The Cute Syndrome FOUNDATION

www.TheCuteSyndrome.com

2021 ANNUAL REPORT

CONTENTS

3 *A Letter from Our Founder*

4 *SCN8A Community*
Volunteer Highlight

6 *SCN8A Funding*
Programs and Grants
Clinical Trial Support
International Efforts
Finances

12 *SCN8A Connect*
Meaningful Change Series
Clinical Trial Readiness Series
New & Updated Family Resources

16 *SCN8A Engaged*
International SCN8A Awareness Day
Social & Emotional Programming
7th Annual Clinician, Researcher, and Family Gathering

20 *SCN8A Research*
2021 SCN8A Research Grant
Survey Series
Ciitizen Natural History Study Campaign

24 *SCN8A Gratitude*
Fundraisers
Sponsors
Executive Committee
Board of Directors

A LETTER FROM
OUR FOUNDER



HILLARY SAVOIE
Founder & President
The Cute Syndrome Foundation

Founding The Cute Syndrome Foundation and running it these last almost 9 years has been one of the greatest honors of my life. This foundation has been responsible for some of the greatest lessons, friendships, and joys of my life. Sometimes it happens that a thing you've made takes on a life of its own. This is what has happened with TCSF. That is why I am writing today with an announcement: I am thrilled to share with you the appointment of Kacie Craig as our new Executive Director.

Many of you know Kacie, mom of Stella, as the host of our Meaningful Change and Clinical Trial Readiness Series. She also runs our weekly book club and has served on TCSF's volunteer executive committee. Kacie will be here to lead TCSF into the next era as we move into expanding our support, engagement, and educational efforts for the SCN8A community.

As I transition to supporting TCSF from a role on the Board of Directors, I am reflecting on the tremendous work we have done over the years. Our family support group and programming has expanded to offer programming year round to serve the educational, social and emotional, and engagement needs of the SCN8A community. Our revenue has increased 10-fold in our time focusing on SCN8A. We are fortunate to have a clinical trial enrolling in our community. This organization is currently run by a number of committed family members of individuals with SCN8A.

I am so proud of what we have accomplished. And I am so thrilled to see what is to come under Kacie's tenure.

With gratitude,

Hillary Savoie
FOUNDER & PRESIDENT

SCN8A
COMMUNITY

Oftentimes a SCN8A diagnosis is earth shattering to a parent or an individual. The world becomes a dark place filled with uncertainty, loneliness, desperation, and grief. Many families hear the news from a neurologist or geneticist within the walls of a sterile hospital room or doctor's office, but immediately turn to an online search to find comfort with others like themselves.

Finding a community, a tribe, that walks beside each other every step of the way is how one survives a SCN8A diagnosis. TCSF is that tribe for hundreds of families by welcoming them with open arms, celebrating successes, supporting during trying times, and mourning together during times of loss. We experience life full circle with all of our TCSF families.



VOLUNTEER HIGHLIGHT



MARY TONE
TCSF Volunteer

Mary Tone is the SCN8A parent to the charming Billy. Billy is the highlight of many of our lives as we've watched him grow into adulthood. He's full of life and laughs—in true CUTE form. Mary served as a police officer for the first 23 years of her professional life and transitioned into a special education teaching assistant for another 15 years.

In March of 2021, Mary decided to finally retire into a full time grandma, companion to her Bill, and reached out to TCSF to inquire about becoming an active volunteer. I believe her request was something along the lines of, "I have some free time, now put me to work!" We did just that! Mary works very closely with our Executive Assistant, Shelley Frappier, to welcome new families into our SCN8A Family Support Group Facebook page and enter their information into our master database. Mary does an outstanding job as we welcome new families and individuals into our TCSF tribe.



LACEY JUROSKE
TCSF Volunteer

Lacey Juroske is a talented artist that makes jewelry, keychains, bag tags, and more for her small business, D&L Impressions. On March 4, 2019 her sweet little Sadie was diagnosed with SCN8A and she found our support group two weeks later. Lacey is a working mom busy with three young children, but she recognized a unique opportunity to honor those that have lost their battle to SCN8A and jumped into action. In 2019, she began creating and donating custom made memorial ornaments for our SCN8A Angel Families.

Losing a loved one to SCN8A is heartbreaking, but due to Lacey's kindness and generosity TCSF is able to honor our Angel Families with a piece of art representing their beautiful life here on Earth.

