THECUTESYNDROME



Daniel Braz of Ajude o Rafa - SCN8A Epilepsia speaks with Brazilian researchers at SCN8A Clinician, Researcher, and Family Gathering

Photo credit: **Ryan Collerd**

ANNUALREPORT

November 2016

A word from Hillary Savoie

on The Cute Syndrome Foundation's third year

This year has been one of a great deal of excitement and change at The Cute Syndrome Foundation. Last year, we expanded our efforts to in supporting the SCN8A Community and this year we continued to work on SCN8A exclusively. Juliann Bradish has joined me as a partner in running The Cute Syndrome Foundation and heading up all of our SCN8A projects as TCSF's SCN8A Advisor. Juliann is an amazing asset to the SCN8A community and our team here at The Cute Syndrome.

Juliann has run an SCN8A Families Support Group on Facebook since 2014, soon after her daughter Adeline was diagnosed with an SCN8A mutation in 2013. The support group now includes 125 parents and caregivers of children with SCN8A. Juliann has brought the SCN8A Families Support Group under TCSF's umbrella, where the group is now administrated with the help of four parent volunteers: Sandy Bush, Karen Varner, Shelley Frappier, and Maggie Nayyer. They join our other volunteers: Dianely Cabrera and Mackenzie Wardrope, our SCN8A Outreach team, and Jessica Jenkins, our graphic designer.

This year we awarded our 2016 grant with our partner organization in Brazil, Ajude o Rafa - SCN8A Epilepsia to Dr. Miriam Meisler for her project: *Functional spectrum of SCN8A mutations*. We have also seen the work of our 2015 co-funded grant with Wishes for Elliott to Dr. Michael Hammer yield movement forward with his rolling our of SCN8A.net, an online SCN8A registry and a vital source of information on SCN8A for clinicians and families. The Cute Syndrome, with the guidance of Dr. Phillip Pearl and Dr. Mandy Harris, produced an SCN8A Clinician Guide to help share the information our SCN8A clinician community has gathered regarding the best practices for treating patients with SCN8A.

Next month we will hold our Second Annual SCN8A Clinician, Researcher, and Family Gathering in Houston, Texas in conjunction with the annual American Epilepsy Society Conference, which has been made possible in part by our generous sponsors: The Rare Epilepsy Network (REN), Xenon Pharmaceuticals, and joint-sponsors Upsher-Smith Laboratories and Pairnomix. Last year's meeting, in Philadelphia, PA, was a tremendous success, bringing together nearly 100 guests-including seven children with SCN8A, 35 family members, and 50 professionals to discuss the needs of the SCN8A community. We are grateful to photographer Ryan Collerd, who donated his time to help us capture beautiful memories of the event.

Finally, we are tremendously grateful for the continued generosity of our donors, whose has helped us make all of these big things happen!

-Hillary Savoie, PhD
Founder and Director of The Cute Syndrome Foundation
www.thecutesyndrome.com

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SCN8AEPILEPSY

The SCN8A Clinician, Researcher & Family Gathering

Last December The Cute Syndrome Foundation hosted the first annual SCN8A Epilepsy Clinician, Researcher, and Family Gathering in Philadelphia, PA. Nearly 100 guests-including seven children with SCN8A, 35 family members, and 50 professionals-met for the inaugural event.The gathering served as a venue for sharing clinical information about the effective treatment of SCN8A epilepsy as well as highlights of ongoing research efforts. And for the first time, the SCN8A community was able to meet with the very professionals who dedicate your professional time to helping our children! This event allowed families the opportunity to share their stories and in witnessing your dedication to this community, they regained hope where it was once lost. We look forward to another ground-breaking gathering this year in Houston!







From left to right: **Jessica Jenkins, and her late son Dustin, Dianely Cabrera, and her son Max, and Mackenzie Wardrope, all TCSF Volunteers** at the first annual SCN8A Epilepsy Clinician, Researcher, and Family Gathering in Philadelphia, PA (December 2015) Photo credit: **Ryan Collerd**

Dr. Meisler Awarded 2016 SCN8A Epilepsy Grant

In January 2016, as a collaborative effort with our Brazilian partners, Ajude o Rafa, The Cute Syndrome Foundation awarded $25,000 the 2016 SCN8A Epilepsy Research Grant to Dr Miriam Meisler of The University of Michigan.

