THE CUTE SYNDROME FOUNDATION



Colleen and Emma Johnston at the 2018 SCN8A Clinician, Researcher, and Family Gathering

Photo credit: **Ryan Collerd**

ANNUAL REPORT

November 2019

XENON

CLAYTON'S CRUSADE
SCN8A
Join The Battle!

PRAXIS
PRECISION MEDICINES

GREENWICH™
BIOSCIENCES

Zogenix



Katie Banks, TCSF volunteer, and her son Wyatt
at the 2017 SCN8A Clinician, Researcher, and Family Gathering

Photo credit:
Ryan Collerd



SCN8A Families at the 2018 SCN8A Clinician, Researcher, and Family Gathering

Photo credit: Ryan Collerd

A word from our Founder, Hillary Savoie,
on The Cute Syndrome Foundation's sixth year

It is hard to imagine that this year The Cute Syndrome Foundation is already six years old! In the last year we have seen such tremendous growth—we have a new leadership team who works directly with me to help define our goals and move our initiatives forward; our volunteer team is now 30 strong; we have added two SCN8A parents to our Board of Directors; and we have already more than doubled our revenue this year, with two months still remaining in 2019.

This Annual Report will highlight some of our accomplishments of this year, including the launch of support programs for bereaved parents, three newly awarded research grants, one particularly exceptional fundraising partnership, our SCN8A Research Roundtable, and our patient engagement efforts with biopharmaceutical partners. We will also highlight our financials—including our 2018 spending and our 2019 trend report—and our growing connections with the SCN8A community world-wide.

At the beginning of next month we will hold our most important event of the year: The Fifth Annual SCN8A Clinician, Researcher, and Family Gathering in Baltimore, which is held in conjunction with the annual American Epilepsy Society Meeting. At this event we are able to provide the unique opportunity to bring children and families affected by SCN8A mutations together with the scientists and clinicians working to improve the lives of those with SCN8A. This event has been made possible by our generous sponsors including out title sponsor, Xenon Pharmaceuticals; our Gold Level non-profit sponsor, Clayton's Crusade; our returning sponsors: Praxis Precision Medicines, Zogenix, and Greenwich Biosciences; and our new sponsor, GeneDx.

We are tremendously grateful for the continued generosity of our sponsors and donors who have helped us make all of these big things happen!

There is a reason that all of this work is so important: Children with SCN8A mutations need our help to remain healthy and safe. Since the last time I sat down to write an annual report update, ten of our children have died: Emerson, Angelo, Hafsa, Laurel, Magna, Julian, Augustin, Wassim, Aneurin, and Eric. These children, and the far too many others that we have lost, are the reason TCSF continues to push the boundaries of what a small organization can do for all of the members of our SCN8A community.

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

The Cute Syndrome Foundation is is tax-exempt under Section 501(c)(3) of the Internal Revenue Code and donations are tax deductible to the extent permitted by law.

SCN8ACOMMUNITY

Volunteer Highlight: Liz and Erin provide support for bereaved parents

In May 2019 TCSF volunteers Erin Rackham and Liz Ramirez started an SCN8A Bereavement Facebook Support Group. Both Erin and Liz are trained as marriage and family therapists, and Erin is working on her doctorate degree.

With the devastatingly high mortality rate associated with SCN8A, it was realized that there was a need to provide a specific kind of support to parents who have lost their children to the syndrome. Although the SCN8A Bereavement Support Group is relatively small at this point, it is a harsh reality that more members will be joining the group as time passes. The Facebook group provides a safe place for members to provide support, comfort, and resources for one another, as the loss of a child can be a very isolating and life-shattering experience.



I lost my son, Will, to Sudden Unexplained/ Unexpected Death in Epilepsy (SUDEP) in July 2017 when he was only 7 years old. Being part of and facilitating the group provides me with meaning and purpose as I continue to grieve the loss of my son. It is my hope that coming alongside other grieving parents might provide them with much-needed support.