SCN8A FUNDING

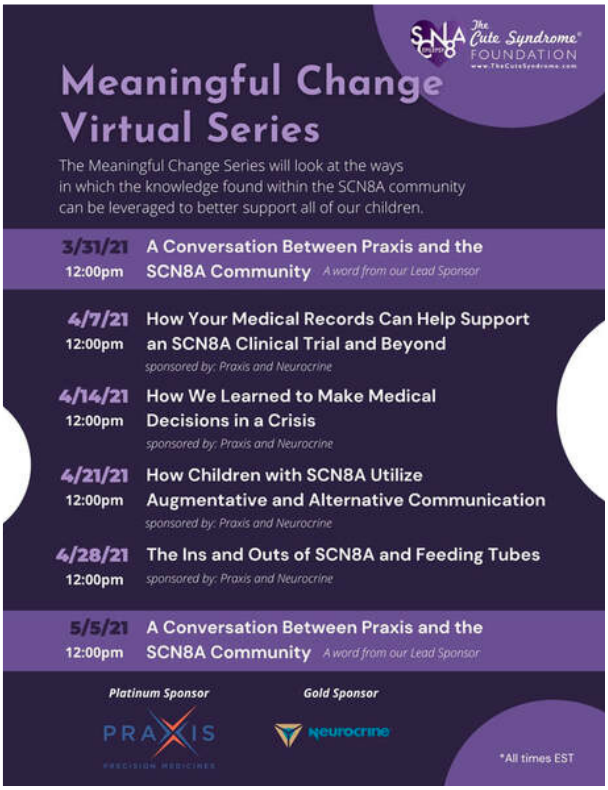
TCSF had another amazing fundraising year. We surpassed our fundraising goal to reach an important new milestone: **$250,000**! We are so grateful for this tremendous show of continued support.

$250,000

TOTAL RAISED

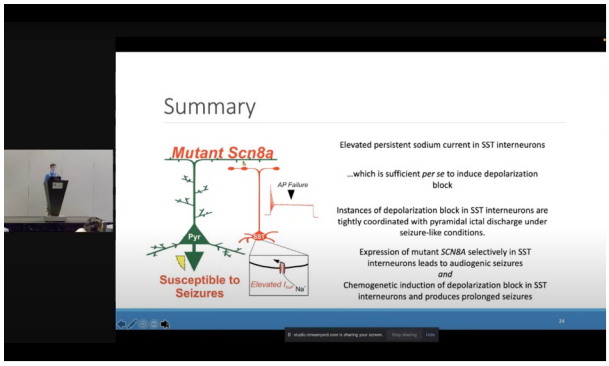
$130,000

TOTAL SPONSORSHIPS RECEIVED









THANKS TO THIS FUNDING TCSF WAS ABLE TO PROVIDE A NUMBER OF PROGRAMS IN 2021, including the Meaningful Change Series, Clinical Trial Readiness Series, social and emotional support programs, The Moth Storytelling workshop, and the Annual SCN8A Clinician, Researcher, and Family Gathering. We were also able to fund a SUDEP-related research grant at the Patel Lab.



One of our main priorities in 2021 was to provide strategic support for our industry partners as they ramped up to **CLINICAL TRIALS FOR THE SCN8A COMMUNITY**. We did so in the Ciitizen campaign, mapping for clinical trial site selection, drug formulation feedback, and providing insight into the needs of the SCN8A community. Together with these partners, TCSF was an author on four posters and a research article in 2021.



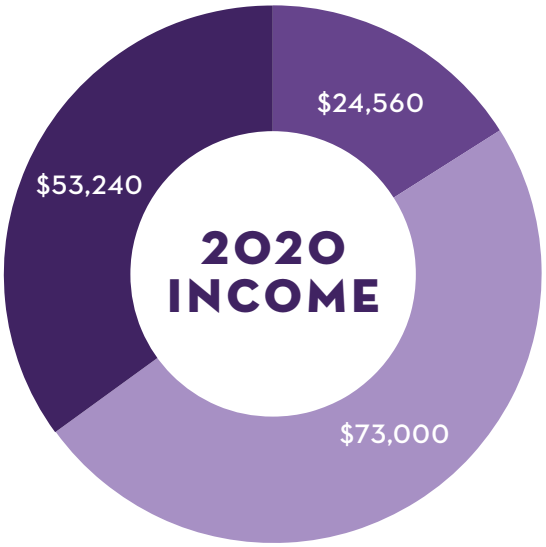


We also worked with our international partners to

COORDINATE EFFORTS ACROSS BORDERS

in ways that support individual needs within countries and regions. All of this happened thanks to the hard work of the TCSF team and our amazing partners.

