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A-T Society News

In this month's edition

- The Family Weekend takes off at Stansted
- Top tips from people with A-T and their families
- How to protect your family and assets
- Clinical Research Conference

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 all people living with A-T. The Society aims to improve quality of life and quality of care for people living with A-T while actively promoting research to lengthen lives and ultimately bring about a cure. We do this by: providing information and support; working to achieve better health and social care services; promoting and funding high-quality research; speaking out to ensure that the voices of people affected by A-T are heard; and raising awareness of A-T.

Editor's Comments

Many thanks to all contributors. The copy date for the next issue is 1st October 2012
Please send comments, ideas, articles and pictures to:

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If you would like to receive this newsletter by email as a pdf file, please let us know.

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Cover picture: Children being entertained at the Family Weekend

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View from the chair

Lian Yarlett, chairman of the A-T Society, looks back at a busy 2012 so far, which has included significant conferences, charity events, the Family Day and proposals by a new joint A-T fund

Hopefully the sun is shining wherever you are so that you can enjoy reading this information-packed edition of the Newsletter. 2012 has so far been packed with conferences and meetings, with the Family Day being one of the main highlights, along with charity events and Charity of the Year adoptions. All this on top of the regular care services we provide!

The conferences have led to concrete collaborations and sharing of information wherever possible, with the aim of advancing research and our understanding of A-T.

There is a big focus on translational research – that is, research which aims to develop treatments directly for the patient. Together with the advances in laboratory-based work, this is all helping us take strides towards our aims and mission.

The international research conference we have just hosted has led to specific projects being discussed or commenced, and a number of interesting proposals for funding by the new the joint fund we have set up with Sparks and Action for AT.

There is a huge amount of work that can be undertaken for our support and research but of course much of this is down to funding. Our care services and clinics give our children and young adults help, and I think this is really important. Some of the research also offers the chance of quite prompt outcomes, as well as creating a basis for future developments.

We are very grateful to everyone who has helped to make a difference, and invite all of you to help us take on the challenges ahead for what is looking to be a really exciting time with many opportunities. Do have a think about what you might be able to do, who you might know, or might not yet know, be it friends or companies. We can always help you with that first idea or make that first approach.

The Family Day is always a great event and we have received some wonderfully positive and constructive feedback. This year's weekend was packed with fun and information, and if you missed it we really hope that you will be able to join us next year – the plans for which we are already working on!

We are making a real difference to the lives of those affected by A-T now and for the future…
Please help us to do more. Thank you.

Lian

"Help us take on the challenges ahead for what is looking to be a really exciting time..."

DATES FOR YOUR DIARY!

Nottingham Clinic
If your child has not been seen at our Nottingham Centre for at least 2 years, or your child's condition has deteriorated and you are concerned: you may wish to book one of the available slots below: (please contact Kay)

15/16 November 2012
17/18 January 2013
7/8 March 2013

Family Weekend 2013
Provisional date set for 22-23 June 2013

Just William

William Davis, the Society's chief executive, reflects on a successful and enjoyable Family Weekend and a Conference that has helped stimulate new ideas and collaborations

I write this with the memory of two highly successful events still buzzing round my mind. The A-T Clinical Research Conference 2012 in Cambridge brought together scientists, doctors and researchers from around the world for two days of engaged discussion on the way forward for clinical research into A-T.

Following straight on from this, our Family Weekend saw another huge turnout of families gather at Stansted to hear from leading researchers and to network and relax with others like themselves.

There are more details about both events later in the magazine but I just wanted to share my overall impressions.

I will admit that I thought last year's family weekend at Chessington had been a bit of a high-water mark and was expecting this year's event to be more low-key. Wrong on both counts! The prospect of hearing first-hand from some of the world's leading A-T experts and the importance of being able to meet and chat to other people living with A-T meant we had more than a third of all the families in the UK together.

Splitting the sessions over two days seemed to work well, as it allowed for a relaxing evening in between. Kay and Suzanne did a fantastic job in transforming a lecture hall almost magically into the scene for a memorable 'banquet'. And while the younger visitors were entertained by a highly creative disco, the rest of us were able to sit around and chat with old friends or make new ones.

"I found myself at a meeting with three delegates from the UK and Australia who had met at the conference... We mapped out a proposal for a two-year research project which would link with existing projects in the USA and Australia"

As to the conference, this was felt by all those who spoke to me to have been a resounding success. The venue was excellent and the staff fantastic. This meant that the delegates were able to put all their energies into the conference. They clearly appreciated the format we had chosen, which allowed plenty of time for group discussion at the end of each session.

The result was that instead of just a series of individual presentations, we were able as a group to map out priorities and likely avenues for future research.

But a conference like this will only have a real value if it changes things; if it stimulates new ideas and new research, gets people working together, and so on.

And on that count, things are looking good. Just 10 days after the conference, I found myself at a meeting with three delegates from the UK and Australia who had met at the conference.

We discussed how the latest advances in MRI and imaging technology could help unlock the mysteries of how and why A-T affects certain areas of the brain. We then mapped out a proposal for a two-year research project which would link with existing projects in the USA and Australia.

At the conference, I also announced the new joint A-T research fund *(see next page)*. This was greeted very positively and as I write this I have had 12 expressions of interest in submitting applications.

Our job now is to make sure that we turn these ideas into real projects and concrete progress. However, this will require hard work and of course money.

Money is not that easy to come by at the moment and that means we are all going to have to roll our sleeves up and redouble our efforts to raise new funds.

But the good news is that there are ideas, enthusiasm and a spirit of cooperation in the A-T research community – and those are things you can't just buy.

Remembering Paula

It was with huge sadness that I heard Paula Hendrick has died after a short illness. Anyone who met Paula will have vivid memories of her – her humour, cheerfulness and warmth, not to mention her determination not to let A-T stop her from being the person she wanted to be and living her life the way she wanted to. Paula's positive attitude to life was an inspiration to many – though her ability to be the 'last-woman-sitting' in the bar at family weekends – "The night is young!" was her rallying cry – resulted in a fair few headaches and bleary eyes for the rest of us.

We are enriched by everyone we meet, but some people are particularly special, and Paula was one of those. I know that the thoughts, wishes and prayers of many, many people will be with her family and I trust that this will be of some comfort to them. **By William**

A-T in Morocco

While to the best of our knowledge there is no organisation dedicated to A-T in any Arabic-speaking country, in Morocco there is an active association supporting people with all Primary immune-deficiencies, called 'Hajar'.

The Secretary of Hajar, a young woman called Bouchra Benhayoun, has a daughter called Rita who has A-T. Bouchra is in touch with around 30 families living with A-T in Morocco, a country where the challenge of living with a disabling condition is considerable.

The association is doing an extremely impressive amount of work to raise awareness of immune-deficiencies in the country and to support the development of a specialist centre at the hospital in Casablanca. They have also helped children with primary immune deficiencies to access information drugs and equipment and to improve standards of treatment and care.

More information on their work is available, in French or Arabic, from their website: **www.hajar-maroc.org**.

If you would like to talk in Arabic to someone who understands A-T, Bouchra would be delighted to do so and can be contacted either through the website or via the A-T Society.

UK A-T service promoted in Brussels

In May, William presented a poster on the A-T service at the conference of Eurordis, the European alliance of organisations representing people with rare diseases. The conference brought together 700 people from 55 countries. The poster highlighted the benefits for patients with rare diseases of having a patient organisation, like the A-T Society, working in close partnership with national expert centres, as we have in Nottingham and Papworth.

New partnership for A-T research

As mentioned in William's diary, the Society has recently joined forces with Action for A-T and the children's medical research charity Sparks to establish a dedicated fund to support A-T research. The fund is offering an initial pot of £90,000 for research projects leading towards treatments for A-T.

Launching the partnership, William said: "Together we can make a much bigger impact than we could working on our own. Collaboration is the only way to make progress with a rare and complex condition such as A-T and our three organisations are showing the way."

By pooling their resources in this way, the charities will be able to offer funding on a scale beyond that which they could achieve individually. It will also enable them to combine their fundraising efforts for A-T research. Already, Sparks have very generously supplied both the A-T Society and Action for A-T with three extra places each for the 2013 London Marathon.

