JOIN OUR COMMUNITY

Our website contains resources for families and professionals who want to learn more about CACNA1A

- Sign up for our monthly newsletter
- Learn about current research
- Enroll in our patient registry
- Access a members-only Facebook support group
- Read family stories
- Volunteer/host a fundraiser
- Make a donation

WHO WE ARE

The CACNA1A Foundation is a parent-led 501(c)(3) non-profit. We are a global community dedicated to creating awareness, supporting families and finding a cure for CACNA1A genetic variants.

CACNA1A FOUNDATION

INFO@CACNA1A.ORG
WWW.CACNA1A.ORG

SUPPORTING RESEARCH

We support translational research that improves patient outcomes and paves the way for clinical trials. Partnerships with families, medical practitioners and researchers are vital to patient-centered outcomes.

We encourage the scientific community to collaborate to achieve targeted treatment options and an eventual cure for those with CACNA1A variants.

WE ARE PARTNERED WITH THE CHUNG LAB AT COLUMBIA UNIVERSITY TO COLLECT DATA FOR RESEARCHERS STUDYING THE DISEASE.

PLEASE VISIT OUR WEBSITE FOR MORE INFORMATION ON HOW YOU CAN JOIN OUR PATIENT REGISTRY AND MAKE A DIFFERENCE.

CACNA1A FOUNDATION

RAISING AWARENESS SUPPORTING FAMILIES FUNDING RESEARCH

RARE TOGETHER IS NOT SO RARE

WWW.CACNA1A.ORG

WHAT IS CACNA1A?

- CACNA1A is, a gene that plays a vital role in the communication between neurons in the brain.
- It is located on the short arm of chromosome 19 and codes for a voltage-gated calcium channel called Cav2.1.
- A change in the gene alters the function of the channel and affects the release of neurotransmitters.
- CACNAIA variants are characterized as either Gain of Function or Loss of Function.

Gain of Function variant:

The calcium channel opens and stays open for a long time, so calcium ion influx is increased, resulting in too much neuron excitability.

Loss of Function variant:

The opposite occurs; there is improper channel opening, causing less calcium to enter the cells and decreased neuronal firing.

TREATMENT

Current options for treating CACNA1A related disorders are limited to treating symptoms. There is no known cure.
However, a genetic diagnosis provides the first steps for solving the puzzle regarding why one has certain symptoms.

Patients with CACNA1A benefit from a multi-disciplinary team approach that includes:

- A Geneticist,
- Neurologist,
- Developmental pediatrician,
- Ophthalmologist,
- Physical and
- Occupational therapists
- A speech & language pathologist.

SYMPTOMS & DISORDERS

CACNA1A variants are associated with multiple neurological disorders. Individuals exhibit a variety of symptoms that fall on a spectrum from mild to severe.

Neurodevelopmental Differences

- Global Developmental Delays (mild to severe)
- Cognitive Impairment
- Intellectual Disability
- Austism Spectrum Disorder
- Hypotonia

Epilepsy

- Mild to Severe Seizure Disorders
- Severe: Developmental & Epileptic Encephalopathy

Ataxia (balance & coordination)

- Congential Ataxia/Tremors
- Episodic Ataxia Type 2 (EA2)
- Spinocerebellar Ataxia Type 6 (SCA6)

Migraine

- Migraines with or without aura
- Sporadic Hemiplegic (temporary weakness or paralysis) Migraine
- Familial Hemiplegic Migraine Type 1 (FHM1)

Cerebellar Atrophy

- Can be Congential, Early Onset or Late Onset
- Progressive
- Can cause Dystonia

Eye Disorders

- Nystagmus
- Paroxymal Tonic Upgaze
- Cortical Vision Impairment

NEWLY DIAGNOSED

Getting a rare disease diagnosis can be a traumatic time in a family's life. Know that you are not alone. Early intervention, intensive therapies and appropriate seizure control are vital to patient outcomes.

The effects of CACNA1A-related disorders are far more than just managing symptoms and can impact multiple areas of a family's life. It is important to find a support system that may consist of family, friends, therapists, and respite care. Find support from other parents who understand the challenges of being a CACNA1A caregiver by joining the private Facebook group.

Link available on website:

WWW.CACNA1A.ORG