Creating Connections

CACNA1A CONFERENCE
JULY 22 - 23, 2022

A GLOBAL HYBRID GATHERING FOR FAMILIES, CLINICIANS & SCIENTISTS

IN PERSON @ THE HILTON SAN ANTONIO HILL COUNTRY, TEXAS USA
VIRTUALLY @ YOUR LOCATION
WWW.CACNA1A.ORG

AGENDA & CONFERENCE INFORMATION
Dear Patients, Families, and Professionals,

We would like to welcome you to our second CACNA1A Creating Connections Community Conference. Whether you are attending in-person or virtually, we are honored that you are with us. We have attendees from around the world and are excited to be learning together! This event provides the opportunity for both families and professionals to foster new relationships and collaborations. For those with us here in the San Antonio Texas Hill Country, this is a momentous weekend because it is the first time CACNA1A patients, families, caregivers, clinicians and researchers have gathered together in person. We have 100 adults and 36 children in-person, and we know that after tomorrow, we will have created lifelong friendships!

We have an incredible line-up of renowned healthcare professionals and researchers who are here to update us on the latest research and treatment options and empower us all. They are also looking forward to meeting CACNA1A families and learning about our experiences. We are grateful that they have taken time from their practices and summer breaks to join us this weekend.

Finally, this conference would not be possible without the generous support of our sponsors, grantors, the Chan Zuckerberg Initiative, and our hardworking conference committee. We thank you all from the bottoms of our hearts for your partnership as we aim to rewrite the futures for our loved ones.

With hope and gratitude,
Lisa, Sunitha, Allison, Amy, and Hala
The CACNA1A Foundation Board of Directors

### IMPORTANT INFORMATION

**Community Participation Guidelines** - Words and actions matter. While at our conference, please remember to treat our community with respect. Please be kind, make space, be inclusive, take responsibility, bring solutions, and lead by example.

**Disclaimers** - The information presented during the sessions is intended for educational purposes only and is not endorsed by the CACNA1A Foundation. In the event of unforeseen circumstances, the conference schedule and speakers may change and/or pivot to a virtual meeting.

### ABOUT CACNA1A

The CACNA1A gene encodes for the main pore-forming subunit of the P/Q-type calcium channel, Cav2.1, which controls calcium entry into neurons. Genetic variants on the CACNA1A gene disrupt this calcium entry, leading to various symptoms, including epilepsy, neurodevelopmental disorders, global developmental delay, ataxias or balance and coordination disorders such as congenital ataxia, episodic ataxia type 2 (EA2) and spinocerebellar ataxia type 6 (SCA6), hemiplegic migraines (stroke-like episodes) and eye movement disorders. The MRIs of many patients show evidence of cerebellar atrophy. CACNA1A is one of the genes commonly associated with epileptic encephalopathy and has been linked to autism spectrum disorder.
ABOUT CACNA1A FOUNDATION

The CACNA1A Foundation, a 501(c)(3) organization founded in March 2020, is a parent-led foundation supporting individuals with CACNA1A genetic variants. Our mission is to find specific treatment options and a cure for CACNA1A 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. Education and collaboration among stakeholders are central to this lofty goal. Our Creating Connections Community Conference is a crucial part of our commitment to address CACNA1A patients’ and families’ unmet needs and to develop collaborative relationships between patients, families, clinicians and researchers.

Currently, there are no established standards of care for CACNA1A-related disorders. Families and their clinicians are experimenting with medications using trial-and-error methods. The CACNA1A Foundation is on a mission to change this! Our Roadmap to a Cure will guide the Foundation on a path to drug discovery with a goal of developing novel treatments for patients with CACNA1A variants. Additionally, we have a Natural History Study, overseen by Dr. Wendy Chung at the Columbia University Medical Center, and are part of the RARE-X data collection program. We are currently developing an open science portal in collaboration with Dennis Lal, PhD at the Cleveland Clinic, which will enable global aggregation of clinical, genetic and molecular data for CACNA1A variants and will link researchers, clinicians and families worldwide to more clearly characterize the clinical spectrum of CACNA1A-related disorders. Furthermore, to become clinical trial-ready, we are developing a library of biospecimens and iPSC lines and are funding the development of animal models aligned with human mutations.

