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A-T Society News

awareness research care

www.atsociety.org.uk
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Just William

As I write this, it is almost 30 years to the day since the inaugural meeting of ten families in Birmingham gave rise to what is now the world's longest standing A-T organisation.

It was the 29th April 1989, that the families came together in response to an invitation sent out by Maureen Poupard. Buoyed up by finding that they were not alone, and by the relief of finding others with whom they could share their feelings, experiences and ideas, the parents gathered there decided to set up a support group.

The aims they agreed were simple:

- to support A-T families
- to raise public awareness of the condition
- to fund research; and
- to fundraise to meet these aims

Thirty years later, the Society is much bigger, supporting 176 people with A-T from over 150 families in the UK, another 15 in Ireland and other individuals in countries around the world. We have the equivalent of 6 full-time staff plus a number of regular volunteers. But our aims are just the same as they were back in April 1989. You can see them enshrined in the strapline of our logo: Awareness Research Care.

Some of the families from those first days of the Society will be with us at the Parliamentary Reception or our Family Weekend in June. I hope that they are proud of all that has grown and happened as a result of that first meeting.

I am certainly proud to be part of the A-T Society. We have an amazing Family Support team that many much bigger organisations would be proud to have. Anne and Kay are familiar faces and voices to all our families, and I know how much their support means to people living with A-T, as they continue to tell us in no uncertain terms. Over the last year or so our work to support adults living with A-T has also developed considerably.

In terms of awareness, it is always going to be an uphill battle to tell the world about such a rare (and unpronounceable) condition. But in addition to all the positive stories we help our families to tell in the local media, we have had some real successes at national level. I think for example of all the radio coverage we got from the Global Make Some Noise Day, the TV sensation that was Ebony Robinson on the BBC Children in Need Rickshaw Challenge, and the splash in The Sun newspaper when the film about Brae and Broghan Sewell came out.

As for research, since making our first research grant in 1991, we have funded research projects every year except one. We have funded projects, equipment, clinical trials, research posts, meetings and a registry. But our impact goes far beyond the funds expended. By organising conferences and meetings, engaging with scientists and clinicians and advocating actively for A-T research we have created new projects and partnerships, and have attracted major figures into the A-T research field. The International Clinical Research Network and Conferences we established are amongst the main drivers of A-T research today.

And I believe that as our aims are still the same today, so are our values. We are an organisation built around people and families

About the A-T Society

Ataxia-telangiectasia, or A-T, is a neurodegenerative, genetic condition which affects many parts of the body, causing increasingly severe physical disability. The A-T Society is a national charity providing information and support to people affected by A-T and funding and promoting research.

Our positive philosophy of Live well with A-T expresses our commitment to provide the information and support to enable people with A-T and their families to live their lives to the full. At the same time, we work to improve treatments and to stimulate and fund research which will ultimately find a cure.

AT society
awareness research care

You can contact us on:

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Registered charity no: 1105528

A-T Pilot Meeting at Acorns Hospice for Children in Birmingham – 29th April 1989.

living with A-T, existing to serve them and their needs and are open and responsive to them. While we strive to be as professional as we can in the things we do, it is people and their lives that come first.

And as it was thirty years ago, it is the meeting of families together that is the beating heart of the A-T Society. So it is right that it will be at our Family Weekend, not far from Nottingham, where we set up the world's first A-T clinic, that we will come together for our 30th birthday.

We will look back and remember the amazing achievements along the way and the many people who have made these things happen.

We will also remember those that we have lost. But above all, we will look to the future. The Society is in a strong position, we are looking to do more, over the years to come, particularly increasing our investment in research. The scientific and technological resources available to us are so much greater than they were 30 years ago. The anniversary must inspire us to use all of these to ensure that in the years ahead of us, we make real progress in defeating A-T.

Our last A-T family weekend attended by over a third of our A-T families.

Cambridge Research Symposium

In 2018 the Society's Research Advisory Board agreed that our research strategy should for the next few years focus on that aspect of A-T which has the greatest impact on every-day life and well-being for people with A-T: the neurological problems.

One of the biggest obstacles to developing treatments for the neurological symptoms of A-T is the fact that after so many years, we still do not understand exactly why it is that in the absence of the ATM protein some neurons (brain-cells) malfunction or die off, while others do not.

To help identify research priorities for this new strategy, we organised a Research Symposium at the University of Cambridge in February. Entitled 'New approaches to understanding and treating the neurology of ataxia-telangiectasia', the meeting brought together nearly 30 experts on the neurology and neuroscience of both A-T and related conditions from across the UK.

Bringing together experts from different fields, the Symposium aimed to both identify the most promising areas for future research in this area, and opportunities to exploit recent technological advances and breakthroughs in related fields, as well as to help establish future collaborations. It was divided into two parts, one focusing on more laboratory-focused research, chaired by Professor Steve Jackson of the Gurdon Institute Cambridge and the other on clinical research, chaired by Dr Anke Hensiek of the Papworth A-T Centre.

The meeting was a great success. There was a wide variety of short presentations which led to some excellent and positive discussions. Amongst other things, particular focus was given to possible approaches to identifying the causes of neurodegeneration, to potential approaches to identifying biomarkers, to the opportunities offered by different kinds of imaging, and to the potential of worn technology such as smart watches or clothes with sensors, to aid the understanding of ataxia and other movement disorders.

A good number of potential areas for research, both clinical and laboratory, were identified, along with a number of other potential resources that could be exploited for A-T research. These were submitted to the Society's Scientific Advisory Board to help them formulate the new research strategy. The Board met in early April for an initial meeting which agreed an initial set of potential priority areas for the Society's research programme. This is now undergoing some more detailed discussion and investigation with the aim of producing the new strategy by the end of the summer.

Natural History of A-T

A *Natural History* that is a detailed description of the condition based on extensive data, is a vital document for any rare condition, invaluable for designing clinical trials, for developing clinical guidance and in many other ways. There is currently no natural history for ataxia-telangiectasia so the Society is delighted to be jointly funding a project to develop one using data from the Nottingham and Papworth clinics.

The project is being undertaken by Dr Emily Petley, with the support of Dr William Whitehouse, paediatrician at the Nottingham A-T Centre. The A-T Society will be jointly funding the second and third years of the project with Action for A-T and BrAshA-T Ataxia-Telangiectasia, the Australian A-T charity.

The project will use data gathered over many years by the clinics at Nottingham and Papworth as well as that from research projects such as the CATNAP brain imaging study and the IMAGINA-T lung-imaging study to create a natural history. The UK is the ideal place to carry out this study given the large numbers of people with the condition who have been seen regularly at the two specialist centres over so many years.