-Liz Ramirez

(ABOVE) Will Ramirez
Photo courtesy of:
Liz Ramirez

(RIGHT) Maura, Ryan, and Luke Pitino
at the 2019 Walk to End Epilepsy

Photo courtesy of:
Epilepsy Foundation of Metropolitan NY



Fundraising Highlight: Luke Pitino's Epilepsy Walk with EFMNY/TCSF

Ryan Pitino, one of our recent additions to the TCSF Board of Directors, met with TCSF Founder/Director Hillary Savoie this summer. It had bee just a few months since his son Luke's diagnosis with SCN8A, and Ryan wanted to know what he and his family could do to get involved in supporting TCSF. Within a month, Ryan and his family had set up a partnership with the Epilepsy Foundation of Metropolitan NY to fundraise in conjunction with The Cute Syndrome Foundation. They organized a team in honor of Luke for the Epilepsy Walk, raising over $72,000—half of which has come to The Cute Syndrome Foundation in support of our SCN8A mission.

We are so grateful to Luke Pitino's family and friends for their wonderfully generous support of EFMNY and TCSF!

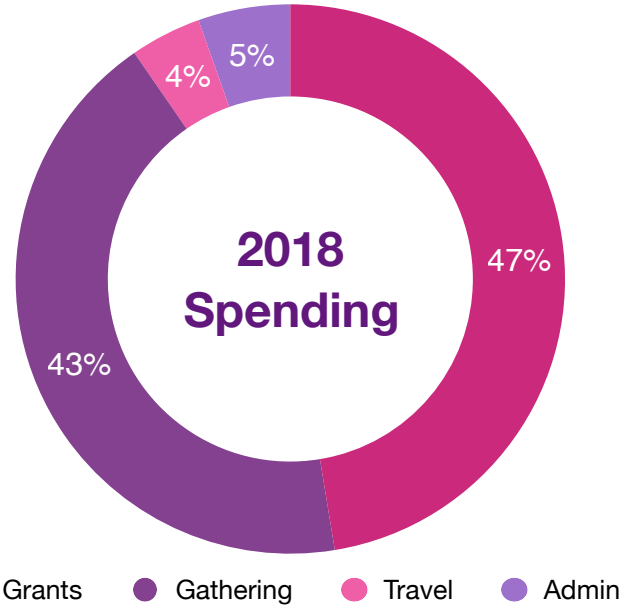
SCN8A FUNDING

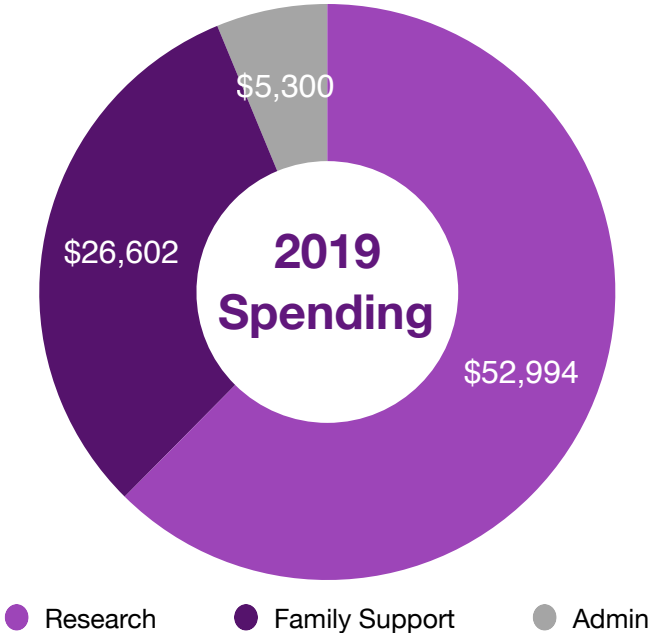
What We Support: 2018 Spending

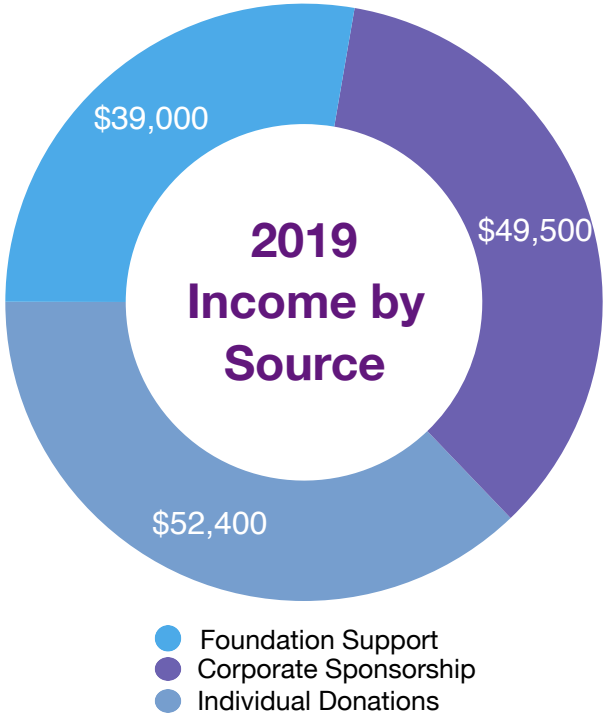
TCSF is focused on providing family support, improving clinical outcomes, and funding medical research for those living with SCN8A mutations. 90% of our spending in 2018 went toward SCN8A Research Grants—including the final payment on our 2017 Zebrafish Research Grant—and our Annual SCN8A Clinician, Researcher, and Family Gathering, which is now a two-day event.

Because we are powered by volunteer efforts, we have been able to keep our administrative costs very low. Currently we have 30 volunteers working on our TCSF projects. Two volunteers each work very close to full-time on TCSF, putting in a collective 2000+ hours/year. An additional five long-term volunteers share another 2000 hours/year working on social media, web design, marketing, fundraising as well as family education, engagement, and awareness efforts.

In 2019 we have also kept our administrative costs very low:







2019 Trends

We do not have full reporting on the 2019 financials yet, however, as of October 18th, we have raised over $140,000—more than doubling our income from last year, with approximately equal parts coming from TCSF-specific individual donations and corporate sponsorships. Thanks to a partnership with the Epilepsy Foundation of Metropolitan New York and Luke Pitino's family, this year we brought in over $35,000 of individual donations through the Epilepsy Walk in September.