As parents of children with CACNA1A variants, a sense of urgency is what drives us. We are committed to raising awareness of this rare disease, providing educational resources and funding research towards a cure (translational research) so that someday, no one will suffer from CACNA1A-related diseases. Please visit our website and follow us on social media to learn more about our initiatives. There you will find educational videos, announcements about upcoming events such as our monthly virtual meetups, and printable resources for you and your physicians.

CONFERENCE SCHEDULE

All times are US Central Time (UTC-5)

JULY 21st
11:00 am – 5:00 pm – Family Fun Day at Morgan’s Wonderland & Inspiration Island. The CACNA1A Foundation will be providing families with tickets to both the amusement park and waterpark. We also have access to a private airconditioned indoor space to gather at throughout the day. The parks are about 20 miles away from the conference hotel and are accessible by car or ride-sharing services (Uber and Lyft). Families are responsible for their own transportation to and from the parks.

JULY 22nd
8:00 am – 9:00 am – Welcome & CACNA1A Genetics: What We Know & What We’re Learning (Laina Lusk, MMSc, CGC, Children’s Hospital of Philadelphia; Liz Butler, MS, CGC, GeneDx)
9:00 am – 10:30 am – The Neurology & Clinical Management of CACNA1A-related Abnormal Eye Movement, Dystonia, and Imbalance (Joanna Jen, MD, PhD, Mt. Sinai; Aasef Shaikh, MD, PhD, Case Western Reserve University; Scott Grossman, MD, NYU Grossman School of Medicine)
10:30 am – 11:00 am – Break*
11:00 am – 12:00 pm – Update on the CACNA1A Natural History Study & Why Participation Matters (Wendy Chung, MD, Columbia University)
12:00 pm – 12:30 pm – Foundation Update (Lisa Manaster, CACNA1A Foundation)
12:30 pm – 1:00 pm – Keynote Address (Leah Schust Myers, FamilieSCN2A Foundation)
1:00 pm - 2:30 pm – Lunch/Break*
2:30 pm – 4:00 pm – CACNA1A-related Hemiplegic Migraine: What it is and What is being Done (Ingo Helbig, MD, Children’s Hospital of Philadelphia; Irene de Boer, MD, MSc, Leiden University Medical Center; Zameel Cader, DPhil, MRCP, Oxford Headache Centre, Oxford University Hospitals)
4:00 pm – 5:00 pm – CACNA1A & Sleep – It’s Way More Than the Channel! (Joshua Rotenberg, MD, Houston Specialty Clinic)
5:00 pm – 6:30 pm – Break*
6:30 pm – 8:30 pm – Welcome Cocktail Reception

JULY 23rd
8:00 am – 8:30 am – Welcome to Day 2 (Sunitha Malepati, CACNA1A Foundation)
8:30 am – 9:30 am – Functional Characterization (Gain of Function/Loss of Function) of CACNA1A Variants and Implications for Therapeutics (Henry Colecraft, PhD, Columbia University)
9:30 am – 10:30 am – Characterizing an Allelic Series of Rare Missense Variants of CACNA1A in a Cohort of Patients with Severe Neurodevelopmental Disorders (Jen Pan, PhD, Broad Institute, MIT)
10:30 am – 11:00 am – Break*
11:00 am – 12:00 pm – CACNA1A Variant Database & the Power of Data Aggregation for Therapeutic Development (Dennis Lal, PhD, Cleveland Clinic)
12:00 pm - 1:00 pm – CACNA1A Therapeutic Strategies – Lessons from Other Ion Channels (Alfred L. George Jr., MD, Northwestern University)
1:00 pm - 2:30 pm – Lunch/Break*
2:30 pm - 3:30 pm – CACNA1A Cure Roadmap (Ethan Perlstein, PhD, Perlara)
3:30 pm - 4:00 pm – Concluding Remarks (Pangkong Fox, PhD, CACNA1A Foundation)
4:00 pm - 6:00 pm – Break*
6:00 pm - 9:00 pm – Family Dinner Dance with DJ