The first year of the project was funded externally but the A-T Society supported this with grants for equipment and the assistance of a statistician.

William commented: “While it may not sound the most glamorous of projects, a natural history really is a vital document to support the development of treatments for A-T. It is great that data from the two UK clinics is now being used to create a natural history for A-T. But beyond this, I am particularly delighted by the fact that the project is being funded in partnership with Action for A-T and with BrAshA-T. I hope we will see a lot more of this collaboration in future.”

A-T International Registry

The International A-T Registry, which was launched in July 2017, is the first ever registry dedicated to gathering clinical data about people with ataxia-telangiectasia and related conditions from around the globe.

By gathering clinical data about people with A-T and related conditions from different countries, the registry aims to improve our understanding of these conditions and how they develop. This in turn will help improve existing treatments, and develop new ones.

Achieving these aims depends on being able to collect detailed information from a large number of people with A-T and related conditions. By comparing the data, researchers expect to be able to answer many of the questions that still exist about the conditions and work out how to treat them better.

AT InternationalRegistry

We have gathered some data but we would love more participants. Please email **jo@atsociety.org.uk** if you would like further information on the registry and how to sign up.

Naples A-T Clinical Research Conference

In November 2018 William and Anne travelled to Naples to participate in the A-T Clinical Research Conference. The focus of the conference was on improving clinical care and developing new treatments for people with A-T by bringing together world leading A-T specialists. Attendees included clinicians, therapists and researchers interested in treatments for A-T, as well as representatives from A-T charities. Over the course of the three days there was much information sharing, many lively and often challenging debates and a considerable amount of discussion about the way forward for new treatments and research.

The programme covered a wide range of recent scientific and technological advances, from cancer to immunology to respiratory problems etc., all aimed at better understanding and treating A-T. Many of our A-T clinicians, therapists and researchers from Nottingham gave presentations about their work and research. For the very first time the programme included presentations about emotional/psychological and social issues in A-T – a topic area which is at the heart of much of the A-T Society’s work.

Seeing this as an opportunity to highlight the need to think outside the 'medical model' box, William opened the conference by encouraging the audience to consider taking a whole person approach and to acknowledge each individual's physical, mental and social well-being, rather than just their diagnosis or medical needs. As a starting point William suggested using the phrase 'person living with A-T' as opposed to referring to 'patients' in an attempt to take on a more holistic view of each individual.

Anne was invited to talk about our new pilot project titled *'Promoting independence to support emotional well-being'.* In her 15 minute slot, Anne delivered an overview of the project and captured the interest of those in attendance. She explained that due to our unique relationship with the majority of people living with A-T in the UK, we have been able to collect and analyse a wealth of anecdotal information built up over many years. Some of the methods used to gather this information include; person centred workshops carried out at recent activity weekends, regular conversations with young people and their families, plus numerous home and clinic visits carried out by Kay and Anne.

The audience learned that the most striking and recurring themes in our support and advocacy work relate to improving Quality of Life, not just health or medical needs and that young people frequently tell us they want *'to be heard'* and to *'have a life'*.

Anne went on to outline the methods used to analyse our findings and identify a way forward with the pilot project. She explained that it quickly became clear that there are 5 priorities most important to young adults living with A-T in the UK:

- Opportunities for independence
- Having meaningful relationships
- Positive emotional well-being
- Mobility – e.g. suitable wheelchairs
- Meaningful occupation

Anne explained that these findings highlighted that having access to an appropriate PA can have a positive influence on young people's sense of health/wellbeing and quality of life but that finding one is often rather challenging. The main aim of the pilot project is therefore to address the psychological and emotional impact of A-T, by assisting a small group of young adults to recruit quality PA's. Having secured financial support from money donated by Rupert Prokofiev, who firmly believed that life was for living, the project is now underway with Rupert's former care coordinator, Ann Highly (**www.annsguidinghand.com**), using her experience and expertise to help with the recruitment and training of PA's.

For Anne, it was a highly nerve racking experience to stand up and speak to such a large, esteemed crowd. Some minor IT issues before she was due to present did not help to steady Anne's nerves, but she was determined to get her messages across and the positive feedback she received afterwards made it all worthwhile. Many people commented on the uniqueness of the A-T Society in that we personally know the majority of UK families affected by A-T and have regular contact with most of them regarding many aspects of their lives – not just health needs. There was also much discussion amongst the audience about how the majority of them only see people with A-T regarding their physical health needs and as 'patients'; therefore Anne's presentation and William's opening speech gave much food for thought.

One of the things that struck Anne the most about the conference was the sheer number of people who travelled from all over the world to participate – including a Mum and her teenage daughter with A-T who came all the way from Western Australia. For such a rare condition it was staggering to witness just how many people from different professional backgrounds are dedicating their time and energy to work together, in order to improve care and develop treatments for A-T.

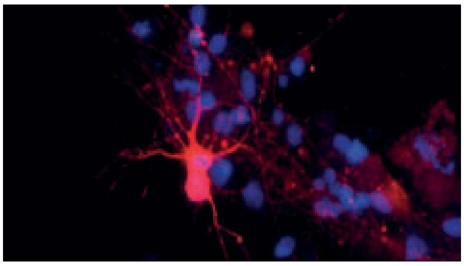

Representatives from patient organisations around the world.


Team from the Nottingham Children's A-T Service.

Purkinje cell project making progress

One of the most interesting and important projects that we are currently funding is that led by Prof. Domenico Delia to create Purkinje cells in the laboratory using stem-cells derived from people with A-T.

Purkinje cells are very big cells, located in the cerebellum. They play a key role in regulating and coordinating movements. For reasons that we don't fully understand, these cells die off in people with A-T.



Purkinje neurons obtained from human iPSC after in vitro differentiation for 69 days using the M. Sundberg protocol (Red: Calbindin, Blue: DAPI) , magnification: 40X

As it is not possible to remove Purkinje cells from the brains of living people, growing them in the laboratory from stem-cells is the only way be able to study them at first-hand. However these are very big complex cells and creating them is at the forefront of what is scientifically possible at the moment.

This complex project, based at the FIRC Institute of Molecular Oncology in Milan, began last year. To create Purkinje cells, the team started with skin cells from people with A-T. These were first turned into 'pluripotent stem-cells' or IPSCs, that is cells capable of producing all kinds of other cells. Then these cells in turn were converted to neural stem-cells, which could finally be used to generate Purkinje cells. The project was made all the harder by the fact that Purkinje cells without ATM protein are particularly vulnerable.

