TIMOTHY SYNDROME ALLIANCE (TSA)

ANNUAL REPORT

2025

For financial period

1 DECEMBER 2024 TO 30 NOVEMBER 2025

Timothy Syndrome Alliance
CACNA1C

Charity Registration Number: 1185523

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Message from the Chair

2025 has been a year of consolidation, growth and increasing responsibility for Timothy Syndrome Alliance (TSA). Behind every statistic in this report is a family searching for answers. As understanding of CACNA1C-Related Disorders continue to evolve, so too does the complexity of the environment in which we operate. What was once viewed as a narrowly defined condition is now recognised as a broader group of related disorders. This brings new opportunities for research and care, but also a responsibility to ensure that information, support, and clinical guidance evolve alongside the science.

TSA delivers its work through six strategic priorities, as outlined in our strategic framework and previous annual reporting. During 2025 we strengthened communications reach, expanded community infrastructure, deepened clinical and research collaboration, and continued to build the systems required to support responsible long-term progress.
Our global registry grew, our community support network expanded, and we secured significant pro bono scientific collaboration to strengthen the evidence base. We continued to prioritise trusted information and careful language, recognising that clarity reduces harm in rare and poorly understood conditions.

Importantly, we have done this as a predominantly volunteer-led organisation. Trustees and volunteers contribute substantial time and expertise, and I remain deeply grateful for their professionalism and commitment. Alongside this, we reached an important milestone in our organisational development. With grant support from the Chan Zuckerberg Initiative, we began to invest in core capacity, enabling us to appoint our first paid staff roles. We are deeply grateful for this support, which strengthens the infrastructure required to deliver our work responsibly and sustainably.

The past year has also reinforced important lessons. Families often reach us at moments of uncertainty and vulnerability. Clinical pathways remain variable. Research interest is increasing, but must be grounded in ethical engagement and lived experience. Infrastructure, though less visible than programmes, remains the foundation upon which progress depends.

Financially, the charity remains stable, with income of £205,304. We continue to rely primarily on restricted funding and recognise the importance of increasing unrestricted income to strengthen resilience. Trustees monitor risk carefully and remain committed to proportionate, sustainable growth.

None of this work is undertaken in isolation. We are grateful to our Scientific Advisory Board, Patient Advisory Board, Research and Public Engagement Officer, Mission Support Officer, funders, research partners, and the global community of families who place their trust in us. Their willingness to collaborate makes responsible progress possible.

Looking ahead to 2026, our priorities are clear: dissemination of emerging consensus guidance, analysis and publication of registry data, continued strengthening of community support, and preparation for our international in-person conference. We remain focused on building understanding, improving care, and enabling research that is rigorous, ethical and community-informed.

TSA was established in response to uncertainty and isolation. Six years on, we are seeing the foundations of a more coordinated and informed field emerging. While significant challenges remain, there is growing evidence that shared understanding can reduce harm and accelerate progress.

On behalf of the Trustees, thank you to everyone who contributes to this work.
Sophie (& mum to Calvin)
Chair of the Board of Trustees

IMPACT AT A GLANCE

250+ families
supported worldwide

39 countries
represented

107
participants in the
global patient registry

19
languages used to
improve accessibility

Over 3,000 people accessed
trusted information

160 direct enquiries from
families and clinicians

£16,000 Speech & Language
research funding raised

£38,043 pro bono
RWE collaboration

37 counselling
sessions provided

66,000 awareness
views online

98.3% of expenditure
on charitable activities

First international consensus
guidance submitted

INTRODUCTION AND OVERVIEW

The Trustees are pleased to present the Annual Report and Financial Statements of Timothy Syndrome Alliance (TSA) for the year ended 30 November 2025.

During 2025, TSA continued to operate as a small, volunteer-led international charity with a singular focus on improving understanding, care, and research for individuals impacted by CACNA1C-Related Disorders, including Timothy Syndrome and Long QT type 8. Despite limited capacity and modest financial resources, TSA delivered significant progress during the year. The charity strengthened its role as a trusted source of information and connection for families, clinicians, and researchers worldwide.

With input from our 227-member Patient Advisory Board, we updated our logo this year 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, we felt it was important that our new look truly represents our whole community.

This new design was created as a pro bono project.

WHO WE ARE AND WHY WE EXIST

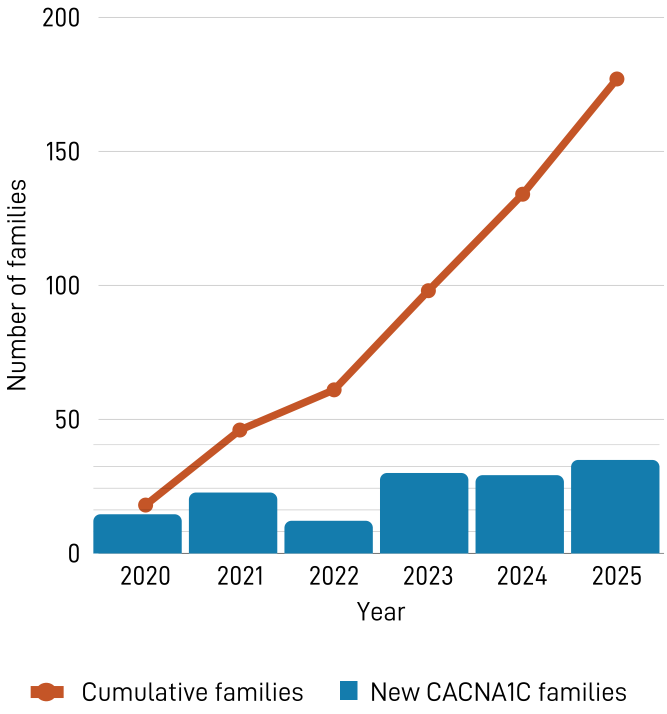
Our Cause

Timothy Syndrome Alliance exists to support individuals and families impacted by CACNA1C-Related Disorders, which include Timothy Syndrome and Long QT type 8, a group of life-altering and life-shortening ultra-rare genetic conditions associated with complex, multi-system impacts including cardiac, neurological, developmental and psychiatric features.

In 2025, TSA continued to provide global support to a growing and geographically dispersed community. The charity's patient registry increased to 107 participants during the year, and the wider community now includes over 250 families worldwide. Trustees note that this growth reflects increased visibility, trust, and unmet need rather than increased prevalence.

Families consistently describe the period after diagnosis as overwhelming. Many immediately begin searching for information, often finding little reliable guidance and feeling deeply isolated.

CACNA1C Community Growth (2020-2025)



The graph shows the annual number of newly connected CACNA1C families and individuals to TSA, with each family counted once, regardless of affected members. It doesn't reflect diagnostic incidence or the overall count of individuals with CACNA1C gene changes.

One parent described the experience starkly:

> "When you type in CACNA1C you immediately see Timothy Syndrome and a life expectancy under the age my child was."

For many families, the difference between distress and stability is not simply receiving a diagnosis, but finding accurate information, informed community, and credible guidance. Trustees consider this point of connection to be a core public benefit delivered by the charity.

Beyond providing support, the charity exists to help change the future of these conditions. TSA combines the power of lived experience with scientific expertise. Led by impacted families and guided by world-class researchers, the organisation focuses on what matters most: earlier diagnosis, meaningful real-world data, and collaborative research that reflects the priorities of those living with CACNA1C-Related Disorders. Through its patient registry, international research collaborations, and growing global community, the charity is helping to build the knowledge base needed to improve understanding, influence care, and accelerate progress toward effective treatments. Trustees believe that when families, clinicians and researchers work together in this way, rare conditions that were once poorly understood can become the focus of meaningful scientific progress.

250+ families worldwide

39 countries

107 patient registry participants

FAMILY STORIES

Families describe the moment of diagnosis as one of the most difficult periods they face. The reflections below illustrate the importance of connection and shared understanding.

“After our diagnosis, we felt overwhelmed at first,” one mother recalls. “It took about three months to find our new family, and we are so grateful — not feeling alone on this journey means everything.” She adds that “having that connection to knowledge is priceless,” and remembers how “Sophie's words of encouragement... truly helped me in my darkest moments.”

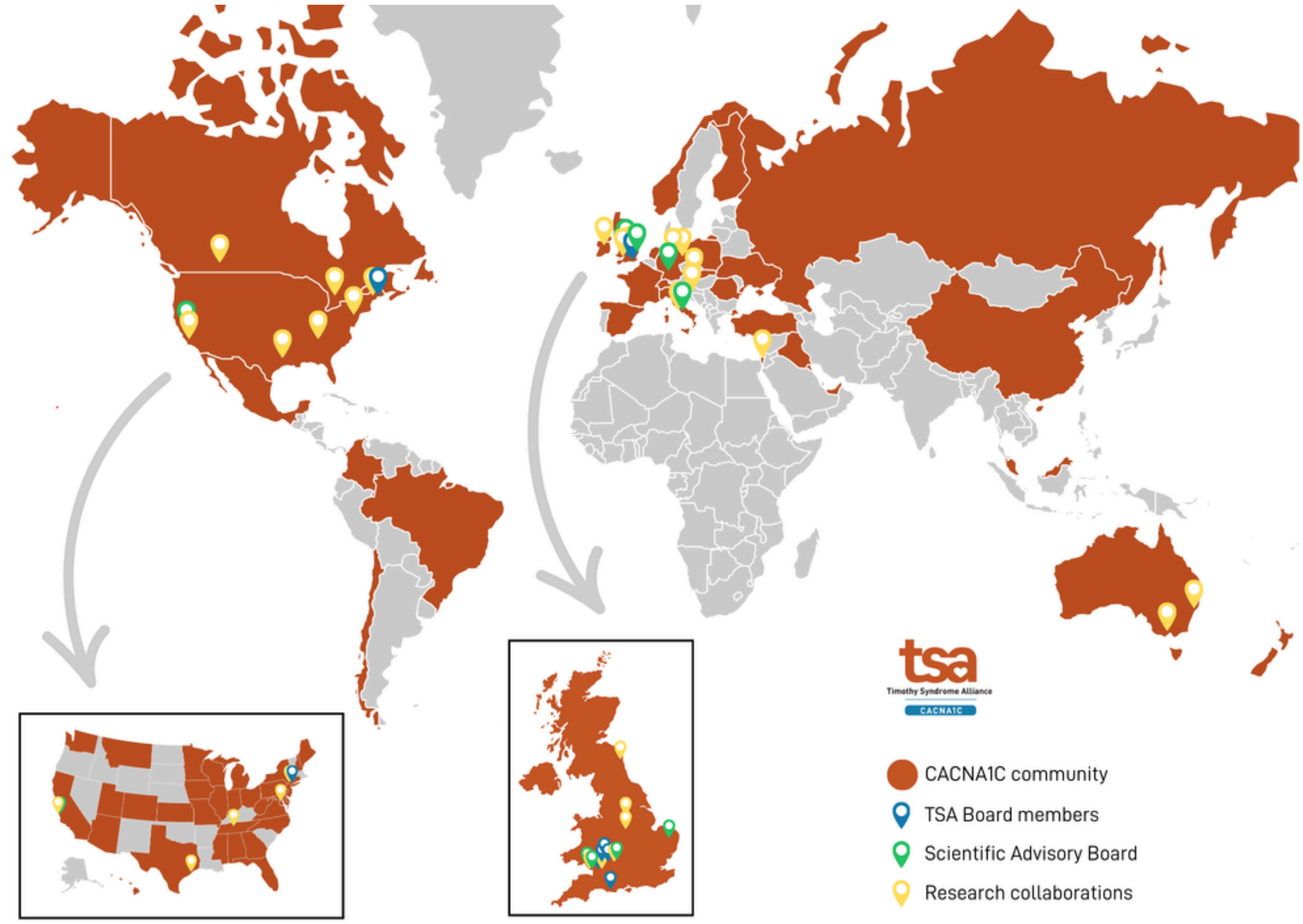
“It took me about six months to find the community, and I was relieved to see that there were other children like mine,” one mother recalls (translated from French). Thanks to a tip from her son's physiotherapist, she discovered the group online. “Understanding the condition a little better through the community helps me,” she adds, especially since her son is the first known case in her country.

