The Cute Syndrome© FOUNDATION
TheCuteSyndrome.com

2023 ANNUAL REPORT











Contents

3 **MESSAGE FROM OUR EXECUTIVE DIRECTOR**

4 **SCN8A COMMUNITY**
Volunteer Highlights

6 **FAMILIES HELPING FAMILIES**
Connor James Smith Patient Assistance Grant Program
Pitino Family Golf Fundraiser
All-Abilities Virtual 5K Race

10 **SCN8A FUNDING**
Finances

11 **SCN8A CONNECT**
Meaningful Change Series
Pathway to Peace
Family and Researcher Roundtable Series
Cute Connections

16 **SCN8A ENGAGED**
Advocacy and Awareness
Support Group Update
9th Annual Clinician, Researcher, and Family Gathering

18 **SCN8A RESEARCH**
2023 SCN8A Seed Grant

19 **SCN8A GRATITUDE**
2023 CUTIES Awards
Donor Recognition
Volunteers
Executive Committee
Board of Directors

Empowering SCN8A Families: **From Surviving to Thriving**

It is a privilege to address you as the Executive Director of The Cute Syndrome Foundation (TCSF) and as a mother to two wonderful children, Lincoln and Stella. My journey, like many of our families, has been profoundly influenced by the diagnosis of SCN8A. My daughter, Stella, was diagnosed when she was just 5 months old. This experience has shaped my family and strengthened my resolve to support those affected by SCN8A.

This year, TCSF's Meaningful Change Educational Series focused on the theme "From Surviving to Thriving". This theme has been the foundation of our efforts, symbolizing not just the struggles we face, but the hope and determination to see a brighter future for all families impacted by SCN8A.

Thanks to the generous support of our donors, we were able to turn this vision into reality through a series of impactful programs and initiatives. One of our significant achievements this year was securing a Global Genes Mental Health grant, which allowed us to train four volunteers in the Grief Recovery Method. This training enabled us to offer personalized grief recovery courses, both individually and in groups, to our community. Additionally, TCSF supported three CUTE Connection family meet-ups in Branson, Missouri; Atlanta, Georgia; and Milan, Italy, bringing together over 125 family members for meaningful connections and support.

We also sponsored eight SCN8A parents to participate in a specialized 6-month life coaching course called "Pathway to Peace" led by life coach, Kara Ryska. This program was designed to foster personal growth and provide support for parents caring for children with special needs.

Lastly, in collaboration with Krista and Shawn Smith, we established the Connor James Smith Patient Assistance Grant program. This grant is available to *all SCN8A families* and continues to make a significant impact, honoring the legacy of their son, Connor, who passed away in 2022.

Thank you for your unwavering support and dedication. Together, we are making a profound difference in the lives of SCN8A families. Your contributions have directly empowered our community to shift from merely surviving to truly thriving.

Warm regards,

Kacie Craig

Kacie Craig
Executive Director



Volunteer Highlights

TCSF believes that it's important to shine a spotlight on individuals who are making a tangible difference within our community.

Nora Lyons, mom of Blake, has been an invaluable asset to TCSF. This year, Nora spearheaded the organization of TCSF's first-ever virtual 5K, a resounding success that united friends, families, and communities around the world. Her dedication extends beyond single events; Nora is always ready to lend a helping hand as a TCSF volunteer throughout the year, and she is especially active at our annual gatherings, often with Blake by her side. Nora's unwavering commitment and enthusiasm embody the spirit of our community, and we are deeply grateful for her continued support.



I volunteer with TCSF because it helps me give back to a community that has given me so much. I have made true friends through volunteering and participating in the foundation's programming. I love finding new ways to connect the community and let others see the bright, joyful life our kids have."

Nora Lyons
Blake's Mom







Madison Whigham, Mia's mom, has been a dynamic force within TCSF. This year, she played a crucial role in fostering connections by facilitating family introductions in our SCN8A Facebook family support group. Madison represented TCSF at numerous Florida events, showcasing her dedication and advocacy. Her entire family contributed significantly at the annual gathering, embodying the spirit of community and support. Additionally, Madison helped organize a successful Cute Connections family meet-up event in Atlanta, Georgia, and creatively fundraised for the gathering by making SCN8A friendship bracelets. Madison's tireless efforts and innovative contributions are invaluable to our mission, and we deeply appreciate her unwavering support.



