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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.

Contents

Editor's Comments

Once again, many thanks to all contributors. The copy date for the next issue is 1st April 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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Cover picture: Joe Bromwich

We are very sorry to report the death in September of Victoria Holliday.
We send our condolences to her family and friends.

News from the Society

The next A-T international research workshop will take place in Japan in April 2008. This is a major event that will drive the research effort forward. Once again the Society will be sponsoring the workshop as well as providing travel grants to enable several UK researchers and clinicians to attend.

We were delighted to collaborate with the A-T Children's Project with the "Eye Abnormality in A-T" Study which took place in Zurich (see page 10) and we thank those participants and their carers who took part.

We have already had one meeting with Ataxia UK looking forward to Ataxia Awareness Day 2008, and the next meeting is planned for January. We hope that together we can build on past success to increase awareness and funds substantially in 2008. Thanks to all those who supported this year's event.

We are presently reviewing our literature. Several leaflets have now been brought up to date and are with the printers. This process would not have been possible without the help of Professor Taylor and the A-T clinic team at Nottingham City Hospital to whom we extend our grateful thanks. Next year we also hope to update our website.

As regular readers of this newsletter you will be aware that many people with A-T enjoy physical activities. In this edition alone you will read about Joe who delights in sailing, Morgaina who gets her kicks through abseiling and Dale who loves jumping out of aircraft. We decided that our annual get together for those over the age of 16 with A-T, should reflect this more keenly. In order to make this possible, we have moved the event to Milton Keynes as it provides the opportunity to try some wonderful activities such as indoor skiing and a sky-diving type experience at Airkix, in a much more accessible environment. We think this is going to be a great weekend and are pleased that so many have already signed up. See back page for more details.

As always, a huge thank you to all our supporters. We could not do any of this without you. I have to say I am lost in admiration for the Hell Runners (see page 16). All that mud - yuck! And they did it for A-T.

Thanks to all.

David Owens,
Chairman

Ataxia-telangiectasia mutated (ATM) gene and lymphoid malignancies in the general population

Dr Tatjana Stankovic

CR-UK Institute for Cancer Studies
University of Birmingham

One of the most important forms of damage to our DNA is a break across the double helix – a double strand break (DSB). Such damage has to be resolved in some manner by the cell. It cannot be ignored and it is the role of the ATM protein to co-ordinate the cellular response to this form of DNA damage.

Therefore, in response to DSBs, the ATM protein modifies different proteins in the cell by adding a phosphate group. There are different protein targets for this modification but the endpoint is that they cause the cell to stop going through the process of dividing. If the level of DNA damage is manageable, the ATM protein supports repair of the DNA damage. In contrast, if the level of DNA damage is above a certain level, ATM induces the death of the cell by activation of another DNA damage response protein.

This induction of cell death is an important phenomenon and is called 'programmed cell death'.

This function of the ATM protein is important for probably all tissues. It may, however, have a special role in lymphoid tissues. Importantly, in the absence of ATM protein DNA damaged cells do not die by 'programmed cell death'. Therefore, cells with damaged DNA (a mutation) survive and the presence of the mutation can lead potentially to the development of a cancer cell.

Ataxia-telangiectasia (A-T) patients are born with inherited mutations in both of their *ATM* genes and develop a range of lymphoid malignancies in both types of lymphocytes, the so-called B and T lymphocytes, probably because these cells survive with genetic damage.

It was anticipated that if the *ATM* gene was mutated in a

The ATM protein and the ATM gene

People with A-T have two faulty *ATM* genes.

Classic A-T is the most severe form of the disease. People with classic A-T are unable to produce the functional ATM protein which is needed for repair of DNA damage.

Less severe forms of A-T are where the *ATM* gene produces protein with some residual function.

Carriers of A-T have one faulty and one normal copy of the *ATM* gene. Although they don't have A-T, they produce less ATM protein than the general population.

In this article, the name of the gene is always written in italics, *ATM*, whereas the name of the protein is always written in regular style, ATM.

lymphoid cell of a member of the general population this cell might give rise to a similar type of tumour as seen in A-T patients. Indeed, *ATM* mutations have been found in a range of lymphoid tumours, including B cell chronic lymphocytic leukaemia, mantle cell lymphoma and T-cell prolymphocytic leukaemia, in the general population.

In recent years we have particularly focused on B–cell chronic lymphocytic leukaemia (CLL), the most common leukaemia in the western world. This leukaemia is remarkable for the extreme variability of its clinical progression. The disease may remain stable for extended periods in some patients whilst in others progression is rapid and requires early treatment. For this reason the molecular mechanisms that underlie the biological variation of CLL are currently an area of intensive research.

We have recently observed that mutations in the *ATM* gene are detected in approximately 12% of CLL tumours in the general population with a prolonged clinical course and in 40% of tumours with a rapid clinical course. This frequency establishes loss of ATM function as the most frequent single gene defect in CLL in the general population.

Importantly, the presence of *ATM* mutations changes dramatically the behaviour of CLL tumour cells. For example, we found that compared with CLL tumours that do not carry *ATM* mutations, *ATM* mutant CLL cells are defective in programmed cell death induced either by ionizing radiation (IR) or drugs currently used in treatment of CLL such as fludarabine, cyclophosphamide and chlorambucil. These leukaemic cells are also unable to efficiently repair IR-induced chromosome breaks.

