

# scan

Autumn 2024

The magazine of the **t**sa  
tuberous sclerosis  
association



Images thanks to recent attendees of the TSA's regular virtual art club

Here for everyone in the TSC community

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# Scan Autumn 2024 highlights

## Meet the researcher

Rory's a final-year PhD researcher, who has 'found his calling' analysing the importance of sleep in children with rare genetic conditions, including TSC



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## Strength after a late TSC diagnosis

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### Fundraising

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 Sally Dutton (Fundraising Manager)  
 Sam Carter (Community Fundraiser)  
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The **TSA Support Line** is  
 free and here for you:  
**0808 801 0700 /**  
**support@tuberous-**  
**sclerosis.org / Webchat**

## A word from your Chair

**“As our current five-year charity strategy for 2019 - 2024 ends, we can reflect on our achievements and how we progress from here**

It's heartening to reflect on the incredible progress we made together over the 2019 - 2024 strategy period, considering our ongoing mission to improve all aspects of the lives of those affected by TSC. We faced many hurdles on the way, such as the pandemic, fighting for treatment access and helping people through the financial crisis. Yet, the TSA was here for you and continues to be here on your TSC journey.

A big focus of the TSA in the 2019 - 2024 strategy was financial stability. We previously invested in vital research opportunities, so it was important to manage our income and outgoings so that we can be here until we find a cure. The charity's reserves are now stronger, although balancing the books is always an ongoing concern for charities of our size. We remain hugely thankful to donors and fundraisers - without you, we simply wouldn't have survived.



**Dr Frances Elmslie**

Looking back at the last few years, I'm particularly proud of our work in launching the TSC Rare Disease Collaborative Network. This brings the UK TSC clinical community together to foster collaboration and innovation in all aspects of TSC care. The TSA continues to manage the network, including a recent face-to-face meeting with a record attendance.

Our strategy for 2025 - 2028 will be shared with you soon. Thank you to everyone who gave their views, as your voice was crucial in making sure we got the strategy right. Our new strategy is ambitious and positive, with our intention to truly progress all-things TSC. Let's look to the next few years with hope.”

- Dr Frances Elmslie (TSA Board of Trustees Chair)

# Getting to know you

We're proud to connect with an amazing number of people in the TSC community. To make the greatest difference, we need to know more about you and how TSC affects you and your loved ones


The more we know about you and your TSC, the bigger our impact will be. For example, you might tell us things about living with TSC that we wouldn't have known otherwise - this'll mean that we can then look at how we can do more in this area.

Your feedback and views will also help us to secure funding for future projects that might not be possible right now, as well as help us to improve our existing services.

Help us today by completing our new 'Getting to Know You' survey. It'll take about 10 minutes, so grab a cuppa and a biscuit and take the time to make a big difference.

To complete the survey, go to: [bit.ly/tsa-get-to-know-you](https://bit.ly/tsa-get-to-know-you) or contact the TSA Support Line to do your survey over the phone (0808 801 0700).

## New TSA team members!



**Danielle's** our fab Grants and Trusts Manager. Danielle finds external funding for different TSA projects: "I love that what we do at the TSA makes a real difference to so many lives in different ways"

**Sam's** our amazing Community Fundraiser. If you've fundraised for the TSA, you might have already spoken with Sam: "I love hearing about the inspiring stories and journeys the TSC community are on, and helping you in your fundraising goals"

**Sally's** our brill Fundraising Manager, helping oversee all-things fundraising at the TSA: "I met many of you at Outlook and Big Day, which was wonderful. Fundraising is a true passion of mine and I look forward to getting to know many people in the TSC community"

# TSA Annual General Meeting 2024: Register your spot

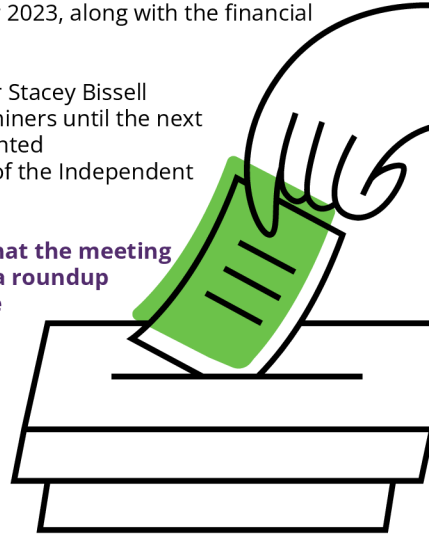
The TSA's Annual General Meeting (AGM) 2024 will take place virtually on **Monday 18 November, 6pm - 7pm**. The AGM is an important opportunity to hear from the TSA's Board of Trustees about how the charity has been performing recently, to hear about our plans for the next 12 months, and to put your views and comments to the board.

Plus, you'll have an important role in voting for important positions and objectives at the TSA for the next year and beyond. Voting will include:

- Review and approve the TSA's financial statements for 2023, along with the financial reports from the trustees and auditors
- To elect if thought fit as Vice Chair: Annemarie Cotton
- To elect if thought fit as Trustees: Maxine Smeaton, Dr Stacey Bissell
- To elect Tudor John as the charity's Independent Examiners until the next General Meeting when financial statements are presented
- To authorise the Trustees to determine the payment of the Independent Examiners in their work for the charity

As part of the AGM 2024, we're delighted to confirm that the meeting will include an update from Dr Chris Kingswood with a roundup of TSC research over the last 12 months, and what the future might hold for TSC breakthroughs.

To register your place at this year's AGM, visit:  
[tuberous-sclerosis.org/AGM2024](https://tuberous-sclerosis.org/AGM2024)



## Big Day and Outlook return!

We're delighted to already confirm that Big Day, our biggest annual event, takes place on **Saturday 14 June** in **Lincoln**. Outlook, for adults mildly affected by TSC, will happen on **Saturday 26 April** in **Glasgow**! The TSA team has worked hard to confirm the events as early as possible, so you can book your places sooner.

We love hosting our face-to-face events and hope to see you there. Visit our website for more information or contact the TSA Support Line to book your places. We're pleased to also confirm that Big Day 2025 will once more include a specialist creche.



# Researcher in Focus:

## Rory O'Sullivan

**TSC research is possible only thanks to the incredible people dedicating their lives to unlocking secrets of the condition**

Rory O'Sullivan (University of Birmingham) is a final-year PhD researcher focused on the importance of sleep in children with rare genetic conditions, including TSC. We caught up with Rory to discuss his PhD research, what he likes to do in his spare time and his plans as a future TSC researcher.

