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A-T Society News

In this high-flying issue

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AT society
awareness research care

Ataxia-telangiectasia is a rare, genetic, neurodegenerative disease. It starts in early childhood and affects many parts of the body causing severe disability.

The A-T Society was established in 1989 and is committed to helping, supporting and advising families affected by A-T. While they may face more challenges than many, people with A-T have lives to live, and the Society's aim is to ensure they have the support they need to live them to the full. We do this through funding research, providing information, practical support and financial assistance, working to improve clinical management and raising awareness.

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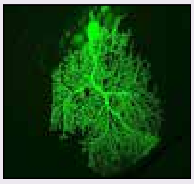
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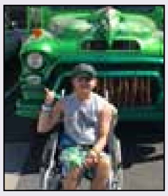
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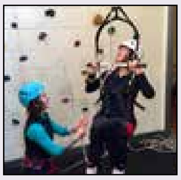
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Editor's comments

Many thanks to all contributors. The copy date for the next issue is 1st October 2018. Please send comments, ideas, articles and pictures to:

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Cover picture: Ben at the A-T Activity Weekend

Just William

William Davis, chief executive of the A-T Society, looks back on three inspiring and uplifting events.

Last autumn, I was lucky enough to attend three amazing and inspiring events. In September, my colleague Anne and I shared a tiring but exhilarating weekend in the Lake District with 11 of our young adults and their carers. Then October brought not just the Society's annual Family Weekend – always a high-point of my year – but a week later I was able to take part in the A-T Children's Project's Caregivers Weekend in Chicago.

The Activity weekend is covered in more detail on page 14. For me it was one of the most memorable and uplifting events I have been on in all my time with the Society. Many things stick in my mind. A group of people with A-T paddling out to an island in Lake Windermere then sitting around on the ground having a picnic lunch, chatting and laughing – like any other group of young people. The amazing sight of people ignoring major disabilities and fatigue, willing themselves up a climbing wall then going back for more, surprising themselves as well as those watching. People arriving shy and nervous and leaving with confidence and assurance.

I have always been impressed by how much people with A-T achieve in their lives, despite all the obstacles put in their paths, but the spirit and energy of this group gave even my expectations a real kick.

The Family Weekend (*see page 12*) always has a real buzz. Every year we wonder if as many people will want to come this time. Every year around a third of all families with a diagnosis of A-T pitch up to enjoy the company and support of old and new friends, and the fact that for one weekend a year having A-T feels normal. Highlights this year included Ebony talking about taking part in The One Show's Rickshaw Challenge (and sharing her joy at the unexpected reunion with the rest of the team), Becky opening the weekend and then speaking later about her life, Kayleigh talking about her work and Jilly about her cake-making.

A week later, I was taking part in the A-T Children's Project's Caregivers Weekend. This was a coming together of parents and carers from all over the USA and from Canada but without the people with A-T they care for. What most struck me here, beyond the strong sense of friendship and community, was the positive and creative approach to life and solving problems shown by so many people. Bringing up a disabled young person in a country so huge and spread out, with no real welfare state, is a real challenge, but one that these people are rising to with real energy and ingenuity. And one of the groups I sat with rocked with laughter as they talked about how they looked after their own needs as carers.

What these three events shared was a strong focus on the possibilities of living full and active lives with A-T. We all too often slip into talking about life with A-T in negative terms: neurodegeneration; immunodeficiency; life-limiting; rare disease. Even more neutral words like disability or condition imply that life with A-T is somehow second-best.

But every life is priceless and unique for what it is, not what it isn't. Lives with A-T may be shorter and more challenging than those of many people with a full complement of ATM protein, but they need not be any less rich or full of joy and achievement.

> **Lives with A-T may be shorter and more challenging than those of many people with a full complement of ATM protein, but they need not be any less rich or full of joy and achievement.**

It is our responsibility as individuals both to live our own lives to the very fullest extent possible and to help others to do so, too. And that's what the A-T Society is about. People with A-T, often with the active support of their families, are showing us how to do this, in very different and individual ways. The A-T Society's Live well with A-T programme, which we are currently developing, aims to recognise and celebrate these achievements and to find new ways to encourage and support people in living their lives their way. Let's find ways to ensure we can all celebrA-Te life!

Purkinje cells in the lab?

A team in Italy is embarking on a project, funded by the A-T Society, which aims to make a huge step forward in A-T research.

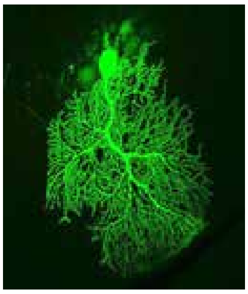
The neurological (coordination) problems in A-T are caused by cells in the brain dying off in the absence of the ATM protein. Particularly affected are cells in the cerebellum, the coordination centre of the brain, called Purkinje cells.

The big question scientists have been asking since the ATM gene was discovered, is why in the absence of ATM these cells die while other cells in the brain don't. To answer this, it would be extremely helpful to have some Purkinje cells to study. The problem is, you can't carry out experiments inside someone's brain, and you can't cut bits off to experiment in the laboratory.

Now though, stem-cell technology is offering a way forward. Stem cells are cells that have the capacity not just to produce copies of themselves, like most cells, but to produce different types of cell. The cells of an early embryo have the ability, ultimately, to produce all the cells required to make the body. However once the embryo has fully developed, stem cells become much rarer and mostly produce a limited range of cell types.

Scientists have for some time been able to take ordinary cells, say skin cells, and reprogram them to become stem-cells, with the rather unwieldy name of induced pluripotent stem-cells (iPSCs). More recently they have been able to induce these to become neural (brain) stem cells. However using these to generate brain cells, and in particular Purkinje cells, which are large with a complex tree-like structure, has proved difficult. Recently though, scientists in Japan did this for the first time.

