Timothy Syndrome Alliance (TSA)

ANNUAL REPORT 2024

For financial period

1 DECEMBER 2023 TO 30 NOVEMBER 2024

www.timothysyndrome.org

Charity Registration Number: 1185523

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What we do

Our charity was formed in 2019 to improve knowledge and understanding of our ultra-rare genetic disease while also providing support to those impacted by it.

We are run by parents of children and young adults from our community, along with volunteers.

We are led by the needs of our community and by working together and collaborating globally, we can harness greater strength. By empowering and nurturing a thriving global community through sustainable growth strategies, we create a lasting positive impact.

We focus on five interdependent areas of activity – raising awareness, improving diagnosis treatment and care, supporting the global community, providing information and advice, and driving research and clinical development.

CACNA1C-related disorders including Timothy Syndrome and LongQT8 are rare and their prevalence is unknown. Raising awareness is challenging due to our limited patient population, lack of steady funding, low recognition within the medical community, being overshadowed by more prevalent conditions, the complexity of our disease, and a general lack of urgency, all of which make it difficult to gain widespread attention and support.

The complexity of our disease is reflected in its wide range of symptoms, which include abnormal heart function, irregular heartbeat, and abnormal heart structure, as well as developmental delays, incoordination, hypotonia, and features of autism spectrum disorder. Additional symptoms include seizures, attention-deficit/hyperactivity disorder, low blood sugar (hypoglycaemia), immunodeficiencies, endocrinological dysfunction, gastrointestinal issues, hypothermia, facial anomalies, syndactyly (joined fingers or toes), and mild dental, skin, eye, and hair anomalies. In addition, conditions like Long QT and Short QT affect the electrical activity of the heart, causing it to take longer or shorter than normal to recharge between beats, potentially leading to irregular heartbeats that can result in fainting, seizures, or even sudden death.

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 the diagnosis, treatment and care of individuals worldwide with CACNA1C-related disorders including Timothy Syndrome and LongQT8, and to support the families and carers of those diagnosed.

Values

Our core values and beliefs combine to form a solid foundation for the way we approach everything we do. We are determined, supportive, empowering and we are a community.

Our strategic purposes and priorities

Our strategic purposes

- Raising awareness.
- Improving diagnosis, treatment and care.
- Supporting the global community.
- Providing information and advice.
- Driving research and clinical development.

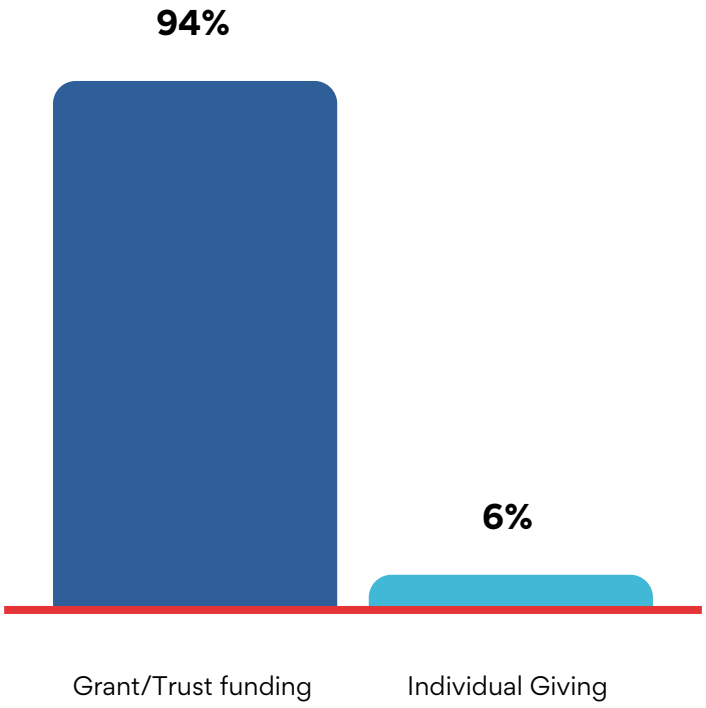
Our priorities to enable our strategy

Uniting families, advancing science, and driving change to transform lives and accelerate progress toward cures.

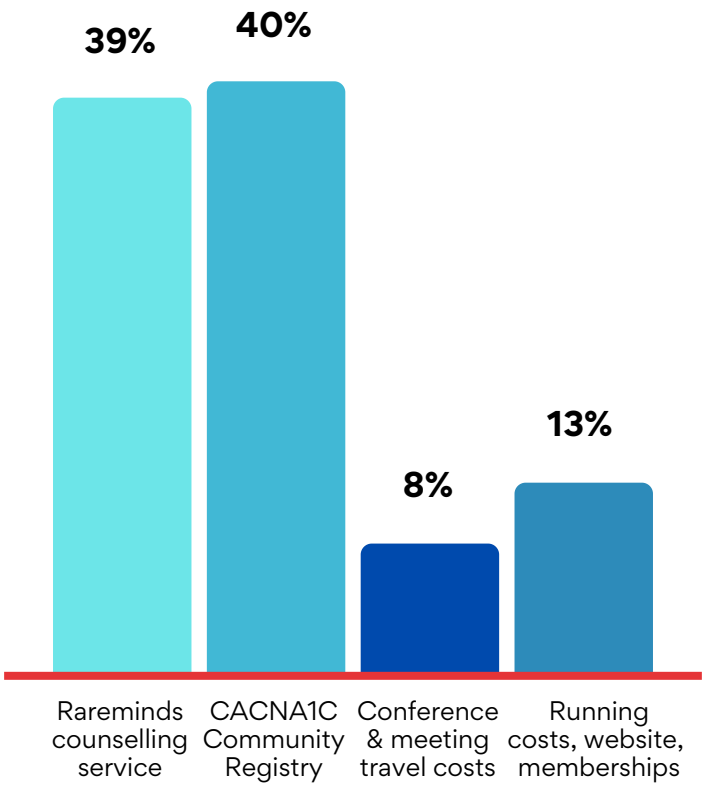
- Expand communications to raise awareness and reach new audiences.
- Build clinical and research networks to improve diagnosis, treatment, and care.
- Strengthen community support through global connection and shared resources.
- Provide trusted information that empowers families and professionals.
- Advance research by supporting funding, collaboration, and participation.
- Influence policy to improve systems and outcomes for our community.

