Inside this Issue:

I. Upcoming events & News.................. p 2-6

II. Active clinical trials and studies.......... p 7

III. About REN / Contact us ................ p 8
The 4th International Lafora Workshop was held in San Diego September 6 – 8, 2018, attended by nearly 100 scientists and clinicians from eight countries as well as 25 family members of Lafora patients.

Lafora research has progressed rapidly since the first Lafora Workshop in June, 2014, organized and funded by Chelsea’s Hope Lafora Research Fund. As a result of bringing together a handful of researchers working on this extremely rare orphan disease, the Lafora Epilepsy Cure Initiative (LECI) was formed and researchers are now working collaboratively under a $9 million NIH grant. There are now two drug development companies (Ionis Pharmaceuticals and Valerion Therapeutics) collaborating in a natural history study to establish clinical benchmarks and biomarkers for disease progression. Enrollment in this study has already begun. The hope is that drugs currently still in preclinical development will be ready to begin clinical trials shortly after the conclusion of the natural history study, which is expected to be completed in two to three years.

One of the major steps forward in understanding the pathophysiology of Lafora Disease has been the realization that it is the build-up of abnormal glycogen (a starch) in brain cells that leads to cellular malfunction and ultimately cell death. It is therefore quite similar to other far more common glycogen storage diseases such as Pompe Disease. Valerion is working on drugs that can deliver amylase, an enzyme that breaks down starch, into cells. One of these is already in Phase 1 clinical trials for Pompe Disease and holds great promise for the treatment of Lafora Disease in the near future. Ionis Pharmaceuticals is working on a different approach - the disruption of glycogen synthesis in brain cells - which has been shown to halt disease progression in mice.

A 5th Workshop will be held in Madrid, and a summary will follow!
Learning Opportunity open to REN:

Find out more and RSVP here
Contact Monica Weldon, Bridge the Gap SYNGAP, with any questions!

Hope for Hypothalamic Hamartomas News

Hope for Hypothalamic Hamartomas Celebrated its 10th Anniversary with the 4th International Research Symposium for HH; inauguration of its Pioneer Research & Advocacy Awards; and the Great Strides 5K to raise HH awareness and funds. It was four non-stop packed days but Hope for HH is pleased to share we had a successful Symposium with 60+ researchers and clinicians from 11 countries and 25 institutes across multiple specialties in Washington DC to help set a new research roadmap for HH and especially the comorbidities severely impacting our patients and their families. Moreover, the Rosenfeld - Rekate Pioneer Awards were presented to Drs. Jeffrey Rosenfeld and Dr. Harold Rekate and advocate Lisa Soeby for their collaboration nearly 20 years ago that transformed HH treatment and care. Their alliance has not only positively impacted hundreds of families around the world but is a demonstration of collaboration across institutions and borders to impact the next generation of surgeons and practitioners. Hope for HH's inaugural honorees are living examples of what happens when people with exceptional skill, pioneering and innovative spirit, and
compassionate hearts partner. To cap off an extraordinary weekend, WUSA9 aired a local story on Ilene Miller and her family [here](#). Stay tuned for videos, summaries, and photos to be available on the Hope for HH website in the coming weeks.

**Rare Epilepsy Landscape Analysis (RELA)**

The RELA survey launched to over 68 rare epilepsy organizations in September, and findings will be discussed at the REN workshop at AES! More information to come soon.

**Rare Epilepsy Network leaders in the community**

“Epilepsy and the family: Caregiver stress and sibling experiences”

Yssa DeWoody (Ring14 USA) and her family, *left*, recently shared their story with the International League Against Epilepsy (ILAE).

[Visit to learn more.](#)
Epilepsy Community Resources

The fifth annual PCORI meeting took place September 18-20. Recordings from the sessions are available [here](#).

**REN members recently attended and presented at the Global Genes Summit, Epilepsy Precision Medicine Conference and Epilepsy Foundation Leadership Meeting!**
Epilepsy Foundation News

Epilepsy Due to Specific Causes

For about half of people diagnosed with epilepsy, a specific cause may be found. Knowing the cause can give valuable information on what to expect over time. Check out our new section describing inherited, genetically acquired, polygenic, structural, metabolic, immune-related and infection causes of epilepsy. Our experts are working on updating and covering the specific causes too, so stay tuned for more. Get started here.

Epilepsy Foundation of America has teamed up with online fundraising platform Omaze to offer fans a once-in-a-lifetime opportunity to win a meet and greet with Lady Gaga and VIP seats to both of her Vegas Residencies — Enigma and Jazz & Piano. The campaign, which kicks off on October 8, 2019, also includes a backstage tour of Enigma and 4-star hotel accommodations and flights to Las Vegas for the winner and a friend. Fans donating as little as $10 to benefit the Epilepsy Foundation get 100 chances to win this unique experience. Proceeds from the campaign support the Epilepsy Foundation’s efforts to END EPILEPSY®, accelerate therapies to stop seizures, find cures and save lives.

Enter now: omaze.com/LadyGaga
Read more: http://bit.ly/2IxEoZ2
Epilepsy Foundation
CLINICAL TRIALS PORTAL

Learn more HERE!

Do you know of another trial or study that should be featured on the Portal?
Email Dr. Kathleen Farrell at kfarrell@efa.org

Other trials/studies currently featured:

• Cannabidiol study: https://www.epilepsy.com/clinical_trials/cannabidiol-add-therapy-tuberous-sclerosis-complex

• Staccato Alprazolam study: https://www.epilepsy.com/double-blind-placebo-controlled-inpatient-dose-ranging-efficacy-study-staccato-alprazolam-stap-001

• Perampanel study: https://www.epilepsy.com/clinical_trials/perampanel-study-infants-epilepsy
Data from the initial REN survey is NOW AVAILABLE for research requests!

View here [https://efa.rexdb.net/](https://efa.rexdb.net/)

To expedite research into the rare epilepsies, 33 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

Epilepsy Foundation 24/7 Helpline
1-800-332-1000

By email: ren@efa.org

Visit us on the web at: [http://www.epilepsy.com/ren](http://www.epilepsy.com/ren)