Dr. Meisler said of her work:
"The discovery that mutations of SCN8A are responsible for many cases of pediatric encephalopathy was a very important step forward towards identification of effective treatment. Unfortunately, many patients do not respond well to existing medications. In order to move forward, we are working on two experimental systems to identify the functional effects of different mutations and to test the effectiveness of new treatments. First, mutant channels are tested in cultured neuron-derived cells to find out which aspect of channel function is abnormal. Second, we generate mouse models of selected mutations representing the three functional classes observed thus far, to find out which neurons are most susceptible and to provide a pre-clinical test system for novel treatments. Both of these research directions will contribute to development of the next generation of treatment for children with these devastating genetic disorders caused by mutations of SCN8A."

SCN8A Interactive Website and Registry

We are excited to announce the launch of the SCN8A Interactive Website and Registry (www.scn8a.net). Supported in part by the donations made by the Cute Syndrome Foundation and Wishes for Elliott, this registry will enable the collection of more complete data about the manifestations of SCN8A and provide the basis for further research into this devastating disorder. By taking take part in the 8A Registry, you will learn of others who share the same mutation and clinical features as your family member. Learn more about the SCN8A registry here (http://www.scn8a.net/registry).

SCN8AEPILEPSY

SCN8A registry and other projects

One of the most essential tools in pushing scientific research on a rare disorder forward, is an accurate accounting of how many people have the disorder as well as their particular mutations, symptoms, response to treatment, etc. Having such an accurate accounting is so important as a foundation for scientific research, clinical understanding, and drug treatment trials.

SCN8A Family Meting:

Esmé and Hillary Savoie, Dr. Michael Hammer, and Adeline, Juliann, and Josh Bradish

Elliott, Esmé, and Kaitlyn

Photo Credits: Anne Scherrer





Therefore, together with our SCN8A partners, Help Adeline Find Answers and Wishes for Elliott, we raised $20,000 to fund an SCN8A research grant, which we are awarding to support the groundbreaking registry and interactive SCN8A website being developed at the University of Arizona under the dedicated efforts of Dr. Michael Hammer. The goal of the project is to make available important information about the clinical features, causes, and treatments of SCN8A epileptic encephalopathy to families, doctors, and researchers. The website will house and curate information on clinical variability, genetic variants, anti-epileptic drugs, and current research findings associated with SCN8A epileptic encephalopathy.

Orienting to our new SCN8A mission

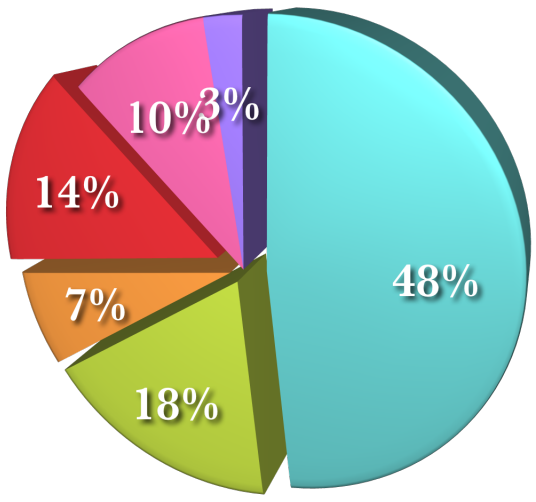
SCN8A Epilepsy is an emerging genetic diagnosis—with only 100 cases diagnosed so far. Like PCDH19 Epilepsy, these numbers are expected to grow exponentially as access to diagnosis is improved. We have connected with Juliann Bradish whose daughter Adeline has SCN8A Epilepsy. Juliann runs an SCN8A parent group and Facebook page called SCN8A-Help Adeline Find Answers, and has been helping researchers collect important data on SCN8A mutations, drug responses, and symptoms. We have also connected with the foundation Wishes for Elliott, which sponsored a wonderful SCN8A conference in Washington DC in April. We attended and we re thrilled to be involved in the wonderful discussions with SCN8A researchers.

We have formed a initiative with our SCN8A partners, Help Adeline Find Answers and Wishes for Elliott, called SPEAK — SCN8A Partnership for Epilepsy Awareness and Knowledge—in order to fund the SCN8A Epilepsy Registry grant with the help of a matching fund from Wishes for Elliott.

We are thrilled that Juliann Bradish and another parent from the SCN8A community are be joining TSCF as SCN8A Adivsors to help drive our future SCN8A projects.

Fund Allocation

- PCDH19 Competative Grant
- PCDH19 Zebrafish
- PCDH19 University of Michigan IPSC
- SCN8A Registry
- Awareness and Education Projects
- Administration costs



We would like to extend our thanks!

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