Consequently, the combination of a failure to undergo programmed cell death and the repair defects leads to impaired tumour killing by standard treatment and to the propagation of cells that carry unrepaired DNA damage. Thus, *ATM* mutant leukaemic cells show the same defect in response to DNA damage as the cells from patients with ataxia-telangiectasia.

We were particularly interested to determine whether these specific characteristics of *ATM* mutant CLL tumours in the general population have an impact on patient survival. Indeed, when we compared survival rates of CLL patients with and without *ATM* mutations we were able to observe that the presence of *ATM* mutation reduces overall survival, not only among milder CLLs but also among patients with more aggressive CLL.

In other words, we concluded that the prognosis of the aggressive form of CLL was even worse if the patient had acquired an *ATM* mutation in the tumour cells.

It became obvious to researchers and CLL clinicians

that CLL tumours with *ATM* mutations in the general population have to be treated in an unconventional way. Recently, a range of anti-cancer compounds have been developed that do not rely on activation of programmed cell death. Many of these compounds have been tested in CLL cells, with different degrees of success. However, the toxicity of these compounds on normal, non-tumour cells remains a major obstacle.

In a search for an effective and non-toxic treatment for *ATM* mutant CLL in the general population we have turned our attention towards the strategies that can provide specific killing of *ATM* mutant tumour cells. We were encouraged by recent reports of successful tumour specific killing of BRCA mutant breast cancer cells that are defective in DNA repair. Treating the patient with a particular drug leads to the generation of DNA DSBs only in the tumour cells and not in non- tumour cells. If the tumour cells have a defective ability to repair DNA DSB repair, which is the case for BRCA mutant cells, then accumulation of unrepaired DNA damage leads to death of the tumour cells whereas non-tumour cells with efficient DSB repair proteins remain intact.

We reasoned that *ATM* mutant tumours might be good candidates for a similar strategy. CLL tumour cells lacking ATM function are also deficient in repair of DNA DSBs and we have hypothesised that treating CLLs lacking ATM function with the same drug is likely to lead to tumour cell death in the same way. This approach would overcome the defect in activation of programmed cell death in *ATM* mutant CLL cells. We are investigating this strategy in the hope that we can eventually improve the treatment of high risk CLL in the general population.

Open Forum

The Open Forum at the Family Day in May offered people with A-T and their families a chance to put questions to a panel of experts. In the June issue of A-T Society News we offered a summary of some of the issues discussed and the remaining questions are covered here.

written by Beatrice Prokofiev

Do children with A-T have problems with puberty that other children don't? i.e. do they develop any differently or at different rates? Are there any problems that we as parents should look out for?

That's something we probably haven't looked at specifically, at clinics, because we've obviously got all the other things to look at, like neurology and the therapy side and everything else. But it is an area that I think we probably should look at, and we would need an endocrinologist (hormone specialist) to help. It's known that there is delay in puberty and quite often girls have irregular periods or not many periods. Others have very heavy periods so it does vary. But I'm not really aware of any studies that look at this particular aspect. Parents or the young people themselves, once they get through puberty, might be the best people to ask about that.

If growth is delayed it may be to do with nutrition rather than A-T.

Are there any exercises, aids or equipment aimed at helping young people to maintain quality and strength of lung function?

A physiotherapist will recommend specific exercises geared to the individual, but it's possible to give some general tips. Swimming is often recommended as is any activity which involves reaching, stretching and that will help exercise the chest, the upper part of the body, for instance, singing, dancing, kite flying! As far as equipment goes, we'd suggest blowers, bubbles, whistles, kazoos, these sorts of things that can be part of play time as well as a good activity for helping lung function.

Equipment ranges from a simple device to show the power of expiration, to a complex device called a cough assist which requires professional supervision. If a patient has weak clearance of phlegm then you use this machine - which requires a mask - to try and turbo the cough. And if someone's breathing muscles were very weak it might be even necessary to consider night time ventilation to assist lung function.

At the Papworth A-T clinic they are investigating lung function in adults with A-T. Looking a couple of years ahead I would expect them to say what they've found in that age group, and then recommend what should be adopted for young children.

The Guide to Therapies highlights the value of walking with assistance in order to minimise foot, knee and hip flexion problems and to promote circulation. Is there any equipment which will allow young people to maintain an upright position, while using their legs when possible for movement? One sees advertised wheelchairs allowing standing positions but these do not allow the use of legs.

This is really a question about walking frames because other than providing extra stability in walking, there's not a lot else you can do. There's a huge variety of walking frames on the market. A physiotherapist should be able to assess you for the one that's appropriate to your needs. It's totally dependant on the way you move, or what you're going to do when you go off balance because the walking frame has to be able to cope with that. So if you're going to have an unstable pattern of walking, the frame has to be stable enough not to go with you, but it also has to not stop you having freedom of movement. Some walkers have four wheels on to give maximum freedom of movement but then you're reliant on hand brakes if you are going to go down slopes. This can be a problem for children with A-T because of the strength needed to pull brakes on. But there are a lot of other walkers that have two wheels and two bungs so you've basically got a natural brake as soon as you go off balance. It really is a question of trying different sorts and finding the one that works for you in the way that you move.

Obviously you can't use your hands when you're using a walking frame, so there are times when it's good to use one and times when maybe it's a bit harder as well – planning it into part of the day when it's not going to be an inconvenience.

A lot of people get by, especially indoors, by using bits of furniture and walls and things. Whatever works is great, and whatever keeps you on your feet for as long as possible really, and isn't going to get in the way.

