Timothy Syndrome Alliance

A Warm Welcome

It’s been a while, but we’re back and excited to reconnect! Whether you're new or have been with us from the start, we’re glad you're here.

We’ve been working behind the scenes to strengthen our impact, and moving forward, our quarterly newsletters will keep you updated on research, collaborations, events, and how we’re achieving our mission for the global CACNA1C community—individuals, families, caregivers, researchers, scientists, healthcare professionals, advocates and supporters.

Our Why

The CACNA1C gene codes for a type of calcium channel found in cells throughout the human body. It is essential to life, and changes in the gene can result in significant health problems. CACNA1C-related disorders (CRDs), which include Timothy Syndrome and LongQT8, present as a spectrum of conditions, from both short and prolonged QT intervals (with the potential for fatal cardiac arrhythmias), to developmental delay, autism, fused fingers/toes, hip dysplasia, hypoglycaemia, abnormally low muscle tone and epilepsy. More symptoms continue to be identified and added to this list as researchers learn more from our community.

Individuals with CRDs typically need medication to manage epilepsy and heart symptoms, and many have implanted cardiac defibrillators. Developmental, neurological and mental health difficulties may impact their ability to engage in school and daily activities. To support their needs, it is essential to provide tailored individual support, which we continue to champion.

Our Strengthened Mission

In order to achieve our mission to improve diagnosis, treatment and care of individuals with CRDs, our focus is on five interdependent areas of activity – raising awareness, improving diagnosis treatment and care, supporting the global community, providing information and advice, and driving research and clinical development. By working together and collaborating globally, we can harness greater strength.

What's New?

RARE AS ONE

In October last year, we were delighted to be welcomed into the Chan Zuckerberg Initiative Rare As One Network (cycle 3).

This support, in its many forms, is already proving to be transformative—it is like installing an engine in the plane we've spent the last five years building. We will share more about our plans in future newsletters.

About Rare As One Project
Rare disease is anything but rare. As many as 10,000 rare diseases affect over 300 million people globally. The vast majority are not well understood, and approved treatments have been developed for less than 5% of them. Yet worldwide, patients are meeting these challenges head on. The Rare As One Project is committed to uniting these communities in their quest for cures. For more information, please visit www.chanzuckerberg.com.

What's on the Horizon?

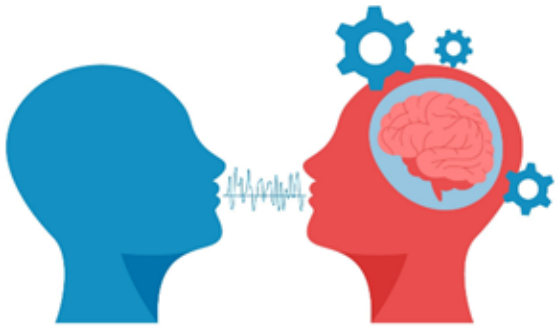
We're incredibly excited to announce that Connections has been shortlisted in the 2025 Smiley Charity Film Awards in the Longform Under £500,000 category. We're officially a finalist!

In this film, a collaboration between the Timothy Syndrome Alliance (TSA) and Cardiff University, we explore the importance of connections between individuals and families with lived experience of CACNA1C-related disorders (CRDs) and Timothy Syndrome (TS), academics, clinical teams and researchers. The more links we make, the stronger our network becomes and the faster we grow our understanding of our rare disease. Without connections, we can't grow or learn.

The judges now have the tough task of deciding the winning films in each category and results to be announced on the 20th March.

Conferences Ahoy!

We will be having another online conference open to all later this year.
In 2026, we will be having an in-person conference at Cardiff University, Wales



We are currently seeking funds to enable us to proceed with our Speech and Language study.

The knowledge from this study will help us learn more about diagnosis and prognosis for speech and language outcomes, and assist in tailoring therapies and supports specifically for our CACNA1C individuals and their families.

Community Registry

Analysis of individuals with CACNA1C variants in the CACNA1C Community Registry (currently 89 participants) will be combined with the neuro-psychiatric and developmental inventory studies that Dr Jack Underwood (Chair) and Dr Rebecca Levy, members of our Scientific Advisory Board, have been working on together. The goal is to publish this to expand the range of known and described phenotypes (symptoms).

CACNA1C Portal

We are building a CACNA1C Portal that will support clinical variant interpretation, enhance patient care management and identify biomarkers for clinical trials.

VGCCC
VOLTAGE-GATED
CALCIUM CHANNEL
COLLECTIVE

The Voltage-Gated Calcium Channel Collective (VGCCC), dedicated to raising awareness and promoting collaboration across the calcium channelopathies, will be launching our website on Rare Disease Day.

This group is composed of caregivers and individuals impacted by the channelopathies, alongside researchers and clinicians focused on the 10 calcium genes (which includes CACNA1C).

Shout Loud on Rare Disease Day -28th February 2025

Rare Disease Day is the globally coordinated movement on rare diseases, working towards equity in social opportunity, healthcare, and access to diagnosis and therapies for people living with a rare disease. Events, illuminating buildings and monuments are happening worldwide to shine the light on people living with a rare disease, including sharing experiences such as via the audible stories 'Rare on Air', which highlight the lived realities of those living with a rare disease.

Did you know Rare Diseases are collectively common? 1 in 17 people will develop a rare disease at some point in their lives - yet most have no cure.

We have in previous years made a couple of films to coincide with Rare Disease Days – please do watch and share them again.

The Diagnosis Challenge is all about the lengthy and frustrating journey from first noticing that something isn't right and going to the doctor to actually receiving a diagnosis—the diagnostic odyssey.

The average time it takes to diagnose a rare disease is 5.6 years.

Rare Strikes Back (made with puppets and lots of golf tees!) is an award-winning film balancing the reality of living with a rare condition with the hope of the Rare Disease Community articulating the conversations about Rare Diseases regarding the pandemic.

Can you help fuel our plane?

We are flying but we need fuel to run. TSA is a lifeline—small, often invisible, yet critical. Your donations keep us strong & impactful.

Behind every successful project is a strong foundation. Your support helps cover core costs—the essential expenses that keep our charity running effectively. These costs allow us to plan ahead, grow sustainably and maximize our impact, rather than just moving from project to project.

In 2024, individual donations contributed £5,454.77 ($6,846.73) toward these vital needs. This funding is the fuel that powers our mission, ensuring we can continue delivering meaningful change. With your support, we don't just survive—we thrive!

Did you know we have been certified as the equivalent of a U.S. public charity, allowing U.S. donors to give with confidence via your Donor-Advised Fund (DAF) or intermediary organisations like CAF America and GlobalGiving?

Important: ensure you select the correct organisation using our Registered Charity number: **1185523**.

💼 Workplace Giving via Benevity – If your employer uses Benevity, you can donate in your local currency and may even get a company match! Just search for Timothy Syndrome Alliance (TSA) in the Benevity portal.

💳 Donate via JustGiving – JustGiving accepts donations worldwide! Your donation will be processed in GBP, but you can give in your local currency and it will be converted automatically at checkout.

We know not everyone can give financially, but sharing our work on social media helps us reach those who can support—as well as those who are impacted by CACNA1C and would benefit from our support. Every post, like, and share makes a difference!

Join the conversation

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Our mailing address is: admin@timothysyndrome.org

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