

scan

Spring 2024

The magazine of the **tsa**
tuberous sclerosis
association



**Welcome to
your new Chair**

**TSC and
genetics**

**Come to Big
Day 2024 in
Reading!**

**TeamTSA at
the London
Marathon!**

Here for everyone in the TSC community

Registered charity 1039549 (England & Wales). Registered charity SC042780 (Scotland)

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tuberous-sclerosis.org/donate

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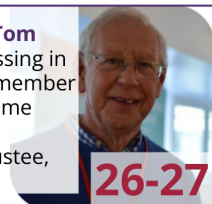
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A word from your Chair

"I'm proud to be the TSA's new Chair. I hope to build on the important progress of the last few years, across all areas of our work"

"Some of you might already recognise me, and I'll certainly know some of you, from my ongoing role as Consultant Clinical Geneticist and TSC Clinic Lead at London's St George's Hospital. At St George's I speak with, support and treat many lovely individuals and families facing a TSC diagnosis.

Although I'm your new Chair, I've been part of the TSA Board of Trustees for some time. As Chair, I'll combine my TSC medical background with board member experience.

It's a time of change for the TSA Board of Trustees. I'm so thankful to departing Chair Sanjay Sethi for his work during what was a very challenging time for our great charity. We're all equally thankful to former Treasurer Rajan Khullar and welcome Simon James to this post.

The entire TSA board, including myself, are still shocked and saddened by the passing of Tom Carter, TSA Lifetime President, in late 2023. Tom can simply never be replaced.



Dr Frances Elmslie

As Chair, I hope to build on the great progress made by Sanjay and the board, and to honour Tom's memory. It's an exciting time for the TSA as we develop new relationships that'll benefit you, the community, for whom we exist. We can only do this thanks to you - **please be sure to continue to support us in all of our efforts.** In return, we remain steadfast in our commitment to you and your families."



Supporting children's mental health

With the demands of school, modern media and also living with TSC, it's important to ensure that you're doing what you can to make mental health an accessible and open topic for children

Approaching mental health challenges in children can feel like a daunting thing to take on. However, by introducing a few simple things into daily life you can help to make mental health a much easier and natural topic.

Before anything else, your child should be registered with their local TSC clinic. This'll mean that any mental health concerns can be supported by the professionals. You can also discuss mental health challenges in children with TSC, and any other topic, through the TSA Support Line.

Acknowledge and validate the child's feelings, as well as your own

Children might not want to show that they're struggling mentally. It then falls to caregivers to recognise possible signs of mental health issues. Flags might include withdrawal, fear, anger, secrecy, difficulty concentrating or signs of self-injury.

Nine out of ten people with TSC will develop TAND (TSC-Associated Neuropsychiatric Disorders). If your child has mental health challenges, recognising it openly helps to reassure them that they don't need to face it alone.

Seeing a child go through mental health challenges can be hard, especially if you're unsure why, or what you can do to help. That's why it's important to share the same compassion for yourself as you have for your children, being honest with yourself that it's a difficult time. However, remember that you and your child aren't alone.

Support your child to express their feelings

Children and young people show their feelings in very different ways. This can be influenced by age, their developmental stage and any learned behaviours that they've got. Emotional expression in a healthy and controlled way is key to regulating our nervous system and restoring balance. This is true for everyone, no matter which additional challenges someone has, like learning difficulties or developmental delay.

Some ways to release stored emotions can include:

1. **Identifying the emotions.** Identifying, naming and acknowledging emotions encourages acceptance and validation which can provide relief. Identifying emotions can be practiced using books or posters. If a child is non-verbal, having picture flashcards they can show you can be helpful - things like different facial expressions, or different colours that represent different feelings. You could cut out the faces on the front cover of this Scan and use this to help
2. **Getting it 'out there'.** This might mean writing feelings down, drawing, or using paints and colours to make marks or scribbles. There's no limit as long as it's done in a safe and controlled manner
3. **Doing a body scan.** Guide and support your child to explore how they're feeling, from the top of their head down to their toes, exploring the different emotions they might be physically feeling. This is also a good way to introduce your child to mindfulness and meditation
4. **Physical release.** 'Throwing the bad feelings away' with a ball, 'Singing the angry out' or any other safe and controlled physical release can be a proactive approach to support the regulation of emotions. It might even be as simple as going for a walk in your local park

Make sharing emotions part of your family routine

Integrating emotional sharing into daily family life can transform the way we connect with each other, fostering a culture of openness and trust. Making this part of the everyday doesn't have to be complex or time-consuming. You might introduce a 'Daily check-in' with family members, sharing how you're feeling and how the day or week is going. You could have a 'Feelings zone' where you all go if you want to share something. Or perhaps a 'Family mood board' to track feelings. Remember that emotion sharing is most impactful when the entire family joins in.