TCSF was so helpful to my family and I when we first got Mia's diagnosis, so I wanted to be able to give back. TCSF does so much for the SCN8A community and I know when I volunteer that it's supporting a good cause. It's also nice that I've been able to make friendships along the way and meet people that I may not have been able to meet in person otherwise."

Madison Whigham
Mia's Mom







Spreading Joy through the **Connor James Smith Patient Assistance Grant Program**

Families helping families is at the heart of the Connor James Smith Patient Assistance Grant Program, which aims to spread joy and provide much-needed support within the SCN8A community. In many cases, insurance covers only the bare minimum for patients with rare genetic disorders like SCN8A. This often leaves families with the financial burden of paying for necessary medical equipment, therapy devices, educational aids, medications, outpatient therapy costs, and home and vehicle modifications.

These out-of-pocket expenses can quickly add up, forcing caregivers to make difficult choices, such as choosing between life-saving medications or necessary medical equipment and sacrificing their physical health due to the inability to afford ADA-accessible home modifications. The Connor James Smith Patient Assistance Grant seeks to alleviate these financial burdens, allowing families to focus on their well-being and enjoy quality time together.

The program's inception is rooted in a heartfelt promise made by Krista and Shawn Smith to their son Connor, a 12-year-old boy who lived joyously despite his significant challenges due to his SCN8A diagnosis. Connor's life was filled

with joy, from watching the Chicago Cubs at Wrigley Field to flying through the Grand Canyon in a helicopter. His bright smile and gentle demeanor touched everyone he met. Upon his passing in December 2022, his parents vowed to honor his legacy by spreading joy and being a blessing to others. They began by donating Connor's special needs and medical equipment to local families and soon reached out to TCSF to create a grant in Connor's name.

Krista and Shawn envisioned a grant with few barriers and a global reach. With the support of TCSF, the vision for the Connor James Smith Patient Assistance Grant quickly became a reality. The foundation matched the grant, and with contributions from friends and family, the program came together in just a few short months.

The Connor James Smith Patient Assistance Grant has already begun to spread joy and make a positive impact on the SCN8A community. By easing the financial burdens faced by families, the program allows them to focus on what truly matters: their loved ones and the precious moments they share. Krista and Shawn Smith's promise to their son Connor continues to be fulfilled, as his legacy of joy and generosity lives on through the grant program, bringing hope and happiness to families in need.

> The 3 week intensive therapy session that the CJSPAG sponsored for us helped McKenzie gain strength, coordination, and balance that she cannot get anywhere locally. In the 3 week session, she made more progress than she has made in a full year of conventional once-weekly therapies."
>
> **Matt Uliasz**
> McKenzie's dad





Connor James Smith



Pitino Family Golf Fundraiser

In June, the Pitino Family hosted a remarkable Charity Golf Outing benefiting TCSF. It was a day full of golf and an evening including a cocktail hour, dinner, live performance by the Silver Arrow Band, and silent auction, all held at the Harrison Meadows Country Club in Harrison, NY. Between golf registrations, ticket sales, the silent auction, and direct donations, this event **raised over $100,000** for TCSF. The event was organized by TCSF Treasurer of the Board Ryan Pitino and Maura Pitino. The SCN8A community was represented at the Golf Outing by their son, SCN8A warrior Luke.

Due to the success of the Golf Outing, TCSF was able to expand the 2023 Dr. Mandy Harris Family Travel Grant, which assists families with travel costs to attend the Annual Clinician, Researcher, and Family Gathering. Thanks to the Pitino family's efforts, every 2023 applicant was awarded a Dr. Mandy Harris Family Travel Grant, leading to record family attendance at the Annual Gathering in Orlando, FL.

The Cute Syndrome Foundation extends a special thank you to The Daniel Pitino Foundation for their generous support through this event.



