

**scan**UK  
magazine  
The magazine of the TSA

**t**sa  
tuberous sclerosis  
association

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*"We have to make  
sure that rare diseases  
in general and TSC in  
particular, do not  
continue to be short  
changed"*

**DAVID SUCHET**



**TAKING OUR #FIGHT4TREATMENT TO PARLIAMENT**

Mental Health & Wellbeing | Funded Research Projects | RESCOM | TSA BIG DAY  
Scottish & Welsh Days | Fundraising Gallery | 40th Anniversary | Campaigning

## A note from our Editor

As we all notice our daylight hours shrinking and the cold nights drawing in, many of us can get those 'winter blues'. Mental health problems are common amongst the general population, with latest statistics revealing that 1 in 4 of us suffer from some form of mental health issue each year. For many people who are affected by TSC, juggling work, caring and medical appointments can all have an impact on mental wellbeing. In the four years I have worked for the TSA, I have continued to be astounded by the strength of character shown within the TSC community. In this edition I'd like to pay tribute to those who feature in our Mental Wellbeing feature and share their stories with the SCAN readership. It takes great courage to talk about who we are at our most vulnerable. We hope that their stories go some way to breaking down the stigma that continues to surround this subject.

And, we haven't stopped there, news and resources from our recent TSA events: TSA Big Day, Scottish Get Together and Welsh Info and Fun Day. And, with Information on Wills and Trusts from Irwin Mitchell and new TSC research projects funded by the TSA, there's plenty to digest in this edition. Pull up a pew and get stuck in. 🍂

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## From our Chairman & Chief Executive

Anniversaries are a great opportunity for reflection, celebration and planning ahead. And as we enter our 40th year as a registered charity, we look forward to doing all three of these things! It was good to meet many new faces at this year's TSA Big Day in Nottingham - one of our most energising events to date. We are planning an even bigger event to celebrate our 40th anniversary next year and hope that you will be able to join us.



Looking to the future it is really important that we are able to continue supporting TSC research and our strategic aim TSCure. One way we hope to be able to sustain our research efforts is by launching a new regular giving programme. Having abolished our annual membership fee we will be inviting our members to commit to a regular gift. Regular small gifts from our community are so very valuable because they add up to a regular and dependable income. This predictability in income will help us plan and deliver our work more effectively.

We hope you will join in with our celebrations across the year as we highlight 40 member stories using a variety of media, old and new. We're also putting together 40-themed fundraising ideas including a 'virtual 40-mile' event. Why not make 2017 special by getting involved in our run and earning yourself a special edition 40th anniversary TSA medal?

Meanwhile, still in 2016, a huge thank you to everyone who has taken part in our recently closed Community Survey. This will help us to review our aims and the allocation of our resources at the mid-point of our five-year strategy. Understanding your experiences and your views will mean that we can continue to place people affected by TSC at the heart of everything we do.

As 2016 draws to a close we find ourselves continuing to campaign for access to treatment in England. We now have a policy enabling Everolimus to be prescribed for the AML kidney tumours associated with TSC. As this edition of SCAN goes to press we are waiting for a decision from NHS England. We hope that they will make the right one and fund Everolimus for inoperable TSC-related brain tumours (SEGA) To the 94 of you who have written to your MPs about this and to the 17,000 who signed our e-petition, to everyone who shared your story - a massive thank you. Together, we will continue to #Fight4treatment. 🍂

**Martin Short** Chairman 🐦 @MartininStow

**Jayne Spink** CEO 🐦 @Jayne451

# Mental Wellbeing and TSC

**We are used to looking after our physical health over the winter months, trying to avoid getting coughs and colds and staying warm and dry when we're out and about, but what about our mental health?**

The subject of mental health can stir up strong emotions. Despite the best efforts of mental health charities, stigma surrounding mental health issues still exists. Such stigma may lead to the feeling that mental health should not be talked about, but we know this is not the case. Acknowledging the challenges and problems that TSC presents to a person's mental wellbeing in no way negates the courage that is so evident in everyone living with this condition, whether they have TSC themselves, or are caring for someone with TSC.

Having a long-term health problem can be very challenging. It can raise issues such as the feeling of being different from others, concerns about one's ability and dealing with the perceptions of others. Additionally decisions about your care may be strongly influenced by the views or wishes of others, such as health care professionals, family members and carers, which can feel disempowering. It is important that we recognise these issues, while also recognising that there are things that people with TSC can do to enhance their wellbeing.

The difficulties associated with TSC means that some people with this condition require long-term care and support. Providing such support for anyone with complex needs can be tough. Caring can at times feel relentless, presenting both emotional and practical challenges. There may be uncertainty, fear, frustration, guilt and anger. It may be physically demanding, time consuming and have financial and social implications. The combined effects of these issues present a challenge to even the most robust person's mental wellbeing.

We also know, however, that there are many positive emotions associated with caring and positive outcomes that come from supporting another person. Self-care and effective support from others is likely to make it easier to identify the positives associated with caring and to balance these against the difficulties.

## Knowing the facts



A recent article entitled Tuberos Sclerosis Associated Neuropsychiatric Disorders (TAND) and the TAND Checklist, published in Pediatric Neurology (2015, Edition 52) states: 'The most common psychiatric disorders observed in association with TSC include neurodevelopmental disorders such as autism spectrum disorders (25%-50% of people with TSC

affected) and attention deficit hyperactivity disorder (ADHD, 30%-50%), as well as depressive and anxiety disorders (30%-60%).'

Furthermore a recent article entitled Caring for Children With Tuberos Sclerosis Complex: What Is the Physical and Mental Health Impact on Caregivers? published by The Journal of Child Neurology (2015) examined the stress levels experienced by people who care for people with TSC. In it, author Anne M Rhentz and her colleagues, say:

'A recent study, by Riley Hospital for Children in Indianapolis, USA, investigated stress that parents of children with tuberous sclerosis complex experience. The study reported that almost half of the parents suffered from clinically significant stress levels.'

Results of the same study indicated that 42% of the 275 people who took part in this piece of research had mild to severe depressive symptoms with more than 19% indicating that they regularly took psychotropic or anti-depressant medication.

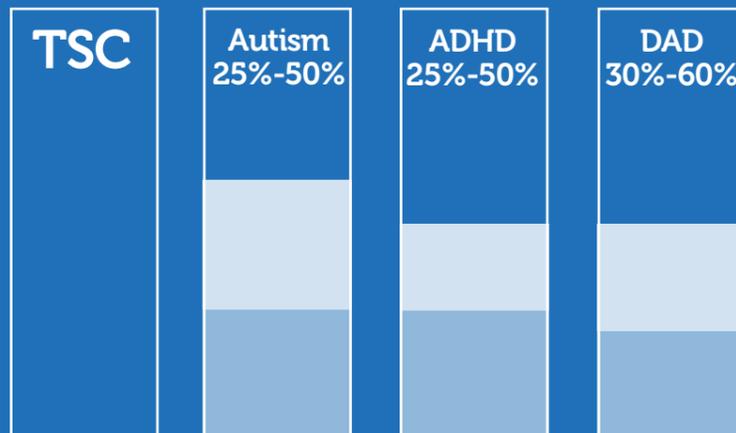
Anne M Rhentz also says: 'Caregivers are taking care of day-to-day needs of tuberous sclerosis complex patients throughout a patient's lifetime, resulting in considerable physical and psychosocial burden over the caregiver life course. Compared to healthy adults in the US population, tuberous sclerosis complex caregivers have

significantly worse physical and mental health and more depressive symptomatology... Our study data seems to indicate that the physical and mental health burden on the caregiver may compound as tuberous sclerosis complex patients age.'

Poorer mental wellbeing is not inevitable, however. For example, from the studies above 58% of caregivers did not experience depressive symptoms and half of caregivers did not experience elevated stress. Further, where someone does experience mental health problems or poor mental wellbeing, there are sources of support and resources that they can access to help alleviate this. Bringing this all together, it is clear this is a subject we should be talking about. This was reinforced visibly when we put out an appeal on our Facebook page [www.facebook.com/thetsauk](http://www.facebook.com/thetsauk) asking if anyone would be prepared to be interviewed for this magazine on the subject of mental health. Within minutes people had come forward, ready to share their experiences and tell their stories, all in the hope of helping others.

