



## Extra! Extra! Read All About It!

So much to shout about that we needed an extra edition!

It's a heatwave in the UK right now, and TSA and the CACNA1C community are turning up the heat too. Whilst the regular quarterly newsletter focused on collaboration, this extra edition will share the latest research, news and updates on our community, team and website, and on our recent fundraising and where those funds are spent.

So grab your favourite drink (hot or cold), settle back and discover even more...

## Noticed Anything Different?

With input from our 227-member Patient Advisory Board, we've updated our logo to better reflect who we are and the families we support. As a charity that advocates for

everyone affected by CACNA1C-related disorders, not just Timothy Syndrome, it was important to us that our new look truly represents our whole community.

### **Timothy Syndrome Alliance: A Home for All with Rare CACNA1C Variants.**

A huge thank you to Vince for his pro-bono creation.

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## **Your Stories...**

*"I want to be loud, so that when other parents get this diagnosis, they do not have to feel alone for even one second."* Susan

Meet Patrick and Yammy - a beautiful partnership. Please read [Susan's Story](#), an honest piece expressing the shock, grief and hope that we all feel when we get 'that' diagnosis. A big thank you to Susan for sharing her and Patrick's personal story.

Hearing your lived experiences brings reassurance, calm, support but above all the realisation that you are not alone. Your stories are so very valuable to so many people in so many ways. If you want to tell yours, please [get in touch](#) - we'd love to hear from you.



## What's New?

### We're Hiring...

a Research and Public Engagement Officer: a newly created role enabled by our RareAsOne multi-year funding from the Chan Zuckerberg Initiative (CZI).

This role will develop and oversee communications and engagement activities focused on Timothy Syndrome and CACNA1C-related Disorders. Key objectives include:

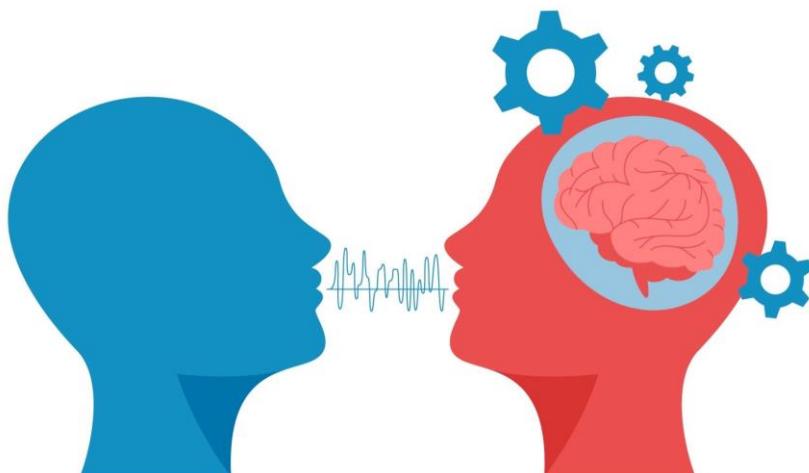
- liaising with the international research community to co-ordinate global research efforts, share knowledge, drive collaboration and accelerate patient-prioritised research.
- engaging with key international stakeholders such as clinicians, researchers, third sector organisations, the CACNA1C community and wider public, to raise the profile of Timothy Syndrome and CACNA1C-related Disorders.
- being responsible for the review of submissions to the imminent TSA's CACNA1C Research Portal.

### We've Hired...

a Mission Support Officer, a behind-the-scenes role, part of the engine room of our charity.

Introducing Amanda Sanders, someone you may come across at some point. As our Mission Support, she is responsible for providing administrative support, dealing with everything from inputting data, maintaining contacts, creating these quarterly newsletters, helping organise conferences and anything in between. Amanda can be contacted on [admin@timothysyndrome.org](mailto:admin@timothysyndrome.org)

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### We are close to achieving...

our fundraising target for our [Speech and Language research study](#), thanks to your incredible support, alongside the generosity of the Heax team and the Sahara Desert Trek fundraiser, Souter Charitable Trust, Renishaw Charities Committee, the Speech, Language

and Hearing Foundation and The Stanley Grundy Foundation. 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.