Papworth Clinic

Papworth Hospital now has a specialist clinic for adults with A-T.

Here Rupert Prokofiev and Graham High describe their experience of attending the clinic.

Rupert writes:

I was Papworth clinic's first visitor with A-T. There were a lot of tests and examinations but they weren't as bad as I thought they would be, and although there was a bit of waiting about I was pleased to have a room and bathroom of my own where I and my parent could both sleep. In between tests during the day we did a crossword, or went to visit the grounds. The food was good too.

I was glad that through all the examinations all questions were addressed to me. Although Mum was there for company she was not involved and as far as the Doctors were concerned they were only interested in me and what I had to say. All the staff were very friendly and relaxed.

I learnt how I could help maintain my own health

On the return visit I really enjoyed the consultations and learnt things about how I could help maintain my own health better.

Before I went I was anxious about how much my immune system might not work very well. I was shown the results of all the tests and was pleased to see that I scored quite high in certain areas and was able to see what are the areas I should watch out for.

Now I know what my strengths and weaknesses are

I got a kick out of realising how healthy I was. Now I know much more what my strengths and weaknesses are and how to deal with things like colds or other infections I might get. Various other things were suggested like a liquid thickener to help swallowing which I use all the time now.

I also feel good about the fact that I don't need to go back to Papworth for a year and that they know me there now and would be able to spot anything that might be of concern.

As Dr Exley said, "It's all to play for" in my life now.

I got a kick out of realising how healthy I was

A Parent's Impressions

Graham writes:

When Rupert went to Papworth Hospital to undergo tests there were, of course, a few anxieties and reservations in my mind. He was to be monitored as to his condition of health in relation to effects known to be associated with Ataxia-Telangiectasia. Would it be an unpleasant experience? Would it result in depressing news? Would it offer false hopes? Would it have been better to let sleeping dogs lie?

Having arrived at the hospital for the weekend of examination there was the immediate doubt in my mind that the visit would all be for the Doctor's benefit of research and not of direct benefit to Rupert.

Nothing could have been further from the truth. Rupert was welcomed, not simply as a source of data, but was comprehensively taken under the care of the Hospital which, with its specialist nature, is able to provide services with an understanding not available anywhere else. They now have a more detailed, joined-up-thinking, portrait of Rupert and his individual case than exists anywhere else.

In the future, through the on-going attention of (in his case) annual visits his health will be checked up on by the best Doctors in the field. This way, any possible complication stands the very best chance of being averted. Unlike the rest of us, who may have underlying conditions not known about, adults with A-T are known to have certain health pre-dispositions. For these to be looked at and diagnosed carefully by experts in the field is a great weapon in the endeavour to keep as healthy as possible.

The thorough check-over included, besides the medical tests, visits by a physiotherapist, occupational therapist, dietitian, and speech and language specialist. Subsequent contact was also made with everyone concerned with Rupert's case (local and college GPs, social workers and care officers) so that all concerned should benefit from Papworth's overall knowledge and liaison. From now on everyone should 'sing from the same song-sheet' regarding Rupert. If one were to ask for such a check up as a private paying patient it would, of course, be very expensive, possibly prohibitively so. As it is the whole thing is NHS funded.

Rupert didn't find the tests themselves (blood tests, breathing and diaphragm tests, Video Fluoroscopy, X-ray etc) as unpleasant as he might have feared. They were certainly exhaustive but handled very efficiently and pleasantly – some, like the blowing tests, were even fun.

Within this wide ranging check up, more specific tests were addressed to known areas of concern with A-T patients. Where it is already known that A-T patients may have a predisposition towards difficulties with the immune system and certain respiratory conditions, it was a great relief to Rupert to be told more specifically where his strengths and weaknesses lay in these respects; to know that his on-going stability was going to be monitored by the staff at Papworth and that any changes could quickly be followed by action that would make it much more likely that he will be able to maintain his normal health level.

Papworth itself is a nice place with very attentive and caring staff all of whom seemed to have plenty of time to really get to know Rupert. It was a very different hospital experience from that at big general hospitals with too many patients and over-worked staff. I am re-assured to know that Papworth has an international reputation as a cutting-edge

If one were to ask for such a check up as a private paying patient, it would be very expensive

continued on p10

research hospital.

The two specialists that are keen to engage with people with A-T are Dr Shneerson, (whose speciality is respiratory complications), and Dr Exley, (An immunologist with special concern in genetic factors). At the second visit these two gave detailed and open feedback following the results of all the tests and were concerned, not only with how to combat the condition of A-T in the future, but also to put their best skills and advice at the service of those patients who, like Rupert, were now under their watching care.

Dr Andrew Exley, Consultant Immunologist at Papworth

Eye Abnormality Study

Last May five people from the UK and Ireland travelled to Zurich to participate in a study on eye abnormality in A-T.

Earlier this year the Society was contacted by the A-T Children's Project in the States asking if we could find five people with A-T willing to travel to Zurich to participate in a study investigating the eye abnormalities seen in A-T.

Why Zurich?
The University of Zurich is the only place in the world which has a three axis motor driven turntable coupled with eye movement recording facilities. Basically this is a large machine containing a chair. Participants sit inside and are rotated in various directions while their eye movements are tracked.

What was the purpose of the study?
The purpose of the study was to undertake a quantitative study of eye movements seen in A-T. The participants had already taken part in a previous MRI study at Sheffield University. The hope is that the neuro-imaging results from Sheffield can be linked to the eye movement findings from Zurich.

