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A-T Society News

The Ataxia-Telangiectasia Society

Ataxia-Telangiectasia is a rare, inherited, neurodegenerative disease which affects many parts of the body and causes severe disability.

The A-T Society was established in 1989 and is committed to helping, supporting and advising families affected by A-T. The Society aims to alleviate the distress and suffering that A-T causes by working to improve quality of life now and in the future. We do this through funding research, supporting families, working to improve clinical management, and raising awareness.

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Editor's Comments

Once again, many thanks to all contributors. The copy date for the next issue is 1 October 2008
Please send comments, ideas, articles and pictures to the newsletter editor:

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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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photographer: Peter Kowalczewski

News from the Society

The Nottingham A-T Clinic team led by Dr Mohnish Suri has applied to the National Commissioning Group for Highly Specialised Services (Department of Health) for funds to develop the clinic. The process began last autumn with the submission of a preliminary application which was successful and the full application was submitted in May. It has been a huge undertaking and I would like to thank all the clinic team and in particular Dr Suri, Professor Taylor and Vincent Poupard.

The Society sponsored several UK researchers and clinicians from both Nottingham and Papworth Clinics to attend the international A-T research workshop which was held in Japan in April. As you will read, there is no major research breakthrough to report but progress is nevertheless being made and moves are presently afoot to begin the groundwork of preparing for clinical trials on an international basis.

In March we held the very successful Xscape Weekend for young people with A-T in Milton Keynes. The skiing and the Airkix experience were the main thrills to be had along with the spectacular "Take That!" musical which is now in the West End. In May we held the AGM and Family Day in Nottingham. Two trustees resigned, our chairman David Owens and Gill Dodge. We thank them for all that they have done for the Society and are very grateful that they both will continue to support us. The Trustees will be seeking an external chair to replace David.

Jo Child has kindly taken on the daunting job of re-vamping the website for which we are very grateful and the task of updating our literature rolls on. We send our best wishes to the new "BrAsh A-T" Foundation in Australia (www.brashat.org.au) which has made a very impressive start and wish it great success in raising funds for A-T research and for family support.

It is with sadness that I report the death in March of Glynis Watkins. Glynis was a "leading light" of the Society in its early days. She and her husband Jeff started the Society's newsletter and it was their idea to establish a multi-disciplinary specialist clinic for A-T in order to advance clinical management. Glynis was also a wonderful support to families. She was always available by phone and her warmth and reassurance helped very many people. We are very grateful for all she did and send our condolences to her family.

Maureen Poupard
Hon Secretary

International A-T Workshop 2008

A team of scientists, researchers and clinicians from the UK, some of them sponsored by the A-T Society, travelled to Japan in April to take part in an international workshop on A-T.

Maureen Poupard was also there, representing the Society.

Maureen writes:

The International A-T Workshop took place from 22 - 26 April in Kyoto, Japan.

These scientific meetings are held approximately every two years and bring together many researchers who are involved with A-T research to share information, forge collaboration and drive the research effort forward.

Talks began at 8am and finished at 10 pm. A typical day contained 23 speakers - making it all rather intense! As well as all the talks, further information about A-T research work that had been carried out was also disseminated via a poster presentation. 127 posters were exhibited during the course of the workshop, and were left on display at the back of the hall for people to peruse. A couple of hours was also laid aside for their authors to be in attendance so that they could be quizzed about their work.

Over 260 people attended from Japan, the USA, Italy, Israel, Canada, Australia, Singapore, the UK, Poland, Germany, Spain, Switzerland, Korea, Netherlands, Denmark, Sweden, France, Norway and Hong Kong.

On the following pages, Professor Malcolm Taylor and Dr Andrew Exley give their accounts of the workshop.

Professor Malcolm Taylor:

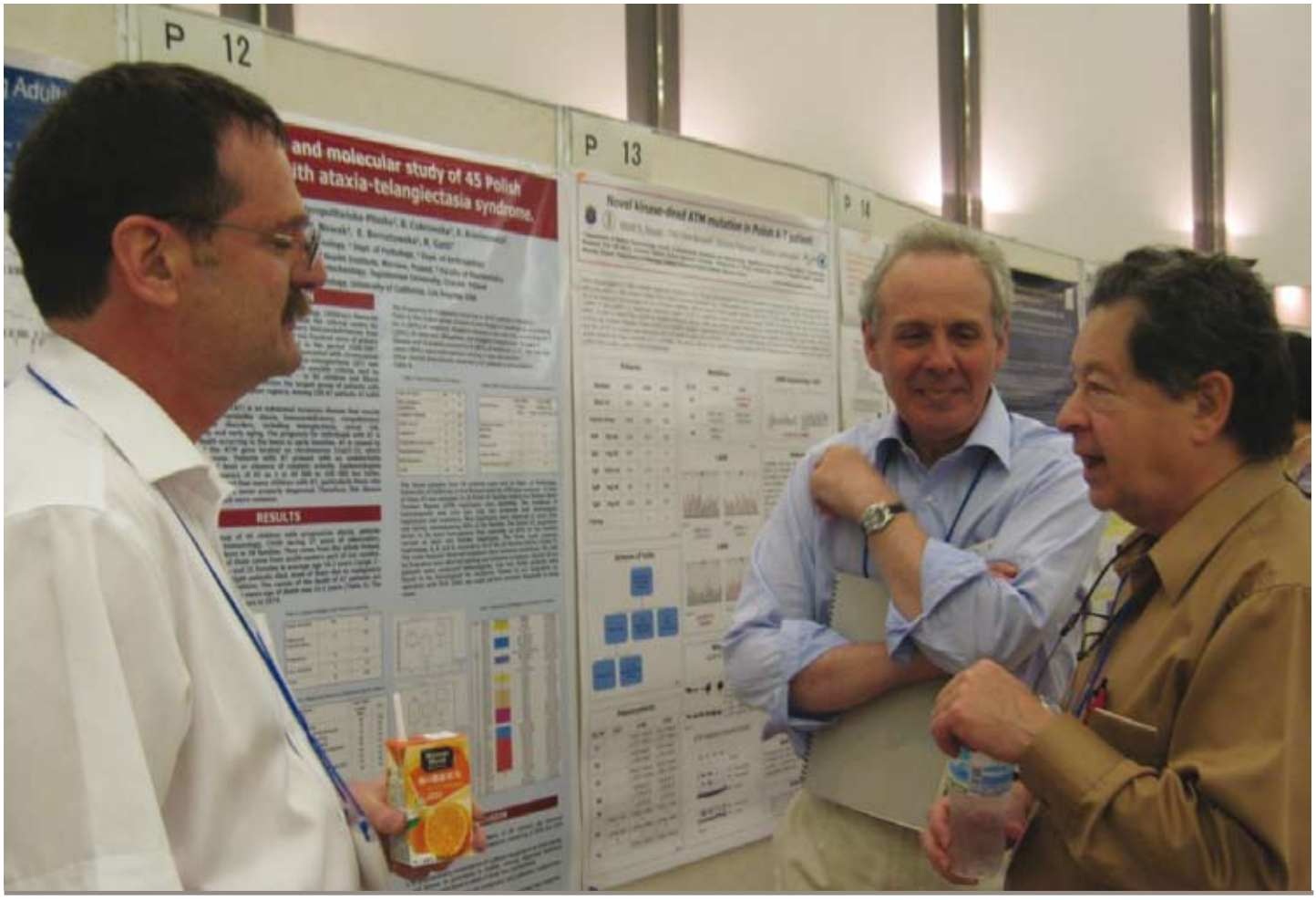
I'll start with some background on the workshop. The gene causing ataxia telangiectasia is the ATM gene and the ATM protein has very important roles in recognising damage to the genetic material of our cells and orchestrating the response to the damage.

