CACNA1A
FOUNDATION

YEAR IN REVIEW
2022

OUR MISSION IS TO FIND SPECIFIC TREATMENT OPTIONS AND A CURE FOR CACNA1A PATIENTS BY BUILDING A COLLABORATIVE NETWORK OF PATIENTS, FAMILIES, CLINICIANS AND SCIENTISTS THAT WILL WORK TOGETHER TO RAISE AWARENESS AND ACCELERATE THE UNDERSTANDING, DIAGNOSIS AND TREATMENT OF CACNA1A-RELATED DISEASES.

HIGHLIGHTS

FEB

Recipient of Orphan Disease Center Grant for Novel Mouse Model Development
The Jackson Laboratory

JUL

First Hybrid Creating Connections Community Conference
San Antonio, TX

SEP

First In-person Research Roundtable
New York City

CACNA1A BY THE NUMBERS

1 COLLABORATIVE RESEARCH NETWORK

37 RESEARCHERS AT 1ST RESEARCH ROUNDTABLE

59 INDIVIDUALS ENROLLED IN RARE-X

150 NATURAL HISTORY STUDY PARTICIPANTS

189 FAMILIES IN OUR CONTACT REGISTRY

200 ATTENDEES AT OUR CACNA1A CONFERENCE

500 5K RUN, WALK, AND ROLL PARTICIPANTS

3000 SOCIAL MEDIA FOLLOWERS

FAMILY SUPPORT AND RESOURCES

1.

Developed Hemiplegic Migraine Resource Library

These resources are based on the published literature and experiences of our clinicians and families. While CACNA1A mutations are associated with Familial Hemiplegic Migraines, data on the clinical care of hemiplegic migraines is quite limited, especially in the pediatric population. Our resources serve as a guide (i) for clinicians to assist with their treatment decisions and discussions with CACNA1A patients and caregivers, both in the emergency and clinical settings, and (ii) for patients and caregivers to prevent and prepare for hemiplegic migraine-related emergencies.
https://www.cacna1a.org/hemiplegic-migraine

2.

Produced 5 Therapy-Focused Webinars in Partnership with DEE-P Connections

In partnership with Theresa Spong, M.ED, PT, CBP, Amanda Luddeke, MS, SLP, CCC, and our friends at DEE-P Connections, we launched a 5-part therapy-focused webinar series focused on (i) impacts of hypotonia on overall development, (ii) unraveling the mysteries of ataxia, hypertonicity, athetosis, and mixed tone, (iii) adaptive equipment and therapy strategies for children with severe developmental delays, (iv) low-tech/no-tech communication systems for children with complex communication needs, and (v) high-tech options for alternative communication and access within the home.
https://www.cacna1a.org/webinar-series

3.

Held 12 Virtual Monthly Meetups for Patients, Parents & Caregivers

We held monthly virtual gatherings for CACNA1A families around the world to share stories, ask questions and create a sense of community. A rare disease diagnosis can leave one feeling exhausted, overwhelmed and socially isolated. These meetings provided a crucial virtual support system for our community.

4.

Awarded 14 travel scholarships to families to attend our first in-person Creating Connections Community Conference in San Antonio, Texas

This gathering provided an opportunity for families to meet in person for the first time after two years of creating deep personal bonds over virtual forums. It was also an opportunity for leading clinicians and researchers to share their work, answer questions, and connect with the true CACNA1A experts - our families!

DRIVING SCIENCE FORWARD

5.

Added Science Engagement Director, Pangkong Fox, PhD

The Rare As One grant we received in 2021 from the Chan Zuckerberg Initiative enabled us to add Pangkong Fox, PhD to our leadership team to serve as our Science Engagement Director. Her passion for advancing our mission is fueled not only by her background in cell & molecular biology but also as a mother to a son who has a CACNA1A variant. As a liaison between the scientific community and the Foundation, Dr. Fox is adept at conversing with industry and academia alike, yet she is also skilled at breaking down scientific concepts into easy-to-understand language for our families.

6.

Expanded our Scientific Advisory Board

This year we added two international researchers to our SAB - Anne Ducros, MD, PhD, and Zam Cader, DPhil, MRCP. Dr. Ducros is a renowned physician-scientist from France who received her PhD in the genetics of hemiplegic migraine in 1998. Since then, she has authored over 168 publications on the subject. Based in the UK, Dr. Cader focuses on pain/migraine, Alzheimer's disease and autism/epilepsy. Using human iPSC brain models, his research aims to develop resources and tools to improve drug discovery for these neurogenetic disorders.