### What was your background before becoming a TSC researcher?

"Before my PhD I did an undergraduate degree in Psychology. During my Psychology degree I spent a year working with Professor Caroline Richards and Dr Stacey Bissell which gave me some early experience in TSC sleep research. My time with Caroline and Stacey motivated me to do the PhD. Through speaking with TSC families, I found that the research we do is really important. After that it almost felt like a calling."

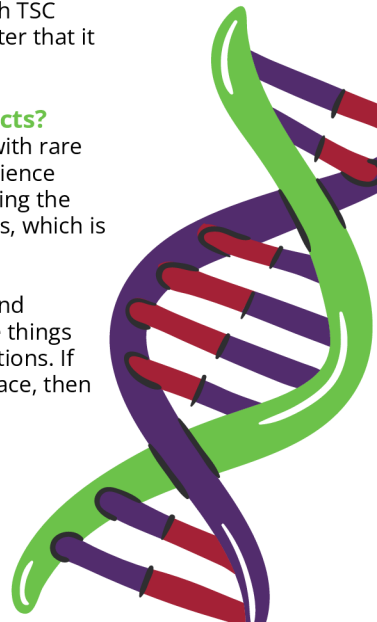
### Can you tell us about your current TSC research projects?

"My research has focused on sleep and behaviour in children with rare genetic syndromes, including TSC. Children with TSC can experience sleep difficulties and this can have an effect on behaviours during the day. However, not much has been done to fully understand this, which is what I want to do.

We're also trying to improve how researchers measure sleep and behaviour. We need to understand how best to measure these things before we can go on to properly address them and other questions. If we're not measuring sleep or behaviour correctly in the first place, then our conclusions might be biased or not quite right."



**Rory O'Sullivan**



### What are your passions outside of research, in your spare time?

"Being in the final year of my PhD, free time is hard to come by these days! When I do have free time I now like to paint, which isn't something I've done since I was a child, it's something I've really picked up over the last few years. I also do calisthenics and love video games. I just got an Xbox set up downstairs last week and I'm trying my hardest not to waste all my time doing that – especially at this crucial stage of writing up my PhD!"

### If you weren't a researcher, what do you think you'd be doing instead?

"For a while, when I started my undergraduate degree, I thought I'd become a clinical psychologist. Before that, I liked the idea of working outdoors and was good at maths so thought I could do something focused on engineering. You know the people that go up into wind turbines to fix them? For a while I thought that could be me!"

### What do you hope we'll see in TSC research in years to come?

"I think the use of wearable technologies to better understand the effect of epilepsy on behaviours, sleep and mood over a longer duration would be really important. Alongside this, there's not too much work looking at sleep in adults with TSC, so I also think this is a very important direction for future research."



**Through speaking with TSC families, I found that the research we do is really important. After that it almost felt like a calling."**



### What's in store for you in the next couple of years?

"I've been offered a four-year position as a postdoctoral researcher at the University of Birmingham focused on how to better measure different cognitive abilities in children with rare genetic syndromes, including TSC. Examples of these abilities include behavioural regulation, attention, working memory and mental flexibility."

### Do you have anything you'd like to say directly to the TSC community?

"I really want to say thank you. Coming towards the end of my PhD research, I really recognise how people's motivations to take part in research has really rubbed off on me, particularly their belief and commitment in the work we do."

## Join the TSA Research Volunteer Network to have a major influence on what TSC research we focus on

The TSA's proud history in TSC research goes back many decades and continues today. Alongside our work in support and information and campaigning, we work closely with the TSC community to ensure that new research into TSC is focused on the things that matter the most to you

Help shape the future of TSC research by joining the TSA Research Volunteer Network. To find out more visit our website or email [research@tuberous-sclerosis.org](mailto:research@tuberous-sclerosis.org)

# You've got this! Be confident and ready for appointments

**Appointments with doctors, teachers or other professionals can be daunting. However, with a few tips and the right preparation you can help make sure that these meetings don't get overwhelming**

It's a scene that many in the TSC community know well. You've been waiting months for an appointment with a TSC professional to discuss a specific topic (perhaps a treatment option, making sure your loved one gets the right support, or something else). After all the anticipation, the meeting is a blur and you leave feeling like you were either unable, or not given the time, to explain your needs. Despite the vital importance of these meetings, it's easy to feel them slip away from you, like sand running through your fingers.

If this has ever happened to you, you might wonder why it happened. You felt prepared and ready, yet the conversation felt stilted or difficult. It's important to not blame yourself or others if you leave a meeting frustrated - in our busy lives, TSC appointments can be extremely challenging and complex, considering all factors that TSC can throw at us.

Feeling prepared is key to building confidence before any appointment. When you know you've done everything you can to get ready, it's easier to walk into that room (or log into that virtual meeting) strong and composed.

## Preparing for TSC appointments

- **Gather documents.** Get all the paperwork or evidence you might need for the appointment, such as medical records, school reports, or a list of medications
- **Choose an outfit you feel confident in.** It may sound simple, but wearing something that makes you feel comfortable can have a big impact on your mindset
- **Know the details.** Be clear on things like the appointment purpose, location, who'll attend and how to get there (even if it's a virtual appointment)
- **Keep notes.** Write everything down that's covered in the meeting, so you don't forget anything later
- **Bring a cheerleader.** Bring someone who can offer emotional support, help you track everything and confirm anything afterwards about what was discussed
- **Practice what you want to say and what you want to achieve.** Rehearse your key points, either to yourself or with a friend. This can help you articulate your thoughts clearly when the time comes





On the day of the appointment, it's normal to feel nervous and that the stakes are high. Keep a cool, calm and collected head, knowing that you've done all of the preparation and that you're allowed to respectfully challenge and check things.

Stay grounded by having your notes with you, including questions or concerns you've got. Your notes are a safety net and a reminder to yourself that you've come prepared, so be sure to refer to them during the meeting. This can help you stay on track and ensure that you don't forget to bring up any important points. It also shows the professionals you're serious and engaged, which can form a feeling of collaboration. As the meeting progresses, try to write down as much as you can. If this is too challenging, politely ask the person you're meeting with if it's ok to record the conversation. The professional might be happy to send you a summary afterwards, although this will be based on their own notes.