Offer choices and control in different parts of their lives

When experiencing mental health challenges, children might feel out of control and disconnected with themselves and others. It can be a scary and frustrating time. Even though they might not be able to control how they're feeling from one moment to the next, by offering choices in other, practical aspects of life you can empower children by reassuring them that their voice is heard and that their voice matters. Offering choices might mean things like choosing what to wear, deciding whether to participate in an activity, or picking what to eat. However, too many choices can overwhelm, so provide the opportunity for choice sparingly.

Mental health becomes easier to support the more we talk about it

Let's remember the important role that we all play in shaping the mental wellbeing of children and young people. By discussing feelings, encouraging emotional expression, offering choices and creating a supportive environment, we provide a way for us to all navigate mental health and wellbeing in our daily lives.



Researcher in focus:

Dr Amanda Almacellas Barbanøj

TSC research is possible only thanks to the people dedicating their lives to helping us better understand the condition. Amanda is a new TSC researcher, looking for the next big discovery

Dr Barbanøj was recently given a joint fellowship award, meaning that she'll be helping to uncover new discoveries in TSC. She was awarded this by a joint research partnership between the TSA and the Epilepsy Research Institute.

Why is your research different and what do you want to achieve?

"Most treatments for TSC-related epilepsy focus on reducing seizures. They don't address changes happening in the brain itself as it develops.



If someone has TSC, they've got changes in the TSC genes, which are called 'TSC1' and 'TSC2'. TSC1 and TSC2 genes are important for regulating the speed at which the body grows, acting like a brake pedal in a car and ensuring cells don't multiply too quickly. In people with TSC, this brake pedal doesn't work as it should.

My research explores ways that changes in the TSC1 and TSC2 genes affect how the cells in the brain that act as messengers form and connect with each other. The project tries to understand how we can support these messenger brain cells to develop normally. If we research this, it might help us to develop new TSC treatments. My work will also generate a new way to study TSC."

What were you doing before you became a TSC researcher?

"I started by studying biomedical sciences in Barcelona, then moved to the Netherlands for a Masters in cognitive neuroscience. I then did a developmental neuroscience PhD in Italy and moved to University College London to focus on focal cortical dysplasia, a condition similar to TSC."

What was your motivation for getting involved in TSC research?

"One cause of focal cortical dysplasia is a change in something called the mTOR pathway, which also happens in TSC. The mTOR pathway is responsible for how quickly the body grows. When it's not working as it should, TSC-related tumours form. I'm seeing if I can apply the discoveries I made about focal cortical dysplasia into TSC."

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

"I enjoy unwinding with friends and colleagues at the end of the week. I also enjoy drawing and strolling in parks, which I find really relaxing."

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

"I think I'd be a psychologist. I think it'd be a really good fit for me. I've always wanted to help and support people."

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

"I'm delighted to see it focusing more on TSC-Associated Neuropsychiatric Disorders (TAND) and quality of life. I really hope it continues to follow this course."

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

"Thank you to you for the trust placed in researchers. I'm so grateful to be funded by the TSA. Research is very expensive, so when money comes from charities it has a huge and very special value. I'll do everything I can for the TSC community."

“Research is very expensive and when money comes from charities it has a huge and very special value. I'll do everything I can for the TSC community.”



Upcoming TSA events

We continue to offer lots of chances for the TSC community to connect, including face-to-face and virtual events that cover a wide range of topics and discussion points!

Our events for 2024 have already started well, with many more to come. This year, we're also hosting a range of new 'lunchtime drop-in sessions'. These sessions will be more relaxed and will happen during the afternoon. Be sure to come along!

