SCN2A RELATED AUTISM & EPILEPSY
FamilieSCN2A
FOUNDATION
WWW.SCN2A.ORG

Welcome to our 2nd Quarter Newsletter! See what we've been up to...

A message from our Executive Director...

While it is difficult to find a silver lining to the Covid19 pandemic, we are learning a lot about adaptability. In the world of parenting kids with complex medical needs, pivoting is nothing new. The FamilieSCN2A Foundation is led by several of these parents so when it came time to jump to plan B, then C, we did so with speed and grace.

It started with evaluating the needs of our community. Many reported jobs lost, schools closed and in-home care halted. The worry of losing our children escalated because of the virus and the disruption to continuity of care. From experience, I can tell you that lack of sleep due to caring for a sick child, without help, and with the added stress of the unknown is a recipe for disaster.

Our first goal was to remind our community that we are here to help so we began hosting virtual meet ups with an open forum for dialogue. Just talking to others who are in similar situations can be therapeutic. The calls have been a huge success with more than 50 families joining in from all over the world.

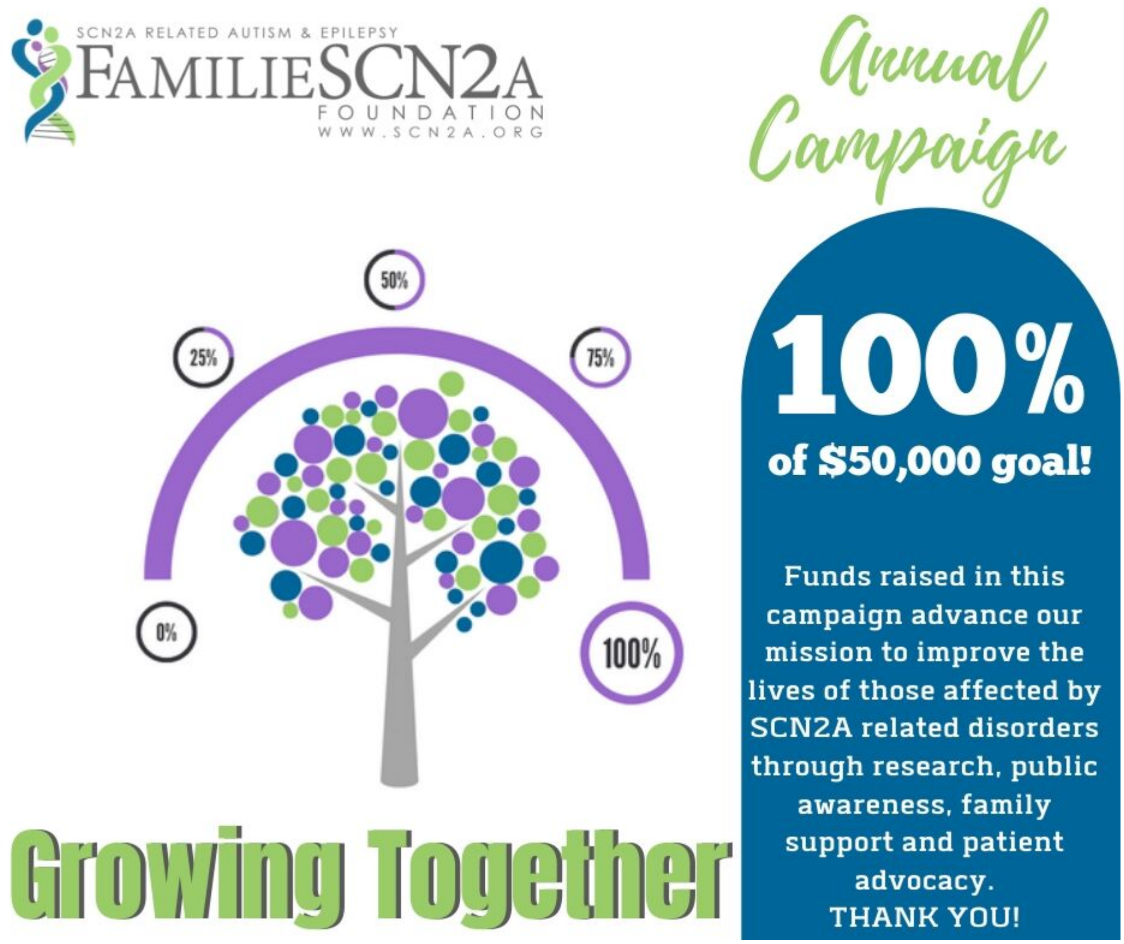
Then, we tackled the financial aspect of supporting SCN2A families. With the assistance of our accounting team, we quickly adjusted our budget and launched the Covid19 Financial Relief Grant. To date, we have awarded immediate assistance to 20 families from around the globe. Thanks to our generous donors and sponsors, we have expanded the program to offer a second round of these grants.