This may range from allowing the time for the cell to repair itself and recover, to initiating a planned death of the particular cell to put it out of the way.

Of course the ATM protein doesn't act alone but in concert with many other proteins and this list is still growing. Damaged genetic material in a viable cell may constitute one of the steps to cancer development. This is how important the ATM gene is. The role of ATM in maintaining the integrity of our genetic material and in its absence the risk of cancer is what interests many scientists across the world.

The emphasis, therefore, is often on the ATM protein rather than on ataxia telangiectasia, the disorder. This may be disappointing for parents. Because A-T is a rare disorder, it will be difficult to get pharmaceutical companies to be interested in, say, screening drugs that may be useful in the treatment of A-T.

However, I believe that it is important for as many scientists as possible to be involved in research into ATM and its partner proteins and this was clearly demonstrated in Japan. The continued emphasis of the



Left to right:
Dr Andrew Exley,
Prof Malcolm Taylor and
Professor Richard Gatti
in discussion

importance of ATM in our cells will generate more work and greater understanding of the roles of ATM and possibly in the longer term what could replace ATM. There are other proteins in our cells with a similar, but not the same function.

There was some limited and speculative discussion at the workshop of trying to boost the activity of these other proteins to replace ATM. Whether this is possible and whether it could slow down the rate of neurological change is unknown.

It would be helpful to have a good animal model of ataxia telangiectasia, one that mimicked the neurological decline in A-T. There isn't such a model at present and, therefore, we cannot test drugs very satisfactorily, that might alter the neurology.

Another approach is to look carefully at drugs in safe current use to determine whether any of these might be of benefit to people with A-T and the possible use of trials was discussed a little at the recent A-T Family day.

I am sure that these ideas will be further developed.

Malcolm Taylor
CR-UK Institute for Cancer Studies,
University of Birmingham

Our team in Japan. Left to right: Dr Kelly Townsend, Dr Mohnish Suri, Dr Nick Davies, Dr Grant Stewart, Prof Malcolm Taylor, Dr Andrew Exley

Dr Andrew Exley writes:

Travelling to Kyoto took 24 hours each way and it was hard work attending the very long scientific sessions - but it was an interesting meeting and great for team building, networking and brainstorming.

We presented our work at Papworth Hospital, Cambridge, on the clinical and laboratory assessment of respiratory disease in young adults with A-T. Standardised clinical assessment is combined with respiratory physiology, video fluoroscopy to assess swallowing, a CT chest scan to assess lung inflammation and airways damage, and specialised immunology. These measures were presented in a graphical, bar chart format to highlight differences between patients according to their genetics and opportunities for intervention.

We met a number of people from Europe and discussed outline proposals to establish a collaborative network. Away from the pressures of every day work, it was great to meet colleagues from Nottingham, Birmingham and even basic scientists from Cambridge! We held discussions about the application to the National Commissioning Group for funds to develop the Nottingham A-T Clinic and talked about the steps needed to establish clinical trials in A-T.

There are no genetic cures for A-T on the immediate horizon but a number of alternative strategies were identified in brainstorming sessions and informal discussions within the umbrella of the meeting. We discussed A Best Current Drug Evaluation Strategy (ABCDEs!) and identified several candidates suitable for preliminary, proof of principle studies. These first trials will require clinical assessments and blood tests or biomarkers sensitive to change so we can identify promising candidates/ approaches for further studies. Some of our established blood tests may be useful as biomarkers.

The Kyoto meeting included many presentations on the fundamental molecular biology of ATM, which [as Malcolm Taylor explains] is the gene responsible for A-T. These presentations prompted thoughts on strategies to boost ATM function or to bypass the block in ATM function. We also discussed the potential to set up new biomarkers based on the molecular biology of ATM.

The meeting provided many opportunities for learning and discussion. It's an exciting time for scientists and clinicians working with A-T!

Dr Andrew Exley
Consultant Immunologist
Papworth Hospital NHS Foundation Trust
Cambridge

"Many opportunities for learning and discussion"

Carrier Status

What does being an A-T carrier mean?

We all carry two copies of the ataxia telangiectasia gene (one on each of our two copies of chromosome number 11). One of these chromosomes with its A-T gene is inherited from mother and one from father. In the vast majority of individuals in the general population both copies of the A-T gene are entirely normal.

The A-T gene of course can be altered or mutated. Occasionally, a child inherits a mutated copy of the A-T gene from mother and also a mutated copy from father. In this circumstance the child will have ataxia telangiectasia.

The birth of a child with A-T does not happen very often and our estimate is that it happens about 5 or 6 times a year in the UK. The frequency of ataxia telangiectasia in the population in the UK is approximately 3 in every million of the population.

A-T is a rare disorder, but one where prenatal diagnosis is possible. If we can identify both A-T mutations in an affected child, then we can test any subsequent pregnancy, from the same partners, for A-T. This involves removing part of the placental material made by the fetus, extracting DNA and looking for the same mutations that are present in the child in the family who has A-T. For each pregnancy the likelihood of the baby having A-T is about 1 in 4.

