

IMPACT REPORT

2023



MISSION:

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

VISION:

A world free of the debilitating effects of cacna1a-related disorders

HIGHLIGHTS



Invited to participate in our first Million Dollar Bike Ride, raising \$42,000 and receiving \$30,000 in matching funds for research from the Orphan Disease Center/Penn Medicine



Hosted second in-person Research Roundtable in New York City with more than 60 attendees



First poster co-authored by the Foundation and members of the CACNA1A Research Network presented at American Epilepsy Society Annual Meeting



CACNA1A Research Network convenes in NYC

CACNA1A BY THE NUMBERS

3

NEW RESEARCH GRANTS AWARDED BY
THE CACNA1A FOUNDATION IN 2023

13

CONFERENCES ATTENDED

45

INDIVIDUALS ENROLLED IN CIITZEN*

*A health tech platform that helps patients collect, organize, store, and share their medical records digitally.

90

INDIVIDUALS ENROLLED IN RARE-X*

*A collaborative platform for global data sharing to accelerate treatments for rare diseases.

154

NATURAL HISTORY STUDY PARTICIPANTS*

*A CACNA1A-specific study run by the Chung Lab (which focuses on human genetics and precision medicine) at Boston Children's Hospital.

250

CACNA1A FAMILIES FROM AROUND THE
WORLD IN OUR CONTACT REGISTRY

845

5K RUN, WALK, AND ROLL PARTICIPANTS

4700

SOCIAL MEDIA FOLLOWERS

FAMILY SUPPORT & RAISING AWARENESS

1. Launched Community Conversation Groups & Global Ambassador Program

We launched new virtual peer support groups to not only foster a sense of belonging but also empower families with knowledge and support. Thanks to our Community Conversation Group leaders, all of whom underwent peer support training through the Child Neurology Foundation, the following groups met regularly: Newly Diagnosed, Grandparents/Extended Relatives, Australian Families, Youth (14-18 years), and Developmental and Epileptic Encephalopathy/Hemiplegic Migraine.

We also announced our first Global Ambassadors in Australia and Italy. Our Ambassadors support CACNA1A families in their home country by helping them connect with other local families and resources. They are critical partners in helping us raise awareness globally and engaging families outside of the United States.

3. Participated in 13 Conferences

Our team attended over a dozen rare disease or healthcare professional conferences, facilitating meaningful collaborations and fostering connections with experts, researchers, and other patient advocates. These meetings are crucial platforms for raising awareness about CACNA1A-related disorders, allowing us to share insights, breakthroughs, and challenges with a broader audience. Some of the events we attended included meetings hosted by the American Epilepsy Society, American Academy of Pediatric Ophthalmologists, National Ataxia Foundation, Rare Drug Development Symposium, St. Jude Pediatric Translational Neuroscience Initiative, National Organization of Rare Diseases, and the American Academy of Pediatrics.

2. Enhanced Foundation's Visibility & Credibility

Being invited to speak at events is a powerful tool for raising awareness of the CACNA1A Foundation's work, opening doors to new opportunities, and providing evidence of our leadership in the field. These engagements also connect us with diverse audiences in different fields. This year, we were honored to showcase our patient-driven research projects at:

- NIH Rare Disease Day
- Global Genes/ODC RARE Drug Development Symposium
- FACES Epilepsy Conference
- CZI Science in Society Annual Meeting
- Rose F. Kennedy IDD Research Center 50th Anniversary Celebration
- COMBINEDBrain Annual Strategic Research Meeting

4. Received Horizon Grant

This year, the CACNA1A Foundation was awarded a \$5,000 #RAREis Global Advocate Grant from Horizon Therapeutics. This generous grant is earmarked for translating our resources and handouts into multiple languages, a crucial initiative to better serve the diverse needs of our global CACNA1A community. Horizon Therapeutics, driven by a mission to make a meaningful difference for patients and communities in need, selected us as one of 50 global rare disease organizations to receive this financial assistance. We are honored to be chosen and know this funding will further our family support and education initiatives.

DRIVING SCIENCE FORWARD

5. Launched the First-Ever CACNA1A Disease Concept Model Study

We were selected to partner with the Orphan Disease Center and the University of Pennsylvania to develop the first-ever disease concept model study for CACNA1A-related disorders. A disease concept model uses a qualitative approach to provide a snapshot of how a disease impacts the lives of patients and their caregivers. It analyzes data to pinpoint the most impactful symptoms or those that create the most burden. In addition, the data can be used to identify appropriate outcome measures or endpoints for clinical trials, ensuring that the most accurate assessments are utilized to determine the effectiveness of potential treatments. The study will be published in 2024.