Our finances 2024: Summary

Source of Income



Summary of Expenditure



Our year in review

Delivery against 2024 priorities

Raising Awareness - We have to build awareness of what CACNA1C-related disorders actually are. The typical diagnostic odyssey for people with a rare disease is around 4 to 5 years, though it can often take much longer, and 40% of rare disease patients are initially misdiagnosed. Data from participants in our CACNA1C Community Registry indicate the mean age of diagnosis for individuals with CACNA1C findings is 9.7 years. We're determined to change that, because we want patients/families/professionals to have improved access to the facts they need especially on receipt of diagnosis. Receiving an accurate diagnosis for a rare condition is crucial for appropriate management and support. And we want them to find us, so we can offer support and a community of people who understand, and to help connect them to what could be a life-saving piece of information, or the chance to engage with research that they might not otherwise hear about.

- TSA was named **Charity of the Year 2024** by Healx and through this partnership we gained increased visibility, benefited from fundraising efforts, and engaged in meaningful collaborations to advance the understanding of CACNA1C-related disorders. Sophie and Dr Jack Underwood (TSA Scientific Advisory Board Chair) attended the Healx Team Day in Cambridge presenting in-depth on the challenges of CACNA1C. Healx is a pioneering AI-driven biotech company focused on accelerating the discovery and development of treatments for rare diseases. Their support amplified our reach and provided valuable exposure including a collaborative post on Rare Disease Day.

- The **2024 Smiley Charity Film Awards** received over 500 entries, and our film **"The Diagnosis Challenge"** released on Rare Disease Day 2023, was a finalist. Trustees Sophie and Galina along with our film-maker Rob Bradshaw attended the awards ceremony – we didn't win this time however we continue to punch above our weight.
  The Smiley Charity Film Awards celebrate the power of film in raising awareness for important causes. As the world's largest campaign of its kind, the awards recognise outstanding storytelling from charities, amplifying their messages and inspiring action. They are the world's largest cause-based film campaign, recognised by BFI and IMDb.
  Our short film highlights the lengthy and frustrating journey from first noticing that something isn't right and going to the doctor to actually receiving a diagnosis. This period of time even has a name, the 'Diagnostic Odyssey'. It's a process that can take many years, with seemingly endless tests along the way. The film is a montage list of healthcare professionals seen by just four of our families during their diagnostic odysseys.

- The production of what we were calling the Rare Disease Research Journey has been completed, and the film aptly named **"Connections"**. It was launched on our 5th birthday of becoming a registered charity. This film, a collaboration between TSA and Cardiff University, explores the importance of connections between individuals and families with lived experience of CACNA1C-related disorders (CRDs) and Timothy Syndrome (TS), academics, clinical teams and researchers. The more links we make, the stronger our network becomes and the faster we grow our understanding of our rare disease. Without connections, we can't grow or learn.

- We again entered the **Student Voice Prize**, an annual international essay competition that gives students the opportunity to engage with the rare disease community. Run by Medics4RareDiseases and Beacon for Rare Diseases, the competition encourages future healthcare professionals to explore the challenges faced by rare disease patients and advocate for improved diagnosis, research, and care. Whilst our 3rd-year medical student Diana did not win the competition, her essay summary below sums up how powerful campaigns such as this can be and the impact on the students taking part.
"The paradox of the low statistical prevalence of a rare disease coupled with its high potential to cause adverse health outcomes and emotional turbulence was called to my attention when I had the privilege of speaking to S about her diagnostic journey with CACNA1C-related disorders. The unpredictability that such a minute gene change can have on a person's life and those that surround them encourages me to be on high alert as a future doctor, especially with the scarcity of information and awareness that coexist with these conditions. Whilst researching and writing this essay, I indubitably knew that change must be made within my practice as well as the medical system itself."

- Being part of an alliance strengthens collaboration, amplifies advocacy efforts, and provides access to shared resources, expertise, and a wider support network. We are delighted to have joined the following alliances:
ePAG ERN-ITHACA, the European Reference Network for Rare Malformation Syndromes, Intellectual and Other Neurodevelopmental Disorders.
AGENDA (Alliance for the Genetic Etiologies of Neurodevelopmental Disorders and Autism), led by the Autism Science Foundation. At one meeting we were joined by one of our SAB members, Dr Rebecca Levy from Stanford University, to talk about the importance of the recent findings of using organoids to understand the role of gene therapies using Timothy Syndrome as an example.
CombinedBRAIN (The Consortium for Outcome Measures and Biomarkers for Neurodevelopmental Disorders). This followed a project we undertook with Terry Jo Bichell and one of her students Lipika who focused on CACNA1C as part of her Translational Neuroscience pre-medical undergraduate course at Vanderbilt University.
CZI Rare As One Network whose vision is to create, nurture, and support an engaged community of collaborative interdisciplinary research teams who will enable scientific breakthroughs that grow from combined expertise.

- Our digital presence plays a crucial role in raising awareness and engaging the community, and this year, our social media platforms and website have proven their impact. Compared to the previous year, we have seen growth in reach, engagement, and website visits, demonstrating that our message is resonating with a wider audience. By sharing research updates, personal stories, and advocacy efforts, we have strengthened our network and inspired more people to take an interest in CACNA1C and the work of TSA. This momentum highlights the power of clear, consistent communication in driving awareness and support.

  A great example of the butterfly effect from social media is the increase in the growth of our CACNA1C community in the months following an intensive campaign. The “For The Love Of CACNA1C” campaign ran every day for the month of February on our social media channels. We expanded this campaign during April with a focus on our families in Brazil ensuring posts were created in both Brazilian Portuguese and English languages. In March and May, each campaign saw community growth double compared to the previous month, including in May a new family from Brazil.

- We continue to be active with existing alliances ePAG ERN GUARD-Heart, UK Rare Epilepsies Together (UKRET), The Neurological Alliance, Genetic Alliance UK, Rare Diseases International, EURORDIS, Global Genes Global Advocacy Alliance, Global Heart Hub and The Voltage-Gated Calcium Channel Collective (VGCCC). Alongside this TSA benefits from services provided by Beacon for Rare Diseases, Costello Medical and CamRARE including mentoring, training, and networking opportunities.

  We were delighted to share how we are involved and engaged in the ePAG ERN GUARD-Heart (European Reference Network for Rare and Low Prevalence Complex Diseases of the Heart) as part of their **podcast** series in June and featured in the April **Newsletter** with a focus on CACNA1C.