A LOOK AT 2020 AND 2021 FINANCES

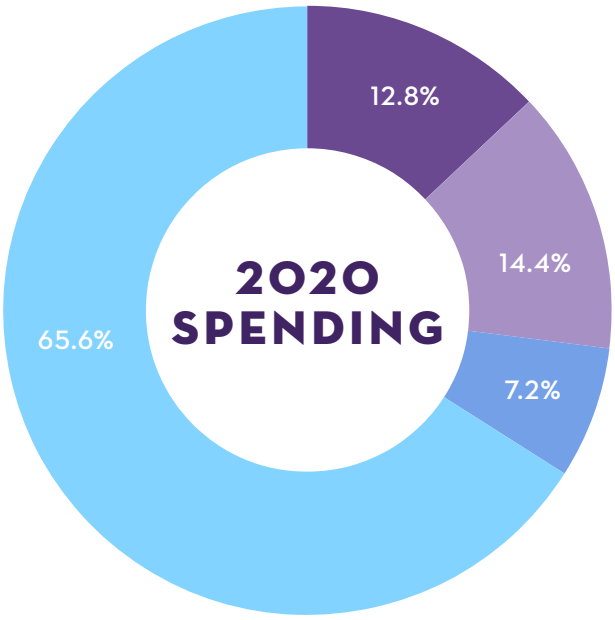


- FOUNDATION SUPPORT
- CORPORATE SPONSORSHIP
- INDIVIDUAL DONATIONS

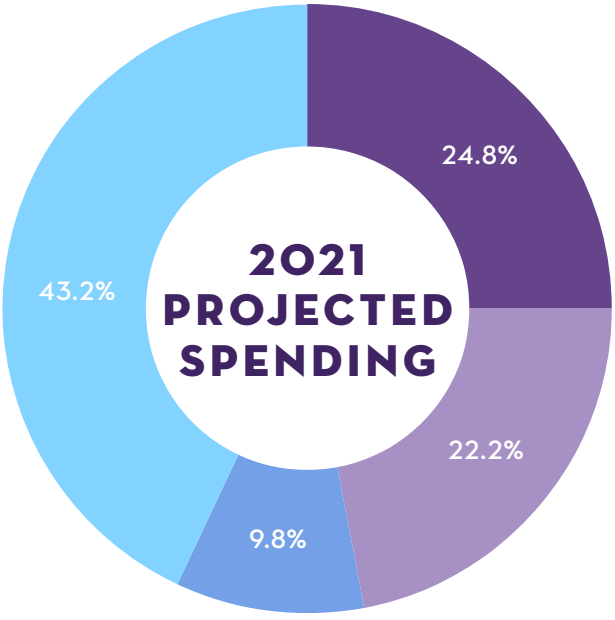


- FOUNDATION SUPPORT
- CORPORATE SPONSORSHIP
- INDIVIDUAL DONATIONS

SPENDING



- RESEARCH
- AWARENESS AND FAMILY SUPPORT
- OPERATING
- FAMILY ENGAGEMENT AND EDUCATION



- RESEARCH
- AWARENESS AND FAMILY SUPPORT
- OPERATING
- FAMILY ENGAGEMENT AND EDUCATION

SCN8A CONNECT

This year, we had the opportunity to connect with our community in a number of ways. We started by hosting our 1st annual Meaningful Change Series, which focuses on supporting our children by utilizing the knowledge found within our community. We also held our 2nd annual Clinical Trial Readiness series, which gave us the opportunity to hear from our partners, Praxis Precision Medicines and Neurocrine Biosciences, as well as learn more about the clinical trial process.

MEANINGFUL CHANGE

1st Annual | Virtual Series

This six part series was held in the spring of 2021 that looked at the ways in which the knowledge found within the SCN8A community could be leveraged to better support all our children. In addition to the conversations with Praxis Precision Medicines and Ciitizen, TCSF heard from 11 SCN8A parents as well as guest, Traci Nagy, the Director of the Feeding Tube Awareness Foundation.

