNF in the coming weeks! To receive updates on ISAN, please join the CNF Partners List, where new and ongoing ISAN efforts will be highlighted.
CURRENT RARE EPILEPSY STUDIES ON THE EPILEPSY FOUNDATION CLINICAL TRIALS PORTAL

FAiRE Program: Learn more & Participate HERE now!

Sibling Voices Survey: Learn more & Participate HERE now!

FAiRE Program

FENFLURAMINE ASSESSMENT IN RARE EPILEPSY (FAiRE)

ZX008-1501/1504

The FAiRE program is exploring whether an investigational drug (ZX008) can improve seizure control in children and young adults with Dravet syndrome.

GOAL

ZX008-1501/1504 TRIALS:

Test if ZX008 is a safe and effective treatment for children and adolescents with Dravet syndrome, when added on to their other seizure medications.

OUTCOMES TO BE LOOKED AT:

- A decrease in the number of convulsive (also called tonic clonic) or non-convulsive (for example absence or focal) seizures compared to before the study medication was started
- More days without convulsive seizures
- Less frequent use of rescue medication
- Less frequent hospitalizations specifically to treat seizures

ELIGIBILITY CRITERIA INCLUDE:

- Age 2-18 years (inclusive)
- Documented medical history that supports a clinical diagnosis of Dravet syndrome
- First seizure happened in the person’s first year of life
- Uncontrolled seizures despite current medication

Notes: 1. Study 1501 does not allow patients taking stiripentol to participate. 2. Study 1504 requires patients to be taking stiripentol as part of their current treatment regimen.

PARTICIPATION ENTAILS:

- Continuation of person’s normal background anti-seizure medication(s)
- Treatment with either placebo or study medication
- Use of a seizure diary to record seizure activity and medication details
- Regular health checks: may include electrocardiograms (ECG) (test of heart rhythms), echocardiograms (an ultrasound of the heart), physical examination (including blood pressure and heart rate), and nervous system assessments.
Children’s Hospital of Philadelphia Sleep Study

Thanks to everyone that participated in the CHOP sleep study, which had the goal of developing a new sleep scale for children. Over 125 REN caregivers participated and 109 had complete analyzable data.

Results will be included in the next REN Newsletter but as a preliminary we know that children with a rare epilepsy have the highest prevalence of sleep disorders compared to typical children or those with other chronic health conditions such as autism or asthma.

Pneumonia Study

Using your carefully entered REN data, findings on pneumonia in rare epilepsy have been analyzed. An abstract has been submitted for AES 2017, and a manuscript submitted for publication.

We found strong associations between immune disorders, neuromuscular scoliosis, gross motor delay, aspiration of food into the lungs, G-tube usage, hyperventilation, breathing device while sleeping, and dysphagia with pneumonia and moderate associations with usage of topiramate or sedating AEDs and pneumonia in the rare epilepsies.

This should raise physicians’ concerns about developing pneumonia among patients with rare epilepsies. Given the association of mostly neurologic problems with pneumonia in this population, it is possible that improved efforts to provide multidisciplinary care, or at least an increase in monitoring, may be an effective intervention to lower the risk for pneumonia. Further work is needed to develop protocols for interventions that target the strongest risk factors for pneumonia in the rare epilepsies in order to decrease the occurrence of pneumonia.

Publication details to follow!

Development of the ‘Caregiver Stress and Caregiver Benefit Scales’ for caregivers of children with epilepsy

Caregivers who provide care to children (18 or younger) with epilepsy participated in a REN-collaborated study that developed a short and flexible questionnaire to measure stress and benefits of caring for a child with epilepsy. This questionnaire can be used to study how lives of caregivers of children with epilepsy compare with caregivers of healthy children and children with different health conditions, and how epilepsy treatments impact caregivers. The purpose was to develop a questionnaire that asks about topics meaningful to caregivers of children with epilepsy and can guide clinical practice, policy and research.

With extensive feedback from pediatric neurologists and caregivers of children with epilepsy researchers developed 50 questions about the stress and benefits of caregiving. The questions were tested with a total of 632 caregivers including 126 caregivers of children with epilepsy. Caregivers of children with Down syndrome, Muscular Dystrophy, and a community sample also responded to the questions.

Average age of caregivers of children with epilepsy who responded to the questions was 41 years, 95% were female, 93% were white, 77% were married, 9% had high school education or less, and 36% were employed full time. Caregivers of children with epilepsy reported the worst mental and physical health of all the caregivers involved. Statistical analyses resulted in two questionnaires, one to measure caregiver stress and one to measure caregiving benefit. The stress questionnaire includes 18 questions, such as “How much are your finances strained because of caregiving?” and “How difficult is it for you to get a good night’s sleep because of caregiving?” The benefit questionnaire includes 13 items such as “How much do you feel that caregiving has made you a stronger person?” and “Are you a better advocate for your child/children because of caregiving?”

Additional information about the caregivers and the children they care for was gathered and will be analyzed and published. The UW Caregiver Stress Scale (UW-CSS) and the UW Caregiver Benefit Scale (UW-CBS) have excellent psychometric properties, and will be freely available to researchers and clinicians. For more information or to receive the manuscript that summarizes the feedback by neurologists and parents of children with epilepsy that has been published in an academic journal, contact uwcorr@uw.edu or toll-free at 844-847-7892.
# Enrollment Update

Please feel free to share this newsletter to others with rare epilepsies! They can [enroll HERE now](#)!

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**Please feel free to share this newsletter to others with rare epilepsies! They can [enroll HERE now](#)!**
ABOUT REN

To expedite research into the rare epilepsies, 26 rare epilepsy foundations 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
and https://ren.rti.org/

Top 10 Reasons to Participate in the REN Rare Epilepsy Network

1. Help researchers understand your rare epilepsy
2. Enable faster and more efficient epilepsy research
3. Help researchers identify individuals at risk
4. Find better treatments for your rare epilepsy
5. Improve the quality of care for people living with Lennox-Gastaut Syndrome and other rare epilepsies
6. Help change the way rare diseases are studied
7. Help others obtain an early diagnosis and avoid delayed or misdiagnosis
8. Unlock the cause and lead to the cure for your rare epilepsy
9. Share your story with researchers who really LISTEN
10. Participate in your pajamas!