  We have also participated in the **Talking Charities podcast** series hosted by St. James’s Place, a previous funder of our work. This series explores key topics in the charity sector and features expert insights on fundraising, governance, and impact. This was a wonderful opportunity to share the impact of our work, including the Mind the Gap counselling, and raise awareness. Discussions took place over two episodes (1 and 2).

Our Scientific Advisory Board Chair Jack was one of the scientists who took part in **ART-TRAnslations: Translating Science Through Art**. Paired with talented artist Naomi Tomkys OBE this is a creative initiative that uses visual storytelling to communicate complex scientific concepts related to rare diseases. This project was sponsored by LifeArc and supported by CamRARE. It was featured at RAREfest24, an event dedicated to rare diseases. By bridging the gap between research and public understanding, ART-TRAnslations makes science more accessible, engaging, and inclusive for diverse audiences.

The plaque reads:
'Ivory Tower
The origin came from Dr Jack Underwood as we discussed scientists being seen as living in Ivory towers, the reality of this is wonderfully different. I've used symbolism as a shorthand to the nature of Jack's work. A bee (dummy) - hard work; a boat (child's plate) - journey; a bear - strength, courage. I've also played with iconography giving Jack's daughter a halo of socks lit up with a computer screen rather than gold leaf.

I've included some of Jack's personal items including a slightly broken old globe. It reflects the way the world thinks about these genetic diseases, often out of date, like the old map of the world.
The drawings highlight that when you look at graphs and academic papers, there are children and families struggling behind every data point. These portraits are named after 4 children with CACNA1C variants that recently passed away. In memory of Halle; Xavier; Theodora; and Griffin.'

Improving diagnosis treatment and care - CACNA1C-related disorders have no geographic boundaries. They impact individuals worldwide regardless of location, ethnicity, or socioeconomic status. Gaps in healthcare systems and workforces vary from region to region and the need for global collaboration in diagnosis, treatment and care couldn't be greater. The more we identify and tackle these gaps in the early detection, diagnosis, monitoring or treatment of CACNA1C, the better the diagnostic and post-diagnostic journey will be for individuals who receive a positive CACNA1C finding.

TSA noted a significant lack of individuals in the UK joining the global CACNA1C community. To investigate this, we set up a project in 2023 to identify if there were any barriers to reporting our rare disease. We began mapping real-world data from Freedom of Information requests in England and found that over 100 individuals with CACNA1C rare variants had been classified as having Variants of Uncertain Significance (VUS).

VUS are a common challenge for ultra-rare diseases like ours. Because awareness is low and symptoms can vary widely, it is difficult to link specific genetic changes to predictable health outcomes. There are no large-scale studies of CACNA1C-related disorders, so genetic test results often do not guide medical care. Many results are classified as VUS, meaning their impact on health is unclear and the result is not reported to the individual.

In August, as one of three rare disease charities we were awarded £38,000 worth of pro bono support from Costello Medical to assist with our **real-world evidence (RWE) generation and communication**. Costello Medical provides scientific support to the healthcare industry.

Our challenge: CACNA1C-related disorders, including Timothy Syndrome and LongQT8, are multisystemic and individuals found to have a CACNA1C rare variant should receive cardiac, developmental, psychiatric, endocrine (particularly diabetic) and neurological (epilepsy) screening [1, 2, 3, 4]. This is currently not understood nor advocated for by key bodies including the National Health Service (NHS) and Genomics England.

Our primary objective: Efficiently disseminate impactful information on the recommended approach in response to a CACNA1C rare variant finding, ideally utilising the NHS and Genomics England data obtained via Freedom of Information Act requests, to increase the screening of individuals with CACNA1C findings in the UK (including those with VUS), to educate healthcare professionals and help patients make informed decisions about their care and to guide future advocacy initiatives.

We are incredibly grateful and excited to be working with Costello Medical on this huge impactful project and look forward to sharing the outcomes in 2025.

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1. Levy RJ, Timothy KW, Underwood JFG, Hall Jeremy, Bernstein JA, Pașca SP. A cross-sectional study of the neuropsychiatric phenotype of CACNA1C-related disorder. Pediatr Neurol. 2022;102542. doi:10.1016/j.pediatrneurol.2022.10.013

2. Bauer R, Timothy KW, Golden A. Update on the Molecular Genetics of Timothy Syndrome. Front Pediatr. 2021;9(May). doi: 10.3389/fped.2021.668546

3. Cipriano L, Piscopo R, Aiello C, Novelli A, Iolascon A, Piscopo C. Expanding the Phenotype of the CACNA1C-Associated Neurological Disorders in Children: Systematic Literature Review and Description of a Novel Mutation. Children. 2024 May;11(5):541. doi: 10.3390/children11050541

4. Timothy KW, Bauer R, Larkin KA, Walsh EP, Abrams DJ, Gonzalez Corcia C, et al. A Natural History Study of Timothy Syndrome. Orphanet J Rare Dis. 2024 Nov 23;19(1):433. doi: 10.1186/s13023-024-03445-x

We are also working on a project with Illumina, exploring the various paths families take to obtain a diagnosis. We are focusing particularly on the reasons why families often receive a Variant of Unknown Significance (VUS) as a result of genetic testing. We're aware that a VUS is both a specific yet also an unspecific result. In order to understand what happens when families receive a VUS as a result of testing, we are hearing about experiences and thoughts from within our community on this topic. Through their experiences we want to develop some themes that might be useful to create change in how people receive results and what information families would find helpful.

39

CACNA1C

families/ individuals

+55

individuals

in our Facebook community

212

members

Patient Advisory Board

Supporting our global community - Every person found to have a CACNA1C rare variant has a unique experience and healthcare journey. We make sure we're there, to offer support, reassurance and information - this includes our Scientific Advisory Board as a point of contact for healthcare professionals and our community when it's needed too. A key bit of knowledge of CACNA1C and shared lived experience can significantly impact your next medical appointment and the care you should expect or request. The right support can be life-changing, even life-saving.

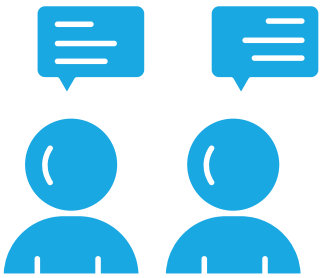
TSA administrates an online private **Facebook Support Group** for individuals and families of CACNA1C-related disorders including Timothy Syndrome and LongQT8, offering 24/7 access to emotional and practical support and information via our community.