In the same million people where 3 children with A-T are present, approximately 0.5% of the population, ie 5000 people, have one of their two copies of the A-T gene that is mutated. These are commonly known as A-T carriers. It is safe to say that these people are oblivious to this fact, and only rarely when a carrier has a child with A-T does this become apparent. The question of carrier status in the rest of the family can then arise.

"should my other children be tested for carrier status?"

The question might be asked, "Is it a good idea for my other children who do not have A-T to be tested for carrier status?" and "What is the chance of them having a child with A-T?" At the level of the individual family, where there is a child with A-T who has an unaffected brother and/or sister, the likelihood of that unaffected sibling being a carrier is 2/3. As long as that carrier individual does not find a partner within the extended family, the chance of them having a child with A-T will be very small (a likelihood of less than 1 in 600).

Just for comparison it is generally accepted that approximately 15% of known pregnancies result in miscarriage.

You might ask if it is possible to find out for sure whether you (with a sibling who has A-T) and your partner (who is not a member of the family) are both A-T carriers. The answer for you is yes, and for your partner is also yes…. in principle, BUT…….and the problem is this. We cannot guarantee to be able to find an A-T mutation, if it is present, in a member of the general public. This is for technical reasons. If we find nothing, it may just represent a failure to detect what is there.

But does it matter that we cannot test your partner to see if he/she is a carrier?

No, I believe that it does not matter. In these circumstances the risk is very small indeed. The appropriate course of action would be to see your GP and take advice from a clinical geneticist or genetic counsellor. By far the best path is to share your worries with a competent professional and hopefully this will allay any fears.

"are there any health concerns for carriers?"

Are there any health concerns for A-T carriers? The answer to this is that there are definitely some facts that female carriers should be aware of and precautions that are appropriate. The risk of breast cancer in female A-T carriers

is about double the risk in the general population. It may be higher in women carriers under the age of 50 years. This is a moderate increased risk.

It would be appropriate for you to discuss this with your GP so that in the event of a tumour this is detected and treated as early as possible.

Malcolm Taylor
CR-UK Institute for Cancer Studies,
University of Birmingham

The Society's information booklet about genetics has been updated. Now called 'The Genetic Aspects of A-T and Pre Natal Diagnosis', the booklet can be ordered from the office. Call Kay if you'd like a copy.

Rare Disease Day

To celebrate the First European Rare Disease Day on 29 February, the Genetic Interest Group organised a reception at the House of Commons which was hosted by Dr Evan Harris MP. We were delighted that the A-T Society was invited and we were very pleased to be joined by Erdal Cevik and his two sisters.

Representing the A-T Society were front: Erdal Cevik; back: from left to right, Maureen Poupard, Nuriye Cevik, Zubeyde Cevik, Angela Sherry and Lian Yarlett.

Photo by Rob Lowe

Our Xscape Weekend Joe Bromwich

Getting ready for rock-climbing

Joe

In January I was 16, this made me old enough to go on the A-T Society weekend away for older kids and adults. I had not been on one of these weekends before. I was looking forward to it, to meet the others who were going, who were not kids. I've got to that age were I no longer feel a kid. I thought it would be good to meet up with other young adults who had A-T, because they have the same problems as me. I go to a mainstream school in Aldridge, I had lots of friends at junior school but they seemed to have disappeared as we've got older, because they go off and do different things that I can't do.

Me and Dad agreed to go to the A-T weekend and were very excited when we found out it would be an Xscape weekend. This was to include bowling, airkix parachute simulator, theatre, rock climbing and skiing. This was all in one place at Milton Keynes.

We got to the hotel early and went for a meal before going to the theatre. I had a big bucket of mussels, which kept me going through the show. I think the show was for girls really, as I am not a big fan of 'Take That'. Kay seemed to enjoy it along with the others, and the special effects were really good, at one point when the second half started I nearly ended up in Kay's lap I jumped that high. We left the theatre hoping to go to the pub, but it rained so much we were all soaked by the time we got to the hotel, and went to bed.

The next day we got up and had breakfast, and went ten pin bowling. It was a bit cramped at the bowling but we moved around in a rota system. We had the afternoon to ourselves and went skiing in the evening. A disabled skiing club were at the Snowdome to help us. We got kitted out, everyone said it

was freezing, but I was ok. We took turns in coming down the slope of real snow. We went zig zagging down really fast and even jumped between the two runs. The helpers were brilliant. Everyone loved the skiing.

The next day we went to Airkix, I had worried about how I would breathe in the air tunnel. This was not a problem as I only went in for a millisecond before I wanted to come out. Everyone had two goes except me and Neil, we weren't big fans. (Get the rubbish joke). All the others had a great time and looked like they were flying. We all got DVDs of our flight. I called mine 'don't blink'. Later that day we went for a meal with everyone on the weekend, at Zizzi's restaurant. This was a good time to socialise with everyone. After a very late night I made sure I was the last one to bed!

We were up the next day to go rock-climbing which was really hard work but good fun. Catherine was a star at the climbing. I think we tested the staff, but I think we all got off the ground.

We all made our way home after the climbing. The whole weekend was just GOOD FUN. It was great to meet others with A-T and we chatted between all the activities. In fact because this was the first time I had been on such a weekend, the talking was as good as the activities. You must remember us people with A-T are a pretty unique bunch.

Many thanks to the A-T Society who made the whole weekend possible.

Maureen Poupard with Joe Bromwich
Nice hat, Maureen!!

Neil Wells - nice hair, Neil!!

Jane, ready for Airkix

Bowling

Jane Henderson writes:

I thoroughly enjoyed the weekend. It was a very busy time but good fun!

I enjoyed all the activities - bowling, rock climbing, skiing, and airkix - but my favourite was the skiing which I thought I would never do!

Ready for Airkix - simulated sky-diving

Left to right: Kay Atkins, Angela Sherry, Tracey Staples, Mike Grist, Maureen Poupard.

The Airkix experience at the Xscape weekend was made possible thanks to a generous donation from the Apollo Charitable Trust. Donna Strong, whose nephew Charlie has A-T, brought the Society to the attention of Apollo Group, where she works. Mike Grist, Apollo Group's Corporate Social Responsibility Director, and Tracey Staples, Marketing Administrator, presented a cheque for £1,000 to the A-T Society.

