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A-T Society News

The Ataxia-Telangiectasia Society

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

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

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Editor's Comments

Many thanks to all contributors. The copy date for the next issue is 1st October 2009
Please send comments, ideas, articles and pictures to the newsletter editor:

Beatrice Prokofiev
Flat 2, 32 New England Road, Brighton, East Sussex, BN1 4GG
01273 746462

email b_prokofiev@yahoo.co.uk

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

A-T Society
IACR-Rothamsted
Harpenden, Herts AL5 2JQ

tel 01582 760733 fax 01582 760162
atsociety@btconnect.com
website: atsociety.org.uk

Registered Charity No.1105528

Cover picture: Beth Carter in her new Hippocampe chair (details on p23)

Looking back, looking forward

The A-T Society began on 29th April 1989 when a group of ten families held a pilot meeting at Acorns Hospice in Birmingham. That this meeting was able to take place was inadvertently due to Professor Taylor and Dr Woods of the University of Birmingham who were researching into A-T and required blood samples from families. Dr Woods' task was to track down as many families who had children with A-T in the UK as possible, not such an easy job at the time. One thing led to another and as a result one of the families was able to make contact with the others involved in this research and organised the pilot meeting at the Hospice.

At that meeting the families agreed that the aims of the A-T Support Group were to be:

- To support A-T families
- To raise public awareness of the condition
- To fundraise in order to meet aims 1 and 2 and to fund research.

Early tasks for the committee were to produce an information sheet about A-T for families and one for the general public and to start a newsletter. Charity status was granted in 1990 and the name was changed to the Ataxia-Telangiectasia Society. Already new families were making contact and requests for information were being received.

Fundraising activities by families began. Of note were those by two young men with A-T, a parachute jump by Dale Phillips and an absail down the side of Charing Cross Hospital by Robert Soper.

By June 1990, the committee had established regular family meetings and were discussing the allocation of funds for research and welfare grants Soon after, they agreed that a multi-disciplinary clinic of expertise for those with A-T should be established to address another major concern, namely the lack of expertise around the clinical management of A-T. This came to fruition in October 1993 at Nottingham City Hospital.

The main elements of the Society's work came into place relatively quickly (family support, information, expert clinical management and research funding) and although they have been expanded and developed since then, they still remain the main focus of our work.

It is a cause of celebration that we have been in existence for 20 years but the challenges have been huge and remain so especially in this uncertain economic climate. Some wonderful people have supported us over the years and to them we offer our heartfelt thanks. They enable us to keep going. We must also thank the first families who came together and all trustees who have served or who are presently serving.

Our children are unwittingly providing mankind with important clues about the mechanisms that govern the development of cancers, neurological deterioration, immune system deficiency and more besides.

A huge amount has been achieved but it is not enough. Please continue to get the message out there, that A-T is deserving of the interest of everybody. We do not want to be here in 20 years' time.

Thank you.

Maureen Poupard
Hon. Secretary

International Research Update

New Trustee Emma Ross gives an overview of the A-T research taking place around the world

Although it is a rare condition, A-T is being researched around the world. Small groups of researchers are addressing a range of A-T associated issues, particularly in USA, Italy and Israel, as well as the research being performed at our clinics here in the UK. 57 research articles have been published since January 2008, specifically investigating Ataxia-Telangiectasia. Here is a brief summary of the main research topics being researched currently:

Pulmonary function in A-T (John Hopkins University, USA)

Ophthalmic issues in A-T patients (Saudi Arabia)

L-Carnitine as a prospective pharmacological therapy (Italy)

L-Carnitine has been shown to have a protective effect on oxidative stress-induced DNA damage in A-T cells suggesting its possible role in future pharmacological applications in A-T therapy

Steroid-induced improvement of neurological function in A-T (Italy)

Steroids (betamethasome) was administered orally for 10 days, and an improvement was observed in speech, finger chase and nose-to-finger tasks.