The benefits of this are huge. It enables:

- meeting and befriending other people with the same rare disorder and similar experiences from across the world
- learning about CACNA1C
- giving and receiving emotional support
- having a place to speak openly about the impact of CACNA1C and one's thoughts and feelings
- learning coping skills
- feeling empowered and hopeful
- advocating to improve healthcare

As well as welcoming 39 individuals/families impacted by CACNA1C over the year, we saw a 22% increase in our community support group membership as a result of a spouse/partner also joining.

Each request to join our rare disease support group is carefully triaged to ensure it is the right fit, helping us maintain a safe, trusted space for our community. While this process takes time and care, it is an essential part of offering meaningful, immediate support, and ensuring no one feels alone.



49
counselling sessions
provided

106
Dealing with Diagnosis
webinar views

We have been providing our **'Mind the Gap' counselling service** to our community since 2022 when we piloted the service, from which feedback found 100% of participants feeling more positive, resilient, and less isolated. We work with Rareminds, qualified and experienced counsellors and psychotherapists with additional training in addressing psychosocial, physical, and mental health challenges faced by those diagnosed with a rare disease, to address significant psychological strain (including bereavement, anxiety, stress, low mood, emotional exhaustion and bearing uncertainty).
Demand for this confidential service continues to increase and this year we have provided 49 counselling sessions for individuals and couples in our community. This support helps participants cope with the challenges of having or supporting someone with our rare disease through tailored counselling which helps foster a sense of belonging, reducing isolation and providing long-term support for personal growth and coping strategies.

“My counsellor has been my lifeline... Thank you to TSA for allowing us to use this service."

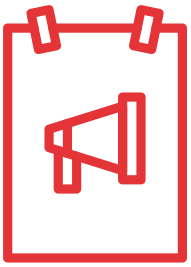
We have additionally created three recorded webinars with Rareminds with open access for our community to enable them to work around caring commitments and time zones; **Dealing with Diagnosis** (received 106 views in the 4 months since production), Looking after Mental Health & Wellbeing, and Managing Uncertainty (will be released 2025). Multiple language transcripts are underway for Dealing with Diagnosis to ensure this is accessible for all our community. A bereavement resource is also under production. When you lose someone you care about as a result of a rare condition, the feelings involved can be challenging and complex.

Providing Information and advice - A variety of new developments this year were shared through our online private Patient Advisory Board, which has 212 members. This hub allows TSA to provide updates and collaborate with our global community on advocacy efforts, ongoing research, partnerships, and campaigns.

- We've also added content to our website, notably our **Glossary** which explains genetic and medical terms in plain English. This original template was created by Genetic Alliance UK and adapted by our TSA Scientific Advisory Board. Plans are underway for our new quarterly newsletter to keep everyone abreast of the very latest happenings, with the first mailing anticipated in February 2025.

- We are delighted to see how much our Wikipedia CACNA1C-related disorders page has grown in views over this last year (92% increase on its first year) and plan to make time in 2025 to contribute further and expand on the content.

- Production of our pathway-focused Educational CACNA1C Interactive Guide for Healthcare Professionals will resume following confirmation of CACNA1C nomenclature expected during 2025. We remain steadfast in our commitment to accuracy and this delay will ensure alignment of the guide with outcomes not yet published.



2 x Scientific poster conference presentations

The impact of CACNA1C real-world data: CACNA1C Community Registry (CCR) design

Driving research and clinical development - This year has been about building on our existing research achievements and partnerships to inform clinical developments and we're proud of all this work. More awareness of CACNA1C-related disorders means more attention, more research and bigger steps towards our vision where shared knowledge and understanding lead to a cure for everyone with a CACNA1C rare variant.

- **CACNA1C Language Consensus** - 20 years ago Timothy Syndrome was first described and named. As the number of people identified with CACNA1C gene variants grows, how do we describe those who don't neatly fit Timothy Syndrome? This is the project we've set out to answer.
  Earlier this year our SAB Chair Jack began convening a panel of international experts with the aim of coming up with clear, well-defined language around Timothy Syndrome and CACNA1C. There are lots of terms being used beyond the original Timothy Syndromes Type 1 and 2, Atypical Timothy Syndrome, Timothy Syndrome Type 3, Timothy Syndrome Variants, Long QT 8, Short QT, CACNA1C variants, CACNA1C-Related Disorders. There isn't a consensus or agreement on what these mean. This isn't helpful for the individuals, their families, their doctors, or researchers.
  Our CACNA1C community has been consulted in this process. The outcome is currently being prepared to be shared with the community and published.

Our Scientific Advisory Board (SAB) has expanded with the addition of Professor Liz Tunbridge, Director of Translational Neuroscience, Boehringer Ingelheim. Liz has a long-standing interest in understanding how the diversity of CACNA1C channels affects different tissues, and therefore symptoms. We are immensely grateful for all the time, effort and enthusiasm our SAB continues to give to our community. It is this teamwork that helps drive us forward.

Scientific Advisory Board

Dr. Jack Underwood (Chair)
Wellcome Trust GW4-CAT Clinical Research Fellow, NMHII, Cardiff University

Dr. Nicola Hall
Postdoctoral Researcher, University of Oxford

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

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

Dr. Gemma Wilkinson
Research Associate, NMHII, Cardiff University

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

Prof Liz Tunbridge
Director of Translational Neuroscience, Boehringer Ingelheim

Following the success of our **Connect CACNA1C Global Network Conference** last year all six presentation recordings are now available with transcripts in 14 languages. Initially accessible via a private link, as much of the work presented had yet to be published, the presentations with transcripts will transfer to a public viewing link at the end of the year to continue to educate and inform both families and healthcare professionals.

October brought Dan Haynes, a new PhD student at NMHII, Cardiff University funded by the Hodge Foundation. His project, which focuses on the role that the CACNA1C gene plays in psychiatric illness onset, will be partly supervised by Dr Jack Underwood. We are the external partner, meaning Dan will support Jack and Sophie by communicating with families, completing administrative duties and helping to organise conferences.