“We couldn't believe it — after 23 years we found an entire group within a day of our diagnosis,” one family says. “It was incredible to discover other families experiencing the same thing. Now there is always someone to share a question with. It's invaluable to know that a dedicated team is moving this cause forward for a better future for our children,” they add.

“I remember feeling very lost and alone, wondering if anyone else was going through this,” one parent says. “Then I found this community and it was a shock — there were so many others sharing our story. This community has given our family hope, showing that despite the challenges of Timothy Syndrome, our child can still live a meaningful life.”

“Within 15 minutes of getting off the phone with genetics, I found the TSA Facebook group,” recalls a mother. “Finding this has been amazing for our family. We are so thankful for the connections we've made, the wisdom we've learned, and for not feeling like we're alone as we figure this all out.”

“About three months after my daughter's diagnosis, I learned about the Alliance,” says a mother (translated from Italian). “I had felt so afraid and lost. Finding the community was like breathing again — it gave us answers, hope, and reassurance.”



Why This Matters

And why it should matter to You

The CACNA1C gene plays a key role in how our brains and hearts function.

Although rare changes in the CACNA1C gene cause severe, life-limiting disorders, the gene itself also plays an important role in common mental health conditions like depression, bipolar disorder and schizophrenia.

Depression affects over **300 million** people worldwide.
Bipolar disorder impacts **1–2 in every 100** people.
Schizophrenia touches **24 million** lives.

These are not rare experiences.

By understanding these rare, high-impact cases, we gain crucial insight into the biology behind more common conditions. This work doesn't just help a small community - it could open the door to breakthroughs in mental health and precision medicine that would benefit millions.

VISION, MISSION AND VALUES

VISION

Our vision is a world where shared knowledge and understanding lead to a cure for everyone with a CACNA1C rare variant.

MISSION

Our mission is to improve diagnosis, treatment and care of individuals worldwide with CACNA1C-Related Disorders including Timothy Syndrome, and to support the families and carers of those diagnosed.

Our core values and beliefs shape how we deliver our mission in practice. In 2025, these values were expressed through the following commitments:

DETERMINED

EMPOWERING

SUPPORTIVE

COMMUNITY-LED

Trustees review these statements regularly to ensure they remain aligned with the evolving evidence base, the needs of our global community, and the charity's objects.

Values in Practice

In 2025, TSA's values were demonstrated through operational and strategic decision-making rather than aspirational language. Trustees undertook a detailed review of the international consensus preprint to ensure terminology reflected both clinical accuracy and lived experience, recognising that language can directly influence care, expectations and psychological wellbeing.

The registry continued to be refined with family-informed input to ensure ethical participation and meaningful engagement. Peer support spaces were actively moderated to maintain psychological safety, and the charity continued to avoid sensationalised awareness activity in favour of proportionate, evidence-based communication.

As one community member reflected:

> *"Knowing this is not my fault. I did the very best I could with what I had available."*

Trustees regard this reframing from blame to understanding as a significant outcome of values-led practice and a clear example of public benefit.

OUR STRATEGIC MODEL
The Problem We Are Solving

Rare diseases affect over 300 million people worldwide, yet each condition often impacts only a small number of individuals. CACNA1C-Related Disorders, including Timothy Syndrome, are ultra-rare.

Because these conditions are so rare, many healthcare professionals are unfamiliar with them. As a result, people are often treated only for individual symptoms rather than receiving coordinated care based on an accurate diagnosis.

Rarity makes diagnosis more challenging. Typically it takes around 4 to 5 years for a rare disease to be diagnosed, though it can often take much longer; 40% of rare disease patients are initially misdiagnosed.

9.7 years on average to receive a CACNA1C diagnosis

Rarity means that we are among the 95% of rare diseases without an approved treatment or cure. Research knowledge is often fragmented with information scattered across different disciplines meaning rare diseases remain poorly characterised at the genetic or molecular level. Drug companies may not see a profitable market, leading to less investment in developing treatments.

Raising awareness to gain widespread attention and support is challenging due to limited patient populations, lack of steady funding, low recognition within the medical community, being overshadowed by more prevalent conditions, the complexity of the disease, and a general lack of urgency.

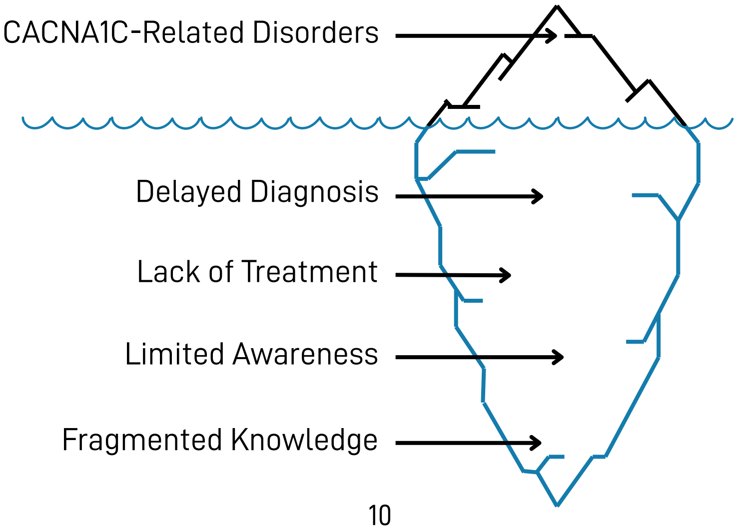
Evidence gathered during 2025 reinforced a consistent pattern across families globally. The period following diagnosis is frequently characterised by fear, shock, anxiety, and confusion. Interviews conducted through collaborative projects, including the Illumina Variant of Uncertain Significance work, highlighted the difficulty families face when clinical teams cannot yet provide holistic advice and when publicly available information is sparse or misleading.

Families reported:

> “Google did not hold that much info, neither did the doctors.”

> “I spent a whole day after the diagnosis crying.”

Trustees recognise that these experiences are not isolated but systemic within ultra-rare disease contexts, where clinical pathways, research evidence and support infrastructure are still emerging.



Our Solution

Families impacted by CACNA1C-Related Disorders consistently describe the period following diagnosis as one of profound uncertainty. TSA's Theory of Change is designed to address this directly.

Because CACNA1C-Related Disorders are rare, poorly understood, and often met with delay, misinformation and isolation, we work globally to help affected individuals and families, clinicians and researchers build understanding, improve care and generate evidence, so that people receive the right support at the point of diagnosis and throughout life, and research and clinical progress can advance responsibly when it matters most.

This model recognises that meaningful research participation depends on informed and supported families, and that improvements in care require shared language, lived experience and credible data.

Current clinical care is hindered by a lack of standardised protocols, insufficient research, and limited access to comprehensive genetic data. This unmet need provides a unique opportunity to create a knowledge base that consolidates data and drives innovation in the diagnosis and treatment of CACNA1C-Related Disorders.

By building strong data foundations, fostering collaborative research, engaging our activated patient community, and leveraging regulatory incentives to reduce development risk, we position CACNA1C as a strategically aligned and investment-ready target.

Theory of Change

How we will do it

Expand communications to raise awareness and reach new audiences

Build clinical and research networks to improve diagnosis, treatment, and care

Strengthen community support through global connection and shared resources

Provide trusted information that empowers families and professionals

Advance research by supporting funding, collaboration, and participation

Influence policy to improve systems and outcomes for our community

2025 IN REVIEW - STRATEGIC DELIVERY

In 2025, TSA continued to deliver its charitable objectives through its established strategic priorities. Trustees consider this structure essential for transparency, proportional reporting, and alignment with both current Charities SORP and anticipated SORP 2026 expectations regarding outcomes, public benefit, and organisational learning.

Our activities are reported below against each priority, reflecting how change occurs in practice within an ultra-rare, globally dispersed community.

Expand communications to raise awareness and reach new audiences

The short film *Connections* produced with Cardiff University, received the People's Choice Award at the *Smiley Charity Film Awards 2025* and was selected for international screening at *Rare by Design's The Film Festival!*, increasing awareness while preserving dignity and authenticity in representation.

SMILEY CHARITY FILM AWARDS

PROUDLY
PRESENTED TO:

TIMOTHY SYNDROME
ALLIANCE - TSA

WINNER
2025

PEOPLE'S CHOICE
LONGFORM
UNDER £500,000

Nicolas Loufrani

NICOLAS LOUFRANI
FOUNDER SMILEY MOVEMENT
CEO THE SMILEY COMPANY

PRESENTED BY
SMILEY

POWERED BY
SMILEY MOVEMENT

During the year, communications remained carefully managed and proportionate, delivered in-house without a dedicated marketing team and alongside programme, governance, and research activities. The focus remained on accuracy, safeguarding, and trusted visibility rather than scale.

Between 1 December 2024 and 30 November 2025:

- Social media channels Facebook and Instagram were viewed over 66,000 times
- With engagement exceeding 1,500 meaningful interactions
- Profiles were visited over 4,300 times
- And organic following grew to just over 1,500.
- Over 3,000 individuals accessed trusted information via the TSA website
- Resulting in more than 160 direct enquiries received through the website from families and professionals across multiple countries.
- On YouTube, our films and conference recordings (excluding shorts) received 1,928 views
- Averaging an audience of 93 viewers per month
- And a watch time of 96.4 hours.
- All our conference and Mind the Gap webinars are available in a minimum of 17 human-checked language transcripts.

These figures show that families and professionals are actively seeking reliable information in a field where accurate resources remain scarce.

YouTube Audience by watch behaviour

Monthly audience

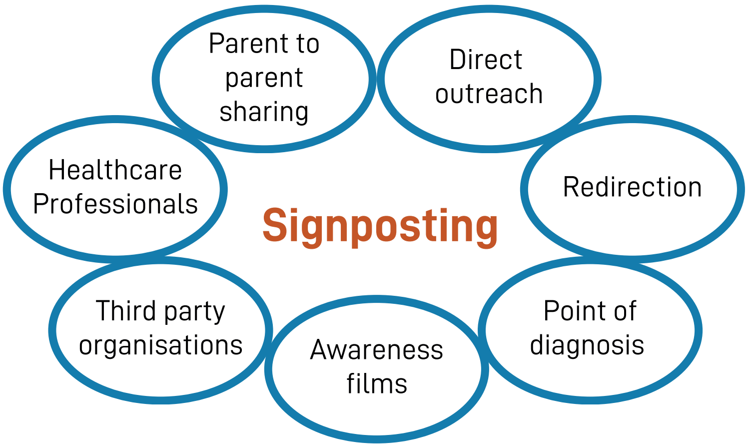
● **New viewers** 77.4%

● **Casual viewers** 19.4%

● **Regular viewers** 3.2%

Expanding communications to raise awareness and reach new audiences also balances the need for us to be signposting to new and existing families not yet aware of, or in contact with, TSA. Signposting pathways are wide-ranging.