As the meeting begins, set out what you want to get out of it. It's easy to get sidetracked as the conversation moves forward. If something isn't clear, don't hesitate to ask for clarification. Remember that this is your opportunity to gather all the information you need. Asking questions isn't just about understanding but also about asserting your right to be fully informed. It's okay to say "I didn't quite understand, could you explain it in a different way?" or "Could we go over that again to make sure I've got it right?"

If the appointment gets overwhelming, it's understandable. After all, these chats are often regarding very important topics. If you start to feel things getting too much, it's okay to ask for a moment to collect your thoughts. You might say, "I just need a moment to think" or "Can I please just take a minute?" Give yourself permission for these pauses to prevent the conversation from becoming too stressful or pressured.

Never forget that you're a key voice in the needs of you or your loved one. You see and experience things everyday that the professional doesn't, which is invaluable for them.

Try to share your perspective, even if it feels challenging. You might say something like, "What I've observed is..." or "In our daily lives we see..." By framing your input this way, you assert your knowledge and make it clear that your experience is a crucial part of the discussion.

As the meeting approaches its end, be sure to clarify next steps. This includes agreeing on specific actions and follow-ups. For example, you might say, "To confirm, the next step is..." or "Could we agree a timeline for when I will hear back about this?" Ensuring that everyone involved knows what to expect moving forward helps prevent misunderstandings and keeps things on track.

With these strategies and tools you can approach your appointments with confidence, knowing that you're well-prepared to navigate them effectively. Also, always remember that the TSA is here to support you if you encounter any challenges in TSC meetings.

# TSC stories: Finding strength after a later diagnosis

**Simon was diagnosed with TSC as an adult and also during a very difficult stage in his life. Although things were tough, support from the TSC community and the TSA has offered a brighter future**

## A sudden collapse

In early December 2014, my life took a dramatic and unexpected turn. What started as an ordinary day quickly changed when I suddenly collapsed at home. I'd been to work, taken my mum shopping, and was sitting in the lounge when everything went blank. When I came to, I found myself confused and disoriented with our neighbour, Bob, smiling reassuringly at me. Mum later told me that I had collapsed "like a sack of spuds," and Bob, hearing her screams from across the road, had sprinted over to help, dragging me into a chair in the sitting room.



**Simon**

## Personal losses, and an epilepsy diagnosis

At the time, I was living with my mother in a large house. Life had already been challenging, as I'd lost my father in a tragic accident and five years later my grandmother passed away. This left just Mum and me in a house that felt far too big. My mother's health had been declining, and I devoted all my free time to caring for her while also holding down a full-time job. Despite the difficulties, Mum was my priority.

Following my collapse, I made an appointment at my local GP surgery. I was referred to Addenbrooke's Hospital, where tests and scans confirmed what I feared: I had epilepsy. This news was a huge shock. After consulting with my GP, I decided to surrender my driving licence, with the hope of reapplying the following year if I remained seizure-free.

For the next year, things were relatively uneventful. My working hours were adjusted, and Mum drove me to and from work. I was looking forward to getting my driving licence back and returning to some semblance of normality. But almost exactly a year later, I experienced another seizure, which led to further tests and scans at Addenbrooke's.

## Discovering TSC

One of the scans I got at this time was an MRI, which was unsettling despite my interest in science. Lying inside the machine, listening to the noise of the massive electromagnets, felt like being in a microwave! After having two MRIs, including one with a radioactive isotope injected into my arm to highlight different areas of my brain, I was contacted by the consultant to discuss the results. I was shown the MRI scans and told that I had a rare genetic condition called Tuberous Sclerosis Complex, which had actually been with me

since birth. The seizures I had, as well as the painful growths on my nails, were symptoms of the condition. It was a lot to take in, especially when I was informed that I would need to be on anti-epileptic medicines for the rest of my life.

The consultant suggested contacting the Tuberous Sclerosis Association (TSA), which I did. Despite their team only being small, the advice they provided was invaluable. I carried on with my daily life and a year later my GP informed me that I could have my driving licence back, as the epilepsy medication was working well.

“Focusing on my own health after years of caring for Mum has been a challenging but essential step in my journey with Tuberous Sclerosis Complex.”

### Coping with loss and finding support

Just before the pandemic, Mum was diagnosed with kidney failure. We'd moved to a smaller house, which made caring for her more manageable as her mobility decreased. In late 2022, Mum passed away peacefully at home. We'd been very close and adjusting to life without her was incredibly difficult.

After a few months, people encouraged me to focus on my own health and future. As part of that, I was advised to contact the TSA again. I hadn't had any more seizures since my diagnosis, and had been taking the prescribed medication for years, but I didn't want to become complacent. Mum's consultant had even expressed an interest in my TSC during her treatment, which made me all the more determined to talk to a specialist again.

After speaking with the TSA, I contacted my GP and got re-referred to the neurology department at our local hospital. There was some confusion over appointment dates and it took a bit of persistence, but the hospital has arranged for me to see the specialist. My appointment is scheduled for early December this year. I'm eager to get a thorough review of my TSC and ensure that everything is being managed and treated as well as possible.

As part of reconnecting with the TSA, I discovered that the charity had an event called Outlook, which is specifically for adults mildly affected by TSC. I'd seen it advertised before, but during the years I was caring for Mum, I never felt I could leave her for long enough to attend. I also didn't think the event really applied to me, since my symptoms were fairly mild in comparison to what others might be experiencing. However, after Mum's passing, I felt a need to do something for myself, something that would connect me with others who understood what I was going through.

My cousin gave me the final push I needed to sign up for Outlook. She said "What have you got to lose, Simon? Think of the people you'll meet, the things you'll learn. You could end up making new friends, new people to stay in touch with!"

### Finding strength in the TSA community

Everyone at Outlook 2024 in Cardiff - a city I'd never visited before - was so welcoming, and it gave me an opportunity to share experiences with others and listen to their stories. I also got to meet Dr. Frances Elmslie, with whom I had a fascinating conversation about TSC. We even discussed how difficult it is to explain TSC to insurance companies, which was something I've had struggles with. Meeting other people with more symptoms of TSC



## SUPPORT AND INFORMATION

than me was humbling - despite their challenges, they're amazing individuals and I hope to remain in touch with many of them. At Outlook I also had the chance to meet the very talented artist Sarah Goy, who herself has TSC. We all had a lot of fun in her art workshop as part of the meeting.