The exciting news is that Prof. Delia and his team have succeeded in creating Purkinje cells, the first time that this has ever been done with cells from people with A-T. At the moment, though, the number being produced is quite low, so the next step is to try and increase the yield. This is what the team is currently working on.

Once Purkinje cells are being produced in sufficient numbers, they can be used to screen potential drugs and for other experiments to help understand why it is that these cells die off when other neurons don't. While there is still a lot of work to do to get to that point, this is an extremely encouraging advance.

New clinical trial for A-T to start soon

Fresh on the heels of the ATTeST trial, another clinical trial for a treatment for A-T is due to start this year. Biopharmaceutical company, Intrabio Inc, plans to trial a drug known as IB1001 in A-T as well as several other neurological disorders.

IB1001, the full name for which is N-Acetyl-L-Leucine, is one form of the drug N-Acetyl-Leucine (IB1000s). Another form, N-Acetyl-DL-Leucine, has been approved in France since 1957 under the trade name Tanganil® as a treatment for acute vertigo, and has an excellent safety record.

In pre-clinical and compassionate-use studies, IB1000s has shown a significant benefit for the treatment of patients with a number of inherited ataxias, including A-T. It is hoped that this trial will confirm the drug's effectiveness.

The company aims to recruit 30 participants with A-T and is planning centres in the USA, the UK and Germany. The UK centre will be at Papworth. The formal name for the A-T branch of the trial is IB1001-203 . Authorisation has already been obtained for the trial to go ahead in the USA and applications have been submitted for the UK and Germany.

People with A-T already involved in the ATTeST (EryDex) trial will not be eligible to participate in the IB1001-203 study. However, as the inclusion criteria for IB1001-203 are different from those of the ATTeST trial, it may be possible for some people who were not able to participate in the ATTeST trial to be involved in this trial. More detailed information on the enrolment criteria can be found on the ClinicalTrials.gov website, searching under IntraBio.

William commented: 'We warmly welcome the interest of IntraBio in testing IB1001 in people with A-T and have been supporting the company with their programme. It is very positive that a trial site at Papworth is planned and that UK A-T patients will have the opportunity to take part.'

Research is the key to unlocking a cure for A-T. It costs £5,000 a month to fund a researcher. Please donate to help fund projects seeking new treatments and ultimately a cure.

ATTest trial going well

ATTest Trial team with Orla

ATTest Trial get a thumbs up from Orla

The ATTest clinical trial of the EryDex System is now going very well at the Nottingham A-T centre. As we go to press, there are 8 children participating in the trial, with another hoping to start shortly. However there is still the possibility for others to take part.

After the hold-ups which delayed the start of recruitment last year, everything now seems to be going smoothly with regard to the treatment. Two of the children participating have portacaths, which means that the team doesn’t need to find a vein each time they come for treament. The rest are happy with a cannula put in for each visit.

Sinead Ward, whose daughter was the first person to receive the treatment in the UK told us: “It’s going really well. Dr Whitehouse and the staff are lovely and have been incredibly accommodating. Because it’s on a Sunday, visits are not too disruptive and Orla is able to go to school regularly. I would encourage any parent whose child might be eligible to be screened. Research is such a positive thing in itself, and it’s also helpful that Orla is getting these regular assessments.”

Dr William Whitehouse, the primary investigator for the Nottingham site, is very pleased with the progress: ”It’s going really well” he said. “The nurses and other members of the research team are enthusiastic and the trial organisers are very supportive. However we still have the capacity to take on a few more participants. If people are interested, I would encourage them to contact me for a chat.”

For more information about the trial, you can contact the A-T Society either through **support@atsociety.org.uk** or on 01582 760733.

Major donation from the Thomas Appeal

As some of our readers will know, the Thomas Appeal was a charity set up many years ago by Beverley Hodson and Peter Cottingham to raise funds for A-T research. Named after their son, over the years the Trust raised many thousands of pounds for A-T research, funding a range of different projects.

The Thomas Appeal has recently closed down and the trustees agreed to donate its remaining funds, a sum of almost £15,000, to the A-T Society. We are extremely grateful to the trustees, and in particular Beverley and Peter, for entrusting the A-T Society with spending these funds, and we can assure them that the money will be put to very good use in supporting first-class A-T research.

A-T Society at the House of Commons

On 13 June, at 12:00 pm, Families living with A-T, will come together with staff, volunteers, supporters, clinicians and researchers at the Palace of Westminster for a reception to mark the Society's 30th anniversary.

The reception is being hosted by Bim Afolami, MP for Harpenden and Hitchin, the constituency where our offices are located. Bim has been extremely supportive of the Society since he was elected in 2017, also hosting a very successful dinner for us, last year.

The event will be an opportunity for people from all parts of the A-T community to come together. The aim is not just to celebrate our achievements over 30 years but to say thank you to all those who have helped us on the way and to tell people about our plans for the future. It is also an invaluable opportunity to tell MPs and the Government about the reality of life and the many many challenges facing people living with A-T and similar conditions.

Our 20th anniversary was marked by a reception at the House of Lords, hosted by the late Lord Parkinson. His widow, the Lady Parkinson, who has been a tremendous supporter of the Society over the years will be there again this time, as will be many people who have at some time worked for the Society and for people living with A-T.

However, it is important that we have as many MPs there as possible, to hear our message and to see the strength and commitment of the A-T community, so if you have a chance, please do encourage your MP to come along to the Terrace Pavilion and join us.

New animated film about A-T

We have been working with Madano, a creative agency, to create a simple animated film to explain A-T. The film explains the different ways the condition affects people and how it progresses over time. The film is primarily aimed at people who are coming across the condition for the first time – new families, teachers, fundraisers, PA's etc. but it will also be relevant for healthcare professionals who may want to know a little bit more about the condition or for children with A-T to explain to their peer group what A-T is. The film will be ready for our family weekend in June and we can't wait to share it.

We are extremely grateful to the team at Madano who have been doing some pro bono work since they heard about the Society and A-T.

Welcome to Susie our new Head of Philanthropy

We are delighted to welcome Susie Norbury to the A-T Team as our Head of Philanthropy. Susie will be focusing on developing our fundraising, in particular, support from corporate and major donors and has a wealth of experience in the sector. Susie will be here 5 days a week and will work alongside Sarah and Sophie in the fundraising team. We asked Susie to briefly introduce herself:

Hi everyone, I am so pleased to have recently joined the A-T Society as their new Head of Philanthropy. I have an extensive background in charity fundraising and am delighted to now be able to bring that experience here. My role at the charity broadly involves raising funds from companies and high net worth individuals.

Our corporate and philanthropic partners really do make a vital difference to our work. If you, or any of your friends or family, work for a company and could help to introduce me to them, I would love to hear from you.