Family Day

23 families attended the family day in Nottingham in May.

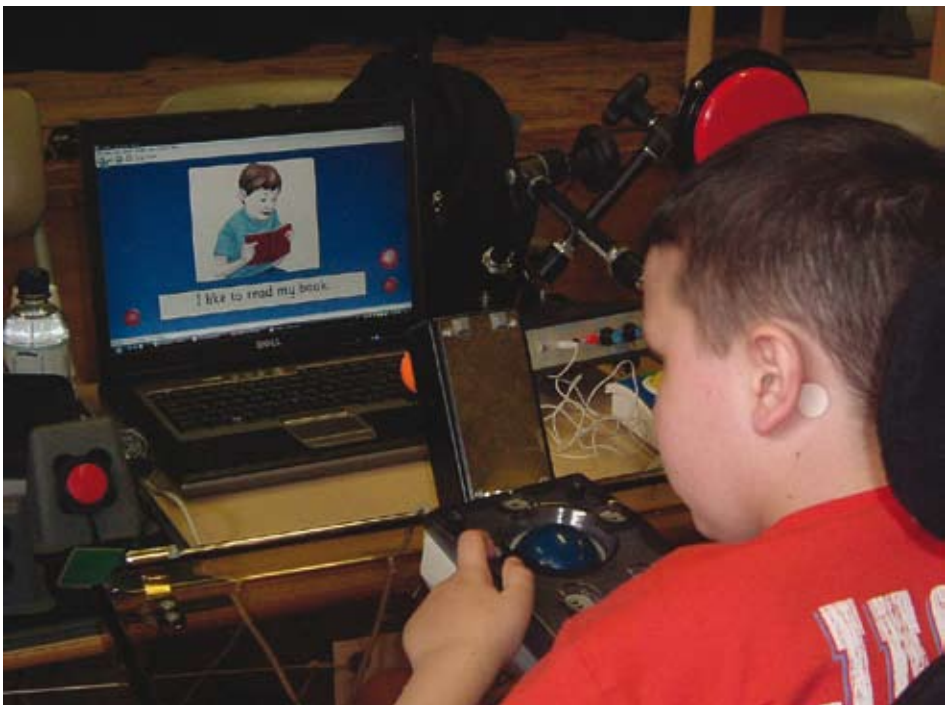
Our annual family days in Nottingham provide an opportunity for families to come together for mutual support, to exchange information with each other and to learn from guest speakers.

This year's family day on 10 May was attended by 23 families. Talks were given on a wide range of subjects, including scientific and clinical research, holidays, and the role of physiotherapy for people with A-T. Mike Ellison spoke about getting help with making adaptations to your home, and his talk is written up on pages 28 and 29 of this newsletter.

The Open Forum offered families a chance to put questions to a panel of medical experts - a short summary of this session is given on page 16.

As usual, children and young people had an alternative agenda while the talks were going on, which this year included morning entertainment and crafts and an afternoon outing for the younger children, as well as a range of therapies for older people with A-T.

To give families more time to socialise, there was an informal dinner in Nottingham the evening before. This is always a very popular occasion.



Young people had a chance to try out computer hardware and software demonstrated by the Aidis Trust. The Aidis Trust offer a free computer helpline for people with disabilities, their families and carers - 0845 120 3719. **www.aidis.org**

Alecia Yarlett helped on the bookstall.

Aimee Cooper enjoyed a massage. Reflexology was also on offer.

What people said about the day:

"It was very informative and interesting and the speakers made the hard bits quite easy to understand. My daughter had a fabulous time and already she's planning next year and who to bring with her! She particularly enjoyed all the craft activities.."

"We got so much from meeting the other families"

Nottingham Clinic

There are still some spaces in the next Nottingham Clinic, on 14 November. If you haven't been for a while, maybe now is the time to come back and see the specialist team. Please contact Kay Atkins on 01582 760733

Open Forum

The Open Forum at the Family Day offered families a chance to put questions to a panel of health experts.

Here are four questions and the answers that were given.

Is there anything we can do to prevent fingers going into a claw?

If you become aware that fingers are starting to curl you could try:
- Slow, frequent stretching;
- Massage;
- Night-time splinting – this has to be tailored to the individual.

Dystonia in A-T can be treated with various types of medication; oral medication is not usually very effective.

Are people with classic A-T (ie people who don't have any ATM protein) more susceptible to cancers?

There is no black and white answer to this question. The majority of children with leukaemias and lymphomas do have classic A-T – don't have any ATM protein. However there have been fewer than 50 over the last 20 years. You may wonder whether having a small amount of ATM protein would give you protection against cancer. We think that it may give some protection against childhood cancers but later in life it may increase risk of a tumour, including breast cancer.

Do all people with A-T get telangiectasia (red extra blood vessels)?

Most, but not all. About 90% of people. Telangiectasia are not always restricted to the eyes – it might be somewhere else, for example the back of the throat, or – very rarely – the bladder. More exposure to sunlight leads to more telangiectasia.

My child's little toes curl under the foot as the day wears on and he needs his shoes removed as they are painful. Is there anything that can be done?

If he suffers fatigue and discomfort you should consider doing something, and there are things that you can do. Shoes should have reasonable arch support and be comfortable, and there should be no pressure on the metatarsal heads. If there are inner soles, they should be soft and not cause any problems. Let him have bare feet sometimes, and consider using resting splints at night.

Fundraising

Kaid Betts met the Pantomime Cow, when Claire Guildford invited him backstage.

Staff at Kaid's brother's school have been helping to raise cash for the A-T Society. Teacher Claire Guildford and her friends took part in a sponsored run for A-T. Claire also played a part in a Panto and profits were generously donated to the Society.

Producer Gillian Williams presents a cheque to Kaid's mum, Jo Betts

Regular Giving

One of the best ways you can help us maintain our level of support is to give modestly and regularly. We receive no statutory funding whatsoever and therefore need a secure income in order for us to plan ahead.

Just £5 a month or a once-a-year commitment will enable us to make a bigger impact on improving the quality of life of children with A-T.

Race Night

A Race Night in aid of the A-T Society was held in Harpenden on Friday 18 April.

It was organised by Richard Cannon who runs "Thebestofharpenden" a local business which champions the best businesses in Harpenden through its own website: www.thebestof.co.uk/harpenden. The A-T Society's offices are in Harpenden.

