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A-T Society News

In this Paralympic edition:

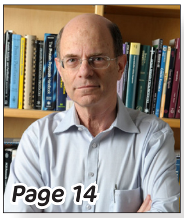
- Two new patrons enter the ring
- Yossi Shiloh looks to go all the way to the line
- Paralympic cyclist Roxy Burns is a star in London
- Neuroscientists in wheelchairs on the podium in Washington

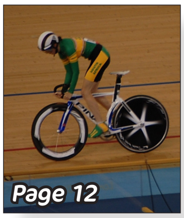
AT society
awareness research care

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.

Contents





Editor’s Comments

Many thanks to all contributors. The copy date for the next issue is 1st May 2013. Please send comments, ideas, articles and pictures to:

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The opinions expressed in *A-T Society News* are those of the individual authors and not necessarily those of the A-T Society.

Cover picture: Emily with TV presenter/Paralympic wheelchair basketball legend Ade Adepitan

View from the chair

Lian Yarlett, chairman of the A-T Society, looks back at a busy year for the Society and forward to 2013 – which promises to be another action-packed year

We are coming towards the end of 2012 already, and it really has been a busy year.

There have been many highlights and for me, as always, the Family Day is at the top of the list. But this year the Cambridge conference has to be there too. This was an incredibly valuable investment – both for the near future and the long term – and was extremely successful in encouraging researchers and clinicians to collaborate more and share knowledge to drive forward treatments and care. It led to interest from new key players, not previously involved directly with A-T. Some very important joint work has already commenced and some exciting developments are under discussion for 2013… We will keep you posted.

Of course, we just can't get everything into each edition of the newsletter – there is simply too much. If you're into computers then keep any eye on our website or join us on Facebook where we post up-to-date news and developments, or contact us direct at the office.

From its foundation, one of the Society's key missions has been to ensure that the care it provides is total – supporting the child and the family with care and advocacy, as well as promoting and funding research to develop our understanding of and treatments for A-T, and raising awareness. I am very proud that the original vision remains strong and that we are able to maintain the balance of providing practical care, which makes a real difference to our children today, with research that delivers improvements and, in the long term, will change the future.

One of the most important people in originally shaping this vision and later making it a reality is Vincent Poupard. Vincent, who has been a trustee and treasurer of the Society for many years, has now stepped down from the Board. On behalf of everyone, I would like to say a very big thank you for his personal strength and commitment for the benefit of others and to wish him well for the future.

2013 is already looking to be a very positive and interesting year. The Ataxia-Telangiectasia World Conference is being hosted here in the UK; there are a series of new research projects we are looking to fund and an exciting fundraising programme looks very promising (although we can always do more!). I hope to see you at our Family Day and until then I wish you all a Happy New Year.

Lian

Mugs, Bags and Wristbands

We are delighted to offer a limited edition A-T 'Keep Calm' mug. These are the perfect gift, ideal to make you smile with every cuppa! The mugs retail at £5 and you can purchase online from our website.

You can also pick up Jute Bags for £4 and our glow in the dark wristbands for just £1.

Please make use of the online shop – it's there for you!

Just William

William Davis considers the findings of the My Life report, which shows that there are very different views on the importance of research

I sat down to write this column, planning to talk about research. A lot has happened this year including the recent very interesting forum organised by the A-T Children's project. But as my computer warmed up, into my inbox popped the first draft of the report on the findings of the My Life project.

If you haven't heard of it – or don't remember – this was a series of interviews carried out by the University of York with young adults with A-T and their families.

The report makes fascinating reading and offers a lot to think about and work on in the months ahead. However, given what I was about to write, one thing particularly struck me: the ambivalent attitude of young people with A-T to research.

Some people interviewed were positive about research, recognising that it gave hope of a cure, if not for themselves, then in the future.

Bryony Beresford presented the My Life project at the family day in June

It is a challenge, particularly for small organisations like ours, to be effective in providing help in all the different ways people want.

However, others thought differently: "if you've got A-T there's no cure for it. So I don't like [research] at all... I'm not interested"; "I don't like being treated like a guinea-pig". While not in the report, another young person with A-T has said to me: "I don't support research. Without A-T, I wouldn't be me."

These comments may come as a surprise to some of the many people who consider that research should be one of the Society's biggest priorities. However, they underline the fact that there are a wide range of views about what our priorities should be. We have to understand and, as far as we can, respect these different opinions.

This is a normal issue for a charity dealing with a long-term medical condition that has no effective treatments. It is also very common that people's views change over time. Often, when someone is first diagnosed, they and their families see research as the prime consideration, hoping to see significant improvements in treatments before the condition worsens. However, over time, when progress perhaps hasn't been as rapid as they hoped, or their needs for support become greater, they may come to give more importance to other work around services, advocacy or practical and financial help.

It is a challenge, particularly for small organisations like ours, to be effective in providing help in all the different ways that people want. It is even harder in a condition like A-T which affects people from early childhood to advanced adulthood. If you try to be all things to all people, the danger is that you get lost.

The answer, I think, is to recognise the challenge, understand what it is that different people want and expect and plan as best you can to meet these varying needs.

I hope that the A-T Society is doing this. We are increasing our commitment to research and by the middle of next year we aim to have a portfolio of active research projects that we are funding. At the same time, we will do more to provide support to people in living with A-T, now – and an important part of this will be talking with adults with A-T about what we can do for them, based on the findings of My Life.

But it is equally important that you keep telling us what you want us to do. We may not be able to do it all, but we are here to do as much as we can.

Have a happy, healthy 2013.

Vincent Poupard retires

It is with a mixture of sadness and immense gratitude that we publish the news that Vincent has stepped down from the Board of Trustees. From its inauguration 23 years ago, Vincent has been the Society's treasurer and at other times its chair. But more than that, along with his wife Maureen, who retired last year, Vincent has been a rock on which the Society was built.

As treasurer, Vincent was a prudent and meticulous guardian of the Society's finances, ensuring the highest standards of accounting and accountability and at the same time cultivating and growing our funds to put us in the strong position we find ourselves in today.

Vincent's concern to ensure that things were done to the highest standards made itself felt across almost every aspect of the Society's work.

The high quality of the Society's services and information and the high reputation in which we are held by so many people, both within and beyond the Society, owe an enormous amount to his influence.

However, in addition to the skills and knowledge that he brought, Vincent also gave huge amounts of time and energy to the Society. This was not just in keeping the Society's books, but he would also get involved with or would contribute to almost every aspect of its work.

However, along with his serious side and his uncompromising commitment to making the Society as good as it could be, we will miss the other sides of Vincent: the wicked sense of humour which he gives rein to frequently when relaxed and more sparingly, sometimes unexpectedly, when working; the deep sense of compassion and concern for others, which he is shy of revealing directly, but which clearly underlay everything he did for the Society; and the breadth and depth of his knowledge and intellect.

Like Maureen, Vincent abhors any fuss, so we have severely limited here what we might have written.

Nevertheless, we are confident that we speak on behalf of the entire community of people that live with or work with A-T in thanking Vincent for the enormous contribution he has made and in wishing him great happiness and fulfilment in future.

ERYDEL make progress in America

Erydel is the company that makes the machines for infusing dexamethasone – a steroid similar to betamethasone – into the patient's own blood cells. We reported on the results of a phase II trial they had carried out in Italy in last December's newsletter.

This study showed significant improvements to the scores of around half the patients on the trial, but importantly there was a complete absence of the typical steroid side effects.

At the end of July, the company had their product – known as Erydex – recognised as an orphan drug by the Food and Drug Administration (FDA) in the United States. This recognition is likely to help and speed up the process of getting authorisation to use it for the treatment of A-T.

Once approval is granted, the company will be looking to carry out a large-scale efficacy trial, though they may be required to carry out some smaller phase 1 studies first.

If this trial does take place, there is a distinct possibility that patients in the UK will be recruited to take part. We will keep you updated on progress with this.

In the meantime, a paper published by Menotti et al in the *Journal of Biological Chemistry* suggests a reason for the effect of steroid treatments; it proposes that dexamethasone alters the process by which the DNA is copied from the ATM gene to create the ATM protein.

The authors suggest that lengths of the DNA are skipped and the result is a protein which is much shorter than it should be but still contains the all-important kinase-domain – the active part of the protein that 'switches on' other proteins.

Research Partnership

As was announced in the July edition, the A-T Society has gone into partnership with two other charities – Action for A-T and Sparks – to set up an A-T research fund.

The first call for applications closed in August and five applications were submitted. Of these, three were sent out for peer review – i.e. sent to other scientists for their comments on the quality of the project.

The administration for the assessment programme was carried out by Sparks. In October, their Medical Advisory Committee of experts met to assess the applications.

Two of the applications were considered particularly interesting, but, in the view of the committee, they needed a bit more work. This is currently being done.

Rather frustratingly, as we go to print, a final decision on awarding the grant has not yet been reached, but we very much hope that an announcement will be made early in the new year. Keep an eye on our website or Facebook page for news of this.

However, whatever decision is made, we will be working with some of the unsuccessful bids to try and put together funding from other sources.

A number of other researchers expressed an interest in applying but didn't get applications in before the deadline. We are therefore hopeful that the next call, in 2013, will attract even more applications.

The project is about more than research, though, and the three charities are collaborating and sharing resources to raise money for the fund.

Sparks has very generously made available some places it has for the 2013 London Marathon, while the three charities are sharing places for the Ride London 100 cycling event in August.

