

REN NEWSLETTER

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Leadership Corner

Janice M. Buelow, RN, PhD, FAAN



Welcome to our first monthly newsletter. It has been my pleasure to serve as Principal Investigator of the Rare Epilepsy Network for almost 2 years. First I want to thank all of you who have joined our data base family. This database is by and for people with a rare epilepsy and their caregivers.

Since beginning our journey our robust database was developed and launched and we have enrolled more than 1000 participants representing 22 rare epilepsies. The data-base compiles important information about rare epilepsies on a large scale making it unique amongst data bases. Further our data-base was and continues to be patient/caregiver driven and managed.

Our goal is to attract researchers such as those who are developing new treatments for epilepsy. We hope to also look at problems that cross not only all rare epilepsies but also other chronic pediatric conditions to develop solutions. We continue to look to the caregivers of those with rare epilepsies as well as people with a rare epilepsy diagnosis to suggest research questions that are important to them. We work continuously with the larger PCORnet (of which REN is a part)

FYI—For Your Information

The U.S. Food and Drug Administration's (FDA) Office of Orphan Products Development (OOPD) announced the launch of a new Orphan Products Natural History Grants program. This new program will provide research grants to fund natural history studies to better inform and support drug development and approval for rare diseases. The FDA's press release is available [here](#). To read more about this new program, including information about funding levels, please visit the FDA's [website](#).

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REN ORGANIZATIONS

[Aaron's Ohtahara Foundation](#)

[Aicardi Syndrome Foundation](#)

[Bridge the Gap SYNGAP](#)

[Carson Harris Foundation](#)

[International Foundation for CDKL5 Research](#)

[Doose Syndrome Epilepsy Alliance](#)

[Dravet Syndrome Foundation](#)

[Dup15q Alliance](#)

[Hope for Hypothalamic Hamartoma](#)

[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](#)

WHAT'S NEW?

Research Project Summary

The Rare Epilepsy Network (REN) PPRN is an initiative created by and for patients with catastrophic rare epilepsies. The REN's goal is to build a patient-centered and -driven database designed to provide the patients and their families an opportunity to participate in research that will improve the lives and quality of care for people with rare epilepsies.

Our PPRN is led by the Epilepsy Foundation (EF), a patient advocacy organization dedicated to the welfare of the almost 3 million people with epilepsy living in the United States.

PCORnet Holds Meeting on Trustworthiness

Jan Buelow and Christina SanInocencio attended the Patient Centered Outcomes Research Network Trustworthiness Meeting on March 28 -29th in Washington, DC. This exciting meeting was meant to stimulate discussion about how to engage patients and encourage trust in the research. Christina and Jan realized that we had work to do to help engage patients and increase trust in research for REN. Some ideas that were generated are as follows.

1. Increase diversity
 - a. Perhaps translating the questions to Spanish, building a mobile app, and helping those who may have trouble completing the questionnaire.
 - b. Engage "foot soldiers" to go into hospitals to discuss REN with physicians and where possible patients.
2. Provide return data more frequently so patients can see what their data is doing.
3. Make sure that those enrolled know the PI's.
4. Finally, what sets PCORnet apart is that it is patient driven and that we care about patients. So we need to build ways to help support patients beyond data.



Jan Buelow and Christina SanInocencio

2016 PAME Conference

Join us for this multi-day learning event, the only meeting focused exclusively on increasing understanding of mortality in epilepsy, including Sudden Unexpected Death In Epilepsy (SUDEP). This program brings together clinical and basic science professionals, patients and their families to advance research, facilitate collaborations, engage patients to participate in critical research, and increase urgency around an overlooked and underfunded public health issue, while building understanding and support for each other <http://pame.aesnet.org/>

- **LEARN** - Identify trends in incidence and mortality, gaps in the knowledge base and progress made in the latest research
- **SHARE** - Come together with patients, families, clinicians, researchers, advocates and policymakers for greater insights and collaboration
- **ACT** - Know risk factors and prevention strategies, identify future research opportunities and strengthen methods for building public awareness.

This conference has been conceived and planned by a large and diverse group of professionals, advocates and family members. Major contributing organizations include the American Epilepsy Society (AES), Citizens United for Research in Epilepsy (CURE), Epilepsy Foundation (EF), the Danny Did Foundation, and the Patrick Ring Foundation. [Please check here](#) for the most current list of all partners or if your organization is interested in becoming a partner. This year's event will be held June 23-26, 2016 at The Westin Alexandria in Alexandria, VA.

[REGISTER NOW](#)

Member News



Giving Hope. Getting Answers.



PCORnet News

Who is PCORnet? PCORnet was funded by the Patient Centered Research Institute to build large and small systems of databases that are patient centered, patient driven, and that will not only encourage important research but also provide a mechanism to make research easier. PCORnet is made up of 13 Clinical Data Research Networks (CDRN) and 31 Patient Powered Research Networks (PPRN). The CDRNs are all very large system data-bases that encompasses multiple health systems and use primarily electronic health record data. The PPRNs are patient centered and much smaller. The PPRN's collect their own data but where possible include electronic Health records. The Rare Epilepsy Network is a PPRN. All of these systems can interact to increase the power of each network. In the next newsletters we will describe some of the important collaborations that happening within PCORnet.

For many families it began as a peculiar giggle or smirk – some as early as birth. Frequently, it wasn't until the kids were toddlers, that parents realized there was something terribly wrong with their child.

These kids are the 1/200,000 that are diagnosed with a hypothalamic hamartoma - a rare benign tumor-like malformation that can cause treatment-resistant epilepsy, developmental and cognitive deficits, severe learning disabilities, memory challenges, behavioral problems, rage attacks, endocrine disturbances, and central precocious (early) puberty. Typically, the lesion is not detected on EEG; missed on brain imaging; and misdiagnosed as a gastrointestinal, psychiatric or behavioral condition. Most patients are diagnosed in childhood, but some patients are diagnosed much later.

For some - surgery, including a newly developed laser, can be curative. Still, not all patients are good surgical candidates, have access to surgery, or have the ideal outcome and therefore have to manage the seizures and side effects for life.

There is Hope! [Hope for Hypothalamic Hamartomas](#) is a 501c3 that provides accurate information, education, support and research to families and health care providers.

Hope for HH also joined the [Rare Epilepsy Network](#) – a registry dedicated to gathering information about all patients diagnosed with this HH and other rare epilepsies to improve our understanding and expedite a cure!



ABOUT US

To expedite research into the rare epilepsies, 22 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.

Contact Us

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Visit us on the web at :

<http://www.epilepsy.com/ren>

<https://ren.rti.org/>

Top 10 Reasons to Participate in the



- 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! 