Why is this important?
If the eye movements can be quantified in this way then it will provide a basis for measuring responses to any future treatment offered to A-T patients. The response to a drug could be measured by its effect on the eye movements.

Who undertook this project?
It was undertaken by Dr Shaikh (Johns Hopkins University Medical School, Baltimore, USA) and Professor Straumann, University of Zurich.

Participants were jointly funded to attend by the A-T Society and the A-T Children's Project.

A big thank you to our five participants for taking part in this important study which could yield very valuable information of benefit to all people with A-T.

And a big thank you to all the Society's medical advisers who helped to make this project possible at such short notice.

Maureen Poupard

Youth Meeting

Kay Atkins writes: In March this year 13 people with A-T and their families met up in London for a consultation meeting and social event.

We had a full agenda which included a discussion about the Papworth Clinic, where people could ask questions of those who had already attended. After lunch there was a computer workshop run by the Aidis Trust and a feedback session with the Trustees of the Society.

After a rest and something to eat everyone headed off to the West End to see a show. Some went to see 'Mama Mia' and others to 'Billy Elliot'. Everyone had a great night out. This was rounded off by a late night in the hotel bar!

Carers had some time to relax and chat with the Society's Trustees

Tea - partying with a Star!

Shaun Williamson lends his support

by Lauren Bell

Actor Shaun Williamson, better known as the former character 'Barry' from Eastenders, was a huge hit with family and friends when he dropped in for tea and a chat at Lian's coffee morning. Lian, who is a trustee of the A-T Society, decided to hold the event to raise funds and awareness of A-T for Ataxia Awareness Day.

Shaun entertained guests with his wit and good humour and said, 'It is lovely to see so many people supporting such a worthwhile cause'. After drawing the raffle, he offered to help out at any future events held by the Society. Lian raised an overwhelming £2,000.

The Society is very grateful to all those who held coffee mornings and other events on Ataxia Awareness Day this year. Thanks too to all those who gave their support.

Making strides for A-T

Lauren Bell writes:

More than 25 ladies and girls stepped out in the Hydro-Active Women's Challenge in September this year, in aid of the A-T Society.

This event has become very popular, attracting over 30,000 participants in London, Birmingham and Liverpool. It appeals to all, from elitist runners to those who just fancy a relaxing stroll to raise money.

Runners, joggers and walkers alike, all had their eyes on the same goal, the 5K finish line - the key to unleashing all the money - and what an inspiration it was to watch!

The swarm of ladies, all with the same aim and motivation, taking part in memory of a loved one or a cause very close to their heart, was a very moving sight to see.

Our participants all enjoyed the day, some having entered last year and most already wishing to participate again next year. This is very promising and so important for such a small charity.

Emily, who has A-T, had her family and friends behind her and they completed the course in an hour and one minute. Emily's mum, Nicola said, 'You all get together and feel as though you are achieving something - everyone's there for a good cause'.

Sheila, Emily and Maggie at Hyde Park

If you would like to join us in 2008, please contact Angie on 01582 760733 for more information.

Wedding gift

Elizabeth and Pat Delaney at their wedding in Summer 2007. Instead of wedding gifts the couple requested donations be made to the A-T Society. Over £1,700 was collected! A very big thank you to the happy couple.

The A-T Society has a new email address...

atsociety@btconnect.com

Easyfundraising

There's a new and easy way for you to help raise funds for the A-T Society – simply by shopping online!

Easyfundraising is a shopping directory that features some of your favourite 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!

Everyclick

Raise money for the A-T Society just by searching the web. 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 or shop online.

Please give it a go and pass the message on!

Dancers

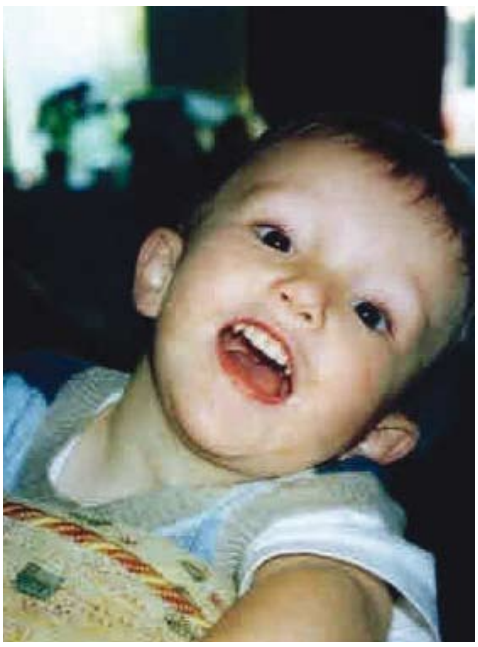
Young pupils from the Heil 'n' Toe Scottish Dancing Club in Harpenden present a magnificent cheque for £3,300 to the Society. This was raised through the Club's show "A Measure of Scotch". Very many thanks to all the dancers and in particular the organisers, Glenday and Michael Thomas.

Raising funds in Ireland

Pauline Lynn organised a coffee morning in October. In the picture (right) Pauline's daughter Sophie holds up one of the many teatowels sold that day to raise money for the Society. Other family members have been working hard to raise money too: John and Gareth (Sophie's uncle, below) reached the heights when they climbed the four tallest mountains in the four provinces of Ireland, raising £706.