As a well-established medical research charity, Sparks already has very effective materials and procedures in place for handling and assessing applications for research funding and also for monitoring the progress of grants that have been made. They will deal with practical administration of the grants programme.

We hope to be able to announce the first grants in October 2012.

ACTION FOR A-T

Sparks THE CHILDREN'S MEDICAL RESEARCH CHARITY

Family Weekend takes off at Stansted

More than 200 people affected by A-T gathered at Stansted in June for the annual Family Weekend

Stansted airport is normally a place to get through as quickly as possible when you can't wait to be on holiday or to get home. But in June, it was the scene of an amazing event: the 2012 A-T Society Family Weekend.

After last year's huge gathering at Chessington, we had expected a fall in attendance– but no, once again we had over 200 people from over 50 families living with A-T from across the UK and beyond.

The programme was packed. The list of speakers and members of the Open Forum panel read like a 'Who's who?' of international clinical research in A-T. We had a great evening banquet with a magician round the tables and disco for the children. And the following morning we had a series of excellent presentations and workshops ending up with a fantastic 'Glee' performance put on by children with A-T and their siblings.

For many people, though, it was the coming together that was the most valuable part of the weekend; meeting other people with the same condition or feeling part of a group with others facing the same problems.

The panel at the Open Forum

Speaker Karl Herrup

Rupert Prokofiev and his mentor Anne giving their presentation on independent living

William Whiltehouse clarifies a point

Saturday afternoon

We were very fortunate to have a line up of speakers, fresh from the Clinical Research Conference in Cambridge.

Luciana Chessa: talked about the recent trial she led of dexamethasone delivered through infusion into red-blood cells. She described how the technique did not produce any of the side effects normally associated with steroid treatment but for some patients it had been difficult to get their cells to absorb the drug. She said the trial showed that there was generally an improvement in the neurological symptoms of those that did absorb the drug and that they hoped to go ahead and carry out larger-scale trials in the future.

Richard Gatti: explained the background to the work his team has been undertaking to find and develop SMRT (small molecule read-through) drugs to help treat people with A-T who have a nonsense mutation. They have identified some promising drugs, but now need the support of a pharmaceutical company to take the work forward. This is hard to do but they are making good progress.

Yossi Shiloh: gave an overview of the complex role of ATM in maintaining the stability of DNA and cells. He also described the work his team is doing screening thousands of small-molecule compounds to identify drugs that have an effect in overcoming the lack of ATM.

Karl Herrup: gave a very clear explanation of the work he has been involved in which involves intervening in the extremely complex chemistry of the genetic processes within the cell to compensate for the lack of ATM. His work has shown positive results in the lab and in mice but there is a long way to go before there is any possibility of a treatment for humans.

Sharon McGrath-Morrow: described the different types of lung problem that could develop in people with A-T and the different approaches that she would take to treating these. She also talked about the studies that they have done to try measure the development of lung pathologies.

Kate Sinclair: described the work of the A-T clinic in Brisbane and how this is closely linked to research work, in particular the tractography imaging that they are doing. She also underlined the benefits of doctors and researchers working together and sharing information and the impact this has had in other conditions.

Their presentations were filmed and can be seen on our website at www.atsociety.org.uk/researchers-speak.

While some speakers had to leave early, the rest were joined in answering questions in the Open Forum by Tom Crawford and Howard Lederman from the US A-T Clinic, Martin Lavin from Brisbane and Malcolm Taylor from the University of Birmingham – truly a 'Dream Team'. They answered a wide range of questions on clinical and research questions.

Saturday evening

Our evening banquet created a magical atmosphere – which was helped by the excellent magician who went from table to table, entertaining children and adults alike.

After the food, the children moved next door to a disco, the DJ of which had everybody involved. This left the older teenagers and adults to sit and chat over a drink – something which is always a highly valued part of our family weekends.

Sunday morning

After a really good breakfast, the morning started with the AGM, an important event in the Society's year. The AGM saw the formal election of the six new trustees who joined the Board during the year. Marian Barber, Naz Hussain, Viv Levy, Ian McInnes, Laura Rafferty and Tania Wheeler bring a wide range of skills and experience and an enormous enthusiasm to the Board.

After this, Bryony Beresford from the University of York gave initial feedback on the My Life project. This project has involved interviewing young adults with A-T between the ages of 16 and 26. The interviews have only just been completed and the information will be collated properly over the summer, but it looks as if it will be extremely interesting.

Bryony's presentation was followed by a presentation by Rupert Prokofiev and his carer and mentor Ann. Rupert, who has classic A-T, talked about living independently, how he had arranged his home and how he recruits and manages his carers. Many people found this an extremely inspiring presentation.

For the final session, we divided into workshops. One of these was run by Bryony, another was with therapists from the Nottingham and Brisbane A-T clinics. Janet Corderoy and Sarah Jessop of the Nottingham clinic were joined by Jo Wilkinson and Janine Anderson from Brisbane. Others stayed in the main hall for a consultation event on what they felt that the Society should be focusing on.

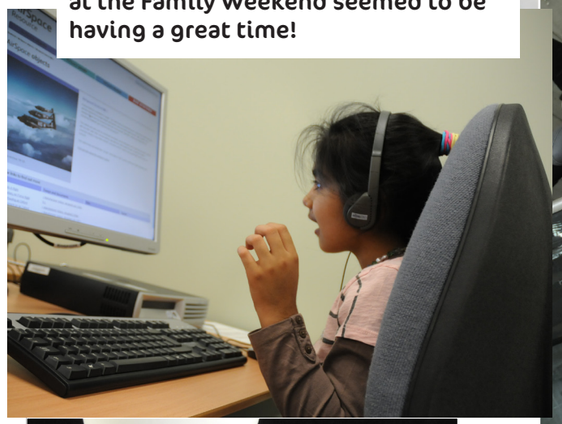
There was also the opportunity for people to make individual or family appointments with our counsellor Helen Hart; and also with the solicitor Robert Smeath of our corporate sponsors Clarke Willmott, who was providing professional advice on making wills and trust-funds for children.

The whole weekend finished on a real high with a "Glee Club" performance by some of the older children to the music of Elbow, Jessie J and Take That. Even though they hadn't had much more than a couple of hours to put together and practise the show, they did an amazing job and the audience were left in no doubt that children with A-T greatly enjoy singing and dancing! The finale sent everyone off home with a smile and a spring in our steps.

Turn over to the next page to see what the children got up to...

The young crowd take to the dancefloor at the disco!

Whether listening to the entertainers, going round Duxford or working on their Glee performance, the children at the Family Weekend seemed to be having a great time!



Voice recognition systems

One family's experience

Merry (age 11) has been struggling with making herself understood in unfamiliar situations for some time and so her speech therapist suggested that a 'Voice output communication aid' (VOCA) might help her. A VOCA is an electrical device that assists people who are unable to use natural speech to express their needs and exchange information by speaking for them.

She organised a review where Merry tried out lots of different machines , eventually opting for one called the ECOII because it had a bigger screen. We also looked and opted for Unity with 64 buttons. We spent a lot of time at different options for the software, to be sure that the system would do what Merry needed. The Unity software she eventually chose takes a bit of learning. It isn't as intuitive as some other options but is more sophisticated linguistically.

The more buttons on the screen the quicker the possible communication but we had to weigh up Merry's accuracy and visual abilities and the 64 buttons with a screen guard gives her a good balance.

At first we had it out every day, and had fun programming in new vocabulary and adding some recorded sounds (fart noises etc!). We found that in some situations Merry was very keen to use it, at home over breakfast and for mental maths lessons for instance. At her mainstream school they also used it a lot to begin with. However Merry was very resistant to taking out and about, refusing to take it to Brownies or shopping , as she didn't want people to think she couldn't speak. Gradually over the three month trial period Merry's use of the ECOII declined, although she still enjoyed the sessions with the SALT.

Merry using her voice recognition software

After the machine went back, we had to decide whether to continue with a VOCA at all. As Merry was about to begin a new special school where they are experts in the use of VOCA equipment, we decided to wait and see what their opinion was. The SALT got Merry, on long term loan, a Liberator 14, an older version of the ECOII, but which still uses the Unity software.