So a huge thank you to everyone who came forward to give their experiences of living with TSC and offering to share their stories in the hope of helping others. And apologies to those of you who we have not featured on the pages of this magazine, this is simply down to a lack of space, and where possible we will publish your stories on the blog pages of our website [www.tuberous-sclerosis.org/blog](http://www.tuberous-sclerosis.org/blog)

### The most common psychiatric disorders seen with TSC



### Key symptoms of the winter blues:

- Depression
- Sleep problems
- Lack of energy and enthusiasm
- Overeating
- Irritability
- Feeling down and unsociable

**Did you know that around 2 million people in the UK are thought to be affected by Seasonal Affective Disorder (SAD), or, the winter blues?**

## Top tips for helping to beat the winter blues:



### Keep active

Going for a walk in daylight can lift your mood and help you to keep fit. Even a 20 minute walk will make a positive difference.



### Listen to music

Research shows that listening to upbeat, cheerful music improves mood, in both the short and long term.



### Keep warm

Being cold can contribute to depression and low mood. Wear warm clothes and aim to keep your home between 18C and 21C (64F and 70F degrees).



### Eat smarter

A healthy diet can boost your mood and energy levels. The NHS recommends balancing carbohydrates with plenty of fruit and veg.



### Make the most of the daylight

Open curtains and blinds and sit closer to windows. Some people use light therapy but this is not available on the NHS. Further details can be found on SADA (Seasonal Affective Disorder Association) website: [www.sad.org.uk](http://www.sad.org.uk)



### Hobbies and interests

Having a hobby, volunteering or trying new interests helps keep our minds active and helps to reduce symptoms of low mood, stress and anxiety.



### Seeing friends and family

Socialising is great for your mental health so keep in touch with people you care about and accept invitations to see others, wherever possible, especially if you are feeling low. Ask family and friends to keep on encouraging you to join them, even if you initially resist.



### Join a support group

Sharing your feelings and experiences with people who understand and have experienced the same symptoms can be helpful.



### Talking treatments

If your symptoms are difficult to manage, discuss with your GP what treatments and support are available to you on the NHS. These may include counselling, psychotherapy and cognitive behavioural therapy (CBT).

See page 9 for a list of organisations that may be able to help.

# Mental Health, a family's story

Freddie Norton-Usher is now a very lively 2 and a half year old, but a little over a year ago the Norton-Usher family's world was turned upside down.

'Freddie was absolutely fine, completely normal, until one day when, out of the blue, he went limp, completely floppy for about 30 – 40 seconds. We did not know what was going on. It was terrifying and we thought Freddie had died.' said Lottie.

In that instant Lottie and her partner Ed's world changed forever. An ambulance ride to Brighton General and 8 months of tests later, and Freddie was diagnosed with TSC.

'It was a horrific year for us,' says Lottie, 39, 'having to deal with the knowledge that Freddie has TSC, and deal with his symptoms – the seizures, his increasingly challenging behaviour, and on top of all that we found ourselves facing homelessness.'

'Looking back now I can see that we spent that year in shock. I had just set up as a self-employed graphic designer, but was not earning much as I had to try to fit it in around Freddie, so on just one salary, we could no longer afford to live in Brighton, where we had lived for years, where our whole support network was. I can't describe how awful I felt for so much of the time, so isolated, scared, and hopeless.'



'After seeing the posts about the #Fight4Treatment campaign on the TSA's Facebook page, I decided to speak out on Facebook about Freddie,' says Lottie.

'After I did it, I felt so relieved, and people were so supportive, both within the TSC community, and existing friends online. It's crazy. As a society we spend so much time talking about celebrities and the trivia of their lives and yet we are ashamed or afraid to talk about what's really going on in our lives and it's wrong. It needs to change.'

'There's loads of help out there, especially on the TSA's Facebook page, and the extremely active Tuberous Sclerosis Mums and Dads page. It's great to be able to meet with other people in the same position as you. Often it just helps to let off steam sometimes. I just want to open every door to every opportunity. You never know who you might meet, or what opportunities could arise, which might make all our lives that bit better.'

'After Freddie's diagnosis Ed and I gave up drinking for a while to help us keep clear-headed at all times. And I have started running which I find helps me so much. Before Freddie's diagnosis, I had never run anywhere in my life. I hated it! But now, I open the door and I go. You don't need anything other than a pair of trainers. Maybe, in a way, it's a kind of like running away, as it's just me, and I'm not carrying anything – in my hands or in my head. But it works. And it means when I come back I am calmer, have more energy, and can think clearly, so I'm much more able to look after Freddie in the way that I want to.'



“There's loads of help out there, especially on the TSA's Facebook page, and the extremely active Tuberous Sclerosis Mums and Dads page”

A move to a shared ownership property in Bexhill, made possible with financial support from both sides of the family, improved things a little, although Ed, 32, a tree surgeon, now has to cope with a daily round trip commute of 2.5 hours.

But it was only when Lottie 'came out' to her friends on Facebook that Freddie had TSC that things really started to feel just that little bit easier for the family.

# Mental Health, living with TSC

Ian Green is 54, has a loving family around him and a full-time job. Eleven months ago he was contemplating taking his own life.

'I had meningitis when I was two years old,' says Ian, 'and after that I started to have epileptic fits. I can go six months without a fit, and other times, I can have two or three in a week. You just never know.'

'I wasn't told that I had TSC until two years ago. But even with the fits, I was going along OK, OK, OK, and then just before last Christmas – suddenly, BANG! 'Out of nowhere, I couldn't settle, I couldn't sleep, I was walking the streets at 3am, I was becoming depressed – and I just felt like ending it all.'

Ian, who lives with his mum, Joan, 76, in Lisburn had reached the lowest point in his life.

'I just felt terrible,' he says. 'And I couldn't understand it. Some of it might have been to do with pressure at work and I don't know if being told I had TSC had anything to do with it because I'd been living with the epilepsy all my life.'

The turning point came when Ian, exhausted and desperate, confided in his sister-in-law Laura, that he was feeling suicidal.

'She was brilliant. She talked me out of it, and encouraged me to get professional help. So the next day I went to see my GP.'

Ian's GP, immediately recognising the seriousness of the situation, prescribed medication to stabilise Ian in the short term, signed him off work, and fast-tracked him on to therapy and a counselling course, with an immediate start.

'The therapy was the best,' says Ian. 'They came and picked me up every morning. I had a free lunch with them every day. I met lots of different people, some had ADHD some had OCD, and it gave you different things to think about every day. And nobody judged you.'

He also started to relax by colouring in.

'At night I do these adult colouring books now. They're really good. They do take the stress off, finding the colours I want to use.'



“Don't hide it, don't deny it”

Now, back at work, and feeling much better, Ian is passionate about encouraging others who are suffering with their mental health to get help.

'Tell people how you feel and go out there and get help,' he says. 'There is no shame in talking about it. Don't hide it, and don't deny it.'

'I feel so much better now. I am back to work and have been on holiday with my mum. I have surprised myself by what I have been able to do, and you don't know how many others there are out there who are like you until you start talking about it.'

Go to: [www.tuberous-sclerosis.org/tsablog](http://www.tuberous-sclerosis.org/tsablog) to read more of Lottie, Ed and Ian's story.

# Mental Health, a carer's story

It took a TSA T-shirt worn by a participant at a fun run to prompt TSC parent, Carolyn Bailey, to finally reach out for help. Carolyn's teenage son Charlie was diagnosed with TSC at 8 months old. Despite the support of her husband and Charlie's stepdad Paul, her daughter Holly, 27, and a handful of close friends, Carolyn, 47 felt like she had spent the past 18 years battling with the NHS, the education system and social services.

'It might sound stupid to some people, but I'd been too scared to contact the TSA, let alone look at the website,' says Carolyn. 'The hospital did tell us about the TSA after Charlie was diagnosed. But it was a kind of denial. I just didn't want to know what could be.'



So strong was her feeling of denial that when she, Dave, Holly, then aged 10, and baby Charlie left the hospital that afternoon 'for a few minutes and I didn't know how we could go on. We were devastated.'

That feeling thankfully passed but Carolyn now admits that the fight for the right healthcare, education and social services support has taken every scrap of mental strength she has. And in the past two years she has seen Charlie, now 18, suffer such mental distress that he has frequently threatened to end his life, antagonised by bullying at secondary school and college, and frustrated by an education system which has seemed, until now, unable to find a place and a purpose for a young man of his needs and capabilities.

'I was in my own "woman cage",' says Carolyn. 'Until now I have always tried to avoid the public side of TSC. Instead of reaching out for help when I should have done. 'I'm fine' is what I would say to anyone who asked. I have a few supportive friends without whom I would have gone under, there's no doubt about that. But I was essentially battling on my own. Now I wonder why I did that.'