Now the A-T Society is funding a project to produce Purkinje cells from the cells of people with A-T. The team at the FIRC Institute of Molecular Oncology in Milan, is led by Marco Foiani and Domenico Delia. They have an excellent track record in A-T research and have already produced iPSCs and neural stem from skin-cells of people with A-T. Now they aim to use these to create Purkinje cells in the laboratory.



Purkinje cell (Photo credit: CIL:39952).

If they succeed they will open the way to studies to understand what is happening in the Purkinje cells of people with A-T and to the possibility of using the cells to screen for drugs that could protect the cells. The team themselves already have plans for a series of tests once the cells are developed.

Penny Jeggo, chair of the A-T Society's Scientific Advisory Board, said: "Why Purkinje cells die in people with A-T when other neurons don't is probably the biggest question facing us as we look to develop treatments. This is a really exciting project which if successful will help answer that question and help us develop effective treatments for one of the most damaging features of A-T."

A new approach to gene therapy for A-T

In the past few years significant advances have been made in gene therapy, and a number of new therapies are being trialled or have been licensed for genetic conditions. For a condition like A-T, where cells die off because the ATM gene doesn't function to produce the ATM protein, the idea of introducing a working copy of the gene into the affected cells is very attractive.

But there are many challenges to be overcome to develop an effective gene therapy for A-T. One of the major ones is the size of the ATM gene. In gene therapy, the new gene has to be transported to the cells where it is needed and this is usually done using a specially engineered virus. The ATM gene, though, is too big for most viruses to carry and even the few that can become very difficult to use with such a large 'payload'.

However, James Dixon, a researcher at the University of Nottingham, has developed a non-viral vector, which does not have these capacity limits. Using techniques that were developed to treat cystic fibrosis, he aims to develop a version of this vector and delivery system with the ATM gene.

The A-T Society has given a grant to fund the creation of this vector and to test if it is able to correct the ATM protein deficiency in cell-lines from people with A-T. This is a short-term proof of concept project, which could pave the way towards trialling the vector in animals and perhaps ultimately in humans.

Penny Jeggo, chair of the A-T Society's Scientific Advisory Board, said: "This is potentially a very exciting approach to gene therapy from a committed research team. We are delighted that the Society is funding testing of an imaginative new approach to treating A-T."

Collecting and sharing A-T data

With something like three cases per million of the population, A-T is classed as an ‘ultra-rare disease’ (though of course we would not call it a ‘disease’). In science, a lot of information is gained by gathering data. In medicine this would involve gathering information about how many people have the condition, how it affects them, how it changes, how it is treated, how it relates to a person’s genes and so on. As A-T is so rare, collecting data is particularly important and there are currently two important projects gathering data:

International A-T Registry

Established and led by the A-T Society, this registry gathers clinical data, which covers both basic information about the person with A-T and more information about the areas affected by A-T (including immunology, neurology, lung-health, posture, skin and cancer).

A major aim of the registry is to build up an accurate ‘natural history’ of A-T – a detailed and evidenced description of the condition and its course. This is a valuable tool to help assess treatments or the effectiveness of clinical trials.

Data can only be input by doctors and centres registered with the Registry, like the centres at Nottingham and Papworth. However it is important that as many people’s data as possible is recorded. Please ensure you have signed and returned your consent form to enable them to do this. For more information email admin@atregistry.eu.

Global A-T Family Data Platform

Set up by the A-T Children's Project and overseen by a group of families and experts from around the world, the Global A-T Family Data Platform collects information from people with A-T or their families. A key part of this project is the collection of data from genome sequencing. Participants can send a sample of their saliva, which will then be sequenced, i.e. all the genetic information carried by their cells will be read. This will produce enormous amounts of data as each individual’s genome contains 2.9 billion base pairs (the basic units of DNA), which amounts to about 725 megabytes. Nevertheless the hope is that by combining the genomic data with clinical and other data for that individual, we may be able to understand why different individuals with A-T are affected differently and this in turn may provide opportunities for new treatments.

We would encourage anyone with A-T to sign up to the platform. You can do this by visiting www.atfamilies.org.

While the two projects are collecting separate data, it would be powerful to bring them together. For the moment this is not possible as each database has its own protocols and they are subject to stringent data protection and ethical regulations. However the two projects are already in discussion about how this can be done and how data from individuals in different systems can be matched up. To do this would require changing the protocols of the two projects and the explicit consent of every individual whose data is shared. So there is still a way to go on this.

ATTeST trial: nearly, but not quite

We reported in the last newsletter that the ATTeST trial of the EryDex System had begun last summer in a number of centres. It is therefore frustrating to report that it has still not started in the UK.

The trial began in other countries last summer. However, as we previously reported, the UK centre at Nottingham needed to recruit a Clinical Research Fellow to help run the study and this was taking time. Since then, all the organisational issues have been sorted and a Clinical Research Fellow, Dr Jeyanthi Rangaraj, appointed. Some 20 families have been in contact with the team and 16 children identified for screening, which was due to start in January.

However the US Food and Drug Agency put a temporary stop on recruitment to the trial allowing only patients already enrolled to continue treatment. This was because some samples taken prior to infusion into patients were found to be contaminated by bacteria. To be clear, it was only samples for testing that were involved, and only a few of these. There is no suggestion that any of the participants could be affected, and indeed children already on the trial are being allowed to continue. However, until the issue is fully understood and resolved, all new recruitment is suspended.

The trial organisers have investigated this thoroughly and reported back with proposals to modify the sampling procedures. However, they are still waiting for permission to proceed.

The A-T Society’s chief executive William Davis said: “The safety of participants in the trial is our priority. However there is no suggestion that the Erydex System itself is affected or that there is a risk to those using it. We urge the authorities to come to a rapid conclusion and let the trial proceed as planned.”