[INSERT NUMBERS FOR NETWORK FOR GOOD HERE]

SCN8ACONNECT

The SCN8A Clinician, Researcher & Family Gathering

Last December The Cute Syndrome Foundation hosted our fourth annual SCN8A Epilepsy Clinician, Researcher, and Family Gathering in New Orleans, Louisiana. For the fourth time, the SCN8A community was able to meet with the professionals who dedicate their careers to helping our children, share our stories and learn more about SCN8A. For the second year in a row, TCSF held a second day of sessions, geared toward families and covering topics of importance to those living with SCN8A every day.

Our 2019 event, which will take place in Baltimore, will offer the same opportunities, and, this time, it will include longer family-focused sessions as well as some family fun!

2019 Research Roundtable

This year, upon the advice of our Scientific Advisors, TCSF held an two-day research-focused event in Chapel Hill, NC. This event brought together a number of experts in the field—from both industry and academia—to discuss the SCN8A research landscape, share their research in detail, and lay the groundwork for future collaborations. We are grateful for a generous donation from Southwest Airlines that allowed us to extend flight vouchers to many of the participants! We would also like to thank to our event sponsors: Xenon Pharmaceuticals, Praxis Precision Medicines, Zogenix, and Greenwich Biosciences.







Top Row from left to right: **Shelley Frappier and Colleen Johnston; Tammy Kooistra holds her son Liam, and Kiran Reddy, MD, of Praxis meets with families** at the fourth annual Gathering in New Orleans (December 2018) Photo credit: **Ryan Collerd**

Right: **Researchers participating in the 2019 TCSF SCN8A Research Roundtable** in Chapel Hill, NC



2018 SCN8A Champions

Each December, at our Annual Clinician, Researcher, and Family Gathering, we hold the CUTIES Awards, which are given to Champions for Understanding, Treating, Investigating, and Empowering those with SCN8A. Our 2018 parent recipient was Dianely Cabrera, for her years of volunteer work for TCSF as our social media volunteer. We also honored JP Johnson, Jr. Ph.D. for his SCN8A-focused research for Xenon Pharmaceuticals in their development of a Nav 1.6-selective sodium channel blocker, as well as his dedication to helping the SCN8A family community understand the science of SCN8A!