**'I'd put on my armour and march out there – nobody in the system was asking how I was'**

Then a purple TSA T-shirt being worn at the Lincolnshire 10k in June of this year changed all that.

'I saw this woman wearing a TSA T-shirt, so I tapped her on the shoulder and asked her if it was anything to do with TSC. We got talking,' says Carolyn, 'and it was amazing. At last I had found someone else with a child with TSC. She then told me about the TSA Facebook page, and the TSC Mums and Dads Facebook group, and that was it. I liked the page, joined the group, and WOW! I've not looked back and now here I am, being interviewed for SCAN magazine.'

Carolyn describes her way of coping as 'I'd put on my armour and march out there'. Nobody in the system was asking how I was. You just can't go under. It's just not possible,' she says. 'But now I have found others in a similar position, and that is incredible. Being able to hear what others are going through, and share what I have faced, is liberating. And two books – Autism: Understanding Behaviour, and Autism: Supporting Your Teenager have also changed my life for the better. At last I could see some way I could help Charlie to cope a bit better with all the pressures in his life.'

And talking of Charlie, the family have now upped sticks and moved 140 miles West from Lincolnshire to Bolton, where Charlie is receiving specialist treatment at the Christie NHS Foundation Trust, hoping to gain a place at Rumsworth Special School, and is slowly feeling better about himself, partly thanks to Carolyn's new-found knowledge about how to support autistic teenagers.



'It's a battle and it's never ending,' says Carolyn. 'And I am still wary. I've been let down so many times before. I don't want to be let down again. Let's hope I get the help I need.'

Go to [www.tuberous-sclerosis.org/tsablog](http://www.tuberous-sclerosis.org/tsablog) to read more about Carolyn and Charlie's story.

# Mental wellbeing, helpful tips

Your GP is the gateway to other services. They will refer patients on to medical specialists, practitioners and other therapists as appropriate. And an NHS programme called Improving Access to Psychological Therapies (IAPT) makes it easier for people to access therapies such as cognitive behavioural therapy (CBT) and counselling. Details for your local IAPT services can be found on the IAPT website: [www.iapt.nhs.uk](http://www.iapt.nhs.uk) This all sounds positive, BUT, in recent years there have been cuts to many mental health services across the UK and waiting lists for treatment can be very long. For a guide to the NHS mental health services go to: [www.nhs.uk](http://www.nhs.uk)

## Other sources of help and support

There are many organisations beyond the NHS who may be able to help you maintain, or get back to, good mental health. These include:



**Anxiety UK** offers support, advice and information on a range of anxiety and anxiety-based depression conditions. [www.anxietyuk.org.uk](http://www.anxietyuk.org.uk) Call: 08444 775 774, Mon-Fri 9:30am - 5.30pm Text Service: 07537 416 905



**Autism West Midlands** have a very useful leaflet on 'Managing stress and anxiety. A guide for parents and carers of children with autism.' Download here [goo.gl/9klqCq](http://goo.gl/9klqCq) Call: 0121 450 7582 / 0303 03 00 111



**Carers UK** [www.carersuk.org](http://www.carersuk.org) Call: 0808 808 7777



**Cerebra** work to improve mental health and wellbeing in children and produce information leaflets and guides for parents. [www.cerebra.org.uk](http://www.cerebra.org.uk) Call: 01267 244200



**Childline** dedicated helpline for children and young people. [www.childline.org.uk](http://www.childline.org.uk) Call: 0800 1111



**hafal** works with people affected by mental illness across Wales. [www.hafal.org](http://www.hafal.org) Call: 01792 816 600/832 400



**Mental Health Foundation** is UK-wide and has a wide range of free resources. [www.mentalhealth.org.uk/publications](http://www.mentalhealth.org.uk/publications) Tel: 020 7803 1100 (London). 0141 572 0125 (Glasgow). 0131 243 3800 (Edinburgh) 02921 679400 (Cardiff).



**Mind** a booklet has been produced and can be downloaded for carers, 'How to cope as a carer'. It provides practical information on coping strategies, self-help resources as well as useful contacts. Download here: [goo.gl/g4DXgt](http://goo.gl/g4DXgt)



Mind InfoLine 0300 123 3393  
Mind BlueLight Line 0300 303 5999  
**National Autistic Society** [www.autism.org.uk](http://www.autism.org.uk) Call: 0808 800 4104



**Rethink** provide support for all people affected by severe mental health problems. [www.rethink.org](http://www.rethink.org). Call: 0300 5000 927



**Samaritans** whatever you're going through, you can call them for free any time, from any phone on 116 123. Or email [jo@samaritans.org](mailto:jo@samaritans.org). Or write to them at Freepost RSRB-KKBY-CYJK, PO Box 9090, STIRLING, FK8 2SA



**SANE** works to improve the quality of life for everyone affected by mental illness. [www.sane.org.uk](http://www.sane.org.uk) Call: 0300 304 7000 between 6pm and 11pm daily.



**SCOPE** has accessible mental health resources which may be useful for talking about mental health issues with someone who has learning difficulties. [www.scope.org.uk](http://www.scope.org.uk) Call: 0808 800 3333



**SAD** for help on how to cope with Seasonal Affective Disorder.



[www.sad.org.uk](http://www.sad.org.uk)  
**The National Institute of Mental Health** is a US-based website but it has a huge range of information about mental health and mental illness. [www.nimh.nih.gov](http://www.nimh.nih.gov)



**Time to Change** works to end the stigma around mental health problems. [www.time-to-change.org.uk](http://www.time-to-change.org.uk)



**Together-UK** helps people with mental health issues to lead independent lives. [www.together-uk.org](http://www.together-uk.org)



Call: 020 7780 7300 general enquiries line  
**turn2me** provides free online mental health help at [www.turn2me.org](http://www.turn2me.org)



**Young Minds** are committed to improving the emotional wellbeing and mental health of children and young people. [www.youngminds.org.uk](http://www.youngminds.org.uk) Call: 020 7089 5050



And finally, our team of TSA Advisers are here to help and support you and will tell you about services which may be able to help you. See the outside back cover for their contact details.

# New research funding in 2016

This year we received a record number of 13 applications for project and fellowship funding from across the world.

Between January and June 2016 we went through a detailed process of external peer review, internal review, research committee consideration and TSA Board approval.

The field of applications was, as ever, very competitive. It made our job to assess and prioritise funding extremely difficult. Of the 13 applications, 7 were of a sufficient quality that had the TSA had unlimited funds we would have funded them all. We therefore had to carefully prioritise our resources.

## We were able to fund three projects from the award round:

	Investigator	Host institution	Project title	Duration	Amount
	Dr Martin Baron	University of Manchester	Exploiting Notch regulation to probe alternative mechanisms of TSC signalling	3 years	£189,737
	Professor Simon Johnson	University of Nottingham	Inflammatory cell recruitment to LAM nodules drives tissue destruction in TSC lung disease: opportunities for drug repurposing	2 years	£109,634
	Dr Mark Nellist	Erasmus Medical Center (The Netherlands)	Functional and structural characterisation of the TSC complex; improved functional assessment of TSC1 and TSC2 variants associated with tuberous sclerosis complex	3 years	£124,707

Details of all the projects the TSA have funded can be found at: [www.tuberous-sclerosis.org/tsa\\_funded\\_research.html](http://www.tuberous-sclerosis.org/tsa_funded_research.html)

We were also able to approve some additional funding of £38,000 to the TRON clinical trial at Cardiff University. This allows continued funding for patients to visit Cardiff to participate in the trial and funding for staff and patients to undertake the work within its extension into centres in Belfast and Cardiff. More details about the trial can be found at:

[www.tuberous-sclerosis.org/take-part-in-research/trial-of-everolimus-in-the-treatment-of-neurocognitive-problems-in-tsc](http://www.tuberous-sclerosis.org/take-part-in-research/trial-of-everolimus-in-the-treatment-of-neurocognitive-problems-in-tsc)

New and continued grant funding means the TSA has committed to a research project spend of £434,000 for 2016 and its takes our portfolio of current research studies (in terms of their total funding) to £1.9 million.

This year, for the first time, the TSA endowed the title of 'Kilmaine Family Fellowship' to Dr Charlotte Tye of King's College London for her work on the TSA funded study 'Designing a protocol to measure the developmental trajectory of infants with Tuberous Sclerosis (EDiTS)'. This title was in recognition of the generous donation to the TSA made by the Kilmaine Family during 2015 and their contribution to the TSA over the years. The late Lord John Kilmaine served on the TSA Board for a number of years until 2000.