ATM gene and A-T review (Greece)

Mutations on ATM gene (as occur in A-T) lead to a loss of ATM protein which normally recognizes DNA damage and activates DNA repair machinery to minimize genetic damage.

Neural stems cells as potential therapy for A-T patients (Tel Aviv, Israel)

Neural stem cells have been shown to be a promising treatment in neurodegenerative diseases. However, the potential for tumours arising from the transplanted stem cells requires more investigation.

Breast cancer incidence in female A-T carriers (NY, USA)

A-T carriers are at a higher risk of developing breast cancer later in life, and this risk is augmented by cigarette smoking.

Review on A-T and cancer risk (Brisbane, Australia)

In Memoriam

We are sorry to announce Samantha Matthews died in February. We extend our warmest sympathy to her family and friends

We have a Memory Book of children with A-T who have died. If anybody would like a copy please contact Maureen, at the A-T office, on 01582 760733.

New Editor

I'm Beatrice and am proud to introduce myself as the new Editor of the Newsletter. My twin brother Rupert has A-T so the Society has been very important to me growing up. I have fond memories of going to family days as a child, surrounded by supportive families going through similar experiences.

The newsletter has always struck me as so informative and encouraging to families, and I'm very proud to now be involved in it. I'd like to take this opportunity to thank Jo for all her hard work as editor for the past eight years - a tough act to follow!

Afternoon tea at the House of Lords

Lord and Lady Parkinson meet Suraen and family at the House of Lords.

To mark the Society's 20th Anniversary, a reception was held at the House of Lords, where Lord and Lady Parkinson greeted guests.

Further photos and a report will be in the winter edition of A-T Society News.

Alton Towers

The group arrives at the park entrance

Family Day, May 2009

In May, families from around the country (and a few from outside) gathered in Staffordshire for the Society's special 20th Anniversary family day, which was held at Alton Towers. While some listened to the talks back at the hotel, others explored the park and enjoyed the many rides. With the biggest turn out for a family day yet, the weekend was a great success!

What people said

" I really enjoyed the weekend and it was great to meet other families."

"I thought the speakers were very good and easy to understand"

"The time in the park was brill, it was really good to see the children, young people and adults, relaxing and enjoying themselves"

"I had such a nice time, everyone was so friendly and welcoming"

"We all had an absolutely wonderful time"

"I have met some wonderful people! I have a feeling of happiness inside myself when I am thinking back"

Adam, Rupert, Ian and Nick enjoying the park

Open Forum

The Open Forum at the Family Day gave people a chance to put questions to a panel of experts. The issues discussed are summarised below:

The panel from L to R: Sue Hall, Physiotherapist, Dr Jayesh Bhatt, Paediatric respiratory specialist, Nottingham, Dr Mohnish Suri, Geneticist, Director of Nottingham Clinic, and Sarah Jessop, Occupational Therapist, Nottingham Clinic

Our son suffers from swollen achy feet and pains in his knees. What can be done to help him and what causes it?

Swelling in lower limbs is a complex issue – there may be a number of factors which should be investigated. Swelling in the feet is fluid retention. It may be related to posture and positioning – or to hydration levels, inactivity or decrease in muscle function. During the day, look at levels of activity and changes in position. At night, you could try raising the end of the bed – two pillows, or something as thick as a phone book, placed under the end of the mattress, could make a difference.

It might help if your son sometimes moves his feet (in a pumping action) when he's sitting still.

He could try other activities such as swimming. The physiological effects of swimming help stimulate renal function. Stimulating footbaths might help too.

Look carefully at diet – checking diuretic effects of various foods. As for pain, it may be about position, activity or lack of movement. It might help if he moves more, and changes position more often.

My daughter has lately suffered from various viruses. Is this caused by having A-T (ie the immune system being weak?)

A lot of people with A-T do suffer from infections. Is this a new problem for her? Advice depends on her age and the social context. At certain ages, children are more susceptible to viral infections. And if she has just started school, nursery, or college, she'll be exposed to more viruses.