Congratulations JP and Dianely! Thank you for your dedication to our SCN8A community!

SCN8AENGAGED

TCSF Visits Industry Partners Praxis & Xenon

In April of this year, our founder and director, Hillary Savoie, flew to Vancouver, British Colombia to visit Xenon Pharmaceuticals, a long-time TCSF sponsor and developer of a potential drug treatment for SCN8A Epilepsy. While there, Hillary visited their laboratories, met with their clinical and drug discovery teams, and spoke to the entire company about TCSF, the SCN8A community, and patient engagement.

In July of this year a team of TCSF volunteers visited another biopharmaceutical partner in Cambridge, Massachusetts: Praxis Precision Medicine. Praxis is also developing a potential drug treatment for SCN8A—and they invited TCSF to come and talk to them more about the SCN8A experience and our goals for clinical trials. Leadership team volunteers Colleen Johnston and Karen Varner and Hillary, brought their daughters with SCN8A mutations, Emma, Lily, and Esmé to the meeting.

We are very fortunate to be working with two industry partners who are not only eager to help develop better treatments for SCN8A, but who want to work directly with our patient/caregiver community to ensure that their work is as responsive as possible to our needs.





(LEFT) Hillary Savoie, TCSF Founder/Director and JP Johnson, JR, Xenon VP of Biology
Photo courtesy of: **Xenon Pharmaceuticals**

(RIGHT) TCSF Volunteers
visiti Praxis Precision Medicines

Engaging Our Growing Community

TCSF's ability to engage with SCN8A caregivers around the world is growing, as more and more families connect with us through our Facebook Support Group and our newsletter. While the world population of those diagnosed with SCN8A is currently unknown, it is believed to be somewhere on the magnitude of 400 individuals. Through our connections with families, we current are in touch with 275 families in 37 different countries!

37
countries represented

88%
research consent to contact

This fall we co-sponsored a survey of our SCN8A families in the US, Canada, UK, and Australia. This survey will help aid Xenon in designing their clinical trial—and help TCSF and other researchers better understand our SCN8A community. 88% of our entire community in those areas has agreed to be contacted for research—and of those individuals we saw 80% participation in the Xenon/TCSF survey.

$4500
family travel grants

80%
survey participation

While connecting with others on Facebook and through our newsletters is wonderful, we also prioritize in-person community connections by supporting families attending our Annual Gathering each year. Attendance at the Gathering is important to families for a number of reasons: 1) learning about cutting edge SCN8A clinical, research, industry advancements, 2) demonstrating our community's commitment to science, 3) engaging in caregiver-focused sessions, and 4) forming in-person connections with others in our community. However, we recognize that not everyone can easily attend our Gathering due to financial and geographic challenges. As a result in 2017 we launched the Dr. Mandy Harris Memorial Travel Grant in order to support members of our SCN8A community to attend the conference. This year we are providing a record breaking six travel grants to families traveling internationally and within the US.

SCN8ARESEARCH

2019 RESEARCH GRANTS WITH AES

In 2019 TCSF was thrilled to co-fund two SCN8A-related seed grants with the American Epilepsy Society.

The first grant, "Targeting micro-RNAs to treat genetic epilepsies," is awarded to Benjamin Prosser, PhD; Ingo Helbig, MD; and Beverly Davidson, PhD of the University of Pennsylvania. This grant has been supported in a partnership with American Epilepsy Society, The Cute Syndrome Foundation, and SLC6A1 Connect.

The second grant, "Testing the role of early mitochondrial dysfunction in SCN8A epilepsy" is awarded to Michael Hammer, PhD, and Fei Yin, PhD of the University of Arizona. This grant has been supported in a partnership with American Epilepsy Society and The Cute Syndrome Foundation.