Remembering Ian, Cleo, David and Becky

It is with great sadness that we announce the passing of four of our children and adults with A-T: Cleo Brady, Becky Goodwill, Ian McInnes and David Hart. They will all be very much missed by their family, friends and pets and have left a huge hole in their hearts. Here though, we remember four amazing lives.

Ian McInnes, who had variant A-T, was for many years a trustee of the A-T Society. He also helped run sessions for young adults with A-T in the past and spoke at many family weekends about his extensive travels and other activities. His positive attitude, determination and outgoing personality meant that he was a very well-known character in the A-T community.

Ian lived a very active independent life in Edinburgh where he was involved in a number of different voluntary roles and was a staunch and determined campaigner for the rights of people living with disabilities. He was very proud to have been chosen as a torch-bearer for the 2014 Commonwealth Games in Glasgow. He was also a keen supporter of Scottish Rugby.

Ian's story was published by the *Scotsman* newspaper in 2012. The article captured the true spirit of Ian, his attitude to life and, in his words, his "sheer bloody-mindedness" to defy the odds. We've published some extracts from the newspaper article below:

Despite being confined to a wheelchair, Ian has refused to let his condition stop him living life to the full, taking part in two 12,000ft and two 10,000 parachute jumps and even climbing the Great Wall of China.

Ian told the Scotsman: "Everyone thought I'd be dead by the time I was 25. I like taking risks - you're not alive if you don't. I've done a lot more than able-bodied people and the reason I'm still here is because of sheer bloody-mindedness."

As well as travelling over the French Alps for his Duke of Edinburgh Award, he also visited Canada and China where he travelled along the Great Wall. Ian said: "During the trip I used 30 different forms of transport, from planes to piggyback. I was determined to walk along the wall, so two of the people I was with stood on either side of me and helped me take a few steps. It's something I have always wanted to do and I'm happy I did it. Living with a condition like mine can be frustrating but you have to make the best of things.

"When people find out what I have and then the life span I was supposed to have they can't believe it. I have made a hobby of proving doctors wrong. It's something that I have done since I was born and I don't intend to stop now."

Cleo Brady's mum, Gemma, writes:

"Cleo was such an amazing soul. She was funny, witty, kind and generous. She loved her family and friends (and her dog, Missy). She showed true courage and determination this past year and she did it without ever complaining. We miss her so much every single day, life is empty without her."

David Hart, son of the A-T Society's counsellor Helen Hart and her husband John, lived a remarkably full and active life.

Like his brother Andrew, who died some years ago, David had the IVS40 mutation which occurs in more than 20 people with A-T in the UK and Ireland, and gives a slower developing 'variant' form of the condition.

In spite of his disabilities, David had a wide range of interests and was involved in many different activities, so much so that he was honoured with an obituary in the *Scotsman* newspaper. You can read extracts of the article here:

As a boy and young teenager, David enjoyed hillwalking with his family and developed a love of nature – especially birdwatching – that he was able to pursue as an adult. In his early teens he climbed most of the big peaks in the Lake District, including Skiddaw and Helvellyn. When he was in his late teens, however, he was diagnosed with A-T.

After finishing school, David went to train as a nurse in Inverness, but the initial effects of A-T, which affected his balance, meant that he could not complete the training. He continued on the path into a caring profession through an access to social work course, but the effects of A-T increased and it was clear that he would not be able to take on employment. However, he used the training in a variety of ways to support others who had to deal with restrictions on their lives.

When David was in his early 20s he began to attend Firrhill – an exemplary municipal day centre that offered opportunities to people with physical disabilities to be part of a busy community, follow interests and learn new skills. During the 25 years David attended Firrhill he worked on reception; became involved with committee work, and represented people with disabilities on a range of external bodies. He participated in various working groups set up by the local authority looking at wider access for people with physical disabilities, partnership groups and creating a plain English guide for service users and providers. Firrhill gave David a sense of identity within the working world and opportunities to use his gifts and make a difference.

David enjoyed writing. He edited The Firrhill Flyer and latterly was "a thorough and prolific reviewer" for Euan's Guide, the online website for disabled access reviews from disabled people and their friends and families. Through his involvement in creative writing, David came into contact with Artlink, the organisation which works to increase opportunities to take part in the arts for those who experience disadvantage or disability in the East of Scotland. In 1995 David joined the board of Artlink.

In spite of his difficulties, David was also widely travelled, having visited many of the countries of Europe at one time or another and getting as far as South Africa, following his love of cricket. David was stoical in his final illness, pancreatic cancer, continuing to set the kind of example which characterised his life.

Becky Goodwill was a fun-loving girl who loved spending time with her family and dog, Buddy. Being out in the garden helping her mum and playing with Buddy was one of her favourite things. She enjoyed trips to London with her family to see shows and visit museums like Madame Tussauds. She was a girly girl who loved all things pink, glittery and she was a keen follower of English fashion and beauty vlogger Zoella. Last year, Becky published an honest blog about her experiences of living with A-T and her day-to-day life. (allthinge.blogspot.co.uk). She passed away in November 2017 and her family is missing their brave, fun-loving girl.

Becky was always smiling and happy. She loved Disneyland so much that she went six times!

Give Boccia a go!

Boccia is a disability sport that tests muscle control and accuracy. It is a great team or individual sport which can be played socially or competitively and is now featured in the Paralympic games. With assistive devices, it can be played by anyone of any ability.

Playing Boccia at the Family Weekend

“After sadly leaving the sport when he finished school, George Keith rediscovered his love of Boccia at the recent A-T family weekend,” George’s mum, Michele, writes.

“George played Boccia at school and really enjoyed it but after leaving school we couldn't see anywhere local for him to play. George had the opportunity to play Boccia again at the Family Weekend and he really enjoyed it. We asked Roy who ran the Boccia session how to find our nearest club and when we looked it up there was a club right on our doorstep!

