



Tuberous Sclerosis
Association



Annual Report 2009/10

Our Mission Statement:

“To promote awareness, research, and the best management of Tuberous Sclerosis Complex, and support all affected individuals and families”

What is the Tuberous Sclerosis Association?

Established in 1977, the TSA works to provide support for today and hope for tomorrow for people affected by TSC. With an ever increasing membership, it has raised nearly £10 million for research and support services for thousands of individuals.

Its scope is:

- to create new ways for people affected by TSC to find known answers
- to provide educational events, information and advocacy
- to set up the means to find answers that are not yet known
- to promote medical, social and educational research
- to provide a way to fight back

1 April 2009 – 31 March 2010

Highlights of the Year

April

- Outlook Group Weekend, Glasgow
- Launch of new quarterly Scottish newsletter
- Features on TSC published in British Journal of Neuroscience Nursing (BJNN)

May

- Family Fun Day at Center Parcs, Suffolk
- Davies family featured in TV documentary, 'Born to be Different' on Channel 4

June

- New TSC Adviser appointed for North-East England, North-West England and Wales
- Parent and Carer Event with National Autistic Society, Glasgow
- Regular Giving brochure launched
- John Lewis Partnership magazine, 'The Gazette', and John Lewis Partnership website ran an article on carers which featured Emma Seager and daughter Charlotte who has TSC

July

- Epilepsy Nurse Specialist TSC Awareness Training

August

- RS Macdonald Charitable Trust grant of £11,000 towards the work of Scottish TSC Adviser
- McGhee Family raised £19,787 through TSC Charity Ball

September

- TS Alliance Conference, Chicago TSC: From genes to new therapeutics

October

- SSBP Conference, Cambridge : Listening to Genetic Disorders: The Etiology and Treatment of Neurodevelopmental Disorders
- Launch of TSA School's Training Pack
- Family Fun Day at Center Parcs, Nottingham
- Local media coverage around TSC Awareness Month

November

- Annual General Meeting and Annual Conference, Harrogate
- Presentation of Maggie Smith Challenge Award to Nick and Jenny Budinger and Lynne Murton
- Scottish Get-Together, Glasgow
- TSA family Emma Seager and daughter Charlotte featured in Contact a Family (CAF) 30th anniversary campaign 'Our Family, Our Future' and launch at House of Lords.

December

- Submitted NHS Choices TSC information summary including family case stories.

January

- Natalie Milham featured on BBC TV South East

February

- Donation of £15,000 from O2e for child-oriented activities
- Media campaign around Rare Disease Day 2010
- Eurordis facilitated meeting in Zurich on patient participation in clinical trials

March

- Scottish members and TSC Adviser attended a reception at Scottish Parliament for Rare Disease Day 2010, Holyrood. The TSA was also represented at a corresponding event in Belfast and supported publicity around the event in Cardiff
- Fauchier Partners chose TSA as Charity of the Year

Chairman's Report

Dear Members

This is my first report to you as incoming Chair, succeeding Chris Naylor who will be a hard act to follow, and to whom we are all grateful for the workload she assumed and discharged so efficiently and elegantly.



Elsewhere in this document you will find details of the splendid efforts that the TSA made in the last financial year. I want to add my own thanks to everybody involved, not least our staff who - inevitably in a small organisation - sometimes find themselves undertaking tasks beyond the call of duty, and deserve our gratitude for doing so.

Let me now be very personal.

As our members will have seen in our TSA magazine, SCAN, our eldest child Jonathan, who had TSC, died last June, aged 23. We had known for some time that this outcome was inevitable, but those who have been through a similar experience - as all too many of us in the TSC "family" have - know that no amount of pre-grieving prepares parents for the awful experience of burying their own child.

I took some time after this to take a quite difficult decision as to my future involvement in the TSA. Emotionally, it was very tempting to "turn the page", and cease my involvement which inevitably entails emotionally re-living all we as a family had suffered. But in the end I decided that this would be self-indulgence, and that I should do my best to ensure that the TSA, which had been such a lifeline to us, should continue to be a lifeline to others.

In particular it was clear to me that, while the TSA owed an enormous debt of gratitude to its founders, its "founder culture" was not a sufficient basis for future success. To do justice to those we seek to serve, and also to our wonderful staff, we need to make ourselves "Fit for the Future"; that was the basis on which I was prepared to assume my rôle, and on which the Board of Trustees appointed me. I am therefore delighted that, as reported in SCAN, the Board unanimously accepted my recommendation for a constructive professional review of our activities, so that we emerge not only with the clear objectives we already have ("Care for Today, Hope for Tomorrow"), but also with a well-worked strategy to achieve them. At the time of writing this in July the review is getting underway; by the time of the AGM I hope to report its outcome.

Philip Goldenberg
Chairman
Tuberous Sclerosis Association

What is Tuberous Sclerosis Complex?

Tuberous Sclerosis Complex is a genetic condition which can affect many parts of the body causing a variety of signs and symptoms. TSC, which can be diagnosed any time from before birth to adulthood, is caused by mistakes in one of two genes which regulate cell growth, resulting in extra growth of normal tissue (benign tumours). Tumours can grow in any organ of the body. Those in the brain may lead to epilepsy, autism, attention problems and learning difficulties. As well as these day to day problems which affect the life of individuals with TSC and their families, there is the underlying worry that larger growths in the kidneys, brain tumours or cysts in the lungs could cause very serious health problems. It is a difficult condition to understand and cope with, both for those affected and for the people who care for them - family members, friends or the range of health and social care professionals who may be involved in their care. It affects 1 in every 6000 people born but in very varied ways. For some families and individuals, the worst thing is the unpredictability of what might come next. TSC may cause;

- epilepsy
- learning disability
- behavioural problems
- autism
- hyperactivity
- skin lesions
- facial rash
- brain tumours
- kidney problems
- physical disability
- lung problems
- heart tumours

Bringing Families Together



“The TS Association is like an extra backbone –They are really supportive and I can always rely on them to find me trustworthy information quickly.”