JANUARY

TSC and LAM
With the National LAM
Centre
Thu 18th (virtual)

TSC Art Club
Thu 25th (virtual)

FEBRUARY

TSC and bullying
With BulliesOut
Wed 7th (virtual)

TSC Art Club
Thu 22nd (virtual)

Meet the TSA team
Thu 28th (virtual)
lunchtime drop-in

MARCH

Power of Attorney
With Renaissance Legal
Wed 20th (virtual)

TSC Art Club
Thu 28th (virtual)

JULY

**Supporting siblings of
children and adults
with TSC**

With charity Sibs
Wed 3rd (virtual)

TSC Art Club
Thu 26th (virtual)

AUGUST

TSC and skin
Wed 7th (virtual)

**Adult service
transition**
Wed 14th (virtual)
lunchtime drop-in

TSC Art Club
Thu 22nd (virtual)

SEPTEMBER

**Children's mental
health**
With Bridge the Gap
Wed 18th (virtual)

TSC Art Club
Thu 26th (virtual)

Reasons to come to Outlook 2024 - for adults mildly affected by TSC

Outlook is one of our longest-running events. If you're aged over 18 and mildly affected by TSC, Outlook is a great way to connect with others in a similar situation to you.

Outlook is an easy-going, warm and welcoming environment. Whether it's your first time or you've been to many!

Reasons to come to Big Day 2024 - our biggest event of the year!

Everyone in the TSC community is welcome to Big Day! Share experiences, make new friends and discuss life with TSC. There'll be workshops, research talks and wider chats on life with TSC.

There'll even be a creche with staff specially trained to work with children with disabilities and additional needs.

APRIL

Outlook 2024

Sat 20th (In-person, Cardiff)

How TSC research happens

Wed 17th (virtual
lunchtime drop-in)

MAY

Big Day 2024

Sat 18th (In-person, Reading)

TSC Art Club

Thu 23rd (virtual)

JUNE

All about fundraising

Wed 12th (virtual
lunchtime drop-in)

Transition to adult care

Wed 19th (virtual)

OCTOBER

TSC research update

Wed 9th (virtual)

TSC Art Club

Thu 24th (virtual)

NOVEMBER

TSC and TAND

Wed 13th (virtual)

TSA Annual General Meeting (AGM)

Thu 28th (virtual)

TSC Art Club

Thu 28th (virtual)

DECEMBER

Outlook Festive Special

Sat 14th (virtual)



Results of first study tracking development of infants with TSC

The Early Development in Tuberous Sclerosis (EDiTS) study is proudly funded by the TSA and led by Dr Charlotte Tye, King's College London. The first results of this important study have now been published

The infant years are a critical time for development. Charlotte and the EDiTS team monitored babies and children with TSC to better understand how the condition might affect their overall development and if they had any developmental delays, such as taking longer to reach certain milestones like walking and talking.

This study is important because if we can anticipate development challenges earlier, it might mean we can support children struggling to reach these milestones quicker.

65 infants with and without TSC, aged up to 24 months, participated in the study. Their behaviour was assessed at different times, and caregivers completed questionnaires and participated in interviews.

Charlotte and her team made some important findings:

- At two years old, infants with TSC used fewer words and had a reduction in certain development skills, compared to children of the same age without TSC
- For children with TSC aged up to one year, there wasn't a pattern or connection between a child with TSC using fewer words and if they had severe epilepsy
- If a child aged one year old had TSC and severe epilepsy, there was a strong connection between this and a delay in social skills, problem-solving and motor functions (how the body moves). There was also a delay in development for the first two years of life

These findings highlight the need for early and repeated development assessments of infants before two years of age. It also suggests that by closely monitoring the development of infants, it'll ensure that treatments and wider support can be accessed at the earliest possible time.

These findings are really important, yet the EDiTS study continues. The researchers

are now onto their next phase - the preschool phase - for children aged three - five years old. By understanding how TSC affects development into early childhood, researchers will be able to continue to learn how TSC can affect children as they grow up beyond the newborn and infant phases.

The findings of the EDiTs study so far were recently published in the important research journal 'Developmental Medicine and Child Neurology'. We're very thankful to Charlotte and her entire team!

Researchers tracing former study participants

Did you take part in a large-scale research study between 1980 - 2000?

One of the most ambitious pieces of TSC research in recent times was an 'epidemiological' study into the condition. Epidemiological studies answer some of the most important questions about a condition, like "Why did I get this condition?" and "What will the future hold for me?" These studies help unlock difficult questions about a condition, as epidemiological research aims to track lots of people with the same condition to see how it affects them over time.

131 people with TSC took part in this study, by Dr David Webb and Dr John Osborne. Participants had lots of scans taken and their full medical history shared. This research was hugely important and helped us understand just how common TSC is and how people with TSC might be affected with different symptoms.

The study was a big success, but with some questions unanswered. Now, new funding has given researchers a chance to continue this vital work.