John and Gareth

We'd like to say a big thank you to Jo and friends who have been energetically fundraising for the A-T Society over the last few months. Amongst other things Jo and friends have been collecting old mobile phones and inkjet cartridges at the local primary school (Hempshill Hall Primary) and Jo has also organised a raffle at the local supermarket of which the grand prize is a Christmas hamper. Well done to all those who have supported Jo.



Jo's son, Kaid, recently diagnosed with A-T

The Hell Run

Paul Dorrell doesn't even like running but after hearing about A-T from a work colleague he decided to do something to raise cash and awareness for the A-T Society. Being a family man he also persuaded his brother Tony to join him.

Sunday 4th November dawned a beautiful day and Paul and Tony joined 2500 other lunatics in the notorious Hell Run at the Longmoor Army Camp in Hampshire. The Hell Run is 12 miles over multi-terrain and varying underfoot conditions. It is steep (very, very steep) rough, slippery and wet.

The highlight for the spectators has to be The Bogs of Doom at the midway point. It's a very long gully, waist deep in foul smelling muddy water. The first few runners are lucky but the more runners who go through, the muddier and more slippery it becomes. Paul and Tony both managed to stay on their feet but many didn't fare so well and were covered from head to toe in black stinking goo. After that they only had to run another 6 miles!

Paul and Tony completed the course in 2 hours 5 minutes and our most grateful thanks to them both for not only subjecting themselves to the Hell Run but for the sponsorship money they raised.

Well done, guys and thank you.

Joan Bridger

Need a gadget?

If you've ever had a problem that could have a mechanical solution, but you can't find the gadget to fit the need, try REMAP.

Remap is a national charity that has, for 40 years, provided one-off technical aids which help disabled people of all ages to enjoy a better lifestyle. Every aid is given free of charge to the user. Remap operates through a network of 1,500 volunteers. The professional engineers, technicians, and craftspeople - along with medical and paramedical staff from community services and hospitals - all belong to approximately 100 panels (groups) linked to regions across the UK, with Scotland and the Isle of Man operating separately.

I spoke to Roy Wrenn who is on the Taunton panel of Remap. He told me about one of the devices he's made:

A lady who has a problem with her eyes, finds it impossible to keep her eyes open in bright sunshine, although her sight is good. She wanted something to enable her to go out into town whatever the weather. Roy painted the inside of an umbrella with matt black paint, and mounted it on her wheelchair, making sure that she would be able to angle the umbrella in such a way that it shades her from the sun. Roy says he likes to work with the client and their Occupational Therapist (OT), as the OT can provide helpful advice. In this case the OT was able to advise on how to position the umbrella so that carers can still help the client in and out of her wheelchair without difficulty.

Roy says that Remap can make aids for any application. It might be something you need for mobility or practical tasks, or just for pleasure – like the revolving table he devised for partially sighted scrabble players.

Remap likes to take referrals from OTs but they'll take self referrals too. There's no charge for the work but clients often make a donation, especially if they're pleased with the outcome!

Look in your local phone book to find the nearest Remap group or look on the website:

www.remap.org.uk

0845 130 0456

Jo Child

See Robert Soper's article on pages 20 to 23 for more on adaptations and special equipment

Sailing

Joe Bromwich writes:

I thought it was about time to bore you all with my tales of sailing again.

Since I last wrote we have left the inland waterways and ventured into the seven seas (well only one really.) We have been out twice with Nick from Boatability on a 90Hp Dory which is suitable to be driven by a person in a wheelchair. The first time we stayed local to Port Solent (Portsmouth) and had some pictures taken, whilst being chased by another boat with the camera man on board. The next time was when I got my official speedboat licence, an RYA level 1 Powerboat certificate. We went out to the Mary Rose Buoy in the Solent, and on the way back I got soaked from the wind blowing the waves in my face. I managed to take the boat through the lock and moored it myself, with help for the rope tying.

We have also been out with Keith from the Ro Ro Project four times over the past two years, also out of Port Solent. They have two yachts: a single hull called Verity K, and a Catamaran called The Spirit of Scott Bader. Both have excellent wheelchair access and electric lifts to take you ‘down below’. On the last trip we crossed a submarine barrier and saw three dolphins (everybody else did but I was at

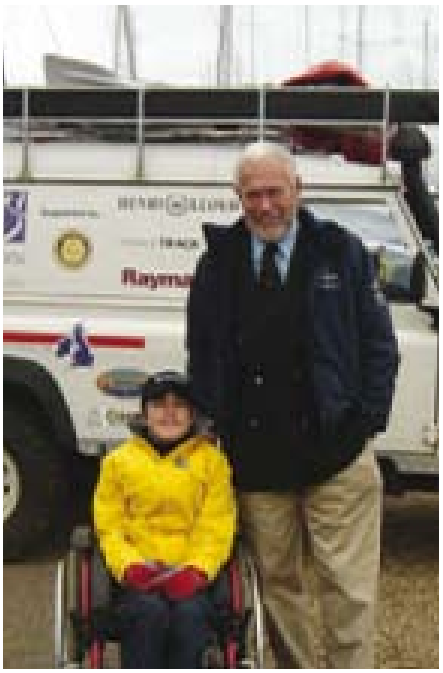
the helm and told to keep looking forward so I missed them).

I still go sailing on Saturdays at my local Sailability Club who all came out with us last time on the Catamaran.

We are looking to use one of the Ro Ro project's 16ft Martin dinghies which is kept at one of the pools in the Cotswold Water Park, but I think this will have to wait till next year due to the winter coming.