All Abilities **Virtual 5K Race**

In 2023, SCN8A mom Nora Lyons presented the idea for a Virtual 5K Race, a unique event that would bring together the global SCN8A community through shared activity while raising funds for The Cute Syndrome Foundation. **This idea grew into TCSF's First Annual All-Abilities Virtual 5K, celebrating the resilience of the global SCN8A community, including affected individuals, caregivers, and their support systems.**

Over 110 supporters from around the world registered to run, walk, roll, or stroll 3.1 miles in support of TCSF. The virtual race platform offered plenty of flexibility for SCN8A families. Participants were encouraged to complete their race any time from April 16th through April 23rd and could do so outside or on a treadmill. Many participants completed their race on their own, while others gathered with friends and family to raise awareness together!

Congratulations to our fastest finishers, **Tyler Younquist** (male - 18:38), **Mendy Rucker** (female - 28:32), and **Henry Youngquist** (assisted wheelchair/stroller - 18:37). Thank you to Nora for organizing this event and to everyone who participated and made TCSF's inaugural virtual race a success.

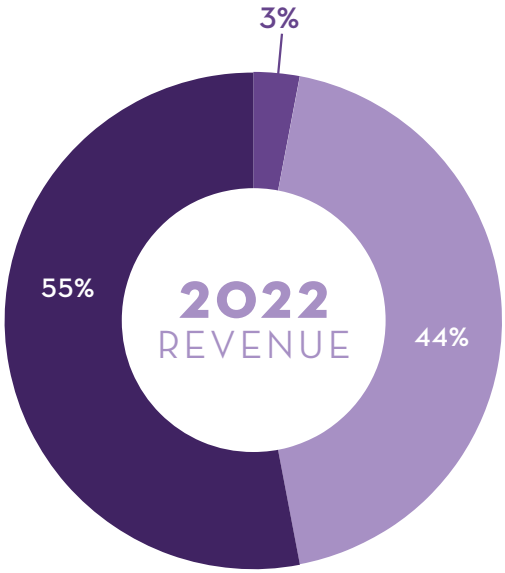
"I wanted to do something active and fun that folks could do in their community to raise awareness and funds. A virtual race meant everyone could participate alone, in groups, or as a big gathering. I'm so proud of what my idea became. Next year will be even better."

Nora Lyon's
Blake's Mom, TCSF Volunteer and Event Organizer

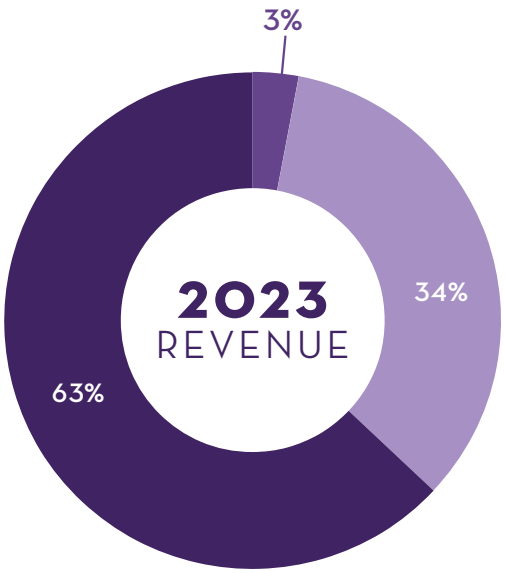
"The first TCSF virtual race was so much fun especially since we could do it together with 2 other SCN8A families. We were able to rent a pavilion at a local park with walking trails, and gathered family and friends to join us. Just a wonderful way to spread awareness and raise money for a great cause!"