TOPICS

A Conversation Between Praxis and the SCN8A Community: Part 1

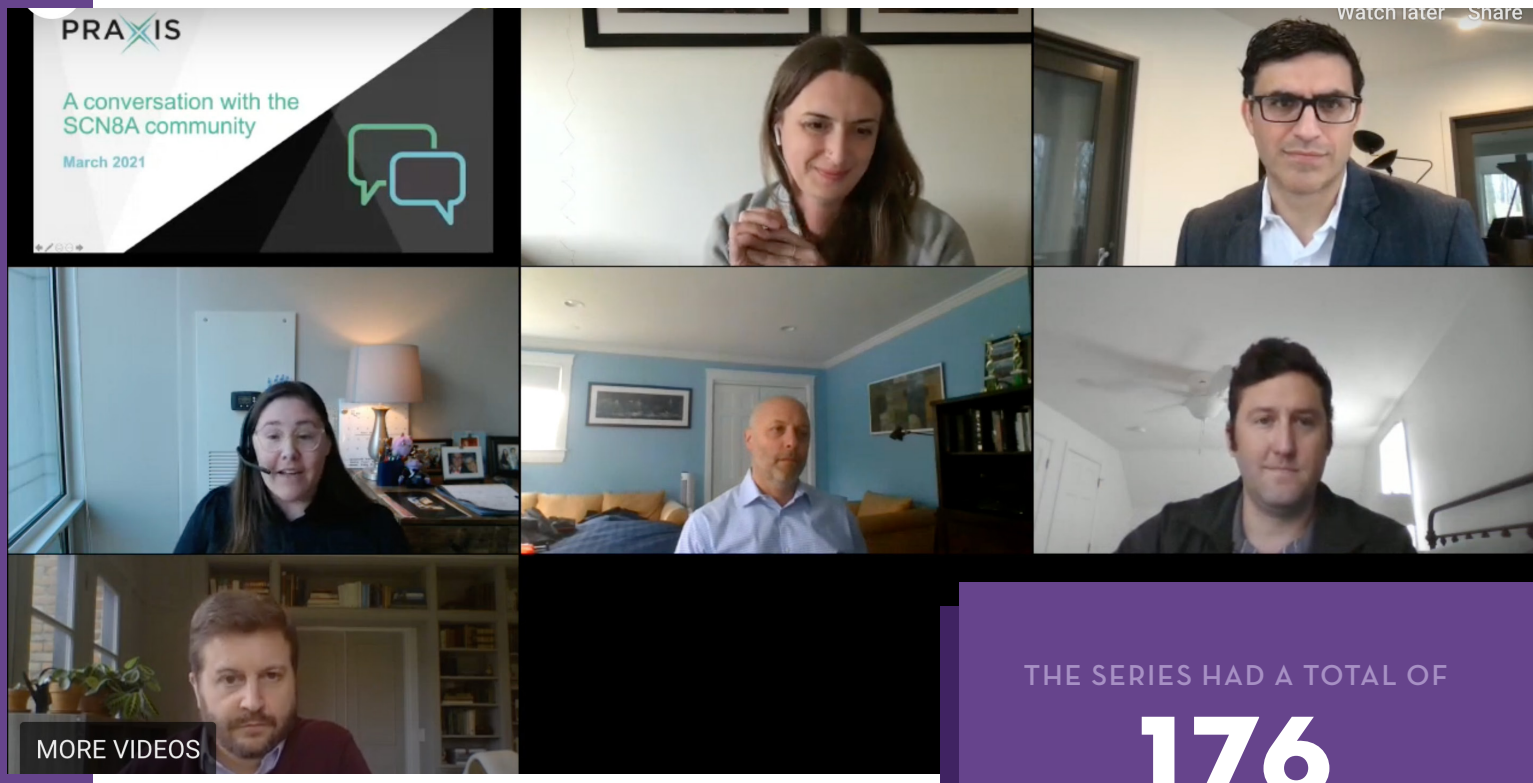
How Your Medical Records Can Help Support an SCN8A Clinical Trial and Beyond

How We Learned to Make Medical Decisions in a Crisis

How Children with SCN8A Utilize Augmentative and Alternative Communication

The Ins and Outs of SCN8A and Feeding Tubes

A Conversation Between Praxis and the SCN8A Community: Part 2



THE SERIES HAD A TOTAL OF

176

LIVE PARTICIPANTS

CLINICAL TRIAL READINESS

2nd Annual | Virtual Series

We believe that helping educate our families about drug discovery and clinical trials is essential to having an informed community that can consent to participate in studies; therefore, TCSF hosted the 2nd annual Clinical Trial Readiness (CTR) series in fall of 2021.

TOPICS

CTR Kickoff: SCN8A International Advocacy Efforts and Update
A Meeting with FDA Representative About The Drug Approval Process
SCN8A and Potential Future Treatments 101
How Families Can Help Beyond Clinical Trials
Community Meeting with Praxis Precision Medicines
Community Meeting with Neurocrine Biosciences

The Hopin virtual platform, used for both Meaningful Change and Clinical Trial Readiness series, allowed TCSF to set up sessions with functionality for presentations, moderated conversations, chat, and video Q&A.



THE SERIES HAD A TOTAL OF

142

LIVE PARTICIPANTS

NEW & UPDATED FAMILY RESOURCES

Resource Page

Having a diagnosis such as SCN8A can be overwhelming. There are many different things patients or their families might need, including seizure monitors, specialty equipment, communication devices, and more. TCSF compiled a robust list of many different resources to help patients and their families with their SCN8A journey.