I'm really looking forward to helping to raise valuable funds to ensure our essential work with children and families is maintained and our crucial research continues. I hope to meet as many of you as possible at the Family Weekend and in the coming months.

Please contact me at **susie@atsociety.org.uk** or phone the office for a chat on 01582 760733. Thank you!

Becoming a trustee by Jilly Shah

Tell us a bit about yourself

Everyone who knows me will tell you I'm a cake obsessed, fun loving, smiley person. I like to think this is true!

I studied pharmacy at university and obtained my pharmacy degree 9 years ago. I used to work full time as a pharmacist, but I now spend my days doing what I love. Most of what I love is baking and cake decorating! I set up my own cake business in 2015, and all the money I make through that, I donate to the A-T Society.

As well as baking, I love hanging out with my friends. I go to the cinema, theatre, concerts and I love going out to eat – especially when dessert is on offer!

Tell us more about your cake ventures

I set up Jilly's Cupcake House in 2015 and I immediately decided that I wanted to donate everything I earnt to the A-T Society. Cakes and cupcakes are loved by all, and everyone will always need a cake or cupcakes to celebrate and/or remember important events in their life. This gives me a massive audience to not only provide yummy cake, but to raise awareness of A-T and show others that people with A-T can do anything they want to – (including designing and making super fancy cakes!) If you can manage to tell just one person about A-T, that has never heard of it before, already you are making the world that little bit more accepting.

I am baking the world a better place, one cake at a time!

How does having A-T affect you?

When I was younger, I showed no symptoms of A-T. I was diagnosed with A-T because as a toddler I was very unsteady on my feet. However, as I entered primary and secondary school, my balance resolved itself and I was asymptomatic.

It was only until I started 6th form – around the age of 17, I started showing symptoms again. Since then my symptoms have been getting increasingly worse.

At present, the main things that I have difficulty with is speaking, chewing and drinking. This sometimes affects how I feel about going out and interacting with people I am not familiar with. Not because I am embarrassed about myself, but because sometimes people see you as someone who may not understand things going on around them, which leads them to talk about you to others in front of you, or to talk down to you. It is understandable that such behaviour is unintentional and not malicious, but this is why A-T awareness is so important.

How has the charity helped you?

Since primary school, the A-T Society has helped me and my parents so much! I've more often than not been told "no" throughout most of my schooling and education. And the superhero that is Kay has always been on our side fighting against the "bad guys". I remember going through my SATs, GCSEs, A-Levels and even my uni degree, all with the support of the A-T Society and my parents in my corner.

Even now, both Kay and Anne and the rest of the team continue to support me and help me with anything I need.

The Family Weekends and Adult Activity Weekends are also another way in which the Society helps me. Meeting up with other people with A-T is a lot of fun. I have made some good friends from the A-T group and I enjoy talking to and hanging out with them.

How have you found the first few A-T Society trustee meetings that you have been to?

In all honesty, most of it has been a lot to take in. The meetings are focused around numbers which isn't my strong suit, but I like to think I am slowly getting to grips with it.

Is there anything you'd especially like to achieve as a trustee?

I feel like I can help provide a different perspective through the eyes of someone with A-T. I know first-hand of challenges that people with A-T face, so I hope to share my experience and provide valuable input into decisions that are made by the Board.

I can also offer other skills to the Board and the Society such as my frequent and positive use of social media, especially Instagram. I feel like Instagram is a massive platform in which A-T awareness can be

raised as it reaches audiences all over the world simply by using hashtags.

What is your philosophy in life?
I would definitely say "be positive". People underestimate the power of positive thinking. This isn't to say "don't feel sad", by all means, feel sad, cry your heart out if you need to, but thereafter, remember what you have in your life and what's important.

As a personal example, I know positive thinking won't make my symptoms better and I know positive thinking won't cure my A-T. But what positive thinking does do for me is help me get through difficult times so that I can come out of the other side as a stronger person.

If you'd like to order one of Jilly's bespoke cakes then please contact her through her Facebook page: **www.facebook.com/jillyscupcakehouse/**

You'll need to live near London as it's collection only (Jilly hasn't worked out how to post them out to arrive in one piece!)

Remembering Lily and Robert

It is with great sadness that we announce the deaths of Lily Pickering and Robert Soper. They will all be very much missed by their family and friends and have left a huge hole in their hearts.
Here we remember them.

Lily Pickering
Lily Pickering's mum, Rachel wrote: *"Our beautiful princess Lily-Grace Pickering sadly grew her angel wings on the 7th November 2018. Lily was a bubbly little girl and lit every room she went in with her big cheeky smile and eyes. She loved going on little holidays and staying in hotels. Her favourite place was London where she had afternoon tea with Princess Rapunzel followed by a horse and carriage ride to Buckingham Palace a memory we will cherish forever. She is very much loved and missed by everyone who knew her especially her family and friends."*

Robert Soper
Robert Soper who had mild-variant A-T sadly passed away at the age of 62 on 26th July 2018. He was very independent and lived on his own in an adapted bungalow which the A-T Society helped him to move into. His favourite quote was "my computer is my window to the world". He shared a love of sport with his friends and was an armchair commentator on many sports including cricket, rugby and football. Determined to live life to the full Robert enjoyed many adventures. For Telethon '90 he abseiled down the Charing Cross Hospital Tower! Robert is very much missed by his mum, family and friends.

New Wheels for Orla

In May Orla received her new wheelchair which she had been waiting a while for. She was desperate for a new wheelchair as she had outgrown her old one.

Her mum, Sinead, messaged us: *'Thank you so much for Orla's new wheelchair which she loves! The small wheels light up which was a surprise for her and went down very well! We are so grateful for the help of the A-T Society for making this such a positive experience for Orla and helping to fund this.'*

We're so glad that we were able part-fund the wheelchair in conjunction with the NHS. We're also happy to announce that Orla's old wheelchair has found a new home with another A-T family.

Exciting times for Kira as she transitions to Further Education

Kira has been attending mainstream secondary school from the age of 11 and has always loved her school experience. Even after having major scoliosis surgery, followed by gruelling cancer treatment a few years ago, Kira was always determined to return to school as soon as she possibly could.

Kira loves Maths, English and French and recently sat her GCSE exams in each subject. Her mum Hayley said that Kira has made some lovely friends during her time at secondary school. However as she approached her GCSE's Kira and her family knew they needed to begin thinking about 'what next'? Kira was clear she wanted to go into further education, but one of the major and most difficult decisions for her and her family was whether she should stay within mainstream school or start looking at specialist provision.

After much deliberation Kira and her mum visited nearby Portland College which offer further education for people with a wide range of disabilities.