Karen Varner
Lily's Mom, Family Support Director

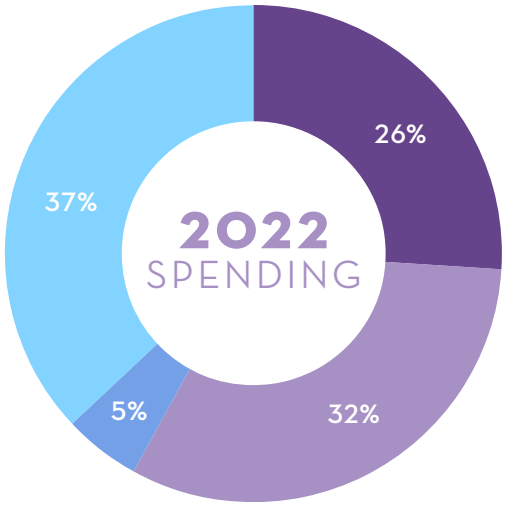
A Look at
2022 and 2023 Finances



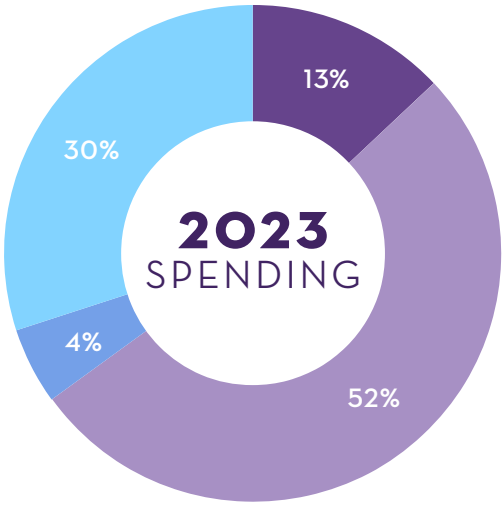
- FOUNDATION SUPPORT
- CORPORATE SPONSORSHIP
- INDIVIDUAL DONATIONS



- FOUNDATION SUPPORT
- CORPORATE SPONSORSHIP
- INDIVIDUAL DONATIONS



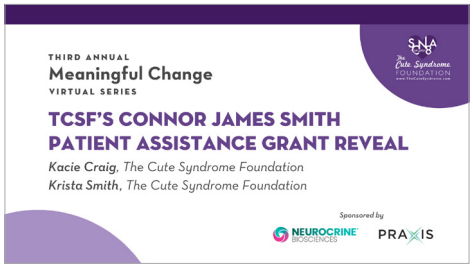
- RESEARCH
- AWARENESS AND FAMILY SUPPORT
- OPERATING
- FAMILY ENGAGEMENT AND EDUCATION

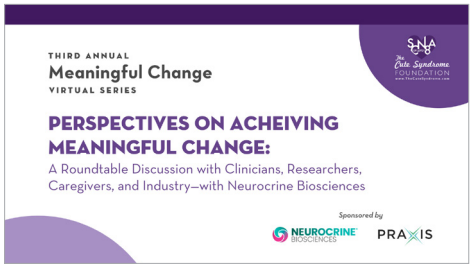


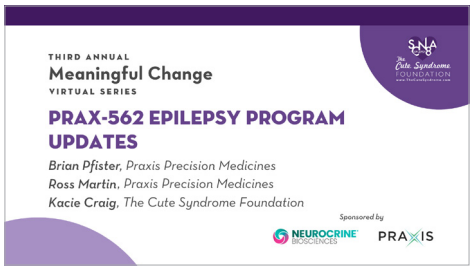
- RESEARCH
- AWARENESS AND FAMILY SUPPORT
- OPERATING
- FAMILY ENGAGEMENT AND EDUCATION

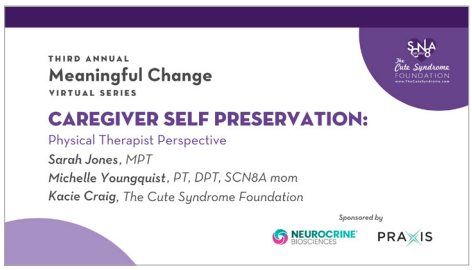
3nd Annual
Meaningful Change Series

The Meaningful Change series looks at the ways in which the knowledge found within the SCN8A community can be leveraged to better support all of our children. **This year's theme was "Surviving to Thriving."** It covered diverse topics such as TCSF's new Connor James Smith Patient Assistance grant, Praxis Precision Medicines's epilepsy program updates, caregiver restoration and self-preservation with life coach Kara Ryska, perspectives on meaningful change with Neurocrine Biosciences and other professionals, and travel tips for caregivers of individuals with SCN8A-related disorders. These discussions emphasized the journey from mere survival to flourishing in various aspects of caregiving.