“So, two weeks after the Family Weekend, George went and joined the Prince Arthur Boccia Knights Club!

He now plays every Saturday, and has made lots of new friends and even met up with old ones from his old school. The club is part of Boccia England and they play in competitions. There are leagues as well and they have an A and a B team so hopefully George will make one of the teams! We bought him his own set of Boccia balls for Christmas so he can get lots of practice at home.

“I would recommend anyone to have a go. It's great fun and myself and Nick often play as well to make up the teams if they are short. So go on, give it a go!”

To find your nearest Boccia club, visit www.bocciaengland.org.uk/clubs.

Don’t you just wish she had said it?

We’ve probably all come across the figure of the condescending medical consultant, who seems to think that patients and their families can’t be expected to understand what is happening to them. This recent example is quite shocking but at the same time funny.

When Ian McInnes (*see page 6*) was in hospital, his mother Elisabeth asked a doctor about the result of some tests Ian had had. Rather than explain, the consultant handed her the written report. After reading the highly technical document, Elisabeth asked the doctor if he could explain some of the technical terms. “May I ask what your profession was?” asked the consultant from on high. “I was a florist”, Elisabeth answered. “Well in that case” said the great man, “I’m going to have to explain it in very simple terms!”

It’s breathtaking that someone working in a hospital in the 21st Century should feel able to say this, but it’s also a shame that Elisabeth is such a well-mannered lady. If not, she might have said to him: “Actually, I was florist to the Queen, who used to discuss flower arrangements with me, and presented me with the medal of the Royal Victorian Order.” We would love to have seen his response..!

George’s birthday trip to Vegas

George Keith celebrated his 21st birthday in style with a trip to the Entertainment Capital of the World – Las Vegas. His mum, Michele, explains how it all came about:

“At the beginning of 2017 we asked George what he wanted to do for his 21st Birthday that was coming up in late September. His answer was ‘a lads’ holiday to Las Vegas!’ so we got to work planning it. Along with dad Nick, his stepbrother Danny and brother-in-law Maz we booked them five nights in Las Vegas at The Excalibur Hotel.

On 25 September, the day after his 21st Birthday, they flew to Las Vegas and their adventure began! After checking into their rooms they met in the bar and started off on the Woo Woos! They had to tell the bartender how to make them. After stopping for some food, they made their way to the Casino in the Hotel and started playing roulette. George took a liking to this game and won most of the time, apart from the time he told his dad to put $50 on number 2 and his dad said ‘no’ and put the money on number 1. And guess what? Number 2 came in so they would have won $1,800! George’s dad will never live this down.

The lads ventured out of the hotel for a spot of sightseeing during their trip. They went up the Stratosphere, looked around several of the big hotels such as Caesar’s Palace, the Bellagio and MGM Grand and did lots of walking up and down The Strip. Hooters Bar was one of George's favourites! He also visited Count’s Kustoms which is one of his favourite TV programmes, where they do up old cars. The man there was impressed with George's knowledge of all the cars they had on show.

All in all, George had a very busy, boozy and enjoyable lads’ holiday. When they FaceTimed me at home, it was great to see them all having such a lovely time, just like any other lads on a lads’ holiday.

It just shows you there are no limits for all our kids, young people and adults with A-T as George didn't experience any boundaries on this holiday and everyone was so helpful. In fact, he enjoyed it so much that he wants to go back again with me and his dad so we have just booked to go in November, staying at the MGM Grand Hotel. I’d better get my skates on and learn how to play roulette, although I have a good teacher in George!”

The day I met the Duke of Kent

By Dale Phillips

A few months ago I went to Stockport near Manchester, along with my carer, to meet the Duke of Kent. I was invited to meet HRH by the Aidis Trust who supply adapted computers for disabled people. They have been very supportive with my computer over the years. For this I am very grateful.

The Duke of Kent shook my hand and I told him how I use the computer. He is the patron of the Aidis Trust. Here we are at the event:

PIP (Personal Independence Payment)

Over the past year we have helped more than 20 families with claiming PIP. As you may know, this is the new benefit that is replacing DLA from the age of 16. It is gradually being rolled out across the country and you will eventually receive a letter asking you to apply.

As we have previously stated, the forms for this can be very complicated and many people are being asked to take part in a 'Home Assessment Visit'. However we have not had anyone who has been refused the benefit and we are confident that everyone who has A-T will qualify.

Please do call us for advice and support: 01582 760733 or email support@atsociety.org.uk.

Here are a few of our success stories:

- **Arthur** from Devon is 16 years old and his mum contacted Kay for help with the forms. Arthur had previously been receiving DLA. Kay helped to fill in the forms and backed them up with a supporting letter and evidence from our Nottingham Clinic. The family were relieved when in December they heard that the application was successful and Arthur is now receiving high-rate PIP.

- **George** from Kent is 21 years old and his mum contacted Anne as, although we had helped with George's forms for PIP, the department of work and pensions (DWP) had contacted the family to arrange a home-assessment visit. The family were very worried and anxious about this and Anne spoke to mum many times to offer reassurance, support and advice. The home assessment went very well and George is now receiving high-rate PIP.

- **Charlie** from Hampshire is 21. His mum filled in all the forms with lots of detail, however the department of work and pensions (DWP) still insisted on a home-assessment visit. In the meantime we were contacted by the DWP and Anne was questioned by a very officious woman regarding Charlie and his diagnosis. However the home visit went very well and the officer who visited Charlie immediately stated it was clear to him that Charlie would qualify for PIP. Charlie is now receiving high-rate PIP and will not be reassessed for another 10 years.

- **Natalie** from Yorkshire is 32 and Kay filled in the forms and submitted supporting evidence, however the DWP still visited Natalie at home. On the day the nurse who visited said 'why am I here?' as it was clear to her that Natalie qualified for the benefit! Natalie is now receiving high-rate PIP.