Signposting pathways



Community feedback indicates that families most commonly find TSA through Facebook searches, Google searches, clinician recommendation, peer referral, and rare disease media content. This reinforces the importance of maintaining accurate and moderated digital visibility.

One parent reflected:

> “The community found me.”

Trustees consider this a priority to deliver clear public benefit by reducing isolation, improving access to accurate information, and enabling earlier connection to appropriate support.

Common online search pathways

“A film called Rare Strikes Back helped us find the community.”
I saw it on TikTok, then searched for the group on Facebook - and realised we weren’t alone.

Build clinical and research networks to improve diagnosis, treatment, and care

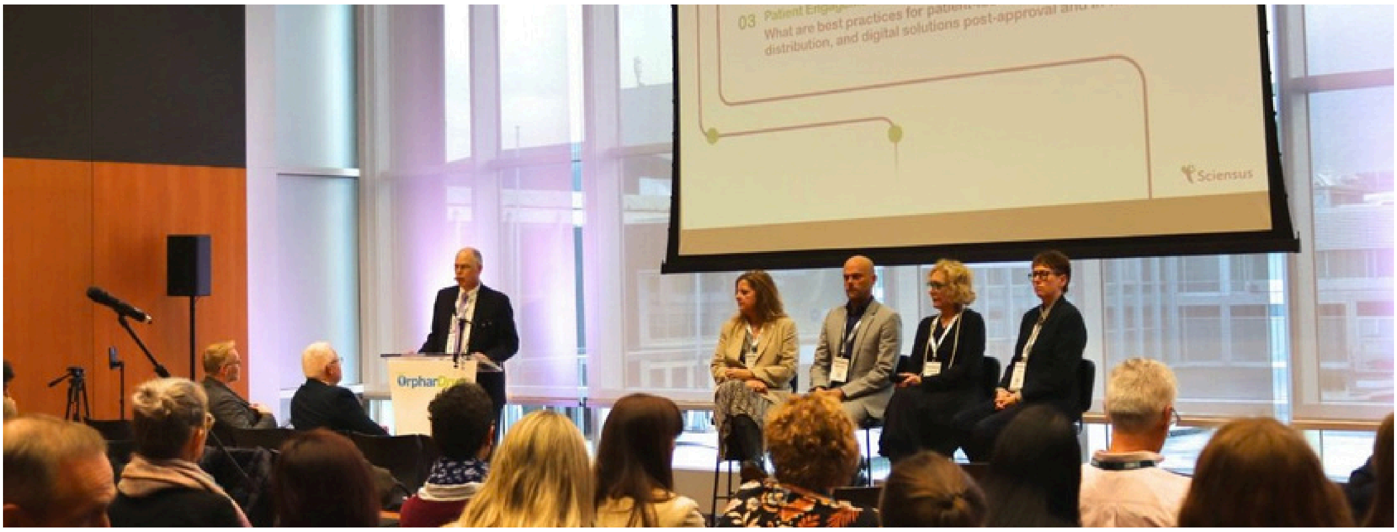
Strengthening clinical and research networks remained a major focus in 2025, particularly in light of evolving understanding of CACNA1C-Related Disorders as a broader and more heterogeneous spectrum than previously described.

Trustees and advisors supported the completion and submission of the *Timothy Syndrome and CACNA1C-Related Disorder: First International Language and Management Guidelines Consensus Statement*, currently under review with the European Journal of Human Genetics. This work is expected to improve consistency of language, clinical understanding, and care approaches internationally.

Every month we joined fellow Rare As One Network members for dedicated channelopathy, science and network calls to connect, collaborate, share research priorities, discuss common challenges, and explore opportunities to accelerate progress.

Additional activities included:

- Conference presentations, including the Royal College of Psychiatrists Congress and World Orphan Drug Congress (thanks to the Sciensus team for their support in enabling this)
- Ongoing engagement with clinicians across cardiology, neurology, psychiatry and genetics
- Collaboration with research partners in the UK, Europe, the United States and Australia
- Scientific engagement through advisory discussions and knowledge exchange
- Wikipedia training in preparation for evidence-aligned public information updates



World Orphan Drug Congress (Europe) Preconference Workshop – October 27, 2025. Photo credit: Sciensus.

Our Chan Zuckerberg Initiative (CZI) Rare As One Network multi-year funding enabled the appointment of Dr Amy Houseman, our Research and Public Engagement Officer, who holds a PhD in Rare Disease Genetics from Cardiff University.
Amy coordinates global research efforts, facilitates knowledge sharing, promotes collaboration, and accelerates research that prioritises our CACNA1C patient needs. This is a key role in developing and managing our communications and engagement with UK and international stakeholders, which include researchers, clinicians, healthcare professionals, our CACNA1C community, and the general public.

Amy Houseman
Research and Public
Engagement Officer

Families consistently report that clinical teams often have limited access to condition-specific guidance at the point of diagnosis. TSA will continue to strengthen professional networks and contribute to evidence-informed language, helping to reduce harm caused by outdated or overly narrow clinical narratives.
This knowledge can be incredibly powerful, as one parent noted:

> “It helped me advocate for my daughter when I know how others are affected.”

Strengthen community support through global connection and shared resources

TSA continued to provide structured, moderated, and globally accessible community support to a dispersed ultra-rare population exceeding 250 individuals worldwide.

Evidence gathered through direct engagement and registry insights confirms that the period following diagnosis is characterised by fear, confusion, and profound isolation, often compounded by limited reliable information and misrecognition of the condition.

Key support activities in 2025 included:

- Continued free at point of access ‘Mind the Gap’ counselling provision through Rareminds
- Private (secure sharing with community) CACNA1C ‘Mind the Gap’ Support webinars ‘Dealing with Diagnosis’, ‘Managing Uncertainty’ and ‘You and your Mental Wellbeing’ have received in excess of 250 views.
- Ongoing moderation of peer support spaces to ensure psychological safety

- Expansion of multilingual accessibility, including conference recordings via website and YouTube with transcripts in 19+ languages
- Development of bereavement resources (in progress)

Community testimonies demonstrate consistent psychosocial impact:

"My anxiety minimised when I joined the group."

"Seeing older children gave me hope."

"The feeling of not being alone is a relief."

Trustees note that demand for support frequently begins immediately after diagnosis and often precedes research engagement or formal clinical pathway stabilisation.

37

'Mind the Gap' counselling sessions

provided

250+

support webinar

views

'Dealing with Diagnosis'
'Managing Uncertainty'
'You and your Mental Wellbeing'

Provide trusted information that empowers families and professionals

Providing accurate, accessible, and evidence-based information remained a core charitable activity and a key area of public benefit in 2025.

Website and digital engagement data indicate that users most frequently accessed:

- Condition-specific information pages
- Research updates
- Conference materials
- Support and connection resources, including our newsletters

Our 'Parent as Partner' approach ensures information is shared proactively rather than reactively. Our information is offered in multiple formats (written, video, verbal) and in preferred languages to accommodate diverse needs. As a community, we now span a total of 39 countries and speak 21 languages.

During 2025 eight global mailings to subscribed families, researchers and healthcare professionals achieved an average 41% open rate, demonstrating engagement levels well above typical nonprofit sector benchmarks, which generally average around 25-30% open rates. The total audience subscribed to our newsletters has also doubled in size over the 12 months.

Direct emails and messages are commonplace. When personalised and translated into the recipient's native language, this supports faster comprehension and relatability, alongside improved accessibility and inclusivity.

Engagement was reflected through attendance at the online CACNA1C conference, with 70+ attendees, both families and healthcare professionals, from 19 countries. The conference was delivered with real-time translation, enabling attendees from multiple countries to listen and read in their preferred language. This significantly improved accessibility and equitable knowledge dissemination in a rare disease context.

Families repeatedly report limited reliable online information about CACNA1C-Related Disorders, reinforcing the importance of a trusted, moderated information source.

As one parent stated:

> "Google did not hold that much info, neither did the doctors."

Trustees consider the provision of trusted information to be a core safeguarding function as well as an educational activity.

Advance research by supporting funding, collaboration, and participation

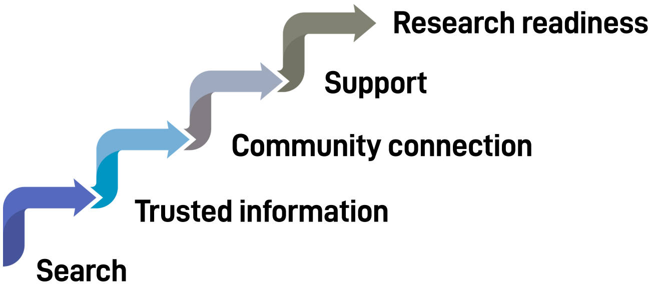
In 2025, research progress focused on building the foundations needed for future breakthroughs: strong data infrastructure, global collaboration and ethical research readiness.

Importantly, qualitative evidence shows that informed and supported families are more confident engaging with research opportunities, reinforcing TSA's model that support enables ethical participation.

One parent shared:

> "Having that connection to knowledge is priceless."

Identified steps to research readiness



Key developments included:

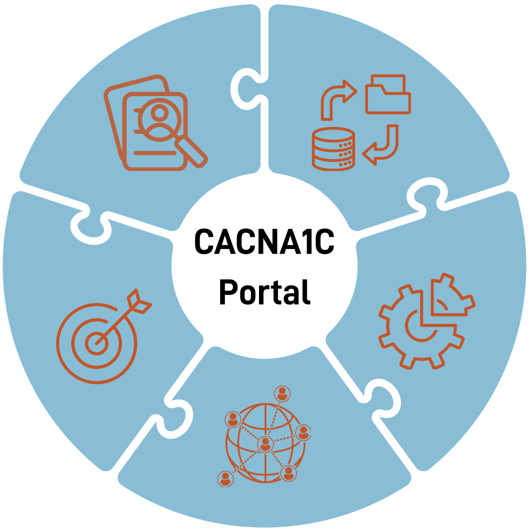
- Submission of the *Timothy Syndrome and CACNA1C-Related Disorder: First International Language and Management Guidelines Consensus Statement* to the European Journal of Human Genetics.

Following our 2023 conference, key gaps in terminology and clinical guidance for CACNA1C were identified, highlighting the need for a dedicated international consensus paper.
In response, this led to a Delphi-style consensus process meaning clinicians, scientists and researchers from institutions across the USA, UK and Europe worked together with the CACNA1C community to develop agreed terminology and minimum standards for diagnosis and care.
Community engagement through TSA networks reached more than 200 families, ensuring the final recommendations reflected both clinical expertise and lived experience.

- CACNA1C Community Registry growth to 107 participants.

The CACNA1C Community Registry is a patient registry for all CACNA1C-Related Disorders, managed by Timothy Syndrome Alliance (TSA) and powered by Pulse Infoframe. The purpose of the CACNA1C Community Registry is to obtain insights to better characterise CACNA1C-Related Disorders, including Timothy Syndrome and LongQT8, and their presentation, management and treatment. The registry was launched in June 2022 and is available for worldwide participation. This decentralised

The CACNA1C Portal



Community-specific Customisation
Tailors the portal to meet community needs

Centralised Data Repository
A comprehensive resource for CACNA1C data

Supporting Precision Medicine
Provides tools for personalised treatment strategies

Data Integration and Analysis
Enables sophisticated analyses of CACNA1C data

Global Collaboration
Facilitates worldwide collaboration among researchers

registry is not tied to a site and enables anyone, anywhere in the world with a CACNA1C-related disorder to sign up for the registry and participate from their home.
The first registry data pull is scheduled for the end of the year. The aim is to run preliminary data analysis and share our findings at the in-person conference this summer 2026.

