CONNECT CACNA1C GLOBAL NETWORK CONFERENCE 2025

CARDIFF UNIVERSITY
PRIFYSGOL CAERDYDD

Neuroscience and Mental Health Innovation Institute
Sefydliad Arloesedd Niwrowyddoniaeth ac Iechyd Meddwl

Language Accessible Virtual Event

Language Accessibility
Powered by Wordly.ai

SATURDAY 20 SEPTEMBER 2025

3pm - 7.30pm BST

To register, email:
admin@timothysyndrome.org

CONNECT CACNA1C GLOBAL NETWORK CONFERENCE 2025

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CONNECT CACNA1C GLOBAL NETWORK CONFERENCE 2025

WELCOME

A very warm welcome to every attendee, speaker and supporter joining us.
We are delighted to bring together our global CACNA1C community – across time zones, borders and languages. Wherever you are, and whichever CACNA1C variant is part of your story, this conference is for you.

Whether you are living with CACNA1C, a parent or carer, researcher, clinician, advocate or friend – you are part of this shared space. Over the course of this four-and-a-half-hour programme, you'll hear four expert presentations and two powerful stories of impact, join small-group conversations on the issues that matter to you, and have the chance to put your questions directly to the experts and lived-experience speakers. We want this time to be useful for you – to inform, to reassure, and to help you find the contacts and information you need.

Thanks to live language access via the Wordly platform, everyone can take part in real time – listening or reading in their preferred language. After the conference, recordings will be published with manually checked transcripts and translations (usually made available in up to 14 languages) to ensure the best possible accuracy for our global community.

What to expect:

- **Concise updates** from members of our Scientific Advisory Board and invited experts across science, research and lived experience.
- **Focused breakout rooms** so you can connect on topics most relevant to you.
- **A dedicated Q&A** where you can put your questions to the speakers.
- **A key update** from Dr Jack Underwood on the CACNA1C Consensus Statement on Language and Management.

We are proud to be a UK-based charity with an international reach – supporting anyone, anywhere, with a rare CACNA1C variant finding. No matter where you are in the world, you belong here.

We hope you leave this session feeling better informed, connected and quietly more confident about the next step in your journey. Thank you for being with us – we're really pleased you're here.

Any queries? Please email Mission Support at admin@timothysyndrome.org

TSA SCIENTIFIC ADVISORY BOARD

DR JACK UNDERWOOD

Wellcome Trust GW4-CAT Clinical Research Fellow, NMHII, Cardiff University

DR REBECCA LEVY

Clinical Scholar, Neurology & Neurological Sciences. Postdoctoral Scholar, Neurology & Neurological Sciences, Stanford Medicine

DR GEMMA WILKINSON

Research Associate, NMHII, Cardiff University

DR NICOLA HALL

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Bambino Gesù Children Hospital & Research Institute, IRCCS, Rome

DR WILFRIED HAERTY

Group Leader of Evolutionary Genomics, Earlham Institute

PROF. LIZ TUNBRIDGE

Director of Translational Neuroscience, Boehringer Ingelheim

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PROGRAMME

3pm

Conference Opens

3:10pm

Welcome and Introduction

Speakers: Sophie Muir (TSA) & Dr Jack Underwood (NMHII, Cardiff University)

3:15pm

Rareminds: Mental Health and CACNA1C Followed by Q&A specific to this topic

Speaker: Rebecca Hargreaves (Lead Counsellor CACNA1C, Rareminds)

3:45pm

The importance of understanding communication in CACNA1C-related disorders. [Pre-recorded]

Speaker: Dr Miya St John (Researcher & Speech Pathologist, Murdoch Children's Research Institute)

4:05pm

Discussion Rooms

Breakout Room 1: Researchers, Scientists, Medical Professionals

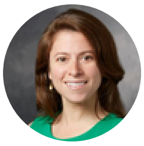
Breakout Room 2: Patients, Families, Parents, Carers

4:35pm

BREAK

Grab a cuppa, have a comfort break and see you in 15 minutes!

4:50pm



Genetics of CRDs and how this informs treatment options.