TSC clinicians at Great Ormond Street Hospital and Bath Hospital are now seeking those 131 people who were involved in the original study, to see if they'd like to now help them to continue to uncover more important information about TSC.

The TSA is closely supporting this project, helping to track former participants and to restart the study. If you or a loved one participated in the original study, please get in touch (research@tuberous-sclerosis.org / [0808 801 0700](tel:08088010700)).

TSC Art Club launched

Professional artist Sarah Goy has launched a special virtual art club for the TSC community - a monthly creative wellbeing session focused on a playful approach to art. No matter what your experience or skills in art, you'll be very welcome!

Part of the TSA Together project, TSC Art Club is an opportunity for adults and carers in the TSC community to express themselves creatively in a safe, understanding and empathetic environment.

For many adults and carers in the TSC community, it's extremely difficult to find time for yourself. However, self-care and time to relax and decompress is important for your long-term health and wellbeing. TSC Art Club gives you this space, not only to explore a fun new skill but to also experiment and interact with art in a way that can benefit you in many different ways.

It doesn't matter if you're already a skilled artist or are just curious about meeting others in the TSC community - Sarah and the TSC Art Club will welcome you. The sessions are about taking time out, meeting other people and learning a few creative tips and ways of thinking along the way.

You don't need lots of art materials or need to do lots of planning. All you need is a sketchbook or sketchpad of some sort, and any other mediums that you can gather - whether this is a pen, pencil or other things like paints and crayons.

Sarah Goy commented on the TSC Art Club: "I'm an artist but I also have TSC. Something that's really got me through many challenges is art, which is why I wanted to launch an art club for the TSC community."

SUPPORT AND INFORMATION



This is aimed at adults with TSC, or carers who want some time out. It's going to be on the fourth Thursday of every month. It's completely free, but if you can donate to the TSA that would be amazing.

If you're thinking 'I'm not arty and don't know how to draw or paint', don't worry! Everyone can come. It's about creating, chatting and connecting, because quite often TSC can be quite lonely and sometimes people outside of the TSC community don't understand it."

To sign up to the TSC Art Club, visit the TSA website or email support@tuberous-sclerosis.org for more information

About Sarah Goy

Sarah is a professional artist, based in Bournemouth. She's a visiting lecturer at Arts University Bournemouth (AUB) – BA and MA Animation.

Running creative online art wellbeing sessions has been a passion of Sarah's for many years. These sessions encourage a sense of play through which new skills are gained, whilst encouraging a sense of community and inclusivity.

TSA Together is a pilot project by the TSA, giving people in the TSC community the chance to organise and run their own virtual and face-to-face events



Your TeamTSA London Marathon 2024 team!

The London Marathon is one of the biggest community fundraising events of the year for the TSA. Here you can get to know some of our 2024 runners a little bit more, like why they're taking on the 26.2 miles to improve the lives of people with TSC

Martin Ready took on the Great North Run and Milton Keynes Half Marathon in 2023. Now he's inspired to do the full marathon in support of daughter Amy, diagnosed with TSC at nine months old. Thanks Martin!



Dwain

Dwain Reynolds and his son both have TSC. Dwain never dreamed that he'd take part in a marathon after being on crutches for two years due to a brain injury from epileptic activity. Only getting his mobility back in 2019, Dwain is determined and looking forward to the event! Brill!



Martin and Amy

Megan Salvidge is pushed on by little daughter Darcie, who is 15 months old and was diagnosed with TSC at just five months old. A keen runner, Megan has taken part in lots of events with her family and friends to raise funds and awareness for the TSA! Great work Megan!



Megan and Darcie

Stephen Keeble is heading to the London Marathon after being lucky with a place through the public ballot! Having been unsuccessful previously, Stephen's patience finally paid off and he's very kindly chosen to raise awareness and funds for the TSA. Great stuff!

Isabella Dohil completed the San Fransisco Marathon in 2021. She's since moved to London and learned all about TSC from her friends Lucinda and Tony, whose son Lucas has TSC. Isabella wanted to support her friends and raise funds and awareness, so signed up for one of our charity places! Well done Isabella!



Isabella

Robert Manchett always wanted to take part in the London Marathon. After his friend Lewis Bastin ran the marathon in 2023 for the TSA, he decided to take part to support his friend's sister, Chloe, who has TSC. Thank you for taking on the challenge Robert!



Robert

Jamie Wareing has already taken part in lots of events to support the TSA and people with TSC. He's now going for his London Marathon hat trick this year! He's motivated to take on the challenge again to support his seven year old daughter Elsie, who has TSC. We're so thankful to you Jamie!