*During the Breaks, there will be opportunities to visit exhibit booths and enroll in our Natural History Study, Rare-X CACNA1A Data Collection Program, and Biobanking Program. There will also be opportunities to meet one-on-one (must sign up for a time slot) with genetic counselors Laina Lusk (CHOP) and Liz Butler (GeneDx). On Thursday afternoon and Sunday morning, Theresa Spong, MED, PT, CBP is available to provide mini-PT evaluations (advance sign up required.)

ABOUT OUR SPEAKERS
Elizabeth Butler, MS, CGC
Senior Director, Genetic Counseling, GeneDx

Elizabeth (Liz) Butler received her master’s degree from the Genetic Counseling Program at the University of Cincinnati in 2003. She spent several years as a clinical genetic counselor primarily working in pediatrics and adult neurogenetics before relocating to GeneDx in 2008. She has many years of experience in variant classification, assessment of gene-disease validity, laboratory reporting for panel and genomic testing, and the development of genetic testing panels for epilepsy and other neurodevelopmental disorders. Liz is now the Senior Director of Genetic Counseling at GeneDx where she leads a team of more than 100 genetic counselors and genetic counseling assistants involved in client support and reporting for a broad array of clinical genetic tests, including whole exome and
genome sequencing. Liz is a member of the ClinGen Epilepsy Gene Curation and Sodium Channel Variant Curation Expert Panels.

Zameel Cader, DPhil, MRCP  
Director, Oxford Headache Centre; Consultant Neurologist, John Radcliffe Hospital; Professor of Neuroscience & Neurology, Nuffield Department of Clinical Neuroscience

Dr. Zameel Cader’s research program is focused on understanding the disease process in headache and pain disorders using human stem cell disease models and preclinical models. His work in headache has included investigating the role of the two-pore potassium channel TRESK in migraine and pain. He is also working on developing improved human blood-brain-barrier models and is the academic lead for the IM2PACT consortium. He previously led the StemBANCC consortium, which established stem cell resources for academia and industry across Europe. His research group, Translational Molecular Neuroscience, aims to bring more effective treatments arising from bench to bedside. He has founded two start-ups – Oxford StemTech, which uses novel technologies to provide human stem cell reprogramming at scale and HumanCentric, which provides a platform for de-risked target identification for neurological disorders.

Wendy Chung, MD  
Kennedy Family Professor of Pediatrics and Medicine, Vagelos College of Physicians and Surgeons, Columbia University

Dr. Wendy Chung is an ABMG board certified clinical and molecular geneticist with over 20 years of experience in human genetic research of monogenic and complex traits including diseases. She has extensive experience mapping and cloning genes in humans and describing the clinical characteristics and natural history of novel genetic conditions and characterizing the spectrum of disease and developing tailored care and treatments for rare genetic diseases. Dr. Chung directs NIH funded research programs in human genetics of birth defects including congenital diaphragmatic hernia, congenital heart disease, and esophageal atresia, autism, neurodevelopmental disorders, pulmonary hypertension, cardiomyopathy, obesity, diabetes, and breast cancer. She leads the Precision Medicine Resource in the Irving Institute at Columbia University. She has authored over 300 peer reviewed papers and 50 reviews and chapters in medical texts. She was the recipient of the American Academy of Pediatrics Young Investigator Award, the Medical Achievement Award from Bonei Olam, the New York Academy Medal for Distinguished Contributions in Biomedical Science, and the Rare Impact Award from the National Organization of Rare Disorders. Dr. Chung enjoys the challenges of genetics as a rapidly changing field of medicine and strives to facilitate the integration of genetic medicine into all areas of health care in a medically, scientifically, and ethically sound, accessible, and cost-effective manner. She received her B.A. in biochemistry and economics from Cornell University, her M.D. from Cornell University Medical College, and her Ph.D. from The Rockefeller University in genetics.

