THE CUTE SYNDROME



Juliann Bradish, Co-Executive Director, TCSF holds Clayton Gard, a little boy with SCN8A at the 2016 SCN8A Clinician, Researcher, and Family Gathering

Photo credit: **Ryan Collerd**

ANNUAL REPORT

November 2017

A word from our Founder, Hillary Savoie

on The Cute Syndrome Foundation's fourth year

This year has been one of tremendous growth for The Cute Syndrome Foundation.

Juliann Bradish, who began as our "SCN8A Advisor" in 2015, has now become the Co-Executive Director, together with myself. This title change for Juliann is reflective of the growing responsibilities she has taken on—including spearheading our Annual Gathering. The SCN8A Family Support Group, which Juliann established in 2014, soon after her daughter Adeline was diagnosed with an SCN8A mutation, has grown to represent around 150 families raising children with SCN8A. The support group is now administrated with the help of four TCSF parent volunteers. Our volunteer team, has also grown to include 16 relatives of those affected by SCN8A. This team works on a number of projects including fundraising, SCN8A awareness, clinician outreach, and parent coordination with Dr. Michael Hammer on the SCN8A Registry, SCN8A.net.

Other highlights of this year include our launch of the first International SCN8A Awareness Day and the awarding of our 2017 grant, together with our partner organization in Brazil, Ajude o Rafa—both of which we are thrilled to present in this report.

Next month we will hold our most important event of the year: The Third Annual SCN8A Clinician, Researcher, and Family Gathering in Washington DC, in conjunction with the annual American Epilepsy Society Conference. 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 in part by our generous sponsors: Xenon Pharmaceuticals, Praxis Precision Medicines, The Rare Epilepsy Network (REN), Greenwich Biosciences, and Pairnomix. Last year's meeting, which took place in Houston, was a tremendous success, bringing together over 100 guests to discuss the future of SCN8A research, potential treatments, and best practices for treatment

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

-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.

SCN8AEPILEPSY

The SCN8A Clinician, Researcher & Family Gathering

Last December The Cute Syndrome Foundation hosted the second annual SCN8A Epilepsy Clinician, Researcher, and Family Gathering in Houston, TX. Over 100 guests met, including professionals working on SCN8A research and treatment and more than 40 members of our SCN8A family community. 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 second time, the SCN8A community was able to meet with the very professionals who dedicate their professional time to helping our children! This event allowed families the opportunity to share their stories and to witness the researchers and clinician's dedication to this community—inspiring hope in all who attended. We look forward to another ground-breaking gathering this year in Washington DC!







From left to right: **Paul Goldberg** of Xenon Pharmaceuticals**, Emma Johnston, and Mackenzie Wardrope**
at the second annual SCN8A Epilepsy Clinician, Researcher, and Family Gathering in Houston, TX (December 2016) Photo credit: **Ryan Collerd**

Dr. Cummins Awarded 2017 SCN8A Epilepsy Grant

This year, as a collaborative effort with our partner organization in Brazil, Ajude o Rafa, The Cute Syndrome Foundation helped award a $40,000 grant to Ted Cummins, PhD and James Marrs, PhD, of Indiana University-Purdue University Indianapolis and Indiana University School of Medicine for their proposal to create an SCN8A zebrafish model.

Dr. Cummins said of his work:
"Mutations in SCN8A can lead to seizures and other serious medical problems. Some patients respond to available drugs, but many do not achieve adequate therapy or suffer side effects. Better treatments are needed. We propose to develop zebrafish models of several different SCN8A mutations. Zebrafish models can be inexpensively and rapidly developed compared to mouse models. They can be used for fast screening of potential drugs in automated assays. Because this high throughput screening is done with intact vertebrate animals, it has tremendous advantages over test-tube and cell-based screens. Our long-term vision is to develop a specific model for each SCN8A mutation to help identify personalized treatments for patients with specific SCN8A mutations."

SCN8A Registry: SCN8A.net

In 2015 we helped to fund the launch of the SCN8A Interactive Website, SCN8A.net. Within the past year SCN8A.net has become a valuable source for discovering current SCN8A literature. This year The Cute Syndrome expanded our relationship with SCN8A.net by collaborating on an effort to increase SCN8A community participation in the SCN8A Registry. Through our efforts with in the Facebook SCN8A Family Support Group and the work of the SCN8A.net team, over 90 families have already started the SCN8A Registry, which went live at the beginning of this year, and approximately 80% of these have already been completed! With each completed registry we are one step closer to understanding the full spectrum of SCN8A-related disorders.

SCN8AEPILEPSY

SCN8A Awareness Day

This year we spearheaded the first International SCN8A Awareness Day! We celebrate SCN8A Awareness Day on February 9th, to honor the birthday of Shay Hammer—the first known person with SCN8A. The awareness day was a great success. It provided a chance for our community to come together around our children and the challenges we share. The campaign also increased the number of families in our SCN8A Support Group by almost 10% and raised essential funding for SCN8A.net.