Over the coming editions of SCAN we will provide a summary of each of the new grants and information about investigators. 



The 'Kilmaine Family Fellowship' was awarded to Dr Charlotte Tye of King's College London for her work on a TSA funded study.

# Professor Simon Johnson: Inflammatory cell recruitment to LAM nodules drives tissue destruction in TSC lung disease: opportunities for drug repurposing.

We are really pleased to be able to start with details of the new project based at the University of Nottingham, along with an interview with Professor Simon Johnson. Simon Johnson may be well known to many members of the TSC community in the UK. He is Professor of Respiratory Medicine at the University of Nottingham and working clinically at Queen's Medical Centre, Nottingham. He is Director of the National Centre for Lymphangiomyomatosis (LAM), Co-chair of the European Respiratory Society LAM Task Force, member of the LAM Foundation scientific board and also a specialist professional adviser to the TSA. He has also recently joined the TSA's research committee (RESCOM).



The University of Nottingham

Lymphangiomyomatosis (LAM) is a lung complication of TSC caused by an overgrowth of cells in the airways, blood vessels and lymph vessels. LAM is more common in women than in men and so the female hormone oestrogen is thought to play a part in its development. Most women with TSC are mildly affected by LAM but for some it can become a serious problem causing collapsed lungs, breathlessness and sometimes respiratory failure. Although drugs such as Rapamycin and Everolimus can slow the progression of LAM, new therapies are still required to address elements of the condition that still progress despite treatment.



Dr Debbie Clements

Professor Johnson and colleague Dr Debbie Clements will be undertaking work in Nottingham looking at LAM at the cellular level and identifying if there are existing drugs that could be used to help control the condition.

The lungs of women with LAM contain abnormal cells with TSC gene mutations called LAM cells. The team at Nottingham has recently shown that LAM cells attract other cells called fibroblasts, which together can produce proteins that damage the lungs and attract inflammatory cells. This study will examine the mechanisms by which LAM cells and fibroblasts together attract mast cells. Mast cells are a specific type of inflammatory cell which are known to cause the growth of fibroblasts and the

activation of lung damaging proteins. The researchers will determine whether this occurs in LAM and importantly if there are existing drugs (used in other conditions) that can block these processes.

We caught up with Professor Simon Johnson about his research, joining RESCOM and got his advice for people with TSC LAM.

## Q. Please tell us about your experience of LAM and TSC, and your research?

We have worked on LAM for around 20 years and have always seen patients with TSC as part of that. That's increasing now as LAM is recognised as an increasing problem for adults with TSC. We're getting more referrals, particularly as screening for LAM is now part of the TSC Guidelines for clinical care. As the prognosis for adults with TSC improves because of better care, lung disease, because its a problem for adults and tends to progress through life, is becoming more of a significant problem for adults with TSC. LAM, like kidney disease are much more significant for adults with TSC.

We're really interested in a group of proteins that damage lung tissue. Our main focus is these proteins, and they're important for a number of lung diseases. The project that the TSA have just funded is a spin

off from understanding how the lungs get damaged in patients with LAM, and recognising that inflammatory cells are actually part of the lung disease - which wasn't on the radar until recently. We're going to look and see how these inflammatory cells contribute to lung damage in people with LAM.

## Q. Please tell us about your clinical role at Queen's Medical Centre Nottingham.

I've got an interest in rare lung diseases, and about 5 years ago we were funded to run a national service for patients with LAM, including patients with TSC. From the outset we've seen patients with LAM that occurs sporadically and also patients with LAM as a consequence of TSC. We've seen about 170 people with LAM at the service, around 20% of which have TSC. One of the things we also do is screen and give an opinion on the lung condition of patients with TSC.

cont...



Queen's Medical Centre, Nottingham

Very often that's LAM, but sometimes there can be other problems that aren't related to TSC. We assess people with TSC for lung problems and we see a lot of people that have been recently found to have LAM through CT screening. The service is UK wide service, people from anywhere can come. It's funded by NHS England.

**Q. If someone has been recently diagnosed with LAM what would you advise in terms of information and help?**

The first point to get across is that it's a really variable disease. I've seen patients with TSC LAM who have only got half a dozen cysts in their lungs and no symptoms, but actually they probably do have LAM. Those patients may never become ill because of LAM and that's reasonably common in TSC. The other side of the coin is that I have seen people die of respiratory failure due to LAM, and others that have needed lung transplants. So, if you have LAM the difference in outcome is potentially huge, so it's not necessarily as bad as you may think. Really what you need to do is sit down and discuss it with somebody who knows about the condition and see what it means for you. All patients, indeed all people, should avoid cigarette smoking. We've seen the disease is hormone driven because it's mostly a problem for women. Because of this, we recommend that people don't take oestrogen in forms of the contraceptive pill, or after the menopause we advise to avoid hormone replacement therapy. We recommend that they keep a healthy diet and that they treat chest infections promptly. They should get vaccinated against flu at the right time of year, and really keep active. We usually warn them of the risk of pneumothorax. LAM makes little holes or cysts in the lungs and sometimes these can leak air out of them and cause the lung to collapse, and that's often the first symptom of LAM. It usually causes sudden breathlessness and chest pain. Pneumothorax can be treated, but we often warn patients what the symptoms are like and what to do when they occur. And if the disease is active and progressive then we have drug treatments like Rapamycin, bronchodilators that open the airways, and oxygen.

**Q. What are the key areas of care someone that has been diagnosed with TSC LAM should be accessing from their local clinician?**

We usually make sure they have a baseline assessment. If they are able to do lung function tests we measure the size and the efficiency that they get oxygen into their bloodstream. And then we usually, depending on how active the disease is, repeat those tests over time. We'll talk to them about LAM and give them all the advice. That's the minimum. Usually we'll follow that up to make sure that their lung function isn't falling over time. If it is then we'll offer them treatment. People need to have an initial assessment, given the advice then followed up. That might not have to be often at all but it's good to have a contact if there is an issue. On the whole, they should be able to access most of these things from their local service. In practice, a lot of people do get referred to us. Basically, if someone is very well and their local person is familiar with the issues and is happy to monitor them, then that's all they need. If they have more advanced disease and treatment is contemplated, then they need to see someone who knows a bit more about LAM and knows about using mTor inhibitors. Everybody is different and you have to take it on a case by case basis. We're always happy to see people.

**Q. You've recently joined the TSA Research Committee (RESCOM), what do you hope to bring to the role?**

I hope that I'll give a slightly different perspective because I'm a respiratory researcher, so a different type of focus. We have an interest in tissue remodelling but those principals also apply to other problems. So, I think that having worked with patients for a long time I think I've got an understanding of what are some of the important clinical questions that researchers should be tackling. Hopefully we can work together to vet research proposals effectively and continue to make sure that the research that's of most benefit to patients gets funded.

**Q. Do you have anything you'd like to say to the TSC community?**

I have never been involved in a disease in which the progression of our understanding has been so rapid. From knowing nothing about it 20 years ago, to finding which gene is abnormal, to finding what the gene does, to having a potential treatment, then showing that that treatment is effective for the majority of patients, is gratifying indeed. That progress and interest from the research community continues. Having LAM nowadays is much better understood, there are treatments, and research is going at pace. It's certainly not the sort of problem that it maybe was 15, even 10 years ago. 🍀

# Goodbyes and Hellos for RESCOM

The last six months have seen considerable changes in terms of the membership of the TSA's research committee (RESCOM).

At the beginning of the year we welcomed a number of new scientific members to the group and in June we had a few further changes.

## Goodbye to...

We must sadly say 'farewell' to those leaving RESCOM, **Professor Harold Baum**, Retired Professor of Biochemistry at King's College London and **Dr Alan Fryer** Clinical Geneticist and TSC specialist at the Liverpool Women's Hospital. We are enormously grateful to both for their support and dedication to TSC research, the TSC community and the TSA.



Dr Alan Fryer at his recent hospital retirement party

Dr Chris Kingswood (TSA Trustee, Head of Research Strategy and Chair of RESCOM) reflects on their work with the TSA: "Alan trained under Professor John Osborne in Bath and

was an active participant in the first research projects in TSC in the UK. Over many years he has contributed to the welfare of the TSC community with his unfailing support of the TSA, and as an expert, kind and much-loved doctor. He has been a professional adviser for many years, taught at professional and family training days and has also previously led the professional advisory panel for RESCOM.