Hayley told us: *"When we arrived at Portland College, the first thing we noticed was how easy it was to get around, no steps, wide automatic doors etc. Also all the rooms had adjustable tables, with plenty of room to get around. Kira has always had the help she needed in mainstream school, but she could never be as independent as I feel she could be at Portland College.*

I would be able to drop her off outside the College and watch her enter the College independently using her powered wheelchair. This is something she has never been able to do before".

Since this initial visit, Kira has had transition meetings with her current school, which Anne was able to attend and with help from her family and school staff she decided to make an application to Portland College.

Kira is now waiting to start at Portland in September this year. Her chosen course is a 3 day week course in Animal Care and Husbandry. She is quite excited about starting college, but a bit sad because of her friends she's leaving behind plus they don't offer French at the College!

Physio and Hydrotherapy will be part of Kira's curriculum and the college also has an amazing fully accessible gym that Kira will be able to access.

Hayley told us: *"It's a scary time at the moment because Kira is 16 and everything is changing: school, clinic, Physio and O.T and of course PIP! Specialist College may not be the choice for everyone, but Kira and myself have spoken about it at length and feel this is the right place for Kira".*

Over the course of a year a gift of £5 a month could pay for information packs to be sent to 12 new families.

2019 is the 30th anniversary of the A-T Society and you can help make it a year to remember! Take part in one of our events or organise your own and we will support you all the way.
For more details visit **www.atsociety.org.uk/events**

Charlie Seal's 'Special Day' at Liverpool Football Club

By Sara Metcalfe

What started out as one of the most stressful weeks of my life became one of the best for my nephew Charlie Seal!

I had booked an Anfield Stadium Tour for Charlie's 20th Birthday in February and arranged to meet him, his brother, Arthur, and their Dad, Richard, the day before in Liverpool. The Travelodge was booked, Frankie and Benny's were warned we would descend on them for virtually every meal and house sitters booked to look after horses and dogs. Then a couple of days before I received an email from Anfield saying all tours on the Friday were cancelled due to "circumstances beyond our control!" To say I went into panic mode was an understatement!

Anyway with a quick re-organisation we rearranged for the Thursday and everyone agreed to set off earlier that day. Then out of the blue the Willow Foundation got in touch to ask if Charlie could get to Anfield on the Friday as they had arranged his Special Day to visit the ground and meet the players! Well we couldn't believe the coincidence as obviously accommodation was already booked and we were all going to be there. However being the control freak that I am known to be, I rang Anfield just to check the tour could happen as the email to me had advised tours were cancelled – I was assured by the club that it would be going ahead and when I checked with Willow I was assured of the same.

I cancelled our booked tour for the Thursday safe in the knowledge that the best day for Charlie was to come the day after…..we all went to the grounds for a recce and a meal at the Boot Room and Charlie bought lots of items for the players to sign the next day. Charlie loved being at Anfield and soaked it all up getting very excited for what was to come.

So off Charlie, Arthur, Richard and Jasper, my son, went on Friday after having a Birthday breakfast at the next door McDonalds! Sadly Charlie could only have 3 people with him so my husband and I would be going to have a tourist day in Liverpool instead. A short while later I got a phone call from Richard saying that all tours on the Friday were cancelled and it was not possible to get in!!!!!!!!!!!!!!!!!!!!!!!!!!!


Charlie and Mo Salah.

What followed was some frantic phone calls to Liz Hooper at the Willow Foundation and to Anfield to find out what I could do to sort this out, knowing what a huge disappointment it would be to the Birthday boy. We eventually found


Charlie and his aunt Sara.

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You shop. Amazon gives.

If you shop at Amazon, why not sign up to Amazon Smile and select the A-T Society as your chosen charity, that way we get a donation from them every time you shop! All you need to do is start your shopping at **smile.amazon.co.uk.** The donation will be made at no extra cost to you.

out that the reason all tours were cancelled was because there was a top secret launch of the new kit for the players and the promotional photos were all being taken at the club.

However, a fantastic chap called John Dowber, who had been assigned to look after Charlie on the day, rallied round taking them everywhere he could in the stadium including some areas not normally open to anyone from the public and in the end they all had a fantastic insight into the club.

Salah. They were there for quite a few hours and I really do think it was one of Charlie's best days ever! He was still buzzing weeks after and was on a high when we all met up after.

The next day Phil and I took him for a free stadium tour generously given by LFC and he saw all the areas he had not been able to access – he got to come down the tunnel and onto the pitch which he had been desperate to do. Every single person at LFC went beyond the call of duty to help us and we could not have been made to feel more welcome.


Charlie, his brother Arthur, Jasper and Jurgen Klopp.

Then off he took them to the players training ground where Charlie was ecstatic to meet every player and the manager and saw them train. He had lunch where the players ate and was looked after incredibly well by all the Liverpool staff. He got lots of free programmes that various players signed, his team shirts were signed by every single player and he had photographs taken with each and every one of them. Charlie was absolutely "buzzing".

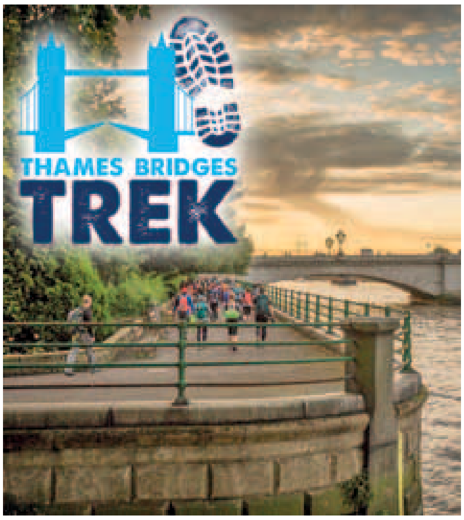
He says the best bit was meeting the players and having lunch with them and his favourite player is Mo

I know that Charlie had a fantastic time and what a birthday day to remember. From us all we need to thank the A-T Society, The Willow Foundation, The Boot Room at Anfield and everyone involved from the players, to the manager, to the stadium guide and the ladies at the tour office. Everyone was quite frankly awesome!

And I have a LFC shirt signed by all the players to auction off for the A-T Society so watch Facebook for details of that in June!!!

Willow's Special Days are designed to provide an uplifting and positive experience to be shared with family or close friends. From afternoon tea and theatre trips to sporting events or theme parks, there's a 'Special Day' experience to suit everyone.
To apply visit **www.willowfoundation.org.uk**

A-T London Bridges Walk



Join our Community and Events Fundraising Manager, Sarah Ajder, and 2,000 other trekkers on the London Bridges walk on Sunday 7th September.