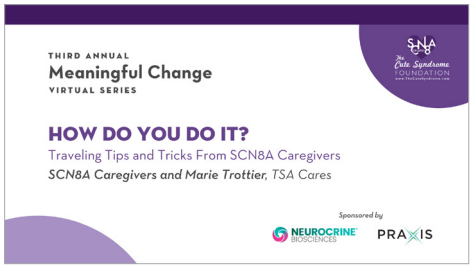












200 *total series views*

38

views of "CJSPAG Reveal"

59

views of "Praxis 562 Epilepsy Program Updates"

16

views of "Restoration Roadmap"

44

views of "Perspectives on Achieving Meaningful Change"

10

views of "Caregiver Self Preservation"

33

views of "How Do you Do It? Traveling Tips and Tricks from SCN8A Caregivers"

Pathway to Peace

This year, TCSF began a partnership with life coach and podcaster Kara Ryska, from the Special Needs Mom Podcast, to offer a group coaching series exclusively for SCN8A parents. The first 8 parents completed a six-month group coaching program in November. TCSF was able to cover the majority of the enrollment costs for these parents.

> I've done therapy of all sorts as well as coaching the majority of my life, but since I've had my medically fragile daughter nothing seemed to fit. Kara's approach for special needs moms was exactly what I needed. It was like my traditional coaching but specifically for moms like me. It was also great to identify with other mothers like me in the group. I can't recommend it enough for special needs moms."
>
> AN SCN8A MOM WHO PARTICIPATED

1st Annual
Family and Researcher Roundtable

TCSF strives to break down barriers between the scientific and patient communities and empower the SCN8A community through knowledge. In November, we held a Family & Researcher Roundtable Series, bringing top SCN8A researchers and families together virtually to discuss some of caregivers' most commonly asked questions. Topics included inheritance and family planning, emerging treatments for SCN8A, and how researchers are working to characterize SCN8A phenotypes.

163

total series views

73

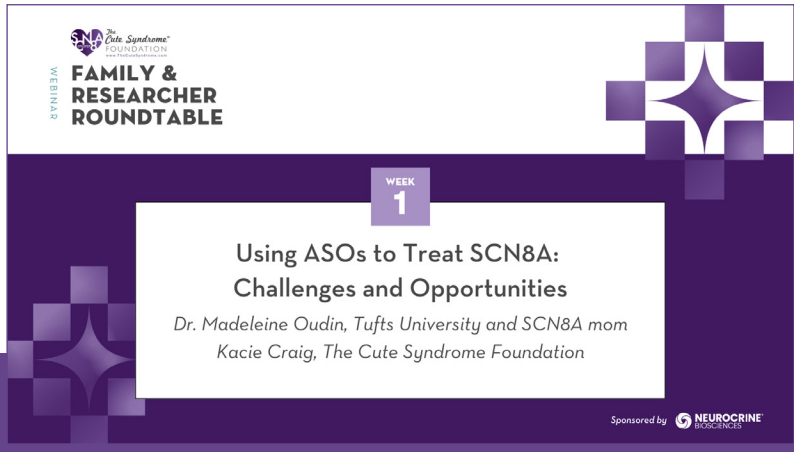
views of "Using ASOs to Treat SCN8A: Challenges and Opportunities"