- Building of a CACNA1C Portal.

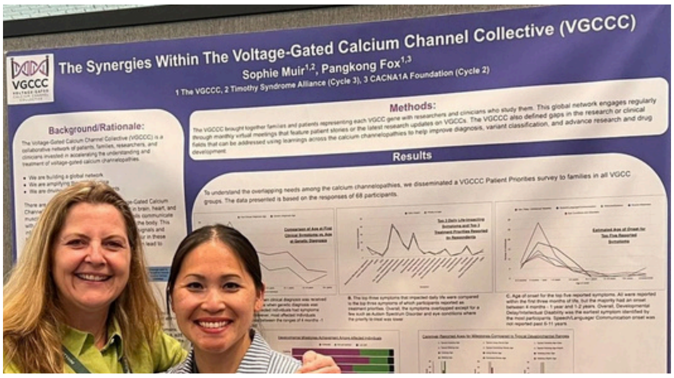
The CACNA1C Portal is a centralised, globally accessible platform that integrates clinical, genetic and molecular data to enable deeper genotype–phenotype understanding, support precision diagnosis and care, and accelerate research and therapeutic development for CACNA1C-Related Disorders.

- Research focusing on understanding the speech and language abilities of children with CACNA1C-Related Disorders.

The study will deliver inclusive, globally accessible gene-specific speech and language research. It will support parents, carers and clinicians through improved understanding of diagnosis, prognosis and treatment planning, addressing the significant impacts these difficulties can have on learning, education, social interaction, self-esteem, mental health and daily living. Findings will also inform speech and language outcome measures for future clinical trials and will be widely shared to maximise clinical impact worldwide.

The research is led by Professor Angela Morgan within the Speech and Language Group at the Murdoch Children's Research Institute (MCRI). With funding now secured, the study is expected to begin in the first quarter of 2026.

- CACNA1C Biobanking viability investigations
- Illumina Variant of Uncertain Significance qualitative work
- Ongoing research engagement in genetics, functional assays, sex differences in the causes and symptoms of psychiatric illness, calcium influx - see articles on the TSA website
- Ongoing epilepsy research as part of South West and Wales Epilepsy Research Network and GW4 epilepsy group
- Early exploration of antisense oligonucleotide (ASO) research pathways
- Continued collaboration as part of The Voltage-Gated



CACNA1C and CACNA1A representing the VGCCC

Calcium Channel Collective (VGCCC) who are dedicated to raising awareness and promoting collaboration across the calcium channelopathies. This group is composed of caregivers and individuals impacted by the channelopathies, alongside researchers and clinicians focused on the 10 calcium genes.

As members of the CZI RareAsOne Network we met everyone in person at the Science in Society meeting in Las Vegas. Both Sophie and Rebecca Levy from our Scientific Advisory Board represented TSA, and felt truly welcomed as Cycle 3 grantees. Over three days, we dived deeper into cross-disease collaborations, explored new ways to move science forward together and shared capacity-building strategies to strengthen the rare disease patient group ecosystem as a whole. The Chan Zuckerberg Initiative created a powerful space for connection, collaboration and hope.

The value of collaboration in working towards shared goals and achieving real impacts is something TSA have been advocates of from the get-go. We were therefore delighted to see Dr Jack Underwood, Chair of our Scientific Advisory Board, receive Highly Commended in the Rare Disease Research UK Early Career Researcher PPIE Awards for his work with our community. These awards highlight the real partnership between researchers, scientists, and people and families living with rare conditions. Rare Disease Research UK, working in partnership with Genetic Alliance UK, seeks to encourage Patient and Public Involvement and Engagement (PPIE), with the aim of involving people with relevant lived experience as partners and advisors in research, and engaging wider audiences with scientific progress.
Read Jack and Sophie's blog.

Jack is also the recipient of a Best Public Engagement Award from Cardiff University School of Medicine for his work with TSA.

Scientific Advisory Board

Dr. Jack Underwood (Chair)
WCAT Clinical Lecturer in Forensic Psychiatry, NMHII, Cardiff University

Dr. Rebecca Levy
Clinical Scholar, Neurology & Neurological Sciences.
Stanford Medicine

Dr. Gemma Wilkinson
Research Associate, NMHII, Cardiff University

Dr. Nicola Hall
Postdoctoral Researcher, University of Oxford

Dr. Anwar Baban
Bambino Gesù Children Hospital and Research Institute, IRCCS, Rome

Dr. Wilfried Haerty
Group Leader of Evolutionary Genomics, Earlham Institute

Prof Liz Tunbridge
Director of Translational Neuroscience, Boehringer Ingelheim

Influence policy to improve systems and outcomes for our community

Participation in influencing policy during 2025 was primarily achieved through evidence contribution, language standardisation, and responsible knowledge dissemination rather than formal lobbying, reflecting the charity's size and governance model.

Key contributions included:
- Input into international consensus language and management guidance
- Dissemination of evolving evidence across clinical and research audiences
- Preparation for updates to public knowledge platforms aligned with peer-reviewed evidence
- Representation at international conferences and rare disease forums

By improving shared language and understanding of CACNA1C-Related Disorders as a spectrum condition, TSA is contributing to longer-term system improvements in diagnosis, care coordination, and research design.

During 2025, TSA participated in a pro bono Real-World Evidence (RWE) project led by Costello Medical, valued at £38,043 in specialist consultancy support. The project brought together leading RWE, regulatory, and genomic experts to explore how real-world data could be used to advance understanding of CACNA1C-related disorders. Through a facilitated roundtable and follow-up work, the project provided the charity with access to expertise that would otherwise be far beyond the reach of a small rare disease organisation.

Discussions focused on the barriers that currently limit evidence generation in rare diseases and how better use of real-world data could support improved diagnosis, clinical awareness, and policy engagement.

For TSA, the discussions centred on a key research question: whether barriers to reporting genetic findings may be masking cases of CACNA1C-Related Disorders in the UK.

Experts provided strategic guidance on challenges such as limited coordination between stakeholders, the lack of standardised approaches for recontacting patients with variants of uncertain significance, and the need to present emerging evidence in ways that can engage policymakers and clinical leaders.

The insights and recommendations generated through this collaboration will help inform the charity's future research and advocacy work, and the meetings themselves would not have taken place without Costello Medical's generous support.

Empowering Rare Disease Charities with Real-World Evidence (RWE)

What Can You Do to Unlock Insights That Drive Transformative Care and Support?

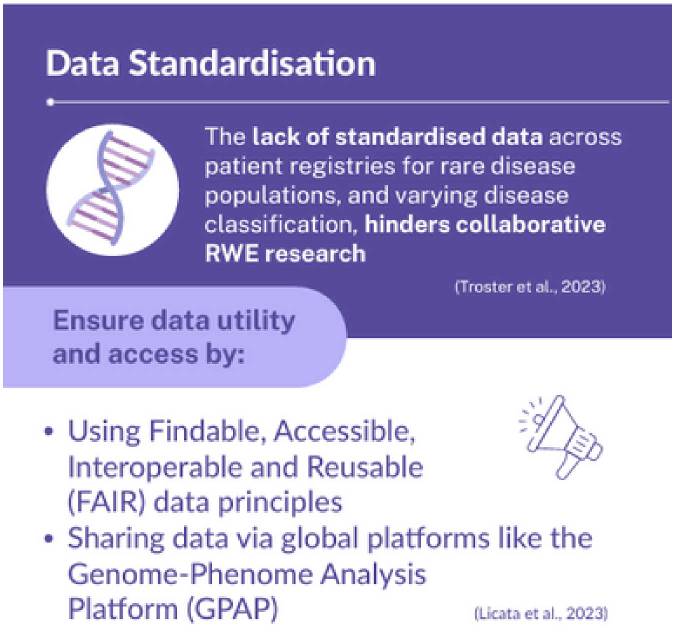
Data Gaps

Rare disease populations often lack comprehensive data on their demographic and clinical characteristics, which **limits understanding** of their treatment and management needs

(Pai et al., 2019)

Support studies of rare disease populations by:

- Sharing data
- Providing funding
- Creating an infrastructure for systematic data collection
- Advocating for rare disease research



Data Standardisation

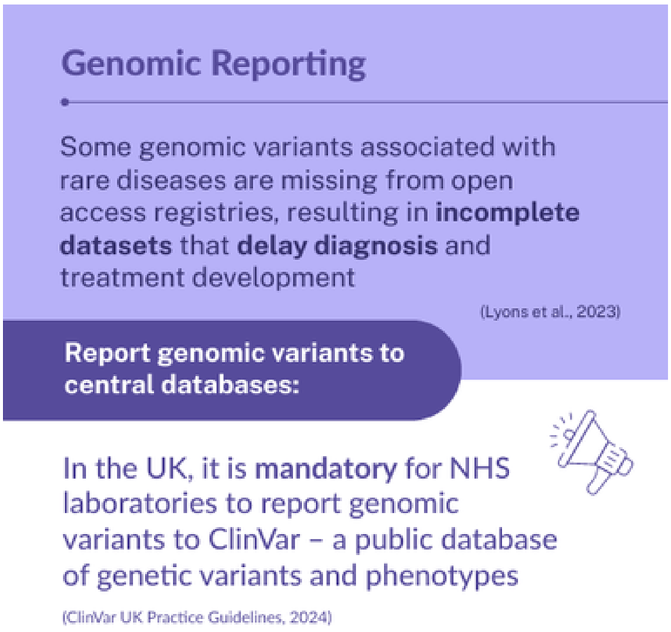
The **lack of standardised data** across patient registries for rare disease populations, and varying disease classification, **hinders collaborative RWE research**

(Troster et al., 2023)

Ensure data utility and access by:

- Using Findable, Accessible, Interoperable and Reusable (FAIR) data principles
- Sharing data via global platforms like the Genome-Phenome Analysis Platform (GPAP)

(Licata et al., 2023)



Genomic Reporting

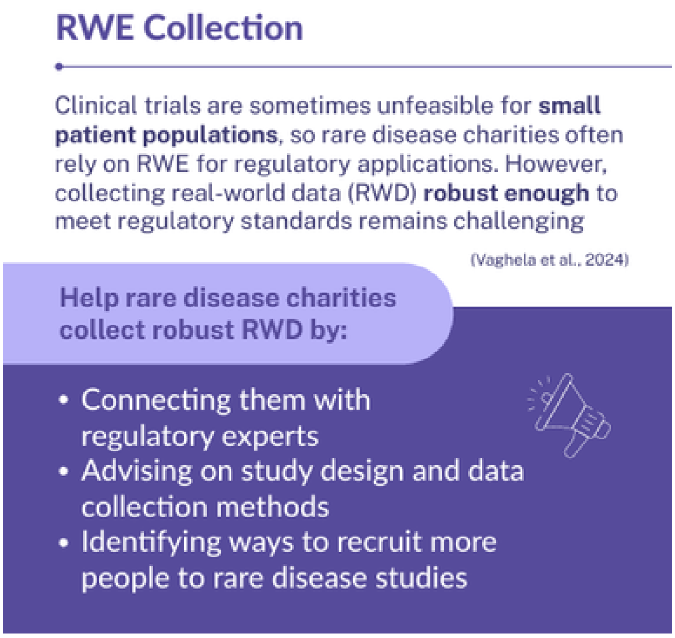
Some genomic variants associated with rare diseases are missing from open access registries, resulting in **incomplete datasets** that **delay diagnosis** and treatment development