Anyone who is getting infections should get their immune function checked. Is it a change of nutrition? Get a check.

My daughter is 16 now and I am worried about her ankles and the angle they are at. At the moment she has solid plastic splints but they don't seem to help, and sometimes they cause pain.

Is there any other origin of pain? Do the splints fit? Why were they originally prescribed and are they still meeting that need? One set of splints doesn't last for ever, and if she has swelling or has put on weight they may not be a good fit. However, no splint fits perfectly.

Exercises, stretches or posture in the chair should also be looked at.

Advice about Flu:

- Any individual with A-T should have the flu vaccine. Hopefully by next autumn the flu vaccine will incorporate the new strain (H1N1).
- The Prevenar vaccine (against the pneumococcus bug) seems helpful for people with A-T.
- If anyone develops symptoms suggestive of flu, discuss this with your GP.
- It's very common for young children to get ill. A cough with a fever lasting about a week is nothing to worry about but if it's still there after 3 weeks this is cause for concern.

My son is 3 years old and has a very poor appetite. It is very difficult to get him to eat – can you help with some advice?

Many children of 3 have a poor appetite and don't eat well; and lots of kids with A-T do have a good appetite.

- Does your child have problems with aspiration?
- Is there a physical cause?

If not, are there ways to supplement his diet, and increase his appetite?

Speech therapists, occupational therapists, and physiotherapists, may all be able to help.

Make sure your child is not too tired to eat. Offer smaller, more frequent meals, and healthy snacks.
Think about ways to make eating enticing.

To get him interested in food, play games associated with food. Little children are very visual, so you could look at pictures of food, and ask him to choose his food from a variety of pictures. Play the pairs matching game with pictures of food.

There are several books you might find helpful:

- Child of Mine: Feeding With Love And Good Sense. by Ellyn Salter, Nutritionist
- How To Get Your Kids to Eat…But Not Too Much. Ellyn Salter. Both of these books are available on Amazon.
- Diagnosis and Treatment of Feeding Disorders in Infants, Toddlers and Young Children by Irene Chatoor (a psychiatrist) - a good book on child-parent interaction over food.

Food is emotive for everyone including children, particularly young children.
Is your son using food as a way of controlling the situation he is in? What do you do as a family - what is your 'eating style'? Could you consider as a family changing your eating style?

What will be the future focus of A-T Research?

By Professor Malcolm Taylor, University of Birmingham

It is over half a century since Elena Boder and Robert Sedgwick described Ataxia- Telangiectasia (A-T) as a syndrome. In 1995 the ATM gene (which when mutated causes A-T), was identified and a mouse model of A-T came a year later. Since 1995 we have learned a great deal about the biochemistry of the ATM protein.

However, in 2009 we are still unable to provide a treatment for patients to slow down the progression of A-T. The giant that we face in A-T is the unrelenting progressive neuro-degeneration.

What should we be doing for patients with A-T?
The answer is obvious: we should be aiming to identify a safe and reliable treatment that will prevent the progression of the disorder in the youngest children, and preferably also reverse the neuro-degeneration in older patients. We can treat some of the symptoms resulting from neuro-degeneration – for example reducing unwanted movements. In addition, Baclofen, a muscle relaxant, is currently being trialed in A-T patients to determine whether it is effective in treating some of the neurological problems in A-T.

We should be aiming beyond this, to at least slow down the rate of progress of the neuro-degeneration. There are various strategies that have been tried and others that could be tried in future. At present, the most likely way of achieving this aim quickly is by identifying, for this purpose, one or more drugs that are currently available that might moderate the progression of the ataxia.

The Challenges Facing Developing a Treatment
The difficulty is that if it is loss of the ATM protein that is causing the problem, how is a drug treatment going to deal with this loss?