It was a long project in the making, but due to the hard work of TCSF volunteers Shelley Frappier, Sandy Bush, Karen Varner, and Megan Varner, it is now readily available to our families on the TCSF website.

Translated Clinician Information & Reference Guides

The first SCN8A Clinician Information and Reference Guide was developed in 2016. The purpose of this guide is to promote a broad understanding and knowledge of SCN8A in an effort to support clinicians in the care of patients with SCN8A. Thanks to our international volunteers Roland Waegner, Cinzia Scarcelli, and Daniel Braz—as well as Dr. Angel Aledo-Serrano and Dr. Arthur Cukiert—this document has now been translated into German, Spanish, and Portuguese.

SCN8A ENGAGED

As we embark on a new year, we are reflecting on the ways our community engaged with each other this past year.

We hosted our annual International SCN8A Awareness Day in February, where families from around the world came together to share their experiences with SCN8A. We also hosted a variety of Social and Emotional Support Programming, which aims to give our families a way to decompress, find support, and build connection. Finally, we held our 7th Annual SCN8A Clinician, Researcher, and Family Gathering event, where we focused on the newest advances in drug development, SCN8A research, expert clinical care, and stories of life with SCN8A.

INTERNATIONAL
SCN8A AWARENESS DAY



On February 9, 2021 we celebrated the fifth International SCN8A Awareness Day. The gene SCN8A was isolated in humans by Michael Hammer, PhD, a geneticist who found the gene in his own daughter, Shay, soon after her death. Shaw passed away suddenly in 2011 and is the first documented case of sudden unexpected death in epilepsy (SUDEP) in a child with an SCN8A mutation. To honor her and all of those with SCN8A, TCSF helped launch the International SCN8A Awareness Day on February 9th, Shay Hammer's birthday.

Each year, a group of TCSF volunteers create an Awareness Day video that is broadcast on social media. This year, our video was viewed over 3.3k times in 45 different countries. Our Facebook post alone reached 84.1K people!