7.

Continued to build our preclinical toolbox

To design efficient clinical trials, there must be validated clinical research tools and knowledge of the natural history of disease, referred to as clinical trial readiness. To optimize the clinical trial readiness of CACNA1A, this year, we (i) grew our Natural History Study enrollment to 150 individuals; (ii) collected blood samples from 12 individuals for the development of iPSC (stem cell) lines for our biorepository, to help researchers study the mechanisms of disease; and (iii) collaborated with scientists to functionally characterize variants to understand their impact on the Cav2.1 channel.

8.

Funded the development of a CACNA1A variant database accessible to families, clinicians & researchers

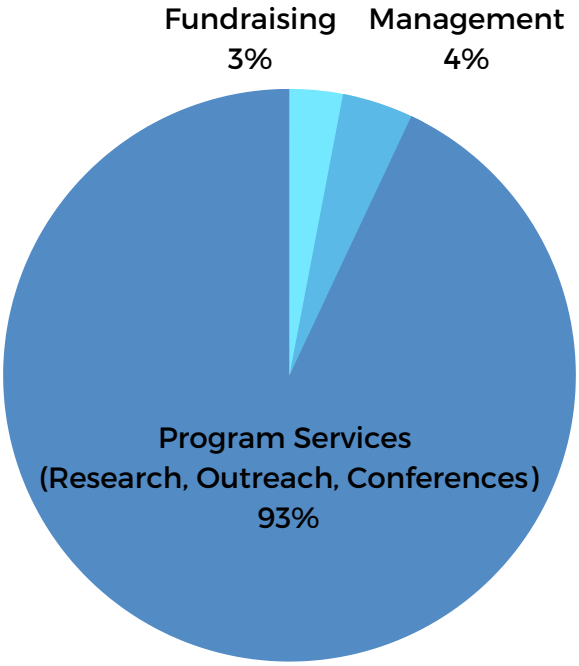
In collaboration with Dennis Lal, PhD, we developed an interactive website for families, clinicians & researchers dedicated to comprehending CACNA1A disorders. It is an "open science" portal, accessible by anyone, and includes comprehensive information on CACNA1A variants, educational resources and natural history study data. The database will launch to the public in 2023.

FINANCIALS & TRANSPARENCY

The CACNA1A Foundation is dedicated to driving science toward new therapeutics and ultimately, a cure for every person affected by a CACNA1A variant. In 2022, we raised $543,000 through grassroots fundraising, grants and sponsorships.

Our work would not be possible without the generous support of our donors. We are an all-volunteer board and donations support our research, family support and advocacy programs. Thank you for supporting our mission to a cure.

How funds were spent:



Platinum Transparency 2022

Candid.

The CACNA1A Foundation is proud to be a Platinum Level Participant on the GuideStar Exchange, demonstrating our commitment to financial transparency.

GREATNONPROFITS 2022 TOP-RATED NONPROFIT

The CACNA1A Foundation has been awarded the distinction of "Top-Rated Nonprofit" by GreatNonprofits.org. Only the top one percent of eligible nonprofits receive this distinction.

MEET OUR TEAM

Board & Leadership Team

Lisa Manaster
President

Sunitha Malepati
Vice President

Allison Buchner
Treasurer

Amy Junge
Secretary

Hala Mirza
Member

Pangkong Fox, PhD
Science Engagement Director

Scientific & Medical Advisory Board

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Montpellier University

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University of Chicago

Joanna Jen, MD, PhD
Mt. Sinai

Sookyong Koh, MD, PhD
Childrens Hopsital, NE

Jeffrey Noebels, MD, PhD
Baylor College of Medicine

Elsa Rossignol, MD, MSc, FRCP
University of Montreal

Michael Wangler, MD
Baylor College of Medicine

JOIN OUR COMMUNITY

203-969-6552

@CACNA1AFOUNDATION

info@cacna1a.org

@CACNA1A

www.cacna1a.org

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@CACNA1AFOUNDATION

@CACNA1AFOUNDATION

Newsletter - Subscribe to our monthly newsletter:
https://www.cacna1a.org/newsletter

Volunteer - Have skills or interests you want to share? Contact us. We have a variety of projects for which we could use your help.

Community Conversations - Join a group to connect with our global community for support:
https://www.cacna1a.org/community-conversations

Enroll - Participate in our research studies:
https://www.cacna1a.org/participate-research

Give - Make a donation to support our work:
https://cacna1a.networkforgood.com/projects/168546-curecacna1a