Winston’s transition to school

By Sara Pemberton-Powell

I have to come clean and say even thinking back to the lead-up to Win starting school is still quite hard. Such a lot was going on for us regarding his health and wellbeing and it was a very scary prospect to be sending him into school.

Win had been struggling at nursery as his physical differences were becoming more apparent. Soon after his diagnosis he began to struggle with anxiety around the more confined spaces in the nursery and he worried about knocking other children over. So the idea of sending him into a whole school full of children felt really daunting. I can remember countless conversations with Anne from the A-T Society as I struggled up the hill to nursery with the buggy – she was always so reassuring and comforting. I would call thinking I had a few quick things to mention and end up bending her ear for the whole walk and without exception she always made the time to talk things through and ease my mind.

When the time came to choose a school, I couldn’t be sure what would be right for Win. Would he be better in a school with a Special Needs facility that was fully accessible? Or should we follow our instincts about Win’s personal needs and send him somewhere a little smaller and more focused on community? Anne gave me helpful pointers as to what was important when viewing the school buildings, as well as sending us the Information for Schools pack which we could take with us to give the settings a better understanding of Win’s needs. But perhaps more importantly, she also gave us the confidence to trust our instincts, and the assurance that the A-T Society would be with us and would support whichever school we chose to make the transition as smooth as possible.

Winston on his first day of school; with his new best friend, Logan.

At this time we were undergoing the process of obtaining Winston’s EHCP. A multi-disciplinary meeting was called and Anne travelled to Hastings to attend. I was so relieved she was there and full of admiration for the way she engaged in the meeting. She gave such concise, thoughtful information and helped keep the conversation on track. She pushed for the best for Win throughout and was able to make a stand for him through her expertise and commitment that even we as his parents would have found difficult. Unfortunately, I had to take Win’s case to appeal and again she and Kay were beside me, giving me all the options and encouraging me to make myself heard. Win now has his EHCP in place and has a wonderful Individual Needs Assistant.

When we had finally selected the school we wanted and he gained a place, Anne suggested I liaise with his teacher and request a few visits so he could come to the classroom when it was empty, get used to the space and meet her properly.

By this time the class teacher had been given all the information provided by the A-T Society and was thinking along the same lines – offering Winston some sessions after school, which he loved. We took photographs of him playing with the toys and printed them, and he remembered where the toilets and coat pegs were. We were lucky enough that the school had a Headteacher and staff who went out of their way to support Win in starting school. Combined with the unwavering support of Anne and the A-T Society, by the time Win’s first day came he was a bundle of excitement who couldn’t wait to put on his uniform and leave the house. At the start of the process I couldn’t have imagined it would ever have been this way.

Win had a hugely successful first week at 'big' school! They arranged for Winston to attend their morning breakfast club so he could avoid the hectic, crowded playground. In true Winston style, he bowled in on his first day and had the year six boys in stitches from the word go! We are so proud of his positive attitude. To top it off he now has a new best buddy called Logan!

Soon Kay and Anne will travel back to Hastings to give a talk in school to give staff a better understanding of A-T. We are delighted for the school to be so well informed and ready to receive Win as he moves through the school. Plus we’re really look forward to seeing Kay and Anne, too!

Family Weekend 2017

Becky Simpkin opening the family weekend.

The beginning of October saw A-T families, friends and carers gathering together once again for our annual Family Weekend. Nearly a third of all UK families with a diagnosis of A-T travelled to the Raddisson Blu hotel at Stansted Airport to attend. For some, the weekend started on the Friday night with a relaxed evening catching up with old friends and making new ones. Pizza Hut very generously supplied the pizzas, and our very own cupcake queen Jilly Shah brought along some delicious homemade brownies.

On Saturday the children headed off to Paradise Wildlife Park and enjoyed a great day out seeing all the different animals. For the young adults there were several workshops on offer for them to join in. From competitive Boccia playing, to cupcake decorating run by Jilly and her family, and an art workshop run by Kay's husband, Kevin, there was something for everyone. A professional masseuse treated those who just wanted time out to relax, to a holistic massage.

For the parents, carers and relatives who stayed behind, a varied programme of talks had been organised including the latest on the Erydex trial, preliminary findings from the RAMP trial on A-T and diabetes, liver problems with A-T and an engaging talk from Tina and Charlie Stubbs who shared their experiences of visiting Papworth Clinic. Over lunch there was a chance to try out some of the computer adaptive technology from the Aidis Trust, chat to the Demand charity about potential practical inventions that could help people with disabilities and find out more about Willow Charity Special Days. The afternoon talks finished with the traditional Question and Answers session with our panel of experts covering a range of topics.

The fancy dress theme this time

> "This was our first time and we thoroughly enjoyed it. It was very informative and made me feel reassured that we are not alone and that they are doing their best to find a cure."
>
> "I valued my time with all the young adults with A-T. I loved the fact that I did not feel alone. I now know more about A-T and what I can do to still have a good life."
>
> "Positive and encouraging weekend, full of hope and fun for everyone. The A-T team go beyond the call of duty, along with their family members, to make it a very worthwhile and enjoyable weekend. Educational and highly recommend it."
>
> "The A-T Society delivered informative perfection through the scientific talks and services that held the stage at this year's Family Weekend. Thank you to all of you that worked tirelessly to plan and stage this great event."

was famous TV, film or book characters, so Minnie Mouse, the Minions, Little Red Riding Hood, Pikachu and Shrek were among the characters who turned up for the banquet dinner. Before food was served, Ebony Robinson gave an inspirational talk about her journey on BBC's *The One Show* Rickshaw Challenge. There was hardly a dry eye left in the house when Ebony's fellow team-mates made a surprise entry onto the stage. Ebony's look of sheer surprise and delight was truly memorable. After dinner it was chance for the adults to relax and catch up while the children were entertained by magician Magic Frostie, a disco, glitter tattoos and a sweet stall.