From the feedback we capture at all events, our members constantly comment on the importance of being able to link up with other people who really understand what it is like to live with TSC in the family. Therefore we strive to create as many opportunities as possible for people to come together and offer mutual support in a relaxed and understanding environment.

Outlined below are this year’s key events and some of the wealth of positive feedback we have received.

Glasgow was the setting for our annual **Outlook Event** when 35 mildly-affected adults with TSC and their friends met up. This year we introduced the day event in line with our other TSA events. The attendees were able to share personal experiences and benefit from presentations and informal contact with expert

speakers, as well as taking part in social activities and enjoying the sights of Glasgow together.

“ ...Couldn’t believe how friendly everyone was. I wish I’d started coming years ago...”

Many families were able to join together at two separate **Family Fun Days** held at Center Parcs holiday villages in Suffolk and Nottingham. They enjoyed the facilities, socialised and handed out encouragement to each other while benefitting from expert tips from TSC Advisers on hand to answer any queries they had about living with TSC.

“Really good day, great atmosphere.... good way of meeting other families – really worthwhile..”

“You always do such a good job, just carry on!”

MOYA

Moya Bannon is nearly five. With an infectious laugh, she blows amazing raspberries, proudly shows off her forward rolls on the trampoline and can now walk a few steps on her own. Her parents Val and Stephen could list many more achievements, all of which are particularly special, when they consider what Moya has overcome to get to this point.

The odds were stacked against Moya before she was even born, when a prenatal scan identified brain and heart tumours. The diagnosis of tuberous sclerosis complex came with a warning that Moya could have severe health problems. Her mother, Val says,

“When she was little we could tell Moya was having seizures and her development began to falter. Then some intensive testing demonstrated that a large brain tumour was causing even more seizures than we could see and we were offered the option of surgery to remove it.”

Val turned to the TS Association (TSA), already a tremendous source of comfort, support and vital information. Among other things the family attended an Annual General Meeting where they listened with interest to the presentations while their children enjoyed the crèche. Val says:

“It was brilliant to chat to other parents, who had been there, seen it and done it themselves. They reassured us that things could be done for Moya and it would be OK.”

This helped them decide to go ahead with the surgery which has been an overwhelming success. Nearly two years later and signed off from the hospital, Moya has only had one major seizure – triggered by the stress of the family home flooding! – and her development has come on in leaps and bounds. In September she will attend her special school full time and also hopes to spend time at her seven-year-old brother Dominic’s mainstream school.

With other brother Sean aged two, Moya reverting to head hitting as they try to wean her off medication, life is never dull for Val. However with the TSA help, she has been able to access some assistance locally. She says:

“The TS Association is like an extra backbone –They are really supportive and I can always rely on them to find me trustworthy information quickly.”

Our **Annual General Meeting and Conference** (see pages 10-11) was well attended with 119 people braving terrible weather to learn more about TSC and the Association while they and their children relaxed together.

“More of the same – feel very grateful for all the people that work so hard to make our Association so great.”

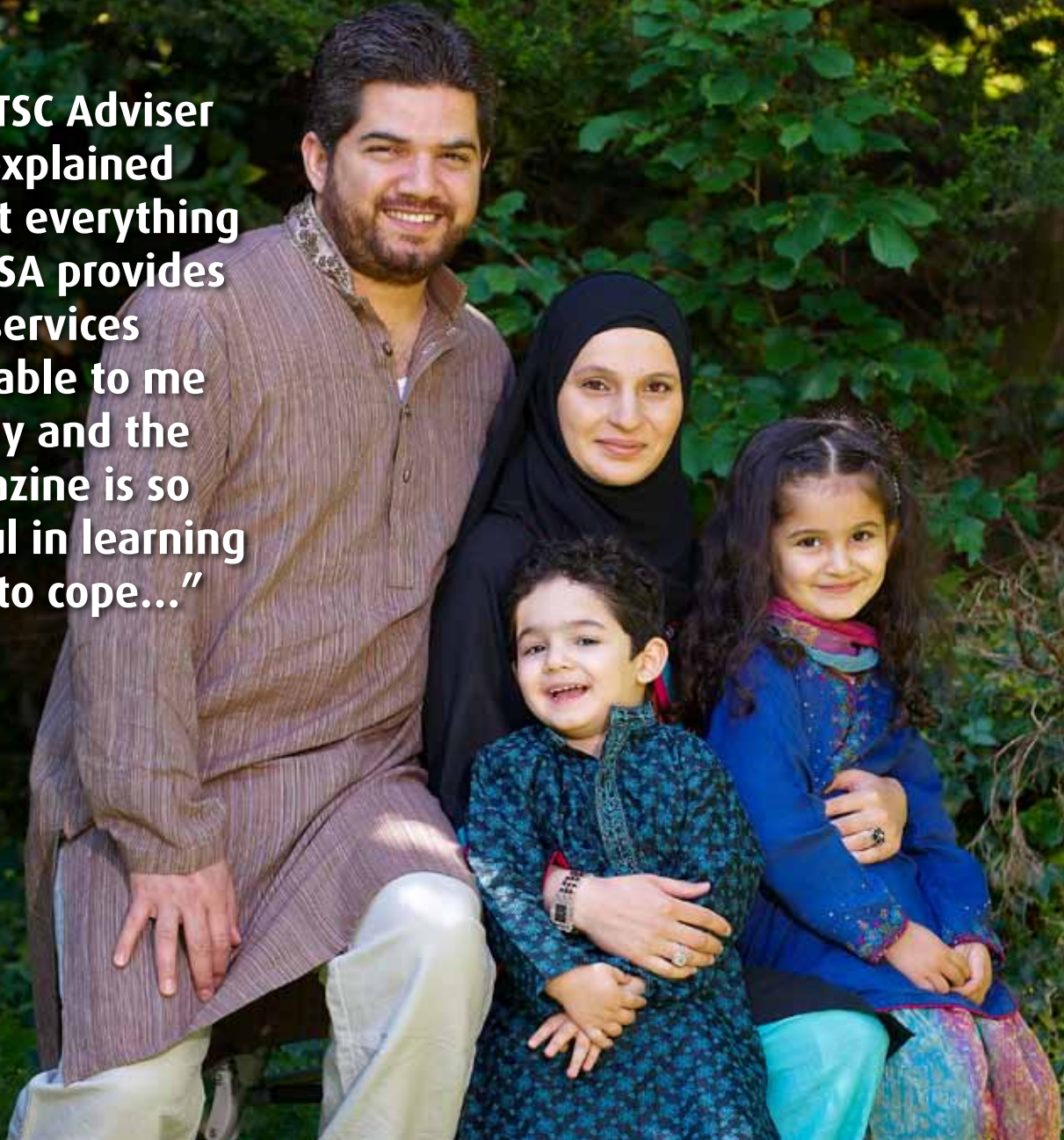
In addition to rotating the location of UK-wide events to ease travelling for people from across the country, we also arrange dedicated activities in different regions and in Scotland and have plans for events in Wales and Northern Ireland. This year’s **Scottish Get Together** was attended by 37 members and focused on legislative and health and social care issues specific to Scotland.