The Use of Antioxidants
One possibility is that a drug might prevent the consequences of loss of ATM. One approach that has been taken is the possible treatment of patients with antioxidant agents. The rationale for this is that one of the roles of the ATM protein is to respond to damage to the DNA in the cell. Such damage can be caused by reactive oxygen molecules that are present in everyone's cells. One method of treating A-T patients would be, therefore, to administer a drug that will mop up these reactive molecules before they cause damage and so make redundant the need for the ATM protein.

Work, with such agents, on A-T mice, has shown encouraging results. No large study has been published so far on treatment of A-T patients with antioxidants, although in initial work some positive effects were seen in cells from these patients but there were no significant changes in the clinical neuro-degeneration.

Other commonly used safe drugs

A major question deserving an answer is what other commonly used safe drugs might help arrest the clinical features of A-T? Are there other antioxidants that might be used as potential treatments? Are there drugs, other than antioxidants that might be considered as treatments? Administration of steroids has been shown to result in some temporary improvement in ataxia. The problem is that steroid treatment cannot be carried into the long term because of the serious complications that are seen. Another important point here is that we do not understand what the mechanism is, of this improvement by steroid treatment.

Identifying new drugs

Aside from utilizing existing drugs, there is the possibility of identifying new ones. Research funding bodies are aware of these issues. The National Institutes of Health, Washington, put out a call in 2007 for research projects on 'Understanding and Treating Ataxia-Telangiectasia'. One of the suggested areas to be developed was the development of assays for high-throughput drug screening. Some would argue that we need to develop a more trialist ethos and this may be correct, but of course with a well chosen, safe drug.

Other approaches to developing a treatment

These may depend either on some genetic intervention/override to switch back on, the production of the ATM protein or biological intervention using regenerative medicine to insert cells (stem cells) with normal ATM protein. For those A-T patients who have no protein as a result of a particular genetic mechanism, a way of inducing genetic override was proposed that would enable the production of the ATM protein to be switched back on again. This can be shown to work in cells in culture but is not yet something that can be applied to patients. This approach would not be applicable to all A-T patients.

An important observation

Again, with respect to the potential for genetically altering cells in A-T patients a very important observation was made on two siblings with A-T, shown to have no ATM protein at all; they did not show the same severe neuro-degeneration as seen in the majority of patients. What this is saying is "I can just about manage without ATM". These patients were neurologically able, to a significant extent, to compensate for loss of ATM. How this happens is not known. Is it by expressing another protein not normally expressed in the cerebellum that does something similar to ATM, or is there expression of a protein in the cerebellum that inhibits the consequences of ATM loss? What is this protein? I believe that this question deserves some effort to determine the answer. Could we then use this to induce this compensation for ATM loss in typical A-T patients?

Stem cell therapy

Stem cell therapy is much in the news for different disorders – most recently for macular degeneration of the retina. There are new methods for making stem cells that do not rely on embryos. It is now possible to induce normal adult cells to become stem cells. Biological replacement of the ATM protein may be an alternative for the future - so called regenerative medicine possibly by inserting stem cells in the brain. At present, this is not without risks. One A-T patient has already undergone such an operation, although without any beneficial effect. Indeed there may be harmful consequences at present if we cannot control the stem cells properly. This is a field that will develop very rapidly and where there may be important advances in the use of stem cells in more common neurological disorders. These advances will be potentially advantageous to families affected by A-T.

Important considerations for these approaches both for A-T families and doctors/scientists

Would families affected by A-T want to participate in drug trials for possible treatments? Ideally a child might best be treated shortly after diagnosis. It may be difficult for parents to accept this and willingness to allow young children to be entered into trials may be important.

The internationalisation of effort

For doctors/scientists, the internationalisation of effort, especially with respect to clinical trials, is likely to be necessary in order to get the numbers of patients required to make the trial statistically sound. Reliable clinical endpoints, based on a neurological feature(s) that responds both quickly and with a magnitude large enough and consistent across patients will need to be developed.