Over 220 punters attended, raising a magnificent £1,150 for the Society - and the roof, as they roared their horses on. (With names such as Lamppost Collision and Dobbin, they needed all the help they could get!)

Photograph courtesy of the Herts Advertiser

The prize for the "best dressed filly" went to Louisa Hill, granddaughter of Alan and Maureen Hammond, who are active supporters of the Society. Louisa (pictured above receiving the prize of a cut and blow-dry voucher from organiser Rachel) arrived in her riding gear from cap to boots complete with whip and grooming kit! Congratulations Louisa, a well deserved win.

Our patron Joan Morecambe (on the right in the picture) received the cheque on behalf of the Society, from Richard and Rachel.

London to Paris

Phil Horne and Les Page took part in a mini Tour de France, representing Morrish and Partners where they both work, and raising a wonderful £1020 for the Society. The two cyclists, shown here with their colleagues, rode from Tower Bridge in London to the Eiffel Tower in Paris. Well done and a big thank you to both of them.

A Pampering Evening for Ladies was held in May at Redbourn Methodist Church in aid of the Society and the church. Participants were able to choose from 14 different therapies and treatments ranging from deep tissue massages to Pilates and eyebrow shaping. Whilst relaxing between sessions they were able to browse through the various craft stalls and enjoy hot chocolate with marshmallows or fruit punch making for a thoroughly chilled out evening! £191.34 was raised for the Society.

Thanks to Kay Atkins who organised this event.

Benefactors

Many thanks to all our benefactors in 2007:

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Mrs Linda Warren
The Mary Webb Trust
The James Weir Foundation
Mr R Wells
Mr W J Wheeler
Mrs HE White
Gladys Wightwick Trust
Franca Williamson
SA Windsor

Zeenat and Muhammad Afzal, whose daughter Aneesa had A-T, raised money for the Society in her memory, in a variety of ways. At the Family Day they spoke about how much they want to help other families affected by A-T, and they presented a cheque to Maureen Poupard.

Friends Email List

Kay Atkins, our Family Support Worker, is updating the 'Penpal' List. In keeping with modern technology, it will now be called the 'Friends Email List'. This list is for anyone aged 12 and over who has A-T. It's a great way to make new friends and to keep in touch.

If you wish to be included on the list, please contact Kay with the following details:

Name
Age
Where you live
Your email address
Brief list of your interests

The list is open to everyone in the UK and abroad, so wherever you live in whichever corner of the world, we would love to hear from you - by the end of July at the latest. Kay will send the list out to everyone by the end of the summer.

Contact Kay on 01582 760733 or email: atsociety@btconnect.com

Ataxia Awareness Day

Thursday 25 September is International Ataxia Awareness Day which is supported by organisations around the world including the A-T Society and Ataxia UK.

The aim of the day is to:

- tell more people about ataxia
- help more people understand the effects of ataxia
- help raise funds internationally for research into treatments

If you are thinking of running a fundraising event this year, how about doing it on or around 25 September?

Perhaps you could organise a coffee morning to raise funds for the A-T Society. And don't forget to take photos for A-T Society News!

A date for your diaries.....

This year, in conjunction with Ataxia UK we are hoping to have an International Ataxia Awareness Day fundraising event in London. We are planning a fun, family event to take place in Hyde Park on Sunday 28 September.

If you are interested and able to get into London, please keep this date free in your diaries.

More information to follow!

www.ataxiaawarenessday.org

Aphra joins Charlton Athletic as a Mascot

Aphra with Charlton player Jerome Thomas

Aphra's day as a mascot for Charlton Athletic football club was one she'll never forget.

A die-hard Addicks fan, Aphra joined the welcoming club for their game against Southampton. It was a dream come true as she made her way onto the pitch in front of a crowd of over 26,000.

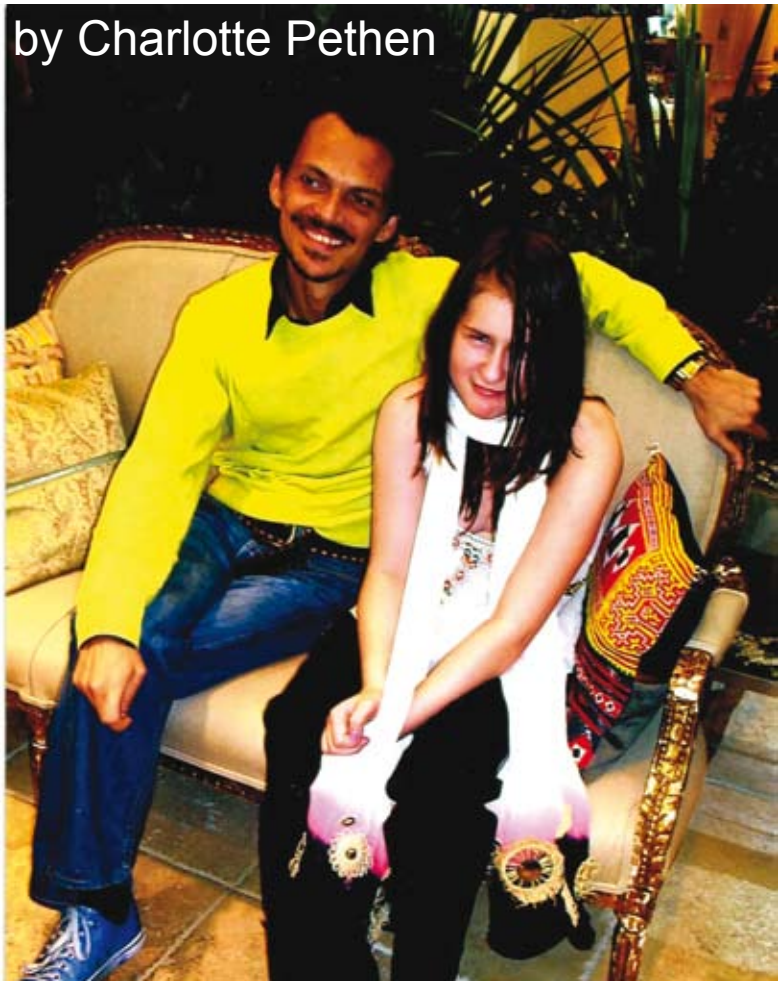
The day began with a tour round the boardroom and then Aphra got to meet the Charlton players and management as well as watching the away team arrive. A packet of Jaffa cakes from the manager Alan Pardew made Aphra's smile even wider and she also got to watch the players warm-up on the pitch before joining the team captains and mascots for a photo just before kick-off.