Speaker: Dr Rebecca Levy (TSA Scientific Advisory Board Member, neuroscientist at Stanford Medicine Children’s Health)

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Timothy Syndrome Alliance
CACNA1C

PROGRAMME

5:10pm

Story of Impact

Linzie: She's not one in a MILLION, she's one in 8.2 BILLION

Speaker: Jill Witt, parent of Linzie

5:30pm

BREAK

Grab another cuppa, a snack, have a comfort break and see you in 20 minutes!

5:50pm

Diverse Effects of Timothy Syndrome Variants on Channel Function

Speaker: Dr Ivy E. Dick, Associate Professor at School of Medicine, University of Maryland

6:10pm

Story of Impact

Rare but Loud: Giving every voice a place to shine

Speaker: Gemma Duncan, parent of Noah

6:30pm

Discussion Rooms

Breakout Room 1: Join **Drs Ivy Dick & Rebecca Levy**
Breakout Room 2: Join **Gemma Duncan & Sophie Muir**

7:00pm

Q & A

Put any other questions you may have to the panel.

7:30pm

Conference Closes

SPEAKERS

DR JACK UNDERWOOD

Wellcome Trust GW4-CAT Clinical Research Fellow, NMHII, Cardiff University. Jack is a Forensic Psychiatry Registrar Doctor and his work focuses on rare and common genetic variants associated with autism and neurodevelopmental conditions. His interest in CACNA1C is to characterise the phenotype-genotype relationship with the goal of targeted therapeutics and a greater understanding of the neurobiology.

SOPHIE MUIR

Sophie's son Calvin was diagnosed with CACNA1C in 2016 through the DDD study and 100,000 Genomes Project, after almost 10 years of searching. In 2019, Sophie registered TSA as a charity and now leads the day-to-day operations, focusing on increasing awareness, growing our community, improving understanding and generating research interest. With an enthusiasm to understand CACNA1C, Sophie is committed to improving the information and support for anyone affected by CACNA1C.

REBECCA HARGREAVES MBCAP (ACCRED.)

Rebecca is an Adult and Young Peoples Counsellor with a particular interest in the psychological impact of health conditions on the family as a whole. She originally worked as a nurse for over 20 years, and was also the Lead for a large school counselling service. Rebecca has experience of working with families impacted by CACNA1C and is the Lead Counsellor for the Timothy Syndrome Alliance Counselling Service.

DR MIYA ST. JOHN

Dr Miya St John is a speech pathologist and post-doctoral researcher at the Murdoch Children's Research Institute and lecturer at the University of Melbourne. Her research interests include phenotyping speech and language in rare genetic conditions and understanding the best evidence for motor speech treatment.

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SPEAKERS

DR REBECCA LEVY

Rebecca is a Member of our Scientific Advisory Board and is a child neurologist, neurogeneticist and neuroscientist studying rare neurologic genetic disorders at Stanford Medicine Children's Health.

JILL WITT

Jill's daughter Linzie was diagnosed in January 2024, after nearly six months of various doctor appointments, following an order for genetic testing from her Neurologist. Jill is an Insurance Agent by day, and a mom to 4 children aged 13-18. In her free time, Jill dabbles in photography, raises chickens, loves fishing and helping around her husband's Auto Repair business.

DR IVY E. DICK

Ivy is Associate Professor at School of Medicine, University of Maryland. Ivy researches the mechanisms underlying the regulation of calcium channels, and how those mechanisms are disrupted in Timothy Syndrome.

GEMMA DUNCAN

Gemma is a UK-based mum to Noah, wife to Paul, and proud trustee with the Timothy Syndrome Alliance. "I'm passionate about supporting the rare disease community, spending time with my family, and giving plenty of cuddles to our 4 cats!"