(Lyons et al., 2023)

Report genomic variants to central databases:

In the UK, it is **mandatory** for NHS laboratories to report genomic variants to ClinVar – a public database of genetic variants and phenotypes

(ClinVar UK Practice Guidelines, 2024)



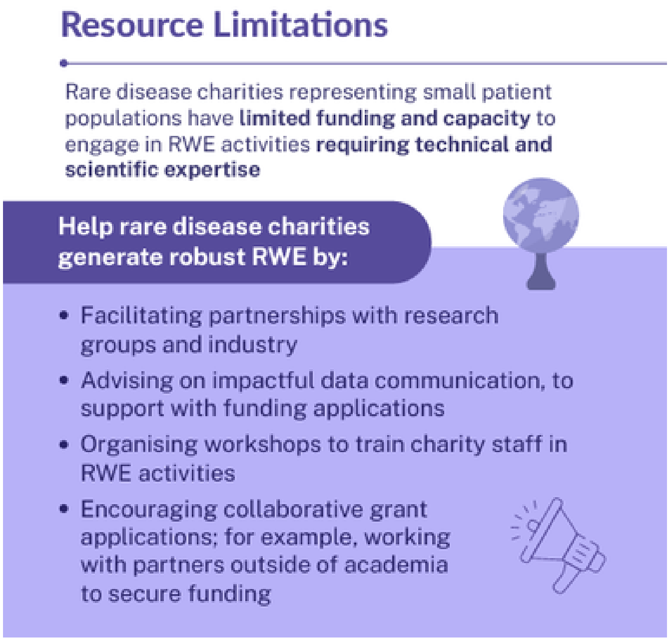
RWE Collection

Clinical trials are sometimes unfeasible for **small patient populations**, so rare disease charities often rely on RWE for regulatory applications. However, collecting real-world data (RWD) **robust enough** to meet regulatory standards remains challenging

(Vaghela et al., 2024)

Help rare disease charities collect robust RWD by:

- Connecting them with regulatory experts
- Advising on study design and data collection methods
- Identifying ways to recruit more people to rare disease studies



Resource Limitations

Rare disease charities representing small patient populations have **limited funding and capacity** to engage in RWE activities **requiring technical and scientific expertise**

Help rare disease charities generate robust RWE by:

- Facilitating partnerships with research groups and industry
- Advising on impactful data communication, to support with funding applications
- Organising workshops to train charity staff in RWE activities
- Encouraging collaborative grant applications; for example, working with partners outside of academia to secure funding

Key Policy Contributions in 2025

1. International Clinical Guidance
   Contributed to the first international consensus language and management guidance for CACNA1C-Related Disorders.
2. Improving Public Medical Information
   Prepared evidence-aligned updates to widely used medical information platforms, including Wikipedia.
3. Advancing Real-World Evidence for Rare Disease
   Contributed to expert discussions on how genomic real-world data can improve diagnosis and reporting of CACNA1C-Related Disorders.

Capacity builds capacity

Despite operating with a turnover of £205,304 and a predominantly volunteer leadership model, TSA continued to strengthen organisational infrastructure in 2025 to support sustainable and accountable growth.

Productivity and programme delivery have been greatly accelerated thanks to support from CZI Rare As One.

Amanda Sanders
Mission Support Officer

Governance remained robust throughout the year, led by a voluntary Board of nine Trustees, including four parents of children with CACNA1C-related disorders, and supported by an international Scientific Advisory Board and a global Patient Advisory Board.
During 2025, the Board was strengthened through the appointment of two additional Trustees. Three Trustees are now based in the United States, reflecting the international composition of the community we serve and strengthening the charity's ability to engage effectively with partners, researchers and supporters across multiple regions.

Operational developments included:

- Implementation of Beacon CRM
- Embedding of Xero accounting systems
- Mailchimp automation improvements
- Increase in external communications, newsletters, programmes,
- Establishment of US fiscal sponsorship via Rare Village Foundation
- Significant unpaid clinical and strategic contribution from expert collaborators
- Operational support

Trustees recognise that infrastructure investment directly underpins programme delivery, research readiness, and safeguarding, and that capacity funding remains one of the most critical yet challenging areas to secure.

Trustee Board

Sophie Muir (Chair)
Mum to Calvin with a CACNA1C-Related Disorder

Gemma Duncan
Mum to Noah with Timothy Syndrome

Galina Gardiner (Treasurer)

Susan Bresnahan
Mum to Patrick with a CACNA1C-Related Disorder

Meg McLaughlin

Achsah James

Nick Muir
Dad to Calvin with a CACNA1C-Related Disorder

Glenn Miller

Christine Ziegler

MEASURING IMPACT (QUANTITATIVE AND QUALITATIVE)

Trustees recognise that meaningful impact measurement in an ultra-rare disease context must combine quantitative indicators with qualitative evidence of lived experience change. Because the CACNA1C community is small and research is still emerging, impact cannot be measured by scale alone.

Community Reach and Connection
These indicators reflect the charity's ability to reach and connect individuals and families affected by CACNA1C-Related Disorders worldwide.

- **250+ individuals connected** through community support spaces
- **39 countries represented** within the community
- **Over 160 direct enquiries** from families and professionals via the website alone
- **107 individuals participating** in the CACNA1C Community Registry

Information Access and Engagement
These metrics show how families, clinicians and researchers access and engage with trusted information.

- **Over 3,000 people accessed trusted information** via the TSA website
- **66,000 social media content views** across digital platforms
- **Approximately 1,500 meaningful digital interactions**
- **More than 4,300 profile visits** across TSA Facebook and Instagram platforms
- **19 language transcripts produced** to support accessible conference content

Research Progress and Infrastructure
These indicators demonstrate progress in building the research foundations needed to improve understanding and care.

- **107 participants enrolled in the CACNA1C Community Registry**
- **£16,000 raised to support speech and language research**
- **Pro bono real-world evidence collaboration secured,** valued at approximately **£38,043**

Qualitative outcomes
Feedback gathered through direct engagement, counselling provision, webinars, and collaborative projects indicates important changes in experience and understanding.

Families report:

- Reduced anxiety following connection to the community
- Increased understanding of condition complexity
- Greater confidence in interactions with healthcare professionals
- Reduction in parental self-blame
- Earlier access to peer support following diagnosis
- Increased confidence to engage in research participation

One family reflected:

> “Finding this community didn’t just help us cope. It helped us breathe.”

Trustees acknowledge that further development of structured outcome measurement tools would strengthen longitudinal reporting, and this will be explored in future years subject to capacity and funding.

LEARNING, CHALLENGES AND RISK

In line with SORP 2026 emphasis on transparency and adaptive governance, Trustees have reflected carefully on key learning during 2025.

Key Learning

- The period immediately following diagnosis is the most destabilising and highest risk for psychological harm.
- Families frequently access community before structured clinical pathways are in place.
- Language used in clinical and public contexts significantly affects wellbeing and understanding.
- Ethical research participation is only possible when families feel informed and supported.
- Infrastructure investment can enable disproportionate impact in small organisations.

Key Challenges

- Infrastructure funding remains harder to secure than programme funding.
- Staff and volunteer capacity limits the pace of strategic development.
- Managing global engagement requires careful safeguarding and moderation.
- Evolving clinical understanding requires continual updating of materials.
- Despite delivering 37 'Mind the Gap' counselling sessions and clear evidence of impact in reducing isolation and strengthening coping strategies, the programme was placed on hold in September due to a lack of available funds. Securing funding for psychosocial support remains a persistent challenge.

Principal Risks

Trustees maintain a risk register which is reviewed regularly. Key risks identified include:

- Concentration of restricted income
- Limited unrestricted reserves
- Dependence on a small leadership team
- Data governance responsibilities associated with registry growth
- International regulatory and coordination complexity

Mitigation strategies include:

- Diversification of funding sources, including US fiscal sponsorship
- Strengthening operational systems including CRM and accounting platforms
- Structured input from Scientific and Patient Advisory Boards
- Formal data transfer agreements and governance review
- Phased capacity growth aligned with available resources

Trustees consider risk management to be proportionate and appropriate to the size and scope of the charity.

LOOKING AHEAD TO 2026

Trustees have identified the following strategic priorities for 2026:

- Publication and dissemination of the international consensus statement
- Registry data analysis and preparation for publication
- Completion of speech and language research project
- Delivery of the in-person international conference planned for July 2026
- Completed production of bereavement resources
- Continued Scientific Advisory Board engagement
- Expansion of fundraising capacity and diversification of income

The charity will continue to prioritise accuracy, safeguarding, collaboration and long-term impact over rapid expansion.

FINANCIAL REVIEW

For the year ended 30 November 2025, the charity reported turnover of £205,304.

The majority of income remained restricted to specific programme areas, particularly research and community support activities. Trustees recognise the continued importance of increasing unrestricted income to strengthen operational resilience.

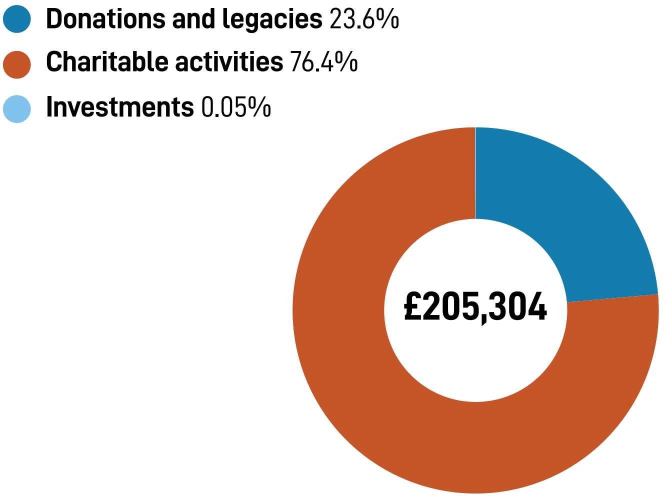
The charity benefited from significant pro bono support during the year, including a £38,043 real-world evidence project delivered in partnership with Costello Medical. Trustees note that such in-kind contributions substantially enhance impact relative to expenditure and represent strong value for money.

Expenditure remained aligned with charitable objectives, prioritising community support, research infrastructure, communications, and operational systems.

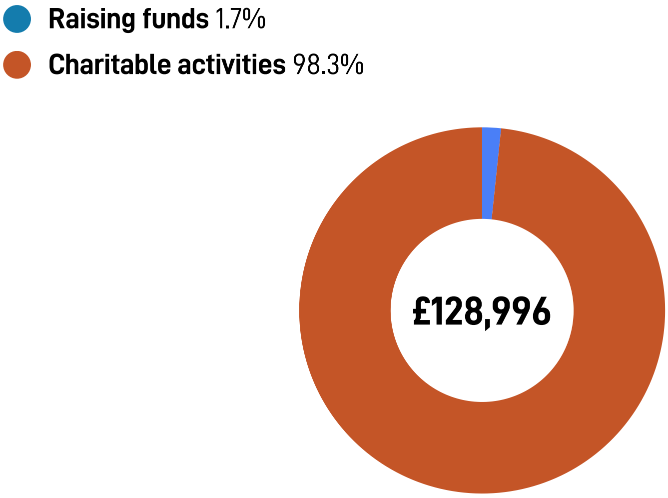
TSA continues to operate as a predominantly volunteer-led organisation, with Trustees contributing substantial unpaid time equivalent to senior executive leadership roles.