87%

of children face challenges with

speech, language & communication

87% of children with our rare disease face challenges with speech, language and communication.
Some individuals do not develop enough verbal speech to rely on it for their daily communication needs. On top of this, parents and carers are commonly also managing cardiac concerns, developmental delay, incoordination, hypotonia, autism spectrum disorder, seizures, and attention-deficit/hyperactivity disorder.
Early intervention and the correct support can help reduce these impacts and improve communication skills and overall quality of life. No studies have thoroughly looked at speech and language problems using consistent methods or formal tests to clearly distinguish between different speech and language issues. Right now, we don't clearly understand the difference between speech problems caused by issues with the muscles used for speaking (like speech apraxia) and those caused by problems with thinking and language skills. These distinctions are crucial, as without accurate diagnoses, parents and clinicians remain unsure about what therapies to implement.

We have launched a **fundraising campaign** to

- DELIVER inclusive and representative gene-specific speech and language research which will be accessible to our globally marginalised community.

- SUPPORT parents, carers and clinicians with clearer diagnosis, prognosis and treatment planning to alleviate the substantial impact on learning, education, social interaction, self-esteem, mental health, and daily living.

- INFORM speech and language outcome measures for future clinical trials through clearly explained data.

- ADVOCATE the widespread sharing of the research upon conclusion to maximise clinical impact worldwide to transform diagnosis and prognosis and enable precision therapies.

The **CACNA1C Community Registry** has continued to grow and at the end of this year had 90 participants enrolled. The registry collects real-world data to obtain insights to better characterise CACNA1C, its presentation, management and treatment. We are working with our SAB to analyse the Registry data and see how this can be published as research and used to inform clinical and medical treatment.

We created a **scientific poster** using registry data presented at our Connect CACNA1C Global Network Conference. Sophie presented this poster at the British Paediatric Neurology Association (BPNA) Conference in January and again at the Genomics England Research Summit Conference in July.

We are members of the **Bristol University Neurodevelopment Hub** which launched in July and brings together a diverse group of researchers and clinicians interested in mechanisms and consequences of brain development and function to better understand, improve diagnosis of, and identify new therapeutic targets for neurodevelopmental, neurological, and psychiatric conditions. Our strong links with the multi-disciplinary cross-GW4 community researching, modelling, diagnosing, monitoring and treating epilepsy has resulted in a GW4 Building Communities Development Award. The project aims to develop the community to better address the lack of fundamental understanding of epilepsies, improve (epi)/genotypic diagnosis, animal, cell and computational epilepsy models facilitating testing of better targets, screen for novel genetic and drug therapies plus develop better devices for diagnosis, monitoring and treatment.

(The GW4 Alliance (also known as GW4) is a consortium of four research intensive universities in South West England and Wales.)

We were selected to attend the Aspire Biosciences **Rare Neurology/CNS Partnering Event** at the Royal Society of Medicine in London. Both Sophie and SAB member Nicola Hall attended the "speed-date" style networking event designed to build collaborations between patient groups, and pharmaceutical and biotech companies interested in finding treatments for rare diseases that affect the brain and nervous system. The day was a whirlwind—exciting, intense, and full-on. So many great conversations took place, and there are plenty of follow-ups to keep the momentum going.

We are also now members of the **BHF Clinical Research Collaborative**, a large national cardiovascular clinical research network that is funded by the British Heart Foundation but is supported by all of the national specialist societies. They aim to link clinicians and researchers more with patients to ensure great collaboration in research.

- Since October 2023 Sophie has been part of the **Rare Diseases Research Network (RDRN)** working group, a partnership project between CamRARE and Patient Led Research Hub, funded by the National Institute for Health and Social Care Research (NIHR) and sponsored by Cambridge University Hospitals NHS Foundation Trust, building an online platform to facilitate rare disease research. This platform was successfully launched in November.

- The **Voltage-Gated Calcium Channel Collective (VGCCC)** unites patients, families, researchers, and clinicians in a collaborative, patient-centred community. We formulated last year and believe we can drive breakthroughs in understanding and treating voltage-gated calcium channelopathies, ultimately improving the lives of those impacted by these conditions. We meet monthly to share updates, discuss challenges, and strategise on our collective efforts. These meetings provide a vital platform for collaboration, ensuring that all voices are heard and that we remain aligned in our mission. On Rare Disease Day we launched our Facebook, Instagram and Bluesky VGCCC accounts and are now underway with the building of our website.

- In June Sophie attended the **EURORDIS Open Academy School on Scientific Innovation and Translational Research** which consisted of one week of intensive in-person training in Barcelona, and followed the e-learning modules and pre-training webinars. The training, delivered by over 20 rare disease expert specialists from across Europe, aims to deepen patient representatives' understanding of how pre-clinical research translates into real benefits for rare disease patients. The training equips participants with knowledge and skills to empower them to effectively participate in discussions with researchers, policymakers, and companies active in rare disease research.

- To complete another incredible year it was confirmed in October that we were one of the first international patient organisations to join the **Chan Zuckerberg Initiative Rare As One Network (Cycle 3)** to help drive forward research (but not fund it directly).
  The Chan Zuckerberg Initiative invites applications for five-year projects from patient-led rare disease advocacy organisations to join the Rare As One Network. Grantee organisations will accelerate research within individual disease areas and work across specified areas of scientific focus (channelopathies; ciliopathies; or inborn errors of metabolism) to develop shared research priorities and proposals.
  For the 2024 award cycle, Timothy Syndrome Alliance (TSA) is one of 30 patient organisations around the world to receive a funding grant and has been awarded $800,000 over five years.
  Further information is available at: Rare As One Project – Chan Zuckerberg Initiative and Rare As One Network RFA – Chan Zuckerberg Initiative
  We look forward to sharing our ambitious and well-informed strategy as a result of this support in the coming year, and believe this will transform the progress for CACNA1C-related disorders.

RARE AS ONE

Fundraising

TSA operates day-to-day with just one core team member and limited internal capacity. While rare diseases are collectively common, our specific condition remains under-recognised, making it harder to attract attention and funding. There is often a perception that rare disease needs will be met by national healthcare systems or government services, but many in our community tell us they have found support nowhere else. Unlike more widely known conditions, we face significant barriers to public fundraising, despite the urgent and unmet needs we address. In an increasingly competitive funding landscape, we are often applying alongside larger organisations with established teams and broader visibility — yet our work delivers targeted impact where few others can reach.