ACTION FOR A-T

Sparks
THE CHILDREN'S MEDICAL RESEARCH CHARITY

Scientific advisory board

We are delighted that Professor Penny Jeggo has agreed to chair a new Scientific Advisory Board for the Society. Penny is a very well-known and widely respected figure in the A-T research community and will bring to the role a very broad knowledge of A-T research.

Penny's background is as a cellular molecular biologist. She has worked extensively on DNA damage response mechanisms and, in particular, the role of the ATM protein in this. She has also identified another genetic condition, Lig 4 Syndrome, which like A-T combines radio-sensitivity and immune-deficiency.

The Board will advise the Society on scientific matters and will shape its research strategy. It will also assess applications for research grants and make recommendations to the Board on which grants they should make.

The Board will meet for the first time in the new year. Its members will include Professor Malcolm Taylor of the University of Birmingham and representatives of the two A-T clinics at Nottingham and Papworth. The final members are currently being agreed.

Do you have a story you'd like to be told?

We are working with a freelance journalist who is experienced in placing articles in the national press and magazines to raise awareness of medical conditions

From our perspective, increasing public knowledge has resulted in potential new diagnoses after parents have read articles about A-T and recognised the symptoms in their own children. At least one of these children had been misdiagnosed for many years.

Of course it also has a benefit to us in terms of raising the funds which are essential to ensure the charity will be here for every A-T family in the years to come.

If you would like your family to be involved, please in the first instance contact Suzanne Roynon at the office.

Two new patrons

The A-T Society is both delighted and honoured that two high-profile people have agreed to become Patrons of the charity

Brian Conley

From top, going clockwise: Merry and Brian; Brian with Helen Carter; Brian with Jo Bloomer

Brian Conley is very talented and multi-faceted entertainer. Comedian, TV presenter, singer and actor, he is one of the most popular stars of the showbiz world. Brian has been a familiar face on television and in theatres across the country for many years, appearing recently in *I'm a celebrity, get me out of here*.

Brian became aware of A-T while he was performing as Fagin in Oliver at the theatre Royal in Plymouth. Jo Bloomer, mother of Lola, who has A-T, had appeared on the front page of the local paper, the *Plymouth Herald*, telling their story (see page 21). Brian read this and was so moved that he decided to put on a special late night show to raise money for the A-T Society .

His generosity and his high profile meant that for a week or so, Lola and A-T were constantly featuring big on the pages of the *Herald*, and the show eventually raised more than £11,000.

Brian said of his new role: "I am honoured to be a part of the A-T Society. When we read about Lola, who is absolutely gorgeous, we felt moved and had to do something. My reaction was that I needed to do something to help find a cure and, if my support and my name can help do this, then I'm with you."

Lord Bilimoria

Lord Karan Bilimoria has had an illustrious business career and is a champion of social responsible business. He is best known as the founder and Chairman of Cobra Beer.

After growing up and graduating in India, he moved to the UK, where he later took a law degree at Cambridge. He was moved by his experience in British restaurants to produce a lager which had less gas and would go well with curries. Thus, Cobra Beer was founded.

In recent years, Lord Bilimoria has been very active outside his business life, with a wide range of charitable and other interests. He has taken on many roles including those of Chancellor of Thames Valley University, Vice Chairman of the London Chamber of Commerce and President of the UK India Business Council.

He has received honours from a number of different countries and was made a life peer in 2006.

On his new appointment, Lord Bilimoria said: "I have been very touched to see the impact that A-T has on children who have it and on their families. I am extremely impressed by the work the A-T Society does to support these people in living their lives to the full now, and to bring hope for the future through research. I hope that my support will help strengthen them in this work."

We are very grateful to both Lord Bilimoria and Brian for their commitment to supporting the A-T Society and look forward to working with them to raise the profile of A-T.

Benefits update

Housing Benefit
From April 2013 there are going to be some changes for those who claim housing benefit for council or housing association homes.

This will involve new criteria for how many bedrooms/children you have and if you are 'under occupying' your home. (i.e. if you have a spare bedroom or young children are not sharing).

This may mean that you will receive less housing benefit. However, if you have a disabled child who has to have their own bedroom because of their condition, this situation can be assessed by your local housing benefit office.

Child Benefit
From January 2013, child benefit is being withdrawn from high income families. This will affect individuals earning more than £50,000 per year.

New Universal Credit
This will be launched in 2013 and will replace:

- Income-based Jobseeker's Allowance
- Income-related Employment and Support Allowance
- Income Support
- Child Tax Credits
- Working Tax Credits
- Housing Benefit

If you have any questions about any of these benefit changes please do contact Kay.

Support Grants

In 2012 we made many support grants to families and adults affected by A-T. Examples of what we funded include:

- Respite/holiday breaks
- Wheelchairs
- Driving lessons
- Therapy sessions
- Adapted trikes

Lisa and her daughter Ella had a wonderful time at Butlin's this year. The Society funded a week away for them as they hadn't had a break for many years and Lisa's condition was deteriorating. Lisa has a milder variant of A-T and her daughter Ella is a young carer.

Lisa said: "We had a fantastic time; it was great to get away from home and spend some time together, without the stress of normal everyday life."

Lisa & Ella at a Butlin's character lunch

If there is anything that you need help with, please go to the 'Support Grants' page on our website to read our criteria and complete an application form. Please contact Kay if you need any advice and help with this.

Introducing our new A-T medical cards

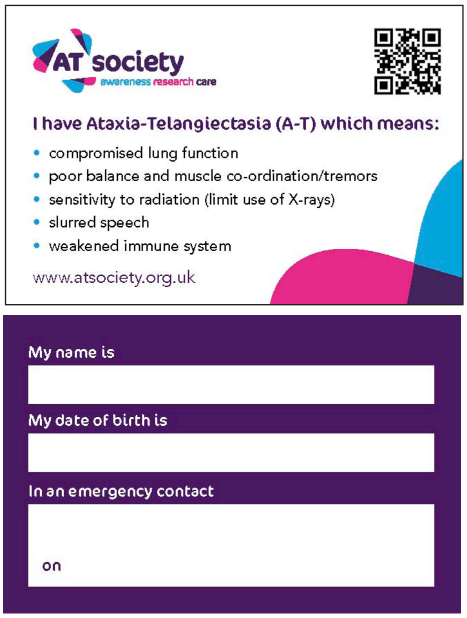
We are delighted to introduce our new 'Medical Card' (see below). This is the size of a credit card and will fit easily into a purse or wallet.

The card is a simple way of telling people about A-T and its most important symptoms, as well as directing them to our website for more information.

In the case of a medical emergency, it will ensure medical practitioners have the information they need to know.

We are in the process of sending these out to everyone in the UK and Ireland.

However if you live elsewhere in the world, or if you would like an extra card, we would be happy to send you one. All we ask for is a donation of £2.00 to cover postage. Please contact Kay if you would like one.



AT society
awareness research care

I have Ataxia-Telangiectasia (A-T) which means:

- compromised lung function
- poor balance and muscle co-ordination/tremors
- sensitivity to radiation (limit use of X-rays)
- slurred speech
- weakened immune system

www.atsociety.org.uk

My name is

My date of birth is

In an emergency contact

on

UP FOR GRABS: Electric, height-adjustable table (40x36 inches). Ideal as work table for computer. Was very expensive when new. Current owner requests the new owner gives a small donation to the Society as payment. If you're interested please contact the A-T Society.

Upcoming events

Family Weekend
22/23 June 2013
Manchester

Next year we will be holding our annual Family Weekend at the Radisson Blu Hotel, Manchester Airport. This is a fully accessible hotel with excellent rail and airport links. We have chosen to come north this time at the warm invitation of some of our northern families.

We will have speakers on a number of topics including the latest research into A-T, education and IT equipment, as well as a wonderful banquet and an opportunity for you to meet up with other families. There will be craft activities for the children as well as ten-pin bowling for all ages. Kay will be sending out further details in January together with booking forms.

Radisson Blu Hotel – Manchester Airport

Young Adults Event
15/16/17 March 2013
Milton Keynes

This event is for people who are already involved in the 'My Life' project, which talked to young adults about their experiences and aspirations.

Central to the weekend will be a workshop with opportunities to discuss the findings of the report and to discuss how the Society can most usefully respond to them. The workshop will be facilitated by Bryony Beresford of York University.

In addition to the workshop, there will be other optional and more energetic activities (see right). Those eligible for the weekend should have received details through the post. If you haven't yet registered for this and are between 16-26 years old and would like to attend please contact Kay ASAP as there may still be spaces available.

Airkix indoor skydiving activity

PS3 helps with schoolwork

By Tina Stubbs

My son with A-T was having real difficulty using a computer at school. We had tried keyguards, intellikeys and joysticks but nothing seemed to work for him. However, since he was small, he has been an avid computer-games player and we realised that he was quite comfortably sending messages to his friends via his PS3, using the controller to write the messages.

After a discussion with the IT department at his school (Treloar's in Hampshire), they said they would be able to write some software to create an interface with the systems used by the school. They have now done this and he is happily doing his schoolwork using his controller. This has opened up a whole new world for him. Another advantage for the school is that PS3 controllers can be bought for as little as £6.99 – whereas specially made equipment often costs hundreds of pounds.

At the moment, we are waiting for the school to produce some software to enable him to use the same controller on a PC at home. Once they have done this, we will make sure the A-T Society knows in case anyone else is interested.