Her new school has offered Merry much more structured sessions using the machine and this has been very successful, especially as other children use VOCA devices too. The ECOII is very clever and can do all sorts of stuff as well as speak. You can put sim cards in and text from it, use it as a remote control and Merry uses it for rolling dice when she's trouncing us at Yahtzee!

I think that we will look to replace the borrowed machine with one of her own and look on it as part of a total communication solution. Merry can talk and does well speaking, except when she's tired, when it can be a real big struggle for her. Of course there is physical effort required in using the machine but when she has to repeat herself several times to make herself understood and then gives up because we're not there to translate, then it will be useful.

Support grants

If you or your child is under 18 and in need of equipment (that cannot be funded by local/statutory funds) then please do contact Kay at the A-T Office. We currently have specific funds in place for items such as; bathroom equipment, mobility aids, wheelchairs, trikes, computers, hoists and any other equipment which is for the child or young person below 18 affected by A-T.

The rough guide to accessible Britain 2012 has reviews by disabled visitors, ideas on days out and parking information. Free to download: **www.accessibleguide.co.uk**

Kay now works full time: Her hours are Monday-Friday 8.30am-4.30pm 01582 760733. Email **kay@atsociety.org.uk**

Contact a Family have produced two new booklets: 'Guide to future benefit changes' and a new 'holidays, play and leisure' guide for children. To get hold of a copy, call 0808 808 3555 or go to: **www.cafamily.org.uk**

Top tips

This new feature aims to encourage our readers to share some of those practical solutions to every-day problems that they have worked out or discovered by chance.

Drooling

Many children with A-T have a problem with drooling. Hyoscine patches can help; they stick to the skin and also help prevent sickness- particularly in the mornings. There can be some side effects, but your Doctor can prescribe the correct dosage.

Sports wristbands are really useful for controlling drooling; children can then wipe their own mouths, without feeling embarrassed. There are many different designs available at sports outlets

Circulation

Some people with A-T can suffer with cold feet and bottom of legs. One of our readers has found MyCoal footwarmers very useful. They can be placed inside splints also. She purchased them from a company on line **www.notonlybowls.com**. They cost about £1.00 each and are single use only.

Uri-Bag

A reader tells us: "We have used this more now as our son has become wheelchair dependent. Sometimes our kids have urgency to go to the toilet and finding wheelchair accessible toilets is not always possible, even at friends houses where they don't have a downstairs loo. This is a great portable urinal available in male and female designs, just rinse out and ready to re-use another time. It has a lid so no spillage. You can get these on prescription from your GP."

Tip for Warts

Use duct tape keep it covered if it comes off just replace, great pain-free way of removing warts.

Family Fund

Depending on your circumstances, (you will need to check qualifying criteria), you can get help from the family fund, they can help with various items for our kids. You can request washing machines, fridge-freezers, bedding, toys, clothing, tumble dryers, holidays and even I-Pads. Contact the family fund for further information. **www.familyfund.org.uk**

New Life Charity

This charity can provide any equipment at home that is not provided by any statutory provider. They have provided one boy with A-T with a spare wheelchair seating system at home. You can get advice about wheelchair seating systems from wheelchair services or O.T's, You could also ask your spinal consultants. **www.newlifecharity.co.uk**

Straws

We have found some great really thin full length straws from Macros. They are great for children and adults controlling the flow of liquid to help with swallowing. They are made by Robinson Young and known as standard straws 210mm x 3mm Product Code No.0807.

Technology

Just wanted to let other people know about play station 3 Home. Would like to know if anyone else uses this or would like to use it as a way of keeping in touch with people with A-T and their siblings. Charlie Stubbs is a user and has great fun meeting his friends and hanging out together. If you want to be friends with Charlie his username is victorqpr, send him a message saying who you are and he would love to keep in touch with his friends. (Contact the A-T office for more information)

Our son uses an iPad, rather than a laptop or computer. He says it is easier to use and he also finds the keyboard easy compared to a normal computer keyboard, as everything is on one screen. He also uses and I Phone and say's it is great because it has voice recognition, and although doesn't work perfectly it is easier than trying to text manually.

Everybody has their own way of dealing with the difficulties A-T can bring – and your tips might be just what someone else needs. So why not write to Kay at the Society's office with your top tips or email **info@atsociety.org.uk.**

Please note that these tips come direct from our readers and the A-T Society takes no responsibility for their safety or effectiveness. Before trying any medical treatment or equipment, you are strongly recommended to discuss it with your doctor or appropriate therapist.

How to protect your family and assets

Liz Smithers, Partner at our Charity of the Year partners Clarke Willmott LLP, gives advice on protecting your family and your assets.

What is actually involved in making a Will? You may be aware that you should be making a Will but the thought of the paperwork, legal process and costs may well be preventing you from taking the first steps.

Talking to a Solicitor who has experience in dealing with people's Wills should make the whole process a lot easier to deal with – and very often it is not as difficult as anticipated – but where do you start?

How can I find the right Solicitor?

Do you have friends or relatives who have recently made Wills? If so would they recommend the Solicitor who helped them?

Society of Trust and Estate Practitioners (STEP): This is the professional organisation for practitioners in the area of Wills, trusts and tax. If a Solicitor is a member of STEP this indicates that they specialise in these areas. The STEP website has a search facility so that you can find a Solicitor in your area who specialises in drafting Wills.

What are the first steps?

Once you have some details of Solicitors, give them a call. Most Solicitors are happy to have a quick chat on the phone to find out what your circumstances are and to give a brief idea of what sort of Will you might want to consider and an indication of likely costs. You will have an opportunity to decide if you feel that the Solicitor is helpful and that the costs are reasonable before you are committed to using them. Do be aware that there is no one cost for a Will. The costs can vary considerably depending upon what type of Will is required. If you wish to go ahead, the Solicitor will either suggest a meeting or will write to you with further details of their service.

What Information do I need to provide?

The more information you can bring to a meeting or provide your Solicitor with the better. The more your Solicitor knows about you, your assets and your proposed beneficiaries the better he or she will be able to assist. The following details will be helpful:

- Nature and estimated value of your assets
- Full names and addresses of everyone you wish to make gifts to in your Will (your beneficiaries)
- Full names and addresses of the people you would like to appoint in the Will to act as your Executors (people responsible for dealing with your estate after death)
- Any details of your beneficiaries that may affect how a gift should be made, such as whether they are in receipt of means tested benefits or would be unable to deal with an inheritance themselves because of age or disability.

What is the process?

- Your Solicitor will advise you on the best type of Will to achieve your aims, bearing in mind your assets and the nature of your beneficiaries.

- You may need advice on tax mitigation.
- Your Solicitor will produce a draft will for you to approve. This should come with an explanation of what each clause of the Will means.
- Once you are happy with the document it will be finalised and you will either be invited to make another appointment to sign or it will be sent to you to sign at home. It is vital that a Will is signed in the correct way before the correct number of witnesses to be valid.
- Once you have a signed and valid Will it must be kept in a safe place. Many Solicitors offer safe custody services to their clients at little or no charge.
- Finally, once you have made your Will and signed it do remember to keep it up to date. Your family circumstances will change and the law in relation to Wills and tax may change and a regular review with your Solicitor is recommended.

A place in the sun

Tips for accessible holidays to brighten up the British summer!

Normandy, France

We have just returned from a week's holiday in a wheelchair-friendly gite in Normandy. The setting was the ideal 'away from it all' location, with local empty beaches only a short drive away. Accommodation was ideal, with friendly British hosts Alan and Liz, and even UK TV. Well worth a visit.

To view the properties please visit **www.bonhom.com**. 'Owl Barn' is the wheelchair-friendly property. Just remember, if you go at Easter France closes for a couple of days!

Gary Bromwich (Joe's Dad)

Owl Barn is wheelchair-accessible, with a ramp leading in and lots of space inside

Pictures, from top to bottom: One of the fully accessible apartments on offer; chairs for hire; a glass-bottom boat with wheelchair access

Cyprus

Avid traveller Ian had a great holiday out in Cyprus and stayed at a wonderful fully adapted apartment. They also have an adapted mini bus for transfers and full equipment hire, such as powered chairs, bathroom equipment, etc.