Use of animal models
Much of this sort of work could be done initially on animals if an animal model existed that reflected the neurological degeneration of A-T better than the current mouse model. If such a model were available we would be able to study the basic pathology of the neuro-degeneration much more thoroughly as well as the associated biochemistry in the neural tissues most affected. It is absence of our knowledge of the detailed pathology and biochemistry of the developing A-T cerebellum that is a major difficulty. I believe, however, that progress will be made in the areas that I have mentioned above, involving patients with A-T.

Indeed after nearly a decade and a half of "the biochemistry of the ATM protein" we ought to be on the threshold of "Understanding and Treating Ataxia-Telangiectasia".

Professor Malcolm Taylor, University of Birmingham

The Race for a Cure: Preparing for Clinical Trials and the Importance of the A-T Neurology Workshop

Cynthia Rothblum-Oviatt, PhD - Summary of 2009 A-T Family Day Presentation

Cynthia speaking at the Family Day

The 2009 A-T Neurology Workshop took place concurrently with the A-T Society's Family Day on Saturday, May 9. The importance of this workshop in terms of preparing for clinical trials was put into context of the bigger picture of 'how drugs reach people' or the drug development process.

The drug development process can be divided into several phases: discovery, preclinical development, clinical development (which includes human trials), approval and market. Drugs that successfully pass through the clinical development phase will be approved by a government regulatory agency and pass into the market place where they will be available to the general population. Drug development is an expensive and lengthy process. However, there are times when a drug which has gone through this long and costly process for one disease is found to be useful for an entirely different disease. This means that a drug that has been approved for a different neurodegenerative disorder may hold promise for ataxia-telangiectasia. The A-T Children's Project and A-T Society are always looking for this possibility as it represents a type of short cut to therapy. As alluded to above, the clinical phase of the drug development process includes three basic types of human studies: phase I, phase II and phase III clinical trials. The primary goal of these trials is to determine drug risk (adverse side effects) and drug effectiveness. But if we were to have a clinical trial today for A-T, how would we measure drug effectiveness? One way would be to use a neurological exam, the development of which is why the A-T Neurology Workshop took place.

Workshops bringing together neurologists and patients with A-T have been essential for the development of what we

call the A-T Neuro Examination Scale Toolkit. The first neurology workshop took place in the US in 2007, and it brought together 19 patients with A-T (ranging in age from 1-27 years) with 12 neurologists, two occupational therapists, two immunologists and a pediatrician. Importantly, several of the neurologists were new to A-T and were therefore able to provide objective input regarding scale development. This year's A-T Neurology Workshop brought together 15 patients with A-T (ranging in age from 2-45 years) with five neurologists, one occupational therapist, one immunologist and one clinical research fellow. This year we had fewer clinicians and all the neurologists were quite familiar with A-T because the goal was to test and refine the existing draft of the toolkit. Ultimately, we hope to develop a toolkit of different neurological exams that can be used together or in various combinations for clinical trials that take place simultaneously all over the world.

Although scale development requires much time and effort, these evaluation tools are desperately needed by A-T clinician/scientists worldwide to help improve patient care and enable clinical trials of new drug therapies.

A Worldwide Patient Registry for A-T

Another way in which we can be prepared for clinical trials is to develop a worldwide registry of patients with A-T. This registry would represent a source of patients for multi-site clinical trials. The A-T Children's Project is searching for a database to house such a registry, but once the database is established, we will be seeking the help of the A-T Society to ensure that every patient in the UK with A-T is a member.

"I want to thank Maureen Poupard and the A-T Society for taking on the extra work that allowed us to run a Neurology Workshop concurrently with their Family Day. I think we made great progress. And I hope we didn't 'stress' the families too much! Maureen, please let your families know that the A-T Children's Project thanks them for participating in this important meeting. Without their help, we couldn't have made progress on this initiative."