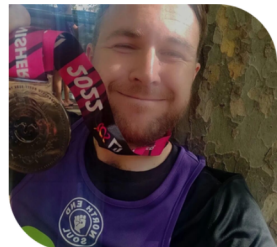


Go Jamie go!

Inspired by our amazing London Marathon 2024 team?

There's lots of ways to get involved in TSA challenge event fundraising, whatever your fitness levels!

Head to tuberous-sclerosis.org and 'Take part in a fundraising event' or contact fundraising@tuberous-sclerosis.org





TSC Stories: Darian Thomas

When Darian's little girl Hallie was diagnosed with TSC, the family entered an unfamiliar world. By connecting with others in the TSC community, the family found their feet once more

"Our daughter Hallie has TSC, which we found out when she was two months old. Before the diagnosis, we had no indications that there were any conditions affecting her. Before Hallie was born, the ultrasounds had been normal and there were no concerns after she was born either.

It all started when we noticed that Hallie's arm was twitching, coming and going throughout the day. It slowly progressed to the point that her whole body was twitching - this is when we went to the Grange Hospital in Cwmbran, Wales.

Hallie was taken to a high dependency ward and it was a long two weeks of no sleep, trying to control what we then found out were seizures which were



happening at least twice every 30 minutes. Hallie had lots of tests – an MRI, ultrasound brain scan and an electrocardiogram (ECG). We were then told that she has TSC. Her TSC was causing epilepsy and lymphangectasia (a thickening in her intestines, causing it to leak out important proteins into her abdomen).

We were then moved to Cardiff Children's Hospital where we met Hallie's new specialist clinicians. Hallie was on so many different medicines throughout the day and night, as well as having baby formula every three hours on the dot. The biggest gap we had in the day was when the doctors would come in to talk to us! We ended up writing down Hallie's timetable on a hospital coffee cup because there was no way we could remember it all.

After another month in hospital, we were discharged. The neurologist told us to look out for infantile spasms (seizures) and that they'd show by six months old. We thought we'd got away with no spasms, but at five months and three weeks they started. We've been very lucky that Hallie has responded well to the seizure medicines she's been put on and we've been seizure free for over a year now.

We attended the TSA Big Day in 2023. It was really nice to see and talk to others who know what you're going through. The biggest struggle is not knowing how the condition is going to affect Hallie in the long run, but talking to others who've been through what we're going through really helps.

While at Big Day, we spoke to the TSA and mentioned that we live and work in a theatre and are happy to host events for the TSA. The theatre committee chose to fundraise for the TSA during one of their productions and the TSA sent us a fundraising kit and information for the audience

“The biggest struggle for us is not knowing how the condition is going to affect Hallie in the long run, but talking to other parents who've been through what we're going through really helps”

to learn about TSC, which really helped to draw attention and raise awareness of the condition. The theatre managed to raise over £400 during the week of the play.

and monthly blood tests to keep an eye on her levels. She's recently had her yearly scans and although the tumours are growing the doctors are still happy with how she's doing. Hallie started walking a couple of weeks ago and is a very confident little girl who has a brilliant personality and sense of humour.

We're so thankful to Darian and his family for their fundraising and for sharing their story. You too can connect with others in the TSC community - join us at virtual and face-to-face events!



Hallie's still on a fair bit of medication, but we're down to twice a day which is a world away from where we started! She has infusions twice a week at home

Play the weekly
TSA Lottery for
your chance to
win **£25,000!**

From just £1 a week you can have the
chance of winning it big, all while helping
to improve the lives of people with TSC

tuberous-sclerosis.org/tsa-lottery

Full terms, conditions and information on self-exclusion available on the TSA website. Responsible gambling means not spending more money or time than we can reasonably afford on gambling activities, keeping in mind all our other responsibilities in life.

GambleAware



Regular giving's big impact

One of the best ways you can support us is by making a regular donation, helping us to focus on improving the lives of people with TSC

We're thankful to everyone who gives to the TSA, including regular givers. Although all donations and fundraising efforts are hugely appreciated, regular giving provides stability and sustainability. We can then focus on the things that matter the most.

Thanks to regular donors, we can plan for the future and be more confident in the long-term health of the charity. This means that more time is spent on support and information, research and advocacy, rather than on our survival.

To set up a regular donation, head to tuberous-sclerosis.org/donate, contact our fundraising team (0300 222 5737), or talk to your bank.