Unfortunately the game itself, like many of Charlton's games this season, did not go quite as planned and they ended up settling for a 1-1 draw.

However, Aphra went home happy with a large bag of souvenirs from the club, some lovely photos and the memories of a very special day.

My Wish

by Charlotte Pethen



How many girls get a designer outfit made especially for them? Charlotte did - thanks to Make-a-Wish Foundation UK

At last! The day I was waiting for - Tuesday 18th March - was here. Just after 9am the taxi collected me and Mum and Dad and took us to York Station to catch the train to London. It was difficult because we had two wheelchairs and a suitcase with us, but they had arranged assistance at York and Kings Cross. After a slight hiccup the taxi took us to the Radisson SAS Hotel.

When we were settled in we caught a taxi to the London Dungeon (very scary) then another taxi took us for a meal which was not so good for me, but the champagne was great. So was the hotel, luxury all the way.

As soon as we finished breakfast the next day, dad went to look for Graham, our limo driver.

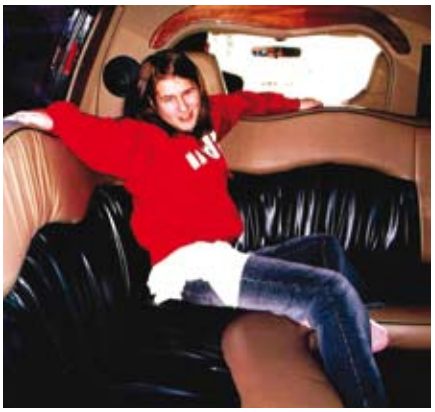
Warning - if using transport in London do remember your blue badge. WE DIDN'T! Graham - a perfect gent - was issued with a parking ticket even though he showed his paperwork which asked to collect two disabled ladies and their wheelchairs. I'm surprised that they only gave him one ticket because the car took three parking spots! Graham took us to Selfridges where I had a makeover and my nails done. Wow. Then upstairs to their sandwich bar where we were met by a lass from Make-a-Wish who accompanied us to the Bruton Street shop of Matthew Williamson who had a special outfit which had been designed and made just for me. Matthew was great and I would love to meet him again sometime.

The outfit was trousers and a chiffon top plus a long scarf all with beads over them to make it really special.

Then back to Graham and the 23 seater mini and off to the Hard Rock Cafe for tea. Then Graham then took us for a tour of the London sights before going to the Ambassador Theatre where we had a box to see the show Grease. It was so nice that the cast had signed a programme for me to keep. Then back to the hotel and bed.

The following morning I was shattered. We finally set off for Kings Cross and..HOME.
All in all a really super trip and many happy memories.

My thanks to all involved.



Holiday exchange anyone?

Vanessa writes from New Zealand

Last issue I wrote about my concerns for my daughter's future as an independent adult. Morgaina recently turned 20, and although she has been able to remain at a fairly local and predominantly mainstream college, she has really struggled with being in a small unit of students aged 13-21 with hugely varying degrees of disability.

One of the problems Morgaina encounters is that there are no other people in NZ of her age group that have A-T. In fact there are only two known out of 4 million people. Over the years I have tried to encourage her to e-mail with other young people with A-T but her desire to mix with kids not afflicted with a disability has been quite strong, especially as she finds the conflicting emotional needs of her fellow students with Asperger's and severe Cerebral Palsy quite tiresome.

However, I have maintained the wish for her to meet other people with A-T, of similar age. I would've loved for her to attend the over 16's week at Milton Keynes but the timing wasn't really good for us as in New Zealand the new school term starts mid-February.

We did a lot of groundwork at the end of last year finding courses that would keep her inspired for the last year of college and it would have been a shame to disrupt them.

I am toying with the idea of a holiday exchange program that would offer these young adults with A-T a chance for some companionship and an opportunity for an overseas experience. We live in a beautiful area in Auckland, surrounded by Native bush and obviously have a deep understanding of the needs of a young person with A-T. I am hoping to meet a family that would be able to support their child with A-T to come here to be looked after by us and vice-versa.

Morgaina spends a lot of time on MSN chat with various friends and I am hoping that she will now connect with some other young people with A-T through this medium.

She is interested in music, dance and writing poetry.

Morgaina writes:

Hi there, I would love to connect with others like myself. A holiday exchange program would be ideal, because not only would it offer me an overseas experience but it would be a chance to meet new people and create new relationships.

However, chatting online would be a good way to get to know one another, before committing to an exchange.

If you would like to connect with me my email address is: bewareofthewitch@hotmail.com

Dale's Holidays

Regular readers of A-T Society News will be familiar with Dale Phillips, who has written several accounts of his holidays in the UK and abroad. Dale has promised us a regular column in every issue. Here he describes a weekend break in Wales.

On Saturday 19 April my carer, Dalpat, drove in our van to Glan-y-Borth Holiday village in the lovely town of Lllanrwst. We arrived early so we decided to explore the delights of the town, somehow ending up in the pub until it was time to check in. We were staying in a bungalow which had been adapted for wheelchair users. After we had unpacked we had time to relax for a while before my friends Debbie and Dave and their children arrived from Criccieth which is about an hour's drive away. They stayed for a couple of hours and it was good to catch up with all their news.

Afterwards Dalpat and I got washed and freshened up and decided to hit the town and go on a pub crawl visiting three different pubs. About 10pm we headed for home where we watched match of the day and had a chicken curry and finally hit the sack.

On Sunday we decided to take a drive to Bangor (about an hour away), where we walked round the harbour and strolled along the pier, wandered into town and had a drink. That evening back in Llanrwst we had supper and went out on the town again for a few beers.

On Monday, our final day, we decided to drive to Rhyl. We drove along the promenade and had a meal, before starting our journey home. All in all it was a terrific weekend break, despite the weather being very cloudy and cold!

I would recommend Glan-y-Borth to any of you looking for a break in this lovely part of Wales. It's not far from Betwys y Coed and

Conway. It's handy to have a car for trips but there are several pubs in Llanrwst and a Spar supermarket, all within walking distance.