Kay Atkins is our Family Support Worker.

Kay's normal working hours are Monday to Friday, 8.30-4.30.

You can call Kay on 01582 760733 or email kay@atsociety.org.uk

Numbers!

You may be interested in the following stats about A-T:

145
Total children/adults registered with us in the UK (with A-T or 'A-T like' condition)

83
Adults (16 & over)

62
Children (Under 16)

Out of 145:
136 have A-T
9 have A-T like conditions

Out of the 136 who have A-T:
68% (approx.) have classic A-T
32% (approx.) have a mild variant

The symptoms of mild-variant A-T are broadly the same as those of classic A-T, except that they tend to progress more slowly and may appear later. While some people with this form of A-T are diagnosed in childhood, others receive a diagnosis as adults.

For some, the symptoms may have been apparent for a while, perhaps appearing during teenage years, but have been misdiagnosed as another condition. For others, they only reveal themselves in adulthood.

Geographical spread of children/adults in the UK:

6 in N. Ireland
8 in Scotland
12 in Wales
119 in England

Blazing a trail

By Rupert Prokofiev

I first found out about the Trailblazers at the A-T family weekend in 2011. Ever since then I have been involved with Trailblazers, a group of young disabled people who campaign for disability rights.

I have mainly been going to Parliament with the APPG (All Party Parliamentary Group) for Young Disabled People. These have all been days when I have had to go in to central London from my home in Greenwich via the Thames Clipper.

At Parliament we have discussed a range of different issues that we face with our disabilities. The issues we have discussed, for example, have included issues surrounding cinemas, housing, hate-crimes, public transport, higher education, and more.

On Wednesday 21 November, I attended another APPG session where we gathered all the evidence we had discussed in the past and it was published in a report, *Removing Barriers, Promoting Independence*, which was released that day.

Through Trailblazers I have also been on *BBC TV* twice with the APPG, talking about hotel accessibility and about the cuts to the disability benefits after the Paralympics.

I discovered and took an interest in the Trailblazers through the A-T Society, so I encourage others to grab the bull by its horns and get your voice out there.

Trailblazers is a national network of more than 400 young disabled people who work together on a national and local level to highlight and address the issues that are important to them.

The group is organised by the Muscular Dystrophy Campaign, but any young disabled adults are welcome. For further information about Trailblazers and how you can get involved go to www.mdctrailblazers.org.

Studying breathing

Dr Emma Ross from the University of Brighton is currently undertaking a study to improve the strength of breathing muscles in young people with A-T, jointly funded by The A-T Society and the A-T Children's Project.

In A-T, weakness and uncoordinated control of muscles also affects those muscles which allow us to breathe. People with A-T often find breathing more of an effort and as a result their cough can become weak. This in turn can lead to repeated lung infections because they are unable to clear mucus effectively.

The research team are working in the laboratory to ensure that the testing equipment and protocols are sensitive enough to detect even small changes in lung and respiratory muscle function in people with A-T.

The research team is looking for volunteers. Please contact Kay at the A-T Society if you're interested, and she will pass on your information.

Dr Ross hopes to be able to present some of the preliminary findings of the study at the 2013 family day.

Fantastic Florida

Two of our children have recently been to Florida – one with 'Make a Wish' and one with 'Dream Flights'. Both said it was a holiday of a lifetime

Kira, Make a Wish

We went to Florida for 7 nights with the 'Make a Wish Foundation' and 'Give the Kids the World'. The whole experience was magical and Kira enjoyed every minute....so did we.

We visited all the Disney Parks twice, Seaworld, Universal studios and Legoland. We met Santa at Mickey's Very Merry Christmas Party. We watched the fireworks and parades at Magic Kingdom, Epcot and Hollywood Studios.

Kira met and got the autographs of all the Disney Princesses and lots of characters that she loves, including Spongebob Square Pants.

All in all, it was a holiday of a life time.

Hayley

For more information on how they got go to, you can visit the websites:

www.makeawish.org.uk
www.dreamflight.org

Mert, Dream Flights

Mert (age 12) went to Florida without his family, with 'Dream Flights' (who provide 24 hour care and medical support).

He said: "I had a fantastic time in Florida and went on every scary ride! I met some wonderful people and made lots of friends. It was definitely a holiday of a lifetime!"

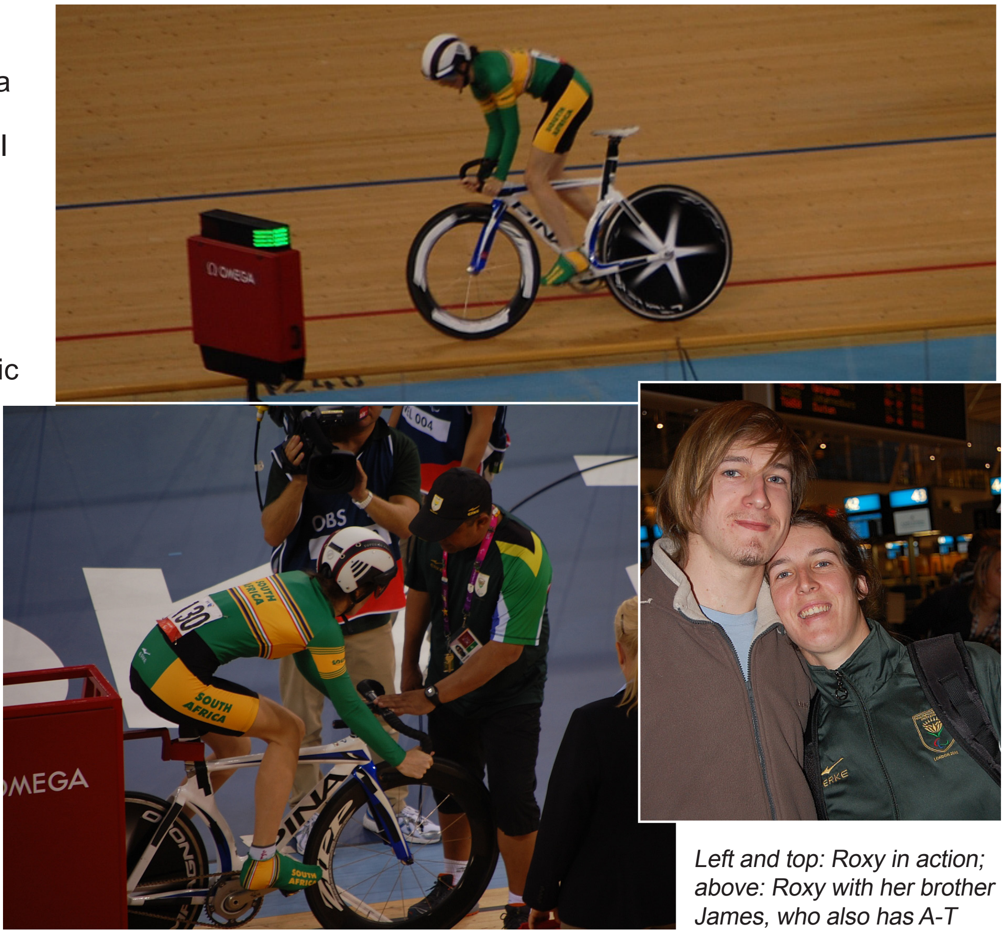
Kira (top pictures) and Mert (above) had fun and made new friends in Florida

Paralympic pride

William Davis caught up with South African cyclist Roxy Burns, who competed in the London 2012 Paralympics and who has A-T

As stars go, South African cyclist Roxy Burns is rather shy. Roxy, who has represented South Africa at both the Beijing and London Olympics, and won a silver medal at the World Championships in Manchester in 2009, is in all probability the most successful sportsperson with A-T that there has been.

We met up with her at the Olympic Park in Stratford, after she had completed a very successful week, competing in three different events. While she didn't win any medals, she broke her personal best in the first event, almost did so in the second and posted an excellent time in the road-race time trial, where courses vary so that you can't really have a personal best.


Left and top: Roxy in action; above: Roxy with her brother James, who also has A-T

I asked her how it had been riding in the velodrome: "Incredible! Wow!" she says. "I've never raced in front of a crowd like that. You can hear the sound following you round the track. In Beijing, the velodrome was much emptier – in fact that they had to bring in bus-loads of school children. And in Manchester, the crowds were much smaller."

And what was it like living in the village? "That is amazing. It's so... vibrant! There are so many people, though what with training and competing you mainly get to mix with people in your own team. But I'm also friends with the other competitors in my group, especially the Aussies."

Roxy has a milder variant form of A-T. Nevertheless, it turns out she was diagnosed at the age of 18 months. Like most people with A-T, she was wobbly when she started to walk. A doctor suggested Cerebral Palsy but her mother refused to believe that, as both pregnancy and birth had been free of any complications. She took Roxy to a children's hospital in Cape Town where, after a series of tests, a doctor asked for a radio-sensitivity test, and A-T was diagnosed.

Later, her parents chose to send her to a special school and it was there that she took up cycling. She was encouraged by the physios to do sports because they said it would strengthen her muscles. Initially she was involved in track and field athletics as well as cycling, but in 2005 she was selected to cycle at the CPIRSA (Cerebral Palsy International Sports and Recreation Association) games in Connecticut USA, and decided to specialise.