Leaving a gift in your Will

Thinking of the TSA and the TSC community in your Will is a powerful way to leave a lasting legacy that improves the lives of people in the TSC community

Everything that we do at the TSA is fuelled by kind individuals and organisations who choose to donate or fundraise for us. One of the most powerful ways that this happens is when someone chooses to consider us in their Will for when they're no longer around.

Leaving a charity gift in a Will is becoming more popular, with the number of adults aged 40 or older who do this rising by 43% over the last decade.

It's an incredibly challenging time for charities like the TSA right now. Whether it's a small or large amount, by thinking about the TSA and TSC community in your Will, you help to ensure that our work across support and information, research and advocacy can continue in the future.

The significance of gifts in Wills is never underestimated by the TSA. We understand that considering whether to leave a gift in your Will is a deeply personal decision, which is why any information we're given is treated with the utmost care and respect. The TSA is committed to supporting you every step of the way and you can be assured that all legacy gifts are used in a way that'll have the greatest impact on the work of the charity.

We've partnered with the professional Will writing service Kwil to make the whole process as easy as possible

Visit www.Kwil.co.uk, enter the code 'THETSA' and you can write your Will completely for free

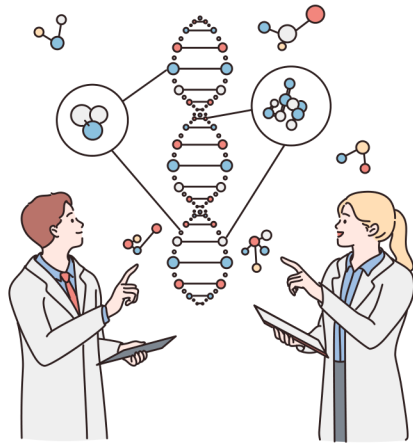
When the time is right for you to draft your Will, and after you've taken care of family and friends, please consider the TSA. If you've got any questions about thinking about the TSA in your Will, please contact Sara Eltman (Head of Fundraising): Sara.Eltman@tuberous-sclerosis.org / 0300 222 5737.



Upskill your knowledge: TSC and genetics

Our website is packed with information about TSC and we work hard to keep it all up-to-date. We recently updated our page on genetics, giving you a solid understanding of genes and their role in TSC

TSC is a genetic condition, meaning it's caused by a change in a person's genes – the instruction manual that determines how we are made up. Everyone who has TSC was born with the condition. A person might have TSC because a change in a gene responsible for TSC was passed down (inherited) from one of their parents. However, sometimes a child has TSC and their parents don't. When someone's diagnosed with TSC, they and their family are usually offered genetic testing to see if TSC runs in the family.



What are genes?

Genes are made up of DNA. The instructions for the body on how it should grow and look after itself are given by our genes. This includes everything about our body, from the colour of eyes and hair to how organs act and work together.

Our genes are given to us by our parents. That's why families might carry certain traits, like ginger hair.

Each of us has thousands of genes and each gene is responsible for different, very specific, things. If something goes wrong in a person's genes, the instruction to the body from that gene doesn't work as it should. When a gene doesn't act as we expect it's known as a 'mutation' - however, it's more often now also referred to as a 'pathogenic variant'.

How do genetic conditions happen?

TSC is a genetic condition. There are many other different genetic conditions, including cystic fibrosis, muscular dystrophy and haemophilia.

When there's an unexpected change ('mutation' or 'pathogenic variant') in a gene, it's no longer able to instruct the body as it normally would. This causes disruption to the body's normal development and functioning. This is what causes genetic conditions like TSC.

Which genes cause TSC?

TSC is caused by mutations in two genes, called TSC1 and TSC2. The TSA is proud of the role we played in the research that uncovered these two genes. All genes come in pairs, including the genes responsible for TSC. One gene in the pair is passed down by the mother, the other gene in the pair is inherited by the father.

In some genetic conditions, including TSC, a mutation in only one gene (i.e. TSC1 or TSC2) will cause TSC. When you or a loved one were diagnosed with TSC, you might have been told that the mutation happened in the 'TSC1' gene or 'TSC2' gene. Generally, but not always, people with a mutation of the TSC2 gene are more impacted by TSC than those with the TSC1 gene mutation.

We get ('inherit') our genes from our parents, but it's possible to have a genetic mutation with no family history of the condition. This is true in TSC, with some people diagnosed when there's no background or diagnosis of TSC in the family.

Did everyone with TSC get the condition from their mum or dad?