Extract of an email from Cynthia to the Society

Helen Hart, the A-T Society Counsellor, is now available on email. You can contact Helen on Helenat32@blueyonder.co.uk.

If people haven't got access to a computer then they can contact us and we can put them in touch with Helen by phone

Immunologic and Respiratory Assessments of A-T

A summary of Andrew Exley's presentation from the Family Day, on his research at the Papworth clinic.

Thank you for the invitation to the A-T family day to present some thoughts from our experience of the immunologic and respiratory assessment of A-T through the A-T adult clinic with John Shneerson and colleagues at Papworth Hospital, Cambridge. This work is in collaboration with Professor Malcolm Taylor, Birmingham, together with Elizabeth Hodges and Susan Harris from Southampton. We have also benefited enormously from your support to present at the International A-T workshop in Kyoto, Japan in April 2008, and support from the A-T Children's Project to present at the Workshop on Pulmonary Disease in A-T, Baltimore, USA, April 2009. Both these meetings have allowed us to have stimulating discussions with colleagues from the Nottingham clinic, and other European specialists in A-T. These discussions have generated new ideas about A-T which provide the basis for preliminary agreement on a European collaboration in a trial of early immunoglobulin replacement in A-T.

We began the programme of immunologic and respiratory assessment of A-T with the multi-disciplinary team at Papworth with some simple ideas about how different aspects of the condition might interact to result in respiratory failure. Immunodeficiency might be revealed by recurrent bacterial infections leading to an accumulation of lung damage including airways damage or bronchiectasis. Difficulty in swallowing, perhaps due to the cerebellar defect, might result in aspiration of food into the chest provoking inflammation with lung damage, and poor nutrition with weight loss. We would therefore expect respiratory function to be impaired by progressive lung damage and muscle weakness revealed by standard assessments including chest CT scans. The immunodeficiency might be exacerbated by the strain of controlling chronic viral infection such as CMV, and common variations in modifying genes.

Our initial experience suggests these early ideas need some modification, particularly if we adopt the principle of health maintenance or pre-emptive care. Immunodeficiency in A-T is profound and relates directly to a limited ability to compensate for the lack of ATM kinase in both T cell development and antibody production. In cases with mild A-T, where the gene mutations indicate some residual ATM kinase function, there is evidence the immunodeficiency correlates with age, probably as another example of early ageing in A-T. Experimentally, recurrent infections drive progression of immunodeficiency particularly when T cell numbers are reduced. Infections also increase metabolic or oxidative stress. The metabolism or workings of cells and the environment they are exposed to inevitably generates metabolic or oxidative stress that is relieved by standard counter measures. If the metabolic stress can not be relieved then cell damage occurs. A-T is characterized by an increased sensitivity to metabolic/ oxidative stress, which exacerbates the ATM kinase deficiency and leads to cell damage. A key consequence of immunodeficiency in A-T might therefore be a damaging increase in oxidative stress from respiratory tract infections. This might result in progression of the neurologic, immunologic and respiratory problems in A-T before clear evidence of structural lung disease as seen in common variable immunodeficiency for example. Just as the neurology of A-T appears unique although it shows overlap with a number of different disorders.

Clinical Trial

Respiratory tract infections in chronic pulmonary disorders are due to viral and bacterial infections. The strategy of strict infection control to reduce metabolic stress, stabilising A-T and / or reducing progression of the condition therefore requires a treatment such as immunoglobulin replacement. In the UK, immunoglobulin replacement is an approved form of treatment for primary immunodeficiency but a clinical trial is required to test whether early use is beneficial. Colleagues in Europe have expressed an interest in such a trial and together we have agreed the best study design would be a comparison of immunoglobulin replacement versus antibiotic prophylaxis. A detailed protocol will need to be generated, funding agreed and regulatory approval established before such a trial can commence.