Grants - Our primary source of income comes from charitable trusts and foundations. Over the past year, we have successfully secured support from several of these organisations.
We would like to extend our heartfelt thanks to the following trusts and foundations for their generous support this year:

The Renishaw Charities Committee
The Waterloo Foundation
Souter Charitable Trust
Jeans For Genes
Chan Zuckerberg Initiative DAF, an advised fund of Silicon Valley Community Foundation

Donations - With rising living costs continuing to strain household budgets in 2024, individual giving became harder for many. Even so, the kindness and commitment of our community shone through, helping us grow despite the odds.
We are so grateful to everyone who found a way to support us this year.
Thank you to those who contributed to the JustGiving Healx appeal (£1,301) and Ethan's incredible Sahara Desert Trek fundraiser (£1,195).
Heartfelt thanks also to Susan, MaryAnn, and Jill for choosing us for their Facebook birthday fundraisers, and to the Minchinhampton Community Choir and the Crown Public House for hosting a fantastic quiz night.
We're equally grateful to those who gave through platforms like Benevity and GlobalGiving, to our generous one-off donors, and to our wonderful regular donors who support us through standing orders.
Your generosity continues to sustain our work and bring hope to families who need it most.

We are eager to achieve the Speech & Language Research campaign target (detailed on page 16). £4,238 of the £5,693 raised by you has been allocated towards this ambitious £16,000 target total. In October we were delighted to learn that the Trustees of the Speech, Language and Hearing Foundation agreed to make a commitment of £4,000 towards the project dependent on us being able to secure the outstanding amount required to launch this project.
Fundraising continues into 2025.

Fundraising events - A ball you say? What a great idea.

A group of amazing friends, that trustees Sophie and Nick met at their local nursery and mother/baby groups with their middle son Calvin, organised a ball for the last evening of TSA's financial year. They'd been witness to Calvin growing up. Their kids had even learnt Makaton sign language to communicate with Calvin as he didn't talk for his first 7 years. Alongside this, they had seen the battles and struggles of hospital and therapy visits, educational support, the near 10-year diagnostic odyssey of not knowing the reason why he was presenting with so many challenges, genetic testing and then the post-genetic odyssey search for information and other children just like him.

They have seen us put our heart and soul into growing Timothy Syndrome Alliance (TSA) as a charity over the last 5 years. Organising a ball was such a kind and thoughtful display of support. Huge thank you to The Hunter Moon Ball team for such an amazing event (which included a viewing of the film Connections), to the sponsors and donors of silent auction prizes and of course to those who came to support in their black tie glamour for live music and dancing. What a ball!

Spoiler alert for the 2025 Annual Report: the event raised an amazing £3,005.

'Twas the night of the Hunter Moon Ball, what a sight,
When you gathered together to make the night bright.
For TSA's cause, you gave up your eve,
Your kindness and giving, hard to believe!

You bought raffle tickets, you bid with great flair,
Your generosity truly filled up the air.
With dancing and laughter, old friends and new,
We hope that the evening brought joy to you too.

To those who donated, both funds and fine wares,
Your gifts have brought hope beyond all compares.
And to our sponsors, who helped pave the way,
Your support made the night a resounding hooray.

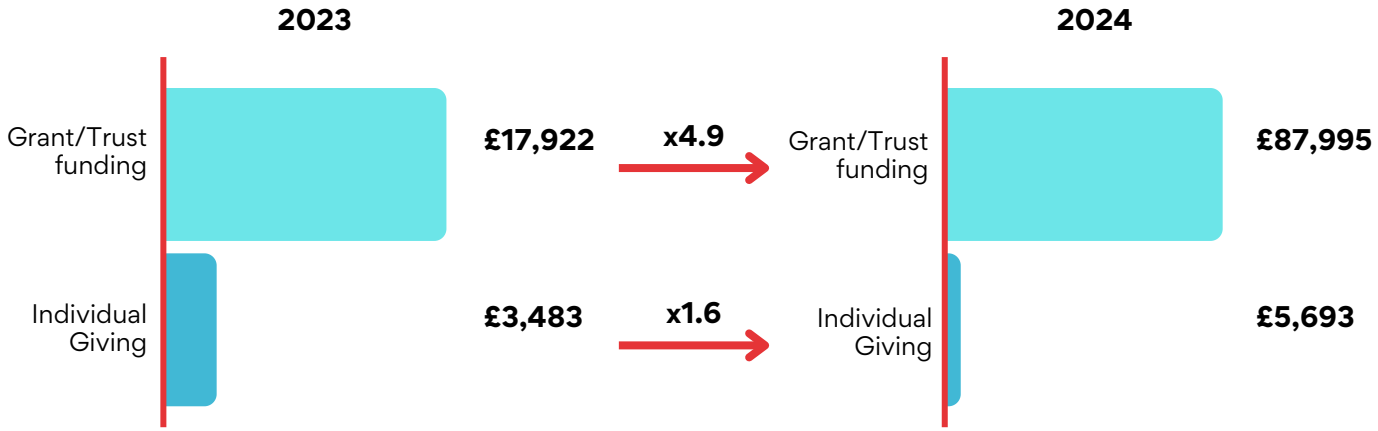
The team worked so hard, their passion so clear,
To bring us together and spread festive cheer.
Their efforts were shining, the event a success,
To them and to you, our heartfelt address.

So thank you for coming, for showing you care,
For helping rare lives with the love that you share.
We wish you a season that's merry and bright,
And thank you again for such a magical night.

'The Hunter Moon Ball team, headed by Rosie, were Charlotte, Kathryn, Annie, Hannah, Sophie B, Cathy and Sue.