Reserves are monitored carefully to ensure financial prudence while enabling delivery of core charitable activities.

Income

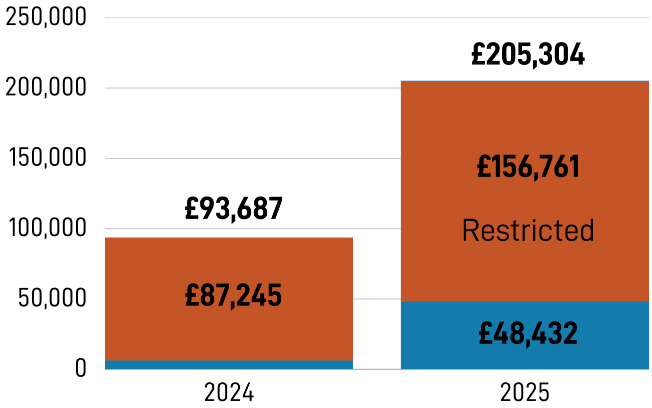


Expenditure

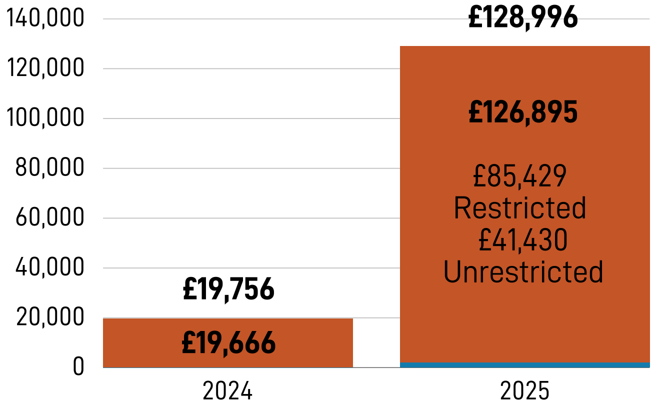


Comparing 2024 and 2025

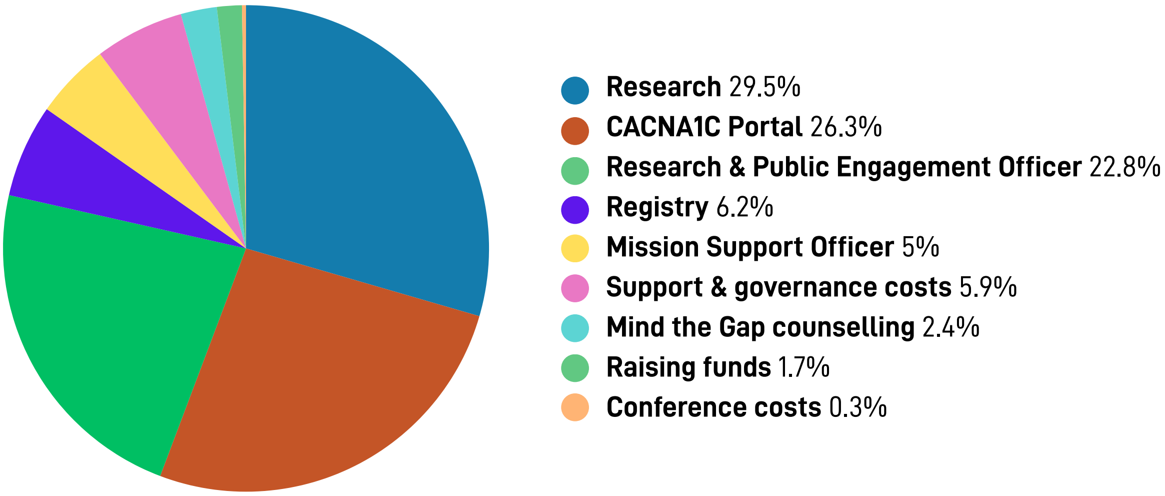
Income breakdown



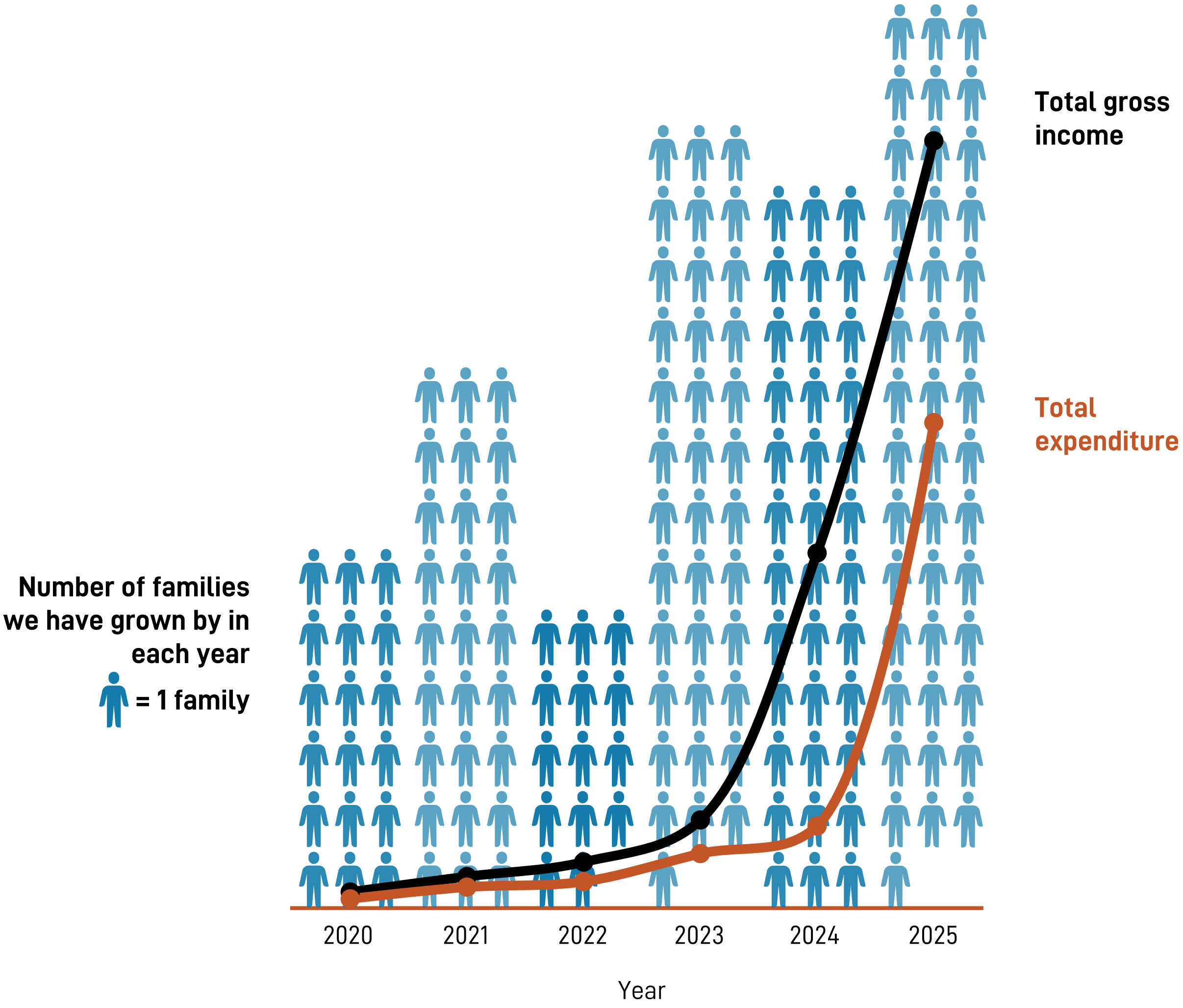
Expenditure breakdown



Expenditure categories



Sustainable Growth: Expanding our community and financial stability



FUNDRAISING AND PARTNERSHIPS

TSA operates within a highly specialised ultra-rare disease field where patient numbers are small and traditional mass fundraising models are neither proportionate nor appropriate. The charity therefore adopts a targeted, relationship-based fundraising approach aligned with its strategic priorities and values.

Income Profile

During 2025, total income was £205,304. The majority of funding remained restricted to specific programme areas, particularly research and community support. Trustees recognise that increasing unrestricted income remains critical to long-term sustainability and operational resilience.

Income was derived from a combination of:

- Strategic grant funding, including support through the CZI Rare As One Network
- Trust and foundation grants
- Corporate and research partnerships
- Community-led fundraising and individual giving
- Platform-based giving through platforms such as GlobalGiving, JustGiving and Benevity
- US-based donations facilitated through Rare Village Foundation fiscal sponsorship

No single income source fully underwrites the charity's core costs. Trustees monitor funding concentration risk carefully and seek to diversify income streams across UK, US and international sources.

Supporting the Vision

Partnership Approach and Ethical Safeguards

Given the increasing interest in CACNA1C-Related Disorders from research and industry stakeholders, Trustees are committed to maintaining independence, transparency and clear governance safeguards in all collaborations.

Partnerships are evaluated against:

- Alignment with charitable objects
- Scientific integrity
- Community safeguarding
- Conflict of interest management
- Long-term public benefit

Trustees retain full oversight of decision-making and ensure that engagement with industry or research partners does not compromise independence or community trust.

Stewardship and Leverage

During the year, TSA secured a £38,043 pro bono real-world evidence collaboration through Costello Medical. Trustees note that such in-kind support significantly amplifies delivery capacity relative to cash expenditure and represents strong stewardship of relationships and resources.

As a predominantly volunteer-led organisation, including significant unpaid executive-level contribution, the charity continues to deliver impact that exceeds what would typically be expected at this level of turnover.

Sustainability and Future Funding Priorities

Trustees recognise that infrastructure funding remains the most challenging yet strategically important funding category. Investment in systems, governance, registry infrastructure and coordination capacity directly underpins programme delivery and research readiness. Priorities for 2026 include:

- Increasing unrestricted income
- Expanding trust and foundation relationships
- Strengthening US-based fundraising capacity
- Developing longer-term funding partnerships aligned with research and registry development

Trustees remain committed to careful growth that prioritises integrity, sustainability and long-term impact over rapid expansion.

TRUSTEES' RESPONSIBILITIES, POLICIES AND FINANCIAL STEWARDSHIP

This report sets out how TSA delivered public benefit during the year, the outcomes achieved, the learning gained, and the challenges faced, in line with current Charities SORP requirements and the anticipated changes under SORP 2026. Trustees have sought to ensure that reporting is transparent, proportionate and grounded in evidence, recognising the particular responsibilities of operating within a rare disease context where misinformation and uncertainty can directly affect wellbeing and care.

Fundraising Policy: We are committed to upholding the highest standards in fundraising, ensuring that all our supporters feel safe, respected, and valued. As a UK-registered charity, we adhere to the standards set by the Fundraising Regulator and follow guidance from other relevant professional fundraising bodies. We never buy or sell personal data, and we take great care to honour individuals' preferences about how and when they wish to hear from us. The principles of the UK General Data Protection Regulation (UK GDPR) are fully embedded in the way we work. We do not use cold calling or cold mailing, and we do not employ third-party fundraising agencies. All of our fundraising is conducted with integrity, transparency, and respect for our community.