One in every three people who has TSC inherited the condition from a parent with the condition. If a person has TSC, there's a 1 in 2 chance in every pregnancy they have that their child will also have TSC. This is because genes are handed down to a child in pairs – one from the mother and one from the father. The gender of the parent that has TSC and the gender of the child doesn't change these odds.

In two in every three people who have TSC, neither of their parents show any signs or symptoms of also having TSC. This means that the change to one of the genes responsible for TSC was random (known as a 'de novo mutation') and not inherited.

In around 1 in 50 TSC cases, couples have a child with non-inherited TSC and then have another child that also has TSC. This is called 'mosaicism' and occurs because the genetic mutation that leads to TSC isn't in all cells of the parents' body.

Want to know anything else about TSC and genes? Be sure to get in touch with us through the TSA Support Line!

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

Superstar **Sarah Bell** raised an amazing **£2,270** by taking on the epic challenge of running a full marathon every month throughout 2023! Sarah's inspiration is her daughter Emily, who was diagnosed with TSC at just four months old.



Sarah Bell

A massive thank you to **Paul Maywood** who took on the Great South Run last year, raising a fab **£1,157!** Paul, a Parkrun regular, plans to keep on running for the TSA during 2024!



Paul Maywood

The amazing **Rebecca Cave-Brown** organised a brilliant charity quiz night with her family and friends and raised a whopping **£2,675**. Well done to her fab little helpers too: Jamie, Zachary, Ruby, Enzo, Leonardo, and Tommy!



Rebecca (middle) with Tommy (left)

Linda Smith continues her fantastic fundraising efforts! Her incredible supporters at The Limes Hotel raised a further **£900** in January and February, with activities including a recent bingo night and raffles. Brilliant!



Linda Smith

The Big Give Christmas Challenge 2023

Thank you to all of our amazing supporters who donated to our **Big Give Christmas Challenge 2023** campaign at the end of 2023. Thanks to you, we raised a fantastic **£10,500!**

Your help means that we can continue to operate the **TSA Support Fund** throughout 2024, giving those in the TSC community at financial crisis desperately needed support.

**Christmas
Challenge**

BigGive

Ed Hildebrand ran the Royal Parks Half Marathon last year and raised a fantastic **£2,743!** On top of his incredible fundraising efforts, Ed is also a research student working hard on new discoveries in TSC-related epilepsy.



Ed Hildebrand

Facebook and Instagram fundraising

Thank you to everyone who recently setup or supported **Facebook or Instagram fundraisers**, with occasions such as birthdays, and anniversaries used to raise funds to improve lives in the TSC community.

In 2023, Facebook and Instagram fundraisers raised over **£7,000!** for the TSA! Be sure to have your own Facebook or Instagram fundraiser for your own special occasion – it only takes a couple of minutes to setup!



FUNDRAISING

The Holts - an inspirational family!

We've recently been blessed to receive incredible support from Laura, Aaron, Jonah and Ruby Holt plus their wider family. Following diagnosis of little Jonah with TSC, the Holts have focused their efforts on improving lives in the TSC community.



Ruby, Laura, Jonah and Aaron

They organised a 14-mile hike with family and friends, gathering over **£4,000!** The family are even planning more fundraising events, with Aaron taking on ten 10k runs, a half marathon, and the Leeds to Liverpool canal walk during 2024. Incredible!



The Holt family and friends during their recent fundraising challenge!

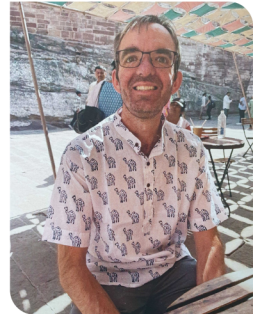
In memory and tribute giving

Our sincere condolences and thank yous go to the family and friends who chose to give in memory of loved ones who recently passed away. This includes: **Tom Carter**, **Gillian Vaughan**, **Joseph Hickson**, **Anne Cole** and **Peter Noble**.

In memory and tribute giving is a heartfelt way to give to the TSA, helping us to continue to be here for the TSC community now and forever across support and information, research and advocacy.

Another in memory donation we received recently was from the loved ones of **Andy Wellington**, a very popular member of the TSA's Outlook group for adults mildly affected by TSC. Andy was a passionate fundraiser for the TSA and will be very missed.

Organising in memory giving is straightforward and free using **MuchLoved.com**, where you can setup donations and even create a tribute page to your loved one.



