Bringing together the global CACNA1C community of families, researchers and clinicians.

tsa

Timothy Syndrome Alliance

CACNA1C

CONNECT CACNA1C GLOBAL NETWORK CONFERENCE 2026

PROGRAMME

PREVIEW

To register attendance, please visit
https://timothysyndrome.org/conference/

Haydn Ellis Building,
Cardiff, UK

22nd-23rd July 2026

Language-accessible in-person event,
with recordings available post event

Cardiff University / Prifysgol Caerdydd — Neuroscience and Mental Health Innovation Institute / Sefydliad Arloesedd Niwrowyddoniaeth ac Iechyd Meddwl
RARE AS ONE
Cardiff University / Prifysgol Caerdydd — Biobank / Banc Bio
Advanced Therapies Wales / Therapïau Datblygiedig Cymru
Midlands-Wales Advanced Therapy Treatment Centre
rareminds — Mental Health for the Rare Disease Community

CONNECT CACNA1C GLOBAL NETWORK CONFERENCE 2026

WELCOME

A very warm welcome to all attendees, speakers and supporters joining us. We are delighted to bring together our **global CACNA1C community across borders and languages**. Whatever CACNA1C variant or research area is part of your story, this conference is for you. Whether you are living with a CACNA1C-Related Disorder, or are a parent, carer, researcher, clinician, advocate or friend, you are part of this shared space.

Over this two-day conference, you will hear from more than 10 world-leading CACNA1C researchers, alongside poster presentations from over 15 additional scientists in the field.

On the second day, we will focus on developing a patient-prioritised research agenda, bringing families and researchers together to shape the future direction of CACNA1C research.

We hope this time will be valuable for you, helping to inform, reassure, and connect scientists and families with the people and information they need.

Any queries? Please email admin@timothysyndrome.org

Timothy Syndrome Alliance (TSA) is a Registered Charity in England (Charity number: 1185523).

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TIMOTHY SYNDROME ALLIANCE

SOPHIE MUIR
Chair of Timothy Syndrome Alliance (TSA)

DR AMY HOUSEMAN
Research and Public Engagement Officer

AMANDA SANDERS
Mission Support Officer

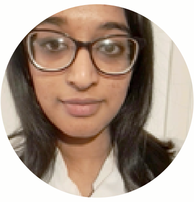
TRUSTEES

NICK MUIR

GEMMA DUNCAN

GALINA GARDINER

SUSAN BRESNAHAN



ACHSAH JAMES

GLENN MILLER

CHRISTINE ZIEGLER

MEG MCLAUGHLIN

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TSA SCIENTIFIC ADVISORY BOARD

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Group Leader of Evolutionary Genomics, Earlham Institute

This is a pre-event preview of the programme.

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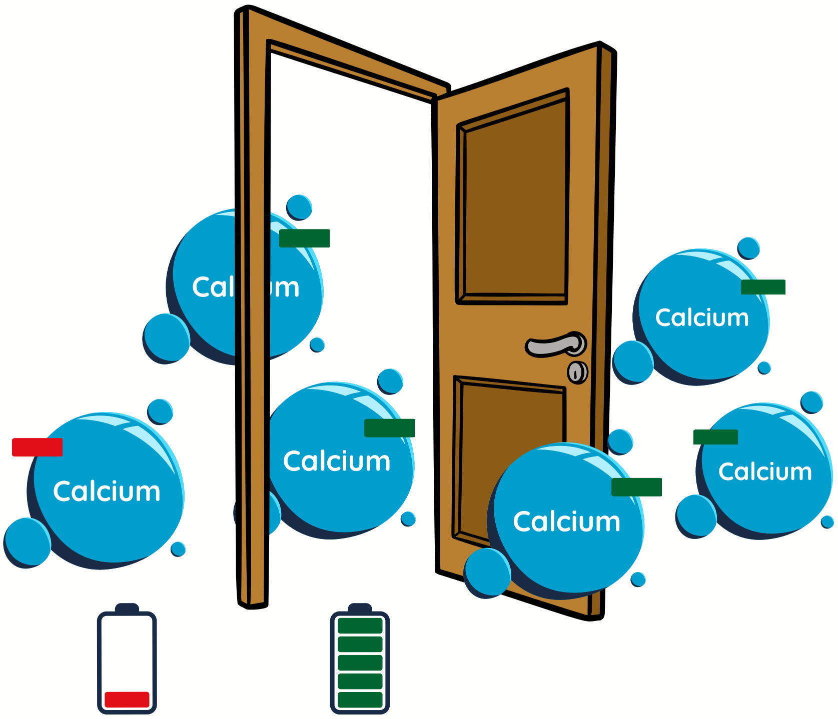
WHAT ARE CACNA1C-RELATED DISORDERS?

CACNA1C-Related Disorders is an umbrella term used to describe individuals who have rare genetic variants in the CACNA1C gene.