“The discussion groups were very good....”

Among the bi-monthly meetings of the **Merseyside Support Group** was a highly successful summer outing for this group of like-minded people who meet to discuss and provide help to other people with TSC, especially during difficult times.

Bringing Support and Advice

“My TSC Adviser has explained about everything the TSA provides and services available to me locally and the magazine is so useful in learning how to cope...”



Complex and highly variable, Tuberous Sclerosis Complex does not lend itself to a smooth passage through the education, health and social care systems. The TS Association tries to ensure that no-one seeking help is left to battle their way alone. Our team of TSC Advisers, located around the UK, are on hand to provide direct practical as well as emotional support and advice. By phone or in person these skilled and knowledgeable professionals work alongside families to guide them through their local services and provide expert information about the medical and other problems families may face.

By attending meetings with the many and varied professionals who work with people with TSC or through letters of support or referral, TSC Advisers positively influence all aspects of family lives, breaking down barriers and making sure they receive all the support they are entitled to with the minimum difficulty.

As well as connecting families with their local services, TSC Advisers also bring together people who live nearby or have similar experiences.

This year we welcomed a new member to bring our team to its full complement of four, when a highly qualified new TSC Adviser took over the role of providing regional support for North-East England, North-West England and Wales.

The scale and impact of this team's contribution testifies to their exceptional professionalism and dedication, particularly when

ISA

A memory which still haunts Navida today is watching her one-year-old son tumble to the bottom of the stairs, bouncing from wall to wall. As Navida picked up his crumpled body, Isa was silent, still under the seizure which had caused him to collapse so suddenly whilst on the landing. Navida, who was still coming to terms with his uncontrolled epilepsy and recent diagnosis of tuberous sclerosis complex, just broke down. She says,

“The doctors were trying medication after medication, even considering surgery, but nothing helped and my baby’s future looked bleak.”

In his first year Isa had developed perfectly and was already walking. Then out of the blue he fell down in the kitchen and began to make jerking movements. These fits continued and so, concerned and perplexed, Navida and husband, Adnan, kept taking Isa back to the doctors. After initial confusion with febrile convulsions, then misdiagnosis of benign, temporary epilepsy, an MRI scan identified tubers around Isa’s brain and doctors gave the devastating and confusing diagnosis of TSC. Navida says,

“At first it was too much to take in. Then I needed details. Information on the internet was confusing until I came across the TS Association site. I felt happy to get clear information and find that there were people to help us. I joined immediately and never looked back.”

Miraculously after several months of uncontrolled and ever-changing seizures one drug controlled Isa’s epilepsy so he now only has limited, mild fits at night.

Contrary to his family’s worst fears, at three Isa attends mainstream nursery, is talking well and following after his adored older sister, Falaah. Sensing the initial nightmare is over, four-year-old Falaah, once so protective of Isa, now treats him more like an irritating baby brother.

However it has been difficult for Navida and Adnan to communicate to family and friends that Isa’s illness has not and will not go away. Undaunted they continue to raise money and awareness for the TSA. Navida says,

“My TSC Adviser has explained about everything the TSA provides and services available to me locally and the magazine is so useful in learning how to cope. Fortunately we’ve not needed much help yet, but it’s great to know it’s there.”

considering that they manage to cover the whole of the UK between just four people working on average 3¼ days each.

TSC Advisers are supported by a dedicated team of senior doctors who form our Board of Professionals Advisers, meeting each year to advise on medical and scientific aspects relating to TSC, including research. Professional Advisers also make themselves available to provide guidance to individuals (via the TSC Advisers) with specific TSC-related concerns and link with the specialist medical support which is provided at the TSC Clinics in Bath, Belfast, Cambridge, Leeds and two in London.

We recognise the additional burdens which face people living with TSC and provide support through the TSA Support Fund for financial assistance to help with expenses such as travel to hospital appointments. The Janet Medcalf Memorial Fund was established to provide a break for individuals with TSC and

those caring for them. This year successful applicants received contributions towards life-enhancing treats such as a break away, a reflexology course and gym membership. These are small things which can make such a big difference.

Spreading awareness and understanding



“The most challenging thing is trying to understand Hannah’s view of the world, which is so different because of her autism.”

We have had to work harder than ever this year to weather the storm of the economic downturn. In these difficult times we are even more reliant on the commitment of our amazing, energetic and inventive supporters. We are therefore delighted to have witnessed a growth in activity from dedicated individuals and families. The snapshot of activities below shows how they have risen to the challenge.

Navida Tabassum encouraged family and friends to donate using the ‘Write Away’ personal email campaign; 350 people attended a charity ball organised by the McGhee family; seven-year-old Megan held a plant sale and Bob Abel and Greg Daniels ran 57 miles across the hills on the Isle of Mull.

