

## ABOUT US

The CACNA1A Foundation was founded in 2020 by parents of children diagnosed with developmental delays, intellectual disability, epilepsy, autism, ataxia, nystagmus, PTU and hemiplegic migraine as a result of a change in their CACNA1A gene. We are a nonprofit 501(c)(3) tax-exempt organization. Our board of directors covers all operating expenses so every dollar donated goes toward our mission.

## OUR 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-linked diseases.

## OUR VISION

A world free of the debilitating effects of CACNA1A-related disorders.



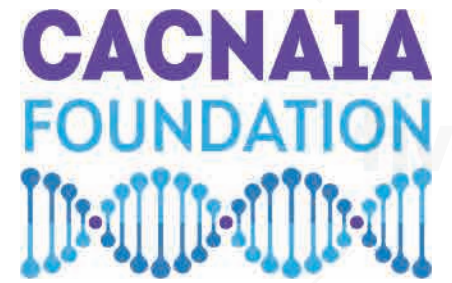
The CACNA1A Foundation accepts proposals for research grants on a rolling basis. Of primary concern is the ability to achieve a better understanding of CACNA1A and treatments for the disease in order to improve the quality of life for those with CACNA1A variants.

Please contact us if you have an idea that would help move CACNA1A research closer toward targeted treatment options.



## GET IN TOUCH

[www.CACNA1A.org](http://www.CACNA1A.org)  
[info@CACNA1A.org](mailto:info@CACNA1A.org)



"Eye movement disorders are an early manifestation of CACNA1A mutations in children. Improved recognition of the CACNA1A phenotype in childhood is important for early diagnosis, counseling and appropriate emergency management." (Manezes, M.P. et al)

**Awareness can help decrease the diagnostic journey for children with CACNA1A-related diseases.**

# WHAT IS CACNA1A?

CACNA1A is a gene located on the 19th chromosome that encodes for the  $\alpha 1$  subunit of Cav2.1, a voltage-gated calcium channel expressed in the brain and abundant in the cerebellar granule and Purkinje cells.

Calcium flux through the Cav2.1 channel is essential for neurotransmitter release. Mutations or variants in CACNA1A lead to a broad spectrum of neurodevelopmental disorders, including:

- Global developmental delay
- Intellectual disability (mild to severe)
- Autism Spectrum Disorder
- Epilepsy (often difficult to control)
- Speech and language delays
- Hemiplegic Migraines (stroke-like episodes)
- Ataxia - episodic type 2 (EA2), congenital, spinocerebellar type 6 (SCA6), cerebellar atrophy
- Dystonia
- Psychiatric disorders
- Eye movement disorders - nystagmus, PTU, CVI

# RELEVANT RESEARCH

## Eye movement disorders are an early manifestation of CACNA1A mutations in children

Tantsis EM, Gill D, Griffiths L, Gupta S, Lawson J, Maksemous N, Ouvrier R, Riant F, Smith R, Troedson C, Webster R, Menezes MP. Dev Med Child Neurol. 2016.Jun;58(6):639-44.doi: 10.1111/dmcn.13033. Epub 2016 Jan 27. PMID: 26814174.

## From Genotype to Phenotype: Expanding the Clinical Spectrum of CACNA1A Variants in the Era of Next Generation Sequencing

Indelicato E and Boesch S (2021) From Genotype to Phenotype: Expanding the Clinical Spectrum of CACNA1A Variants in the Era of Next Generation Sequencing. Front. Neurol. 12:639994. doi: 10.3389/fneur.2021.639994

# RARE DISEASE FACTS

- Affects < 200,000 people in the USA
- 10,000 + identified rare diseases
- 1 in 17 people will suffer from a rare disease at some point in their life
- 1 of 2 patients diagnosed with a rare disease is a child
- 85% of rare diseases are caused by a genetic mutation
- 8 years - The average time it takes for rare patients to receive an accurate diagnosis
- On average, 8-10 different physicians are consulted before receiving a proper diagnosis

The first sign that parents notice in their child eventually diagnosed with a CACNA1A genetic mutation is the presence of Paroxysmal Tonic Upgaze, usually as an infant and before their first seizure.

An ophthalmologist is often the first specialist our patients see.

**IF YOU HAVE A PATIENT WITH DEVELOPMENTAL DELAYS AND PTU OR NYSTAGMUS, PLEASE REFER THEM TO GENETIC TESTING OR NEUROLOGY!**

**EARLY DIAGNOSIS IS KEY!  
DON'T DISMISS THE SYMPTOMS...  
HELP DECREASE THE DIAGNOSTIC JOURNEY OF PATIENTS WITH CACNA1A.**

**CACNA1A AWARENESS**

*Did You Know?*  
Nearly 90% of participants in our NHS\* have reported CACNA1A-related vision disorders.

The top issues include:

- Nystagmus
- Eye Movement Abnormalities
- Strabismus
- PTU

\*Natural History Study (2021)

#ThisIsCACNA1A #CureCACNA1A