So how does her A-T affect her cycling? The bike she rides is a normal track racing bike – and that means it has neither gears nor brakes. However, she manages that fine. The main problem she has is with her balance. This makes it difficult for her to hold a straight line, even when travelling at speed – and she reaches speeds of more than 48 kph. Fatigue, she says, doesn't affect her – at least no more than it would anyone who trains for three hours a day, six days a week.

As to diet, she has no problems eating. While she doesn't have a special training diet, she does eat healthily. She also tries to eat little and often. "So what about the food at the Olympic Village?", I ask her. "It's great!" she says. "There are all these different sections you can go to for different styles of food – British, Mediterranean, African, Indian and so on."

"Is the British section where you go when you want some quiet?" She laughs: "Actually, it's the busiest..."

She trains both in the gym and on the track. This obviously requires real dedication and, as she tells me, it can get very lonely. I ask her what inspires her when she's pounding away at the training. "It's moments like these. It's going overseas and performing in front of crowds. This all makes it worth it. And then there are the other people in the team. Natalie du Toit (winner of 13 gold medals for swimming) and Oscar Pistorius are inspirational. They have just done so well."

When she isn't on the track training, Roxy is studying to be a teacher. She hopes to use the coming year, when there are no major international sporting events, to complete her postgraduate certificate of education so that, once the Rio Paralympics are over, she can start looking for a job – perhaps overseas.

But Rio is firmly in her sights. She tells me that her times are continuing to improve, so I ask her when cyclists reach their peak. "Late 20s." she tells me. "So you'll be reaching your peak about the time of the Rio Paralympics?" "I guess so." "Well, you'll have a huge fanclub by that time", I tell her, "and we'll all be expecting you to perform. There'll be a lot more pressure!" Roxy just smiles. You wouldn't bet against her...

More fun at the Games

We were lucky to get tickets for three events at the Paralympics Athletics, Swimming and Wheelchair Basketball, and a number of our families were able to go. We were even joined by a young man from France, Laurent Poursoubire, who was featured in last December's edition.

Sadly we were unable to get tickets to see Roxy Burns, the cyclist with A-T (see previous page), as her events were sold out. Everyone was impressed by how accessible all the venues were and all the volunteers, the 'Games Makers', were great.

Our Family Support Worker Kay went with her husband Kevin to help Stephen and Neil (who have A-T) and their Mum. Kay said: "We went by public transport, which, with 2 wheelchair users, was an experience in itself! Everyone had a fantastic day, we went to see the Athletics, and had excellent accessible seating with a great view of the track. It was a very hot day but luckily we were in the shade! The atmosphere was electric and we were all screaming when team GB were on the track. We saw some wheelchair races, running races and long jump (some of whom were amputees or blind). It was very inspirational seeing these athletes, who have trained for many years, going out and giving their all. It was a very long day and I think Stephen and Neil were exhausted by the end, but they were adamant it was well worth all the effort!"

George also went, with his Dad Nick, to the Athletics Stadium. George said afterwards: "It was a really good day out, I really loved seeing David Weir and Oscar Pistorius – the 'blade runner' – they were great!"

RideLondon 100

Prompted by the success of Team Sky and the Olympics, the Mayor of London – with the help of Team GB and the organisers of the London Marathon – has set up the first ever closed road cycling event in the centre and surrounding areas of London. Starting in the centre of London, professional and amateur cyclists will ride the 100 mile challenge on the same closed roads as the Road Race for the 2012 Olympics.

Taking place on the weekend of the 3-4 August, the ride will start in the new Queen Elizabeth Olympic Park, following roads through the capital into Surrey's picturesque road and hills. The route is designed to be challenging but also rewarding for all those taking part.

The A-T Society has 18 places available for the inaugural RideLondon 100 and it's expected to become an annual event. We will be asking all place holders to raise a minimum of £600 in sponsorship. We will be participating alongside our two partner charities – Action for A-T and Sparks – who will also be raising funds for A-T research. The Sparks event team will be running the administration for the event.

There are some great incentives for taking part in the joint charities A-T RideLondon 100 team, including a Tour de France jersey signed by Gold Medal winner Sir Bradley Wiggins. We're hearing rumours of other incentives to make you itch to take part... you heard it here first!

Unravelling the secrets of A-T

William talks to Yossi Shiloh, who is best known for leading the team that identified the ATM gene

Yossi Shiloh's name will be familiar to everyone whose life is affected by A-T. Yossi (a diminutive for Yosef) is best known for leading the team that in 1995 identified and cloned the ATM gene, mutations in which cause A-T, and showed that ATM was the only single gene responsible for the disease.

However, over the years Yossi and his team have done a huge amount of work to explore the role that the product of this gene – the ATM protein – plays in the cell and how its absence contributes to the clinical effects of A-T. In fact, as he made clear when I spoke to him, unravelling the secrets of A-T is his life's work.

And Yossi is deeply serious about this work. As an opener, I asked him how he would describe what he does to someone at a dinner party who didn't know him. He launched into detailed discussion of his work and 10 minutes later was still talking. And yet everything he says is beautifully clear, even to the lay-person. He has a gift for explanation and illustration that hints at the sharpness and clarity of the mind beneath.

Based in Tel Aviv, Yossi works closely with the team at the Israeli National A-T Clinic. Israel is an interesting country with regard to A-T. There are a number of different communities within the country where A-T is found more frequently than normal. As a result, Israel has a higher number of cases of A-T than you would expect for its population size. And, being a small country with a unified health service, most of these patients are in close touch with the national clinic.

Yossi talks to an A-T family at the Family Day in June

I ask him what first got him interested in A-T. Like many other A-T specialists it was contact with an A-T family: "I had just finished my Masters and was looking for a subject for my PhD thesis. My Masters mentor, Professor Maimon Cohen, who was interested in A-T, asked me to join him on a field trip to visit a family with A-T. This was the summer of 1977. We visited the family, who were of Moroccan-Jewish origin. They had 10 children, of whom four had A-T. The huge personal impact of the condition on their lives made a real impression on me personally.

"At the same time I was fascinated scientifically. It was already known that A-T was caused by mutations in one gene and it was clear to me, from the range and complexity of the symptoms of A-T, that this gene had a very important function. I knew I wanted to work on it."

This complexity led to some real challenges. There are a number of conditions where the same symptoms can be caused in different patients by mutations in different genes. This usually happens when the proteins encoded by these genes work together in a particular process. If any one of the genes is mutated, the process will be affected, leading to the symptoms of that particular condition. Thus, although in each affected family only one gene is mutated, different genes may be defective in different families with the very same disorder.

Yossi continues: "Some very meticulous work done in several labs appeared to show that A-T was this sort of condition. It was widely accepted that there were four separate A-T genes – known as A-TA, A-TC, A-TD and A-TE. A major breakthrough took place in 1988 when the A-TA gene was localised by Professor Richard Gatti's team at UCLA, to a specific chromosomal region on human chromosome 11. Our group was then able to show that the A-TC gene, which was thought to be mutated in the Moroccan Jewish families, was localised in the same region. Subsequently, it became apparent that all four A-T genes were located closely together on chromosome 11.

"This led to a long period of intensive work in many laboratories involving considerable international cooperation. This cooperation is most notable, since it accelerated the efforts to identify the elusive A-T genes. Eventually our laboratory was able to identify a big deletion in one of our candidate genes in a big Arab A-T family. The deletion was associated with the disease and, in all the patients in this family, it was found in both copies of the corresponding gene. Since this family was known to represent a mutation in the A-TA gene, we assumed that we had identified this gene.

"We had to decide whether to publish this finding as a discovery of the A-TA gene, or go on to check out the possibility that this was not one of four A-T genes but rather a single gene, which was mutated in all families with A-T. We decided to do the latter. We looked for mutations in this gene in other A-T patients representing the three other supposed A-T genes – and found them! We therefore concluded that A-T was caused by mutations in one gene alone – the one we had identified. We called it "ATM", which stands for A-T, mutated.

Identifying the gene for what turned out to be such a large and important protein was a highly prestigious achievement. It also marked a major step forward in the journey towards understanding A-T. Importantly, it meant that DNA-based prenatal diagnosis and carrier detection could now be carried out in A-T families around the world.

For Yossi and his team, though, it also meant a change of focus. Now that the gene had been identified, they increasingly turned their attentions from molecular genetics to protein chemistry, as they tried to understand the complex role played in the cell by the ATM protein. And complex it is. ATM reacts with hundreds of different proteins and modifies their activity or stability. This gives the growing community of A-T researchers around the world plenty to work on.

I ask what his current priorities are? "Continuing basic research into ATM and ATM-related processes. We have several students working on the function of ATM in the cerebellum. Our thinking here is in a process of transition, based on accumulating evidence."

ATM's best documented function is mobilising a complex network of responses to an important form of damage to DNA called a 'double-strand break', which is caused by ionising radiation. Yossi is increasingly convinced that ATM is involved not just in the cellular response to double-strand breaks but also in other processes. "We think it plays a role – maybe not a direct role, but a supporting one – in dealing with other forms of DNA damage. There is also increasing evidence for the involvement of ATM in processes outside the DNA damage response arena."

We concluded that A-T was caused by mutations in one gene alone – the one we had identified. We called it "ATM", which stands for A-T, mutated.

His team is also trying to establish new mouse models that show the neurological problems typical of A-T patients. This is crucial if we are to be able to test new treatments.