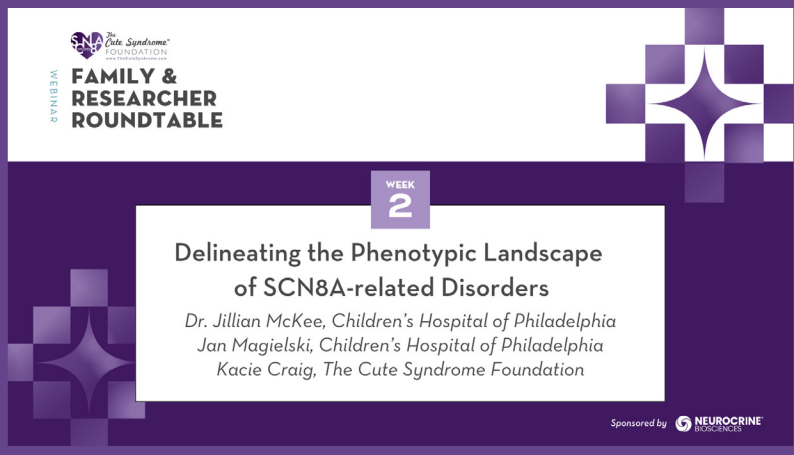
32

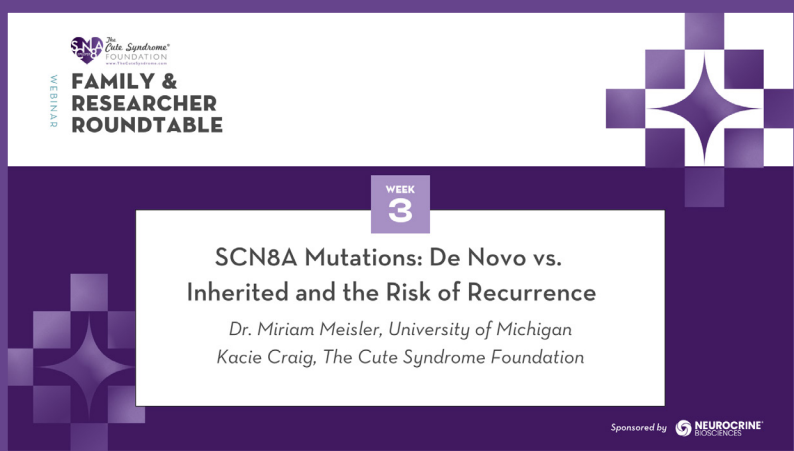
views of "Delineating the Phenotypic Landscape of SCN8A-related Disorders"

58

views of "SCN8A Mutations: De Novo vs. Inherited and the Risk of Recurrence"

















Cute Connections

Although providing year-round virtual activities is imperative to our families because we are far too often isolated, even in the best of times, TCSF recognized an urgent need for families to engage in face-to-face activities. The pandemic has left our families suffering from extreme social isolation and loneliness and requesting in-person fellowship activities in addition to virtual events.

The goal of Cute Connections family activities is to, first and foremost, give our community a worry-free opportunity to give and receive actual hugs, spend time catching up, and offer each other moral support. We want to provide a safe and accessible environment for our kids to do what they do best, with minimal stressors for parents.

Overall, 2023's CUTE Connections created opportunities for SCN8A families, **totaling over 125 people**, to enjoy a bit of summer fun and respite from the day-to-day stressors our families typically face when living with an SCN8A diagnosis. The locations of the 2023 CUTE Connections included **Atlanta, Georgia**; **Branson, Missouri**; and **Milan, Italy**.

> ***We are very fortunate that The Cute Syndrome is so focused on supporting the mental health of the families of children with SCN8A. It was such a nice change of pace to spend time with other families sharing stories of both trials and triumphs while also doing fun activities."***
>
> **-MEGAN GARD, CLAYTON'S MOM**

Advocacy and Awareness

Throughout the year 2023, TCSF actively engaged in advocacy and awareness efforts through participation in conferences, speaking engagements, and awareness events. These initiatives provided valuable opportunities for networking and collaboration with other advocacy groups, professionals, and families affected by SCN8A.











> In 2023, I applied for and received a travel grant through the Danny Did Foundation to attend the Partners Against Mortality in Epilepsy (PAME) Family Day and Conference. PAME is a community of individuals and organizations that come together to increase awareness and promote the prevention of premature death related to epilepsy. The event's Family Day session was a convening of families and advocates, while the conference day had a scientific focus that was informed by the experiences of people impacted and bereaved by epilepsy.
>
> Attending this event was impactful on both a personal and professional level, as my son, Will, lived with uncontrolled epilepsy that was caused by an SCN8A mutation and passed away due to sudden unexpected death in epilepsy (SUDEP) at the age of 7.