I have included some photos for you to see. In my last report I said look out Sir Robin Knox-Johnson. Well I met him recently at the Royal Southern Yacht Club. We had gone to see off Geoff Holt on his solo round Britain trip. (By the way, Geoff is in a wheelchair.) Sir Robin had just completed his second solo circumnavigation of the world. I would love to do that one day myself, but probably not in my little access boat!

Keep you updated - Joe Bromwich



Joe with Sir Robin Knox-Johnson

Useful Websites

www.disabledsailing.org
Ro Ro Project: a charity for disabled sailing.

www.boatability.co.uk
A company which offers reasonably priced boat charter and training courses.

Adaptations to the home

Robert Soper lives in a bungalow that has been specially adapted for him. Here he shares his thoughts about getting adaptations to your home.

Trying to predict what path a disability will take in the future is difficult, especially with A-T.

I think that I am probably amongst the oldest people with A-T, and nobody has been able to successfully predict what is going to happen to me in the future. Each person with A-T is different and A-T progresses at different rates in different people. There are other things to consider too: the way the people around you react to your disability will affect the way your life will go in the future, and so will your own attitude towards it.

In my experience of getting adaptations in the home, I wonder sometimes if the people doing the adaptations realise how important they are and that you actually need them.

Because of the progressive nature of A-T it is often a race between the rate at which the disability is progressing and the time that it takes to do the adaptation. I do feel that the process could be speeded up considerably if the contractors would ask you and/or your carer what's needed instead of just ignoring what everybody is telling them and doing the job that they think they should do. The jobs that have been done over the past twelve years while I have been living independently are a case in point; jobs that should have taken one visit have ended up taking many visits, wasting both time and money.

"I had to enter the bathroom backwards to use the toilet, then come out, turn around and go in forwards to wash my hands"

When I moved out of my family home I was housed "temporarily" in a converted flat. This was adequate initially, but became unsuitable. I ended up living there for 10 years. During this time I struggled to get the correct toilet fixture, the shower room was not originally adapted correctly and the passageway and entrances into bedroom and bathroom were too narrow. As a result I had to enter the bathroom backwards to use the toilet, then come out, turn around and go in forwards to wash my hands and complete my ablutions.

From there I moved into the bungalow I'm in now. I have lived here for more than two years now and I am very happy, I have two ceiling hoists, a Closomat toilet, and a telephone which I can use. I feel that if I could have moved to the bungalow earlier, things might have taken a different turn.

I am a great believer in not dwelling too much on the past; we live now and have to make plans for the future. I have come to rely more and more on my mechanical adaptations, but I also rely on the people who are around me for help and support, my family and my carers.

Here are some of my thoughts about adaptations:

Adaptations are to enable the user and afford an improved quality of life.

Consultation

In order for an adaptation to be really successful the consultation process is critical. All too often this has not been considered and this results in inappropriate adaptations and/or reluctance on the part of the user to use the adaptation fully. If consultation is not done properly, the user feels alienated, feeling that “no one listens to me”. The adaptation may be seen as more mentally disabling than enabling.

This contributes to the stress levels of the user, which compounds their situation instead of relieving it. Stress and frustration are two facets of life of the user to which insufficient attention is paid, while the “immediate” physical enablement is attempted.

Often the professional feels they have the correct piece of equipment or adaptation through

" After consultation, the adaptation should be completed promptly"

prior experience; however with proactive consultation it may very well be that another piece of apparatus would be ideal, and would prevent a waste of both time and money. More importantly, the user will be part of the process and will have ownership of the solution.

It is important the consultation process should be short, not protracted.

Looking ahead

Another area which is overlooked is planning the adaptations for the future, rather than completing them piecemeal as the patient situation declines. This too realises itself in the patient becoming stressed, and their situation becoming increasingly frustrated. Too often, stopgap measures have been employed and this becomes expensive. The equipment may end up being abandoned and its full worth or potential never being achieved.

Implementation

After consultation, the adaptation should be completed promptly. Too often the impetus of the project is lost in an inordinately long paper chain, blurred areas of responsibility and decision making.

It took over a year for me to get a Closomat toilet. All the paperwork had been done but the paper trail was incredible. Kent Social Services did their bit, Highweald Housing had done their bit, as had the Occupational Therapist, and when each one was asked what was happening we were told that they had done what was required of them and the documents were forwarded to the relevant party. The only problem was the wrong party had the documents. Once Maidstone Borough Council had the completed documentation the toilet was installed two months later. This situation will be familiar to many of you.

Now for the physical requirements:-

Premises

As this is a capital intensive venture, a long term view needs to be taken, for two reasons:

- the cost of the equipment is likely to increase over time, and
- the physical condition of the user is likely to deteriorate.

The paramount rule is to create a stress-reduced environment, not only for the user but for the carer too, as the carer's stress is often passed on to the user - albeit unintentionally. The user too will transfer stress on to the carer.

Accommodation consists of a main room and a side room. The main room would be the living space and bed room. The side room would be modified to become the wet room with shower, hand basin and toilet. Ideally this room should have an external wall, to improve the plumbing access and reduce costs.

The idea is to run a ceiling hoist above the bed from left to right with sufficient space to park the wheel chair next to the bed with the charging point behind the wheel chair. The hoist would run from over the bed, through on a straight line to the wet room with the toilet first, then hand basin and lastly the shower or wet room. Even if there no current need for the rail to go right through, it will still pay dividends to have a straight connection into the wet room.

