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 due to 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.

GET IN TOUCH

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Learn more about the CACNA1A Foundation

CACNA1A FOUNDATION

Understanding CACNA1A-related Ataxia

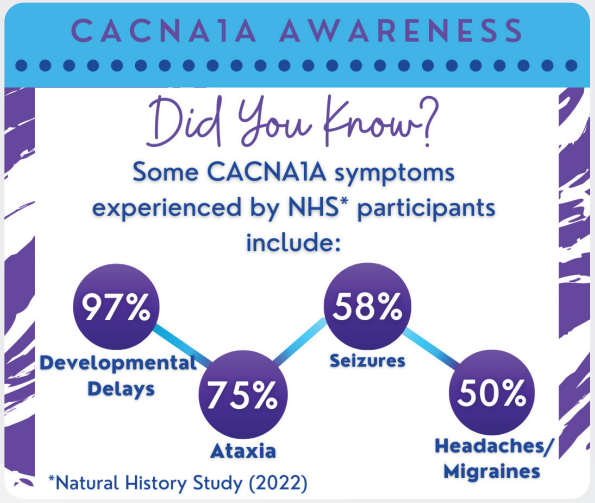
- Congenital Ataxia
- Cerebellar Ataxia
- Episodic Ataxia Type 2 (EA2)
- Spinocerebellar Ataxia Type 6 (SCA6)

WHAT IS CACNA1A?

CACNA1A is a gene located on the 19th chromosome that encodes for the α1 subunit of Cav2.1, a voltage-gated calcium channel expressed in the brain and abundant in the cerebellar granule and Purkinje cells.

Calcium flow 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 disorders
- 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

Clinical and genetic characterization of CACNA1A-related disease (1)

Lipman AR, Fan X, Shen Y, Chung WK. Clinical and genetic characterization of CACNA1A-related disease. Clin Genet. 2022 Oct;102(4):288-295. doi: 10.1111/cge.14180. Epub 2022 Jun 26. PMID: 35722745; PMCID: PMC9458680.

Rare CACNA1A mutations leading to congenital ataxia (2)

Izquierdo-Serra M, Fernández-Fernández JM, Serrano M. Rare CACNA1A mutations leading to congenital ataxia. Pflugers Arch. 2020 Jul;472(7):791-809. doi: 10.1007/s00424-020-02396-z. Epub 2020 May 26. PMID: 32458086.

From Genotype to Phenotype: Expanding the Clinical Spectrum of CACNA1A Variants in the Era of Next Generation Sequencing (3)

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

CACNA1A-Related Channelopathies: Clinical Manifestations and Treatment Options (4)

Indelicato, E., Boesch, S. (2023). CACNA1A-Related Channelopathies: Clinical Manifestations and Treatment Options. In: Handbook of Experimental Pharmacology. Springer, Berlin, Heidelberg.

https://doi.org/10.1007/164_2022_625

CONGENITAL ATAXIA

Congenital ataxia is a term associated with children who exhibit early signs (before the age of 2) and evidence of ataxia as the result of cerebellar disease. (2) One of the earliest symptoms of CACNA1A-related disease is hypotonia, either in the newborn period or the first year of life. (1) This symptom, combined with delayed motor milestones, paroxysmal tonic upgaze and speech and language delays, are markers for a pathogenic CACNA1A variant, yet they are often diagnosed retrospectively, typically after a child has had their first seizure or hemiplegic migraine, which should alert the medical community that there may be a genetic cause for the patient's issues. (2)

CEREBELLAR ATAXIA

Cerebellar atrophy is detected with magnetic resonance imaging (MRI) as a widening of the interfolia spaces in the cerebellum. However, it is often difficult to diagnose in early MRIs. (2) Nystagmus and gait ataxia are the most chronic symptoms. (1)

EPISODIC ATAXIA TYPE 2

Episodic ataxia type 2 (EA2) is characterized by sporadic attacks of ataxia that typically last minutes to days, with typical onset in childhood or early adolescence. The attacks are associated with nausea and vomiting, double vision, dysarthria, tinnitus, dystonia, hemiplegia, and headaches, including migraine headaches. Stress, caffeine, alcohol, exertion, fever, heat, and phenytoin can trigger attacks. (3)

SPINOCEREBELLAR ATAXIA TYPE 6

Spinocerebellar ataxia type 6 (SCA6) is an adult-onset neurodegenerative disorder caused by the expansion of a CAG repeat sequence of three DNA nucleotides at the end of the gene. This results in the degeneration of cerebellar Purkinje cells (important for motor coordination and balance). (3) For more information, visit www.sca6.net.

TREATMENT OPTIONS

Treatment options are limited. The CACNA1A Foundation is funding research on gene therapy and the discovery of new therapeutics. Current treatments include:

- acetazolamide
- 4-aminopyridine
- flunarizine
- topiramate
- acetyl-DL-leucine (Tanganil)

In addition, physical therapy is an important part of a rehabilitation program due to the loss of coordination and balance over time. (4)