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FUTURE LIVE ONLINE PRESENTATIONS

DR AMY ARNSTEN

Albert E. Kent Professor of Neuroscience & Professor of Psychology;
Member, Kavli Institute of Neuroscience, Yale University

We are delighted to announce that Amy will be giving a virtual talk on *“Cognition and CACNA1C”* on **Friday 17 October 2025 at 10:00 am EDT (New York) / 3:00 pm BST (London) / 2:00 pm GMT (Cardiff) / 8:00 am MDT (Denver) / 11:00 pm AEST (Melbourne, Australia).**

Dr. Arnsten will speak about their new research findings of the importance of CACNA1C (Cav.12 calcium channels) to the cognitive functioning of the prefrontal cortex. She will outline 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 then talk about 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 loss- or gain-of-function mutations in CACNA1C impair prefrontal cortical abilities.

About: Dr Amy Arnsten studies the molecular regulation of higher cortical circuits, identifying the unique mechanisms needed for cognition and the executive functions. She is discovering why these circuits are particularly vulnerable to genetic insults, and to changes with stress and inflammation that increase risk of cognitive disorders such as schizophrenia and Alzheimer’s Disease. Dr Arnsten received her BA in Neuroscience with Honors from Brown University in 1976, her PhD in Neuroscience from UCSD in 1981, with postdocs at Cambridge with Susan Iversen, and then Yale with Patricia Goldman-Rakic. In 1986, she became Assistant Professor at Yale, and is currently the Albert E. Kent Professor of Neuroscience. She is a member of the National Academy of Medicine, and received the Goldman-Rakic Prize for Outstanding Research in Cognitive Neuroscience. Her research has led to the development of guanfacine (Intuniv™) for the treatment of ADHD and other prefrontal disorders. Dr Arnsten's recent research has shown that the prefrontal cortex requires just the right amount of Cav1.2 (CACNA1C) actions, and that either too little or too much impairs prefrontal cortical function.

FUTURE LIVE ONLINE PRESENTATIONS

DR MIYA ST. JOHN

Dr Miya St John is a speech pathologist and post-doctoral researcher at the Murdoch Children's Research Institute and lecturer at the University of Melbourne.

If you found Miya's pre-recorded talk on *"The Importance of Understanding Communication in CACNA1C-related Disorders"* valuable, you'll want to join her and the team at Murdoch Children's Research Institute for a live follow-up session on **Sunday 28 September 2025 at 2:00 pm BST (London) / 9:00 am EDT (New York) / 7:00 am MDT (Denver) / 11:00 pm AEST (Melbourne, Australia).**

In that session, they'll go deeper into speech and language issues in CACNA1C-related disorders, and explain more about the upcoming Speech & Language research study we're preparing. This study (once we reach our funding goal) aims to deliver insights on diagnosis and prognosis of speech and language development, so we can better tailor therapies and supports for individuals and families living with CACNA1C variants.

You'll also have the chance to ask your questions — about this research, about speech and communication more broadly, or anything else related to CACNA1C.
We'd love you to join us and engage with what's coming next.

SAVE THE DATES!

So mark those dates in your diary for these talks from Dr Amy Arnsten and Dr Miya St . John.

Use these links to book your spot with Dr Miya St. John and/or Dr Amy Arnsten

CACNA1C Awareness Day is on the 1st October
Please show your support

DISCUSSION ROOMS

SESSION 1:

Room 1 will be for researchers, clinicians and scientists to share their own experiences and work on the CACNA1C, with an aim to forging links and connections for future work.

Room 2 will be for families and individuals from across the world to connect, as an opportunity for people to meet, and share stories and experiences.

Attendees will be free to choose which room they wish to enter within the Teams software, so we ask you to attend the room suitable for your background.

Please keep your cameras on wherever possible in these breakout sessions: it's really great to be able to see our community and makes such a difference when talking to people - we may be virtual but we are human!

SESSION 2:

Join our guest speakers and attending Scientific Advisory Board members in separate rooms where attendees will have the opportunity to ask questions and discuss issues in smaller groups.

Room 1: Doctors Ivy Dick and Rebecca Levy

Room 2: Gemma Duncan and Sophie Muir

Does the Wordly App for Teams work in the Discussion Rooms?

Unfortunately the Wordly app does not support Teams Breakout Rooms. In order to provide translation, Wordly needs access to the audio stream, which is not available in Teams Breakout rooms.

