SCN2A RELATED AUTISM & EPILEPSY

FAMILIESCN2A FOUNDATION

WWW.SCN2A.ORG

SAVE THE DATE for our 1st Virtual Family Meeting: October 6, 2018

In preparation for the SCN2A Virtual Family Meeting this October, we are hoping to have all families completely through the Simons VIP registration process by **August 6th**. Even if you are not planning to join the virtual meeting, your family's data is important and valuable to compile for researchers. If you haven't registered yet, please do so now by clicking "Let's Get Started" below. If you have not completed all of your surveys, please contact Simons VIP to get them completed as soon as possible.

Let's Get Started

More About Simons VIP:

They will launch a new platform in early September where all surveys and documents will have the option for translation into Italian, French, Dutch, and Spanish! Other improvements will include a much more robust seizure survey which was created in partnership with REN (Rare Epilepsy Network), graphics to increase ease in completion, and additional surveys to collect data on trends we have noticed in SCN2A.

Simons VIP is the **official**, central SCN2A registry. The FamilieSCN2A Foundation partnered with Simons years ago to collect longitudinal data that defines the disorder. Simons makes collected de-identifiable data available to qualified researchers around the world to expedite finding treatments and a cure. The more researchers looking at this disorder from multiple angles the better. Today they have collected valuable data from close to 150 families. Is your family included?

Trends in
Neurosciences
Volume 41 Number 7
July 2018
ISSN 0166-2236

SCN2A in Neurodevelopmental Disorders

CellPress
REVIEWS

The FamilieSCN2A Foundation is very excited to announce that some children with SCN2A and their siblings had artwork featured on the July 2018 issue of *Trends in Neurosciences* magazine. Here is a a quick overview of the article:

SCN2A encodes the neuronal sodium channel NaV1.2. Dysfunction in this channel contributes to an array of neurodevelopmental disorders, including autism spectrum disorder, intellectual disability, and epilepsy. In this issue, Sanders and colleagues highlight recent advances in our understanding of how changes in the function of this single channel contribute to its associated disorders. The authors also discuss existing treatments and future therapeutic paths that will hopefully benefit affected individuals, their caregivers, and families.

Collage courtesy of Rich Morganstern, Art Director at MediaGarden.co. Individual panels contributed by children with *SCN2A* variants—including Benjamin, Bert, Colin, Eliana, Emily, Gianna, and Joel—and their siblings—including Mark's sister Jillian and Sienna's brother Jamie. Artist age ranges from 3-13 years.

Full pdf of this article will be available soon on the web site: www.scn2a.org

Genetic Epilepsy Team Australia's Family Conference on Genetic Epilepsy in Melbourne, Australia on May 26th was a great success. There were over 100 attendees present to hear talks on genetic epilepsy from internationally renowned experts, and social media about the conference reached over 200,000 people around the world. Speakers included Professors Ingrid Scheffer, Steven Petrou, Annapurna Poduri and Gavin Rumbaugh. Various family groups were also represented by international delegates Monica Weldon of Bridge the Gap - SynGap and Scotty Sims of KCNQ2 Cure Alliance. The entire conference was live-streamed around the world and recorded thanks to the generous support of the FamilieSCN2A Foundation. This allowed people from Europe, North America, South America, Asia and Australasia to attend and participate in the conference. Details about the conference and videos of all the presentations are available at: geneticepilepsyteam.com.au/conference-2018/

Do you shop on Amazon.com? Did you know that shopping through AmazonSmile is the same but offers a percentage of each sale back to your non-profit of choice?

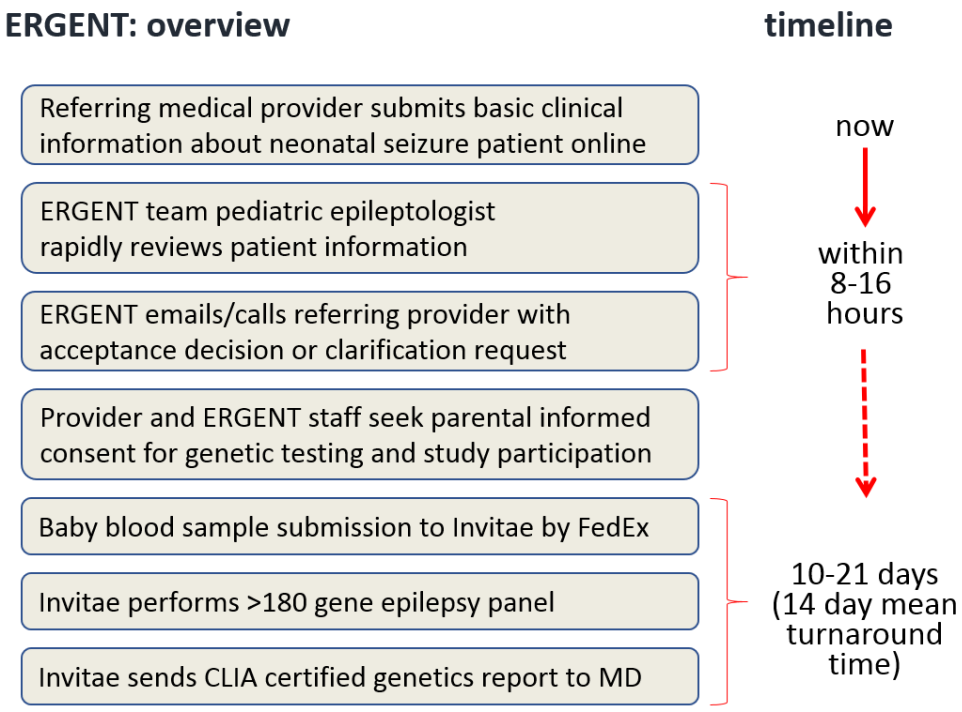
The AmazonSmile Foundation has donated over $150 so far this year as a result of AmazonSmile program activity. Your shopping matters!

Add AmazonSmile to your saved bookmarks
http://smile.amazon.com/ch/47-3169795 and start giving back today!

Early Recognition of Genetic Epilepy in Neonates (ERGENT) Study is now accepting patients across the United States!

One of the pillars of our mission statement is to aid in the early detection of SCN2A. Genetic testing is the only way know if SCN2A is the cause of a child's seizures. Knowing the cause can inform the choice of treatment and may profoundly affect the course of a child's development. For many reasons, most

genetic testing today is delayed until after a treatment has begun, often after a child has left the hospital. We are helping to change that. The FamilieSCN2A Foundation has recently contributed funding to a study coordinated by Dr. Ed Cooper of Baylor University to provide early genetic testing for newborns. This project hopes to not only quickly identify infants with genetic causes of epilepsy, but to also demonstrate the efficacy of early testing and influence the standard of care going forward.



Learn More

Join the FamilieSCN2A Birthday Club!

Who doesn't enjoy a special shout out on their birthday? We would love to honor your child or children affected by SCN2A by having them be a part of our

special Birthday Club. It's super easy to sign up today.

Sign Up Now!

Our mailing address is:
FamilieSCN2A Foundation, P.O. Box 82, East Longmeadow, MA 01028