Space is the first consideration. The idea is to be able to lift the client onto the toilet easily and without fuss. Initially the user may want to manually transfer from chair to toilet so sufficient space for the chair is necessary.

One area of importance here is when there are difficulties in toileting, insufficient fluid intake may occur, the result of which may be renal problems. Privacy issues may also be brought to the fore and these too may impair the physical health of the user. Automatic toilets are easy to use and improve hygiene, offering better quality of life. They are very desirable despite the cost.

The use of a shower cubicle also adds to the ease of bathing. Bearing in mind the run off from the shower, simple pull curtains work best around a gently sloped partitioned cubicle.

Door frames need to be wider, door handles lower; grab rails “L” shaped (to give vertical and horizontal support).

Plans must take into account which side of the wheelchair the user dismounts (this depends on where the electrical control is) and the turning circle of the chair.

Support Services

One other area that has a critical bearing is the quality of the Support Services, e.g. Social Services, Occupational Therapists, and service providers (heating, plumbing etc.)

Often the user lives, or aspires to live, on their own. Once that happens then the reliance on the parties listed above increases.

Their competence and attitude is crucial and unfortunately often leaves a lot to be desired! One doesn’t want to be overly critical, but when one has nationwide guidelines why does the service vary so much?

Equipment

Electric Wheelchair
Seating
Telephone and Communication equipment
Computer Equipment
Overhead Hoists
Closomat Toilet
Shower
Taps on Bathroom Basins
L shaped Grab Rails
Drop down rails
Suitable Cupboards for Clothing

Do you or your family have a similar or a very different experience of getting special equipment or adaptations to your home? What practical advice would you give to others planning to make adaptations?

The Society would like to hear from you. This information could be useful for other families.

Please contact Kay in the office on 01582 760733.

holidays

An interview with Dale Phillips

Dale Phillips has written in A-T News about his holidays in Southport and Spain, and readers of the newsletter may also remember his parachute jump in 2003. Dale, who lives with his parents and his brother Travis (who also has A-T), describes himself as outgoing – he likes holidays that involve meeting other people.

This year Dale went back to Southport and to Las Piedras in Southern Spain.

Who did you go with?

I went with Eddie, my friend who lives in Ipswich. I've known Eddie for a few years now – I can't remember exactly when we first met but I think it was at Sandpipers, the holiday centre in Southport that I go to. We get on very well.

What did you do in Southport?

The first day of my holiday was spent chatting and relaxing round the bar. The second day, I went into Southport with my brother for some lunch. In the evening there was some entertainment. The next day we went tenpin bowling and walked along the promenade. On Tuesday, my brother and I went with our two carers to the cinema and walked along the sea front. On Wednesday we went to town for a pub lunch. On Thursday after a hearty breakfast we headed by coach to the historic city of Chester. We had a stroll around the town to see the sights - including a Church which is 900 years old. The final day of the holiday I spent in Southport. I really enjoyed my week at Sandpipers - plenty to do and a whole lot of fun!

What did you do in Spain?

The owners of the villa picked us up at the airport. Over the week they took us on four trips – the other days we stayed at the villa. The villa is specially adapted with wide doors, ramps, and a chair hoist to help you get in the pool.

Tell us about one of the trips.

We went to a fish restaurant on the sea front. It was a small place, very noisy – lots of people shouting in Spanish! When you wanted to order anything you had to put your hand up to catch their attention. I had prawns, sardines and perch. It was very nice.

Where next?

I'll definitely go back to Southport to stay at Sandpipers. For a European holiday I'd like to somewhere different next time – maybe to Amsterdam.

I also go away for two weeks at a time for respite. My brother and I go to different places as we have different tastes. I like mixing with other people. There's a couple of places I regularly go to, one quite near where we live and the other near Oldham – I'm going there soon.

Tell us what else you enjoy doing

I like to keep myself pretty fit. I have done two parachute jumps, and I've done some abseiling. Travis and I go to a Phab club every other Monday. We do all sorts of things, like ten pin bowling, trips to the cinema, going out for meals or on a canal boat.

If you're considering a holiday like Dale and you would like to consult him about his experiences, he'd be happy to talk to you on the phone.

Dale: 0161 338 3183.

More information

Las Piedras is a fully accessible guest house in Southern Spain.
www.laspiedras.co.uk
00 34 952 033 100.

Sandpipers is a Vitalise holiday centre in Southport.
Vitalise offers a variety of accessible breaks and services for disabled people and their carers. **www.vitalise.org.uk**
0845 345 1972

Phab is a national charity which was set up 'to promote and encourage people with and without physical disabilities to come together on equal terms, to achieve complete inclusion within the wider community and to make more of life together". They support a network of over 200 clubs in all age ranges offering activities which all members can enjoy together.
www.phabengland.org.uk
020 8667 9443

Wimbledon

Wheelchair users have a special ballot for spaces on Centre Court, Court No 1 and Court No 2.

If you would like to apply for a place, you need to act quickly!! The deadline for applying is 15th December 2007.

For a 2008 Wheelchair Ballot form you must first obtain an application form. Write to the Wimbledon ticket office, and mark the letter and envelope 'Wheelchair'. The closing date is 15th December 2007.

Entry into the Ballot does not automatically entitle applicants to tickets for Wimbledon, but a place in the draw for a pair of tickets.