Reserves Policy: The charity receives funding for specific purposes which are restricted funds – these are not available for expenditure on other purposes. The general reserves are the unspent unrestricted funds of the charity. The charity currently owns no fixed assets, so the general reserve is held in cash. The general reserve is therefore the free reserves of the charity plus any designated funds, also termed 'unrestricted funds' in the charity's balance sheet.
The purpose of the general reserve is to:
a) fund shortfalls when income does not reach expected levels.
b) fund unexpected expenditures, for example, when projects overrun or unplanned events occur.
c) ensure that the Charity is not unnecessarily holding back on spending in favour of using the resources it has to meet its charitable objectives.
We aim to hold a general reserve, estimated to be in the range £9,000 - £10,000, to cover operating costs for a 3 to 6 month period. At 30 November 2025 total free reserves were £8,150. We are able to operate on a low-funds base as we have kept overheads to a minimum and are not committed to fixed-term costs such as rent or other long-term contracts. This enables us to make sure the majority of income is directed to charitable activities. Our reserves policy is reviewed annually and updated as necessary.

Going Concern: The trustees have reviewed the circumstances of the Charity and consider that adequate resources continue to be available to fund its activities for the foreseeable future. The trustees are of the view that the Charity is a going concern.

ACKNOWLEDGEMENTS AND THANK YOU

Trustees extend sincere thanks to the families who share their experiences, to clinicians and researchers who collaborate generously, to funders and partners who provide financial and pro bono support, and to volunteers who contribute time and expertise.

We also acknowledge the dedication of trustees, advisors, and collaborators whose commitment guides and strengthens the charity's work.

The charity's progress depends on this collective effort and on the willingness of individuals and organisations to support greater understanding. The work continues because of shared commitment, lived experience, and a belief that understanding reduces harm and enables progress.

This Annual Report was created with care in-house at no cost to TSA.

STRUCTURE, GOVERNANCE, AND MANAGEMENT

For the year ended
30 November 2025.

Registered Charity Number: 1185523

Trustees: Sophie Muir – Chair
Nick Muir
Galina Gardiner
Meg Mcloughlin
Gemma Duncan
Susan Bresnahan
Achsah James
Christine Zeigler (appointed 10 September 2025)
Glenn Miller (appointed 10 September 2025)

Registered Office: 8 Butt Street, Minchinhampton, Gloucestershire GL6 9JP

Objectives: To relieve the needs of those affected by deleterious CACNA1C gene changes resulting in CACNA1C-Related Disorders including Timothy Syndrome and Long QT8, their families and carers worldwide in particular but not exclusively by:- (1) Promoting greater understanding of the causes, symptoms and treatment of CACNA1C-Related Disorders including Timothy Syndrome and Long QT8, by the promotion of research and sharing and disseminating of the results of such research for the benefit of the general public; (2) Raising public awareness of the symptoms, needs and related medical conditions of those living with CACNA1C-Related Disorders including Timothy Syndrome and Long QT8.

Structure, governance, and management: Timothy Syndrome Alliance (TSA) is a registered charity number 1185523, governed by the Charities Act 2006. The charity is a Charitable Incorporated Organisation registered on 27 September 2019 under the Foundation Governing Document. The Trustees delegate the charity's day-to-day management to Sophie Muir. Trustees met four times during the year and corresponded regularly via email and other digital means, particularly to keep financial performance under review. New trustees are appointed by the serving trustees, considering the skills required by the board. Trustee induction includes online training (NCVO) to give an overview and understanding of charity governance, regulation and best practice alongside Essential Information for Trustees from the Charities Commission.

Public Benefit: The Trustees confirm that they referred to the Charity Commission's general guidance on public benefit when reviewing the Charity's aims and objectives for the year. Public benefit has been achieved through the activities outlined in the Achievements and Performance section of this report.

Statement of responsibilities of the trustees

The trustees are responsible for preparing the trustees' report and the financial statements in accordance with applicable law and United Kingdom Accounting Standards, including Financial Reporting Standard 102: The Financial Reporting Standard applicable in the UK and Republic of Ireland (United Kingdom Generally Accepted Accounting Practice).

The law applicable to charities in England and Wales requires the trustees to prepare financial statements for each financial year, which give a true and fair view of the state of affairs of the charity and the incoming resources and application of resources, including the net income or expenditure, of the charity for the year. In preparing those financial statements the trustees are required to:

- select suitable accounting policies and then apply them consistently;
- observe the methods and principles in the Charities SORP;
- make judgements and accounting estimates that are reasonable and prudent;
- state whether applicable accounting standards and statements of recommended practice have been followed, subject to any material departures disclosed and explained in the financial statements; and
- prepare the financial statements on the going concern basis unless it is inappropriate to presume that the charity will continue in operation.

The trustees are responsible for keeping proper accounting records which disclose with reasonable accuracy at any time the financial position of the charity and which enable them to ensure that the financial statements comply with the Charities Act 2011, the Charity (Accounts and Reports) Regulations 2008 and the provisions of the constitution. The trustees are also responsible for safeguarding the assets of the charity and hence for taking reasonable steps for the prevention and detection of fraud and other irregularities.

The trustees are responsible for the maintenance and integrity of the corporate and financial information included on the charity's website. Legislation in the United Kingdom governing the preparation and dissemination of financial statements may differ from legislation in other jurisdictions.

Members of the charity have no liability to contribute to the assets of the charity in the event of winding up. The trustees are members of the charity but this entitles them only to voting rights. The trustees have no beneficial interest in the charity.

Auditors / Independent examiners

Godfrey Wilson Limited were appointed independent examiners to the charity during the year and have expressed their willingness to continue in that capacity.

Approved by the trustees on 30 March 2026 and signed on their behalf by

Sophie Muir

Sophie Muir - Chair of the trustees

Independent examiner's report

To the trustees of

Timothy Syndrome Alliance (TSA)

I report to the trustees on my examination of the accounts of Timothy Syndrome Alliance (TSA) (the CIO) for the year ended 30 November 2025, which are set out on pages 33 to 43.

Responsibilities and basis of report

As the charity trustees of the CIO you are responsible for the preparation of the accounts in accordance with the requirements of the Charities Act 2011 (‘the Act’).

I report in respect of my examination of the CIO’s accounts carried out under section 145 of the 2011 Act and in carrying out my examination I have followed all the applicable Directions given by the Charity Commission under section 145(5)(b) of the Act.

Independent examiner’s statement

I have completed my examination. I confirm that no material matters have come to my attention in connection with the examination giving me cause to believe that in any material respect:

(1) accounting records were not kept in respect of the CIO as required by section 130 of the Act; or
(2) the accounts do not accord with those records; or
(3) the accounts do not comply with the applicable requirements concerning the form and content of accounts set out in the Charities (Accounts and Reports) Regulations 2008 other than any requirement that the accounts give a ‘true and fair view' which is not a matter considered as part of an independent examination.

I have no concerns and have come across no other matters in connection with the examination to which attention should be drawn in this report in order to enable a proper understanding of the accounts to be reached.

Rob Wilson

Date: 30 March 2026
Robert Wilson FCA
Member of the ICAEW
For and on behalf of:
Godfrey Wilson Limited
Chartered accountants and statutory auditors
5th Floor Mariner House
62 Prince Street
Bristol
BS1 4QD

Timothy Syndrome Alliance (TSA)

Statement of financial activities

For the year ended 30 November 2025

	Note	Restricted £	Unrestricted £	**2025 Total** **£**	2024 Total £
Income *(and endowments)* from:					
Donations and legacies	3	73	48,359	**48,432**	6,442
Charitable activities	4	156,761	-	**156,761**	87,245
Investments		-	111	**111**	-
Total income *(and endowments)*		156,834	48,470	**205,304**	93,687
Expenditure on:					
Raising funds		-	2,137	**2,137**	90
Charitable activities		85,429	41,430	**126,859**	19,666
Total expenditure	6	85,429	43,567	**128,996**	19,756
Net gains / (losses) on investments		-	-	**-**	-
Net income / (expenditure)		71,405	4,903	**76,308**	73,931
Transfers between funds		-	-	**-**	-
Net movement in funds	7	71,405	4,903	**76,308**	73,931
Reconciliation of funds:					
Total funds brought forward		79,941	13,086	**93,027**	19,096
Total funds carried forward		151,346	17,989	**169,335**	93,027

All of the above results are derived from continuing activities. There were no other recognised gains or losses other than those stated above. Movements in funds are disclosed in note 11 to the accounts.

Timothy Syndrome Alliance (TSA)

Balance sheet

As at 30 November 2025

	Note	**2025** **£**	2024 £
Current assets			
Cash at bank and in hand		**171,155**	94,287
Liabilities			
Creditors: amounts falling due within 1 year	9	**(1,820)**	(1,260)
Net assets / (liabilities)	10	**169,335**	93,027
Funds	11		
Restricted funds		**151,346**	79,941
Unrestricted funds			
Designated funds		**9,839**	6,000
General funds		**8,150**	7,086
Total charity funds		**169,335**	93,027

Approved by the trustees on 30 March 2026 and signed on their behalf by

Sophie Muir

Sophie Muir - Chair of the trustees

1. Accounting policies

a) General information and basis of preparation

Timothy Syndrome Alliance (TSA) is an unincorporated charity registered in England and Wales. The registered office address is 8 Butt Street, Minchinhampton, Gloucestershire, GL6 9JP.

The financial statements have been prepared in accordance with Accounting and Reporting by Charities: Statement of Recommended Practice applicable to charities preparing their accounts in accordance with the Financial Reporting Standard applicable in the UK and Republic of Ireland (FRS 102) (effective 1 January 2019) - (Charities SORP (FRS 102)), the Financial Reporting Standard applicable in the UK and Republic of Ireland (FRS 102).

Timothy Syndrome Alliance (TSA) meets the definition of a public benefit entity under FRS 102. Assets and liabilities are initially recognised at historical cost or transaction value unless otherwise stated in the relevant accounting policy note.

b) Going concern basis of accounting

The accounts have been prepared on the assumption that the charity is able to continue as a going concern, which the trustees consider appropriate having regard to the current level of unrestricted reserves. There are no material uncertainties about the charity's ability to continue as a going concern.

c) Income

Income is recognised when the charity has entitlement to the funds, any performance conditions attached to the item of income have been met, it is probable that the income will be received and the amount can be measured reliably.

Income from the government and other grants, whether 'capital' grants or 'revenue' grants, is recognised when the charity has entitlement to the funds, any performance conditions attached to the grants have been met, it is probable that the income will be received and the amount can be measured reliably and is not deferred.

For legacies, entitlement is taken as the earlier of the date on which either: the charity is aware that probate has been granted, the estate has been finalised and notification has been made by the executor(s) to the Trust that a distribution will be made, or when a distribution is received from the estate. Receipt of a legacy, in whole or in part, is only considered probable when the amount can be measured reliably and the charity has been notified of the executor's intention to make a distribution. Where legacies have been notified to the charity, or the charity is aware of the granting of probate, and the criteria for income recognition have not been met, then the legacy is treated as a contingent asset and disclosed if material.

Income received in advance of provision of a service is deferred until criteria for income recognition are met.

d) Donated services and facilities

Donated professional services and donated facilities are recognised as income when the charity has control over the item, any conditions associated with the donated item have been met, the receipt of economic benefit from the use by the charity of the item, is probable and the economic benefit can be measured reliably. In accordance with the Charities SORP (FRS 102), general volunteer time is not recognised.