If you haven't supported yet and would like to help nail this final £2083 (\$2812) and hit the £16,000 target, you'll find it helpful to know supporters using JustGiving can choose from five currencies: British pounds (GBP), US dollars (USD), euros (EUR), Canadian dollars (CAD), and Australian dollars (AUD).

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## We are working with...

**Chai Ann Ng (Andy), PhD, Senior Staff Scientist**, Victor Chang Cardiac Research Institute - read about [Functional assays and drug screening for ion channels](#)

**Daniel Haynes, PhD student** funded by the Hodge Foundation, NMHII, Cardiff University read about [Sex differences in the causes and symptoms of psychiatric illness](#)

**Richard Dolder, Graduate Student in Human Genetics**, Glazer Laboratory, Vanderbilt University Medical Centre - read about [Detecting differences in calcium influx](#)

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## We've added...

to our support webinars. In partnership with Rareminds, these webinars are available on a private channel with open access for our community to fit around caring commitments and time zones. They are all complete with 17 language transcripts for international accessibility. In addition to *Dealing with Diagnosis*, which has received 201 views in the 10 months since launch, we have *Managing Uncertainty* and *You & Your Mental Wellbeing*. If you do not have access to our Facebook Support Group and are impacted by CACNA1C please contact [Mission Support](#) for access. **and...**

to our website with new pages added to [Our Community](#) section:

- [Newsletters](#) : sign up here to receive updates or find back issues and catch up on all the news, research and stories that affect our community.
- [Advocacy](#) : where you'll find reports from other organisations that are speaking up for the rare disease communities. Read the report from the Real-World Evidence (RWE) for Rare Disease Roundtable highlighting the challenges rare disease charities like us face in accessing data and generating RWE, and emphasising the importance of collaboration.
- And an up-to-date [CACNA1C map](#) showing the extent of our community.

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## We are growing...

The [\*\*CACNA1C Community Registry\*\*](#) is unique: it combines the precision of genetic data with **deep, patient-reported clinical detail**, enabling a richer understanding of rare CACNA1C-related disorders. This registry now exceeds 100 participants.

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### Past Issues



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## TSA Report Card 2024

We're proud to share our [Annual Report for the 2024 financial year](#). This report is not just a regulatory requirement - it's a transparent account of how funds were raised and, importantly, how they were spent to advance our mission. We're committed to ensuring every penny goes where it's needed most, and we invite you to read the report to see the difference our collective efforts have made, and how your support – financial or otherwise – continues to help us grow, advocate and make meaningful change. The report has been submitted to the Charity Commission and is available on both their website and ours.

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### P.S

Just to clarify from our earlier newsletter, our recent trip to Las Vegas for the Chan Zuckerberg Initiative (CZI) gathering was completely funded by CZI. We're so grateful for their support, and we want to reassure our community that no donated charity funds were used for travel or expenses.

### P.P.S

Don't forget to [save your seat](#) at our online Conference on Saturday 20th September 2025 from 3pm-7.30pm (BST) making it accessible for all those at work Mon-Fri.

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## How you can help

TSA may be small but we are mighty - but it can't happen without your support. TSA is run by an amazing team of parents and volunteers. Which makes your donations so important to cover our essential core costs that keep our charity running effectively. These costs **Past Issues** allow us to plan, grow and above all maximise our impact.

And there are so many different ways you can donate...



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 Donor-Advised Fund (DAF) or intermediary organisations like CAF America and [GlobalGiving](#)?

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

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[DONATE](#)



[DONATE](#)



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[Benevity](#) makes it easy for you to support our charity through your workplace—your donation can go further with employer matching, and every contribution helps us make a bigger impact.



## Spread the Word...

We know not everyone **can't** give financially, but sharing our work on social media spreads the word, reaching those who can support—and those who are impacted by CACNA1C and need our support. Every post, like, and share makes a difference - so hit that button! 

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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 



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Our mailing address is:  
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