SCN8A Community

At our Gathering last December we honored three individuals with CUTIES Awards, Awards given to Champions for Understanding, Treating, Investigating, and Empowering those with SCN8A. Our recipients included research recipient, Manoj Patel, PhD, for his dedication to collaborative SCN8A research and parent recipient Jessica Jenkins, for her volunteer work for TCSF both before and after the loss of her son Dustin, due to complications from his SCN8A mutation. We also honored clinician Mandy Harris, MD, for her work both as a physician to those with SCN8A and in an advisory capacity with TCSF and others who are trying to improve the health and safety of those with SCN8A. Sadly, early this year Dr. Harris passed away unexpectedly. Her loss has been deeply felt by our SCN8A Family.







2017 SCN8A CUTIES

Manoj Patel, PhD (pictured with and Juliann Bradish and Hillary Savoie)
Jessica Jenkins (pictured with and Hillary Savoie)
Many Harris, MD (pictured with and Juliann Bradish)

Photo Credits: Ryan Collerd

TCSF is honoring the memory of Dr. Harris by creating the Dr. Mandy Harris Travel Grant, which will help two families attend this year's SCN8A Gathering in Washington DC. We believe that Dr. Harris would be thrilled to know that we are helping travel grant awardees feel as though they can fully participate in the all aspects of the SCN8A community—with families, researchers, and clinicians. As one of this years recipients, Katie Banks said, "To be at the gathering would make me feel like I am doing something proactive and important to help our son as well as all of the other children in our SCN8A family."

SCN8AEPILEPSY

We would like to extend our thanks to:

Fundraiser Organizers

Melissa Peverill
Karen Varner
Sandy Bush
Dianely Cabrera
Colleen Johnston
Juliann Bradish
Kacie Craig
Mackenzie Wardrope
Megan Gard
Federica Curione
Diego Costa

Donors

A&DbyChristinaU
Alan Aronson
Alfred and Georgia Scrivo
Allyson Polotaye
Amanda Rotondo
Amy Svrjcek
Andrew and Maggie Bailey
Angela Savoie
Angelina McDonald
Aubry Bracco
Blanca Diaz
CarVal Investors
Casey Brands
Cheryl Rebsamen
Christen Komosinski
Colleen Johnston
Daniel Kelley
David and Karen Varner
David Kennedy
Denise Breeding
Diane Gajewski
Diego Ruiz de Somocurcio
Eileen Berg
Elizabeth Mercy
Emily Reed
Erika Horowitz
Federica Curione
First Lutheran Church and School
Florence Kooistra
Floyd and Sherrill Smithey
GE United Way Campaign
Greg Ricketts
Gwen Willey
Heidi Arvin
Hela Trerotola
Jeanette Bordeaux
Jennifer Donohue
Jennifer Sciotto
Jessica Jenkins
John McCollum
Jon Bepko
Juliann Bradish
Kacy Craig
Kara Rogan
Kate Weinstein
Katherine Robart
Katherine Roome
Kathleen Gleeson
Kathryn O'dwyer
Kendall Sign and Design
Kerri Kuzmar
Kim Boatman
Kristi McCullough
Lauren Vandewalle
Linda Restifo
Linda Secor
Lisa Giese
Lorene Middleton
Margaret Ells
Marianne Sutcliffe
Mary Robinson
Matilda Jane Clothing
Meghan Cheek
Michael Ace
Michael Hammer
michael ramirez
Michelle Levi
MicSavoie Photography
Nancy Brown
Nancy Kennedy
Pairnomix
Pennywise Shoppe
Praxis Precision Medicines, Inc
Ray and Tina Clark
Rev & Mrs. Paul Bushur
Richard Rothwell
Roland Waegner
Ronald Wardrope
Ruth Hess
Sam Richardson
Sharon Cuddy
Sherri Schecter
Sophia Sattar
Stacy Thomas
Stanley Gajewski
Steven Hall
Suzanne Bellofatto
Tanya Paulin
Thirsty Canvas
Thomas Preston
Trisha Rathke
Vincent Oliver
Virginia McCaffrey
Wan Ling Fahrer
Xenon

Board of Directors

Hillary Savoie
Philip Gitlen
Monica Lamontagne
Lan Aronson

Grant Scientific Advisors

Michael Hammer, PhD
Miriam Meisler, PhD
Manoj Patel, PhD
John Schreiber, MD
Phillip Pearl, MD
Princess Costello
Daniel Braz
Hillary Savoie, PhD
Juliann Bradish, PharmD

Volunteers

Dianely Cabrera
Colleen Revene Johnston
Maggie Nayyar
Jennifer Smith-Stout
Megan Varner
Karen Hess Varner
Shelley Frappier
Sandy McClurg Bush
Alisha Wheeldon
Jaden Bitton
Jessica Jenkins
Daniel Braz
Natan Pinheiro

For more information visit us online:
www.thecutesyndrome.com
or find us on Facebook at
www.facebook.com/thecutesyndrome