Henry Colecraft, PhD  
John C. Dalton Professor and Associate Vice Chair in the Department of Physiology and Cellular Biophysics, and Professor in the Department of Molecular Pharmacology & Therapeutics at Columbia University Irving Medical Center

Dr. Colecraft is an international leader in the molecular physiology of ion channel proteins that underlie signaling in nerve cells and the heart. He has directed the Ion Channel Physiology & Disease Lab since 2001. His research group has contributed seminal advances to understanding molecular mechanisms underlying regulation of voltage-dependent Ca2+ and K+ channels by accessory subunits, posttranslational modifications, and signaling molecules. His group also studies how inherited mutations in ion channels lead to devastating diseases (known as ion channelopathies) that span the cardiovascular, neurological, and respiratory systems, and in devising new therapies for them. He has an active research program focused on understanding mechanisms underlying CACNA1A-linked neurological disorders and discovering new molecular treatments for these diseases. Dr. Colecraft
obtained his BSc in Physiology from University of London King’s College, and his PhD in Pharmacology from the University of Rochester. He completed his postdoctoral training at Johns Hopkins University, where he remained as an Assistant Professor in Biomedical Engineering when he was recruited to Columbia as an Associate Professor in 2007

**Irene de Boer, MD, MSc**

PhD candidate and physician researcher neurology, Leiden University Medical Center (LUMC), Attending physician at the LUMC headache clinic with a focus on hemiplegic migraine and cerebral hereditary angiopathy

Dr. Irene de Boer is a treating physician at the Leiden University Medical Center (LUMC) headache clinic and the Cerebral Hereditary Angiopathy Clinic. With a background in both Medicine and Biomedical Science, she started her PhD project at the LUMC. She is now finishing her dissertation, which focusses on monogenic migraine disorders, including hemiplegic migraine and small vessel disease Retinal Vasculopathy with Cerebral Leukoencephalopathy and Systemic manifestations (RVCL-S). She is focused on unraveling the pathophysiological mechanism leading to monogenic migraine disorders and on identifying biomarkers for disease progression. She is a member of the International Headache Genetics Consortium (IHGC) and the International Consortium for Cluster Headache Genetics (CCG) and vice-president of the International Headache Society Junior group. She recently received the prestigious Dekkergrant to continue her research into the contribution of vascular mechanisms to neurological disorders, including migraine.

**Alfred L. George, Jr, MD**

Chair, Department of Pharmacology; Director, Center for Pharmacogenomics; Alfred Newton Richards Professor of Pharmacology, Feinberg School of Medicine, Northwestern University; Director of the Channelopathy-associated Epilepsy Research Center

Dr. George is an internationally renowned leader in the channelopathy field. His work focuses on genetic disorders caused by voltage-gated ion channel mutations that are responsible for disorders of membrane excitability including diseases affecting muscle, heart and brain. At Northwestern, Dr. George’s research program is focused on the structure, function, pharmacology, and molecular genetics of ion channels. Dr. George’s long-term involvement and commitment to investigating channelopathies including channelopathy-associated epilepsy provides him with an unparalleled knowledge and perspective on the scientific trajectory and major opportunities in this field. His labs’ studies of the molecular basis for genetic epilepsy have revealed new targets for antiepileptic drug development. Dr. George is the Director of the Channelopathy-Associated Epilepsy Research Center, a multi-institutional and interdisciplinary research center that combines high-throughput technologies on non-neuronal cells with studies of human neuron and animal model systems. According to Dr. George, “Results from this work will contribute to improving the accuracy of genetic diagnosis of epilepsy and foster a better understanding of disease mechanisms at molecular and cellular levels. We will also strive to determine the optimal drug therapy for specific mutations — a step in the direction of precision medicine.”