Psychologist Jane Flint led the talks on Sunday morning with a thought-provoking presentation about the importance of looking after your emotional well-being and making the days count. Following on from Jane were a series of talks from our adults with A-T. They all gave inspirational speeches about their lives and demonstrated how the challenges of living with A-T can be overcome. By lunch it was time to head home and there were a lot of emotional farewells as families said goodbye to each other for another year.

Clockwise from top left: Frances and family chatting to Jarno; art workshop; rickshaw challenge team; Brooke and Caleb; catching up over dinner; cake decorating workshop; playing Boccia; Shah family

Family Weekend 2018

When: 29th June - 1st July
Where: Stansted Airport (Essex)

This year we will be heading back to Stansted Airport! We very much hoped to be heading north or to the midlands, however despite all our efforts (and the agency we work with) we have not been able to find a hotel with enough adapted bedrooms (as we have so many people wanting to attend, which is great!) and the available dates.

However this year we will be extending the event to enjoy the whole weekend together! We will officially be starting on the Friday evening, with a guest speaker, talks and an indoor BBQ. (Registration will be from 3.00pm on the Friday afternoon.) A full, fun-packed day on the Saturday will follow, and there will be an informal breakfast with talks on Sunday to finish off the weekend.

We hope that as many of you as possible can join us and if needed we will be able to help with the cost of the Friday night's stay. So put the dates in your diary and look out for more details coming to you soon!

Adult Activity Weekend

The Adult Activity Weekend is a popular event for many and this year didn't disappoint, with the A-T adults heading to the Lake District for an action-packed adventure.

In September 2017, 11 adults with A-T headed off to the Bendrigg Trust activity centre in the Lake District for an action-packed weekend. The first day of the activity weekend was a day of two halves. While one group went off to Lake Windermere for a day of canoeing, stunning views and sunshine, the other group climbed, zipped and competed in a tough archery challenge.

In the evening the intrepid group gathered round the campfire trying to answer some challenging brainteasers before kicking back and catching up with old friends and getting acquainted with new ones. One of the carers, Toni, taught them all how to do the best toasted marshmallows and s'mores and everyone happily tucked in. A brilliant first day was had by all and everyone went to bed tired but happy.

The wifi proved to be as reliable as the weather forecast and, on the second day, the adults woke up to a very wet morning – luckily it didn't dampen their spirits. One group canoed in the driving rain while the other lucky group spent Sunday morning on the excellent indoor climbing wall at Bendrigg Trust. There was some brilliant climbing from everyone. Stephen powered his way up 3 climbs; Merry, an experienced climber, started by sprinting up a tough one; Amy and Emily were quick learners and Ebony challenged her carer Jenny and volunteer Katherine to a race and burned them up.

After lunch the climbers spent the afternoon trying their hand at indoor archery before heading out into the horizontal rain and overcoming their fears by zipping down the zip wire.

Luckily for the rather wet canoeists, they were able to dry out in the warm lounge and watch smugly out of the window while their comrades braced themselves against the rain.

By the end of the weekend, spirits were running high with everyone feeling the benefit of all the fresh air and the camaraderie. Every single one of them left with a sense of achievement at accomplishing something new.

"One of the most amazing places I've been to. Facilities, activities, accommodation etc are second to none. The compassion and enthusiasm of all the staff and volunteers was captivating, humbling and genuinely amazing. Seeing people overcome their fears, accomplish new things and jump so far from their comfort zone was so inspirational." Toni (Carer)

Two of our adults with A-T, Helen and Natalie, wanted to share their own personal experience of the weekend:

Natalie Fall

"The main reasons I attend the Activity Weekend is because it's a weekend away from home and a chance to meet up with current friends and make new ones that have recently been diagnosed with A-T, or that I haven't met before.

I have done all the activities before on previous trips, however I enjoyed canoeing and the campfire the most.

Canoeing was an all-day adventure on Lake Windermere and luckily it was a nice sunny day. We canoed to an island in the morning to have a picnic lunch, then afterwards while coming back we played games and sang on the boat until we got back to shore.

In the evening, I enjoyed getting together and having a drink and a laugh and making s'mors (marshmallows and chocolate) on the campfire.

During the weekend I was on such a high as there was so much to do and it was great being with all my friends, but afterwards I was so tired!!! But being tired was a small price to pay for such a great weekend. One of the nicest things for me was spending time with my personal assistant, we had such a laugh. She is not only my PA but a good friend as well.

The location where we stayed was beautiful, and I bet it's even more stunning in the summer. A perfect setting for a weekend away. The accommodation we stayed in was clean, wheelchair friendly and more than adequate for our needs.

Even though I had done most of the activities before it was a very enjoyable weekend and I would definitely do it again. Thanks to the A-T Society for organising such a brilliant weekend and especially to William and Anne – it was a pleasure to be in their company."

From left to right, row 1: Ben preparing to zip wire; Amy, Ebony & Natalie; Becky; climbing wall. Row 2: Ebony & Amy play skittles; Ben gives archery a go; Merry & Stephen canoeing. Row 3: climbing wall; preparing to canoe.

Helen Carter

"I really enjoyed my trip to the Bendrigg Trust with the A-T Society. Once everyone arrived after some hectic journeys due to traffic problems, we all got to know each other and one of the coordinators, Jenny, read out the activities for the next couple of days.

There was a lot to fit in the next day. My morning began with me rock climbing in a harness on an indoor wall. I was really proud of that and afterwards used a very technical piece of equipment called a "pully-up thing". Abseiling for the first time that afternoon was a real achievement followed by zip wiring which I love – so much so, I had three goes. I surprised myself at how energetic I'd been and how determined I was to try everything.

We socialised over meals in the big kitchen or dining room and there was a Co-op run for treats and wine – we needed a glass of wine after the exhilarating and tiring day we'd had! We sat around the campfire toasting marshmallows and giving our brains a workout with quizzes.