So what approaches does Yossi think are most likely to deliver effective treatments? He believes that the one his laboratory is taking is a promising one. It is based on a search for small molecules that can compensate for the loss of ATM. "We have been conducting a drug search, in collaboration with the National Institutes of Health in the USA, screening hundreds of thousands of small molecules. Cells from A-T patients are treated with DNA damaging agents that activate ATM and then are exposed to these molecules. We look out for molecules that appear to allow A-T cells – which usually do not respond well to this challenge – to respond better. We are currently analysing the results."

ATM is a member of a family of proteins with distinct but

Continued on page 16...

Continued from page 15...

overlapping functions. Their hope is that it may be possible to use a small molecule to stimulate one or more of the proteins with an overlapping function to compensate for the absence of ATM.

However, the problem with small molecules is that they usually affect more than one process. So even once you identify a ‘hit’, i.e. a molecule that seems to have an impact on the A-T cell, you have to try and understand how and why it does so and there may well be a lot of complex chemical manipulation to do to try and prevent side effects.

What about approaches like gene therapy? “I am not sure that it will be very easy to get gene therapy to work for A-T. There are enormous technical problems to overcome. Since the most devastating effect of A-T is on the nervous system, this is what we need to correct first. In principle, it can be done, but there are big challenges in introducing a gene into the large number of cells that make up the nervous system. And once it is there, you have to control where it ends up. Uncontrolled manipulations of the DNA in tissue cells may lead to unwanted consequences. And A-T patients, where it is precisely the stability of their DNA that is compromised, are likely to be particularly vulnerable to this.”

It is hard to imagine Yossi not being a scientist, but out of curiosity I ask what he would like to do if he weren’t doing science. He pauses... “A conductor – conductor of an orchestra,” he says. I assume that this means he must play music himself, but no: “When I was young, I used to play the flute”, he says, “but I really wasn’t very good. However, I love listening to music – especially watching concerts and operas on DVD.”

So what would he most enjoy conducting? He thinks about it. “Mahler symphonies. That is what I have most enjoyed listening to in the past few years. Just recently my wife and I attended an incredibly exciting concert in Barcelona, with the San Francisco Symphony Orchestra playing Mahler’s 6th symphony. It was almost a religious experience.”

But it’s not just concerts. Yossi enjoys all the performing arts: theatre, opera and dance – modern dance in particular. “Tel Aviv is a fantastic city in that regard,” he says. “It offers an enormous amount of culture.”

It’s fortunate that he has these other outlets, because as a scientist he is very focused. When I ask if he has other research interests beyond A-T, the answer is ‘no’. “I did when I started the lab. A-T was just one of four research projects. But after Dick Gatti’s initial localisation of the gene it became clear that it would be a long and demanding project, so we stopped all the others.”

So what keeps him awake at night? “I sleep fine at night”, he tells me, “but I do wake up early. I love the beginning of the day. I jump up and the first thing I do is take a quick look at PubMed (a database of biomedical literature) to see what’s new in the ATM world. I do that before I find out, later during the day, what is new in my own lab. But I love that time of the morning, when it’s still quiet and you can read and think.”

And finally, I ask him what message he might have for people living with A-T. “The same as it has been for many years. We have to study and understand the disease and then, based on this, hopefully we, or others, will be able to design treatments.”

This might not be the sound-bite that people living with A-T are hoping to hear, but as long as Yossi is working on it, we can be confident that progress will be made.

Betamethasone study update

In the last edition of *A-T Society News* we published an interview with Luciana Chessa. In August she was one of the authors of a paper reporting the results of a trial of double-blind placebo controlled trial using oral betamethasone.

The trial involved 13 children between the ages of 5 and 16, all of whom were able to walk. The dose used was low, but not as low as the dose used in an open study published by Broccoletti in 2010.

The study showed significant improvements in the participants scores using the ICARS scale. This is a scale used generically to assess ataxic symptoms in a variety of conditions, and it must be pointed out that most people with A-T show a range of neurological symptoms in addition to ataxia.

While this is positive, participants on the trial did also start to exhibit some of the side effects of prolonged steroid use.

The authors conclusions are: “Oral betamethasone could be a promising ataxia symptom-relief therapy in A-T patients; however, larger-scale studies are warranted. Long-term effectiveness and safety must be established.”

A-T Neuroscience Forum in Washington

In early November, 60 experts in neuroscience and A-T came together in a hotel in Washington to review the current state of our knowledge of the neurological aspects of A-T

The aim of the forum was to identify the most promising avenues for developing new treatments and any obstacles that are preventing the discovery of new approaches.

Organised by the A-T Children's Project, this was not a traditional scientific conference with formal presentations. Each session took the form of a series of chaired discussions with a panel of experts for each subject with knowledge not just of A-T but also of other conditions and scientific techniques. This refreshing approach enabled information and ideas to be shared and tested, and allowed many voices – including those of people living with A-T – to be heard.

Brad Margus, chairman of the A-T Children's Project, hosted and chaired the whole forum. One nice touch was that he asked members of the panel for each session to sit on the stage in wheelchairs. As Brad said, we should never forget why and on whose behalf we were all here nor the urgency of finding treatments and a cure for A-T.

The subjects discussed covered a wide range of issues, including animal and cell-based models, oxidative stress, inflammation, stem cells, gene therapy and neuro-circuitry. This gives some idea of the wide scope that there is for developing treatments for A-T.

There is clearly a lot of work going on in all these fields and much progress being made. There were a lot of interesting ideas and suggestions for new research. Nevertheless, the prospect of a rapid break-through still feels just beyond our reach.

Part of the problem is that there is still much that we don't know about how and why A-T affects the parts of the brain that deal with movement. However, this is also true for many other much more common diseases affecting movement, such as Parkinson's or Motor Neurone disease. There is much that we can gain by working more closely with those studying these other conditions.

On the positive side, some excellent imaging work is now being done by Norah Volkow in Bethesda, USA, which is starting to show differences in activity in the different parts of the brain between people with A-T and others. Similar work is being done in Brisbane and we hope to fund a new imaging study to complement these in Nottingham next year.

Another area which seems to have good potential is the use of 'neurotrophic' and growth factors. These are substances produced by the brain which help brain cells to grow and flourish and protect them from damage. Studies in Parkinson's disease have been done and are showing some promise and this is an approach which should be looked at for A-T.

A number of different researchers are currently producing stem cells which can be used to gain a better understanding of A-T and to test potential treatments and drugs. However, the prospect of being able to use them to replace damaged cells in the brain is still some way off. Stem cells, though, have many properties beyond that of creating other cells, and there is potential that they can be used much sooner for their ability to strengthen and protect existing cells.

Brad Margus, A-T Children's Project

Gene therapy is another area that many people are looking to with hope. Again, while there has been some good progress, there are still many challenges to overcome. You can read more about these in the interview with Yossi Shiloh on page 14.

There is also positive news about the development of animal models for A-T research. One of the problems facing researchers is that mice which are bred to produce no ATM do not show the same neurological problems as people with A-T. However, one team in America thinks they may have found a way to breed mice that do, and a team in Australia is making good progress with ATM-deficient rats. There is also now a strain of fruit-flies that are A-T deficient. While this may sound odd, fruit-flies can be extremely useful for research, and have the advantage that they can be bred very quickly.

All in all, then, it was an extremely useful event, which has raised the profile of A-T within the research community and should give a new impetus to research and encourage new projects and new collaboration. Brad and his team at the A-T Children's Project are to be thanked and congratulated for their efforts in putting it on.

Fundraising

Busy busy busy! Where have the past few months gone?

The A-T Society was introduced to many delightful new fundraisers during 2012, and the 2013 calendar is filling up with exciting events, many of which you can get involved with or be inspired by.

We are big fans of BBC choirmaster Gareth Malone, so it was exciting to learn that the Military Wives will be performing at a Ball in aid of the Society in the early part of the year. We're also hoping to gather a lot of interest for Dragon Boat racing at various venues around the country.

There should be some strong media coverage for the Mega-Tri in April and we have a journalist working with us to try to place some awareness-raising articles in the national press and various magazines. If you have a story you would like to tell, please let me know.

It's great to see so many families and supporters requesting collection pots to place around their local area. If we can get enough collection pots on shop counters, we have the potential to raise enough each year to cover the cost of an additional medical research project. Can you help every parent's dream come true?

Finally, we have a new permanent member of the fundraising team. You can read all about Atey Bear in his own words on the back page; please give our latest recruit lots of support in the coming year!

Suzanne

Tough Mudder

Not to be outdone, Paul Gordon and his friends took on the 'Tough Mudder' – a 10 kilometre race through mud, cold water and general filth.

We applaud their determination to make the world a brighter place for everyone with A-T, and send our commiserations to whoever had to do the washing afterwards!

Larkfields School, Nottingham

This is Kaid with his teacher Mrs Parnell and his teaching assistant Miss Wright from Larkfields Infant School.

Even before Kaid started at Larkfields, the school chose the A-T Society for their Charity of the Year and donated a whopping £1,603.15 to the charity.

Headteacher Mr Taylor thought it would be a good way for the children to learn about A-T and have a greater understanding of the condition before Kaid started at the school.

If you would like to get your school involved, please contact Suzanne for information.