Harold has been a trustee and stalwart of RESCOM, offering wise counsel from his very broad knowledge of science and medicine. He has used his extensive network of friends and contacts in the scientific world enthusing them and drawing them into TSC. He was fundamental in setting up the landmark conference on molecular biology of mTOR, with Ann Hunt, at the Royal Society of Medicine (garnering grant from the Wellcome Foundation, MRC and Cancer Research UK). This was attended by the world's top scientists in the field and changed the direction of TSC research leading ultimately to treatment trials with Sirolimus and Everolimus."

The TSA would like to thank Harold and Alan for their huge contribution to the TSA over the years, particularly in helping to shape our research agenda, promoting its cause and their role in assessing external research applications whilst on RESCOM. They will both be missed.

## Hello to...

**Professor Simon Johnson** has joined RESCOM as an additional scientific member. A very warm welcome to Professor Johnson.



We also welcome a new lay member to the committee. After advertising in Spring 2016, the recruitment process was concluded with the Board of Trustees approving the appointment of **Corinne Swainger** to the post.



Corinne is a long-time active member of the TSA community. She has contributed to the TSA for over 20 years as a previous TSA trustee and helping to start TSA Outlook events for mildly affected adults in the early 1990s. She is ideally suited for the RESCOM role as she has worked as a medical writer/editor for over 25 years specialising in pharmaceutical communications.



Corinne at the 2016 Outlook event

Corinne said of the RESCOM role: "I have benefited from TSA-funded research in a number of ways. These have included new medications, genetic research, and clinical-patient support. I'm keen to see how new TSA research can help various TSC patients, carers and healthcare professionals."

Please see Corinne's blog at [www.tuberous-sclerosis.org/new-tsa-blog/medical-writer-corinne-swainger-joins-rescom](http://www.tuberous-sclerosis.org/new-tsa-blog/medical-writer-corinne-swainger-joins-rescom) on the TSA's website about her life with TSC and her involvement with the Outlook group. 🍀



# tasa BIG DAY

The TSA Big Day, held in September at the Nottingham Belfry gave members of our community, TSC researchers and specialists the perfect opportunity to discuss all things TSC.

## Fun TSA Big Day facts!

- 179 people came along
- 7 conference rooms
- 5 workshop leaders
- 20 children attended
- 6 TSC researchers showed their work
- 6 speakers
- 20 animals (including a skunk)
- 1 doughnut eating (without hands) competition

For those who couldn't attend, our workshop leaders gave us their top tips for the TSC community:

Living with TSC, including mental health issues, presented by Stacey Bissell from the Cerebra Centre for Neurodevelopmental Disorders (University of Birmingham):



1. There are lots of causes of difficult behaviour and sometimes it can be hard to work out what is causing the behaviour of the person you care for. Measures such as the Questions About Behaviour Function questionnaire, and the Face, Legs, Activity, Cry, Consolability pain scale may provide some insight into the causes of behaviour.
2. TSC is variable, try not to focus on comparing the progress of the person you care for to that of other children or adults with TSC. Instead identify their own individual areas of strength and difficulty. If they need support with speech and language, education, difficult behaviours, self-help skills etc, focus on these and track their progress relative to their own developmental history.



“Great to be talked to as an intelligent adult by enthusiastic researchers. Thank you to those who shared their experiences”

Benefits for carers and individuals, Charlotte Throssell from Disability Direct:



1. Get help to complete the forms
2. Don't believe all that you hear or read in the papers
3. Never give up
4. Be prepared to appeal
5. Ask someone who knows you well how you do things
6. Keep a diary and time yourself while completing tasks



“Thank you so much for the opportunity to hear all the great information and meet some inspirational people today. Will certainly attend in the future”

# TSA Big Day round up

Our biggest event of the year was full to capacity with workshops on wills and trusts, epilepsy, benefits for carers and individuals living with TSC. We've tried to bring you all the action, even if you were unable to attend. Here's some information from the TSA Big Day that you might find useful. Go to our YouTube channel [goo.gl/15IPwf](http://goo.gl/15IPwf) or search YouTube using #TSABigDay16 for all of our videos or see [www.facebook.com/thetsauk](http://www.facebook.com/thetsauk) for our Facebook Live videos from the day.



**Chair of the Trustee Board  
Martin Short and Jayne Spink**  
See what Martin and Jayne had to say about 2015, and hear and update on our work in supporting families, funding research and our #Fight4Treatment campaign.

“ This was my first Big Day and glad that I could be part of it. Glad my experience helped others. Would like to keep in contact with people as never met people with TSC ”



**Presentation by Professor Ben Whalley** of Reading University. See what Ben had to say on the use of Cannabinoids in the treatment of epilepsy.



**Hear the community stories**  
Members of the TSC community, Sarah Goy and David Dunn, stepped up to give their moving and inspirational accounts living with TSC.



 For news on Wills and Trusts by Gillian Coverley from Irwin Mitchell, head to page 24 of this edition of SCAN.

 **Our Facebook Live stream**  
We used our social media channel [facebook.com/thetsauk](http://facebook.com/thetsauk) to live stream many of the speakers. Click the 'like' button on our page and see the speakers in action.

 **Twitter**  
All attendees were encouraged to share their experiences of the day. You can find their tweets using #TSABigDay



**Our Impact Video for 2015**  
See how we made a difference in 2015 providing support, funding medical research, campaigning and celebrating our volunteers and fundraisers.

# The TSA turns 40!

In 2017 the TSA will be celebrating 40 years of supporting the TSC community. Launched in 1977 with a membership of 50 families and £25 in the bank, we now support over 1000 TSC households every year across the UK.

Far from a '40 year old mid-life crisis' we're going to be using this birthday milestone to shout about everything TSC. We want to raise as much awareness of the condition and the work of the charity as possible. We have some exciting plans afoot and we hope that you get involved too.

## 40 Stories to celebrate the 40<sup>th</sup> Anniversary

We need as many of you as possible to tell your stories. What's it like to live with TSC? Receive a diagnosis? What is it like to take part TSC research? Or, in a fundraising event for the TSA? These stories may be told by video, blog, Twitter, Facebook, SCAN or even through the local media. Contact Sarah Roberts, our Comms Officer, to put your name forward to feature as one of our TSA 40 stories. Email: [sarah.roberts@tuberous-sclerosis.org](mailto:sarah.roberts@tuberous-sclerosis.org) Call: 07870 210308

## Regional Group Birthday parties

We'll be hosting 40th birthday celebrations at all of our regional groups, and asking members of the community to give their experiences and memories from over the years. And yes, there will be cake!

## We'll be hosting a new fundraising event

The TSA have decided to host a brand new fundraising event to celebrate the big 40. Keep your eyes peeled for news on potential firewalks and zip wires!

## 40 Fundraising ideas

Our wonderful Fundraising Officer, Kathryn Harrison, has been putting her creative hat on and has come up with 40 awesome fundraising ideas for the year - with ways to get your business, school or local community involved too.

## TSA Big Day and Outlook weekend

We will be bringing together our Outlook event and the TSA Big Day at one single venue and hosting a celebration event. More details on this exciting event coming soon.

## Demonstrating impact and fighting for TSC funding

We'll be using the opportunity to gather facts and stats about what we've achieved over the last 40 years to demonstrate our worth to funders and raise more funds for TSC research and support services.



Keep your eyes peeled for this special 40<sup>th</sup> Anniversary logo for all celebratory activities, events and communications, and follow the #TSA40 hashtag for the latest updates on the year's events.



We'd like to say huge thanks to all who came along to our annual #ScottishGetTogether which we held on 27th August at The Lighthouse, Glasgow.

TSA Adviser for Scotland, Lynn Shields says:

'Our grateful thanks go to our amazing speakers. The evaluations from the day highlight how much everyone benefited from all the expertise and information. It was wonderful to hear that carers rights are being strengthened in the near future, and, the possible changes to the benefits system in Scotland.'



Please have a look at the easy to use benefits calculator from Turn2us [www.turn2us.org.uk](http://www.turn2us.org.uk)

**Speakers on the day included:**

- Dr Joss from the TSC Clinic Glasgow
- Heather Noller from the Carers Trust Scotland
- Rhona Matthews from Talking Mats
- Linda Mitchell from the Scottish Commission for Learning Disability
- Ross Craig from Turn2us
- Chloe Burton from Carr Gomm

We audio recorded speakers on the day, and you can now hear their presentations on our YouTube channel - just search #ScotGetTogether2016

If you want more information about the event, or have suggestions for events in Scotland in the future, contact Lynn Shields. Phone: **01236 608257** Email: [lynn.shields@tuberous-sclerosis.org](mailto:lynn.shields@tuberous-sclerosis.org)



## Welsh Info and Fun Day

Looking to the future was the message that came across at the Future Inn, Cardiff Bay on 10th July when the TSA Wales Fun and Information Day was held. The day proved to be a success and brought together families from across the country.