Papworth Everard
Cambridge
CB23 3RE

Papworth Hospital NHS
NHS Foundation Trust

Pulmonary Disease in Ataxia Telangiectasia Workshop

By Jayesh Bhatt, Consultant Respiratory Paediatrician, Nottingham University Hospitals

23rd to 24th April 2009, Baltimore, USA

This workshop was organised by the A-T Children's project USA. The workshop objectives were to develop and publish a consensus statement on management of lung disease in A-T and to look at developing further research questions to explain the reasons for lung disease and methods to prevent it.

The invited participants included A-T clinicians including Paediatric Pulmonologists (clinicians with expertise in lung disease in children), therapists, researchers and parents of children with A-T from the USA and Europe, including the UK.

After a welcome address from the Volunteer President and Founder of the A-T Children's project, there were some excellent presentations reviewing:

- Lung disease in A-T,
- The molecular basis of A-T,
- Neurological features in A-T, brain function related to breathing and swallowing, muscle strength testing, swallowing abnormalities
- Impact of aspiration on lung function
- Managing and maintaining lung health in multisystem problems
- Sleep problems in medically complicated patients
- Interstitial Lung Disease in A-T
- Oxidative stress
- Vaccinations / preventative therapy
- Treatment of immunodeficiency
- Microbiology
- Lung function testing in A-T
- Nutrition and gastrostomy tubes
- Peri-operative management in complex chronically ill patients

It was suggested that we should learn from the management of lung disease in other chronic multisystem disorders (Cystic Fibrosis, Duchenne Muscular dystrophy) where proactive expectant management has led to hugely improved outcomes in terms of survival, overall wellbeing and quality of life. The goal should be not to lose any ground on lung function or nutrition levels at every clinical encounter and intervene proactively e.g. doing gastrostomy before it is too late and the body mass index has dropped significantly.

Lung disease was well described in some of the initial reports on A-T (BODER E, SEDGWICK RP. Paediatrics. 1958) but it is only relatively recently that the focus shifted onto the management of lung disease to try and improve survival. Sino pulmonary disease is common and can be silent. Nearly 50 % of children with A-T have respiratory morbidity by the mean age of 10 years. There are several reasons for lung involvement as follows:

- Primary effects
- Neuro-degeneration
- Decreased respiratory muscle strength
- Poor airway clearance
- Abnormal swallowing and aspiration
- 80 % have immune defects which contributes or leads to bronchiectasis
- Some have autoimmune lung phenotype and develop interstitial lung disease.

Secondary effects on lung: There is increased oxidative stress so the lungs are particularly susceptible to:

- Environmental factors e.g environmental tobacco smoke, pollution
- Abnormal cell repair, premature ageing of lung
- Systemic proinflammatory cytokines

Three phenotypes of lung disease seem to emerge in those with A-T though there are often overlapping phenotypes:

Chronic sinopulmonary disease (frequent infections, moist cough, chronic or intermittent chest congestion, immunodeficiency, bronchiectasis)

Neuromuscular and bulbar weakness (trouble clearing secretions, aspiration of food, liquids or saliva)

Interstitial lung disease (ILD- dry cough, rapid breathing, poor response to antibiotic therapy)

It is important to recognise that "lung health" should not be equated to absence of symptoms as persistent symptoms are indicative of advanced lung disease. Routine microbiological cultures should be performed in all children and treatment thresholds should be clearly defined. There should be a clear definition of an exacerbation and a structured approach to treatment. Respiratory Paediatricians should be involved early in the management and should have annual inputs in the management.

Early management and monitoring

of lung function is necessary to minimize lung damage. Nutritional impairment should be avoided and consideration to gastrostomy tubes should be given early especially when there swallowing dysfunction with or without aspiration.

Monitoring of lung function should include lung function tests, thorough symptoms review and clinical examination, overnight oxygen studies / polysomnography as indicated, chest and sinus xrays and in some instances chest CT scan (to identify bronchiectasis or ILD). Chest xrays are not necessarily to be avoided but will be required in a child who is not improving on antibiotic treatment, anybody with more than 2-3 episodes of respiratory tract infection per year or with chronic chest symptoms. Lung biopsy is usually not required for ILD as the changes are very nonspecific.