Worldpay: an action-packed year for A-T

A round-up of the amazing fundraising activities the Worldpay team have got up to this year

The A-T Society came to be the Worldpay Cambridge Charity of the Year after a determined campaign by Lexy Morgan (a friend of Jo Betts) and Alan Staples – who was working for Worldpay and supports the A-T Society with his IT skills. Sarah Jarvis, Worldpay's charity coordinator, gives an overview of their charity activity this year:



The brave skydiving team prepare for the big jump...

In February we kicked off our fundraising with a quiz and Mardi Gras pancake fiesta. We found out early that the way to our colleagues hearts is via their stomachs and have maximised this information (along with their waistlines) throughout the year!

March saw us taking advantage of the Cheltenham Festival and as avid virtual race-goers we tipped our way to success.

April saw an Easter raffle and yet more turf-inspired fundraising with a Grand National Sweepstake. By June, the Jubilee fever took hold and we hosted a mammoth tea party, selling plates of homemade cakes, savouries and sandwiches with great success.

It was great fun and very inspirational to be able to support the A-T Society Family Weekend. I went along with my husband Peter to escort the brilliant Charlie and Arthur to Duxford on the children's outing, and Lexy Morgan was there to escort Kaid Betts and his brother Ben.


The 10K runners

As June slipped into July, we gave Sir Alan Sugar a run for his money with the A-T Society Challenge. Three teams were given one month and £50 to raise as much money as possible. The teams raised a whopping £1,722.55 between them by selling homemade cakes, dosa batter, pizza, strawberries and ice cream and sloe gin. They also organised a quiz, Euro 2012 sweepstake, pool competition, raffle and international food fair.

July also saw us field a team of six in the British 10K London Run and then, just two weeks later, we got on our bikes in a team of 11 to take part in the London to Cambridge bike ride. Several people took part in both events and were super fit by the end of the month!

In August we celebrated the 'summer' with a Golf Tournament at the Cambridge Lakes course, and in September we had another, larger, general knowledge quiz event.

October dawned bright and beautiful and 14 intrepid staff made the short journey to Chatteris in Cambridgeshire to take part in our first tandem parachute event. With blue skies and a light breeze it was the perfect day for skydiving and the event really brought us together as a team.

As October drew to a close, Rob Honour launched the "Comfort Cafe Cruiserz Car Club" based at Abingdon just outside Cambridge. We provided a stall of A-T merchandise and were thrilled that William Davis was able to attend.

The tail end of the year will see a craft fair and Christmas raffle.

The staff really got behind the A-T Society in 2012, taking the children and the cause to their hearts. We already know this is our best ever Charity of the Year total and it's a huge achievement for us.

However, we're not letting on just how much we've raised quite yet. That's a secret we are keeping until our James Bond-themed Presentation Night in January 2013 when we will surprise William, Suzanne and Kay with a cheque!

Raising funds and awareness

The Clarke Willmott team in Bristol have gone to great lengths to support the A-T Society

Our two-year Charity Partnership with renowned law firm Clarke Willmott LLP's Bristol office officially began in January 2012 after lawyer Philippa Hann nominated the A-T Society and mounted a strong campaign to see us adopted as one of two charities to receive support until 2014.

Philippa is a close friend of A-T mother and trusteeTania Wheeler, and was determined to raise funds and awareness for A-T throughout the company and further afield through their links with individuals and businesses throughout the South West.

Clarke Willmott has an established Charity Committee and a strong background in corporate social responsibility. This is a summary of their accomplishments so far from one of CW's A-T Society champions, Catherine Zakarias-Welch:

In March we hosted our Virtual Cheltenham Race Day. Clients and contacts joined us for an Irish-themed, three-course lunch followed by an afternoon watching the racing. Bets were placed with our trainee solicitors who each took a table and looked after the bets. Guests were extremely generous and supportive of our charities.

Clarke Willmott lighting the way

The Clarke Willmott Monopoly Challenge

We also held a quiz and raffle in March. We pitted our minds against each other with music, geography and picture rounds to name but a few.

May 2012 saw another fantastic Bristol 10k and the Clarke Willmott Business Challenge. Two teams from the Clarke Willmott property litigation team took part for the A-T Society. Kevin Jones, head of the Bristol office, said: "The CW marquee was, once again, a hive of activity. It was a real focus point for runners and supporters. Well done to all!"

The 12 month countdown to the Bristol 10k 2013 is now on…

June saw Clare Norman and the marketing team volunteer their support for the A-T Society International Research Conference, as we assisted William with organisational support and lent our administration skills. We were also pleased to be part of the Family Weekend when one of our solicitors, Robert Smeath, gave his professional advice to families who wanted to learn about making wills and provision for the future of their children, and our graphic designers, Minty Design, created a flyer for Trevor Sanderson's Marathon des Sables.

In June we also hosted the opening England Euro game (on television) in Bristol, Birmingham and Taunton. Each office saw good attendance with clients and contacts enjoying an evening of networking over drinks and nibbles. Each office raised money through sweepstakes and donations.

Philippa Hann took a short break from the office to prepare for the arrival of our youngest fundraiser, baby Agatha, who arrived in early summer. Never one to rest on her laurels, Philippa used the time to focus on fundraising plans for the year ahead.

On 4 July 2012 we held a

Pampered Chef event. Imagine if you will the Generation Game (but without any cuddly toys), Catherine MacMillan was our skilled professional from Pampered Chef and, one by one, the audience was asked to participate by attempting to do the same. There was chopping, cutting, peeling and preparing scrummy treats for the rest of us to enjoy. No aprons were involved but we managed to keep it (fairly) clean and without chopping off a finger! Funds were raised through orders of Pampered Chef products and a raffle.

In July 2012 we held the third Monopoly Challenge evening in Bristol. Fourteen teams took part in numerous challenges following a CW Monopoly board which took them to four pubs en route. Challenges included building an Olympic torch, lego mascots (see pictures on opposite page), general knowledge quiz, basketball and table football.

Teams were, as always, extremely generous (especially when it came to bribes), raising nearly £1,000 for our joint charities in just one day. Teams were very competitive, some tactically aware. During the basketball challenge some teams impressed with their ingenuity. Some chose to throw in a different approach to the traditional way, others tried to distract the scorer using their Olympic torch, buying them drinks and bribing with cash. Other teams stole opposing team's torches, stole the balls and even took the signs down.

We feel it is important to use the skills we have as a company to offer help and support to our charity partners, and it was with great pleasure that Liz Smithers, a partner specialising in Wills and Trusts, wrote an article for the summer edition of *A-T Society News*.

As we move towards the end of the first year of our charity partnership, we anticipate the staff of Clarke Willmott's Bristol Office will have contributed over £9,000 to the A-T Society. It's a sum we are all incredibly proud of. 2013 will see teams taking part in the Bristol Half Marathon, Triathlon and Dragon Boat Race as well as numerous other team events and individual fundraising endeavours. It feels really good to be helping children with A-T right now, and giving hope for the future.

The CW boys at the Bristol 10k run

Lawyer Philippa Hann

Media babe

When Jo Bloomer received her daughter Lola's diagnosis earlier this year her whole world turned upside down. Jo decided that the best way to cope was to tell all her Facebook friends and to actively raise awareness of A-T locally.

Within days, a number of friends had offered their help – both practically in terms of emotional support, and financially by making donations – and starting to fundraise for the A-T Society.

Jo was invited to do an interview with the *Plymouth Herald* newspaper and, subsequently, her moving article was repeated in the *Daily Express* and the *Daily Mail.*

One of the people who read the article was comedian Brian Conley who was performing in Plymouth at the time. Brian was so overwhelmed by Lola's story that he arranged to meet her and, having done so, pledged to perform in a special charity show with the cast of Oliver! raising money for the A-T Society.

The performance took place in early September at the Theatre Royal, Plymouth. As he opened the show, Brian announced his intention to become our new Patron and committed to do as much as he could to spread the word about A-T (you can read more about this on page 7).

The Herald, Monday September 3 2012

THX-E01-S2

Last chance for a seat at stars' show for Lola

by EDD MOORE
Chief Reporter

TIME is running out to snap up tickets to a star-studded one-off charity theatre show.

Actor Brian Conley, who is starring as Fagin in Oliver! at the Theatre Royal, was inspired to organise a special late-night show for little Lola Bloomer after seeing her story in The Herald.

The three-year-old, from Plympton, has been diagnosed with the rare and incurable condition of Ataxia-Telangiectasia, or A-T.

Lola is one of only 60 children in the UK living with the fatal genetic disorder, which causes increasingly severe disability and premature death.

TV and stage star Mr Conley said he "had to do something to help" after reading Lola's story in The Herald.

The dad-of-two joined forces with Theatre Royal staff to organise a special cabaret-style performance, set to hit the stage tomorrow evening.

Nearly 500 tickets had already been bought by the weekend – well over half of the 750 total.

Theatre Royal staff said that the box office phones had been "ringing off the hook" last week, after the tickets went on sale on Thursday.

Today is the final day people can get their hands on the remaining few.

Billed as 'A Charity Night with Brian Conley and Friends from Oliver!', the spectacle will raise money for the A-T Society, as well as the theatre's work with young people and communities.

The stars will be performing from 10.30pm, after the curtain has come down on the evening's performance of Oliver!.

Tickets for the charity cabaret night are priced at £20.

For more information, call the Theatre Royal box office on 01752 267222 or visit www.theatreroyal.com.

Around 30 kind-hearted staff members at the Derry's Cross venue have offered to give up their time for free to stage the show.

Mr Conley last week described the support the city was giving Lola as "incredible".