An inspirational talk was given by Gerwain Wilson (TSA volunteer) on his experience of growing up with TSC, followed by a presentation by Professor Julian Sampson of Cardiff University on access to medicine as well as current and proposed future research into the condition.

Icaris Duo flute and guitar combo and Charlie the magician provided fantastic entertainment for both children and adults alike, and there were information stands run by Epilepsy Action Cymru, The National Autistic Society and The Tuberous Sclerosis Association. The session concluded with a Q&A session with Gerwain Wilson, Professor Sampson and Jenny Jones TSA Adviser Wales taking part.

Thank you to all those who attended on the day, to Gerwain Wilson and Professor Julian Sampson for their presentations, TSA and Gene Park Wales volunteers and Marie James NAS Carmarthenshire and Glyn James, Epilepsy Action Cymru, for the information stands. The day was made possible due to funding from the Waterloo Foundation via Emma Hughes, Development Officer Wales at Genetic Alliance UK.

**Thank you to Emma for securing this funding and for her part in organising the event.**



## Janet Medcalf Award

Award will buy toys for Tommy's sensory room.



The latest winners of our quarterly Janet Medcalf Award are Stephanie Lucas, Chris Wilde and three-year-old Tommy Wilde, who has TSC, from Erith, Kent. Stephanie writes:

'At 2 days old Tommy started to have seizures but it wasn't until he was 8 weeks old that he was kept in hospital and was admitted for a week waiting for a bed at the Evelina London Children's Hospital. Once there, within a few hours Tommy had had an MRI and an EEG and he was diagnosed with tuberous sclerosis complex.

At the time, the diagnosis didn't sink in to what we were going to have to go through, and three years later sometimes we feel that it still hasn't sunk in.

We are often asked how we cope with Tommy's condition and watching him have seizures up to 80 times a day. The truth is that we don't know any way of parenting other than to deal with it. We have had to witness Tommy having seizures since he was 2 days old and he has never had a seizure-free day. To us this is our normal everyday life now.

Each day we don't know how Tommy is going to be awake, whether he is going to sleep all day or not sleep at all, or be poorly and end up in hospital. We can never plan holidays, parties, or family days out as we never know how Tommy is going to be or even if we are going to be home. He has spent 2 out of 3 birthdays poorly and asleep all day.

Tommy has already overcome many obstacles in his life. Now three years old he has only recently learnt to support his head and to giggle. This makes buying toys and presents for him extremely hard as everything is much more expensive. We are currently trying to turn a room in our home into a sensory den for him, but all of this comes with a big price tag. Affording the toys he needs is made even more difficult because all the time we need to spend with Tommy in hospital impacts on our earnings.'

Tommy's grandmother Roberta Lucas writes: 'Both myself and my husband Steve, and all the family are extremely proud of my daughter Stephanie and her partner Chris for all they have gone through - and always come out the other side. Stephanie was 19 and Chris was 22 when Tommy was born. It breaks our hearts to see everything that they have to go through, but they are both excellent parents and always put Tommy first.'



**The deadline for the next draw for our Janet Medcalf Award is 31 December.**  
**To enter please email [moira.dennison@tuberous-sclerosis.org](mailto:moira.dennison@tuberous-sclerosis.org) and tell her how £300 would improve either your life, or someone you care for who has TSC. All entries are entered in to a randomised draw.**  
**Or write to her at our head office address (please see the outside back cover for full details).**

# Campaigning

The #Fight4Treatment continues.

Here at the TSA we believe that everyone affected by TSC has the right to the best possible treatment and care. We launched our #Fight4Treatment campaign 3 years ago when it became clear that patients in the UK were not able to routinely access Everolimus on the NHS. It's been a long road, filled with delays, broken promises and changes in the prescribing process at NHS England. But, through it all, we've had ever-growing support from you, the TSC community, other rare disease charities, clinicians, politicians and journalists. And, we've had some success with Individual Funding Requests and securing routine prescribing for TSC-related kidney tumours (AMLs). However, despite a commitment to develop policies for routine prescribing, our latest battle has been over the negative decision in June 2016 to deny routine funding for Everolimus for inoperable brain tumours (SEGA). We estimate that this decision will affect around 20 children and young people each year. NHS England has decided that Everolimus is 'unaffordable' and there is no right of appeal. For us, this decision is wholly unacceptable.

Through an unrelated legal challenge, the group that made this decision (CPAG) now have to rerun the process, offering a chance for NHS England to make the right decision. And in response we have applied pressure both publicly and by lobbying decision makers. We want to thank all of you that have taken part in this vital campaign - we can't do it without you. Here we give a roundup of our campaigning activities.

## All Party Parliamentary Group Meeting, 11<sup>th</sup> October 2016

Our CEO, Jayne Spink, Dr Chris Kingwood and family campaigners Nick, Jo and Sam Dale attended the Rare Diseases All Party Parliamentary Group (APPG) meeting on Access to Medicines at the Houses of Parliament on 11th October. Here Nick explains why they went and what they hope will come from it.



### Tell us about your trip to the APPG in London. Why did you attend?

It is very easy for access to medicines issues to be reduced to numbers and policies, so much so that those involved sometimes forget that they are talking about people. We were invited to attend the APPG Access to Medicines meeting by Rare Disease UK. The idea of any APPG is for MPs to gather and consider the facts. We decided to attend because we were asked to represent patients and their families, who despite being the stakeholders closest to the front line are generally under-represented. We are lucky that our employers supported us to do this and Sam's health and behaviour is good enough for him to travel to London (just!).

### What did you get from attending the APPG?

We didn't attend to get anything from the meeting for our family. We attended because we could and lots of people who need representation can't attend or wouldn't feel comfortable speaking in that type of meeting. I hope that we made it clear to all present that this is an issue that affects real people and that the current NHS policy development process is not only not fit for purpose, it is endangering people's health and lives. Some of those present thanked us for reminding them that it is about people and not politics. So hopefully that is a positive outcome. I invited our MP, Julian Sturdy, who attended, and has now joined the APPG which is another positive outcome. I also invited the Prime Minister who, oddly, was too busy to attend. However, at least she should now be aware of the issues we face.

Julian Sturdy MP ended up talking to Greg Mulholland MP, who has been a great advocate throughout our campaign, and between them they decided to stage a protest outside Parliament before Prime Minister's Questions the following week. They decided to do this in half term to allow more families to attend. We worked hard to get as many families and MPs as possible to the protest and the meeting afterwards. Greg Mulholland MP was convinced that this would increase the profile of the #Fight4Treatment campaign. I invited the Prime Minister again, but at the time of writing this had not received a response from her. However, the MP who heads up the APPG, Ben Howlett MP, agreed to attend.

### How do you think this helped our #Fight4Treatment campaign?

It appears that the only way anything will change is if the NHS is put under pressure to change a process which is unfair, lacks transparency and one that leads to serious consequences for patient health. The past 4 years seem to have proved that policy makers don't always make evidence-based decisions and that they may also ignore expert and patient opinion. This means that we need to influence the politicians and encourage them to take action and hold NHS England to account for the way it works and the decisions it makes.



## Taking Tea on the Terrace, Parliamentary Reception, 19<sup>th</sup> October 2016

For the first time in our history, the TSA brought together 60 invited guests to take tea on the Terrace of Parliament. We used this opportunity to raise awareness of the condition, the issues our community face and the work of our charity. The invite list included TSC researchers, families and individuals affected, politicians, journalists and funders of our work. We made presentations on the importance of funding TSC research and the current issues around access to treatment and medicines our community encounter. These events are vital to raising awareness of the condition and building lasting relationships with people that can help us to improve the lives of people with TSC.



### “ Actor David Suchet has a grandchild with TSC and has become an invaluable supporter. He spoke at the event:

“200 years ago or even possibly 100 years ago, these babies (with TSC) would probably never have survived. Not a day goes by when our family does not celebrate the fact that we have the most delightful and loving little boy. He and every other child with this condition has the gift of a life that can now be truly supported. Because of ongoing research we now have the knowledge and hopefully the drugs to transform the quality of lives of those with TSC. Sadly these drugs have to be fought for because they are not readily available from the NHS. All of us have a duty and a vitally important one. We have to make sure that rare diseases in general, and TSC in particular, do not continue to be short-changed when 'high up' decisions (even in this very building) are made about funding and resource allocation. This is a most important core message. We can deliver this message together and we MUST!”