After the success of attending Outlook, I then decided to go to Big Day 2024, taking place in Reading. Big Day is for everyone in the TSC community and it was another incredible event. I met even more people from the TSC community there. The day was filled with interesting speakers, including TSC researchers working on potential future TSC treatments. One of the speakers was the artist Sarah who I met at Outlook. Sarah spoke candidly about her experiences of having TSC. It was inspiring to hear her story, like how she managed to navigate university life while dealing with her TSC symptoms and how she's now a successful artist and lecturer.

Attending TSA events makes me realise how important it is to connect with others going through the same experiences. The sense of community and support is invaluable, giving me strength to focus on my own health and wellbeing. My TSC journey no longer feels isolated and I'm grateful for friendships I've made.

### Looking forward

My TSC has a relatively minor impact on my daily life. The epilepsy is kept under control with medication which allows me to drive - a necessity for my job, as there's no public transport links to where I work. I'm very thankful that the condition hasn't progressed further and that I can continue to live independently.

In the coming years, I plan to attend more TSA events and support the incredible work of the charity. I hope that by sharing my journey, others might find some comfort and encouragement, knowing they're not alone.

“Attending Outlook and Big Day made me realise just how important it is to connect with others who share similar experiences. The sense of community and support has been invaluable.”



Simon at Outlook 2024

I want to thank the TSA team and all Outlookers for making me feel so welcome. I'd also like to express my gratitude to the wider TSC community I've met through the TSA - your stories, strength, and resilience have been a source of inspiration for me.

I wish everyone all the best, whether you've just received a diagnosis or are further along in your TSC journey. Remember that there's a community here to support you, and events like Outlook and Big Day can provide connections and strength to face whatever challenges come your way.

# Get involved in TSC research

**There's always opportunities to help unearth the next big TSC discovery. Two major studies happening soon are focused on TSC-Associated Neuropsychiatric Disorders (TAND) and TSC-related epilepsy**

## Long-awaited TAND clinical trial now open to participants

The highly anticipated clinical trial, EPICOM, is now recruiting across the UK.

The EPICOM study is exploring if there's changes in TSC-Associated Neuropsychiatric Disorders (TAND) when a medicine is taken in addition to other treatments. If the trial is successful, it might mean that the medicine can be used to help with some of the things that are under the TAND umbrella, such as behaviours that can be challenging and anxiety.

If you, or a loved one, with TSC has seizures, you might be eligible to be part of EPICOM. The trial is taking place across the world, including in the UK at Bristol and Sheffield, but there's also a virtual option so you can take part completely remotely!



[epicomstudy.com](https://epicomstudy.com)



[astroscapestudy.com](https://astroscapestudy.com)

## ASTROSCAPE hopes to hit new heights in TSC-related epilepsy

Radiprodil is a potential medicine for TSC-related seizures. A global clinical trial of radiprodil for TSC, called ASTROSCAPE, is going to take place. This is following radiprodil showing promise in helping people with another similar condition.

Radiprodil was found to reduce seizure frequency in almost 3 out of 4 participants who have the genetic condition 'GRIN-related developmental disorder'. In this study, radiprodil was also found to have few side effects.

ASTROSCAPE will take place in the UK. If your child is aged between 6 months and 18 years and has TSC-related epilepsy that isn't managed with at least two anti-seizure medicines, they might be eligible to be part of the study.

# The impact being involved in TSC research

**TSA Ambassador Marie James and her family are long-standing supporters of the TSC community. Their story shows just how important it is for people to participate in TSC research**

The first time Marie, husband Perry and son Trystan took part in research, Trystan's blood was analysed by researchers in a landmark study that helped to identify one of the genes responsible for TSC. This was a huge moment in our understanding of TSC, and one that the TSA was proud to be a major partner in.

Marie comments: "Trystan was seven years old, it was a game-changing day. Everything changed. It felt like research was no longer a bottomless pit, how it felt before then." After this first gene was identified, the second gene responsible for TSC was very quickly discovered. These moments helped progress TSC care to where it is today. Yet, it hasn't always been possible for the family to participate in research projects, because they've sometimes not met participation criteria. For example, Trystan was once excluded from a clinical trial for the medicine everolimus, due to his significant learning disabilities. This is something that the TSA has worked hard to change, as learning difficulties or other challenges shouldn't be a barrier to participating in TSC research.



**Marie and Trystan**

Marie recalls Trystan being unable to participate in this research: "Luckily, Trystan's consultant secured everolimus on compassionate grounds. By this time, his kidney tumour was life-threatening. Unfortunately, this life-saving treatment then sat on a shelf for three months as we campaigned against the Welsh Government who wouldn't commit to funding the medication once the compassionate need supply finished. We felt like we were playing Russian roulette." Trystan's now been taking everolimus for 11 years.

A major way that Perry's participated in TSC research is as a founding member of the TSA research committee, a vital way that we expanded TSC knowledge. Perry was also a patient representative in the TRON trials, which focused on whether a medicine could improve cognitive problems in TSC. The family's lives really have touched so many different areas of TSC research, support and awareness!

Thanks to extensive work of the TSA, the TSC community and TSC clinicians, learning disability is used less as exclusionary criteria in research. Around 1 in 2 people with TSC



**“Trystan is still alive because of the research work that has been undertaken. Continued involvement with anything that we can do to share our experiences within research is really important.”**

have a learning disability, meaning that this is a key step in better trial design and more people than ever before can be part of TSC research.

Marie says that Trystan’s access to everolimus “has been lifesaving as well as life-enhancing.” But, being on everolimus means that it’s harder for Trystan to take part in other clinical trials. However, Marie has continued to participate in a wide range of studies and research projects.

Marie was recently part of the ‘Caregiver experience in TSC (Epidyolex)’ study, where she shared her experiences of Trystan’s behaviour since he began taking Epidyolex (also known as cannabidiol). The study involved completing two questionnaires followed by an interview, which Marie recalls: “It was good to have the interview because I expanded on the benefits and side effects that I couldn’t share through the surveys. All in all, I’ve had a very positive experience, and I would encourage anybody who meets the eligibility criteria to share their experiences as this could open the doors to others with TSC to have access to the medicine, and that’s so important.”

Participants in the caregiver experience study that Marie took part in were reimbursed for their time, which is becoming increasingly common as researchers work to recognise the time and effort involved in participating in research. This is also something that the TSA encourages researchers to do. Marie comments: “When I started to see financial incentives to take part in the research surveys, I felt a bit hesitant. But because of the endorsement of the TSA I felt comfortable getting involved.” Reimbursement is usually in form of something like a voucher or a small payment of another kind.

