THECUTESYNDROME



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ANNUALREPORT

July 2015

A word from Hillary Savoie

On The Cute Syndrome Foundation's second year

The Cute Syndrome Foundation has had another successful year. PCDH19 Epilepsy research has made some wonderful advances in the last year due to the efforts of the Cute Syndrome Foundation and our partner organizations. We look forward to updates on our efforts to support important PCDH19 research, contributing to raising awareness of PCDH19 Epilepsy, and supporting the current clinical drug trials within the PCDH19 community.

The Cute Syndrome Foundation has also expanded its efforts to help support awareness of and research for a second rare genetic disorder: SCN8A Epilepsy. SCN8A Epilepsy causes epilepsy, severe developmental delay, and movement disorders. It is a relatively newly discovered disorder and has been documented in approximately 100 cases to date. We have brought on several new staff to help support our growing mission: Lourdes Smith, our PCDH19 Advisor, and Juliann Bradish and Mackenzie Wardrope, our SCN8A Advisors. We are thankful for the skills and hard work they bring to TCSF.

In the coming year we will be helping to fund an SCN8A patient registry, starting an SCN8A Awareness campaign and coordinating with our international partners to continue to support PCDH19 research.

We are tremendously grateful for the continued generosity of our donors. Your support has helped us make big things happen, and we look forward to providing you updates on all of the research projects we have funded, the awareness initiatives we support, and our efforts to strengthen our partnerships with other organizations.

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

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PCDH19EPILEPSY

The first competitive PCDH19 research grant

In last year's annual report we announced that, together with our partners in Italy, Insieme per la Ricerca PCDH19 (Together for PCDH19 Research), we were granting a two-year $135,000 award to Dr. Maria Passafaro and Dr. Elena Battaglioli at the CNR Institute of Neuroscience Milan, Italy for their PCDH19-related research. Less than one-year into this research, they are discovering important scientific insight into the role of PCDH19 mutations and brain function—which they will present this fall at the Insieme per la Ricerca PCDH19 conference in Rome. We look forward to seeing what more they will discover over the remaining year of this grant.





Paola of Insieme per la Ricerca and Hillary Meeting in San Francisco for a conference (September 2014)

Hillary and Esmé at Boston Children's Hospital Zebrafish Lab (April 2014)



Other PCDH19 Projects

It is important to investigate a variety of approaches to understanding how a particular genetic mutation leads to a disease. For this reason, in addition to our major research grant, we are also helping to fund a number of seed grants to help bolster other promising ongoing PCDH19 research:

Dr. Poduri and PCDH19 Zebrafish at Boston Children's Hospital

Last year the Cute Syndrome awarded a $25,000 research grant to Dr. Annapurna Poduri at Boston Children's Hospital in order to help support her work to study the effects of various drug-therapies on zebrafish with PCDH19 mutations. Dr. Poduri's research has shown such promise that she has now been awarded the Richard A. and Susan F. Smith President's Innovation Fund Grant to continue this research. In 2015 we have continued to support Dr. Poduri's research by awarding a grant to her Post-Doctoral Fellow, Christelle Achkar, who is working to better understand the phenotypic spectrum associated with PCDH19.

Dr. Parent and IPS Cells at University of Michigan

With our partners in Italy, we awarded a $20,000 grant to Dr. Jack Parent of the University of Michigan to support his PCDH19 research. Dr. Parent's research uses two cutting-edge scientific approaches, induced pluripotent stem (iPS) cell and sophisticated gene-editing methods, in order to create models to better understand how PCDH19 mutations change brain function and to provide platforms to screen for new therapies to treat PCDH19 Epilepsy.

Ganaxzolone Clinical Trials

Marinus Pharmaceuticals has launched Phase II clinical trials for the use of their drug, Ganaxzolone, in the treatment of uncontrolled epilepsy in patients with PCDH19. The first patients have been enrolled and we are looking forward to seeing how this trial progresses. We are working closely with Marinus to help in any way we can.

Establishment of the PCDH19 Epilepsy Awareness and Research League (PEARL)

This spring we formalized our partnerships with Insieme per la Ricerca PCDH19 (Italy), PCDH19 France, and Asociacion Epilepsy Rosa (Spain) with the establishment of a league aimed at coordinating international efforts to fund research for and raise awareness of PCDH19 Epilepsy.

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

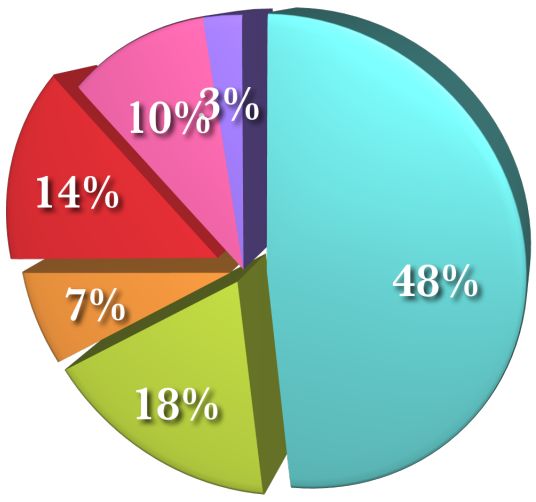
SCN8A Epilepsy is an emerging genetic diagnosis—with only 100 cases diagnosed so far. Like PCDH19 Epilepsy, these numbers are expected to grow exponentially as access to diagnosis is improved. We have connected with Juliann Bradish whose daughter Adeline has SCN8A Epilepsy. Juliann runs an SCN8A parent 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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