- Our e-petition reached 17,142 signatures
- We've had in excess of 40 mentions in parliamentary debates, questions, early day motions and Prime Minister's Questions
- A letter signed by 13 consultants, strongly objecting to the decision was published in The Times
- 94 people have written to their MPs
- 12 MPs have written directly to the Secretary of State for Health on their constituents' behalf
- Our e-petition reached 30,626 people on Facebook
- And 39,752 users on Twitter

We have had national coverage in the Daily Mail the Daily Express and several regional and local newspapers. The issue has also been covered on ITV Calendar Region news, BBC Radio York and Leeds-based commercial station, Radio Aire.





## #Fight4Treatment Protest, 26<sup>th</sup> October 2016

Following the APPG meeting, Greg Mulholland MP, the Dale family and the TSA decided to organise a protest outside Parliament. Over 40 people attended and the protest got the backing of 16 MPs. After the demonstration families and supporters were invited back to a committee room at House of Commons to meet with their MPs after Prime Minister's Questions and discuss the issue. We were delighted when Greg Mulholland was allowed to table an oral question to the Prime Minister, Theresa May, about the negative decision for prescribing Everolimus for inoperable SEGA. Theresa May said that she was very happy to look at the issue and look in detail at what can be done to take it forward.



Watch Greg's question to Theresa May at Parliament here: (12.41pm) [goo.gl/jZpxpK](http://goo.gl/jZpxpK)



The Dale family appeared on ITV Calendar News and made the front page of the **York Press**, twice.



Juliette Felce and daughter Keira made the front page of the **Bedford Citizen and Times**.



Lottie Norton and Ed Usher made the front page of the **Bexhill-On-Sea Observer**



Yvette Cooper MP of Pontefract and Castleford said: "This is a decision I simply do not understand"



And Jessica Holt and Darcy made lead news item on Radio Aire as well as the **Wakefield Express**.



The Ward family made headlines on **BBC Radio York**.

## TAKE ACTION NOW

Anybody who has been directly affected this decision please contact your regional TSA Adviser. Their details can be found on the back on this magazine.

Enormous thanks to all who attended the protest, shared our updates on social media and to the MPs who are supporting the **#Fight4Treatment**

At the time of going to print we are waiting on the results of the rerun Clinical Priorities Advisory Group (CPAG) meeting on the whether or not NHS England will prescribe Everolimus for inoperable SEGA. We will update you in the next edition of SCAN and through our e-newsletters when we have the latest decision.

# TSA Events

## Tuesday 22nd November, Brighton Regional Group

12pm-2pm at Stepping Stones Children and Family Centre, Gardner Road, Fishersgate BN41 1PN

## Saturday 26th November, East Midlands Regional Group

1pm-4pm at Broadway Baptist Church Hall, Broadway, Derby DE22 1BP - super hall, facilities and on site parking.

## Tuesday 6th December, Berkshire Regional Group

11am – 1.00pm at St. Andrew's Methodist Church, Merton Road, Slough. SL1 1QW

## Saturday 10th December, Bristol (South West) Regional Group

10.30-12.30pm Christmas Get-Together at Holy Trinity Church community Hall, Stapleton, 23 Park Road, Bristol BS16 1AZ

To find out more about our Regional Groups, contact our Volunteer Manager, Tanya Damton

Call on 02380 730823 or email [tanya.damton@tuberous-sclerosis.org](mailto:tanya.damton@tuberous-sclerosis.org)



## The Great North Run

We are thrilled that this year we fielded our biggest number of runners ever to tackle the iconic Great North Run for us on 11th September. Huge thanks to our runners: Amy Ennis, Andrew Smith, Piaras Connelly, Sarah McCartney, Shaun McCartney, Steve Watson, Luke Styles, Vickie Hand, Rebecca Featherstone, Karen Deasy, Vince Lawlor, Nicola Daykin, Glyn Daykin, Steve Sargant, Hollie Light, Angus Filshie and Andy Hara.

Together, our runners have raised over **£4000** – thank you to all who ran, and, supported them.



## Ride London - Surrey 100

On 31st July Michael Donnelly, Elliot Parfitt, Tom Stafford, Mark Hindley, Chris Pratt and Sam Hodgkinson completed the 100-mile bike ride, Prudential Ride London - Surrey 100, which started at the Olympic Park in London and finished on The Mall!

It was a great day and together the team has raised more than **£5,000** for TSA. THANK YOU ALL!



Join **#TeamTSA** and take part in a fundraising event today! Call or email our Fundraising Officer Kathryn Harrison on 0161 681 6015.

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



# Wills and Trusts

Over the last year we've been working with Irwin Mitchell to bring to the TSC community an option for setting up Wills and Trusts to look after your loved ones when you are no longer here.

The TSA asked that any financial incentive from Irwin Mitchell for people taking up their services be passed back to you, the TSC community.

We were delighted that expert Gillian Coverley, Partner at Irwin Mitchell, joined us at the TSA Big Day to host a law workshop and launch the Will writing service with the TSA.

Here's what she had to say:

## What services are Irwin Mitchell offering?

The Will writing service has been developed in partnership with the TSA. Both as a means to support the charity by leaving gifts in Wills, and also to look at how we can work with the charity to help community members who might need to set up Trusts, through Wills or independent of that, to support their children or other people with TSC when they are no longer here.

## Why do we need to set up will?

It is really important to set up a Will. I recognise that many supporters will already have one, but as well as setting one up it's important to make sure it's up to date. Some people won't have a Will because naturally people don't like thinking of their own death, and think it's something they'll do in the future.

Sadly, only 3 out of 10 people make a Will, that means plenty of people die without one. It's really important to make one. It's your record to your family about how your estate should be distributed. It gives peace of mind to your family that you've written down exactly what you want. It's particularly important if you've got vulnerable people to think about and how they will be protected when you're no longer here. Without a Will the intestacy laws come into effect, they're a very broad brush and are basically just a list of people that will inherit. Usually, they're not adequate and it's much better to make a Will because you're in charge.

## How does Irwin Mitchell support people to set up a Trust?

We recognise that you will need to support people beyond your death if you have a vulnerable person to look after. At Irwin Mitchell we have a Trustees service. And it's very important when you're thinking about setting up a Trust whether or not you want to appoint a professional body to act with you. The role of the Trustee is to look after assets on behalf of another individual who isn't able or capable of looking after the assets themselves. There are lots of responsibilities that go along with being a Trustee, you have to consider all aspects including finance, tax, the balancing of various interests and make sure you're acting in accordance with what you think were the instructions given to you in the Will document. So Irwin Mitchell offer the Trustees service to help Trustees and also act alongside them to give them advice and support, in setting up the Trust but also running it.

## Why should our community choose Irwin Mitchell?

In terms of Wills and Trusts, we are absolute experts in this area. We are a national firm of solicitors and we can offer this service nationally. It's a very quick and simple process. One of the main barriers to setting up Wills and Trusts is that you have to go and see a solicitor, sit down with them for a long time and it can be quite expensive. This is a very simple and cost effective way to get something sorted out very quickly with a leading firm of solicitors in the UK.

To view the whole interview search in YouTube [#TSABigDay16](#)

You can also watch an Irwin Mitchell video on Wills and Trusts here: [youtu.be/vIXgCM2fSa4](https://youtu.be/vIXgCM2fSa4)



# Nicholas Annear, marathon runner

Driven by working with patients in the St George's TSC Clinic, and inspired by hearing more from the TSA's very own Jayne Spink and Martin Short, Dr Nick Annear decided to take on the challenge of a lifetime.

The 26.2-mile Virgin London Marathon is not for the faint-hearted, but in 2017 Nick will undertake the greatest physical challenge of his life to raise money for TSC research and our TSC support services. We caught up with Nick after completing his first 5-mile training run. Having hung up his trainers in 2010, after running the London 10K for his local kidney patients association, he was nervous but excited by the preparations for race day.

*'I feel good, I'm very pleased to be at 5 miles already. I haven't yet been out in my TSA running vest, but soon I'll feel confident enough to take part in preparation events and put the full kit on, and finally admit that I'm going to run the marathon!'*

Nick first became interested in TSC while doing his PhD at University College London, performing research into kidney tumour syndromes. It was some years later - on returning to his clinical training in renal medicine - that he attended an inspiring lecture by Dr Chris Kingswood about TSC, and learnt more about the novel treatment options that are becoming routinely available for certain patients with the condition.