When people take part in a clinical trial, it sometimes means that they’re given either the new medicine that’s being investigated or a blank medicine that doesn’t do anything, called either a ‘placebo’ or sometimes ‘sugar pill’. Marie is keen to help people understand what this means in practice: “I sometimes think about that Trystan may well have had a placebo drug. However, if you do get started on the placebo, you’re often given access to the actual medicine at the end of the trial.” When someone takes part in a clinical trial, they usually also take the medicine alongside their existing treatments. This means that getting the placebo shouldn’t stop you getting medicines that treat TSC.

TSC research and the development of new ways to diagnose, treat and manage the condition wouldn’t happen without the support of amazing people and families who volunteer to take part, like Marie and Trystan. As well as helping TSC research, participating in trials and projects has benefited Marie in other ways: “Getting involved in research has been empowering and therapeutic. I’ve gotten to know the researchers, and for most studies, you get feedback which can be helpful in understanding and learning more about TSC and your own child and how the one you love is affected.”

Being so involved in research has left Marie extremely thankful: “Trystan is still alive because of the research work that has been undertaken. Continued involvement with anything that we can do to share our experiences within research is really important.”

# TSC stories: How Lucy's gone from strength to strength

**After many ups and downs since her TSC diagnosis at six months old, Lucy now lives a comfortable and active life**

Lucy Marshall was born on 4 March 1994 to Claire and Brian Marshall. Her journey with TSC began early, with infantile spasms at just 10 weeks old. At six months, Lucy was diagnosed with TSC at Queens Medical Centre Nottingham, after skin markings typical in TSC were found on her.

Since Lucy's diagnosis, she's faced numerous challenges. Aged 11, she experienced severe complications from a large SEGA (subependymal giant cell astrocytoma, a growth on the brain). This required brain surgery, followed by an 11-week stay in hospital and shunts inserted. Her teenage years included further surgeries, particularly to manage seizures and other related issues.

A turning point came when Lucy was prescribed everolimus, after lots of campaigning by her family, local MP and the TSA. The medication helped reduce Lucy's SEGA. Lucy has now been on everolimus for eight years and her TSC has stabilised in that time.

Lucy is now 30 years-old and thriving. Her grandmother Mary is very proud of her: "Lucy lives at home with her mum Claire, and with other close family very nearby. She has a busy social and family life, including classes and time with carers, and is enjoying life to the full. We are always prepared for the next crisis but so happy to watch Lucy's current zest for life."

The family has a long history in supporting the work of the TSA. Mary is the president of her local Rotary Club in Grantham. The club chose the TSA as their Charity of the Year for 2024, organising several fundraising events to raise money and awareness for TSC. Recently, the club had a charity golf day, raising £1,000 for the TSA!

Lucy's journey reflects the incredible resilience of those living with TSC and their families. Despite many tough situations because of her TSC, Lucy and her family continue to move forward with hope, and with the TSA by their side from their early days to today.



**Lucy and her family**

# #TeamTSA at the London Marathon 2025

**Introducing you to the incredible superstars taking on the iconic London Marathon in 2025, all to support the TSC community!**

## Aaron Holt

Aaron's son, Jonah, was diagnosed with TSC at six months old: "Although I'm nervous, I'm absolutely over the moon to be running for the TSA!"



**Aaron Holt**

## Rebecca Cave-Brown

Rebecca runs for her six-year-old son, Tommy, who has TSC. Tommy often joins her on training runs in his buggy: "Every time I put my trainers on, it's for the TSA."

## Sam Segrave

Mum to 12-year-old Toby-Jay who has TSC: "I want to run for my son, for those we've lost too soon, and for the families that rely on the TSA's vital work."



**Sam Segrave**

## Daniel Marks

Daniel's daughter Cece was diagnosed with TSC at four months old: "After a few runs, the marathon was the next logical step."

## Daren Little

After 16 attempts, Daren finally got his ballot place: "To say I'm excited is an understatement. I cannot wait to raise as much money as possible for the TSA."

## Kyle Williams

Kyle is running in memory of his daughter, Katie, who had TSC: "The London Marathon has always been on my bucket list. I'm ecstatic to finally have the opportunity."

## Karl Richens

Karl's family member has TSC: "I was ecstatic when I received the marathon email. I can't wait to take part and support the TSA."



**Ollie Law**

## Ollie Law

Oliver's son, Teddy, was diagnosed with TSC at seven weeks old: "I've always said I'd never run a marathon. But I'm excited to see how much money we can raise."

# Fundraising thank yous

We celebrate some of the incredible fundraising taking place that means that we can continue to be here for the TSC community

**Lou Caterall** hosted a charity day in July at the Dolman Theatre in Newport. Lou held the event as her daughter Hallie has TSC. The event also helped Lou's friend Holly get a fundraising boost for her efforts in the Cardiff Half Marathon, raising a brilliant **£1,500!**



**Lou, Hallie and Holly**



**Thank you Jo!**

**Jo Woolley** held a fabulous Ladies Day at Wellington Cricket Club, featuring a three-course meal, live music, and a raffle. The sun shone, spirits were high, and the event raised **£1,586!** Thank you to everyone involved, it looks like it was a lovely day!



**Well done Katie!**

**Katie Morris-Taylor** took on the Birmingham Half Marathon, pushed on by her personal experience of living with TSC. Katie's goal was to support the TSA's work in groundbreaking research and providing essential support to those affected by TSC. Katie did brilliantly, completing the race and raising over **£600!**

Well done Liam Salvidge and Liam Kettle for completing a skydive! In Liam's words "It was an absolutely insane experience. I loved it!" Liam and Liam raised a brilliant **£2,000!**



**Liam, Liam and little Darcie**



**Jade Scott** wanted to show support for the TSA as son Albie has TSC. Jade, her father-in-law Craig and the Pre-War Austin 7 Group in Yorkshire held a charity day, raising a fantastic **£500!** Thank you to everyone involved!



**Pre-war Austin 7 Group**

During lockdown, **Leah Hickson** learnt how to do diamond painting, a special craft art that creates mosaic-like designs. Leah found her passion and over the years accumulated over 20 paintings. When SAI Infinity Care had a fun day, they asked Leah if she'd like a stall to sell her art. From the stall, Leah sold her some of her paintings and raised a fab **£120!** Plus, Leah's planning on trying to organise other ways to sell more in the future! Thank you Leah!