CACNA1C provides instructions to make a protein called a voltage-gated calcium channel (CaV1.2) - which is like a doorway on the surface of a cell. The channel opens and closes, allowing calcium to move in and out of a cell. The movement of calcium through the channel also creates a change of electrical charge, which can transmit signals between cells. These electrical signals control your heart's rhythm, communication between brain cells, and the function of your muscles, amongst many other processes in the body!



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WHAT ARE CACNA1C-RELATED DISORDERS?

Because this calcium channel is found in many different tissues, CACNA1C-Related Disorders typically affect more than one system in the body. Individuals can experience a combination of symptoms across the following areas (and more!):

- **Neurological:** hypotonia, epilepsy and seizures.
- **Neurodevelopmental**: intellectual disability, developmental delay, autism and ADHD.
- **Cardiac**: prolonged QT interval (and possibly shortened), abnormal heart rhythm, and structural cardiac abnormalities.

Some individuals may also have fused fingers/toes, low blood sugar, hip dysplasia, gut or bowel problems, and/or be susceptible to infections.

Symptoms associated with CACNA1C-Related Disorders are usually not inherited, and occur randomly by chance at conception (*de novo*). Some variants have been found to run in families, but this is extremely rare.

Importantly, not all individuals with a CACNA1C variant will present with the same symptoms!

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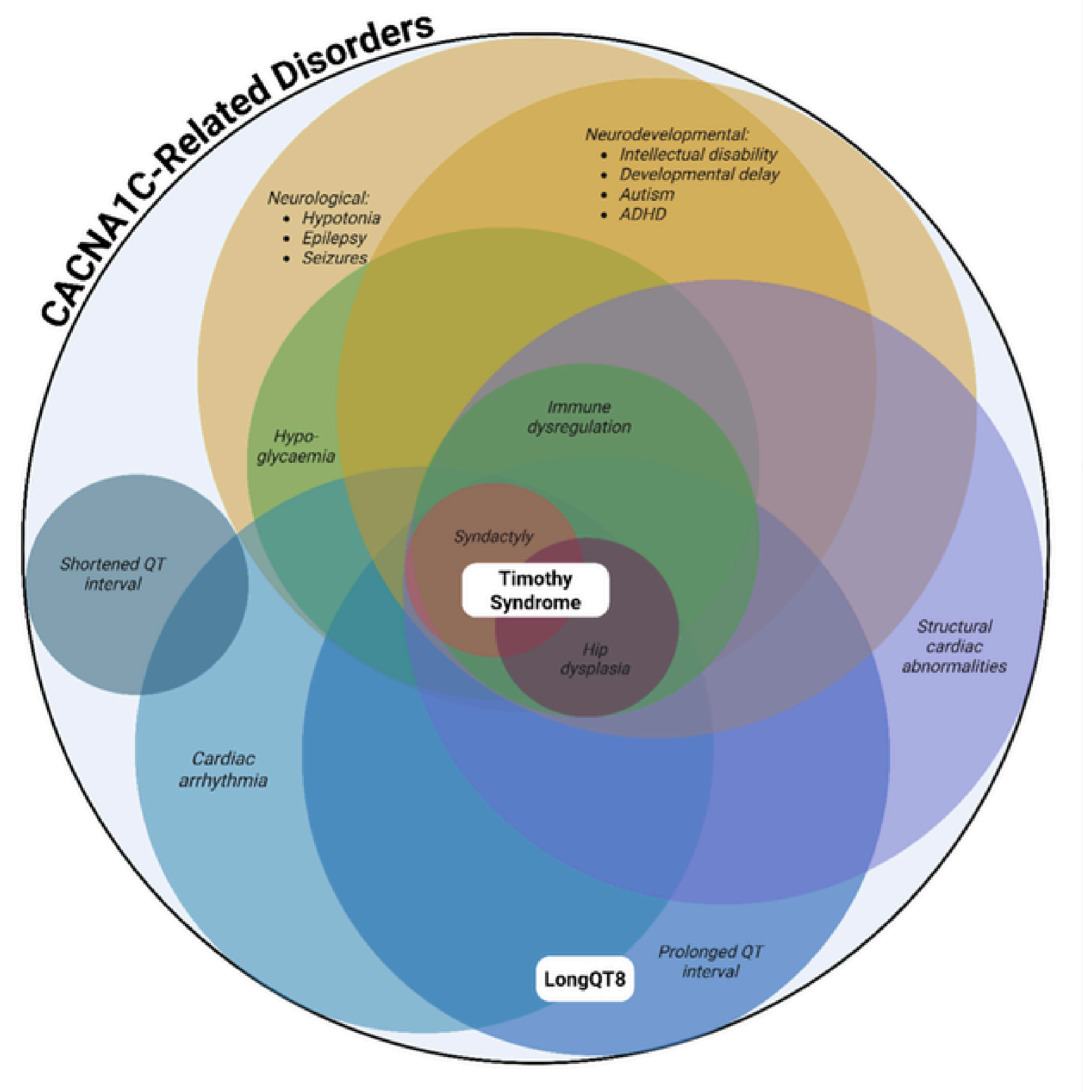
WHAT ARE CACNA1C-RELATED DISORDERS?