7. Hosted 2nd Research Roundtable & Formed New Working Groups

In October, we brought together over 55 researchers, clinicians, trainees, and representatives from industry and the NIH to exchange ideas and build collaborations to develop CACNA1A-specific treatments and push them into clinical trials for FDA approval. At the meeting, three distinct working groups were formed to tackle the key scientific questions that need to be addressed in order to be ready for clinical trials. The groups are (i) Natural History Study/Biomarkers & Outcome Measures, (ii) Preclinical-to-Clinical, and (iii) Variant Classification. These groups meet monthly and will report on their progress at the 2024 Research Roundtable meeting.

6. Awarded Three New Research Grants

The research grants we awarded this year exemplify our commitment to advancing scientific breakthroughs for the CACNA1A community.

- Samuel Young Jr., PhD for “Development of novel gene therapy strategies to treat CACNA1A-related disorders”
- Henry Colecraft, PhD for “Fixing Cav2.1 functional expression to treat CACNA1A disorders”
- Bart van de Warrenburg, MD, PhD for “Development of an in vitro compound screening assay utilizing an electrophysiological fingerprint in induced pluripotent stem cell-derived neurons carrying a CACNA1A gain-of-function variant”

8. Presented Two Research Posters

At the Chan Zuckerberg Initiative Annual Science in Society Meeting, we presented a poster titled “Breaking Down Research Silos: Bringing Together Clinical and Molecular Researchers to Build an Open Science Portal” to showcase our work with the CACNA1A Research Network to build a portal that aggregates clinical, genetic and molecular data on CACNA1A disorders, led by Dennis Lal, PhD.

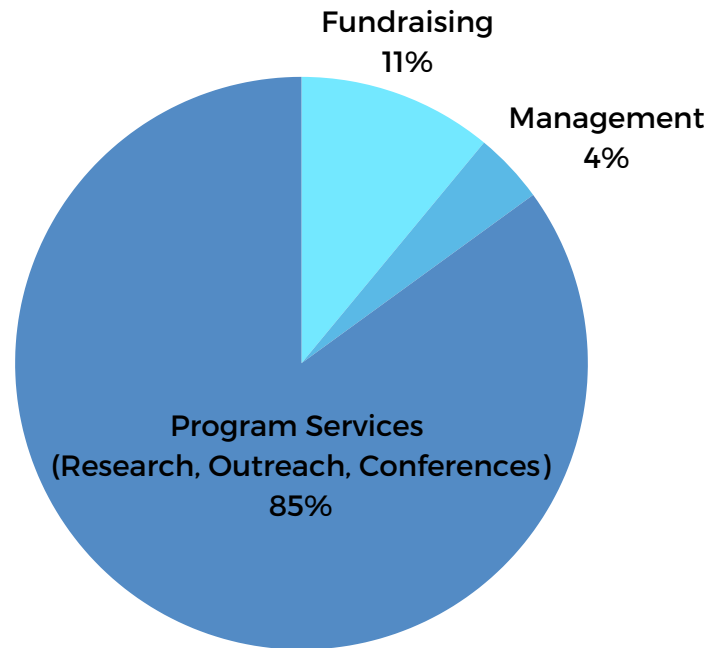
At the annual American Epilepsy Society meeting, Laina Lusk, MMSc, CGC, presented a poster co-authored by the Foundation and members of the CACNA1A Research Network titled “Phenotypic Analysis of 454 Individuals with CACNA1A-Related Neurodevelopmental Disorders,” which conceptualizes CACNA1A-related disorders as a disorder of several homogenous groups.

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 2023, we raised \$585,860 or 12% more than the prior year 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:



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

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

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Baylor College of Medicine



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University of Montreal



Michael Wangler, MD
Baylor College of Medicine

LOOKING AHEAD TO 2024

- **Events**

- CACNA1A Awareness Day (March 19th)
- Annual 5K Run, Walk & Roll (May 18th & 19th)
- Million Dollar Bike Ride (June 10th)
- Research Roundtable at the NIH, Bethesda, MD (July 18th)
- Creating Connections Family Conference, Bethesda, MD (July 19th & 20th)

- **Research**

- Publish CACNA1A Treatment Guidelines
- Publish CACNA1A Disease Concept Model
- Participate in metabolomics project to look for plasma biomarkers
- Co-Develop a Channelopathy Center of Excellence at UT Houston
- Increase enrollment in our patient-driven research studies (Natural History, Rare-X, Ciitizen)
- Add clinical assessment data to the Natural History Study
- Launch CACNA1A Variant Portal
- Fund drug screening and EEG research projects

- **Support**

- Expand the Global Ambassadors Program to additional countries
- Publish educational resources in additional languages

JOIN OUR COMMUNITY



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Volunteer - Have skills or interests you want to share? Contact us. We have a variety of projects for which we could use your help. [Info@cacna1a.org](mailto:info@cacna1a.org)

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>

Thank you for your continued support:

The CACNA1A Foundation relies on the generous support of our donors to advance our mission. Whether you are making your first donation or are a major donor, we are honored to have your support. We can't do this work without it. Please make a donation to support our work today! **Give Here** - <https://cacna1a.networkforgood.com/projects/168546-curecacna1a>