Financial review

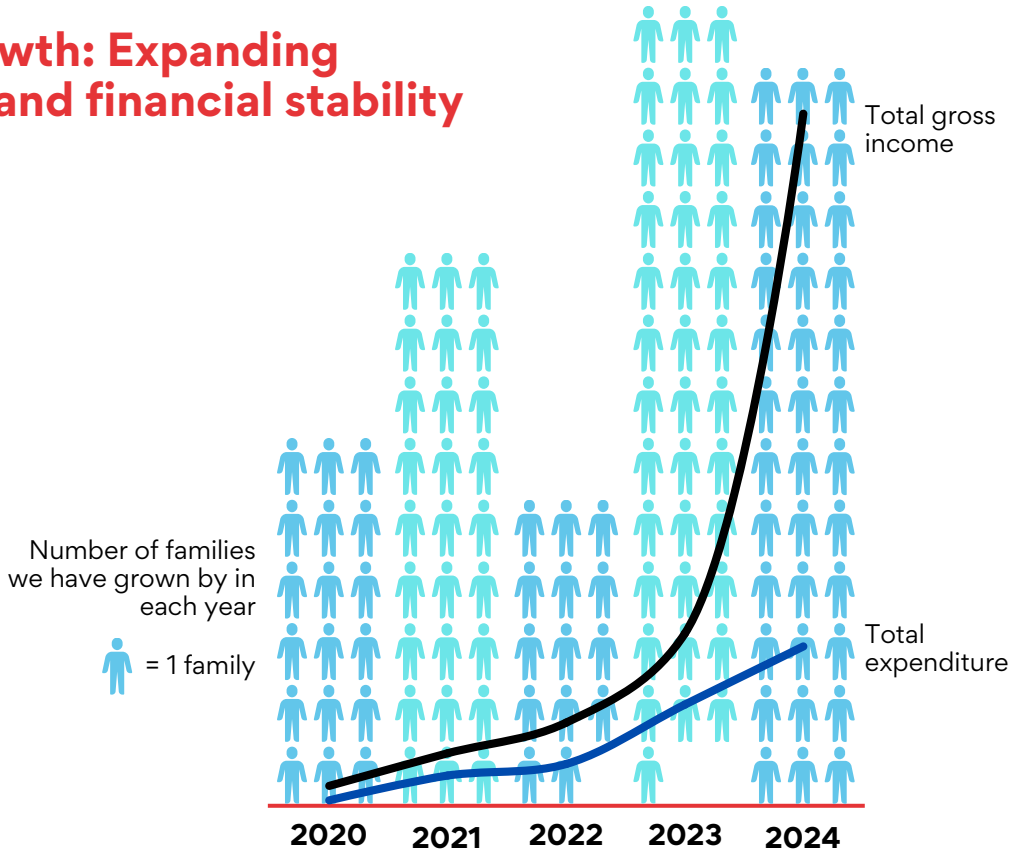
Income by source



Our determination and drive have fuelled significant growth this year, including a notable increase in our community size.

- Our income from trusts and foundations has increased nearly five times and individual giving (including gift aid) has increased by 60%. Thank you to all of our supporters this year for their generous contributions.
- This has powered an expenditure increase of 60% on last year, ensuring we are meeting our aims by uniting families, advancing science, and driving change to transform lives and accelerate progress toward cures.

Sustainable Growth: Expanding our community and financial stability



- **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 2024 total free reserves were £7,086. 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.

Looking forward to 2025

We are focused on the same strategic priorities as last year, but have added a fifth as we want to become better equipped *to do more, reach more people, and achieve greater impact*.
We will be having an **online conference** with an **in-person conference** the following year.
We will also aggregate and harmonise clinical, genetic, and molecular data, and develop **a global CACNA1C Portal** to provide a unified resource for characterising the clinical spectrum of CACNA1C-related disorders, linking specific genetic variants with their phenotypic manifestations.
The portal will support clinical variant interpretation, enhance patient care management, and identify biomarkers for clinical trials.

- 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.
- Capacity builds capacity.
  Building capacity creates the conditions for further growth and effectiveness.
  We will hire a **Mission Support Officer** — a vital role that will helps us grow our capacity and focus on what matters most. By having dedicated support behind the scenes, we can work more efficiently, respond faster, and take on more without burning out.
  Making sure research stays grounded in the real lives and voices of the people it aims to help we will welcome a **Research and Public Engagement Officer** to the team — a role designed to strengthen the bridge between science and our community. They'll play a key role in making complex research easier to understand. By sharing updates in clear, accessible language, they'll help, families stay informed and involved, while keeping researchers connected and aware of community needs.
  We will support our core operations with a **CRM and accounting software**, helping us stay organised, accountable, and ready to grow. Our CRM, which stands for Customer Relationship Management, will help us stay connected with our community, thank people properly, and make sure no one falls through the cracks. Our accounting software will help us manage our finances clearly and accurately — tracking income, expenses, and budgets so we can report transparently and meet all our financial obligations. It means we will spend less time on admin and more time delivering impact.

Thank you

None of this impact would be possible without the incredible people and organisations who stand beside us — funding our work, offering pro bono support, and generously sharing their time, expertise, and energy. Huge thanks to Dmitry for continued voluntary commitment to our TSA website and as well as the build of the website for The VGCCC, to our conference language transcript volunteers Hugo, Therese and Tricia for their human attention to detail and to Rob at Limestone Media for going above and beyond with our filming. We're deeply grateful to our Scientific Advisory Board members for their unwavering dedication to advancing understanding of CACNA1C and guiding our research efforts.

As Chair, I would also like to personally thank my fellow trustees for their continued guidance, expertise, and commitment throughout the year. It is a privilege to serve alongside such a dedicated team. An extra thank you to Costello Medical for their pro bono support with our trustee recruitment project, through which we came up trumps with Achsah, and the amazing resources we now have for future recruitment rounds

Our voice wouldn't be as strong without the support and engagement of our CACNA1C community. Besides sharing stories and photos which greatly helps signpost new families, encouraging and supporting each other, attending our conferences, supporting our social media channels, joining the registry, and contributing to research, they make the impossible possible. Thank you. We are Stronger Together.

How to connect and support

We have been certified as the equivalent of a U.S. public charity, allowing U.S. donors to give with confidence via your Donor Advised Fund (DAF).
Ensure you select the correct organisation using our Registered Charity number: 1185523.

NGOsource
Equivalency Determination on File

Help us make a difference

GlobalGiving tax-deductible in the US

Create online fundraising page

Benevity Workplace Giving

Take on a challenge in support of TSA

Raise awareness on social media

Be the difference. Volunteer

This Annual Report was designed and created with love at no cost to TSA

TIMOTHY SYNDROME ALLIANCE (TSA) TRUSTEES' REPORT General Information

For the year ended
30 November 2024.

Registered Charity Number: 1185523

Trustees: Sophie Muir – Chair
Nick Muir
Galina Gardiner
Meg Mcloughlin
Gemma Duncan (appointed 11 March 2024)
Susan Bresnahan Appointed 11 June 2024)
Achsah James (appointed 6 November 2024)
Katherine W Timothy (resigned 24 November)

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 6 May 2025 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 2024, which are set out on pages 28 to 38.