*'It's a fascinating condition - each patient can be so different, and affected across such a wide spectrum - from showing no outward evidence of the condition at all to being very severely impaired. It's interesting to see how one class of medication (mTOR inhibitors), often used to prevent rejection in kidney transplantation, have made such a difference to TSC patients. It's a wonderful example of how good science has been applied to ameliorate a complex condition that affects so many different parts of the body, and in many cases can prevent serious complications from developing.'*



Nick started as a Consultant in Acute and Renal Medicine in June 2015, and is now part of the St George's TSC Clinic in London. Working with Dr Chris Kingswood and Dr Frances Elmslie, he regularly sees TSC patients. He supports the TSA because he's seen first-hand what a difference our support team can make in both referring patients to their local TSC clinic, and helping in other ways to support patients and their families in often very challenging circumstances.



*'It's a partnership, a collaboration between the Clinic and the TSA'*

Aside from raising funds for the TSA, there are a couple of reasons Nick wants to run the Virgin London Marathon.

*'I want to raise awareness of the condition and TSC Clinic at St George's Hospital. Also, my wife ran the London Marathon in 2009, so in the true spirit of family competition, from that moment on I knew I had to run it too.'*

Nick will be supported on the big day by his wife Vinny, and daughters Freya, 5, and Emelia, 2. Finally, is there anything he'd like to say to you, the TSC community?

*'It is an enormous privilege to work in the St George's TSC Clinic, and to support families and individuals with TSC. I hope that I can help improve the visibility of this condition so that more people can benefit from earlier formal diagnosis, specialist input and intervention.'*

We're enormously grateful to Nick for his fundraising effort.

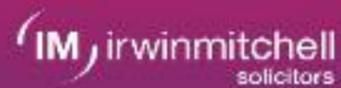
If you'd like to support Nick today you can visit his JustGiving page: [www.justgiving.com/Nicholas-Annear](https://www.justgiving.com/Nicholas-Annear)

If you'd like to know more about the St George's TSC Clinic, you can visit their website here: [www.stgeorges.nhs.uk/service/tuberous-sclerosis-complex-tsc-clinic](https://www.stgeorges.nhs.uk/service/tuberous-sclerosis-complex-tsc-clinic)

Find your nearest TSC Clinic here: [www.tuberous-sclerosis.org/clinics-map](https://www.tuberous-sclerosis.org/clinics-map)

Want more information on the Wills & Trust Service with Irwin Mitchell?

Call 0370 1500 100 or visit [www.tuberous-sclerosis.org/wills-and-trusts](https://www.tuberous-sclerosis.org/wills-and-trusts)



# FUNdraising Gallery



◀ **Emily Dyson**  
Emily Dyson raised an amazing £243.24 by doing a skydive for us in July. Emily told us: 'The skydive was amazing – I absolutely loved it! My sister has TSC, so I am doing this skydive to raise some money and awareness whilst doing something that I am excited about.'



◀ **Lindsey Crawford**  
Lindsey Crawford faced her fear of heights and did a skydive for the TSA in July and raised £988. Lindsey was inspired by her son Lewis who has TSC.

▶ **Beeston Community**

We would like to say a huge thank you the community of Beeston, Nottingham, Wilko's, the British Legion, TSA supporter Nicola Daykin and David Clifford for all their support raising £1,000 for TSA. David organised a Cabaret night at the local British Legion Club in Beeston, and the staff at Beeston Wilko's held a Family Fun day.



▶ **Barry Gilmour**

Thank you to Barry Gilmour who rode 68 miles as part of the annual Ride Staffs Weekend Festival at Shugborough Hall, Stafford. Barry raised £325 for TSA because his close friends have a son with TSC, and Barry sees the struggles they go through. Barry said: 'My friends are the most hard working parents and are fantastic with their son, and its unbelievable how they manage.'



◀ **Vicky Wood**

Thank you to Vicky Wood who raised £2,415 by doing the challenging Bolton UK IRONMAN. Vicky - who until last year did not own a bike and had barely swam a length in her local pool - swam 2.4 miles in open water, cycled 112 miles and then ran a marathon - all in 15 hours and 3 mins. Vicky's daughter Millie, now 6 years old, has TSC.



◀ **Daniel Marks**

Daniel Marks and his father Len Kirby completed the Vitality 10k London Run in July. Inspired by Daniel's daughter, Cece, who was diagnosed with TSC in August 2015, they have raised an amazing £1,300. Daniel said he wanted to 'continue to make a difference by supporting the research programme and help them continue the vital services, giving families the support they need until they can find a cure.'



◀ **Elaine Russell**

Following her Tea & SCones event at Cardiff University Cancer Genetics Department, Elaine Russell challenged herself to run the Caerphilly 10k. Running it in under an hour and raising over £325 - we'd like to say THANKS Elaine!

▶ **David Cropper**

Huge thanks to David Cropper who ran 110 miles for the TSA. Friend to the Dale family, David decided to run for their son Sam, who has TSC. He is running to support our #Fight4Treatment campaign and has already raised over £1,900.



**In Memory of Alex**

We would like to say a huge thank you to Dale Mitchell, Sarah Hatter and friends who ran their 4th annual crib evening in memory of Dale's brother, Alex, who had TSC, raising £452. We'd also like to thank Barclays Bank who match funded the £452. Match funding makes a real difference, so if you are raising money for TSA ask your employer about their match funding policy.



◀ **Roisin Hagen**

Roisin Hagen has raised over £1,000 in memory of her cousin, Matthew, who had TSC, by holding a Own Clothes Day at the school she works at, and by organising her very own Strictly Come Dancing Evening in Portadown. Roisin said 'Thanks for all the work you do for those with TSC. I know my Aunt and Uncle, parents of my cousin Matthew, speak so highly of your charity.' Keep dancing Roisin!

▶ **Lisa Campbell**

Lisa Campbell got a team of 7 women to run with her in the Annual Glasgow Women's 10km, in memory of Lisa's uncle Billy, who had TSC and who passed away earlier this year. The team did fantastically well on the day and raised an amazing £1,634. What a tribute!



◀ **Janice and Peter Oliver**

Celebrating their wedding on 30th April 1966, Janice and Peter Oliver asked for donations in lieu of presents for their Golden Wedding Anniversary and raised £419. Congrats guys!

▶ **Oliver, Zara, Kyrrah, and mum Jennie**

Well done Oliver, Zara and Kyrrah, and mum Jennie Binnie, who took part in the Colour Me Rad run in Aberdeen and raised an amazing £455! Jennie said 'The kids did fantastically. We are all so proud of them and the effort they put in. They loved getting completely covered in the powder and were rolling around the floor at each station. So were the adults!'



▶ **Wendy Lane**

Well done Wendy Lane, her son Dylan, 14yrs, and friend Anna Higgs, who all took on the Colour Run in Windsor and raised £164. Wendy's Mother and Aunt both had TSC.

▶ **Philip Goldenberg**

Our wonderful Deputy Chair, Philip Goldenberg, raised an amazing £3,125 from donations in lieu of birthday presents for his 70th Birthday. Philip had a fantastic time celebrating with family and friends, and was very pleased to have passed his personal target of £3,000. Thank you for everything you have, and continue to do, for the TSC community.



▼ **The Mayor of Didcot**

The TSA are proud to announce that the new Mayor of Didcot, Steve Connel, and his wife Sarah have selected TSA as one of their chosen charities for the Mayoral year May 2016 – 17. So far, Steve and Sarah, have raised over £800 at local community events. We look forward to working with Steve and Sarah over the coming year.



▶ **Leon Upton and Luisa Vass**

Massive thanks to Leon Upton and Luisa Vass who have raised a fantastic £4,674 by completing a marathon along the Great Wall of China. Luisa said: 'It was an absolute pleasure to raise money to help such an amazing charity, which means so much to us all.'



▶ **Darren Mackay**

ran the Edinburgh Marathon Festival 10k and raised a fantastic £228. Darren said 'the reason I decided to fundraise for TSA is that they supported a very good friend of mine when her daughter was diagnosed, and every little that can be donated to help them is a plus as far as I'm concerned.'

Feeling inspired? Contact our Fundraising Officer to discuss your fundraising ideas!  
kathryn.harrison@tuberous-sclerosis.org 0161 681 6015



# Your Contacts At The TSA



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## Get Involved



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## Fundraising



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## Research



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## Get Support

Our TSC Advisers work across the UK. Although all Advisers work part-time, we will answer your query as quickly as we are able.



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Please note our address for postal donations or enquires is:

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