On receipt, donated professional services and donated facilities are recognised on the basis of the value of the gift to the charity which is the amount the charity would have been willing to pay to obtain services or facilities of equivalent economic benefit on the open market; a corresponding amount is then recognised in expenditure in the period of receipt.

e) Interest receivable

Interest on funds held on deposit is included when receivable and the amount can be measured reliably by the charity: this is normally upon notification of the interest paid or payable by the bank.

f) Funds accounting

Unrestricted funds are available to spend on activities that further any of the purposes of the charity. Designated funds are unrestricted funds of the charity which the trustees have decided at their discretion to set aside to use for a specific purpose. Restricted funds are donations which the donor has specified are to be solely used for particular areas of the charity's work or for specific projects being undertaken by the charity.

g) Expenditure and irrecoverable VAT

Expenditure is recognised once there is a legal or constructive obligation to make a payment to a third party, it is probable that settlement will be required and the amount of the obligation can be measured reliably.

Irrecoverable VAT is charged as a cost against the activity for which the expenditure was incurred.

h) Allocation of support and governance costs

Support costs are those functions that assist the work of the charity but do not directly undertake charitable activities. Governance costs are the costs associated with the governance arrangements of the charity, including the costs of complying with constitutional and statutory requirements and any costs associated with the strategic management of the charity’s activities. These costs have been allocated between cost of raising funds and expenditure on charitable activities based as follows:

	2025	2024
Raising funds	**1.8%**	0.0%
Charitable activities	**98.2%**	100.0%

i) Debtors

Trade and other debtors are recognised at the settlement amount due after any trade discount offered. Prepayments are valued at the amount prepaid net of any trade discounts due.

j) Cash at bank and in hand

Cash at bank and cash in hand includes cash and short term highly liquid investments with a short maturity of three months or less from the date of acquisition or opening of the deposit or similar account.

k) Creditors

Creditors and provisions are recognised where the charity has a present obligation resulting from a past event that will probably result in the transfer of funds to a third party and the amount due to settle the obligation can be measured or estimated reliably. Creditors and provisions are normally recognised at their settlement amount after allowing for any trade discounts due.

l) Financial instruments

The trust only has financial assets and financial liabilities of a kind that qualify as basic financial instruments. Basic financial instruments are initially recognised at transaction value and subsequently measured at their settlement value with the exception of bank loans which are subsequently recognised at amortised cost using the effective interest method.

m) Foreign currency transactions

Transactions in foreign currencies are translated at rates prevailing at the date of the transaction. Balances denominated in foreign currencies are translated at the rate of exchange prevailing at the year end.

n) Accounting estimates and key judgements

In the application of the charity's accounting policies, the trustees are required to make judgements, estimates and assumptions about the carrying values of assets and liabilities that are not readily apparent from other sources. The estimates and underlying assumptions are based on historical experience and other factors that are considered to be relevant. Actual results may differ from these estimates.

The estimates and underlying assumptions are reviewed on an ongoing basis. Revisions to accounting estimates are recognised in the period in which the estimate is revised if the revision affects only that period, or in the period of the revision and future periods if the revision affects both current and future periods.

2. Prior period comparatives: statement of financial activities

	Restricted £	Unrestricted £	2024 Total £
Income from:			
Donations and legacies	-	6,442	6,442
Charitable activities	87,245	-	87,245
Total income	87,245	6,442	93,687
Expenditure on:			
Raising funds	-	90	90
Charitable activities	18,286	1,380	19,666
Total expenditure	18,286	1,470	19,756
Net income / (expenditure)	68,959	4,972	73,931

3. Income from donations and legacies

	Restricted £	Unrestricted £	**2025** **Total** **£**
Donations	73	10,316	**10,389**
Gifts in kind	-	38,043	**38,043**
Total income from donations and legacies	73	48,359	**48,432**

Gifts in kind comprise of donated services from Costello Medical for a research project carried out for the charity.

Prior period comparative:

	Restricted £	Unrestricted £	2024 Total £
Donations	-	5,692	5,692
Grants	-	750	750
Total income from donations and legacies	-	6,442	6,442

4. Income from charitable activities

	Restricted £	Unrestricted £	**2025** **Total** **£**
Grants	156,761	-	**156,761**
Total income from charitable activities	156,761	-	**156,761**

Prior period comparative:

	Restricted £	Unrestricted £	2024 Total £
Grants	87,245	-	87,245
Total income from charitable activities	87,245	-	87,245

5. Government grants

The charity received no government grants in the period.

6. Total expenditure

	Raising funds	Charitable activities	Support and governance costs	**2025 Total**	Raising funds	Charitable activities	Support and governance costs	2024 Total
	£	£	£	**£**	£	£	£	£
Research	-	38,043	-	**38,043**	-	7,871	-	7,871
App development costs CACNA1C portal	-	33,869	-	**33,869**	-	623	-	623
Research and public engagement officer	-	29,375	-	**29,375**	-	-	-	-
Registry	-	7,976	-	**7,976**	-	-	-	-
Virtual assistant mission support officer	-	6,500	-	**6,500**	-	-	-	-
IT software & consumables	-	-	5,194	**5,194**	-	-	4	4
Mind the Gap counselling	-	3,095	-	**3,095**	-	7,654	-	7,654
Trusts fundraiser	2,000	-	-	**2,000**	-	-	-	-
Accountancy	-	-	1,320	**1,320**	-	-	1,260	1,260
Training	-	-	427	**427**	-	-	-	-
Subscriptions	-	-	411	**411**	-	-	131	131
Conference costs	-	344	-	**344**	-	-	-	-
Travel	-	-	214	**214**	-	-	1,709	1,709
Online donation platform fees	137	-	-	**137**	90	-	-	90
Printing, postage & stationery	-	-	36	**36**	-	-	80	80
Legal fees	-	-	35	**35**	-	-	35	35
Insurance	-	-	20	**20**	-	-	299	299
Sub-total	2,137	119,202	7,657	**128,996**	90	16,148	3,518	19,756
Allocation of support and governance costs	-	7,657	(7,657)	-	-	3,518	(3,518)	-
Total expenditure	**2,137**	**126,859**	**-**	**128,996**	**90**	**19,666**	**-**	19,756

Total governance costs were £1,320 (2024: £1,260)

7. Net movement in funds

This is stated after charging:

	2025 **£**	2024 £
Trustees' remuneration	**Nil**	Nil
Trustees' reimbursed expenses	**214**	1,655
Independent examiner's remuneration:		
▪ Independent examiner's fee	**1,320**	1,260

Two trustees (2024: one trustee) were reimbursed for travel expenses.

8. Taxation

The charity is exempt from corporation tax as all its income is charitable and is applied for charitable purposes.

9. Creditors: amounts falling due within 1 year

	2025 **£**	2024 £
Trade creditors	**500**	-
Accruals	**1,320**	1,260
	1,820	1,260

10. Analysis of net assets between funds

	Restricted funds £	Designated funds £	General funds £	**Total funds £**
Current assets	151,346	9,839	9,970	**171,155**
Current liabilities	-	-	(1,820)	**(1,820)**
Net assets at 30 November 2025	**151,346**	**9,839**	**8,150**	**169,335**

Prior year comparative	Restricted funds £	Designated funds £	General funds £	Total funds £
Current assets	79,941	6,000	8,346	94,287
Current liabilities	-	-	(1,260)	(1,260)
Net assets at 30 November 2024	79,941	6,000	7,086	93,027

11. Movements in funds

	At 1 December 2024 £	Income £	Expenditure £	Transfers between funds £	**At 30 November 2025** **£**
Restricted funds					
Chan Zuckerberg Initiative DAF	75,540	148,821	(78,015)	-	**146,346**
GlobalGiving	1,901	73	(1,974)	-	**-**
The Renishaw Charities Committee	500	-	-	-	**500**
Souter Charitable Trust	2,000	-	-	-	**2,000**
St. James's Place	-	2,140	(2,140)	-	**-**
The Stanley Grundy Foundation	-	2,500	-	-	**2,500**
The Waterloo Foundation	-	3,300	(3,300)	-	**-**
Total restricted funds	79,941	156,834	(85,429)	-	**151,346**
Unrestricted funds					
Designated funds:					
Registry project	4,000	5,043	(2,000)	-	**7,043**
Speech and language project	2,000	796	-	-	**2,796**
Total designated funds	6,000	5,839	(2,000)	-	**9,839**
General funds	7,086	42,631	(41,567)	-	**8,150**
Total unrestricted funds	13,086	48,470	(43,567)	-	**17,989**
Total funds	93,027	205,304	(128,996)	-	**169,335**

Purposes of restricted funds

Chan Zuckerberg Initiative DAF	An advised fund of Silicon Valley Community Foundation, over a five-year period, is supporting capacity building of our patient-led rare disease organisation, the development of a prioritised research agenda, convening our community, and accelerating collaborative research in our area of scientific focus.
GlobalGiving	Donors giving via our GlobalGiving (a US 501(c)(3) intermediary organisation that facilitates tax-effective donations from U.S. donors to U.K. charities) project page, specifically supported the running of our worldwide CACNA1C patient registry designed to obtain insights to better characterise CACNA1C, its presentation, management and treatment.
The Renishaw Charities Committee	Contributed towards a funding goal to support breakthrough speech and language research for young people affected by CACNA1C-related disorders.

11. Movements in funds (continued)

Souter Charitable Trust	Contributed towards a funding goal to support breakthrough speech and language research for young people affected by CACNA1C-related disorders.
St. James's Place	Contributed towards the continued provision of our ‘Mind theGap’ Counselling Service, delivering specialist mental health support to individuals, couples and families impacted by CACNA1C.
The Stanley Grundy Foundation	Contributed towards a funding goal to support breakthrough speech and language research for young people affected by CACNA1C-related disorders.
The Waterloo Foundation	Concluded its three year support of the CACNA1C Community Registry, enabling meaningful change for our existing and future CACNA1C community.

Purposes of designated funds

Registry project	The CACNA1C Community Registry (CCR) enables meaningful change for our existing and future CACNA1C community - the registry collects real-world data to obtain insights to better characterise CACNA1C, its presentation, management and treatment. Funds paid were for the annual platform fee.
Speech and language project	The Speech and Language Research Project delivers inclusive, globally accessible research to support families and clinicians, inform clinical trials, and advocate widespread sharing to improve diagnosis, treatment, and outcomes for individuals with CACNA1C rare variants.

11. Movements in funds (continued)

Prior year comparative	At 2 December 2023 £	Income £	Expenditure £	Transfers between funds £	At 30 November 2024 £
Restricted funds					
Chan Zuckerberg Initiative DAF	-	77,423	(1,883)	-	75,540
GlobalGiving	1,886	3,322	(3,307)	-	1,901
Postcode Local Trust	8,531	-	(8,531)	-	-
The Renishaw Charities Committee	-	500	-	-	500
Souter Charitable Trust	-	2,000	-	-	2,000
The Waterloo Foundation	565	4,000	(4,565)	-	-
Total restricted funds	10,982	87,245	(18,286)	-	79,941
Unrestricted funds					
Designated funds:					
Registry project	-	-	-	4,000	4,000
Speech and language project	-	-	-	2,000	2,000
Total designated funds	-	-	-	6,000	6,000
General funds	8,114	6,442	(1,470)	(6,000)	7,086
Total unrestricted funds	8,114	6,442	(1,470)	-	13,086
Total funds	19,096	93,687	(19,756)	-	93,027

12. Related party transactions

There were no related party transactions in the current or prior period.