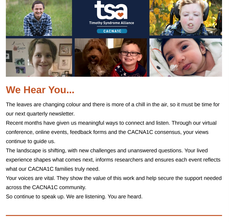
Timothy Syndrome Alliance

CACNA1C



We Hear You...

The leaves are changing colour and there is more of a chill in the air, so it must be time for our next quarterly newsletter.
Recent months have given us meaningful ways to connect and listen. Through our virtual conference, online events, feedback forms and the CACNA1C consensus, your views continue to guide us.
The landscape is shifting, with new challenges and unanswered questions. Your lived experience shapes what comes next, informs researchers and ensures each event reflects what our CACNA1C families truly need.
Your voices are vital. They show the value of this work and help secure the support needed across the CACNA1C community.
So continue to speak up. We are listening. You are heard.

Connect CACNA1C Conference 2025

On Saturday 20 September, we were delighted to bring together 70+ attendees from 19 countries for our global CACNA1C community online conference.

Over the course of this four-and-a-half-hour programme, we had four expert presentations and two powerful stories of impact, all translated in real time thanks to the Wordly platform so attendees could listen and read in their preferred language. A great Q&A session completed our conference, giving participants an opportunity to put questions to our speakers.

For anyone who missed it or would like to watch again, the programme speakers are listed below. You can click on each name to access the recording, all of which include subtitles in 19 languages.

- TSA Scientific Advisory Board Updates by Dr Jack Underwood, WCAT Clinical Lecturer in Forensic Psychiatry at Cardiff University and Chair of the Scientific Advisory Board for the Timothy Syndrome Alliance
- Rareminds: Mental health and CACNA1C by Rebecca Hargreaves, Lead Counsellor CACNA1C at Rareminds, UK
- The importance of understanding communication in CACNA1C-related disorders by Dr Miya St. John, Researcher and Speech Pathologist at Murdoch's Children's Research Institute, Australia
- Genetics of CHDs and how this informs treatment options by Dr Rebecca Levy, Member of TSA Scientific Advisory Board and Neurocientist at Stanford Medicine Children's Health, USA
- Story of Impact: Lincie - she's not one in a MILLION, she's one in 8.2 BILLION by Jill Wilt, Parent of Lincie, USA
- Story of Impact: Rose but Loyal: giving every voice a place to shine by Gemma Duncan, Parent of Noah

Following on from our Conference, we had a further live presentation in mid-October:

- Cognition and CACNA1C by Dr Amy Arnsten, Albert E. Kent Professor of Neuroscience & Professor of Psychology, and Member of the Kavli Institute of Neuroscience, Yale University, USA.

Joined by 20+ attendees from 10 countries, Dr Arnsten spoke about new research findings of the importance of CACNA1C to the cognitive functioning of the prefrontal cortex. She outlined the numerous functions of the prefrontal cortex in humans, including language, working memory, abstract reasoning, social cognition, the executive functions, and top-down control, and shared data on the narrow "inverted U" relationship between CaV1.2 and prefrontal function. She has found that CaV1.2 are essential to prefrontal function, but that excessive actions, as occurs with stress, impairs prefrontal function. This may explain why either too- or gain-of-function mutations in CACNA1C impair prefrontal cortical abilities.

In late September, there was a Q&A with Dr Miya St John following on from her talk at our Conference. To protect the personal information of attendees, this Q&A session was not recorded. Instead, we have prepared a summary of the key questions and answers.

We need your voice...

Please tell us what you thought about the conference and the recordings. Your feedback helps us create future events that are relevant, engaging, and meaningful to you.
As we rely 100% on grants and donations, this feedback also helps us share how these conferences and events benefit you.

Click here to share your feedback and help shape what comes next.

REGISTER YOUR INTEREST TO ATTEND

Connect CACNA1C *In-Person* Global Network Conference
22/23 July 2026
@ Cardiff, UK

We're thrilled to announce that the next Connect CACNA1C Global Network Conference will be held 22-23 July 2026 in Cardiff, UK. This inaugural in-person event aims to unite all those committed to improving the lives of individuals with CACNA1C-Related Disorders and Timothy Syndrome.

But why travel all the way to the UK? Because there is so much more than the conference itself.