Andy Wellington

Corporate giving

Thank you to our recent amazing corporate supporters, including:

- Equistone Partners Europe
- GSC Grays - Alnwick
- Kennedy Hygiene Services

Corporate fundraising is an amazing way to get your workplace involved in improving the lives of people with TSC.

Whether it's a one-time donation, choosing us as your 'Charity of the Year', payroll giving or something completely different. Every contribution goes a long way in our work!

Trusts and foundations

Thank you to all of the trusts and foundations who recently supported us, we are very grateful. This includes:

- The Masterserve Foundation
- The James Dyson Foundation
- The Rest Harrow Trust
- AJ & KN Barnett Charitable Trust

THANK
YOU!

Remembering Tom Carter

It was with great shock and sadness at the end of 2023 that the TSA announced the passing of Tom Carter, TSA Trustee and Lifetime President, following a short illness. The TSA now pays tribute to Tom

Tom was a vital part of the TSA, from the charity's early years right to his passing. Tom held many important positions in the charity and can be credited with being one of the reasons that the TSA developed into the charity that it is today.

Born in 1947, Tom spent his life in the Nottingham area. Tom trained and worked in textiles, going on to achieve a Business Management degree from Nottingham Polytechnic (now Nottingham Trent University). Tom met Anne, his wife of over 50 years, when both were teenagers. They married in 1970 and in 1977 welcomed daughter Sally-Anne. In the early months of Sally-Anne's life, she was diagnosed with TSC. This life-changing moment – one that parents and caregivers in the TSC community know all too well – put Tom and Anne onto a very different life path than they had expected.

After Sally-Anne's diagnosis, Tom and Anne were determined to make sure that she and others with TSC receive the care and support they deserve. Tom and Anne were part of the original group of TSC families that pushed support, awareness, treatment and management of the condition to levels never previously seen.

In 1978, just a few short months after the official launch of the TSA, Tom was welcomed to the TSA's Board of Trustees. This was a role that Tom would hold for the next 45 years. Anne joined the TSA as Appeals and Publicity Officer one year later. Tom became Chair of the TSA Board of Trustees in 1987, then becoming President in 1992. The TSA was then proud to award Tom with the title of Lifetime President, a role he held until his passing.



Over the following decades Tom and Anne continued to be central to the TSA, including raising significant funds. This had a life-changing and life-saving impact on thousands in the TSC community.

As well as his focus on TSA fundraising and TSC awareness-raising, Tom was a strong TSC campaigner - especially in education and the rights of young people. Ensuring holistic support and campaigning is something that the TSA continues to do, with our focus on all areas of life with TSC largely in part thanks to Tom's early work.

Tom's influence even spread to the global TSC cause, with Tom being a strong ally of E-TSC - the European-wide umbrella organisation for all TSC groups.

Tom achieved so much 'behind the scenes' for the TSA, but also did possibly even more as a public face of the TSA and TSC community. Many people knew Tom after speaking with him at TSA events. Tom would remember someone even if years had passed. This embodies the spirit of the TSC community being a big family, which the TSA embraces.

Tom offered support to everyone in the TSC community through a combination of warmth and humour. Tom would help people reconsider situations that felt impossible – perhaps a recent TSC diagnosis, a challenging time in treatment management, or something else – into ones that felt achievable or manageable.

In later years, Tom remained steadfast and important to the TSA team and Board of Trustees. During difficult times, Tom was on-hand to provide direction, structure and passion, giving the team the belief and courage to conquer all challenges.

Tom will continue to be a major influence on the charity, although there is now a gap left by his passing that will never be filled. Tom's massive contribution was always acknowledged, but it is now with his absence that it is so keenly felt.

“I will remember meeting Tom for the first time in 1987. I was already in awe of Tom and his wife Anne's incredible fundraising and awareness efforts for the TSA.

Throughout his time in the TSC community, Tom was friendly, approachable and had a special ability to put people at ease. His sense of responsibility, commitment and concern for others with TSC and their families along with the awareness and fundraising, led to involvement in many TSC 'firsts'.

We owe a huge debt of gratitude and thanks to Tom, and of course Anne, solid at his side, for the lifelong commitment to supporting others through their undaunted work for the TSA and far beyond.

- Marie James, TSA Ambassador

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

Switchboard:

0300 222 5737

admin@tuberous-sclerosis.org

TSA Support Line:

0808 801 0700

support@tuberous-sclerosis.org

Webchat (tuberous-sclerosis.org)

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