And the youngster's mum, Jo Bloomer, said she was "absolutely overwhelmed" by the response to Lola's plight.

"The A-T Society are over the moon too," she added. "I think other parents have been inspired."

Jo has been persuaded by Mr Conley to give a brief speech at tomorrow's show about her daughter and A-T.

More information about the condition is available at www.atsociety.org.uk.

Jo Bloomer's story shows how big little acorns can grow. She has fantastic support in her local community and there are a number of fundraising events planned for the years ahead.

Extraordinary people

Vaughn and Jo Rawson from Wrexham, North Wales are the kind of big-hearted friends no person should be without. In April last year they hosted the first A-T Spring Ball at Chester Racecourse. This spectacular night was a towering success and they lost no time in organising the 2013 event which now has a waiting list for tickets.

Not one to rest on his laurels, dynamic Vaughn wanted another challenge. Working in tandem with Expedition Wise of Chester, he is organising and taking part in a London to Paris cycle ride in June 2013: to raise funds for our joint research fund with Sparks and Action for A-T. Vaughn would love you to join the team and you can contact him via the A-T Society to reserve a place.

London to Paris cycle challenge

The three-day London to Paris challenge will see you cycling through the beautiful countryside of Surrey, Hampshire and Normandy. Rather than the normal Dover, Calais route, you will be taking the ferry from Portsmouth to Caen. You won't be short of spectacular sites, either, as you will pass through Hampton Court and finish up in the shadow of the Eiffel Tower.

The challenge starts on Friday 7 June and finishes on Sunday 9 in Paris. After an evening of celebration in Paris, you will return to London by Eurostar on Monday 10th.

You can find full details of the itinerary at www.atsociety.com/cycle-events. There are about 20 places available. Because of the cost of organising the trip, you will be asked to commit to raising a minimum of £1,700 for this adventure of a lifetime.

Pentagon Shopping Centre

A call out of the blue was the prelude to an exciting invitation to become the Charity of the Year for the Pentagon Shopping Centre in Chatham Kent.

The vibrant shopping mall has five major events each year and the management team are keen for the charity to be represented there as frequently as possible throughout the year.

We are asking our Kent families to each take on one day during 2014 to help us with fundraising at the Pentagon and we hope you will support us to make the most of this fantastic opportunity to raise money and awareness of A-T locally. Please contact Lian Yarlett to book a convenient date.

London Marathon Runners

Thanks to a determined campaign by A-T mum Tania Wheeler, we have been given three London Marathon places by Sparks for 2013, which along with our own silver bond place gives us four A-T Society runners. We would like to introduce you to:

Graham McIntyre, from our Charity of the Year partners, Clarke Willmott, Bristol, Ian Barnes, a solicitor from Wrexham, Vera Hall, also from Wrexham and finally James Sloan who works in Manchester.

Each runner has made a commitment to raise a minimum of £2000 for the A-T Society and we send them our very best wishes for their training and the race. If you would like to show your support and appreciation for our runners you can find their fundraising pages on Virgin Money Giving.

New fundraising video

The Paralympics opened minds to 'ability' rather than disability and the young people who took part in the Glee performance at the Family Weekend played their own small part in changing attitudes.

We felt it was important to record the event, and as digital media is the way forward in raising awareness and advertising the work of the charity and the amazing young people we work with, we were delighted to receive support from Simon Greene Associates and Brightspells to make this happen.

The outcome is two fabulous new videos, one is a 'commercial' length fundraising video and has already received more than 800 views on You Tube. The other longer film is a mini showreel created as a reminder of a wonderful day for the families. You can find both videos on You Tube.

Glenday and the Hiel' and Toe Club

Glenday Thomas has been a supporter of the A-T Society almost since the charity was formed. She's a passionate exponent of Scottish dancing and shares her enthusiasm with her students and friends in the local 'expat' Scot community.

Every other year, Glenday produces the show "A Measure of Scotch", a feast of Scottish heritage. The Alban Arena, St Albans resounds to the skirl of the pipes and the swish of tartan as singers, dancers and musicians come together for two performances of this remarkable and entertaining show.

2012 was described by the audience as 'the best ever' and Glenday spoke movingly of her experience of A-T and why raising awareness is so important to the charity. For the Society, Glenday's support and that of her husband Michael and friend Robert Hedger – who donates the profit from sales of his cards and prints – has been a cornerstone of fundraising for almost 20 years. Thank you for all the energy and devotion you put into supporting the A-T Society.

Spreading the word

Peter Brady is a postman in Manchester and is on a mission to see A-T wristbands worn by everyone he comes into contact with. Starting on a small scale in March, Peter sold 30 bands to his colleagues. At the time of writing, Peter had sold over 1,000 wristbands at local pubs, clubs and events and has no intention of stopping until Manchester is a sea of purple, pink and blue. Peter said: "Every band I sell means I'm giving children with A-T a chance, I'm not going to sit on my hands and do nothing if I can change things for the better".

If you are around Manchester and spot someone wearing an A-T wristband, please snap a picture and post it on our Facebook page.

If you want to "change things for the better" like Peter, please contact Suzanne for information.

Charity bar

Jen and Bob Owens ran a Charity bar at the Durberville Village Hall, Dorset on 11 November. It was a fantastic musical evening. Reg Foy of Crackerbang Entertainments set up a Neil Diamond Tribute Evening and Grandad David Owens was in charge of the teas and coffees. Through the generosity of the people of Wool, the family raised £227.

Please look out for our networking questionnaire online, if you complete it and send it back to us, it might be just what we need to attract some exciting new corporate support.

The kids are united

Chris Wooster – Harpenden

Chris explains “A group of friends and I at Sir John Lawes School in Harpenden were chosen to present an assembly to the whole of our year. We decided to do our assembly on the A-T Society.

One of the main reasons we decided to do this was to raise awareness for the charity as it is not very well known. During the assembly we gave an outline of the disease and how the A-T Society helps people with A-T, and their whole families. We also advertised ‘A Measure of Scotch’, a show which raises money for the charity and which I have been involved in since I was four years old.

After the assembly, we were approached by the school’s learning support team who wanted more information about the charity. As a consequence, our school is now supporting the A-T Society by donating a portion of money it collects on fundraising days such as ‘Jeans For Genes’ to the charity. Also, a lot of teachers came to the show, which helped support it and meant that once again it was a great success.”

Junior runners Jasper and George

Kaycee and Sadia

Kaycee Carrington and her friend Sadia Islam (pictured right) did a fundraiser at their local Asda store. You can follow Kaycee and Sadia’s example by contacting your local superstore and requesting a collection day.

Junior Great North Runners

Jasper Metcalfe and George Spink, aged 11, took part in this race for the first time and raised over £3,000 with their determined fundraising campaign. Thank you to everyone who sponsored the boys!

Magnificent Maguires

The letter which went out to fundraisers with the summer newsletter really caught the imagination of the Maguire family of North Wales. We had this email from mum Germaine whose cousins have A-T:

“The Maguire children have been busy thinking of things they can do to raise funds since reading your letter and some of the fantastic articles in the magazine.

Poppy (aged 12) decided that she would like to do a sponsored bike ride. She rode a very windy uphill 26 miles with her grandad and will be sending her sponsor money soon.

Lucy (aged 13) did a sponsored read. Lucy is severely dyslexic and so this was quite the challenge for her. She read four whole books over four weeks and wrote a review on each book. Lucy also held a cake sale at Rydal Penrhos School and raised a super £192!

Harry (aged 10) didn’t want to be left out and has decided to do a sponsored swim (following in his big brothers footsteps – Harvey!) We aim to fit this in over next couple of weeks giving Harry a chance to get some sponsors. Finally, Charlie has something up his sleeve and we will be in touch with that another time. The kids may only raise pennies but it is all money in the pot and it gets the word out there.”

We would like to say a huge thank you to the Maguires for being such an inspiring family. You can read their stories in full on the website.

Kaycee and Sadia

Poppy with her bike

Lucy did a sponsored read

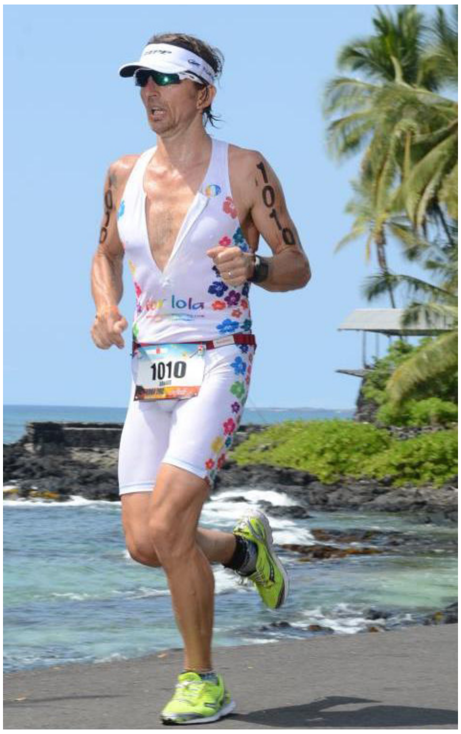
Mega –Tri for Lola Update

In April 2013, a team of 25 men and women coming from Singapore, France, Ireland, Australia, the Netherlands and the UK will embark on a mega-triathlon. The event will start at 6am on Saturday 13 April at the Life Centre swimming pool in Plymouth with a 2.5 mile swim. The team will then cycle 220 miles along the South Coast to Brighton, arriving in time to run the Brighton Marathon on Sunday 14 April. Team leader Kevin McGregor has set their fundraising target at £100K!