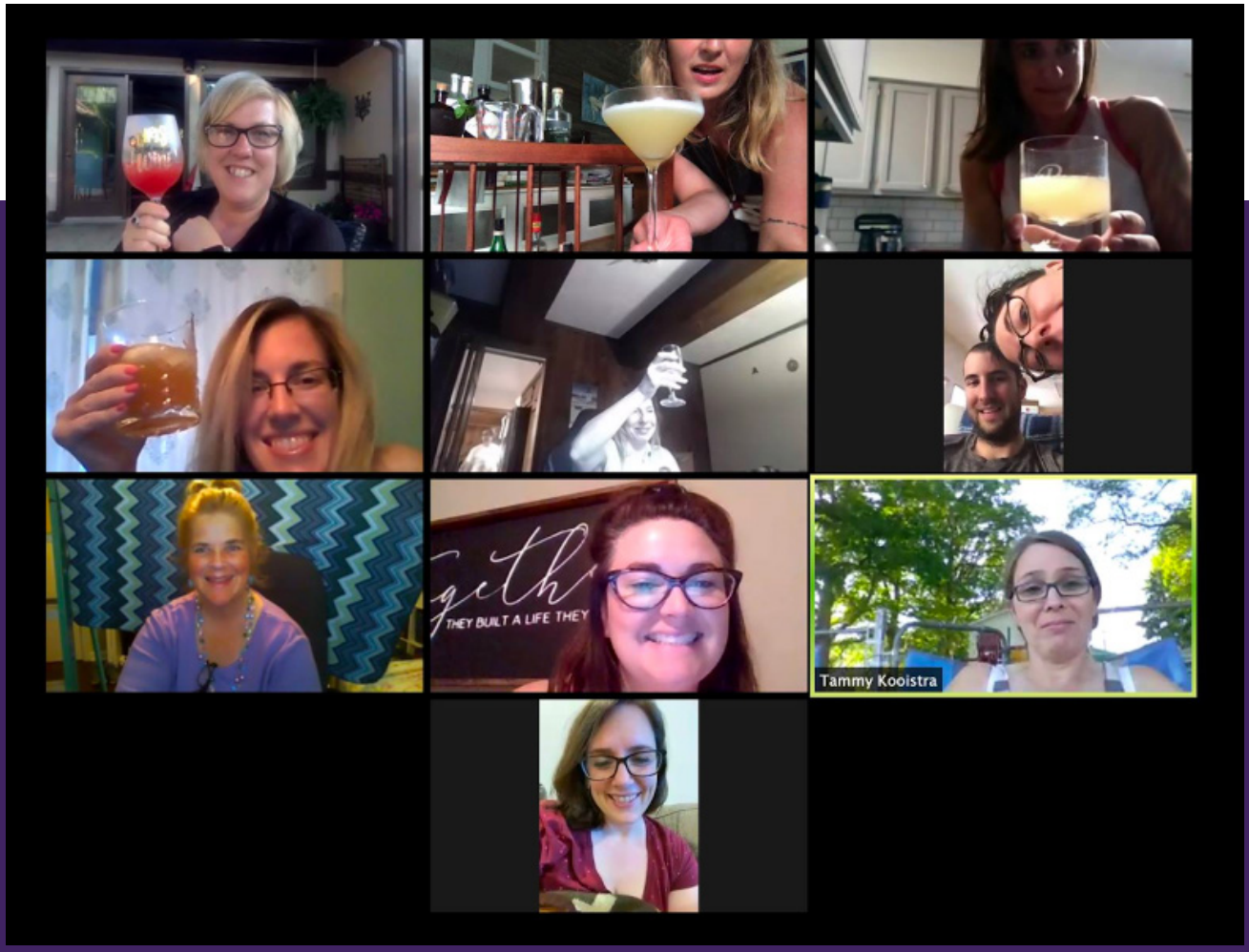
3.3K
VIDEO VIEWS

VIEWED IN
45
DIFFERENT COUNTRIES

1.5K
WEBSITE CLICKS

SOCIAL & EMOTIONAL SUPPORT PROGRAMMING

TCSF's Social and Emotional Support Programming for our families started in the summer of 2020 with the support of the Child Neurology Foundation's Rising Tides Grant, and we were able to continue it in 2021 thanks to sponsorship from Neurocrine Biosciences. This year's new programming included a Self-Care Group with Shelley Frappier, Dad's Group with Ryan Phillips, and Yoga with Kelly Love. Back by popular demand, we continued Food and Drink Club with Hillary Savoie, Sandy Bush, and Brian Bush, and Book Club with Kacie Craig.









7TH ANNUAL SCN8A **CLINICIAN, RESEARCHER, AND FAMILY** GATHERING

In response to the COVID-19 pandemic, TCSF held our annual Gathering as a hybrid event, once again using the virtual platform Hopin. Hopin was so inspired by how we've used their platform that they used TCSF not only as a case study on their website, but also as an internal team example of what they are trying to achieve as a company: bringing people closer together!

Our hybrid event was a complete success with ***146 total attendees: 31 in person and 115 virtual***! We wanted our virtual guests to feel as connected to the gathering as possible during our two-day event, so we shipped our customized TCSF "Conference in a Box" to 74 at-home attendees. They were a complete hit with the families and professionals joining us from afar!

In addition to the live presentations focused on the newest advances in drug development, SCN8A research, expert clinical care, and stories of life with SCN8A, the Hopin Expo area housed videos covering SCN8A 101, SCN8A Italia updates, and heartwarming Moth Storytelling videos from SCN8A parents.

Saturday's family workshop showcased the Moth Storytelling hosts, Juan Rodriguez and Devan Sandiford, where they provided guidance and empowered SCN8A parents to share their impactful stories with the world. We all have a story worth telling!

SCN8A RESEARCH

TCSF is dedicated to supporting and funding various areas and methods of SCN8A research in order to help those with SCN8A live better, healthier lives. This report highlights three key research advancements from this past year: our 2021 Research Grant, our Global SCN8A Survey Series, and the Ciitizen Natural History Study Campaign.



2021 SCN8A RESEARCH GRANT

The Cute Syndrome Foundation is thrilled to share the awarding of a research grant to Ian Wenker, Ph.D, of the University of Virginia to further research related to SUDEP in SCN8A epilepsy. Sudden Unexpected Death in Epilepsy (SUDEP) is a devastating and unfortunately all-to-common outcome for those with epilepsy. Patients with SCN8A epilepsy have heightened risk of SUDEP compared to the general epilepsy population.

> There is increasing evidence that apnea is the primary cause of death from SUDEP in many cases. In a mouse model of SCN8A epilepsy, I have shown that apnea occurs during tonic seizures and SUDEP occurs when breathing does not recover postictally. I have also found that the main breathing muscle responsible for inhalation is tonically contracted during apnea. I have now also shown that inhibition of the forebrain does not affect this apnea, and now will investigate relevant brainstem neuronal groups that regulate breathing. The identification of the neural circuitry that causes this potentially-fatal apnea will serve as targets for future SUDEP prevention therapies."
>
> —IAN WENKER, PHD, UNIVERSITY OF VIRGINIA