**Leah and her brilliant art**



**Laura and Aaron**

Thank you to the wonderful **Holt family**, who've done an incredible amount of fundraising to improve the lives of people in the TSC community. Laura, Aaron, Ruby and Jonah have done so much, ever since little Jonah was diagnosed with TSC at just a few months old.

You might remember the Holts from our Scan Spring Appeal 2024, as well as other activities like a sponsored hike, charity nights, football tournament and online raffles. Aaron even sorted a charity event through his employer **Coveris!**

We're so thankful to the Holts and their loved ones, who've so far raised over **£12,000!** Their ongoing efforts show how absolutely anything can be made into a fundraiser that'll make a massive difference to people with TSC.

### Corporate giving

Thank you to our recent amazing corporate supporters, including:

- DHL
- Total Movement Solutions
- Precision AQ
- Phoenix Group
- P B Services Wales
- Permission PLC
- J P Gatrell Ltd
- Columbia
- Pro Gaming Ltd
- Copper Ashton Developments
- Coveris
- Foresters Financial Services
- Pavers Foundation

Corporate giving's a great way to get workplaces involved with the TSA. Whether it's a one-time donation, choosing us as 'Charity of the Year' or something else

# Amazing £50,000 donation shows community power

**Foresters Financial donated a jaw-dropping £50,000, all thanks to a committed TSC family**

When Lorraine Boffa saw that Foresters Financial were holding a global competition for charities, she knew she had to get involved. To celebrate their 150th anniversary, Foresters members were asked to submit videos advocating for chosen causes. Alongside her sister Rebecca, niece Ruby and nephew Tommy, Lorraine submitted a video highlighting the impact of TSC. A few weeks later, they were thrilled to find out that they won!

Lorraine shared her gratitude: "Foresters is a fantastic organisation, we couldn't be more grateful. They've got charity and community at the heart of what they do. We'll never take for granted this amazing donation and what it can do for the TSA." Rebecca, whose six-year-old son Tommy has TSC, added: "When Lorraine called me to say we'd won, I was speechless."



**Tommy and Rebecca**

This is a powerful example of how personal connections and talking to lots of different people about TSC can lead to significant support for the TSC community and the TSA. Remember to tell friends, family, colleagues and everyone else about TSC.

Luke Langlands (TSA Joint Chief Executive) commented: "When we found out about this incredible donation and the huge efforts of Lorraine and Rebecca we were all so thrilled. Every donation is important, and this'll help us to significantly progress everything that we do. We're thankful to Foresters for seeing and understanding the impact of TSC."

The cheque presentation took place at Foresters UK's office. Nici Audhlam-Gardiner, Chief Executive Officer of UK Foresters Financial, remarked: "This contribution is an investment in the hope of a future free from the burden of TSC. We are honoured to partner with the TSA in this critical mission."



**At the cheque presentation**

The TSA is deeply grateful for this donation, as well as to Lorraine, Rebecca, Ruby and Tommy for making such an effort in raising understanding of TSC.

# Big Give Christmas Challenge 2024: Double Your Impact!

**As the festive season approaches, we're thrilled to announce that The Big Give Christmas Challenge is back for 2024!**

We're taking part in the UK's biggest match-funding campaign once more between **3 - 10 December**, and we need your help to make it a success. Last year, many of you took part, we hope we can count on you again!

We're hoping to raise £10,000 for our support and information services, which includes our dedicated TSA Support Line - a vital lifeline for many individuals and families in the TSC community.

The demand for our services is high and expected to rise, so your help is more important than ever. The best part? Every donation made during The Big Give Christmas Challenge 2024 is doubled! This means a £25 donation becomes £50, and your impact on the TSC community is twice as powerful with no additional cost to you.

More details will be shared soon. In the meantime, please mark your calendars and help to spread the word. Thank you for your continued support, and we look forward to another successful Big Give Christmas Challenge!

**Christmas  
Challenge**

**BigGive**

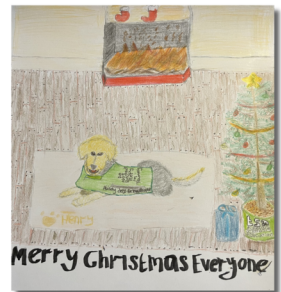
## Christmas cards and other goodies now on sale!

**Our ever-popular range of festive goods are available now, but only while stocks last!**

We've got a lovely range this year, from traditional Christmas illustrations to more modern designs. Our cards also include the winner of this year's design competition, Alison Cracknell!

Alison's design (here on the right) was inspired by her assistant dog, Henry: "He was born at home, and he is everything to me."

Order our full range of festive items by filling in the form that came with this issue of Scan or heading to [charitycardsonline.com/tsa](https://charitycardsonline.com/tsa)



# What you're asking on the TSA Support Line

Recent key questions and answers between us and you



**“My child’s friends are starting to notice that they’re a little bit different - things like TSC-related skin issues. What I can I do to support my child?”**

Although this is a very common and understandable issue in TSC, it’s also something that can be extremely challenging and upsetting. It can sometimes be helpful to frame this as an opportunity to ensure that your child builds a strong feeling of who they are, and that everyone is different - even those without TSC!

Open, honest and patient conversations can be useful. Rather than waiting for friends and others to notice that your child has challenges related to TSC, one option is to be proactive and encourage talking about it. Explain to your child’s friends what TSC is, how your child is affected, and that it’s not a bad thing to be curious. It might be useful to reassure your child and their friends that it’s ok to be different and to ask questions.

Consider talking to your child’s school and organising a simple, age-appropriate session to educate their class about TSC. It might even be an opportunity for other children and their caregivers to share things that they find challenging themselves. This might help to

foster a shared feeling of togetherness across everyone in the class. If your child has siblings who don’t have TSC, you could use this also as a chance to highlight some of their challenges, as we know that siblings of children and adults with TSC can experience major challenges too.

Roleplaying questions and answers might help. You could plan questions like why your child goes to hospital, why they have marks on their face, or why they take medicines. You could practice these with your child too, so they have a ‘bag of answers’ in their head to take from if they’re asked any questions.

Above all, speak often with your child and ensure that they’ve got a strong and valid voice in how their TSC is explained to others. This helps develop trust within the family and shows that everyone’s thoughts and feelings are important and valid.







## “I haven’t been seen by a TSC clinic in many years / ever. Am I too old to be seen by a clinic? What support is out there for me?”