For more information go to:

- (for apartments) **www.ca-touristapts.com/Apartment.htm**
- (for equipment hire) **www.paraquip.com.cy**

Ian McInnes (Trustee)

Dale's Diary

June 2012: My week's holiday at Sandpipers in Southport began on Saturday 16 June when my auntie and uncle drove myself and my brother to Sandpipers. On the day of arrival, we sat about chatting and making friends, and played a few games. On the Sunday we had a nice stroll into town. We wandered along the pier but it was very blustery. The next day was a trip to the Lowry Centre and the Imperial War Museum, which was very interesting. Back at Sandpipers, the evening entertainment was a couple of ballroom dancers called 'Havana Nights'. Not my scene really so I made my exit and went to bed!

On Tuesday, we went to Chorley market, where we had lunch and explored the town. The entertainment that night was a singer called Paul Rodgers, who was very good. I stayed in the bar and had a few drinks. The following day's trip was to Port Sunlight, near Ellesmere Port. We had lunch in the grounds of the building, then went to the art gallery to look at paintings and sculptures of the local artist, William Hesketh. Back at Sandpipers, the evening entertainment was another singer, who was excellent. On Thursday, after a day staying in as the weather wasn't very good, I went out for a walk to the smallest pub in Britain! I had a brilliant holiday and can't wait to go back.

www.vitalise.org.uk *– Vitalise is a national charity providing short breaks [such as Sandpipers] for people with physical disabilities and carers.*

My Life

The 'My Life' project, carried out by York University, looks at the views and experiences of young adults with A-T. **Bryony Beresford** presented her initial findings at the Family Weekend.

One particularly interesting item at our Family Weekend in June, was the first report back on the findings of the My Life project. Carried out by the University of York Social Policy Unit, My Life is a study of the views and experiences of young adults living with A-T and their families.

The project leader, Bryony Beresford, was very keen to take the opportunity to come and talk to families about the project. However, at that time, the interviews had only just been completed and they hadn't had the chance to completely analyse the data. She made plain that she was just giving an initial and very general overview.

The first thing she said was that her researchers believed this was "one of the best projects they had every worked on" and the people had been a real pleasure to talk to. The focus of the discussions, Bryony said, had been the young people's lives, the things they did, their routines and what they looked forward to. They were also keen to talk about the people who were important to them and the equipment and services that helped them to live their lives.

It was clear also that the young people had aspirations. These included working, whether paid or as volunteers, accessing further education and training, living independently and widening their social circles. They showed a keen interest in knowing what their peers were doing and in meeting up with people with similar interests and similar conditions.

However they also felt that there were a lot of barriers to their achieving their ambitions. These included problems with equipment, transport and services, rules about benefits, money, employer's attitudes and so on. They also felt that it could be difficult for them to have good information about what their options were.

One positive fact was that they generally had a good opinion of the A-T Society and the support that it provided to them and to their families. They would be keen, though, if the Society was able to do more to help them with the specific issues they face and also to have more input into how the Society works,

William Davis welcomed the report and said that he was delighted at what he had heard so far. "It sounds as if there is a lot of excellent feedback to come. This will give us really useful information to work on. I look forward to working with these young people over the months ahead, to help them achieve their ambitions and support them in living their lives as they wish to."

The full report will be published towards the end of September. Once it is, the Society plans to hold consultation meetings with adults with A-T to discuss how to take these matters forward. We aim to have a more detailed report available for the December magazine.

Bryony presenting her project at the Family Day in June

Olympic values

At Abingdon school they are working on the Olympic values. The value last week was Inspiration and the teachers had to each nominate one child from the school who they thought demonstrated this. There were five children in total who got the wristband and Cleo's name was pulled out three times! She is really proud of herself for receiving it and hasn't taken it off since.

We're really proud of you too Cleo!

Tom graduates in style

Hats (and mortar-boards) off to Tom Hodson-Cottingam who graduated from the University of Birmingham in June, after four years of study, with a BSc in Animal Sciences. This is an extremely impressive achievement by Tom, successfully completing a 'hard-core' science course in university recently ranked as one of the top 10 in the UK,

Tom, who has a 'milder' form of A-T, took an extra year to complete the course. Just before his graduation he came along to the clinical research conference, where his in-depth knowledge of biology was extremely useful. We hope to feature more on his experience at university in our next edition but, in the meantime, our warmest congratulations to Tom!

Tom was diagnosed with A-T at age 2 when he developed a T-cell lymphoma. He was living in Nottingham at the time and the team at Queens Medical Centre did a brilliant job in both diagnosing A-T and ensuring his cancer was successfully treated without adverse side effects.

Tom was always very interested in animals and wildlife and developed a huge knowledge of the different species and their behaviours even when very small. So it was not surprising that he chose to study Animal Biology at university.

After getting his 'A' levels, Tom took a gap year and went to stay with his godfather, Richard Gatti, for the summer, where he worked in the A-T Research lab before heading off to Birmingham.

Tom is a clever, independently minded and generous young man. In his five years at university he has led a full student life with friends who share similar interests. He has been an active member of the University Gaming Society, of which he was President, overseeing the merger of two societies and organising regular events and activities. This final academic year was celebrated with a Society Ball in May and his Graduation on July 5.

Tom plans to stay in Birmingham because he enjoys the variety and stimulus of life in the UK's 2nd largest City.

Breathe easier in Brighton

After a "brief" pause, while lead investigator Dr Emma Ross was on maternity leave, the research into using breathing training in young people with A-T is underway. The project aims to improve the strength of breathing muscles, and reduce the effort of breathing in people with Ataxia-Telangiectasia.

People with A-T have problems with weakness and uncoordinated control of muscles, and this also includes the muscles which allow us to breathe. A-T patients often find breathing more of an effort and their cough can become weak. This can lead to problems in clearing mucus effectively which in turn can cause repeated lung infections.

The research study is looking at the effect of breathing through a handheld respiratory muscle training device for 15 minutes, twice a day, for 12 weeks.

The researchers are currently recruiting participants for this study, so if you would be interested in getting involved, please contact Emma via email **e.z.ross@brighton.ac.uk.**

They are looking for people with A-T who are aged between 14 and 30 years and feel they would be able to commit to doing the training (about 30 minutes) on a daily basis for 18 weeks. The training device is portable, so you can take it wherever you like!

During the study you will be visited at home to have some lung function testing, and also to teach you how to use the breathing trainer. The project's research assistant will then stay in constant contact through the training period, and will return to your home to do more testing halfway through, and at the end of the training.

The project is jointly funded by the A-T Society and the A-T Children's Project.

Scientists inspired in Cambridge

"A wonderful conference. This will increase the pace of our progress and bring our efforts to fruition"; "really riveting"; "great science"

The words of this participant at the A-T Clinical Research Conference 2012 held in Cambridge in June reflect the enthusiasm expressed by nearly all of those taking part.

Organised by the A-T Society, the conference brought together 100 delegates from across the world, including nearly all the leading specialist A-T researchers, together with clinicians and researchers from related fields.

As well as covering the various clinical areas of A-T, the programme focused on particular approaches that offer promise of significant progress, such as stem cells, gene therapy, MRI imaging and clinical trials. Experts from beyond the immediate field of A-T research were invited to provide different perspectives.

The structure aimed to promote discussion and to encourage concrete outcomes to be agreed. Each session concluded with a generous time for group discussion, the outcome of which was fed back to a final session on conclusions and future directions.

This worked very well. The points of agreement and proposals for future research activity gathered in the final session should help shape research in the years to come. There was also a distinct sense of renewed enthusiasm and of shared purpose amongst delegates. A-T is such a rare condition and its experts are so thinly spread that they can feel quite isolated. Coming together and working collaboratively to map the way ahead was a very positive and motivational activity.

Professor Robin Franklin offers his views in the discussion on stem-cells

Discussing the potential of neuro-imaging A-T

Topics covered

Opening the conference, Yossi Shiloh addressed the question of how the loss of ATM leads to the specific symptoms of A-T. He stressed the complexity of the picture, given the very wide range of process that ATM is involved in, and also the important role ATM plays in dealing with oxidative stress. These would be major themes for the conference.

Neuro-imaging

In a very positive session on neuro-imaging, Nora Volkow presented some initial data from her imaging work at the Brookhaven National Laboratory in the US. Other speakers highlighted the enormous potential of new techniques to help reveal what happens in the brain of a person with A-T. Following on from the session they agreed to set up a special interest group on neuro-imaging in A-T and are already designing a potential research project.