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Timothy Syndrome Alliance
CACNA1C

WHY YOUR SUPPORT MATTERS

- Because we have a rare opportunity to lead bold and lasting change. To build something that's never existed before. But we need your support.
- Because we have a strategy – and it's working. We're leading a multi-pronged plan that includes real-world data, precision research, international clinical guidance and family-led advocacy. With your help, we're moving from scattered effort to focused progress.
- Because there's no safety net. Without dedicated clinical pathways, families are left to piece together care themselves. With your help, we're building global resources that clinicians and families can rely on.
- Because research needs roots. Data is fragmented and underfunded. You help us grow the CACNA1C Community Registry — one piece of a broader research infrastructure including cellular modelling, variant analysis and therapeutic development.
- Because what we learn here matters far beyond rare disease. CACNA1C is also linked to major mental health conditions (e.g. schizophrenia, bipolar disorder and depression). When you support this work, you're investing in discoveries that could impact millions of lives.
- Because without community, there is no progress. You help bring families, researchers and clinicians together — shaping a future where science is guided and informed by lived experience.
- Because everything is ready — except the funding. We have the partnerships. The roadmap. The global network. What we need now is the fuel to move faster.

Because this is **urgent**.

Because this is **ready**.

Because **without us**, nothing moves.

And **with you**, everything can.

CONNECT CACNA1C GLOBAL NETWORK CONFERENCE 2025

tsa
Timothy Syndrome Alliance
CACNA1C

WHY YOUR SUPPORT MATTERS

Choose the right way to give
Wherever you live, there's a simple way to support this mission.

UK-based donors

DONATE

Eligible for Gift Aid
Supports TSA core programmes

US-based donors

DONATE VIA RARE VILLAGE 501(C)(3)

Immediate US tax receipt
Powered by our US fiscal sponsor

Global donors

Accepted currencies:

- British pounds (GBP)
- US dollars (USD)
- Euros (EUR)
- Canadian dollars (CAD)
- Australian dollars (AUD)

PayPal

DONATE

JustGiving™

DONATE

Join the conversation

We know not everyone can give financially, but giving a moment of your time is just as valuable. Sharing our posts on social media or forwarding our quarterly newsletter helps spread the word, reaching those who can help support our work - and those who are impacted by CACNA1C and need our support.

Every post, like and share makes a difference - and it takes less than a minute to hit that button! 💟

CONNECT CACNA1C GLOBAL NETWORK CONFERENCE 2025

CELEBRATING EVERYONE WHO MAKES THIS POSSIBLE

Where we are today is because of you. Every connection made, every insight shared, every conversation started has brought us to this moment. This conference is another step forward on a journey that belongs to all of us.

We are grateful to our guest speakers and to every member of our Scientific Advisory Board – whether presenting or attending – for giving their time and expertise so freely. A special thank you to Jack Underwood, our Scientific Advisory Board Chair, whose leadership helps unite science with our patients and lived experience voices to drive progress that matters.
Thank you to the families who will soon share their stories of impact. Your honesty and courage guide the priorities that shape research and care for the future.

We are thankful to NMHII, Cardiff University, for hosting this year's event, and to Daniel Haynes, PhD student funded by the Hodge Foundation, NMHII, Cardiff University, and Amanda, our Mission Support Officer funded by CZI, for their careful planning and coordination. And to everyone who connects with us and collaborates with us throughout the year – thank you. Your partnership is how we make lasting change.

This is our sixth year as a charity, and each year our global community grows stronger. Since January, we've welcomed families at a pace of around one per week. When TSA was registered in 2019, there were just 43 known individuals worldwide living with CACNA1C-related disorders. We are now over 250. Together, we are creating connection where there was isolation – and momentum where there was uncertainty.

We also celebrate our first year as a Cycle 3 grantee of the Chan Zuckerberg Initiative Rare As One Network. Their support is helping us strengthen an already thriving international network and expand opportunities for patient-led collaboration and research worldwide.