Firstly, you’ve NEVER left it too late to start thinking about TSC. It’s very important to be regularly checked by healthcare professionals so that they can track how TSC is affecting you. However, if you’ve never or only very rarely been seen by TSC specialists, there’s absolutely no reason why you can’t change this.

It’s recommended that everyone with TSC is registered with an NHS TSC Clinic. These clinics are managed by TSC specialist doctors and nurses. All of these clinics are part of something called the NHS TSC Rare Disease Collaborative Network (RDCN), which the TSA is proud to organise and manage. You can find a list of TSC clinics and their contact details on our website or by contacting us on the TSA Support Line. Depending on how TSC affects you, it’s recommended that you’re seen by a TSC clinic roughly once every 6 - 24 months. However, this depends on your individual circumstances.

You might wonder why it’s so important to be registered with a TSC clinic. This is because the doctors and nurses in the clinics ‘speak the language’ of TSC - in other words, they know the condition well and the impact that it can have. The doctors and nurses in TSC clinics are also knowledgeable about the right scans and tests that you need, and can track how TSC is affecting you as time goes by.

### If you haven’t been seen by a TSC clinic in a while, or if you’ve never been referred to one, the steps to getting a referral are usually quite straightforward:

- ✓ Find your local TSC clinic and their contact details (see our website)
- ✓ Explain to your GP that you’d like to be referred to specialist TSC clinicians to monitor your TSC. Give them the details of your local clinic
- ✓ Your GP will then arrange for the referral to happen. You should then later receive a letter with next steps from the clinic itself. Some clinics might see if you’d like to have some of your visits virtually, to make things easier for you

It might take a long time to be seen by the clinic, especially for your first appointment. This is usually because there’s only a small number of doctors and nurses at the clinics - in fact, many of these medical professionals are part of clinics voluntarily or while juggling lots of other commitments. Your patience will be worthwhile as it means that you’ll be then seen by TSC experts.

If you have any challenges at all in getting registered with a TSC clinic, don’t hesitate to get in touch with the TSA Support Line.

There are currently **22 TSC clinics** across the UK. We’re proud to work with these clinics, including as manager of the **TSC Rare Disease Collaborative Network (RDCN)**

The RDCN is a special group recognised by the NHS that connects TSC clinics. It means that **clinics have an easy way to discuss and progress the best ways to diagnose, treat and manage TSC**



**“My child’s teacher / learning assistant wants to know more about TSC but doesn’t know where to start. Can you share resources or tips on explaining TSC to education professionals?”**

If a child has TSC it’s important to make sure that their teachers and school know about the condition. This could be for many reasons depending on how TSC affects them, such as TSC-related seizures, behaviours that can be considered challenging, learning disabilities, or regular time outside of school to attend doctor and hospital appointments.

When educators are keen to learn more about TSC, it’s a great sign that they’re taking your child’s TSC seriously. However, if you find the school unwilling to learn more, consider being firm but non-confrontational, emphasising the necessity for them to better understand TSC, not only for your child but for the wellbeing of the class as a whole. Of course, the TSA is always here to help emphasise to them the importance of TSC training.

The TSA is proud to offer the world’s first and only online TSC training course specifically for teachers, teaching assistants and others in education. We launched the course last year and feedback has been excellent, with the course covering many different areas of TSC. The course is even fully accredited for Continual Professional Development (CPD) points, which is a mark of approval that allows educators to use the course as part of their mandatory annual training and development.

As well as the TSA’s dedicated training course for teaching professionals, there’s lots of information about specific aspects of TSC on our website. You might want to point your child’s school towards the pages focused on how TSC affects your child.

It can sometimes be easy to forget just how complicated TSC is to understand for someone new to the condition, something we all know too well when explaining the condition to someone for the first time. It can be useful to remember this when encouraging your child’s school or teacher to learn more.

If your child has TSC-related epilepsy, you might want to also encourage your child’s teachers to learn basic epilepsy first aid from an organisation such as Epilepsy Action.

Find out more about our training course for education professionals, go to [tuberous-sclerosis.org/education](https://tuberous-sclerosis.org/education).





**“My sibling has TSC. I’m now thinking about starting a family, but TSC is in the back of my mind. What should I tell doctors if I want to start a family? Will my child be at risk of having TSC?”**

Being a sibling of someone with TSC has its own unique challenges. Siblings starting a family might wonder if they should consider TSC for any children they have. The best place to get correct advice in this situation is your sibling’s TSC clinic or your local genetics clinic.

TSC is a genetic condition, meaning there’s a problem with a particular gene. Genes are building blocks of what makes a person, like LEGO bricks put together to make a model.

Around 1 in 3 people with TSC have the condition because they inherited it from one of their parents who also has TSC. The parent with TSC has one altered copy of a TSC gene and one normal copy. This is known as ‘dominant inheritance’. When we have children, they only inherit one copy of each gene from each parent.

If one of your parents has TSC there’s a 50% chance you also have the condition. In that situation, your children also have a 50% chance. You should have an assessment to find out the chance of your children having TSC. You may be offered a genetic test if available in your family. If not, you can arrange for an assessment and scans by a genetic doctor.

In around 2 in every 3 people with TSC, neither parent shows any signs or symptoms of the condition. This means that the alteration in the TSC gene occurred randomly when they were made as a baby. In other words, they have TSC but it wasn’t inherited from Mum or Dad (this is known as ‘de novo’ or ‘sporadic’). This happens as a chance event.

In this situation, the chance of you (a sibling) having TSC is quite small (around 1-2%). If you have no symptoms of TSC it’s even more unlikely you have the condition. If you don’t have TSC then your children can’t inherit it. TSC doesn’t “skip” generations. If genetic testing has been possible in your family, we can confirm that you’re not affected with a blood test. If genetic testing hasn’t been possible then you can be examined by a genetic doctor for any missed signs.

When genetic testing happens in TSC we’re looking for the precise alteration (a bit like a spelling mistake) in the TSC1 or TSC2 gene which caused the condition. The alteration is found in 80-85% of individuals with TSC. Genetic tests are then offered to others in the family to know if they’re affected and what the chance of any future children having TSC is.

In the remaining 15-20% of people, current genetic testing is not able to find the precise cause of their TSC, but it also may be that the testing has just not found the alteration. In this situation we cannot offer a genetic test to other members of the family. However, they can be examined by a genetic doctor and have a baseline round of screening tests, such as scans, to determine if they have the condition or not.