Neurology

The neurology session, stressed the complexity of the neurological picture in A-T. While cerebellar ataxia is a very important element, there are many other features, which are associated with other parts of the brain. For example dystonia, is common, particularly with some mutations, It was suggested that a special interest group be set up to study this, as it might teach us a lot about the effects of A-T on neurons.

Lungs and immunology

The sessions on the lungs and immunology both identified priority areas for research. However they also both stressed the importance of setting up an infrastructure for sharing data from different centres and also for establishing international guidelines on best practice in the treatment of A-T. This was repeatedly underlined throughout the conference.

One very interesting presentation from Gil Sokol, pulmonary physiotherapist at the A-T clinic in Israel, showed how he used exercises and a device called a Cough Assist to help children with A-T to relearn to cough, a vital element in maintaining lung health. This emphasised the capacity for supportive treatment of A-T patients to improve quality of life and probably life expectancy.

Stem cells

Stem cells were another important key issue. There are up to four different projects around the world aiming to create stem cells from ordinary cells taken from people with A-T. These cells, known as induced pluripotent stem cells, have a number of uses. They could be used to screen drugs, to find those potentially effective in treating A-T. Alternatively, it should be possible to turn these stem cells into the particular cell types that are most affected in A-T, such as Purkinje cells or granular neurons. Understanding the characteristics of these patient-specific cells could help better understand what is abnormal about their processes, hopefully leading to treatments.

Discussion with the panel on respiratory problems at the conference

Cancer

The cancer session highlighted that while there has been progress in the treating of cancers –mainly lymphoma and leukaemia – in A-T, there needs to be more sharing of information and internationally-agreed protocols need to be developed. Jan Loeffen from the Netherlands will be developing a registry to do this. It was also agreed that other countries should be urged to adopt the increased screening for female 'carriers' of the ATM gene now agreed by the NHS in England.

Clinical trials

The final session, on clinical trials, heard about two steroid trials, one completed and one planned. It also heard about Richard Gatti's progress in developing small-molecule read across drugs. Bringing a new drug to market needs the huge amount of money only a drug company can provide and while he is hopeful of establishing such a partnership, it is likely to be years before any successfully trialled drug could be on the market.

Karl Herrup's also spoke of his work studying the processes by which the lack of ATM causes particular cells in the brain to die. He has identified a number of different chemical interactions which make it difficult for cells to read and transcribe DNA. Using drugs that inhibit these processes has had some effect in improving performance in ATM-deficient mice, suggesting that this may be a pathway for future therapies.

Conclusions and future directions

There are too many ideas and conclusions from the different sessions to be able to list them all here but among the key and more general conclusions are:

- Better communication and more cooperation between all those engaged in A-T research is essential
- There is a great value to conferences like this for sharing information and ideas.
- An international register of people with A-T is essential
- There is a need to agree clinical guidance documents and get information out to non-expert physicians
- Supportive treatment of A-T patients can improve quality of life and probably life expectancy
- Better diagnosis, particularly of atypical or asymptomatic cases is needed and may give insights into the nature of A-T
- We need to raise awareness of A-T as a potential diagnosis for a wide range of neurological symptoms

A new Scientific Advisory Board for the A-T Society, chaired by Penny Jeggo, who chaired the final session, will meet shortly to discuss how we make the most of the outcomes from the conference to push forward the search for effective treatments.

We will also work closely with the A-T Children's Project and other organisations to use the A-T Clinical Network to improve communication and co-operation, as outlined above.

And finally, the conference and the announcement of the new A-T research fund, featured on page 5 seem to have elicited a burst of activity. As we went to print, we had been informed of a dozen potential new research projects. This is very exciting – and just what we hoped to achieve. Keep an eye on our website for more news as it happens.

Dedicated to A-T research

Meet Luciana Chessa, a professor of medical genetics from Italy, whose goal is to find a cure for A-T

Luciana Chessa could be considered the 'grande dame' – or perhaps we should say 'grande signora' of A-T research. Since she first became interested in A-T in the mid 1980s she has built up a remarkably detailed database of A-T patients and families in Italy, been involved in work to isolate the A-T gene, and is now very active in organising clinical trials.

Last year she led a trial of dexamethasone to treat the neurological symptoms of A-T, using a ground-breaking new technique to almost entirely eliminate side effects. And she is currently working to put together an international partnership for a larger clinical trial of steroids, which she believed can have a significant impact on the neurological problems A-T brings.

Luciana, who is director of the clinical genetics lab at Rome's Sant' Andrea Hospital, came to the Society's offices to meet me. She was in the UK for the Clinical Research Conference, despite having her right arm in plaster, from a recent bad break.

"That's dedication" I said, but Luciana has no time for praise. "I'm not sure what it is", she retorts. "I fell over in the street on Friday evening. It hurt, but I just got on with the weekend. It was only when I drove into work at the hospital on Monday that my colleagues persuaded me to have it looked at. When the orthopaedic surgeon got his hands on it, I suspect you could hear me in Naples..! However, there was no way I was going to miss the conference."

And how had she found it. "Extremely interesting and very fruitful for my work. I met a lot of excellent scientists and by working together I feel we established new insights."

I asked Luciana what had drawn her to working with A-T. "It's an emotional thing. Once you are in contact with these children it just gets you. They are so clever and sensitive. It feels as if it is they who are worried about their families, not vice-versa.

"Having to work with a progressively degenerative disease is incredibly frustrating. But while I've seen so many diseases, there is something special in these children – you could say I've fallen in love with them... The most important thing is to find a cure. This is the goal of my life."

Luciana's initial training was as a laboratory geneticist, rather than a doctor. She was working in a laboratory looking at a range of 'conditions like A-T where breaks in DNA, can't easily be repaired, leading to chromosomes becoming damaged.

In the course of this work, Luciana met a few patients with A-T, and this really got her interested. She created a repository of lymphoblastoid cell-lines which can be kept alive as long as you need them, unlike the blood samples or fibroblasts they had been using before.

A presentation of her work in the UK led to her spending six months at the Centre for Genome Stability in Brighton. While there, the director of the centre, Brian Bridges, suggested that she set up a register of people with A-T.

This proved no simple task as, although Italy does have a national health service, it has a strong regional focus and communication between different centres is not very good.

Undaunted, Luciana questioned colleagues and by travelling to and fro across Italy, got the clinical data and blood to establish a sample bank. Today the bank contains samples from around 150 A-T patients of whom 70 are alive, and also other individuals with related conditions.

The quantity of samples she gathered from patients and their families was for many years unrivalled. It was a very helpful resource in the search for the ATM gene and she worked closely with laboratories of Yossi Shiloh, Richard Gatti and Malcolm Taylor who were seeking to isolate it. And of course in 1995 the gene was found.

This discovery enabled her to start doing genetic analysis to identify the specific mutations of people with A-T and also allowed her to analyse samples from relatives of people with A-T and work out who was a carrier of the gene. Her register now has over 800 cell-lines from different family members.

She was already offering genetic counselling and pre-natal genetic diagnosis but, realising that with the knowledge she now had she could do much more to help people, she went back to university and in 2001 qualified as a doctor.

Since then, she has combined the different areas of her work; diagnosis, analysis and also medical care. And her focus now is on clinical trials with cortico-steroids.

"What we have found is not a cure, but in my view cortico-steroids are an important treatment option. It's a symptomatic therapy but it's the only one we have. I am confident that it will be found to have a positive effect – if I weren't, I wouldn't be working with it.

But while she is very encouraged by the possibilities, there are still problems to overcome. "The next step, in my view, is to try to find the optimal dose for each patient, to enable them to be treated without too many problems. I am confident that we can find a way to treat even very young patients, and slow the disease progression as early as possible.

What would help take her work forwards? "Well, we are completely dependent on the collaboration of families. We need their trust and confidence, even when the results are not initially astonishing. However, they shouldn't have unrealistic expectations."

The other thing, of course is: "Money, money, money! It's worse in some countries than others. The ideas are there, and there are a lot of excellent scientists working on A-T but they don't have the money, labs and opportunities. It is getting more and more difficult with the economy going so badly.

We need to recognise and finance good ideas – even if these do not always offer big prospects for a financial return. A variety of approaches is essential. We don't know where the answer is going to come from. It can seem as if we are spending money on pointless things but they then turn out to be really useful. Most people work in good faith.