Glan y Borth, Llanrwst, North Wales
tel 01492 641543
www.glanyborth.co.uk

The Holiday Village is situated on the banks of the river Conwy on the edge of Snowdonia National Park. Amongst the 20 bungalows are six specially designed and created for disabled people. They are all on one level, with doorways 33 inches wide. Bathrooms have full access for wheelchairs into the shower, and all kitchen units are at a comfortable level. The landscaped gardens are ramped for ease of movement around the site. Prices range from £140 pw to £560 per week. Short breaks are available at any time, except school or bank holidays.

News of two students

Alexandra Wills

Alexandra Wills has recently graduated from the University of Wales, Newport, with a BA/BSc Hons in English and Psychology. Alexandra was diagnosed with A-T just over a year ago, after six long years of trying unsuccessfully to determine the cause of her problems.

She was 16 when her symptoms first started to manifest themselves, a crucial period for her during which she was taking her GCSEs. After gaining GCSEs she went on to pass three A-levels. During this time she was struggling with her walking and fine motor skills but the only help she wanted was someone to hold onto in order to get around the school.

At university, Alex lived independently in halls for the first two years, but did need an arm to hold onto around campus and someone to take notes for her. Her last year in university was spent partly at home and partly renovating a house that she and her fiancé were buying. Mrs Wills, Alexandra's mum, says: "Alex was very hands-on with the renovating, she would sit on the floor and paint to eye level, bag up the rubbish and rubble and project manage everything else!!

"During all the studying for GCSEs, A-levels, and degrees, there were numerous visits to hospitals to London, Cardiff, and Newport. In one year she was in the National Hospital for Neurology in London four times.

"Her courage and determination is very humbling and we're very proud of her."

Tom Hodson, who is 20 and has A-T, has just taken his first year exams at Birmingham University where he is studying Biological Sciences. He hopes to specialise in animal science, possibly with a view to working with animals or something to do with conservation and the natural environment.

During his gap year in 2007, Tom visited Malcolm Taylor's lab for a week and went to Los Angeles where he worked in Richard Gatti's lab sequencing A-T gene mutations.

Since he's been at uni, where he shares a flat with 6 other boys, he's become quite a good cook. His specialities include spaghetti bolognaise but during the exam weeks he readily admits to living on sandwiches and ready meals!

His hobbies are gaming and role playing and he loves films, music and comedy. For the third year running he will go to the Edinburgh Fringe this year, where one of his friends from school will be performing. He's also a big Terry Pratchett fan and reads fantasy fiction very widely.

Tom, with his father Peter, setting off for Birmingham University

Disabled Facilities Grant

Mike Ellison explains how to get help meeting the costs of adapting your home.

A mandatory disabled facilities grant is designed to help meet the cost of adapting a property for the needs of a disabled person.

Who is eligible for a grant?

To be eligible for a disabled facilities grant, the applicant must be:

- an owner occupier; or
- a private tenant; or
- a landlord with a disabled tenant; or
- a local authority tenant; or
- a housing association tenant.

Some occupiers of caravans and houseboats are also eligible.

The person benefiting from the adaptations i.e. the disabled person must come within the following definition of disability:

- sight, hearing or speech is substantially impaired; or
- has a mental disorder or impairment of any kind; or
- are physically substantially disabled by illness, injury, impairment present since birth, or otherwise; or
- are registered (or could be registered) disabled with the Social care services department.

What can you get a grant for?

A grant can be awarded for:

- facilitating a disabled occupant's access to and from the dwelling;
- making the dwelling safe for the disabled occupant and others residing with them;
- facilitating a disabled occupant's access to a room used or usable as the principal family room;
- facilitating a disabled occupant's access to or providing a room used or usable for sleeping in;
- facilitating a disabled occupant's access to or providing a room in which there is a lavatory, bath or shower, and wash-hand basin or facilitating the use of any of these;
- facilitating the preparation and cooking of food by the disabled occupant;
- improving the heating system to meet the disabled occupant's needs, or providing a suitable heating system;
- facilitating a disabled occupant's use of a source of power, light or heat;
- facilitating access and movement around the home to enable the disabled occupant to care for someone dependent on them, who also lives there.

The test of financial resources

Disabled facilities grants for adults are means tested. There is no means test where an application is made by the parent or guardian for the benefit of a disabled child or qualifying young person.

A qualifying young person is someone who is aged 16 but not yet 20 years old and is still in full-time non-advanced education i.e. A level or below.

The 'relevant person'

For applications from owner-occupiers and tenants for the benefit of an adult, a test of resources is applied to the person with disabilities and their partner, if they have one. This is so even if the disabled person is not the applicant for the grant.

For example, a disabled person lives with his brother, who has sole ownership of the property. The brother can apply for a disabled facilities grant to carry out adaptations to his property for the benefit of his brother who has a disability. The test of resources only applies to the brother who has a disability (known as the relevant person) not to the brother who made the application.

Subsequent grants

If a 'relevant person' has had to make a contribution to a previous grant on the same dwelling (in the last 10 years for owner occupiers or 5 years for tenants), the value of that contribution is deducted from the assessed contribution on a subsequent grant application. The works under the first grant must have been carried out to the local authority's satisfaction for this offsetting to apply. If the contribution on the earlier grant was more than the cost of the works, leading to a 'nil-grant approval', the value of the works properly carried out can be offset against a subsequent grant contribution.

Applying for a grant

Disabled facilities grants are administered by the local housing authority rather than the social services department where these are different authorities. An application form should be available from the local housing authority. An application must be supported by a certificate stating that the disabled occupant intends to live in the property for at least 5 years after the works are completed, or for a shorter period if there are health or other special reasons.

Approval of a grant

The maximum grant payable under a mandatory disabled facilities grant is £30,000 in England, Northern Ireland and Wales. Local authorities could provide further assistance for extra costs under their discretionary powers.

In order to approve an application for a disabled facilities grant, the local housing authority must be satisfied that the works are both necessary and appropriate for the needs of the disabled person, and reasonable and practicable in relation to the property. In determining whether the works are necessary and appropriate, the local housing authority must consult with the social services authority. This is why some local authorities will direct people to the social care services department first for an occupational therapy assessment.

However, it is important to make a formal application for a grant because the 6-month time limit for the local authority to make a decision only begins from the date of the formal application. They cannot refuse to allow you to make a formal application or refuse to give you an application form.