Kirsten Koh

But that's not all! Members of the team have been fundraising well ahead of the event. Arnaud Selukov, who is based in Singapore, raised sponsorship by participating in the Hawaii Ironman Triathlon World Championships He swam 2.4 miles, cycled 112 miles and ran 26.2 miles in an incredible 9 hours and 33 minutes, and achieved 11th in his age group.

Kirsten Koh lives in Tasmania and set herself a three-hour static cycle challenge. Kirsten was critically injured in 2011 when she was hit by a truck and, after numerous operations and a long period in ITU, is fighting her way back to fitness. You can read her incredible story at www.truecoloursfoundation.com/kirstens-story. Kirsten and her best friend Orla Gilmore are planning monthly events in preparation for the Mega-Tri.



Arnaud Selukov

How can you get involved? We want as many people as possible to turn out and cheer the Mega-Tri team along their cycle route along the south coast from Plymouth to Brighton. Precise route details will be available nearer the time. Please bring your families and friends out and give this phenomenal group of people your support. Contact Suzanne for further information.

Grainger PLC Altringcham

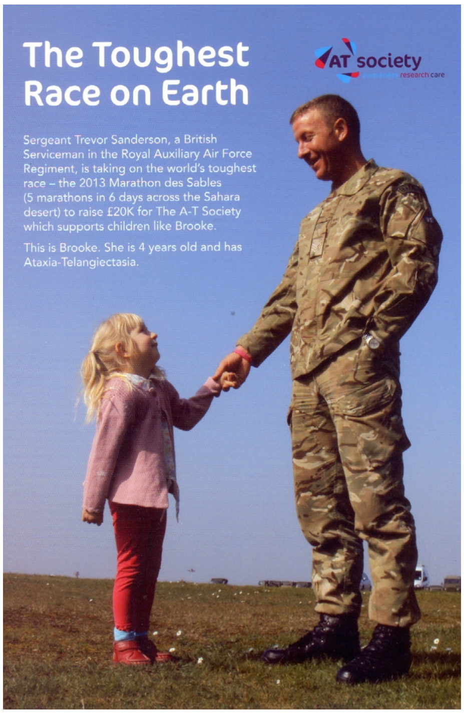
Members of staff from Grainger Plc in Altrincham completed the North West Relay Triathlon in September and raised £2,045 for the A-T Society.

The teams were made up of Phil Nelson, Simon Aldridge, Melissa Elliott, Emma Burdge, Louise Spooner and Claire Henry (who competed as two relay teams) and Mark Weeden (who very bravely undertook the challenge by himself).

(Right: the Grainger Plc team!)

The Marathon des Sables

Trevor Sanderson has transformed fundraising the Marathon des Sables into something close to a military campaign. With partner Hayley firmly on side, he has organised events, collections and sales opportunities every month since he booked his place. Trevor's target is £20K and he is looking for corporate and community sponsorship to make this happen. Trevor has asked us to thank everyone who met and sponsored him when he spoke at the Family Weekend, and looks forward to going beyond the call of duty for the A-T Society.



The Toughest Race on Earth

Sergeant Trevor Sanderson, a British Serviceman in the Royal Auxiliary Air Force Regiment, is taking on the world's toughest race – the 2013 Marathon des Sables (5 marathons in 6 days across the Sahara desert) to raise £20K for The A-T Society which supports children like Brooke.

This is Brooke. She is 4 years old and has Ataxia-Telangiectasia.

Fall Family Fortune

Helen and Tony Fall organised a Charity Dinner at the Pavillions, Harrogate and raised £2,000 as well as lots of local publicity for A-T.

Thank you Helen and Tony – and all who donated!

Running for A-T

Jordan Button of Nottingham jumped in at the last minute to take place in the British 10K London Run in July. The race had over 33,000 entrants who braved appalling weather to take part. Jordan, who works for Nobel Foods (creators of The Happy Egg Co), said: "Nobel Foods believes it is important for their staff to fundraise and raise awareness for charities. To encourage this, they very kindly match 50% of the employee's total fundraising to a maximum of £500. Because I was able to raise a brilliant £1,200, they contributed an additional £500, bringing my total up to £1,700. I would like to thank them massively for this donation, and for allowing me to share information about the A-T Society."

Action at Asda

Do you or your friends work for Asda, First Group or any other company which actively supports charities or has a charity of the year? Many companies ask their employees to choose the charities, and we would love you to help us get involved.

You can also nominate the A-T Society for the green token community fundraising at your local Asda. Simply fill in the form and hand it into Customer Services.

Collector's item

Gulsen Deren lives in Kent and is mum to Ahmet aged 16. She said: "The A-T Society has been so good to me ever since Ahmet's diagnosis. I don't work and my own health isn't very good, but I wanted more than anything to give something back. I talked to local shops, cafes and businesses about the charity and before I knew it they were all filling collection pots for me. The money is coming in, but the best thing is that more people know about A-T and want to support us. It's such a simple way of raising money and awareness and it feels great because I know I'm helping despite all the other challenges in my life.

Scotland's brave men

The Brownlie children were the motivation to get men on the run across Scotland this year. Grandad Martin Doherty took on the Great Scottish Half Marathon, while cousin Martin Cairney and friend James Miller did the Great Scottish 10K.

First fundraiser

Brian Sewell and Siobhan Kelly held their first fundraiser, raising more than £2,000 for the A-T Society. Hundreds of family and friends attended the event in Croydon.

Manchester United defender Rio Ferdinand donated a signed shirt along with Chelsea Football Club who sent a shirt signed by the team. Both items went into a charity auction.

A natural!

Sorrell Hodgson took up running just a few months ago..

She did a very respectable time in the Robin Hood half marathon and raised money for A-T!

Student helpers

Hertford Regional College provided the student helpers for the 2012 Family Weekend. The students were so excited about their work with us, that they chose the A-T Society as their charity for 2013. Tutor Denni Morrison says they have lots of events planned and she hopes this will be an enjoyable year for everyone involved.

Olympic saddle cloth donated

Nicola Wilson, a member of the Olympic Equestrian Eventing team, has donated a saddle cloth signed by many members of the medal winning team, including Zara Phillips.

The Saddle Cloth will be auctioned in Yorkshire.

Graham High – Sea Passages

Artist Graham High, whose stepson has A-T, recently held an exhibition at the Institute of Physics in London.

Entitled Sea Passages, the pictures were based on his love of the sea and landscapes of the North Norfolk countryside where he lives. Graham very generously donated a significant part of the price of the paintings he sold to the A-T Society, raising more than £1,400.

Film review: Untouchable

By Rupert

Untouchable is a film about the relationship between a carer and a caree. It explores the taboos associated with this topic, which is not very often represented in films.

At the beginning, one of the two main characters, who is disabled, lives in a care home, and the main problem of a care home is one is told what to do. I've been in college, where the relationship with my carers was really professional. In uni, there was more freedom. Now, I can finally choose which carers I want and my carers do not necessarily do the caring stuff only, but social stuff as well, which I prefer.

The second main character turns up as his carer, and he is the kind of person who does what he likes to do. One wishes that social services really worked like that, because there are carers who just do their job and nothing more than required. This fellow has no experience and therefore no preconceptions, so he tackles his role with a lot of energy from square one. The first character does not know what to expect, so he's going to have to find out for himself.

The disabled guy in the film has a big van he can put his wheelchair into, but he also a sports car. The carer tends to just carry him into the sports car, which is what he wants to do, rather than what he is *meant* to do.

The carer gives him the courage to meet in person a girl he has met on the internet. The carer has serious issues with his family and, when he leaves, the disabled guy is on his own again and understands that he is going to have to do things himself. Before, the disabled guy didn't know what was out there, but from their time together he realises he can be more active and live what they call a "normal life".

Untouchable was really interesting and insightful to me, being a disabled person myself. After seeing the film, I breathed a big sigh of relief, because in a way it represented quite well how I thought about my life and my carer life. It was an acknowledgement of disabled people's lives, shown as they are. This film could make a difference to disabled people's lives by promoting how carers could make life better for them. The impact of the carer gives the main character the drive to have fun, and the confidence to make it happen.

Even though the movie is in French and it can be hard to follow the subtitles, it is not necessary in order to comprehend the action. Think of silent movies and how they show instead of telling. This film also explains itself through the actions of its characters rather than weighty explanations.

Another interesting film on the topic is The Best of Men, about how they came up with the idea of the Paralympics.

Have you been inspired by a film, book or perhaps a concert? Why not send in a review!

Introducing Atey Bear!

We are delighted to introduce a new and furry member of the fundraising team. Atey Bear.

Atey Bear told us in his interview that while he is happy to do anything for A-T, even jump out of a plane, he's happiest standing outside shops with a bucket. He enjoys it because children will often come up and talk to him – which he really likes. He likes grown-ups too, but finds them a bit scary...

He says he particularly likes collecting outside supermarkets, because there's always a chance someone will come up to him and put a jar of honey or jam (he likes blackberry best) into his bucket.

You are likely to find Atey Bear at the Pentagon shopping centre in Chatham next year, where we are Charity of the Year. But we are hoping to persuade him to come along to the Family Day in Manchester, and perhaps we might see him at other A-T fundraising events too.