I ask her what she is most proud of in her distinguished career. She pauses: "I am proud of what we have achieved, given the conditions that we are working in Italy- underfunding and general disorganisation." I ask if she would like me to use another milder word. "No. Disorganisation. That's the right word.

"Having to work with a progressively degenerative disease is incredibly frustrating. But, while I've seen so many diseases, there is something special in these children [with A-T]"

"I am happy to be able to go on with this work, even though I am working almost totally alone. Going round Italy to find families, data and blood; setting up our register and sample bank; assisting the cloning of the gene; these are all very important. And being able to create a small unit for diagnosis, and the fact that we are in a position to carry out trials on steroids. These are all achievements."

I ask about research interests outside A-T. She hesitates. "Mainly breast-cancer, its diagnosis and demographics. And I am also interested in other A-T-like diseases such as ATLD and AOA1 and AOA2." However, I am not convinced. She comes across as a woman with a single focus and driving passion.

So what does Luciana do to switch off and relax? Books and travel are her two outside interests. "I need to read to relax", she says. "It doesn't matter if it is a book, an article, a magazine, I just have to have written words to read.

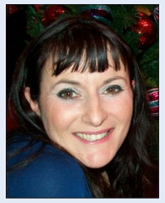
She is currently reading The Great Game by Peter Hopkirk, about the 19th Century struggle for control of central Asia. Why this? The answer is her other passion, for travel. "I have just come back from Uzbekistan. It was fascinating – the people, buildings, landscape and of course the food..." Like every good Italian, she has views on the food "The meat was excellent – it reminded me of how it used to taste when I was a girl – and they have some fantastic soups."

And what about the future? What does she think are the most exciting lines of research? "I am sure that small read-through molecules, like those used by Richard Gatti, are a very positive approach for people with nonsense mutations."

Otherwise, there are two areas that are particularly exciting. "Firstly, stem-cells mean that it is possible to create neuronal cells and other brain tissue and to try to use this to test substances and develop treatments. Secondly, we need to be looking at the use of small molecules that can pass through the blood-brain barrier and work directly on neuro-degeneration."

And finally, if she had a message for people with A-T, what would it be? "To be confident and trust in our efforts. As I've said before we have good people and we have ideas. With the money and support, we can make real progress."

Fundraising



Hello!

A-T HQ has seen many interesting events in the first half of the year, and I hope this newsletter will give you a flavour of some of the fun and excitement shared by our fundraisers around the country.

We have reduced the fundraising pages for this edition to free up space for the family weekend and international conference information and photographs. Nevertheless, we've packed in as much information as we can, and we hope you enjoy reading about our wonderful fundraisers and their dedicated support for the A-T Society.

We never underestimate the time and effort which goes into raising money for us, from a ball to a coffee morning and everything in between. We hear about the hours spent training for sponsored events and share the huge sense of achievement when someone exceeds their personal best.

So in this Olympic year, we wish you happy fundraising for the rest of 2012 – and remember the team is here to support and encourage you every step of the way.

Suzanne

Media opportunities

To keep the level of support we offer to families and to fund more medical research we have to keep increasing the money we raise. We really appreciate the many families who work hard to help with fundraising and stay in touch with the charity to give us new ideas.

Even if you are not in a position to raise money for the charity, if you are willing, you can help us in other ways.

It's really helpful when we are able to place strong stories in the media, whether TV, radio or local or national press, to spread the word about A-T and the work of the A-T Society.

The best stories are ones which show the impact on families, the strength of character we see in every child and adult with A-T and stories which clutch at the heartstrings. As well as engaging people, they raise awareness of Ataxia-Telangiectasia, the importance of family support and research towards a cure.

If you were willing to share your experience or a story in this way, that would be a real gift to the Society. To start this process, we ask you to complete a media information form. This contains your contact details and gives a space to tell your story. In this way, if we have an enquiry, after a quick confirmation phone call with you, we are able to pass on information immediately. You can download a form from the website, or contact Suzanne at the A-T Society office.

Ian McInnes did just that and within a few days was interviewed by a journalist for a very positive article in the Edinburgh Evening News. You can find the whole article on the paper's website – but here's one quote we loved from Ian "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 I have done since I was born and I don't intend to stop now!"

WorldPay

Welcome!

Thanks to A-T mum Jo Betts' friend Lexy Morgan and our longstanding IT supremo Alan Staples, the A-T Society was selected as Charity of the Year by the Cambridge office of Worldpay. Worldpay provide online payment systems for internet shops and the staff threw themselves into supporting us.

Worldpay have teams taking part in both the British 10K London Run and the London to Cambridge cycle race. Employee and author Graham Jones is donating a percentage of each sale of his first novel "From the Painter's Heart" to the charity and they also regularly hold fundraising events at their offices.

You might have met some of their volunteers at the Family Weekend and we were provided with a credit card machine to use at the A-T Spring Ball. We wish Worldpay a happy and enjoyable year with us.

Sophie and the Olympic Torch

By Simon Lynn

On Sunday 3 June 14-year-old Sophie Lynn had the honour of carrying the Olympic Torch in Glenariff, Northern Ireland. This was the culmination of a long process starting with her nomination almost a year ago.

Sophie was nominated because she has a great 'can do' spirit despite the limitations of having A-T. She has suggested and helped organise many fundraising events over the years, raising money for the A-T Society at every chance. We felt that Sophie was a good candidate to carry the Olympic Torch and hoped the Nominations Committee would recognise her. Just before Christmas we found out that Sophie was going to be one of the lucky 8000 torchbearers but we were sworn to secrecy until 19 March this year.

When we could finally let the cat out of the bag her school had a special assembly to announce the news. It went round our local community like wildfire with many people calling to congratulate her and wish her luck.

The week before the Torch came she received her uniform and instructions of what to do on the day. Her little sister Zoe was nominated to push her in the relay so she had to get a white outfit too.

On the day itself both the girls were really excited and couldn't wait to get left off at their collection point, to be picked up by the Olympic convoy. When we got there the Olympic Torch Relay Team could not have been nicer and Sophie got special attention because she was the youngest torchbearer that day. Sophie and Zoe got on the Olympic bus with the rest of the torchbearers for that day while we rushed off to await Sophie at Glenariff.

Sophie said the atmosphere on the bus was brilliant with torchbearers laughing and joking until it was their turn to carry the torch. Eventually it was Sophie's turn and when she got off the coach all her friends and family were there to give her a cheer. Sophie's godfather flew in from Indonesia (a 14 hour flight) especially to see her and I know Sophie and the family will never forget this. Other guests came from London and Edinburgh as well as places closer to home. While she waited for the incoming torchbearer to light her flame she was the centre of attention with media and onlookers taking photos of her.

The flame soon arrived, Sophie got her flame lit and Zoe and her set off down the road at what seemed like 100mph. The 300 metres that she carried the flame was lined with well-wishers and she got cheered all the way from start to finish. At the end she passed on the flame and got to keep her torch. She said it was one of the best moments of her life but it was over so quickly.

All of us would like to thank the Olympic Torch team for the way that we were treated and looked after on our special day. Thanks as well to all our friends and family for supporting and encouraging the girls as they carried the flame.

Action for A-T Shared event

Emma Anderson, a Trustee of Action for A-T, and supporter of the A-T Society writes; "On May 19th we held a Family Fun day in Haslemere in aid of Action for A-T and The A-T Society. This is a cause the community have really taken to heart since Evie Read's diagnosis and many people wanted to help and show their support.

"The Entertainer came and supported us with a stall (all money to the charities) as well as other local businesses. We had a bouncy slide and castle, face painting, beat the goalie, coconut shy, kid's races, cream teas, BBQ and many more things going on. The Mayor of Haslemere drew the raffle and there was even a visit from Snow White. We had a very successful Silent Auction with some amazing prizes donated by local people. The event was continuously busy all day with an overwhelming turn out. It truly was a community event that was brilliant fun and raised over £5000 as well as achieving awareness of A-T."