Connections
A unique opportunity to connect with families, researchers, clinicians, and healthcare professionals, building relationships that provide support, foster collaboration, and help advance research, therapies, and ultimately a cure.

Sharing
Connect with others who understand your experiences and exchange knowledge, stories, and guidance to support one another.

Being Involved
You can make a real difference by taking part in research. Attending the conference in person gives you the opportunity to donate blood samples, which may be reprogrammed into iPSC cells to help advance scientific understanding of CACNA1A-Related Disorders.

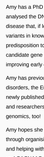
Be a tourist!
Not only does Cardiff and beautiful Wales have a lot to offer, but why not take the opportunity to have an unforgettable family holiday, making memories in London, England - or even Europe. Don't waste the trip - maximise it!

Team TSA are hard at work already making arrangements, so keep an eye on your inbox, our social media profiles and these newsletters for further details about the conference programme, registration, hotel bookings, travel bursaries/ assistance and childcare.

Click here to register your interest in attending

What's New?

We've Hired...



a Research and Public Engagement Officer, adding another vital part to the engine room of our charity. Introducing **Amy Houseman** who will help develop and oversee communications and engagement with all our key international stakeholders, such as researchers, clinicians, healthcare professionals, our CACNA1C community and the wider public.

Amy has a PhD in Rare Disease Genetics from Cardiff University. For her PhD, she analysed the DNA sequences from over 300 patients with colorectal polyposis, a rare disease that, if left untreated, can develop into bowel cancer. Amy searched for pathogenic variants in known and new genes to determine how some patients may have a genetic predisposition to bowel cancer. Her work has led to the identification of a promising candidate gene that may play a role in bowel cancer, representing a step towards improving early diagnosis and personalised care for those at-risk patients.

Amy has previously volunteered with Unique RareChromo to create leaflets for rare disorders, the European Journal of Human Genetics to help with their engagement of newly published articles and has organised numerous science events for both the public and researchers. She has also been all the way to Shanghai for her placement year in genomics, too!

Amy hopes she can make a difference to patients and families with CACNA1C variants through organising communication and engagement activities, liaising with researchers, and helping with the research portal! She will help raise the profile of Timothy Syndrome and CACNA1C-related disorders and also help coordinate global research efforts, knowledge-sharing, collaboration and accelerate patient-prioritised research.

This appointment has been enabled by our RareAsOne multi-year funding from the Chan Zuckerberg Initiative (CZI).

We're analysing...

thanks to a grant from the Hodge Foundation, your data in the CACNA1C Community Registry will be analysed by Dr Jack Underwood to look at the wide range of presentations, symptoms and experiences of individuals with CACNA1C variants. All necessary permissions and approvals have been obtained to ensure your information is used safely, legally, and ethically.

If you haven't signed up and completed the surveys and included your genetic report already, now is a really good time to do so.

Make sure you are counted.

Coming soon...

the results of the language consensus project: a set of guidelines for the diagnosis of individuals presenting with variants in CACNA1C, developed by an international panel of experts with the input and involvement of the CACNA1C community. The principal aims are to have the language used around syndromic presentations linked to CACNA1C variants formalised, the classical Timothy Syndrome phenotype reaffirmed and a new umbrella syndrome defined. Furthermore, the work will present minimum expected standards of clinical care for individuals with CACNA1C-Related Disorders or Timothy Syndrome, which has far-reaching implications for treatment and care.

Why support TSA?

- Because we're not filling a gap. We're building the system.
- Because families and scientists deserve to work side-by-side.
- Because what's rare to the world is everything to us.
- Because with you, the impossible becomes possible.

Donate Today!

UK Donors (GBP)

US Donors (USD)

International Donors (EUR, CAD, AUD)

Spread the Word...

We know not everyone can give financially, but sharing our work on social media helps more than you might think. It spreads awareness, reaching those who can support our impact and those living with CACNA1C-Related Disorders who face an immense burden, managing crisis-level symptoms within systems that still don't understand their needs. Every post, like, and share truly makes a difference, so please hit that button!

Thank you for reading this edition of our TSA newsletter and giving your continued support. TSA is run by parents and volunteers and we thank every one of you.

Team TSA

Join the conversation

Our mailing address is:
admin@tsyndrome.org