And this is only the beginning. In 2026, we will come together for our in-person conference at Cardiff University — our first since 2019. It will be a rare and special opportunity for families, researchers, and clinicians to share the same room, deepen collaboration, and work side by side to accelerate progress on the questions that matter most to you.

To everyone living with a CACNA1C-related disorder and Timothy Syndrome, to parents, carers, and supporters: thank you for being here. This is your community, your network, your movement.

Together, we are stronger – and together, we are shaping the future.

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TIME ZONES

3pm-7:30pm	British Summer Time (BST)(example: London, UK)
7am-11:30am	Pacific Daylight Time (PDT)(example: Los Angeles, USA)
8am-12:30pm	Mountain Daylight Time (MDT) (example: Denver, USA)
9am-1:30pm	Central Daylight Time (CDT) (example: Chicago, USA)
10am-2:30pm	Eastern Daylight Time (EDT)(example: New York, USA)
11am-3:30pm	Atlantic Time (AT)(example: Halifax, Canada)
11.30am-4pm	Newfoundland Time (NT)(example: St. John's, Canada)
4pm-8:30pm	Central European Summer Time (CEST)(example: Paris, France)
5pm-9:30pm	Eastern European Summer Time (EEST)(example: Athens, Greece)
7pm-11:30pm	Moscow Time (MSK)(example: Moscow, Russia)
7:30pm-12am	Indian Standard Time (IST)(example: Mumbai, India)
10pm-2:30am	China Standard Time (CST)(example: Beijing, China)
11pm-3:30am	Japan Standard Time (JST)(example: Tokyo, Japan)
21st Sept-4:30am	Australian Eastern Standard Time (AEST)(example: Sydney, Australia)
2am-6:30am	New Zealand Standard Time (NZST)(example: Auckland, New Zealand)

WORDLY AI TRANSLATION USER GUIDE

How to Attend a Microsoft Teams Meeting with Wordly

Key Things to Know

- No Wordly account is needed to join—you just need the Attendee Link or QR code.
- You don’t need a paid Wordly account or minutes—attendees aren’t charged. You are joining a Wordly translation session hosted by us.
- You can switch languages mid-event if available—look for language or interface options once you're in the session.
- To exit, simply close the browser tab or tap the exit button provided in the interface.

1. Receive the Attendee Link

As an attendee, you’ll be provided with an Attendee Link. This link often arrives via email or through event communications and typically looks like:
https://attend.wordly.ai/join/ABCD-1234

If needed, the Session ID (e.g., ABCD-1234) may also appear at the end of the link.

2. Joining the Session

Whether you use a desktop or mobile browser, the flow is similar:

On Desktop (Browser):

1. Click the Attendee Link—it will open in your default browser.
2. Once the page loads:
    - Choose your preferred language from the dropdown (e.g., if the speaker is in English and you want to read captions in French).
    - If not already filled in, enter the Session ID (four letters/digits) if required.
    - Click Attend to join the session.

When you're ready, close the browser/tab or click the exit button to leave.

WORDLY AI TRANSLATION USER GUIDE

On Mobile (Browser):

- Tap the Attendee Link (or use a QR code if provided)—it opens in your mobile browser.
- Select your translation language from the dropdown.
- Enter the Session ID if required.
- Tap Attend to begin the session.
- To leave, close the tab or use the exit button.

Via QR Code (Mobile Only):

- Scan the QR code using your phone’s camera or barcode scanner.
- The Wordly Attend page opens automatically.
- Choose your preferred language, enter the Session ID if needed, and tap Attend.
- Exit by closing the browser tab or using the exit button.

3. Alternate: Wordly App (iOS)

If you're using the Wordly iOS app:

- You can enter the Session ID manually, or—
- If you're signed in, select your session and tap the green “Start” button.

Quick-Start Summary Table

Platform	**Steps**
Browser (Desktop)	Click Attendee Link → choose language → enter Session ID (if needed) → click Attend. Close when done.
Browser (Mobile)	Tap Attendee Link (or scan QR) → choose language → enter Session ID (if needed) → tap Attend. Exit when done.
Wordly iOS App	Open app → enter Session ID (or select session if signed in) → tap Start.