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Representatives from the 26 rare epilepsy organizations that make up the REN, the REN Professional Advisory Board, and researchers came together in Houston on December 1 for the first ever REN Symposium.

The goal of this workshop was to develop a REN research strategic plan, and implementation strategies for the next 1 and 5 years.

Deborah Hirtz, MD, board member of the Pediatric Epilepsy Research Foundation (PERF), moderated the open discussion on the short (1-2 year) and long-term (3-5 year) priorities of the group for REN’s work over the next several years. The discussion was introduced using the following questions: What are the research questions most urgently needed, including caregiver issues as well as other research questions? What other research questions need to be answered to improve the health of the affected child? Where are the gaps, and what data is needed to fill them? Where are the areas of common interest?

REN PAB member, Zachary Grinspan, MD, gave a talk about “Your Role as REN Ambassador” which focused on: How do we recruit patients and increase enrollment into the REN? How do we increase enrollment for greater demographic diversity in the REN? How do we increase requests from researchers who want to submit questions, obtain data, propose trials, etc.?

Suggestions from the talk included the generation of computable phenotypes for the rare epilepsies, which is underway through a CDC-funded grant to Dr. Grinspan and co-Investigator Dale Hesdorffer, PhD. The phenotypes are expected to be available by 2018.

Gail Farfel, PhD provided insight on “The Relevance of REN to Drug Companies” in a talk focusing on: Do drug companies value the REN registry? If so, why and how would they use REN? What does a drug company consider when working with patient advocacy groups to evaluate the potential for outreach and RCTs? How should REN reach out to other drug or device companies?

The meeting concluded with a thank you to Dr. Jan Buelow, who will be stepping down from her role as Epilepsy Foundation PI for REN on December 31, 2016. Dr. Brandy Fureman of EF will succeed her in this role.

A more detailed summary of the strategic planning priorities that were identified will be available in the REN Newsletter Spring 2017 issue.
PCORnet News

PCORnet hosted a webinar “Best Practice Sharing Session: Community Engagement Using Digital Technologies” on October 19. The webinar provided an overview of digital and social media approaches to engaging patient stakeholders in research networks. Presenters shared information about the strategies and vendors they have used for patient outreach and recruitment. The presentation included utilizing traditional social media (Facebook, Twitter), text message short codes, mobile apps, sharable videos and gifs, and other liberated orbital digital content as well as a summary of lessons learned and promising practices. For more information, visit http://bit.ly/2eUgHtu.

The Patient Centered Outcomes Research Institute’s second annual meeting was held in Washington DC’s National Harbor from November 17-19, 2016. The theme of the meeting was “Changing the Conversation about Health Research.” Drs. Jan Buelow and Brandy Fureman attended on behalf of the Rare Epilepsy Network. The meeting emphasized the goal of making health research more stakeholder-driven and patient-centered. Highlights of the meeting included keynote talks about making patient needs and values central to health research and decision making from Stephanie Buxhoeveden, a woman living with multiple sclerosis who is also a nurse practitioner caring for people with neurological disorders, and on building a culture of health and finding common ground among stakeholders from Robert Wood Johnson Foundation President and Chief Executive Officer Risa Lavizzo-Mourey. REN’s own Megan O’Boyle of the Phelan-McDermid Syndrome Foundation gave insightful lessons learned about building a successful international patient registry, which also helped the PMS Foundation secure PCORI funding for the Phelan-McDermid Syndrome Data Network. Video from many of the keynote presentations and Megan O’Boyle’s meeting blog is available to view at http://www.pcori.org/events/2016/2016-pcori-annual-meeting.

5th Global Symposium on Ketogenic Diet Therapies

The 5th Global Symposium on Ketogenic Diet Therapies was held September 20-24, 2016 in Banff, Alberta, Canada. The conference was designed to raise the level of science regarding ketogenic diet therapies and to promote collaborations among scientific and medical professional, parents, and people living with epilepsy. See review by Dr. Eric Kossoff for more details http://bit.ly/2eUjWNs.
3rd International Symposium on Hypothalamic Hamartomas

On September 16, Hope for Hypothalamic Hamartomas (Hope for HH) collaborated with Hope for HH UK and the University College of London (UCL) to bring together more than 60 researchers from around the world to set a strategic roadmap for HH.

Two exciting outcomes emerged from the meeting:
1. plans to explore the development of a HH tissue consortium (biobank) - researchers need tissue from surgeries so they can understand how HH develops and how to best treat HH
2. an international study to better assess the pros and cons of each of the surgical interventions.

For a video recap of the meeting by Chair Dr. Helen Cross, click HERE.

On September 17, nine HH experts presented on best practices in HH diagnosis, treatment, management and care.

Each of the nine PowerPoint presentations can be accessed on Hope for HH's You Tube Channel HERE. Each presentation is available in FULL (long versions) or you can watch a QUICK INTERVIEWS for the key points.

This is the most up-to-date research on hot topics including laser ablation, imaging (FMRI) in diagnosing and targeting HH, behavioral and cognitive side effects of HH and more!

The Epilepsy Foundation was a proud co-sponsor of the 3rd International Symposium on Hypothalamic Hamartomas.