The walk sets out from Putney Bridge and heads east towards the City, zig-zagging over the array of historic bridges – each with its own fascinating story – and a mid point stop in Vauxhall for some snacks and drinks. 25km later, it's time to celebrate as you reach the finish line in Southwark past the majestic Tower Bridge.

It's a great opportunity to set yourself a new challenge and see some of London's iconic landmarks on foot.

If you fancy putting your best foot forward and keeping Sarah company on this walk then please do get in touch – **sarah@atsociety.org.uk**.

There is a choice of registration packages with varying fundraising commitments to choose from and the fundraising team are ready to cheer you on!

For more details and to sign up visit **www.ultrachallenge.com/thames-bridges-trek**

Our sporting A-T Heroes

The A-T Society's philosophy is that everyone with A-T should have the opportunity to live their life to the full and participate in the activities that they love. It is both inspiring and motivating to see them taking on personal challenges and pushing themselves to reach their goals. Here some of our children and young adults with A-T share their sporting journeys and achievements.

George Keith

George Keith rekindled his love of playing Boccia after taking part in the sport at one of our Family Weekends. His mum, Michele, tells the story.

After the Family Weekend 2017 where George took part in Boccia, he really wanted to find a club to join. We found one 5 minutes away from where we live! So in October 2017 George started playing Boccia weekly, where he met some old school friends and made lots of new ones!

Nick, George's dad, also enjoys going with George as I do, but in April 2018 Nick decided to take the Level 1 Referees course with Boccia England. After this he became more involved with the club, helping out as a referee. He enjoyed it so much that in December 2018 he took the Level 1 Coaching Course with Boccia England and passed with flying colours! Since Christmas Nick has really concentrated with coaching the teams at the club, with great results. Two of the players are through to finals of the Boccia England Cup in Sheffield this May!

After only taking up the sport 18 months earlier, George took part in his first singles competition, the regional competition in Crawley in March. He won his pool and came second in his group and finally came 4th overall! We were so proud of him as we didn't expect him to win a match as most of these players have been playing for years – one of his competitors had been playing for 20 years! We also met the lovely Ebony who was also competing there!

The following week we got an email to say George had made it through to the Finals of the Heathcote Cup taking place in Nottingham in April! We were so shocked and overjoyed. George's face was a picture! He was so excited and his self esteem was sky high!

The three of us went to Nottingham. George was the only person from his club to get through to these National Finals, although 8 took part in the regional competition! George played very well and most matches were very close, but he didn't win a match. However, he came 9th overall and that's 9th in the country so something to be very proud of!

Nick and George have learnt so much from this experience, especially from playing against people who have played for years. George loves playing Boccia because it is a good social event, helps him mix with different ages and disabilities and is good physio for him. Also it has helped him mentally as well, it's made him feel good about himself and that he can achieve what he wants to if he puts his mind to it!

Nick really enjoys coaching as he loves interacting with the people in the club and helping them improve their game. He is hoping to take his Level 2 Coaching Course this December and then he could coach at Academies! He is also hoping to set up his own Boccia Club, so fingers crossed it all works out.

Playing Boccia has changed our family's life, so why don't you give it a go!!

Boccia is a Paralympic target sport, similar to bowls, which is played indoors. It is currently the fastest growing disability sport in the world and is played in over 50 countries worldwide. There are currently over 4,600 people regularly playing the sport in England. To find your nearest Boccia club, visit **www.bocciaengland.org. uk/find-a-club**.

Ebony Robinson

Ebony Robinson has been playing Boccia for over 10 years and is now training at her local club to become a Level 1 Boccia coach.

I have enjoyed playing Boccia since I was 12yrs old. I have recently been offered the opportunity to become a coach with my local Boccia club, which I attend every week. I really feel that I have what it takes to become a good team leader and help others like myself to achieve and follow their dreams.

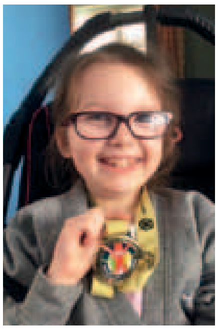
I compete in Boccia and most of my teammates say I have good sportsmanship, but I say I am not competitive like a 'man' LOL!

If you have found an inclusive sport that you love to take part in please consider sharing it with the rest of the A-T community – who knows your story could inspire someone else to give it a try. Email **kate@atsociety.org.uk** with your story and send some photos too!

Toni-Jo

Gold Medal for Toni-Jo!

Congratulations to Toni-Jo and her team mates from Burnham Primary School. They competed in the South Essex Primary Ten Pin Bowling competition as part of the Pentathlon Challenge and won gold medals! Well done – what a fabulous achievement!



Charlie Stubbs

Charlie with his brand new football wheelchair

Charlie Stubbs has always had a passion for sports – whether it's watching his beloved football team Liverpool play or taking part in competitive sports like wheelchair football or Boccia. Here Charlie tells us about the sports he likes to participate in:

Wheelchair football

I play for Portsmouth in the South East League. I like to play wheelchair football because I feel not only the passion to win, but also the enjoyment of playing with my mates.

Another thing about wheelchair football is no matter what age or gender, you can play. You need a specialist chair to play in, with a guard to hit the ball, and most clubs will have chairs you can have a go in. Below is a link that you can search to see if there is a club local to you to have a go. It's good fun. **www.thewfa.org.uk/Pages/FAQs/Category/club-list**

Boccia

This sport is good for my fitness and of course competing at a competitive level. When you enter competitions you have to go though a classification process to make sure you are competing with others with similar abilities. I first played Boccia when I was at school and college. I am going to start training again soon and hope to compete in the near future. I have secured funding for a set of balls and am going to hire a sports hall to do some training.

Gym

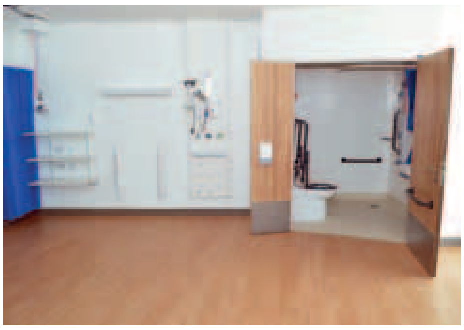
I go to the gym every week to keep me fit and healthy and now that I have left college it's good for my social skills.

Swimming

I incorporate some of my physio programme in the pool and swim independently with my easy-breather mask. Below is a link to the mask I use. **www.decathlon.co.uk/easybreath-mask-blue-id_8304664.html**

As you can see I am very active as I love sports. If you have any questions please forward them to the A-T Society and I will try to assist you where possible.