**Scott Grossman, MD**

Assistant Professor of Neurology, NYU Grossman School of Medicine

Dr. Scott Grossman is an Assistant Professor of Neurology at NYU Grossman School of Medicine. He earned his medical degree at the University of Pennsylvania and then trained in Neurology, Clinical Neurophysiology and Neuro-ophthalmology at NYU. After finishing training in the summer of 2020, he joined the faculty and has a special clinical and research interest in efferent neuro-ophthalmology, including the validation of ocular motility patterns as a biomarker across the spectrum of neurologic disease.
Ingo Helbig, MD  
Pediatric neurologist in the Division of Neurology and the Director of Genomic Science, Children’s Hospital of Philadelphia (CHOP)

Dr. Helbig went to medical school in Heidelberg and Mannheim, Germany, and Lexington, Kentucky, USA. He trained at the Epilepsy Research Centre, Melbourne, Australia, and was Assistant Professor at the Department of Neuropediatrics, Kiel, Germany. Between 2011 and 2015, he co-headed the EuroEPINOMICS-RES Consortium, the European counterpart of the NIH-funded Epi4K consortium involved in collaborative genomic studies to identify genes for human epilepsies. He was part of the Genetics Commission of the International League Against Epilepsy (ILAE) from 2014-2017 and currently leads the Epilepsiome Task Force of the ILAE Genetics Commission, which aims at increasing genetic literacy in the epilepsy community. After heading the epilepsy genetics group at the University of Kiel, Germany, he transferred to the Children’s Hospital of Philadelphia (CHOP) in 2014 and became faculty in the Division of Neurology in July 2017. The main focus of his prior work was to understand how genetic changes lead to severe epilepsies in both children and adults, contributing to several new gene findings in the field in the last seven years including GRIN2A, CHD2, KCNA2, HCN1, and DNM1. Dr. Helbig uses clinical and research expertise to curate epilepsy-related genes in variants within his leadership role of the Epilepsy Clinical Domain Working Group.

Joanna Jen, MD, PhD
Dr. Morris B. Bender Professor of Neurology, Professor of Otolaryngology & Neurosurgery, Chief, Division of Neuro-otology & Neurogenetics, Department of Neurology, Icahn School of Medicine at Mount Sinai, New York City

Dr. Jen is a neurologist with formal training in neuro-otology, the neurology of hearing, balance, and eye movement control. She evaluates and treats patients with dizziness due to a variety of causes, spanning very common disorders (for example, benign paroxysmal positional vertigo), vestibular migraine or migraine-associated dizziness suspected to be genetically complex, and other exceptionally rare hereditary neurodegenerative conditions such as episodic ataxia and pontocerebellar hypoplasia. Dr. Jen’s clinical focus in neuro-otology is complemented by research performed in her laboratory on the genetic and physiological bases of disorders affecting balance and eye movement control in neurodevelopment and neurodegeneration. Her research spans many levels, from clinical observation to genetic characterization, cellular and animal studies, and clinical trials, to bring everything full circle back to patients. The ultimate goal of her research is to improve diagnosis and develop treatments that will improve patient function and quality of life.

Dennis Lal, PhD
Assistant Professor in the Department of Molecular Medicine in the Cleveland Clinic Lerner College of Medicine of Case Western Reserve University (CCLCM), Assistant Staff in the Genomic Medicine Institute with a secondary staff appointment in the Neurological Institute’s Epilepsy Center both Cleveland, OH, Visiting Scientist at the Broad Institute of Harvard and M.I.T., Cambridge, MA, Group Leader at the Cologne Center for Genomics, Cologne, Germany

Dr. Lal is an early career investigator with expertise in genomics and bioinformatics. He has focused his recent research effort on developing and implementing novel computational strategies to investigate variants in genes associated with epilepsy and neurodevelopmental disorders. He has also established strategic collaborations with large groups in Europe with large databases of variants in channelopathy-associated epilepsies.
Laina Lusk, MMSc, CGC  
Genetic counselor in the Division of Neurology at Children's Hospital of Philadelphia

Laina is a genetic counselor in the division of neurology at the Children’s Hospital of Philadelphia, with a research interest in understanding the genetic mechanisms and clinical phenotypes of childhood epilepsy and related neurodevelopmental disorders. She received her BS in neuroscience from Bucknell University and her MMSc in human genetics and genetic counseling from Emory University. Laina has prior clinical experience working as a cancer genetic counselor, and prior research experience in the field of neurodevelopmental disorders. In the Helbig Lab, she serves as a clinical liaison and assists with phenotyping projects.

