The impact of *CACNA1C* real-world data: *CACNA1C* Community Registry (CCR) design

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The *CACNA1C* Community Registry is a patient registry for all *CACNA1C* gene related conditions, managed by Timothy Syndrome Alliance (TSA) and powered by Pulse Infoframe. The purpose of the *CACNA1C* Community Registry is to obtain insights to better characterise *CACNA1C*-related disorders, including Timothy Syndrome and LongQT8, and their presentation, management and treatment.
The registry was launched in June 2022 and is available for worldwide participation. This decentralised registry is not tied to a site and enables anyone, anywhere in the world with a *CACNA1C*-related disorder to sign up for the registry and participate from their home.

REGISTRY AIMS

- Allow researchers to study common aspects among the different conditions caused by variations in *CACNA1C*.
- Increase the visibility of *CACNA1C* so those navigating the many health concerns may be improved through research and clinical trials.
- Document how different variants present with different symptoms and outcomes.
- Assist researchers anywhere in the world interested in studying variations in this gene.

ESTABLISHING *CACNA1C* COMMUNITY REGISTRY

TSA set out to create a patient registry to help increase the visibility of *CACNA1C*-related disorders and gain insights that will help facilitate research into this group of rare genetic disorders. The Pulse Infoframe platform follows a governance framework that allows data to be collected and housed from multiple different rare disease communities. As a result, researchers are enabled to study multiple conditions. These capabilities made Pulse Infoframe the ideal candidate to build the *CACNA1C* Community Registry. With this partnership, Pulse Infoframe and Timothy Syndrome Alliance hope to facilitate better research for the *CACNA1C* community.
Different variants on this same gene present a variety of outcomes. By collecting data on one platform, TSA and researchers can work with patients anywhere in the world which has proven to be critical when studying diseases with patient populations this small.

PRELIMINARY FINDINGS

Results are based upon individuals with *CACNA1C*-related disorders (CRD) who consented to participate in the *CACNA1C* Community Registry between June 10, 2022, and May 5, 2023.
Through May 5, 2023, 43 individuals with CRD consented to participate in the registry, of which 32 completed forms that provided information on demographics, clinical characteristics, and symptoms.
Data presented here are based on these 32 participants.
As of June 2024 there are 73 participants in the *CACNA1C* Community Registry.

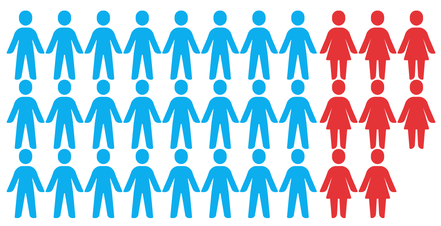
CONCLUSION

Families living with a *CACNA1C* variation face many difficulties, including having to educate many healthcare professionals about the condition. They also have limited treatment options. Continued enrollment and participation in the *CACNA1C* registry will help those affected by a variation and researchers in this gene in many ways:

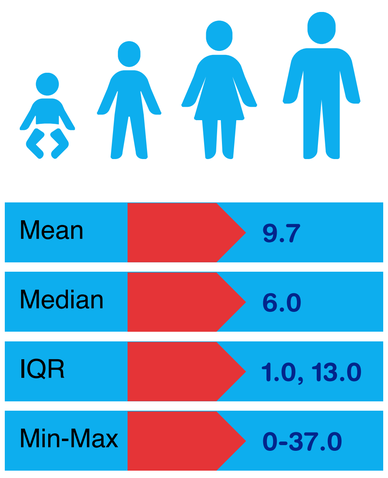
- Increase understanding of all *CACNA1C*-related disorders
- Encourage efficient and timely diagnosis
- Enable progress in research and clinical trials
- Facilitate the development of treatments for *CACNA1C*-related disorders

Demographics

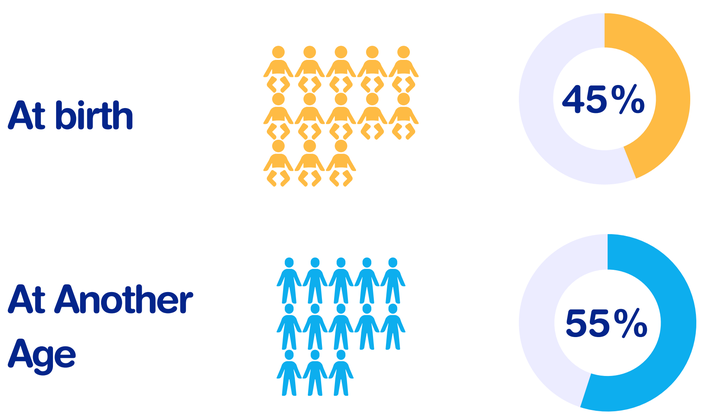
Gender



Age at Diagnosis



Age at First Symptom



Overview of current symptoms.

Symptom Categories *	Number of Participants reporting this symptom
Bowel problems (Gastrointestinal)	53.1%
Dental	34.4%
Dermatological/skin symptoms and abnormalities	18.8%
ENT (issues of the ears, nose and throat)	15.6%
Eyes (Ophthalmological)	15.6%
Genitourinary	9.4%
Heart symptoms (Cardiac-related symptoms or abnormalities)	25%
Lungs and breathing problems (Laryngeal & Respiratory)	46.9%
Multisystemic symptoms and abnormalities	12.5%
Musculoskeletal (i.e. diseases of the muscles, ligaments, tendons, cartilage and bones)	18.8%
Neurological (neuropsychiatric and neuropsychological) /Developmental, Behavioral or Mental Health	46.9%
Neurological (motor/movement symptoms and abnormalities)	34.4%
Neurological (seizures or epilepsy)	25%
Neurological (sensory deficits/disturbances to sensation)	28.1%
Neurology (diseases of the spinal cord)	6.2%
Bone Skin Muscle (Orthopaedical)	9.4%
Sensory conditions (i.e. diseases of the special senses)	9.4%
Speech and language	50%
Vascular or cardiovascular (together with heart problems)	9.4%

*Participants can present with multiple symptoms

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pulse infoframe

CACNA1C Community Registry
https://ccr.healthie.net/register

tsa Timothy Syndrome Alliance

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