Liz Ramirez
Grants Advisor and
Bereavement Support

The Cute Syndrome Foundation's **Family is Growing**

Founded to offer support, guidance, and a sense of community for families dealing with SCN8A-related conditions, TCSF's Family Support Group saw significant growth by the end of 2023. With 501 members from 53 countries and families in 42 states across the U.S., the group has evolved into a global center for support and solidarity. Through online platforms, meet-ups, and resource-sharing initiatives, the group continually promotes a sense of belonging and empowerment for individuals and families affected by SCN8A-related conditions around the world.



OVER 500 MEMBERS

REPRESENTING 53 COUNTRIES

WITH MEMBERS IN 42 US STATES



9th Annual
SCN8A Clinician, Researcher, and Family Gathering

The Cute Syndrome Foundation's annual gatherings play a pivotal role in keeping the Foundation deeply engaged with the SCN8A community. These events, such as the transformative 9th Annual Gathering in Orlando, serve as a hub for families, clinicians, researchers, and industry leaders to unite in a shared journey of discovery, unity, and resilience. Through insightful presentations on the latest advancements in SCN8A research and treatments, heartfelt family narratives, and enriching industry insights, TCSF fosters a sense of camaraderie and mutual support. These gatherings not only illuminate the path forward for SCN8A but also celebrate the strength and unity inherent within the community, ensuring that TCSF remains at the forefront of advocacy, education, and support for those affected by SCN8A.





SCAN TO VIEW MORE PHOTOS FROM THE EVENT!

2023 SCN8A **Seed Grant**



Chris Dulla, PhD
Tufts University

Chris Burge, PhD
Massachusetts Institute of Technology

Madeleine Oudin, PhD
Tufts University

The Cute Syndrome Foundation was excited to announce the awarding of an SCN8A seed grant to Chris Dulla, PhD, and Madeleine Oudin, PhD, of Tufts University, along with Chris Burge, PhD, of MIT for "Validating a new mouse model of SCN8A infantile spasms and epilepsy." This grant has been supported in partnership with the **American Epilepsy Society** and the **Cute Syndrome Foundation**.

These funds will support PhD student Haley Dame and her work in characterizing the new SCN8A mouse model their lab has developed, as well as testing out the impact of disease-correcting Antisense oligonucleotides (ASOs) for SCN8A.

This project holds a special significance for Dr. Oudin and Dr. Burge as their daughter Margot is affected by SCN8A epilepsy.

Dr. Oudin said of their work: ***"We are really excited to be starting new SCN8A research at Tufts University. While Prof. Chris Dulla has been studying epilepsy for 20 years, he has never worked on ion channel-based epilepsies. The AES Seed grant gives him the opportunity to bring his extensive knowledge and expertise to the SCN8A community."***

2023 CUTIES Awards

After the first SCN8A Clinician, Researcher, and Family Gathering, held in 2015, TCSF saw a handful of individuals rising to the opportunity to work collaboratively to improve the health and safety of those living with SCN8A. **In recognition of that work, we created the CUTIES, our annual awards for Champions for Understanding, Treating, Investigating, and Empowering those with SCN8A.**



Dr. Scott Perry

Cook Children's Hospital— Medical Director, Neurology; Medical Director, Genetic Epilepsy Clinic

Dr. Perry is widely respected for his compassionate care and deep knowledge of SCN8A-related epilepsy, drawing families from across Texas. Parents like Lacey Juroske, whose daughter Sadie has been seizure-free for four years under his care, credit his expertise with transforming their children's lives. Dr. Perry is celebrated not only for his clinical skill but also for his heartfelt dedication to SCN8A families.