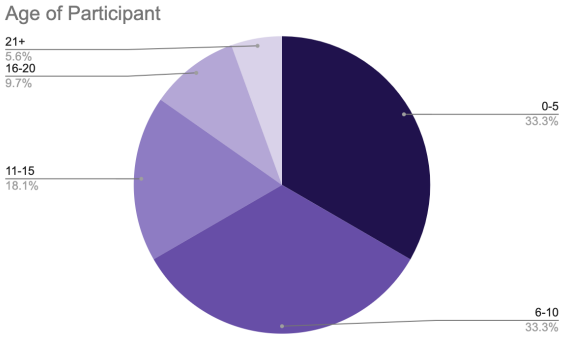
SURVEY SERIES

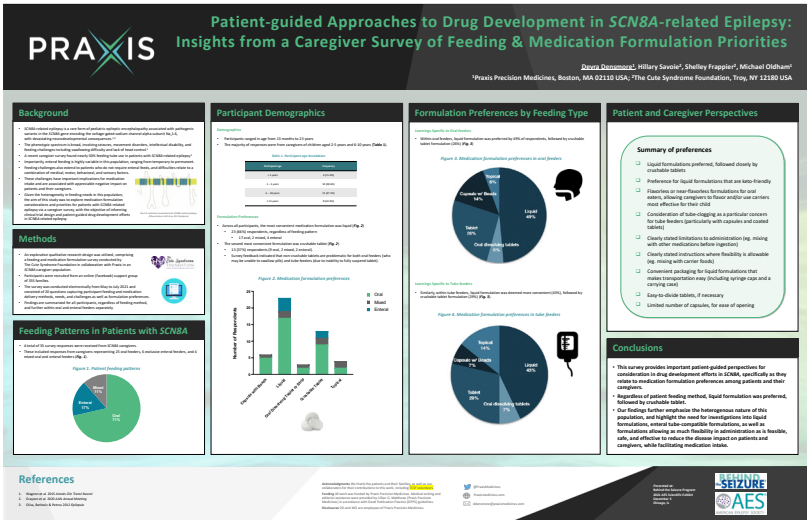
Launched in February 2021

In February 2021, TCSF launched a survey series through the NORD iamrare platform. The idea for this series came out of the tremendous success of the short Xenon survey which gathered such important data for designing upcoming clinical trials—in a fast and effective way. We needed the ability to nimbly gather information about our community in order to help focus foundation goals, clinical research, bench research, and drug development efforts. The first survey launched was in conjunction with a researcher at Children's Hospital of Philadelphia (CHOP) and looked at the relationship between Quality of Life and Seizure Burden.

The second survey focused on feeding and medication formulation with questions that our pharmaceutical partners helped us design in order to better inform their medication formulation and commercialization plans. TCSF developed a report for our partners that provided real-world feedback on how medications are actually delivered, preferences for formulation, and common concerns--as well as our recommendations.

Being certain that family needs and daily experiences are given a strong and complete voice with our partners in clinical care, research, and drug development is fundamental to the mission of TCSF. This project could not have happened without Shelley Frappier, TCSF's Executive Assistant, lifting this project off the ground! We have 10 additional IRB approved surveys ready to launch going forward!





CIITIZEN NATURAL HISTORY STUDY CAMPAIGN

Observational studies and real world evidence are critical to the drug development process. Ciitizen has developed a new method to make this process easier. The Ciitizen platform provides the following benefits to patients and families: comprehensive patient health record collected and digitized under caregiver control; a 'Personalized Disease Summary' that enables quick access to key information; easy sharing of health record and Personalized Disease Summary with diverse medical teams or to solicit second opinions; detailed analysis of SCN8A-related disorder patients to improve day-to-day are decisions; and helping to accelerate treatments for SCN8A-related disorders.

On April 1st TCSF kicked off the Ciitizen campaign to help fill the 50 Ciitizen registration spots available to our community and to their surprise, ***TCSF filled nearly all 50 in the first 24 hours***! Due to the high enrollment numbers, more slots were opened and in approximately two weeks, we recruited 87 patients total!

We all want to see our efforts lead to big things for all of our patients and children. Fortunately, the state-of-the-art technology behind efforts like Ciitizen can make our small lifts take us to big places quickly.

SCN8A GRATITUDE

We are so grateful for the support we have received this year. Your donations have allowed us to reach a milestone of over $250,000 raised! Without your help, we wouldn't be able to continue our work and make a difference in the lives of those living with SCN8A. We look forward to seeing the impact of this achievement in the coming year!