UK's National Ataxia-Telangiectasia Service for adults is moving to Cambridge



Natasha Everett is the Ataxia-Telangiectasia Coordinator at Royal Papworth Hospital. Here, she talks through the hospital's move to Cambridge and offers re-assurance that services will remain exactly the same at the new location.

The UK's National Ataxia-Telangiectasia Service for adults, which we run at Royal Papworth Hospital, will be based in Cambridge from May 2019.
We are moving from our current site in Papworth Everard in Cambridgeshire to a state-of-the-art hospital 15 miles away on the Cambridge Biomedical Campus, to the south of Cambridge city centre.

Royal Papworth has been managing the national A-T service for adults for more than a decade and has gone through many changes over the years. It was commissioned as a National Service by the National Specialised Commissioning Team in 2012, a coordinator was introduced to the multi-disciplinary team in 2014, as well as adding neurological investigations to the programme and MRI breast screening for women with A-T.

The team comprises many specialities including; a consultant respiratory physician, immunologist, neurologist, physiotherapist, occupational therapist, speech and language therapist, dietician, social worker, and service coordinator.

A-T is a rare condition with around 200 people in the UK affected, therefore the centralisation of expertise is essential in order for patients to receive care by staff who fully understand the disorder and who can support them throughout their life.

At present, Royal Papworth sees more than 80 A-T patients a year for their annual reviews, ensuring that all aspects of care are addressed to provide the best management plan. The service will continue to be hosted by the Respiratory Support and Sleep Centre, which will be based in the northern section of the third floor at the new hospital, known as 3 North.

Patients can expect bigger rooms with larger windows, allowing for plenty of daylight and panoramic views of the outside world compared to the restricted views currently. Each room will be fitted with brand-new equipment including beds, monitors and other medical devices.

Treating patients from all over the UK can mean that some people who travel from metropolitan areas feel cut-off in Papworth village, but the new hospital will have amenities onsite including WHSmith and Costa, in addition to the restaurant and more options at Addenbrooke's Hospital next door, plus free wifi for everyone.

Being based on the same campus as Addenbrooke's will also allow for an easy transfer for people who need to go there for further tests and investigations.

Cambridge is also much better served by public transport with regular bus services, a guided busway, nearby railway stations and an airport.

We are aware that familiarity is absolutely critical to our A-T patients when they visit the service. Crucially, therefore, despite the move, the exact same schedule as now will be followed from the moment a patient arrives through to the moment they leave. Additionally, the landscaped grounds surrounding the six-floor building includes a tranquil duck pond, just like at the current site.

It's also important to reassure our patients that whilst the new site will look and feel different initially, our staff will be making the journey across too, and they will continue to provide expertise and safe care to people living with the symptoms of A-T and their families.

We look forward to welcoming you to our new hospital from Sunday 12 May.

If you have any further questions, please contact Natasha on:
01223 639452 or
Natasha.Everett1@nhs.net

Fundraising

THANK YOU! We think you're all AMAZING!

Another successful HCO concert was held at St Nicholas's Church in Harpenden, this time boasting an impressive performance from Internationally-renowned 'cellist Guy Johnston.

Helen Skippen inspired her colleagues at Equiniti to raise money with another dress down day at work.

David Campbell-Smith organised a successful Golf Day.

Peter Cowan, Chairman of the Concrete Society, paid us a visit and presented us with a rather splendid cheque.

M&S Bridgnorth generously supported us as their Charity of the Year for the second year running.

Inspired by her niece, Daisy, Claire Magee put on a fabulous Body Shop party to boost her Brighton Marathon fundraising.

Thank you to all those who have used Facebook to think of us and raise money by asking friends and family to donate money instead of giving birthday gifts.

A fantastic, festive celebration with Peter Knapp was held at the Rothamsted Theatre organised by Mike Hodge.

Santa's Sleigh travelled around St Albans spreading joy and collecting money, coordinated by the St Alban's Round Table.

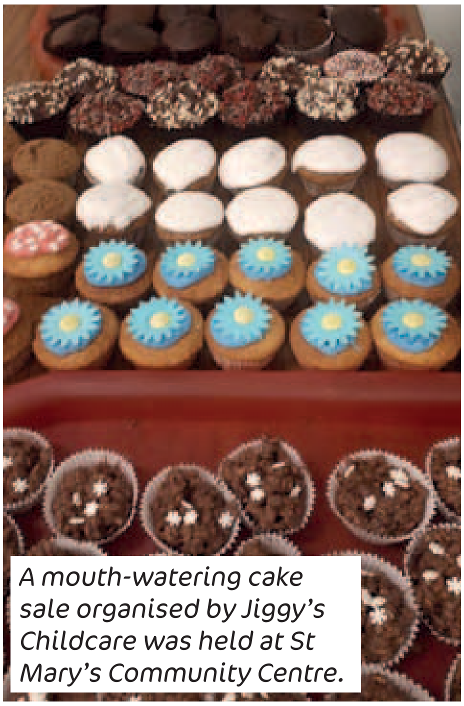
Harry Muddle dressed as his sister Lizzie and spent the day in her wheelchair telling pupils what it is like to be her brother, he also organised a cake sale.

Charlie Reoch took on the Village Bakery Half Marathon in Wrexham

Santander in Bromley hosted a charity day and cake sale organised by Emma Roe.

A glamorous evening at Elizabeth Arden, Boots, St Albans, saw our very own Kate McEleney being transformed as a makeup model! Thanks to Wendy Seabrook and team for organising.

There are plenty of ways you can support us and not all involve sweat and tears!! Why not shop 'till you drop from the comfort of your own armchair? Every pound you raise will help families live well with A-T all year round. Sign up to Give as you live or Amazon Smile and select the A-T Society as your chosen charity and every time you shop they will send us a donation.



A mouth-watering cake sale organised by Jiggy's Childcare was held at St Mary's Community Centre.

Superfit supporters Rachel and Chris Gutteridge completed a gruelling 10-hour CrossFit Challenge.

Joy Ferguson and the amazing Concordia Choir sang their hearts out at a number of carol singing events over the festive period.

A brilliant quiz night was organised by Nora Roe and The BT Tower Social Committee. 22 teams battled it out and made the evening a huge success.

A Charity Business Breakfast was hosted by the Harpenden Business Breakfast club.

A wonderful concert of Music in Film was put on by Tea Killick and the Horam Vox and the Whealson Youth Choir.

Brighton Marathon 14th April 2019

We do love to be beside the seaside, especially when we are supporting our A-Team of Brighton Marathon runners!