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: 6 May 2025
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 2024

	Note	Restricted £	Unrestricted £	**2024 Total £**	2023 Total £
Income *(and endowments)* from:					
Donations and legacies	3	-	6,442	**6,442**	3,483
Charitable activities	4	87,245	-	**87,245**	17,922
Total income *(and endowments)*		87,245	6,442	**93,687**	21,405
Expenditure on:					
Raising funds		-	90	**90**	-
Charitable activities		18,286	1,380	**19,666**	12,305
Total expenditure	6	18,286	1,470	**19,756**	12,305
Net gains / (losses) on investments		-	-	**-**	-
Net income / (expenditure)		68,959	4,972	**73,931**	9,100
Transfers between funds		-	-	**-**	-
Net movement in funds	7	68,959	4,972	**73,931**	9,100
Reconciliation of funds:					
Total funds brought forward		10,982	8,114	**19,096**	-
Total funds carried forward		79,941	13,086	**93,027**	9,100

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 2024

	Note	**2024** **£**	2023 £
Current assets			
Cash at bank and in hand		**94,287**	19,096
Liabilities			
Creditors: amounts falling due within 1 year	9	**(1,260)**	-
Net assets / (liabilities)	10	**93,027**	19,096
Funds	11		
Restricted funds		**79,941**	10,982
Unrestricted funds			
Designated funds		**6,000**	-
General funds		**7,086**	8,114
Total charity funds		**93,027**	19,096

Approved by the trustees on 6 May 2025 and signed on their behalf by

Sophie Muir

Sophie Muir - Chair of the trustees

Timothy Syndrome Alliance (TSA)

Notes to the financial statements

For the year ended 30 November 2024

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:

	2024	2023
Raising funds	**0.0%**	0.0%
Charitable activities	**100.0%**	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 £	2023 Total £
Income from:			
Donations and legacies	-	3,483	3,483
Charitable activities	17,922	-	17,922
Total income	17,922	3,483	21,405
Expenditure on:			
Raising funds	-	-	-
Charitable activities	10,940	1,365	12,305
Total expenditure	10,940	1,365	12,305
Net income / (expenditure)	6,982	2,118	9,100

3. Income from donations and legacies

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

Prior period comparative:

	Restricted £	Unrestricted £	2023 Total £
Donations	-	3,483	3,483
Total income from donations and legacies	-	3,483	3,483

4. Income from charitable activities

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

Prior period comparative:

	Restricted £	Unrestricted £	2023 Total £
Grants	17,922	-	17,922
Total income from charitable activities	17,922	-	17,922

5. Government grants

The charity received no government grants in the period.

6. Total expenditure

	Raising funds £	Charitable activities £	Support and governance costs £	**2024 Total £**	Raising funds £	Charitable activities £	Support and governance costs £	2023 Total £
Research	-	7,871	-	**7,871**	-	8,135	-	8,135
Counselling	-	7,654	-	**7,654**	-	1,280	-	1,280
App development costs	-	623	-	**623**	-	-	-	-
Translation services	-	-	-	**-**	-	1,096	-	1,096
Consultancy	-	-	-	**-**	-	176	-	176
Accountancy	-	-	1,260	**1,260**	-	-	-	-
Online donation platform fees	90	-	-	**90**	-	-	-	-
Insurance	-	-	299	**299**	-	-	-	-
Legal fees	-	-	35	**35**	-	-	35	35
Printing, postage and stationery	-	-	80	**80**	-	-	23	23
IT software and consumables	-	-	4	**4**	-	-	98	98
Subscriptions	-	-	131	**131**	-	-	136	136
Travel	-	-	1,709	**1,709**	-	-	1,326	1,326
Sub-total	90	16,148	3,518	**19,756**	-	10,687	1,618	12,305
Allocation of support and governance costs	-	3,518	(3,518)	-		1,618	(1,618)	-
Total expenditure	**90**	**19,666**	**-**	**19,756**	**-**	**12,305**	**-**	12,305

Total governance costs were £1,260 (2023: £nil)

7. Net movement in funds

This is stated after charging:

	2024	2023
	£	£
Trustees' remuneration	**Nil**	Nil
Trustees' reimbursed expenses	**1,655**	1,207
Independent examiner's remuneration:		
▪ Independent examiner's fee	**1,260**	-

One trustee was (2023: one trustee) was 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

	2024	2023
	£	£
Accruals	**1,260**	-
	1,260	-

10. Analysis of net assets between funds

	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**

Prior year comparative	Restricted funds	Designated funds	General funds	Total funds
	£	£	£	£
Current assets	10,982	-	8,114	19,096
Current liabilities	-	-	-	-
Net assets at 30 November 2023	10,982	**-**	8,114	19,096

11. Movements in funds

	At 1 December 2023 £	Income £	Expenditure £	Transfers between £	**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**
The Stanley Grundy Foundation	-	-	-	-	**-**
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**

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.
Postcode Local Trust	A grant-giving charity funded entirely by players of People's Postcode Lottery, supported the continued provision of our 'Mind the Gap' Counselling Service for our CACNA1C community.

11. Movements in funds (continued)

The Renishaw Charities Committee	Contributed towards a funding goal to support breakthrough speech and language research for young people affected by CACNA1C-related disorders.
The Stanley Grundy Foundation	Enabled our conference to be inclusive, accessible and global through a translation service.
Souter Charitable Trust	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.

Prior year comparative	At 1 December 2022 £	Income £	Expenditure £	Transfers between £	At 30 November 2023 £
Restricted funds					
GlobalGiving	-	1,886	-	-	1,886
Postcode Local Trust	-	9,636	(1,105)	-	8,531
The Renishaw Charities Committee	-	500	(500)	-	-
The Stanley Grundy Foundation	-	1,200	(1,200)	-	-
Souter Charitable Trust	4,000	-	(4,000)	-	-
The Waterloo Foundation	-	4,700	(4,135)	-	565
Total restricted funds	4,000	17,922	(10,940)	-	10,982
Unrestricted funds					
General funds	5,996	3,483	(1,365)	-	8,114
Total unrestricted funds	5,996	3,483	(1,365)	-	8,114
Total funds	9,996	21,405	(12,305)	-	19,096

12. Related party transactions

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