The Fearsome Four West Highland Way Challenge

Let me introduce myself! My name is Horse. I am a singer song-writer. As a creative person I tend to wear my heart on my sleeve. My first awareness of A-T, I am sad to admit, was being approached by Suzanne Roynon to contribute a track for a video piece for the A-T Society to be shown at an A-T telethon in America. As the track she wanted was owned by EMI and we wanted to avoid potential copyright infringement issues, myself and guitarist Gordon sang/recorded the track live just after a show in London's Bush Hall for the charity to use

I had absolutely no knowledge of A-T and was shocked at how it goes unnoticed by the general populace. With all of this in mind, when my partner AJ, good friends KB and Gemma and I thought we would do some of the West Highland Way across Scotland, we thought we should try to raise some funds for a charity. The choice of charity was easy for me,The A-T Society.

Each of us is very busy with work and we didn't have more than 3 days here and there for walking so we decided on four stages to fit in with our schedules;

Horse and her team after breakfast

Horse takes note of the signs!

Drymen to Balmaha

Armed with waterproofs and packs on our backs, off we set on our first stage, buoyed with the spirit of adventure and our initial excitement. Lots of talking and lots of snack bars later (and a couple of wrong turns) off we set with a real spring in our collective steps for around 4 miles. Then we came upon Conic Hill and decided to go over rather than around it. From the foot of the hill the path gradually but steeply slices through the hill, and it was with a lot less talking and a much more irregular pace that we eventually made the top.

The view over Loch Lomond was tremendous. It had been some time since I had 'done' any hills and I found it really exhilarating to be elevated and amongst the clouds again.

Crianlarich – Tyndrum

Several weeks later and a few more practice walks between us, the real walking began.

Fortunately, having spoken to several people who had done this stage, we were a little more prepared than if we had trusted our book's commentary that this was to be a fairly easy walk and fairly flat – wrong! Uphill, downhill zig-zagging, and seemingly endless It felt much longer than the suggested 6 miles.

The midge issue first presented itself on a brief stop over a burn whilst applying Jungle Formula – I was engulfed! I resembled Pig-pen from the Charlie Brown cartoon with a cloud of bugs around me. Panic ensued and we all ran away covering up as we went. Please note my language at this point has been moderated – whilst a midge feeding frenzy is taking place one's politeness can slip somewhat!

Tyndrum to Bridge of Orchy

After a night's rest and fuelled by a sumptuous Scottish breakfast we set off at a rate of knots. However, as we passed the Green Welly Shop, we saw a fairly large contingent of buses, police and 'locals' We asked a policewoman what was happening. It transpired,the Olympic torch was going to be passing through. Having missed it in Glasgow by doing the walk, we were thrilled and vowed to keep an eye out for it. As we were travelling along the route 2-3 miles outside Tyndrum we got our wish and saw the Olympic entourage wending their way up north! The sun began to shine on us and we stopped for a break

Whether we were tired from the walk or fit to bursting with our lunch – we all had a little snooze in the sun. This day was exceptionally pleasant – apart from a few rising gradients. The path was fairly undulating and of course the sunshine was delightful.

Further on we met McGowan the Highland coo (I named him after the toffee). He seemed very curious about us. We watched as he made his way over the river. AJ began walking backwards – I was wearing a red vest! We moved a little more quickly onwards after that – I had to catch up with them as they had left me behind in their haste!

That night we indulged in a wee glass of the local ale – Blonde Bitter and Twisted – which slipped down rather nicely thank you very much.

Bridge of Orchy to Inveroran then on to Kingshouse

We stepped out at a jaunty pace – all of us far fitter in the few days in which we had spent on our adventure. Everybody was delighted at the increase in stamina as this was the stage we were worried about.

For our last day we put two stages together, Bridge of Orchy to Inveroran – a wee stepping stone of about 3.5 miles leading on to what is dreaded by loads of people – Rannoch Moor – about 13 miles of moor which on a dreich day can be hell on earth with absolutely no shelter whatsoever!

From the hotel is a roadway which then becomes a path nudging you towards the moor. There is a further path with forest either side which then disappears leaving you facing the moor. To this point we had been expecting rain but it seemed to just hang over us. We had a moment in which we had a skirmish with our auld enemy – Scotus Midgimus. I was trying to flap them out of my anorak as quickly as they were swarming in.

So on we trod, all in a line, one after the other. It was both tedious and solitary work. All joking aside, this was a real challenge. Several guides warn that if you reach the halfway mark at Ba Bridge and the weather comes in you should turn back. I was anxious but determined, as was everyone else.

The wee break at Ba Bridge with a few energy bars certainly perked us up but the bunches of wilted flowers lying in memory of someone just to the side of where we rested, made us feel very thoughtful indeed.

The sky descended over us and its dark clouds hovered ominously. We moved on wrapped up tightly. The rain came down like stair rods and we hunched our shoulders forward. Then eventually, seemingly as if by magic, we began to see traffic a mile or so to our right, This could only mean one thing, that we were getting closer to the A82 and the end of our trip.

The weather never let up after this. We trudged on until we hit the main road and eventually we reached the end of the stage at the Kingshouse Hotel.

We enjoyed ourselves so much, we all want to do the remainder of the West Highland Way whenever we can find the time again. So far we have raised £1,127 for The A-T Society.

'McGowan' the highland coo!

Mega-Tri inspired by Lola

Like every other parent, when Lola's Mum Jo was told that her beautiful daughter has A-T the world stopped. After digesting the news, Jo bravely decided to tell all her Facebook friends what had happened and asked them to help give Lola a future by supporting the A-T Society. She was deluged with offers of support, including a Ball and a Skydive which you can read about in a later newsletter. However, for this edition we are concentrating on Kevin McGregor's Mega-Tri plans for April 2013.

Kevin lives and works in Singapore and has friends and contacts all over the globe. He has formed a team who plan to raise £100K for the A-T Society over the next year. The Mega-Tri will start in Plymouth with a 4km swim, then the team will cycle the 220 miles to Brighton in time to run the Brighton marathon on Sunday 14 April. A music gig and auction are provisionally planned for the Sunday evening.

If you are a strong cyclist and would like to join all or part of the cycle section, please contact Suzanne to be put in touch with Kevin. You can keep track of the event on Facebook at 'Mega-Tri for Lola and A-T'.

The South Downs Way Cycle Path

Matt Wright, a friend of an A-T family in Hampshire, gathered a team to take on the South Downs Way. He writes: We started the ride from Winchester on Saturday 7am, 103 miles, approximately 3500 metres of climbing (GPS died before the end!), 7000 calories burnt, 15 litres of fluid, copious energy gels, Haribo sweets, jam sarnies, a malt loaf and 16 hours later we reached a dark and blustery Eastbourne, to be met by our other halves and very patient children.

The team on their bikes with the stunning South Downs backdrop

At times it was very physically and very mentally tough, but the majority of the time it was a real blast and a great "box" to be ticked, especially with the A-T Society in mind. On the ride we only suffered 3 punctures between 5 of us, so the puncture god was looking down on us. The weather gods were also looking down on us through the day as it was near perfect conditions for riding. I now know what it's like to get to the peak of a hill wishing for some friendly lights the other side to find it's nothing but more hills stretching out in front of you!

A massive thank you from the bottom of my heart to everyone who supported me and to Gracie who gave me the inspiration to the do the ride to raise money for the Charity. On to the next challenge... Any ideas welcomed!

Tables are already being reserved for the 2013 A-T Spring Ball. Get your tickets now!

A-T Spring Ball

The first A-T Spring Ball was a sparkling evening of entertainment for more than 300 people who had travelled from all over the UK to be there. Chester Racecourse provided the ideal venue for the event organized by Vaughn and Joanne Rawson and Tania and Andy Wheeler.

Guests were entertained by Britain's Got Talent stars Signature and Les Gibson together with local band Universal Exports who filled the dancefloor for each of their sets. A charity auction included sporting lots from premiership football clubs and the worlds of boxing and formula one and framed posters signed by stars of stage and screen.

The Ball was so popular that the venue was immediately re-booked for 27 April 2013 for next year's event.

The 2013 A-T Spring Ball will feature the "Mugenkyo Taiko Drummers", a top performing group bringing drumming to an exhilarating new level, and compère/comic John Martin will entertain throughout the night and run the auction. The band, Universal Exports, make a welcome return to lure even the most unwilling dancers onto the floor!