Jen Pan, PhD  
Institute Scientist, Broad Institute of MIT and Harvard & Director of Translational Neurobiology, Stanley Center for Psychiatric Research

Jen Q. Pan is an Institute Scientist at Broad Institute of MIT and Harvard and the director of Translational Neurobiology at the Stanley Center for Psychiatric Research. The research of her group focuses on translating emerging genetics into biology and to enable next-generation therapeutics to treat psychiatric and neurological illnesses. Dr. Pan studies genes whose dysfunction has been implicated for psychiatric illnesses using molecular, cellular, and electrophysiological approaches, both in vitro and in animals, with a focus on membrane proteins and ion channels. She leads the ICE-T (ion channel electrophysiology and technology) effort at Broad. Before joining the Broad, Dr. Pan led drug discovery efforts involving sodium and calcium channels in psychiatric and neurological disorders at two start-up biotechs and Amgen. Dr. Pan obtained her PhD in Neuroscience from Brown University, and BS in Chemistry from Nanjing University.

Ethan Perlstein, PhD  
Co-Founder and Chief Executive Officer, Perlara

Dr. Perlstein is a graduate of Harvard University in the Department of Molecular and Cell Biology with a PhD. He developed an approach to studying old drugs and discovering new drugs in model organisms called evolutionary pharmacology. He is an author on 19 peer-reviewed scholarly publications, including the discovery of a novel mechanism of action for the antidepressant Zoloft based on studies in yeast cells. In 2014 he founded Perlara, the first biotech PBC on a mission to codevelop drugs with highly motivated families racing to cure the long tail of genetic diseases.

Joshua Rotenberg, MD, MMS  
Pediatric Neurologist at Houston Specialty Clinic, TX

Dr. Rotenberg is a child neurologist with subspecialty certifications in epilepsy, brain injury medicine and sleep disorders in Houston, TX. He manages children and teens with neurologic illnesses including brain injury, cerebral palsy, epilepsy, migraine, ataxia, spasticity, neuro-developmental disorders and autism. Dr. Rotenberg earned his undergraduate, graduate, and medical degrees from Brown University. He received the United States Air Force Health Professions Scholarship and was commissioned as a 2nd Lieutenant in the United States Air Force in 1991, where he served for 10 years. Dr. Rotenberg completed a fellowship in child and adolescent neurology at the Uniformed University of the Health Sciences, Walter Reed AMC, Bethesda National Naval Medical Center and the NIH. Applying his
advanced training in vagal nerve stimulation, ketogenic diet, video EEG, sleep and in chemodenervation, Dr. Rotenberg served as the Chief of Pediatric neurology at Wilford Hall USAF Medical Center. He is a pediatric neurologist at Houston Specialty Clinic, and is affiliated with Texas Children’s Hospital, Children’s Memorial Hermann and The Woman’s Hospital. Dr Rotenberg holds a volunteer appointment as an assistant clinical professor of pediatric neurology at the University of Texas at Houston. Dr. Rotenberg is an active advocate for children and families with neurologic illness. He is the founder and admin of the CACNA1A/Cav2.1 Facebook Group, an educational and support group for patients, families, and caregivers of individuals with Cacna1a mutations that currently has over 600 members. He started the group after his fourth child was diagnosed with a CACNA1A variant in 2014.