WE WOULD LIKE TO EXTEND OUR THANKS TO:

2021 Donors

Aaron Williams
Alan and Nancy Brown
Alanna Stockford
Alex Youngquist
Alexander Knee
Amy Glynn
Andrea Hanley
Angela Duren
Anthony Maroon
Ashish Sagrolikar
Bill and JoAnna
Bill Langan
Brian Caufield
Caren Caplan
Casey Speer
Catelyn Troxell
Chelsea Music Hall
Cheryl Rebsamen
Chris Howard
Christian Nubar Hovsepian
Christine Grayson
Christopher and Madeleine Burge
Cormac McNaughton
Crown Brewing
Cynthia Maddox
Darrell and Marcilyn Matson
David Cowger
David Gittens and Erika Hansen
David Young
Debbie Stroemer
Deborah Gorga
Deborah Page
Deirdre Lynch
Denis Seidalieva
Dennis Morton
Diane Doyle
Doug Aragones
Dr. Bruce and Lori Hayward
Earl Watkeys
Edmund Abdua
Ekaterina Stepanova
Eleanor Repole
Elizabeth Balk
Elizabeth Lock
Eric Laskowski
Erin Burgess
Frank Duff
George and Elizabeth Owczarzak
Gina Morton
Holly Ventura
Inna Mitelman
Jacob Mack
Jaimie Sams
Janet Eid
Jeanette Mendez Sarmiento
Jeff Silber
Jerrold and Stacy Gaza
John Russo
John Stimac
Joshua Harris
Joyce Smith
Judith Pierle
Juliann Bradish
Kathie Glynn
Kathy Couture
Keith Hymes
Kelly Ly
Ken and Liz Patin
Kenneth Balmforth
LaRita Kimes
Laugh Dot Events
Laurel Young
Lauren Dalton
Lauren Myers
Lauren Vandewalle
Liana Zee
Linda Stadler-Perry
Lisa Conticello
Lori Crawford
Lydia Toso
Makenzie Mackin
Marlene McInnes
Marvin and Gloria Bennett
Mary Corace
Mary Leffner Powell
Mary Waegner
Matt Moran
Matt Uliasz
Megan Sell
Michael Ginsberg
Michael Ramirez
Michelle Fritsch
Mike Mustillo
Monica Wolyniec
Mr. and Mrs. Jackie Black
Mr. and Mrs. William Baird
Nancy Curtin
Nina Gibson
Pamela Hart
Patricia Hayward
Paul and Carol Cuneo
Pennywise Shoppe
Pete Tuana
Phoebe McLaughlin
Pinnacle Construction Group
Rachel Klekot
Rachel Saufley
Radoslav Yankov
Red Fox Romance
Richard Leckerling
Robert and Sarah Wolfe
Robert and Susan Surowiec
Robert Hayward
Rodney and Viola Reid
Rosemary Lombardo
Ryan Corcoran
Samuel Richardson
Savannah Cliggett
Sean Connolly
Shelby Lacerte
Stephanie Noye
Stephanie Patton
Sue Johnston
Superior Signs, Inc
Susan B Hurd
Susan Hearon
Susan Refearn
Suzanne Depue
Swiders Christmas Classic
Teresa and Andrew DeYoung
Terese Hayward
Terry Jennings
Travis Kassay
Trisha Rathke
Wendie Strickler
William Minardi
William Parker
Wing Lee

Fundraiser Organizers

Kacie Craig
Nora Lyons
Karen Varner



THANK YOU, **VOLUNTEERS!**

The Cute Syndrome Foundation's work depends on all of our volunteers. Thanks to each of you, we are able to advance our mission of raising awareness of SCN8A mutations, funding the dedicated and talented scientists researching SCN8A, and supporting the families around the world who are affected by this disorder.

EXECUTIVE COMMITTEE

Hillary Savoie, PhD
Kacie Craig
Karen Varner
Liz Ramirez
Megan Varner
Shelley Frappier

BOARD OF DIRECTORS

Lan Aronson, PhD
Marianne Sutcliffe
Philip Gitlen
Ryan Pitino

2021 SPONSORS

NEUROCRINE® BIOSCIENCES

PRAXIS

GREENWICH™ BIOSCIENCES

epilepsy ALLIANCE AMERICA

ZOGENIX

GRAPHIC CREATIONS

The Cute Syndrome FOUNDATION

www.TheCuteSyndrome.com



MAKE A GIFT

thecutesyndrome.com/donate



FIND SUPPORT

thecutesyndrome.com/caregiversupport



SPREAD AWARENESS ABOUT SCN8A

thecutesyndrome.com/shop



LEARN MORE ABOUT TCSF

thecutesyndrome.com/about-us

The Cute Syndrome Foundation

PO Box 842

Ozark, MO 65721

www.thecutesyndrome.com