Having no choice but to cancel in-person fundraisers and events planned for this year, we will have a gap in our earnings, but we plan to stay flexible and continually reevaluate to ensure we are making progress toward our vision: A cure for all SCN2A disorders. The Board of Directors and our small, but fierce community are

> *banding together to hold one another up during this difficult time. Please reach out if there is anything we can do for you. ~ Leah*

FAMILY INPUT NEEDED! As you know, due to the public health crisis caused by COVID-19, our Family Conference has been postponed until next summer. In an effort to keep you informed and engaged, we will host a **virtual conference event** at a date TBD. **PLEASE CLICK BELOW** for a quick survey of topics you would like to see presented. Thanks! We value your input.

Virtual Conference Survey



Funds raised in this campaign advance our mission to improve the lives of those affected by SCN2A related disorders through research, public awareness, family support and patient advocacy.
THANK YOU!

The 2020 Annual Campaign was a success! The need for financial assistance among families is greater than ever right now as is the need to

continue our research initiatives. We are grateful to be in a position to do both. Click here to hear how you have impacted the SCN2A community: Heartfelt thanks to our donor and sponsors!

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Allies in Rare Disease

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BIOSCIENCES

PRAXIS

Supporting the global SCN2A community is a key part of our mission.
Apply here for emergency funding for SCN2A families in great need.

All SCN2A Families: the *Patient Assistance Grant* is available to help you purchase equipment and cover therapy costs denied by insurance. **Apply Today!**

We are hosting a series of virtual "Chats" via Zoom to provide a forum for community connection. Families affected by SCN2A are invited to join for informal conversation with others who understand. These calls are not intended to provide medical advice. ***The next 'Chats' are Saturday, June 20 at 12 noon ET and Tuesday, June 30 at 10:00 pm ET.*** *Please check social media posts or email community.support@scn2a.org for the Zoom link and future Chat schedules.*

RESEARCH

SCN2A in the news

"SCN2A Channelopathies in the Autism Spectrum of

Neuropsychiatric Disorders: A Role for Pluripotent Stem Cells"

(Kruth, Grisolano, Ahern, Williams,

Molecular Autism, volume 11, Article number: 23 (2020))

"Biological concepts in human sodium channel epilepsies and their relevance in clinical practice."

(Brunklaus A, Du J, Steckler F, et al., Epilepsia, 2020; 61-387-399)

We caught up with a busy Chris Ahern, PhD, as he was reopening his University of Iowa Lab after the Covid19 hiatus and preparing for an upcoming meeting with the NIH. He's excited to get back to working with his team in person and seems undaunted by the challenge of having to do some things differently now in order to keep everyone safe. Be sure to check out the Ahern Lab's refreshingly clever website.

Besides writing grants, zoom lab meetings and virtual happy hours, the pause in the usual daily routine gave Chris precious time to think more deeply about his current projects, reflecting on how far they can go and what steps it takes to get there. To escape the seemingly endless screen time, Chris enjoys cycling, something he used to do competitively but is now a pleasant escape. “It’s a great way to clear the mind, help work off those baking projects with the kids and think about science."

A self-described ‘biophysics geek,’ Chris notes that science often leads you down a path and you tend to stay in your lane but sometimes you have to look at new opportunities. While using transfer RNAs to study *how* sodium channels work, his lab saw the possibility of using them to *fix* premature termination codons (which has been associated with autism spectrum disorders). His team is trying to develop a new kind of gene therapy–a way to give a one-time dose of a small molecule that could heal a genetic defect. Their approach looks not at a specific gene, but at the type of defect, specifically those caused by premature stop codons. In the academic lab they work on building the datasets and the technology with the goal of getting it to a biotech company who can accelerate drug development.

A big step towards that goal came in 2019 with a Simons Foundation SFARI Program grant to fund work using SCN2A patient iPSC (induced pluripotent stem cells). Along with Iowa collaborator Dr. Aislinn Williams and her lab, this support allows both labs to focus their energy on figuring out ways to deliver possible new therapies to neurons. "It's an incredibly exciting project because it draws on our long-standing interest in sodium channel research."

Chris attended our Family and Professional Conference in Seattle last summer and took that positive energy back to his lab. “As scientists we don’t often get to meet the patients that our research may eventually help and it’s so inspiring to know who we are working for.” Chris could tell by speaking with some of our families that there is some healing in simply knowing that they are not being ignored, that there are people working hard towards a cure.

Being a scientist in the academic world often means living a vagabond life, but Chris, his wife and two children have been settled happily in Iowa for the past 9 years. Originally from the Midwest, Chris has lived near both coasts and in the Southwest, but enjoys the quality of life in a college town without a lot of traffic. “It’s a great place to live because I have time to focus on the science.”

Chris shared his #QuarantineJoy: Spending time with the family; riding Iowa gravel roads and single track; and marveling at his daughter taking virtual ballet lessons in the living room.

GREATNONPROFITS
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A SIMPLE WAY TO HELP:

Please [click here] to share your good stories about The FamilieSCN2A Foundation and help increase our visibility in the giving community. Let donors, volunteers, and beneficiaries know how you celebrate our work! These reviews will be shared on Foundant, Guidestar, GlobalGiving and JustGive where thousands of donors do their own research on how much to give and to whom.

Donations Welcome