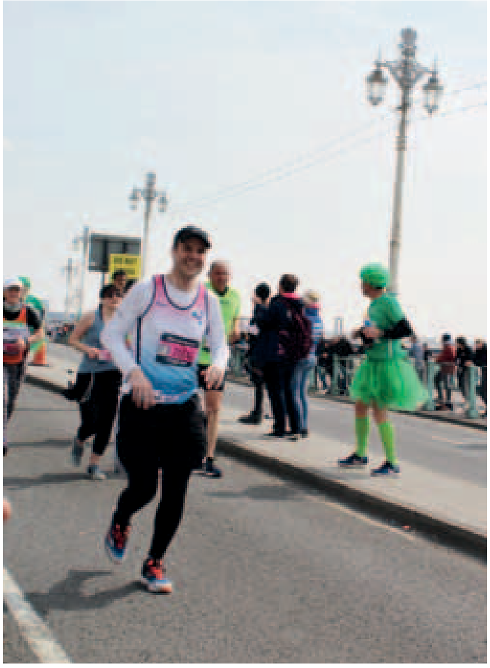
Congratulations to Alice Budden, Charlie Beevor, Claire Magee and Pedro ZuaZua for completing their first ever marathon. The many hours they dedicated to training through the winter months were worth it when they crossed the finish line to receive their chunky finisher's medals.

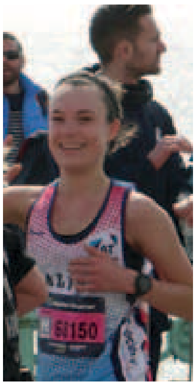
William, Lian and Sarah were pleased to thank three of them in person at our post-race tea party and meet their families and friends, some of who joined us at our cheering spot outside The Old Ship Hotel on the seafront.

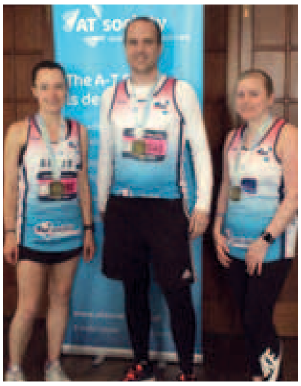
Julian Thomas ran in our 2016 Brighton team. We were delighted he chose to run for us again as one of four charities he was supporting this year. We tracked our team on the event app and saw when Julian and Pedro crossed the finished line but we didn't see them on the course or at our post-race party. They remain the invisible men in our team who we didn't manage to catch on camera!

Our team also committed a huge amount of time and energy to their fundraising and didn't stop when they reached the minimum required but kept on going to raise as much as they could. Thanks to their marathon efforts they have jointly raised over £6,000!

Claire and William have already claimed their place in our 2020 team! If you would like to join them do let us know.







Inspired to run a marathon? Come and join our Brighton Marathon 2020 A-Team.
We'll support you all the way and will be there to cheer you over the finish line.
For more information call Sarah Ajder on 01582 760733 or email **sarah@atsociety.org.uk**

Glamorous Fundraising A-T Ball

A-T parent Emma Roe has taken to fundraising like a duck to water!! Having organised a successful charity day at work Emma set her sights on organising a fundraising A-T ball.

She showed herself to be a very capable fundraiser and soon had the venue sorted. Tickets sold like hot cakes and with the help of family and friends some fabulous raffle and auction prizes were donated.

Farleigh Golf Club made a superb venue and looked fabulous with A-T balloons as table centres and very attractive displays of the many raffle and auction prizes on offer.

Guests enjoyed a wonderful evening. Emma shared the value of the support given to her and her family by the A-T Society since her young son Jack was diagnosed with A-T, and William was pleased to thank everyone for helping to make the evening such a huge success.

Emma took our breath away when she told us that £14,001.09 had been raised during the evening! Her pride beams out from the beautiful photo of her with her sons Jack and Harry with the giant cheque showing the total raised. We share their gratitude for the love and support given by their friends and family and thank Emma and her husband Tommy for being superstar fundraisers.

A donation of £10,000 could buy an all-terrain powered wheelchair and give a young adult with A-T the freedom to travel wherever they want.

‘Incredible’ Hulks run Hastings Half Marathon for Chief Hulk Winston and the A-T Society

Inspired by 5 year old Winston who has A-T, a small team dressed as The Incredible Hulk ran the 2018 Hastings Half Marathon to raise money for A-T Society. Winston loves the Hulk so dressing up as his favourite superhero was a great way to raise awareness, and it worked!

Encouraged by their success Joe and Sara Pemberton-Powell decided to ‘go large’ and aim for 100 Hasting Hulks in 2019. This incredible green team caught the imagination and on a glorious sunny day in March this year more than 50 Hulks ran the Hastings Half Marathon for Winston and the A-T Society.

Lian Yarlett, one of our trustees and an A-T parent, reports from a memorable day.

“It was a glorious sunny day and a team of enthusiastic volunteers arrived early to help set up A-T Hulk HQ. There was lots to do but we had some helpful Hulks on board! The marquee looked fantastic, decorated with handmade Hulk bunting and was soon filled with a selection of homemade cakes kindly baked by local supporters.

Hulks arrived with family and friends and a couple of the Hulks’ dogs joined the humungous hoard! Green face paint was expertly applied and we were impressed by the effort made by the team to dress the part. They were Hulktastic!

Drumming band Sambalanco revved up the atmosphere and Sarah, from the A-T Society, said a resounding thank you before everyone cheered the Hulks off to the start line. Joe, Winston’s dad, was injured so he was pushed around in a wheelchair – no football injury was going to stop him from participating! The incredible team rallied round to make sure he made it and Winston sat proudly on his lap as he crossed the finish line”.

These amazing Hulks did themselves and everyone proud. To date they have raised the truly incredible total of £20,880! In recognition of this big team effort they were awarded the Dyer Charity Shield by the Hastings Lions Club as the team who raised the most for charity in 2019. A very well-deserved award.

Well done and THANK YOU to the Pemberton-Powell family, the Hulks, cake bakers, bucket collectors and everyone who contributed in any way to make this event a HULK SMASH!

If this sounds like an event that is too good to miss then why not join the Hulks in the Hastings Half Marathon on Sunday 29th March 2020 and help them reach their goal of 100 Hulks? There is no focus on running ability or athletic prowess so any Hulk who signs up to take part is a hero in the family’s eyes.

Visit their Facebook page **www.facebook.com/hundredhulks** or e-mail **sarah@atsociety.org.uk**

£250 could buy Clicker 7 adaptive software to help a child with A-T build confidence with literacy at school.



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 and all data collected will be stored and protected in accordance with the UK data protection laws. If you are happy for us to do this please tick this box. ☐