Genetic testing has improved more recently so if genetic testing has not been successful in the past in your family there may be more testing that can be done now.

As this topic is complex, you should ask your GP for a referral to either your sibling’s TSC clinic or your local genetics clinic. You’ll then receive advice for your personal situation.

# The big topic: Making decisions for loved ones

**Many parents and carers make more decisions for their children with TSC than would be expected for those without TSC. This is generally straightforward in childhood, but gets increasingly complicated as their loved one becomes an adult**

Renaissance Legal are experts in advising families on planning for the future. In this piece, Sarah O'Sullivan (Solicitor at Renaissance Legal) answered some of the commonly asked questions that parents and carers in the TSC community often have about how to manage loved ones' personal affairs as they grow older.

## **If my child turns 18, does that mean I can't make decisions on their behalf?**

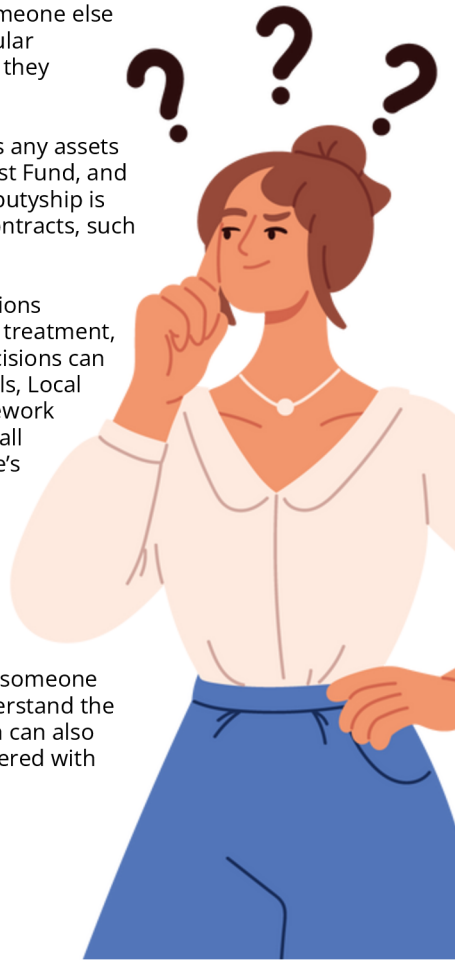
The starting point for making decisions on behalf of someone else is mental capacity. If your child is able to make a particular decision, or participate in the decision-making process, they should be encouraged and supported to do so.

It also depends on the type of decision. If your child has any assets in their own name, such as a bank account or Child Trust Fund, and they lack mental capacity to deal with those, then a Deputyship is likely to be needed. This is also true for entering into contracts, such as a tenancy agreement.

If your child lacks mental capacity and can't make decisions relating to health and welfare matters (such as medical treatment, the care they receive or their education) then these decisions can be made together with the relevant clinical professionals, Local Authority, and social workers. The 'best interests' framework under the Mental Capacity Act 2005 is followed to help all involved objectively agree on your child's needs. If there's a dispute, an application can be made to the Court of Protection to make a specific ruling. You can also submit an application to be appointed as Personal Welfare Deputy for your child.

## **What's the difference between Lasting Power of Attorney and Deputyship?**

A Lasting Power of Attorney (LPA) can only be made by someone who is aged 18 or over and has mental capacity to understand the meaning and power given in the document. The person can also choose those given LPA for themselves. LPAs are registered with the Office of Public Guardian (OPG).





A Deputyship comes from an Order made by the Court of Protection. This is made when a person lacks mental capacity to make decisions for themselves, and also lacks mental capacity to make an LPA.

There's two types of LPAs and Deputyship. One deals with property and finances, and the other deals with health and welfare decisions. It's very common to have the same Attorneys or Deputies listed for both types, although it's possible to have different people making different types of decisions.

### **Will I automatically be appointed as an Attorney or a Deputy for my child? Can anyone be an Attorney or Deputy?**

No, an appointment is not automatic. If your child can make an LPA when they've reached 18, they must independently choose who their Attorneys will be. If they lack mental capacity and need a Deputyship, then you can apply to the Court of Protection to be appointed as their Deputy.

Anyone aged 18 and over who has mental capacity can be appointed as an Attorney, or apply to be a Deputy. However, a person who is bankrupt or under a Debt Relief Order cannot be an Attorney or Deputy for someone's property and financial affairs.

### **Do I have authority to make decisions as a Deputy or Attorney once I've submitted an application or registered?**

The short answer to this is "no". A Deputyship is only in place once the Court of Protection has made an Order appointing you, and an LPA can only be used once it has been registered by the OPG.

This 'best interests' framework can be used for health and welfare decisions while a registered LPA or Deputyship is pending. For example, if a particular decision needs to be made urgently and cannot wait for the Deputyship or the registered LPA (as applicable), then there are some alternative options. This is complex, as the options vary depending on individual circumstances. It's extremely important that you take specialist legal advice in those circumstances to protect you and your child or loved one.



## **About Renaissance Legal**

Renaissance Legal is part of the Renaissance group of companies alongside Renaissance Trust and Renaissance Financial. Together, the group offer legal services, financial services, Inheritance Tax planning, welfare benefits advice and Trusts administration

The expert team at Renaissance Legal can assist you with all aspects of planning for the future, including Wills, Letter of Wishes, Trusts, Powers of Attorney, Estate Administration and Court of Protection work. They can also act as an Attorney or Deputy if required. They will discuss your circumstances and put in place the necessary legal documents which are in the best interests of your family. To discuss this further please call Renaissance Legal on 01273 610611 or email [info@renaissancelegal.co.uk](mailto:info@renaissancelegal.co.uk)

We're here for everyone affected by TSC  
only thanks to the **amazing people and  
groups** who kindly provide us with  
**regular and one-off donations**

Help us to continue to be here for you  
and others, now and the future:

**[tuberous-sclerosis.org/donate](https://tuberous-sclerosis.org/donate)**

**Switchboard:**

0300 222 5737

[admin@tuberous-sclerosis.org](mailto:admin@tuberous-sclerosis.org)

**TSA Support Line:**

0808 801 0700

[support@tuberous-sclerosis.org](mailto:support@tuberous-sclerosis.org)

Webchat ([tuberous-sclerosis.org](https://tuberous-sclerosis.org))

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association