Dr. Madeleine Oudin

Assistant Professor, Tufts University and Margot's Mom

Dr. Oudin originally focused her research on cancer. After her daughter Margot was diagnosed with SCN8A, she shifted her lab's focus to SCN8A research. For the past 1.5 years, she has concentrated on developing ASO therapies targeting alternative splicing to treat SCN8A epilepsy. Dr. Oudin is committed to advancing research and actively engages with the SCN8A community to share knowledge in an accessible way.



Shawn and Krista Smith

Connor James Smith Patient Assistance Grant Founders and Connor's Parents

Krista and Shawn Smith have truly demonstrated exceptional dedication to our SCN8A community. They have become steadfast advocates for families facing obstacles in accessing essential specialized equipment. Understanding the difficulties that arise when insurance and other resources fall short, they created the Connor James Smith Patient Assistance Grant in memory of their son, Connor. Through this grant, they continue to provide critical support where it's needed most.

A Heartfelt Thank You **to Our Generous 2023 Donors**

Aaron Williams
Alex Youngquist
Allison and Troy Donohue
Alonzo Guess
Ashish Sgrolikar
Ashley Flynne
Barbara Hallberg
Bonnie Johnson
Brad Gianiny
Brenda Beck
Brian Bachman
Brian Lasky
Brian Pizzitola
Casey Youngflesh
Catelyn Troxell
Catherine Banks
Catherine Bave
Charles Moran
Chelsey Hanneken
Cheryl Gallus
Chris Giordano
Christina dupre
Christopher Padden
Clay Courville
David and Karen Varner
David Bauer
DeAnna Daniels-Fischer
Denise Dance
Devon and Tom Hosford
Edward Detmer
Elizabeth Monaghan
Erin Jones
Erin Mara
Fazlin Ahlgren
Francine dion
Frederick Adair
Gillian Minardi
Hannah Wingo
Heather Grant
Héloïse Jannin
Indri Rudy
Isabelle Hauswald
Jacqueline Barnett
Janice Miller
Jenny Hadiaris
Joe and Rosemary Ranson
Joshua Sell
Joyce Purkey
Judith Avery
Julie Anne Lewis
Katherine Roome
Kevin Del Mauro
Kevin Hubschmann
Kimberly Overton
Kris Kahlig
Leigh Harrington
Lisa Gidaro
Lisa Jones
Loraine Hughes
Lori Hayward
Lori Wilson
Lydia Toso
Maite Schenck
Margaret Toriello
Marie Bundhoo
Marie Duncan
Marisa Antonelli
Mark Curtin
Mary Holly
Matthew Ott
Matthew Wright
Megan Sell
Michael Gann
Michael Leach
Michael Pizzitola
Michelle Parnes
Mike Wrightson
Nick Pinto
Patricia Sowers
Pricilla Merryman
Richard and Barbara Leckerling
Richard Bosman
Richard Pitino
Rick Avare
Rick Pitino
Rickie Le
Rob Minardi
Ryan Pitino
Samantha Oberkramer
Sayde Dunlap
Scott Maddox
Sophia Sattar
Stacey Gagnier
Stefan Hrdina
Stephanie Noye
Stuart Jackson
Summer Kotkin
Susan Allen
Susan Hurd
Susan Watson
Terese Hayward
The Hayward Family
Theresa Banks
Thomas O'Grady
Thomas Preston
Todd Johnson
Tyler Duke
Valintina Munoz
Victoria Hight
William Minardi



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

DANIELLE HAYWARD
Director of Fundraising

KACIE CRAIG
Executive Director

KAREN VARNER
Family Support Director

LIZ RAMIREZ
Bereavement Support and Grants Advisor

MEGAN VARNER
Marketing Director

SHELLEY FRAPPIER
Director of Patient Engagement and Data Management

Board of Directors

HILLARY SAVOIE, PHD
Founder and President

LAN ARONSON, PHD
Vice Secretary

MARIANNE SUTCLIFFE
Secretary

PHILIP GITLEN
Vice Chair

RYAN PITINO
Treasurer



The Cute Syndrome Foundation
PO Box 842
Ozark, MO 65721

www.thecutesyndrome.com