On left: Benjamin Prosser, Ph.D.

Center and Right: Michael Hammer, Ph.D. and John Schreiber, M.D. at the fourth annual Gathering in New Orleans (December 2018)
Photo credit: **Ryan Collerd**

2018 Clinical Research Grant

At our 2018 SCN8A Clinician, Researcher, and Family Gathering in New Orleans, The Cute Syndrome Foundation announced the awarding of a clinical research grant to Dr. John Schreiber and Children's National. The vision for this grant developed after the devastating loss of two infants in our SCN8A community last fall, and we are grateful to Dr. Schreiber for working with us to look for ways to improve access to expert advice in the treatment of individuals with SCN8A.

The official announcement follows:
Children's National Medical Center and The Cute Syndrome Foundation are delighted to enter a 6-month pilot research project to evaluate education and access to expert care for SCN8A. Led by Dr. John Schreiber, Neurologist and Assistant Professor of Neurology and Pediatrics at George Washington University, Children's National will develop a focused effort to provide families and clinicians with the Clinician Information and Reference Guide—developed by The Cute Syndrome Foundation— and attempt to uncover the barriers to accessing expert advice. The study will result in a better understanding of those factors that prevent access to consensus recommendations and expert care. It will also increase provider knowledge of SCN8A-related disorders, improve utilization of appropriate anti-seizure therapies, and may eventually reduce mortality. As the first ever study of its kind, this pilot project will pave the way for future interventions that will raise the profile of SCN8A-related disorders, facilitate collaborative care and research, improve patient outcomes, and reduce mortality.

SCN8AGRATITUDE

We would like to extend our thanks to:

Facebook Fundraiser Organizers

XXXXX

2019 Donors To Date

Thank you, as well, to those who made donations through YourCause & Benevity

AbbVie Employment Engagement Fund
Accuform Industries, Inc
Alan and Nancy Brown
Amy Gestosani
Amy Glynn
Breanne Zebrowski
Daniel Mills
Debbie Stroemer
Diana Friedman
Emily Wellman
Feisal Afzal
GSCSA
Gennine Uliasz
Inna Mitelman
Janice Fish
Janice Miller
Kaitlyn Livoy
Kathie Glynn
Kevin Swider
Laurel Gitlen and Samuel Richardson
Lauren Vandewalle
M Isel-Margolis
Marcio Carvalho
Marlene McInnes
Mary Robinson
Meaghan Kearns
Megan Sell
Michael Ramirez
Olivia Valenti
Patricia Livoy
Robert Wolfe
Rosemarie Montanero
Ryan Pitino
Sarah Wolfe
Stuart Chaffee
Swiders Christmas Classic
Thomas Lee
Timothy Wilkins
Trisha Rathke
William Gelsomini
Ryan Pitino
Marianne Sutcliffe

Volunteers

Alisha Wheeldon
Colleen Revene Johnston
Daniel Braz
David Baboneau
Denise Short
Dianely Cabrera
Erin Rackham
Hillary Savoie
Jaden Bitton
Jennifer Costello
Jennifer Smith-Stout
Juliann Bradish
Kacie Craig
Karen Hess Varner
Katie Banks
Lacey Juroske
Liz Ramirez
Maggie Nayyar
Mali Finn
Marianne Sutcliffe
Megan Gard
Megan Varner
Miranda Pond
Nancy Christensen
Natan Pinheiro
Ryan Pitino
Shelley Frappier
Sandy McClurg Bush
Tonya Baboneau
Trisha Rathke
Wendy Woody

2019 Sponsors

Clayton's Crusade
GeneDx
Greenwich Biosciences
Praxis Precision Medicines
Southwest Airlines
Xenon
Zogenix

Board of Directors

Hillary Savoie
Lan Aronson
Monica Lamontagne
Philip Gitlen

For more information on SCN8A, please see our updated *SCN8A Clinician Reference Guide* at **www.thecutesyndrome.com/reference-guide.html**