Tables of 10 are already being reserved, so put the A-T Spring Ball on your list of 'must do' events for 2013, contact **Suzanne@Atsociety.org.uk** and your details will be passed on to the organisers.

Troopers for A-T Bridgnorth Walk

Team leader Tim Hughes writes: The 22 mile Bridgnorth Walk was held on 4th June 2012 and the weather was good for walking – no rain, plenty of clouds, a steady breeze and relatively low temperatures for the time of the year.

After a steady start from Bridgnorth the summit soon came into view and eventually on reaching the top we could just see into Wales.

The way down was as tough as ever and tested ageing knees but we all survived – the younger lads achieving some good times.

12 months gives the more seasoned of us just about enough time for the blisters to heal and for the joints to recover for next year!

We are hoping to raise more than £2,500 with gift aid, either via the sponsor forms or online, by the time all the sponsor monies have been collected.

Solicitor firm picks the A-T Society

Bristol-based solicitors Clarke Willmott LLP have adopted The A-T Society as their charity for the next two years. We share this honour with their local children's charity Jessie May.

Clarke Willmott aim to maximise their corporate social responsibility (CSR) benefits to their charities and have offered us skills and personnel.

This couldn't have come at a better time for us, and Clarke Willmott staff have been very involved in the planning and preparation of the conference, supporting William with printing and practical advice; Robert Smeath, a partner specialising in Will planning gave free consultations at the family weekend; and advice from another partner, Liz Smithers, features on page 12). Clarke Willmott also arranged the design of a special promotional flyer for the Marathon des Sables.

The flagship Bristol 10K was one of a variety of fundraising events that are planned for the next two years and we look forward to working with both Clarke Willmott and Jessie May throughout 2012 and 2013.

Joy's Super-story

I like to do what I can to support the A-T Society because they have given us so much help since our children were diagnosed.

I found out that our local branch of Sainsbury is very keen to be involved in the community. I wrote to the store and asked for permission to sell A-T goodies and do a collection. So far, they have given me two dates and as a result more people around my hometown now know about A-T, raising awareness as well as valuable funds.

To my surprise I was invited to the Sainsbury's charity night, and was presented with a further cheque for £50! It's good fun going out and chatting to new people and I know I'm helping a charity very close to my heart. I'd recommend it to anyone!

Joy Ferguson

Joy Ferguson at Sainsbury

Les Gazelles Triathlon

The A-T Society had an all French fundraising team supporting us in the Cotswold 113 Half Iron Man event.

Marie and Celine joined Perrine Hue, a friend of Stef Sprawling, to take part. The ladies each selected a charity for their support and organised a cocktail party to add to their total.

They created an informative website about the event which you can find at: **www.lesgazellestriathlon113.com**

Team Walope Great North Swim

Nick Walsh, whose niece has A-T, challenged girlfriend Siobhan to join him in chilly Lake Windermere for the Great North Swim. Nick said “Siobhan and I managed to complete the swim on Sunday after the Friday and Saturday events were called off due to the appalling weather. Unfortunately Siobhan took a bit of a beating from another swimmer who tried swimming through her, but she still completed the swim in a very good time.”

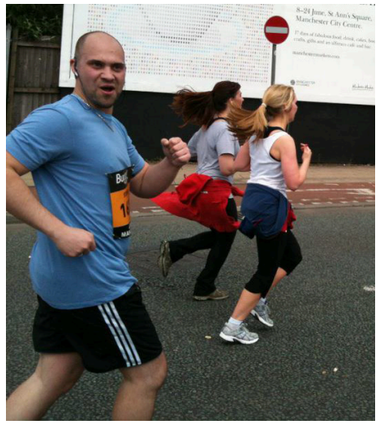
Congratulations to Nick and Siobhan on their new webbed feet!

‘Out of your Mind’

Wool Village Hall was the venue for a night of magic and entertainment staring Graham Jolley and Herbie Adams. The event was organized by Reg Foy, with Jen and Bob Owens doing sterling work behind the bar!

Miss Surrey

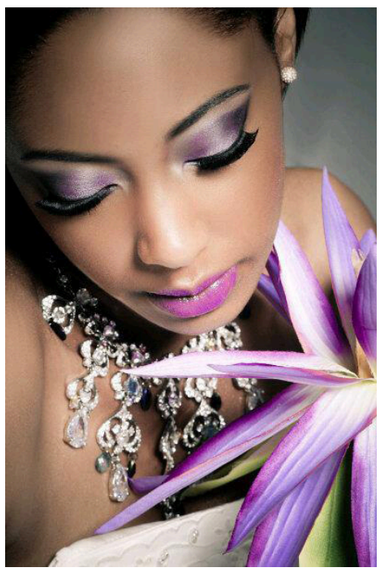
Stunning Shenise Bennett (right), Miss Surrey 2012 has chosen to support the A-T Society as one of her two charities in the run up to the Miss Great Britain competition. She has already completed a sponsored cycle and has more plans for fundraising and networking for the year ahead.



Steve Ogden – Great Manchester Run

It’s wonderful when friends of friends get involve with fundraising – it shows your passion can enthuse others to go out and do something incredible. Steve Ogden decided to take part in the Great Manchester Run after sponsoring his friend Nicky Edge last year.

Steve did a great time and will be supporting us again in the future!



Brownies love wristbands!

The 1st Bensfieldside (St Cuthbert’s) Brownies hosted a cake sale and raffle. While they were at it, they all invested in an A-T wristband and celebrated the Olympics with a visit from a local torchbearer.

Tough Mudder

Paul Gordon and his team mates crossed boggy hill and soggy swamp in The Tough Mudder, a 10K mud race in Scotland.

The Marathon des Sables

Those who attended the banquet at this year’s family weekend were treated to a short and very humbling talk by Sergeant Trevor Sanderson, a great friend of the Sprawling family. Trevor found his time serving in Iraq and Afghanistan made him revaluate his life and as a result he has made a commitment to spend the next few years raising money for the A-T Society with a range of events and challenges.

This is a man who doesn’t do things by halves! The MdS involves running 5 marathons in 6 days across the Sahara desert carrying all your own equipment. Trevor hopes to get some corporate sponsorship from companies all around the country. You should find a leaflet with your newsletter explaining how you can encourage Trevor and perhaps get your company or friends involved in his marathon fundraising efforts.

Why we made Wills 'in trust' – by an A-T parent

When my husband and I first got married we made wills. It was a relatively important thing to do as we were young and didn't want any government to get their hands on our assets!

When our children came along, our wills were amended to include them as beneficiaries. As my father had died shortly before we changed them, it made me suddenly realise just how fragile life can be.

We also had the unenviable job of thinking about what would happen if we died whilst our children were still young and who would look after them. At the time no relatives were considered suitable for one reason or another, so we asked some dear friends if they would act as guardians and they kindly agreed to do so. Then a few years ago our lives were turned upside down with the news that our children had A-T which meant they may not be able to look after themselves independently or manage their own financial affairs when they grow up. At that point we had re-think everything.

First we asked our friends if they were still willing to carry out their duties as guardians, if the worst should happen. Fortunately they were prepared to take on this task. Next we had to rewrite our wills. This proved more complex as we had to find a solicitor qualified to carry out the type of will that was necessary and our original solicitor did not have the right qualifications. We had to consider who we could trust to manage our children's financial affairs. After much soul-searching we approached four people we felt we could rely on, and most importantly were not going to benefit financially once we had passed on.

I'm quite an organised person and felt for my peace of mind I should also name three other people so if any of the trustees were unable to carry out their duties, there would be someone else to carry out our wishes.

The reason for leaving things 'in Trust' is that if the children need a lot of care, our estate will not be used up in care fees because the Trust means the capital is kept secure.

At the moment, if someone has more than £25,000 in savings (without a 'Trust' the children would have this from the proceeds of selling our house) then you are expected to fund your care and it doesn't take long to drain the finances.

I also ensured that when we are no longer around, the people we love and care about get whatever is left and, of course, if they have died before us, we have named a charity to benefit. My husband and I did a mirror-image style will so if one of us passes away and the other remarries, the original wishes are still carried out!

At the time, it was quite difficult thinking about what can be a taboo subject, but once it was done, we could rest easier and carry on enjoying what will hopefully be a long and happy life, secure in the knowledge that we have done all we can to safeguard the future of our children.