The next day canoeing was on the itinerary for me. Of course I wasn't daft – I didn't do any of the hard work – I sat back enjoying the lovely Lakeland view.

Me and my carer, Joanne, really enjoyed ourselves and would definitely go back. I really felt good about myself and mum said I had a real buzz when I got home. Thank you A-T Society for making it happen."

We are already planning the next Activity Weekend and will be announcing full details soon.

Fundraising

THANK YOU! We think you're all AMAZING!


Chris Bentley ran 32 miles in the Tring Ultra Marathon.


Marv Hine completed the Stour Valley Path 100km Ultra Run in 13.5 hours.


Michael Byrne conquered the Manchester Half Marathon.


A wonderful celebration for Sara Metcalfe's 50th birthday marked the end of the 50:50 challenges she took on in 2017.

Scott Horney and the Merton Lions looking very clean before powering around the South London Tough Mudder.

Halton Baptist Church, Hastings, held a Christmas fayre.


Santa and his helpers braved the cold to collect a fantastic amount in St Albans.


The Concordia Choir sang their hearts out at Much Wenlock Christmas Fayre.

Scott Horney's fantastic football tournament was fun for all the teams.

Alfie Arnold co-hosted the Chard Car Fest in aid of the A-T Society. It was a great day, and another is planned for summer 2018.

John and Mary Humphries celebrated their 65th wedding anniversary by asking for donations instead of presents.

Maxine & Graham from the Moody Mare pub organised a Beer and Music Festival with great music, excellent food and a lively atmosphere.

Aiden Wilson pedalled to glory in the Prudential Ride London 100.

The Rotary Club of Weston Super Mare generously included us in the proceeds of their Annual Golf Day.

Kay Thornton hosted a very successful cake and coffee morning in Hastings.

Susan Semrau, Sandra Eglington and Judith Mears took part in the Gung-Ho 5k inflatable obstacle course.

Brae & Chloe had a blast at a Family Funday organised by A-T parents Siobhan Kelly and Brian Sewell.

Flo and Eliza raised money by charging an entry fee to their spooky Halloween party.

Hertfordshire Chamber Orchestra brightened a cold January night with a feast of classical music in Harpenden.

Jim Thompson treated guests to an evening river-boat trip from Totness to Dartmouth to view the Regatta firework display.

Two separate, mind-bending quiz nights were presented by Adelle Rouse and Andrew Field. Both were a resounding success.

The Caritas Harmony Choir gave guests a beautiful evening of song.

Robin Arnold kindly requested donations instead of gifts for his 75th birthday.

The Paediatric department at Oxford Children's Hospital and A-T mum Nav Cooper held a fun-filled Christmas Jumper Day and hamper raffle.

Heather Allsop and Marsh House Orthodontics in Surrey celebrated the festive season with a plethora of Christmas jumpers.

A very elegant cheese and wine party was hosted by Micky MacCormick in Glasgow.

Jilly Shah set up her business making cakes in 2016 and has donated every penny raised.

Catherine Muddle generously gave donations in lieu of birthday gifts.

Ferguson family of fundraisers

Take a walk around the pretty town of Bridgnorth and you will find A-T collection pots in several shops and cafes. Some days you will also find James, Rachel and Joy Ferguson collecting in the local branch of M&S Simply Food! Their latest store collection amounted to £311 and they have more collections planned in March before our second year as the Bridgnorth store's annual charity ends. The store staff are very active and will be undertaking a sponsored 800 Steps Challenge Walk and a hike up Snowdon. Heartfelt thanks to all the staff and customers for their generous support.

Joy relaxes by singing with the Concordia Choir, who were pleased to give her fundraising a boost by carol singing at Christmas, bringing some festive cheer to Christmas shoppers.

Joy really enjoys fundraising and over the years has been a great ambassador for the A-T Society. Each year she opens her home for an annual Fundraiser which raised nearly £1,500 last year!

We love supporting fundraisers in the local community and Joy and her family are a shining example of how much can be achieved raising awareness as well as vital funds. Knowing they're giving back helps raise the family's spirits as well as vital funds.

Clockwise from top left: Concordia Choir; James collecting in M&S; Joy with raffle winners; Joy and Rachel with raffle prize.

Strictly Come Dancing gala dinner

Darren Haynes hosted a glittering night of entertainment in support of the A-T Society and St Luke's Hospice, Plymouth. Guests dressed to impress, enjoying a three-course dinner and being treated to a 'Strictly-Style Celebrity Dance-Off' with, among others, some rather dashing Sugar Plum Fairies dancing their hearts out!

Jayne Rainbow (*pictured, far right*) stole the show representing the A-T Society with a dazzling Cha Cha. Having spent four months learning her routine she did us proud and captivated the audience with her A-T Society inspired dress, resplendent with the same blue, pink and purple as our logo! The evening was a huge success and we are very grateful to all the dancers who trained so hard and everyone who made the event happen.

Church charity of the year

All Saints Church in Blackheath chose the A-T Society as their charity of the year in memory of Rupert Prokofiev, who had been a pupil at All Saints Primary School.

An impressive and varied calendar of fundraising events has been organised throughout the year including carol singing, a wine tasting, table top sale with the primary school, cake and craft sale, a new-year ceilidh, evening supper party, two concerts performed by London Joy Singers and a gallery tour with The Reverend Dr Cranfield. This marathon fundraising year is on course to raise over £5,000 and concludes with a wonderful charity concert for children and adults, 'Peter and the Wolf and Friends', appropriately narrated by Rupert's brother Gabriel Prokofiev.

Sarah Evans' Three Peaks Challenge: a Win-win situation

Intrepid Sarah Evans is proof that it's never too late to fundraise! Inspired by her friendship with Winston Pemberton Powell and his family, Sarah set up her online fundraising page just hours before braving sub-zero temperatures, rain and high winds during her attempt to scale the three highest peaks in Scotland and England and raised over £2,000!