If you do not get a decision within 6 months of applying, write and ask why and request that a decision be made. Seek legal advice if you still do not get a decision, or if you have been prevented from applying in the first place. Alternatively, you can make a complaint of maladministration to the Local Government Ombudsman.

If approved, the adaptations should usually be completed within one year by one of the contractors who supplied an estimate for the application. It is important not to start works until the local authority has approved the application.

If, after the application for a disabled facilities grant has been approved, the disabled person's circumstances change in some way before the works are completed, the local housing authority has a discretion as to whether to proceed with paying for all, part or none of the works. The local housing authority must take into account all the circumstances of the situation before deciding how to proceed in such a situation.

From 22 May 2008 in England, local authorities may place a charge against a property where a disabled facilities grant exceeds £5,000. The maximum charge is £10,000 and would apply for 10 years. This means the value of the charge would be repayable if the adapted property is sold within 10 years. Local authorities should decide whether to place a charge on a property on a case-by-case basis considering the individual circumstances of each applicant.

Many thanks to Mike for speaking at our Family Day, and providing this information. If you are thinking of applying for a grant and you would like advice, contact Kay Atkins at the A-T Society office. She will be happy to help.

Organisations that offer support

A selection of organisations that provide support to individuals and or families

information compiled by Angela Sherry

Lifeline 4 kids
www.lifeline4kids.org
020 7794 1661
Provide specialised equipment for children with disabililties. Also help with welfare appeals for clothing and household items for the benefit of disabled children. No cash awards.

React
www.reactcharity.org
020 8940 2575
Provide grants for specialist equipment and everyday items like washing machines for families on low incomes. Immediate response.

BDF Newlife
www.bdfnewlife.co.uk
Tel: 01543 468888
Provide grants for equipment for children where a delay could worsen the child's condition.

Make a Wish Foundation
www.make-a-wish.org.uk
Tel: 01276 405060
Grant wishes for children aged between 3 and 17 with life threatening illnesses

Cerebra
www.cerebra.org.uk/grants
01267 244200
Provide equipment such as computers, wheelchairs, trikes, for children under 16 with a neurological condition.

Willow Foundation
01707 259777
Organise special days out for seriously ill adults between 16 and 40 years old

ACT
www.theactfoundation.co.uk
01753 753900
Will help individuals with funding for building and modifying homes, specialist equipment . Application available on website

Variety Club Children's Charity
www.varietyclub.org.uk
020 7428 8100
Support individuals – children needing mobility aids.

ELIFAR
www.elifarfoundation.org.uk
No phone number available.
Provide funding or part funding for specialist equipment, toys, adaptations to the home, holidays or therapies to children and young adults.

Starlight Children's Foundation
www.starlight.org.uk
020 7262 2881
Grant wishes for children with life threatening illnesses between 4 and 18 years old

Daytrippers
www.daytrippers.org.uk
020 7758 0030
Provide funding for trips/days out for children 3 – 21 years old with life limiting diseases.

In brief

Gastrostomies

The A-T Society has produced a new booklet called 'Gastrostomies and A-T' which is available from the office. Please call Kay if you would like a copy.

Genetics and prenatal diagnosis

The Society's information booklet about genetics has been updated. Now called 'The Genetic Aspects of A-T and Pre Natal Diagnosis', the booklet can be ordered from the office. Call Kay if you'd like a copy.

Self Help Directory

www.ukselfhelp.info is a directory of self help groups, with a useful section on disability. It lists many websites with subjects ranging from holidays to housing, boating and snowsports.

Really Useful Things

www.reallyusefulthings.com is a website that has...some really useful things! Have a look at the flextendable straws, the straw holders and the ingenious shower sandal.

Christmas Cards

Sadly it is no longer viable for us to continue to commission our own Christmas cards. Very many thanks to trustee Maureen Jenkins and Pamela Dickerson of Tudor Galleries, Norwich for all their hard work producing Christmas cards for the Society over many years. The trustees are investigating if there are any other ways we can have our own range of cards at an economic price and should there be anything available, will let you know later on in the year.

need a bath?

A family has an Appollo adapted bath that they would like to see go to a good home. This bath uses water pressure to lift the seat out of the water and is suitable for varying degrees of mobility.

It's a spacious bath with a water powered seat which lowers and raises the bather in and out of the water.

The seat lowers flush with the base of the bath, giving the bather maximum freedom of movement.

A conveniently placed lever on the side of the bath operates the lift system.

With the seat in its top position, manual rotation through 90 degrees aids the bather's entry into and exit from the bath.

The seat lifts up to 20 stones weight.

If you're interested, phone Kay at the A-T Office on 01582 760733

Recycle your old mobile phones and inkjet cartridges and raise money for the A-T Society.
For a full list of wanted inkjets visit the website – www.recycle4charity.co.uk
You could ask your local club, school, store or supermarket if they would be willing to have a recycling box somewhere on display which can be filled with old cartridges or mobile phones on your behalf. This will be collected free of charge from you as long as it contains at least 50 items.
Give the A-T office a call if you require any bags or boxes.

Easyfundraising

Don't forget you may be able to raise funds for the A-T Society when you shop online.

Easyfundraising is a shopping directory that features some well known online stores, including Amazon, NEXT, Debenhams, John Lewis, Toys'R'Us, HMV and over 400 others. All you have to do is use the links on the easyfundraising site whenever you shop online and, at no extra cost to you, we'll receive a free donation of up to 15% from every purchase you make. It really is simple!

It's completely FREE to register and use and you still shop with each retailer in exactly the same way. Many retailers even offer additional discounts and money saving e-Vouchers when you shop using easyfundraising.

If you shop online anyway then why not raise valuable extra funds for us by using this fantastic scheme? All you need to do is visit http://www.easyfundraising.org.uk/atsociety and when you register, select A-T Society as the organisation you wish to support.

Thank you for your support!

Are you using Everyclick?

Everyclick.com is a search engine that gives half of the revenue it generates to charity. If you are not already using it, please give it a try - it's a great way to give every day and it doesn't cost you a penny!

All you have to do is choose the A-T Society as the charity of your choice and make everyclick your home page . You can then use it whenever you search the web.
Please give it a go and pass the message on!

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atsociety@btconnect.com
tel 01582 760 733 fax 01582 760 162
Reg Charity No 1105528