SCAN ME

Scan the QR code to read our preprint, or visit DOI: 10.21203/rs.3.rs-805853-6/v1

The new umbrella term, CACNA1C-Related Disorders, encompasses all individuals with pathogenic rare variants in CACNA1C, beyond those with Timothy Syndrome Type 1 or Timothy Syndrome Type 2 phenotypes, as shown above. This serves to bring those with Long QT Type 8 into the wider community, and to include those individuals who previously did not have a formal diagnosis, allowing them improved access to medical care and therapies.

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CACNA1C RESEARCH TALKS - WEDNESDAY 22ND JULY

Session A

9.30am - 9.45am	Opening welcome talk	Prof. Jeremy Hall
9.45am - 10.15am	Science talk	Dr. Rebecca Levy
10.15am - 10.45am	Science talk	Dr. Patapia Zafeiriou
10.45am - 11.10am	Break	
11.10am - 11.15am	Group photo	

Session B

11.15am - 11.45am	Science talk	Dr. Nadine Ortner
11.45am - 12.15pm	Science talk	Prof. Ivy Dick
12.15pm - 14.15pm	Lunch and posters	

Session C

14.15pm - 14.45pm	Science talk	Dr. Antoine Delinière
14.45pm - 15.05pm	TSA research update	Dr. Jack Underwood
15.05pm - 15.35pm	Break	
15.35pm - 15.45pm		Dr. Gemma Wilkinson
15.45pm - 15.55pm		Dr. Demetrio Julián Santiago Castilllo
15.55pm - 16.05pm	Short flash talks	Dr. Adam Bates
16.05pm - 16.15pm		Sophie Smith
16.15pm - 16.25pm		Dr. Emily Cottrell
16.25pm - 16.30pm	Day 1 closing remarks	Sophie Muir

Socialising and Dinner

16.30pm - 17.00pm	Socialising in the conference building
18.00pm onwards	Dinner at Premier Inn (Queen Street) restaurant

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CACNA1C FAMILY/RESEARCH PRIORITIES - THURSDAY 23RD JULY

	Families	Researchers/Clinicians
Parallel priorities session (Part 1)		
9.00am - 10.00am	During the first half of the parallel session, families will be divided into groups with post-it notes, A3 paper, and time to discuss together what is important to them. **Chaired by Lauren Roberts, CEO of RareMinds.**	During this parallel session, researchers and clinicians will have the opportunity to discuss recent and future advances in the field, including TSA's recent language consensus paper. **Chaired by Dr Jack Underwood**
10.00am - 10.30am	Break	
Parallel priorities session (Part 2)		
10.30am - 12.00pm	During the second half of the parallel session, families will, as a group, discuss the common themes identified. Using a microphone that will be passed around, we will encourage families to share their insights on specific topics. These suggestions will be noted and used by TSA to form our patient-prioritised support and research agenda. **Chaired by Lauren Roberts, CEO of RareMinds.**	During the second half of the parallel session, researchers and clinicians will have the opportunity to discuss their own research collaboration opportunities. **Chaired by Dr Jack Underwood**
12.00pm - 14.00pm	Lunch	
Speed dating		
14.00pm - 15.00pm	Around the room, there will be tables of healthcare professionals ranging from genetic counsellors (AWMGS), genetic educators (Wales Gene Park), rare disease counsellors (RareMinds), Cardiff University Biobank and more! You will have 5-10 minutes to go around each table and ask as many questions as you want!	
Pitch a priority		
15.00pm - 16.00pm	To round off the second day, we will have a session inspired by Dragon's Den and Shark Tank, where researchers and clinicians will have pre-submitted questions that they want to ask the families. Each pitch invites feedback, challenge, and discussion - putting families in the role of experts to shape research priorities, highlight what matters most in day-to-day life, and guide what research should happen next.	
16.00pm - 17.00pm	**Closing of the conference and thank you!**	

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TRANSLATION

At the Connect CACNA1C Global Network Conference 2026, we are committed to making the experience as accessible and inclusive as possible for everyone attending.

Professional in-person translators will be available throughout the event for families who have let us know in advance that they require this support.

Following the conference, recordings of the talks will be made available on our website (https://timothysyndrome.org/) and YouTube channel (https://www.youtube.com/@timothysyndromealliance) in a range of languages, ensuring that attendees can revisit the content at their own pace, and that those who were unable to join us in person can still benefit from the discussions.

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WHY YOUR SUPPORT MATTERS

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.

Our last in-person meeting in 2019!

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 Dr. 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: your honesty and courage guide the priorities that shape research and care for the future.

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WHY YOUR SUPPORT MATTERS

We are thankful to the Neuroscience and Mental Health Innovation Institute at Cardiff University for hosting this year's event. Thank you also to Dr Amy Houseman (Research and Public Engagement Officer) and Amanda Sanders (Mission Support Officer), both funded by CZI, for their planning and coordination.

This is our seventh year as a charity, and each year our global community grows stronger. Since January, we've welcomed families at a pace of around 3 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 complete 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.

To everyone living with a CACNA1C-Related Disorder, including Timothy Syndrome and Long QT Type 8, 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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