To celebrate these incredible gifts of time and money we grant the Maggie Smith Challenge Award, given this year to Nick and

Jenny Budinger for money raised through sponsored bike rides and Lynne Murton for organising the first Northern Ireland ball.

A new leaflet to encourage more people to commit to regular giving was launched this year along with flyers promoting legacies and fundraising events.

Often fostered by personal links, we continue to receive much-needed donations from companies and Trusts. For example, O2e made a significant donation towards child-oriented activities; Fauchier Partners selected the TSA as Charity of the Year and we received funding towards TSC Adviser posts from The Robertson and RS Macdonald Charitable Trusts.

Through continuous review of the most effective fundraising mechanisms available we have initiated several new schemes and utilised the website to promote ideas to supporters. These

HANNAH

As a self-confessed 'unsporty' girl, with an injured knee and extremely painful shin splints, Freya Davey would not be the first person you'd imagine would decide to run a marathon. However that is what happened this year, when Freya completed the London Marathon and raised over £4,000 for The TS Association (TSA).

When you learn more about Freya, however, her motivation to put herself through that much pain is obvious.

Growing up, Freya witnessed how her older sister, Hannah, who has TSC, and their parents had to cope with countless hospital visits and operations to remove cysts or tumours from Hannah's skin, head and fingers. Family outings were always planned for a possible quick exit, should Hannah's behaviour suddenly become too challenging. Hannah's epilepsy has never been properly controlled and she progressed through school needing constant support for her learning difficulties and autism.

Hannah's diagnosis came as a complete shock; made worse by the conflicting information initially given. 24 years later Helen's first port of call for advice or information remains the TSA. She says

"The most challenging thing is trying to understand Hannah's view of the world, which is so different because of her autism."

Recently at her supported living home near her parents, the authorities only accepted the family's pleas for 24 hour support when Hannah's inability to cope on her own with daily life, began to cause her great distress. Her mother, Helen says:

"With someone alongside her Hannah can do things like cook or garden, but left to her own she forgets simple things like getting dried before dressing after a shower."

Freya, who spends much of her free time from university working with other families of disabled children, has witnessed her mother spend many years volunteering for the TSA, including a long stint as editor of Scan.

The fight to get Hannah the help she needs is ongoing but her family make sure they enjoy the good times while they last, knowing that new challenges are always around the corner.

Freya may have said, "Never again!" as she completed her marathon, but her contribution to the TSA will go on helping other families long into the future.

include Dinner4Good, a scheme which allows people to raise money by simply inviting friends around to dinner; a renewed challenge events programme featuring numerous overseas pursuits and abseiling in the UK and TSA as the official charity of the Big Fun Run across 21 events countrywide.

The story of how TSC impacts lives has been promoted using all media available. Many families kindly featured in local newspapers and radio, often linked to events like Rare Disease Day. Natalie Milham appeared on BBC South East news and the continuing story of the Davies Family was told on Channel Four in the documentary series 'Born to Be Different.' The Seager family featured in Contact a Family's 30th anniversary campaign and John Lewis company magazine. We communicated exciting advancements in medical research to the national media and through specialist publications such as the British Journal of

Neuroscience Nursing which ran informative articles on TSC.

The financial climate remains tough but we are confident that with such widespread and deep-seated support we can continue to champion the needs of people with TSC.

'I am running the 2010 London Marathon for people with Tuberosus Sclerosis as I have seen first hand how this cruel condition can affect a person and their family. ...it will be the hardest and most challenging thing I will ever do in my life but my cousin Paul's whole life has been a constant challenge, so I want to give something back...'

'I'm raising money for TSC because last year a member of my family almost died because of it.'



“At the Annual Conference we really appreciated the opportunity to speak to leading specialists in such a relaxed way that we could pop back with anything we’d forgotten over the day.”

The TS Association’s website is the first point of contact for many people, often the first glimmer of light when confronted with a devastating diagnosis and a void of information. As life progresses and the way TSC will affect each individual begins to unfold, this rapidly developing resource often remains the tool of choice to find the answers people need as well providing a link through the forums to others similarly affected and to experts.

This year we supplemented the invaluable contribution from our volunteer webmaster with a new dedicated staff member to assist with day-to-day management and undertaking the upgrades needed to accommodate ever increasing demand.

Our comprehensive review of publications has continued this year. It resulted in several key initiatives to improve the quality, accessibility and relevance of our information. We reviewed and updated, where appropriate, the factsheets on ‘Insurance,’ ‘Behaviour in Adults with TSC’ and ‘Intellectual and Behavioural Issues in Pre-School Children’ and made our ‘Guide for You’ brochure available online, a simple guide to TSC in a format appropriate for children or learning impaired adults.

Another vital role the TSA performs is to link our members to expert information from other specialist groups. Our magazine, Scan, is full of news and contributions from groups such as Kidney Research UK and at our Annual Conference information was distributed from The Challenging Behaviour Foundation alongside travel insurance advice from The Genetics Interest Group. All handouts were made available to members unable to attend.

SAM

Not many parents would tell you their son was fortunate to be dropped on his head as a baby. However for Nick and Joanne Dale, the precautionary brain scan on baby Sam following his accident showed TSC-related tumours which gave them early notice of his condition before he had any symptoms.

There is never a good time to hear such news but Nick had just accepted a new job, and committed to uproot his family far away from their support network of friends.

This move had already happened when Sam had his first seizure and was admitted to hospital. Over the coming months his parents were grateful for the introduction to the TS Association (TSA) as they became embroiled in the trauma of being registered in two different health areas and desperately needed reliable guidance on what Sam needed and when. Nick says:

“On top of the confusion from moving area came the realisation that even with a diagnosis, doctors couldn’t predict Sam’s future. The TSA were a great sounding board to double check everything we were told.”

Sam’s seizures and family life have now settled down. He is generally happy and healthy and now beginning to talk - a real little man with ‘football’ and ‘beer’, featuring alongside ‘mummy’ ‘daddy’ and ‘Is Is’ (his sister Isabelle) in his first words.