Her initial fears of letting people down if she didn't complete the challenge were quickly dispelled as sponsors started adding support, their messages helping to keep her going when the going got really tough.

An hour into the final climb up Snowden, weather conditions become so hostile the guides decided it wasn't safe to continue and reluctantly Sarah had to turn back.

There is no doubt that Sarah earned her sponsorship. Everyone visiting her My Donate fundraising page would have understood why she chose to undertake this gruelling challenge and her determination to complete it if humanly possible. As she said, 'My motivation is Little Win. It's a win/win situation.'

Corporate fundraising

Meet the brilliant companies that have chosen to support and raise funds for the A-T Society.

Aggregate Industries

Little Daisy Goble charmed her way into the hearts of staff at Aggregate Industries Garside Sands Quarry in Leighton Buzzard and motivated the entire team to take part in a static bike cycle distance from 'Land's End to John O' Groats'. The team started at 5am accompanied by pumping music and the encouragement of trainers Darren and Eddie from DW Sports Fitness at Milton Keynes. Daisy's Dad and Mum Lee and Terri joined in along with members of the local fire brigade who turned up to offer their muscle mid-shift.

Just 12 hours and 874 miles later the team was victorious. Their incredible fundraising achievement enabled us to purchase a trike for Daisy which will grow with her and give her a little independence (*see picture, right*). As the money kept pouring in, other children will benefit too. Thank you to Claire, Gary and the Garside Sands staff who went the distance and more!

Willmott Dixon Gateshead

Another inspirational duo are Charlie and Arthur Seal whose connection with the North East of England brought them to the attention of the fundraising team at national building company Willmott Dixon's Gateshead office.

Just a fortnight ahead of their annual charity day, staff selected the A-T Society as their chosen charity for a 24 hour static triathlon. Keeping it local, the staff rowed the length of the Tyne (73 miles), ran Hadrian's Wall (84 miles) and cycled the distance to and from the company's head office in Letchworth (580 miles).

Local girl Merry James and her parents popped in to cheer the team on, and later that night Sara Metcalfe took up the challenge and helped the team cruise to the finish. At the time of writing, the event had smashed the company's previous fundraising target with more money flowing in. Thank you to Sarah, Tammy and Willmott Dixon's staff and suppliers.

Audience with Bim Afolami MP

A chance meeting before the election led to Hitchin and Harpenden's new MP Bim Afolami offering his support to the charity and suggested a fundraising networking dinner for corporate and potential major donor supporters at the Rothamsted Conference Centre.

'An Audience with Bim Afolami' featured the MP being interviewed by BAFTA award-winning TV journalist, ITN's Head of News, Richard Frediani. Richard quizzed the MP with his own questions and those of the audience.

Bim answered questions on housing, disability rights, education, Brexit, medical research funding and the impact of social media on the life of a politician. He was put on the spot with quick-fire questions including whether he wanted to be Prime Minister, and reinforced his determination to support the A-T Society now and in the future.

This was a witty and entertaining evening generating new and exciting contacts for the charity locally to the office and further afield. We've had great feedback and requests for another 'Audience with' later in the year. We very much appreciate Richard and Bim's support and believe they will be strong advocates for A-T, disability and rare diseases in the media and in parliament.

Pizza Hut

Summer in Harpenden features a huge car enthusiast's event 'Classics on the Common' just over the road from the A-T Office. Harpenden Cricket Club donated Pimm's to sell and space on their pitch for our gazebo. Volunteers from Pizza Hut Delivery sold their delicious home-baked cakes and cookies to visitors and raised awareness of our work.

Pizza Hut Delivery features the A-T Society on their website, so everyone ordering a pizza online has the option to make a donation. We were overwhelmed when donations totalled over £21,000 in 2017 and are thrilled to continue our corporate partnership with the company in 2018.

AT society
awareness research care

Standing order form

To: {insert name of your bank}

____

Bank address: ___________________________________

____

Account name: __________________________________

Sort code: ___/___/___ Account no: ____________________

Please pay a regular gift to the A-T Society of: {tick appropriate box}

☐ £3 ☐ £5 ☐ £10 ☐ £25 ☐ £50 ☐ Other amount

I wish to donate £ ________ (please also state amount in

words) ____

each month to the A-T Society

Please start on ___/ ___/ ___ and pay this amount each month

until further notice.

Signature: _______________________ Date: ___/ ___/ 20___

The A-T Society bank details are as follows:

HSBC, 1 High Street, Harpenden, AL5 2RS

Sort code: 40-23-11 Account no. 41248693

Your details to set up the standing order:

Title: ______ Name:_____________________________________

Address: ____

____

Post code: ______________ Phone: ____________________

Email: ____

giftaid it

Boost your donation by 25p for every £1 you donate

Gift aid is reclaimed by the charity from the tax you pay for the current tax year. Your address is needed to identify you as a current UK tax-payer.

I am a UK tax-payer and understand that if I pay less Income Tax and/ or Capital Gains Tax in the current tax year than the amount of Gift Aid claimed on all my donations, it is my responsibility to pay any difference.

I wish to Gift Aid my donation to the A-T Society ☐

Please notify the charity if you:

- want to cancel this declaration
- change your name or home address
- no longer pay sufficient tax on your income and/ or capital gains

If you pay Income Tax at the higher or additional rate and want to receive the additional tax relief due to you, you must include all your Gift Aid donations on your Self-Assessment tax return or ask HM Revenue and Customs to adjust your tax code.

Please return this form to:

The A-T Society
Rothamsted
Harpenden, AL5 2JQ

Thank you!

We would like to keep your details and use them to inform you occasionally about our activities. We will not pass your details to any other organisation. If you are happy for us to do this please tick this box ☐