Successful applicants are selected at random by computer. It is not possible to request tickets for specific days or courts, as the day and court offered are also chosen randomly by a computerised selection process.

For a ballot form, send a self-addressed, stamped envelope - DL size (110mm x 220mm, 4 1/4 " x 8 5/8") - to:

Ticket Office
AELTC
P.O. BOX 98
London SW19 5AE

Only ONE application per household is permitted.

Letters

If you have a query or a good idea you'd like to share, write to the newsletter!
See page 2 for the address to write to.

Morgaina on the Skytower

from Vanessa King
Kia Ora from New Zealand

It is so great to receive the A-T News. Some of you may be familiar with my daughter, Morgaina, who has featured several times in the News, once swimming with the dolphins at Australia's Sea World, and another time abseiling from a 192m platform off Auckland's Skytower, to celebrate her 18th birthday. (She did it again for her 19th). High points aside, we have felt very isolated with this condition as there are only two people in New Zealand (population 4 million) with A-T, and I only found out about the second person two months ago! Unfortunately for us, he is only five years old, and lives at the other end of the country.

I recently visited the UK to stay with relatives. It was the first decent break I'd had in many years of caring for Morgaina. The July issue of A-T News was waiting for me when I got home, and I was very interested in the article written by Frances Prokofiev about the educational experiences of her son, Rupert.

Here in NZ, there are no Specialist Residential Colleges, and educational funding for disabled people is very limited. I have been greatly concerned about Morgaina's future, as she moves into adulthood, and although there are supported living environments here, I'm not confident that the funding allocated to her would be sufficient to ensure that her life was anywhere near that which I can provide at home. However, I so badly want to see her growing into an independent young woman, and she so badly craves to be with people her own age.

After discussions with my own parents, I discovered that we are British citizens by descent, and as such, Morgaina might be entitled to funding from the LSC to attend a residential college in the UK. I am particularly interested in The Orpheus Centre, Hereward College and National Star College. I am wondering if anyone could give us some feedback about these places and the funding processes involved?

We seem to be in a bit of a Catch 22 with needing a Connexions Officer to initiate the process, but as we don't have a "local" office (as we are in NZ) we can't get the ball rolling. If anyone can provide some advice or assistance it would be greatly welcomed. You can contact me at fishdontfly@orcon.net.nz.

Many thanks,

Vanessa King

In brief

Finding a Vein

Did you know that if you are having blood taken, or an IV drip put in, it helps if you drink a lot of fluids beforehand? Drinking plenty of fluids will make it easier to find the vein.

Special Child Website

This is a website that provides advice for parents and family of a disabled child, addressing their problems, worries and fears:

- How to cope with feelings
- What help is available
- Taboos like death and failing to cope

The website gives a refreshing perspective as suggestions are based on practical experiences rather than just general theories.

Who made it and why?
Diana Kimpton compiled this information when she found out her two sons had cystic fibrosis. She felt that she needed to understand why she was struggling so much emotionally and how to cope, rather than simply reading facts about the disease. Finding there was very little information written for parents, she wrote a book covering these issues in relation to her personal experiences. This book is currently unavailable to purchase but the website has a full content and detailed summary of the book. It is a personal, honest and detailed account of how she has dealt with having a special child in the family, covering issues such as the initial emotional acceptance to a diagnosis; the role of doctors and therapists; educational aspects; its effect on family matters; ways of coping, and looking to the future.

It also provides a list of website links that relate to family support.

www.specialchild.co.uk/index.htm

Help with Reading

Victa do scanners that will read text out loud.
www.victa.org.uk

National Blind Children's Society take books of your choice and enlarge the print.
www.nbcs.org.uk

Information in many languages

An overview of A-T is available now in 14 different languages.

Arabic
Bengali
Farsi
French
Greek
Gujarati
Mandarin
Punjabi
Somalian
Spanish
Tamil
Turkish
Ukranian
Urdu

If you would like copies of any of these translations, contact Kay Atkins in the office on 01582 760733.

Email forum for Clinicians

The A-T Children's Project has set up an international email forum for clinicians dealing with A-T patients. Through this forum they can seek advice from other A-T experts should they need it.

Any clinicians wishing to register should contact Jennifer Thornton, Director of the A-T Children's Project, by email:

Jennifer@atcp.org

A timely reminder from Dr Graham Davies, Consultant Paediatrician/Immunologist, Great Ormond Street Hospital:

Flu Vaccinations for People with A-T

It is again that time of year when the prospect of a flu outbreak looms. The Medical Advisory Panel recommends that all people with A-T should receive the flu vaccine each autumn. This autumn we understand that there are delays in getting supplies through. Nevertheless we would recommend that you contact your GP to get your name put down for the vaccine when it does arrive. The vaccine is a non-live vaccine and is perfectly safe for people with A-T. Children having it for the first time need two doses, one month apart, while for everyone else a single dose will suffice.

Don't miss the..

A-T Family Day 2008

Saturday 10 May
Nottingham

more details to follow

Xscape Weekend

8-11 March 2008

The 'Xscape Weekend', taking place in Milton Keynes in March 2008, promises to be an exciting opportunity for over 16's to escape and relax!

There are lots of fun activities available – from bowling and shopping, to cinema and theatre trips. For the more daring participants, there are even opportunities to try out skiing in the snow dome or even the 'Airkix' sky diving simulation! Those taking part can also contribute ideas about the A-T Society's work, at a meeting with the Board of Trustees.

For more information, contact Kay Atkins at the A-T Society.

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