His parents still struggle with the additional burdens involved with even simple things like taking a child with TSC abroad or

knowing whether disturbed sleep is just normal two-year-old behaviour or the sign of a health problem.

However Nick’s greatest unease relates not to the everyday but to the future. He says:

“I find it hard to think too far into the future as wondering whether Sam will marry or have kids himself is too upsetting and detracts from enjoying the present.”

The next big decisions in Sam’s future are likely to be about the best education for him. Fortunately both his parents have worked in schools for many years and can draw on this experience.

Nick and Joanne have recently volunteered to join the TSA’s fundraising committee. Nick explained what motivated them:

“At the Annual Conference we really appreciated the opportunity to speak to leading specialists in such a relaxed way that we could pop back with anything we’d forgotten over the day.”

Scan draws on the medical expertise among our Board of Professional Advisers who make frequent contributions, together with regular news round-ups from TSC Advisers on issues relating to services, finances, policies and law. Members, staff and trustees also regularly contribute to Scan.

Alongside dedicated training and information events, we strive to include a learning element in all our events and gatherings. For example, this year we changed the format of our AGM and Annual Conference to allow more time for questions as well as incorporating invaluable presentations on TSA accounts, interactive workshops on topics such as genetics and kidney monitoring for the adults while children were well catered for in a dedicated professional crèche.

Scottish members welcomed the launch of our first-ever quarterly newsletter to provide updates on relevant Scottish

legislation and services. Events in Scotland included a speaker from Epilepsy Connections addressing the 2009 Outlook Weekend and a Parent/Carer Day organised in conjunction with the National Autistic Society.

“A very worthwhile event, which has given me lots of information and confidence to go on and make changes.”

“This was an ideal event as all was relevant to the current stage of my autistic son. Handouts were great allowing time to listen and understand”

“Each year I gain more and more knowledge of TSC...”

Professional Education and Influencing



“When your family is affected it feels good to do something, not just to let it take you over. It feels like you are attacking the TSC.”

It is imperative that we continue to raise awareness and provide training on TSC to health, social care and education professionals and carers working alongside people with TSC. We achieve this first by direct contact and training dedicated to the different needs of staff in residential homes, supported living accommodation, schools and other professionals such as nurses. In addition, we provided an input to policy-makers and others who influence services for people with TSC

Greater awareness and understanding of TSC and the needs of people affected has wide-ranging benefits for people living with TSC and their carers by encouraging good practice.

This year response to our training service has gone from strength to strength. We have continued with wide promotion of what we can offer, responding to numerous requests for guidance and generating additional training tools. We have developed a TSC Awareness training session feedback and evaluation form for all occasions. In conjunction with our American sister organisation (TS Alliance) we adapted their school training pack for the UK with advice from a TSA Professional Adviser and specialist teacher colleague.

Attendance at our training sessions is excellent, boosted by the fact our TSC Advisers normally travel to the recipients' place of work, individually tailor each session to the audience and individual context and provide certificates of attendance and appropriate, practical training resources.

This year over 85 teachers, nurses, support workers, managers

AOIFE

Like all parents of a child with TSC, Julie had no choice but to become a near expert so she can help her second daughter Aoife who was diagnosed at nine months old. This knowledge might have helped throughout Julie's life because doctors treating Aoife soon spotted the growths on her mother's nails, white skin patches and a facial rash and established that Julie, too, has TSC, albeit mildly. Reflecting on the moment she realised that TSC was genetic, Julie said:

"I came out of the room feeling totally destroyed and guilty."

As time has gone on Aoife's dad Alan has actually felt more helpless than Julie who now has to focus on fighting on her own behalf as well as her daughter's.

Fortunately doctors advised them to look only on the TSA website if they wanted information that was guaranteed to be accurate, sensitive and practically helpful. In this way they made contact with their TSC Adviser and after an initial three hour conversation, Julie knew she had found the support she was going to need. She says:

"If I have any concerns or question, my first point of contact will be my TSC Adviser. She even comes with me to appointments. She is such a lovely person, who genuinely cares - I don't know how I'd have coped without her."

and head teachers received individualised training about TSC at venues as diverse as Care Homes, Epilepsy Nurse Specialist centres, Nurseries, Primary and Specialist Schools and Supported Living accommodation.

Feedback from these sessions testifies to the quality of the training and its positive impact on the way the teams are able to address better the needs of people with TSC. Specific comments included:

"Excellent booklets and handouts....good in practical terms in how to help patients with TSC."

"Lots of great information, very well presented, this will be incredibly useful for our team and the person we support."

In spite of fears that she may never walk, at two-and-a-half Aoife is developing well physically. Sometimes however, overcome with frustration at being unable to communicate, Aoife's boundless energy can be misplaced into aggressive behaviour. Julie feels particularly sad for her older daughter Niamh who is desperate to play with Aoife but is often rejected.

Recently Julie appreciated the chance to attend a meeting of MSP's in Edinburgh to make them more aware of the needs of people with rare conditions like TSC. She says:

"When your family is affected it feels good to do something, not just to let it take you over. It feels like you are attacking the TSC."

Meeting people with other rare conditions, many without an organisation like the TSA to turn to, helped reinforce Julie's positive outlook that she and her family should deal with each day as it comes and never take anything for granted.

'Refreshing awareness, symptoms to look out for, very informative... will plan activities differently.'

Increasingly we have channelled our influencing into collaborations with other specialist groups where we share common aims and needs. This is an effective use of our resources and generates a greater strength of voice. This year the TSA and the Genetic Alliance UK began working together on a Department of Health funded project that will assist in creating networks of professionals with expertise in rare neurological conditions. For us the aim of the project is to improve access for people with TSC to healthcare professionals with experience and understanding of TSC.

REBECCA

Rebecca Parry and her family will never forget the dreadful day when, after months of driving an hour from home every other day for dialysis, they were told there was a donor kidney available. Dreadful because without the results of recent tests on a newly discovered brain tumour, the doctors wouldn't risk the transplant operation.