**Leah Schust Myers**  
Founder & Executive Director, FamilieSCN2A Foundation

Leah has spent her entire career working in health care administration and never imagined she would find a use for her skills in an entirely different way. From medical secretary to hospital manager and everywhere in between, Leah learned how to manage the needs of large populations within a medical setting. When her son, Ben, was diagnosed with an SCN2A Disorder in 2012, it became abundantly clear how to leverage her 20+ years of experience to help not only her family but hundreds of others. In July of 2019, Leah transitioned from her volunteer position as President of the FamilieSCN2A Foundation Board, to a full time Executive Director role. The experience she brings with her, especially from her most recent position as Executive Project Manager for a fortune 500 Healthcare company, is giving the foundation the professional edge to meet the growing needs of the community. Communication and collaboration being her area of expertise, Leah has been instrumental in forming relationships with clinicians and scientists to advance progress in studying SCN2A and novel treatments. These alliances with researchers and industry leaders have been influential in catapulting SCN2A on to the map of exciting targets to study.

**Aasef Shaikh, MD, PhD**  
Penni and Stephen Weinberg Chair in Brain Health, and Vice-Chair for Research, Department of Neurology, Case Western Reserve University, School of Medicine; Staff Neurologist, University Hospitals Cleveland Medical Center

Dr. Aasef Shaikh is Penni and Stephen Weinberg Chair in Brain Health, and Vice-Chair for Research in The Department of Neurology at The University Hospitals and Case Western Reserve University in Cleveland. His academic work at The Daroff-Dell’Osso Ocular Motility Laboratory at the Case Western Reserve University focuses on approaching the complex disorders of the vestibular system, eye movements, head movements, gait and balance with biomedical engineering concepts. The overarching goal is to discover the novel network connections and leverage their influence to modulate the motor circuits artificially for the treatment of intractable neurological conditions. Dr. Shaikh had received the prestigious American Academy of Neurology (AAN) Alliance Founders Award, The American Neurological Association Grass Foundation Award in Neuroscience, The AAN Career Award, Jon Stolk Award for Excellence in Movement Disorders from the AAN, and George C. Cotzias Memorial Fellowship by The American Parkinson's Disease Association.

**CACNA1A NATURAL HISTORY STUDY**

Patients with a CACNA1A genetic variant are invited to participate in the CACNA1A Natural History Study (NHS) led by Dr. Wendy Chung at Columbia University. The CACNA1A NHS will follow patients over time and collect data to accurately define our patient population, develop an in-depth understanding of the disease and its progression. This information is crucial to inform research and translational science. Individuals are asked to fill out detailed health questionnaires and provide comprehensive medical data, including MRIs and EEGs. Our research coordinator from the Chung Lab will attend the conference to facilitate the enrollment of new patients and answer any questions.
CACNA1A + RARE-X DATA COLLECTION PROGRAM

The CACNA1A Foundation has partnered with RARE-X to build a Data Collection Program for the CACNA1A community. By increasing access to research-ready data, we hope to accelerate the discovery of ground-breaking treatments for CACNA1A! We chose to partner with RARE-X because we hope that by leveraging RARE-X’s data collection platform in conjunction with other rare disease communities, we can increase the likelihood that CACNA1A will be included in cross-disease research. We encourage all families to enroll in this program.

CLINICAL TRIAL READINESS

The CACNA1A Foundation is focused on making our community clinical trial ready. This entails compiling comprehensive natural history data, developing a library of biospecimens, iPSC lines and animal models. A phlebotomist will be available throughout the conference to collect blood from patients who have consented to participate in the biorepository at Sampled (formerly IBX). In addition to the CACNA1A Foundation’s biorepository, patients and their immediate family members will also have the option to submit blood specimens to the NIGMS Human Genetic Cell Repository housed at Coriell. The CACNA1A Foundation is committed to the open sharing of biospecimens, cell lines, animal models, and de-identified data.

MAKING A DIFFERENCE

Our work would not be possible without the generous support of our donors. We are an all-volunteer board and donations support our research and family support programs. Please consider making a tax-deductible gift in support of the CACNA1A Foundation. We are #StrongerTogether!

More information on how to donate can be found on our website at www.cacna1a.org.

EIN# - 8404985747.

If you would like to volunteer with the CACNA1A Foundation, please reach out to us at info@cacna1a.org

We welcome the involvement and have a variety of projects for which we could use help.

Thank you for your support!

STAY IN TOUCH

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