To learn more about activities of Hope for HH, sign up for our newsletter: http://www.hopeforhh.org/get-involved-3/

Read the full story here: http://conta.cc/2eqtUcn
TESS Research Foundation 1st Family and Professional Conference

TESS Research Foundation had a small but productive 1st Family and Professional Conference in Houston in December 2016. We brought together clinical and scientific teams from Stanford Children’s Health, Baylor College of Medicine, Rady Children’s Hospital, and University of Calgary to discuss SLC13A5 Deficiency (Citrate Transporter Disorder) and our path toward a cure.

We are currently funding 4 ongoing research projects, and we have partnered with AES for Early Career Funding.

The phenotype of this disease is consistent and unique:
• Seizures begin within a few days of life
• Persistent seizures of multiple types, resistant to medications
• Severe difficulty with speech production, with better receptive language
• Movement and coordination problems (low tone and unsteady)
• Episodes of body stiffening or weakness
• Brain MRI that appears normal or has subtle changes in the white matter
• Tooth enamel abnormalities
• Mildly elevated Citrate levels in blood samples

At this time, there is NOT a cure for the disease. Inheritance is autosomal recessive.

Our top 5 goals for 2017 include:
1. Continue to create the best animal and cellular models of SLC13A5 Deficiency
2. Better understand the mechanism of this disease and why a lack of citrate transport leads to epilepsy and the other neurologic symptoms
3. Screen currently available medications through our disease models
4. Help more families receive an accurate diagnosis and join our registry
5. Organize our disease community for a clinical trial

For more information http://tessresearch.org/
First International SYNGAP1 Conference

Bridge the Gap – SYNGAP Education and Research Foundation were excited to have held the world’s First International SYNGAP1 Conference, November 30 – December 1, 2016 at Texas Children’s Woman’s Pavilion in Houston, Texas. In partnership with Texas Children’s Hospital, Baylor College of Medicine and The Scripps Research Institute Florida brought all stakeholders together in one room.

There was an international representation of the world’s most renowned neurologists and experts on brain development and brain disorders as presenters. In attendance we had over 25 families from 10 different countries, scientists and clinicians.

The First International SYNGAP1 Conference has begun the process that will enable us to close the gap of time between discovery research and clinical impact. This new approach that brings together multi-disciplinary groups across the globe and also engages the patient families’ world-wide as equal stakeholders is needed to accelerate the process especially for rare disease.

The aims of this conference were to unite the international SYNGAP1 clinicians and researchers to foster collaborative approaches to identify the molecular underpinnings of the disease and potential new avenues of treatment. Secondly, to support junior investigators through meeting participation and provide opportunities to build networks of collaboration with KOLs within and out of their particular research areas. Thirdly, to unite as many patient families that can participate to learn about SYNGAP1 (MRD5), what resources are available, for support and most importantly engage them as partners driving solutions forward. Lastly, to understand the clinical phenotypic diversity of the children amongst researchers and clinicians can provide insights into the strategies pursued.

During the first day a special session was planned specifically for the clinicians and researchers to meet the patient families’ one on one. This is probably one of the most important aspects of our meeting as it will help clinicians drive earlier diagnosis and researchers with a glimpse of the clinical phenotype that can be critical in building translational models that can be used to understand the molecular pathways relevant to this disease. The Foundation also awarded and recognized their founding Scientific Advisory Board Members for beginning the journey to find answers.

For more information visit: http://www.bridgesyngap.org/
Wishes for Elliott Hosts a Second Successful SCN8A Scientific Conference

On December 1st, just in advance of the 2016 AES conference, Wishes for Elliott hosted a second scientific conference of SCN8A experts from around the globe. While a tiny organization (less than 3 years old and run entirely by Elliott’s family all engaged in his direct care), hosting this event has optimized our impact, far beyond our very limited resources.

Attendance more than doubled from our first meeting a year and a half ago. Scientists openly shared the progress and new directions on their ongoing work. The community of experts identified “outside-the-box” strategies for accelerating the pace of progress – to hasten the day when children like Elliott have access to new and effective treatments. Moreover, there was widespread interest in continuing to work together to further develop and implement the best strategies for jump-starting new avenues for research.

Several factors contributed to the success of the event:

- We engaged a child neurologist with a strong interest in research from Children’s National Medical Center to help plan and organize the meeting.
- We enlisted a leading clinician and bench researcher to serve as Co-Chairs of both our first and second meeting (Dr. Miriam Meisler and Dr. Ingrid Scheffer).
- We worked with these partners to set the stage for an intimate gathering where scientists would openly share directions and issues with their ongoing work as well as an explicit focus on identifying and probing strategies for accelerating the pace of progress of SCN8A research in bringing answers to our families.
- The objective of the meeting was to engage the scientists in a frank “outside-the-box discussion exploring opportunities for accelerating research.
- The identified strategies provide an excellent blueprint for our own priorities for the coming year – supporting and fostering efforts to implement the identified “acceleration strategies”.

http://www.wishesforelliott.org/scn8a-conference-2016
www.Facebook.com/WishesforElliott
For more information, contact: JayEtta at wishesforelliott@gmail.com
The 2017 International Research Conference on TSC and LAM: **Innovation Through Partnerships** will be held at the Hyatt Regency Washington on Capitol Hill in Washington, DC on June 22-24, 2017. 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.
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.

Top 10 Reasons to Participate in the 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 others obtain an early diagnosis and avoid delayed or misdiagnosis
7. Help change the way rare diseases are studied
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!