Rebecca's mother, Liz, did what she had done countless times before. She says:

"My immediate response was to call the TSA who told me everything I needed to know. As always it was such a relief to have someone there who really knew what they were talking about."

Days later, the news that her tumour wasn't growing came too late for that kidney. Fortunately, a week before Rebecca's sister Emily was due to donate her kidney, Rebecca had a successful transplant from an unknown donor and Emily's kind offer remains in reserve.

Rebecca's kidney problems came at 18 after a childhood punctuated by TSC-related health problems. Rebecca has battled with epilepsy – now controlled; dyslexia - now she can spell better than most of her friends; facial rash – removed with laser surgery and successfully completed her education with an NVQ2 in Hospitality and Catering and NVQ3 in Massage, Anatomy and Physiology. Liz says:

"Rebecca is an absolute trooper. She'd always go down to theatre smiling and coped brilliantly with her studies even when her kidneys were failing."

At 28 Rebecca faces new problems. She cannot count on everyone showing the same support and understanding as her friends and family. Even her successful laser treatment has a downside. Rebecca says:

"Now I look like there's nothing the matter with me. Several employers have reacted unsympathetically when I've been ill, even though I've mentioned TSC on applications. Sadly people seem scared of words like 'tuberous sclerosis' so they avoid dealing with them. "

Although Rebecca's immuno-suppressant drugs are now affecting her stomach, she is planning her future undaunted. Focused initially on getting herself well again, Rebecca wants to do a work-based retail qualification linked to her part-time work at Oxfam.

Liz is comforted that Rebecca's future will also include the TSA. Liz says:



"The TSA is a wonderful organisation, still keeping its personal touch even when it has grown. I don't think I'd have survived without them."

In a Washington conference in 1999 Frank Collins, head of the human genome project, spoke to TSC researchers and families about the discovery and cloning of the TSC genes. He paraphrased Winston Churchill saying: "This is not the end, it is not even the beginning of the end but it is perhaps the end of the beginning". Now we can say: "This is the beginning of the end".

This has been the year of the mTOR studies, which featured prominently at the inspiring TS Alliance Conference in September

David Franz leads a research group in Cincinnati that has published papers showing both Sirolimus and Everolimus can shrink subependymal giant cell astrocytomas (SEGAs – a type of brain tumour): potentially a much better treatment than surgery or surgery alone. The optimal use of mTOR inhibitors in the treatment of SEGA needs a large randomised controlled trial (RCT) to work out how best to use them. An RCT shows the balance of benefit against risk of any particular drug treatment strategy. Just such an international study is underway: the EXIST-1 trial (now fully recruited), with Finbar O'Callaghan in Bristol as the UK lead.

Other exciting studies in Cincinnati and the UK (Tesstal Trial) have shown that mTOR inhibitors shrink angiomyolipomas (AMLs – a type of kidney tumour). The EXIST-2 study (UK centres at Nottingham, St George's in London, Brighton and Cardiff) is the RCT that will show their optimal place in the management of this major TSC problem. Recruiting is planned to continue until December 2010 unless we get a clear answer before then.

Several studies are focussed on the benefit of mTOR inhibitors in Lymphangiomyomatosis (LAM - a rare lung disease). The early signs are very encouraging. This is fantastic; if their early promise is fulfilled mTOR inhibitors will be the first really effective treatment for LAM.

The trials in SEGA, AML and LAM have already shown that in some people mTOR inhibitors help the facial rash (the angiofibromata), epilepsy, autism and possibly learning disability. Further research is needed to find out how many people might benefit, at what dose, when to start treatment - and whether the benefits outweighs the risk of side effects.

Back in the laboratories researchers are looking for other types of drugs that will act on the mTOR pathway and other pathways that may also be important to target in TSC. Their research will eventually culminate in clinical trials as well.

We continue to liaise with, learn from, and add to the combined voice of TSC organisations. This year we were represented at a European meeting on patient participation in clinical trials. We also learned more about the differences and commonalities between TSC and a variety of other genetic behavioural conditions at the SSBP conference, which paved the way for ongoing collaboration in a study of lifelong health and wellbeing.

We need more social sciences research and natural history research into how TSC affects people and their families and the need for community support. This is so national health services can plan funding for medical and social care.

Despite severe financial handicaps we have made huge and exciting progress in the past two decades.

In a Washington conference in 1999 Frank Collins, head of the human genome project, spoke to TSC researchers and families about the discovery and cloning of the TSC genes. He paraphrased Winston Churchill saying: "This is not the end, it is not even the beginning of the end but it is perhaps the end of the beginning". Now we can say: "This is the beginning of the end".

Fundraising



Acknowledging our Supporters

Thanks to Our Supporters!

Many thanks to everyone who has worked so hard this year to raise money for the Tuberosus Sclerosis Association. We are extremely grateful for every single contribution as everything adds up to help us support people with TSC.

Only a small selection can feature in this Report but our thanks go to everyone who organised or took part in the wide variety of fundraising activities and events held this year to help people with TSC.

Thanks to our Sponsors!

We have received financial support from a range of individuals, corporate and charity sponsors. In addition to recognising each donation in our newsletter Scan, we would like to express our thanks to each of you once more. A selection of this year's major sponsors from 1st April 2009 to 31st March 2010 has included:

- 02e
- RS Macdonald Charitable Trust
- The Roald Dahl Foundation
- Robertson Trust
- Garfield Weston Foundation
- James Ellis Charitable Trust
- Felicity Wilde Charitable Trust
- James Weir Foundation
- Hugh Fraser Foundation
- AJ & KM Barnett Charitable Trust
- Adint Charitable Trust
- Thames Wharf Charity
- Sir Julian Hodge Charitable Trust
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- John Jarrold Trust
- The Company of Actuaries Charitable Trust Fund
- Souter Charitable Trust
- EA Chamberlain LLP
- Chapman Charitable Trust

... our thanks go to everyone who organised or took part in the wide variety of fundraising activities and events held this year

😊 = Target fully achieved with the year

😐 = Target partially achieved within the year

Support Services

IMPACT 2009/10

This year we have managed either to achieve or to make good progress towards everything we had planned. Our targets were as follows:

- **Continue to update and develop the website** 😊

A major upgrade was performed this year by our volunteer webmaster as well as a peer review to ensure information is accessible and easy to find.

- **Induct and train two new staff members (Fiona Clark to replace TSC Adviser Hilary McGlynn and Ben Kobus in a new post of Communication and Research Administrator)** 😊

This was completed for both posts.

- **Develop the School training pack** 😊

This has been completed and is now in use and receiving high acclaim.

- **Pilot a Scottish newsletter** 😊

Four newsletters have been completed to date, following positive feedback from the pilot. This initiative allows us to manage the fact that the health, education, local government, housing and social work are governed by the Scottish Government and that services, support and legislation therefore differ greatly to those in England and Wales.

- **Create a kidney monitoring fact sheet in conjunction with the research team** 😊

This fact sheet was included as a pull-out resource in the special edition of Scan and received highly positive feedback.

- **Develop a "What you need to know about my condition" factsheet based on the Neurological Alliance template that TSA staff helped develop** 😊

This has been completed as a template which is then individualized by each member.

- **Arrange a gathering for members in Wales** 😐

This is now planned for late 2010 depending on interest.

- **Arrange a gathering in Northern Ireland** 😐

This took place in June 2010.

- **Arrange a family gathering in or around London** 😐

Initial planning to determine what will work best for families is underway.

Our plans for next year and onwards are to:

- Continue to contact members requesting interest in a Wales gathering
- Contact members requesting interest in a London gathering
- Review and develop our existing residential training programme
- Update the nurses leaflet and continue to review all TSA publications
- Update the genetics fact sheet
- Perform a thorough review of the TSA website
- Continue to theme editions of our member magazine ,Scan, with mental wellbeing planned for August and respite/ holidays planned for subsequent editions
- Provide a parent/ carer study day at the NI International Research conference in Belfast
- Continue to work alongside the Outlook group to develop their event
- Provide a Family Day in the North of England
- Continue to liaise with TSC Specialist Clinics and the TSA Board of Professional Advisers
- Continue to liaise with other specialist organisations to develop the teachers pack with practical strategies for teachers who support children with TSC
- Organise a Family day in Wiltshire in June

Research

IMPACT 2009/10

In addressing this year's objectives we have achieved the following:

- **To take forward plans for a topical treatment for facial angiofibromas**

Awaiting results of pilot study undertaken in Texas. Early experience with EXIST2 in the UK shows promising effects of systemic treatment. 😊

- **To facilitate new studies of the treatment of TSC tumours with mTOR inhibitors**

We continue to actively recruit for the EXIST 1 & 2 studies of Everolimus for the treatment of AML and SEGA. 😊

- **To set up a training fellowship in TSC research or a project grant in a TSC priority area**

Initial groundwork undertaken; due to economic climate rescheduled for next year. 😐

In addition:

- A new paper has been drafted incorporating the neurocognitive aspect of the TESSTAL trial 😊
- Planning has commenced to collaborate in a controlled trial of an mTOR inhibitor for neurocognitive aspects of TSC 😊

For next year our priorities are to:

- Award first TSA training fellowship
- Induct and train the new Research Administrator
- Finish planning for the International Research Conference in Belfast September 2011
- Contribute to the UK National Strategy for Rare Diseases
- Support ongoing research into the natural history of TSC
- Promote research into neurocognitive problems and the setting up of appropriate provision
- Promote social sciences research

Fundraising and Awareness Raising

IMPACT 2009/10

We have achieved all our targets for 2009/10 as follows:

- Continue to research and implement new fundraising ideas 😊

New fundraising ideas initiated this year include the following:

- Write Away campaign and flyer
- Fundraising Events Calendar and flyer
- New fundraising posters – ‘Molly’ poster & blank event poster
- Update and reprint of Appeal Brochure ‘Just a few reasons to help’
- Inspirational Fundraising campaign and flyer
- University Challenge
- Collection box brigade for supermarket/garden centre collections
- ‘Bucket drop’ brigade for specific events
- Ebay ‘Dump the junk’ campaign
- Legacy campaign and flyer updated
- Regular Giving Brochure

- BIG FUN RUN
- ‘Dinner4Good’
- Abseils

Create a Regular Giving brochure 😊

This has been successfully produced and is now in use.

Continue to work with other NGOs, including umbrella organisations, to help build awareness of TSC. 😊

This was achieved through the following activities:

- Contact a Family podcasts on their website – we sourced two families with TSC
- TSA representation with Genetic Alliance UK at the Rare Diseases Event at The House of Commons
- NORSACA Asperger’s Conference – Liaison with Professor Digby Tantum
- Attendance at the Innovative Therapies & Rare Diseases Conference, London
- The Association of British Neurologists (ABN) organised a fun run for neurologists to raise awareness of neurological conditions. We worked with them and provided TSA-branded materials
- Rare Disease UK – We have been in regular contact with this organisation. We attended the Scottish Parliament reception in March 2010 and were represented at the corresponding event in Belfast and supported publicity around the event in Cardiff
- Brain Tumour UK has linked their website to ours.

Further define the PR strategy 😊

This has been achieved through meetings with staff and supporters with relevant professional expertise. A preliminary PR strategy has been created and is being further refined.

Promote a Legacy Campaign 😊

An updated Legacy flyer and codicil form were mailed to TSA members and supporters. There is also ongoing promotion on our website and member magazine, Scan.

Our priorities for next year are:

- Our first priority is to continue to increase ‘Friends & Supporters’ of the TSA by working to the operational plan
- Our second priority is to seek to increase income from Trusts
- Our third priority is to continue to expand the programme of fundraising opportunities.

Principal aims and activities: public benefit

The Trustees confirm that they have complied with their duty, under Section 4 of the Charities Act 2006, to have regard to the Charity Commission's published public benefit guidelines in determining the activities undertaken by the Association.

The Association has three main objectives

- To support individuals affected by Tuberous Sclerosis Complex, together with their families or carers
- To encourage and support research into the causes and management of TSC
- To provide education, publicity and information to promote awareness of the problems caused by the condition

The benefits of our activities are described in our Impact Report and relate directly to our objectives. Our Support Services team have run effective training, research update and family/carer events, issued publications and provided individual and group support to families and carers. We have continued to support appropriate research projects. Anne Carter, as Head of Appeals and Publicity, has secured publicity for our activities in many helpful forms and has thereby increased awareness of TSC and our rôle in improving outcomes.

Financial report

It is very easy for the interpretation of the TSA's finances to get lost in translation. In what has been another very difficult year for all charities, the TSA's total income for the year fell by some £140k (28%) to £356,562 (2009: £495,758). Although this gave a technical surplus of £28,682, in real terms there was a deficit of some £200,000; the difference is caused by timing differences on research expenditure under the somewhat artificial accounting requirements of SORP.

We have to budget for deficits of around £100,000 in the next couple of years, to maintain services at the current levels.

Income from the TSA's investment capital originating in the H I Leech bequest in 1993 similarly fell to £86,705 (2009: £110,081). As noted below, the Trustees have arranged for the investments to produce a relatively stable income; a stellar performance by our new investment advisers resulted in the value of the investment portfolio rising by £969,366 (2009: loss of £748,071).

Financial Statements

The summary financial statements on the facing pages have been extracted from the statutory accounts of the Association, which were approved by the Trustees on 5 July 2010. The summary financial statements may not contain sufficient information to allow a full understanding of the financial affairs of the Association. For further information, a copy of the statutory accounts and the auditors' unqualified report on these accounts should be consulted. Copies of the statutory accounts will be filed with the Register of Companies and the Charity Commission. Copies may be obtained from the Head of Appeals & Publicity.

On behalf of the Trustees

Jonathan Swainger, Treasurer
Tuberous Sclerosis Association

Summary Financial Statements

Independent Auditors' statement to the Trustees of Tuberous Sclerosis Association

We have examined the summary financial statements of Tuberous Sclerosis Association.

This statement is made solely to the Trustees, as a body, in accordance with the terms of our engagement. Our work has been undertaken so that we might state to the Trustees those matters we have agreed to state to them in this statement and for no other purpose. To the fullest extent permitted by law, we do not accept or assume responsibility to anyone other than the Trustees as a body, for our work, for this statement, or for the opinions we have formed.

Respective responsibilities of Trustees and Auditors

The Trustees are responsible for preparing the summary financial statements in accordance with the charities SORP.

Our responsibility is to report to you our opinion on the consistency of the summary financial statements with the full financial statements and the Report of the Trustees. We also read the other information contained in the Annual Report and consider the implications for our report if we become aware of any apparent misstatements or material inconsistencies with the summary financial statements.

Basis of Opinion

We conducted our work in accordance with Bulletin 1999/6 'The auditors' statement on the summary financial statement' issued by the Auditing Practices Board for use in the United Kingdom.

Opinion

In our opinion the summary financial statements are consistent with the full financial statement and the Report of the Trustees of Tuberous Sclerosis Association for the year ended 31 March 2010.

PKF (UK) LLP Registered Auditors, Birmingham - 23 August 2010

Summary Balance Sheet

as at 31 March 2010

	2010 £	2009 £
FIXED ASSETS		
Tangible assets		
Investments	2,964,608	2,005,841
Total Fixed Assets	2,964,608	2,005,841
CURRENT ASSETS		
Debtors and prepayments	8,316	78,925
Cash at bank, in hand & held by investment managers	231,825	308,491
Total Current Assets	240,141	387,416
CURRENT LIABILITIES		
Research grants due within one year	(176,097)	(314,115)
Other creditors due within one year	(103,540)	(119,277)
Net Current Assets	(39,497)	(45,976)
Total Assets less Current Liabilities	2,925,111	1,959,865
OTHER LIABILITIES		
Research grants due beyond one year	0	(32,802)
NET ASSETS	2,925,111	1,927,063
FUNDS		
Unrestricted funds		
General funds	27,243	38,906
Designated Capital Fund - HI Leech Bequest	2,732,642	1,658,924
	2,759,885	1,697,830
Restricted funds	165,226	229,233
TOTAL FUNDS	2,925,111	1,927,063

Summary statement of financial activities

year ended 31 March 2010

	2010 £	2009 £
INCOMING RESOURCES		
Incoming resources from general funds:		
Voluntary Income		
Legacies	2,000	10,000
Grants	3,000	4,200
Subscriptions from members	17,924	11,900
Other income	230,111	275,055
	253,036	301,155
Activities for generating funds	15,975	43,229
Investment income	86,705	110,081
Incoming resources from charitable activities:		
Education	0	33,269
Family care & support	846	8,024
TOTAL INCOMING RESOURCES	356,562	495,758
RESOURCES EXPENDED		
Cost of generating funds:		
Cost of generating voluntary income	28,162	28,078
Cost of fund-generating activities	10,297	22,590
Investment management costs	11,150	10,204
Charitable activities:		
Research	22,430	164,838
Promoting awareness	36,837	37,557
Education	31,029	82,479
Family care & support	169,352	171,052
Governance costs	18,624	18,942
TOTAL RESOURCES EXPENDED	327,879	535,740
NET INCOMING RESOURCES	28,682	(39,982)
Transfers between funds	0	0
Gains (losses) on investment assets	969,366	(748,071)
NET MOVEMENT OF FUNDS	998,048	(788,053)
Funds brought forward 1 April 2009	1,927,063	2,715,116
Funds carried forward 31 March 2010	2,925,111	1,927,063

Support



Further information on Tuberous Sclerosis Complex and the work of the TSA can be obtained from Diane Sanson, Support Services Co-ordinator

The Tuberous Sclerosis Association is a Company Limited by Guarantee Registered in England & Wales No.2900107
Registered Charity No. 1039549

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