Management should include ensuring routine immunisations including Prevenar and Pneumovax II as well as influenza vaccine annually. All the household contact should be vaccinated against influenza. The value of routine preventive antibiotic use in preventing respiratory tract infections remains unproven and this should be balanced against the potential risk of adverse events including antibiotic resistance, C Difficile infection etc. Immunoglobulin replacement therapy will be required in some individuals. ILD will require treatment with corticosteroids.

Airway clearance should be taught to carers and may include assisted coughing devices.

A 'Consensus document' is being prepared and will be submitted for publication to scientific journals for wider dissemination.

Swallowing in Ataxia-Telangiectasia

Dr Maureen Lefton-Greif with Jayesh Bhatt at the Family Day

Maureen A. Lefton-Greif explains the swallowing process and problems that can occur for those with A-T

We swallow approximately 600 - 900 times daily. Swallowing is critical for survival. Its two primary functions are to: (1) direct food, liquid, and saliva from the mouth to the stomach while keeping the airway protected; and (2) deliver enough of the right types of liquids and foods for adults to stay healthy and children to grow and develop. Although, the act of swallowing appears to be simple, each swallow requires the coordination of multiple muscles in the mouth, throat, and esophagus. In addition, each swallow must be coordinated with breathing to avoid aspiration (i.e., liquid, food, or saliva entering into the trachea or windpipe).

Swallowing occurs in four overlapping and inter-related phases - oral preparatory, oral, pharyngeal, and esophageal. During the oral preparatory phase, foods are chewed and formed into a ball (called a bolus) for swallowing. The bolus is moved to the back of the mouth during the oral phase. During the pharyngeal phase, the airway is closed while the bolus is transported from the mouth, through the pharynx, and into the esophagus. The primary role of the esophagus is to move the bolus into the stomach.

Dysphagia [dis-fā'ja] is the term used to describe swallowing problems. These problems may involve any or all the phases of swallowing described above. For example, if problems involve the oral preparatory phase of swallowing, a person may have

trouble preparing foods (e.g., meats or raw vegetables) that require chewing. Problems with the pharyngeal phase of swallowing may cause someone to cough or choke when thin liquids are aspirated into the airway. Unfortunately, some people aspirate without coughing. Aspiration that does not result in a cough response is called “silent aspiration”. Silent aspiration is a problem because the protective response of the cough is missing and there is no indication that a swallowing problem has occurred. A prompt and strong cough helps clear the airway when something was aspirated and it signals caregivers that a problem has occurred.

In our experiences at the A-T Clinical Center at the Johns Hopkins Hospital, dysphagia is common in A-T and typically presents during the second decade of life. That said, not every person with A-T develops swallowing problems. In A-T, common mealtime signs of swallowing problems are: coughing (particularly with thin liquids), difficulty chewing solid foods (e.g., meats, raw fruits and vegetables, nuts or seeds), excessively long meal times (longer than 40 minutes), very short mealtimes (due to fatigue), and meals that are stressful. Over time, dysphagia can cause respiratory or nutrition problems, or both. In turn, respiratory and nutrition problems may cause significant health problems and adversely affect a person’s quality of life.

When dysphagia is suspected, the Videofluoroscopic Swallow Study (VFSS) is the gold standard for evaluating swallowing function and guiding decisions about how to treat swallowing dysfunction. The VFSS is a specialized x-ray study. As with any examination, and particularly those that involve radiation, one must weigh the potential risks and benefits associated with the test. A VFSS should not be completed as a screening procedure. In general, most children with A-T who are younger than 10 years of age do not need a VFSS. It is most helpful for children (older than 10 years) and young adults who have problems that appear to involve the pharyngeal phase of swallowing. Relative to the VFSS, ways to decrease radiation exposure are: complete a VFSS only after a clinic feeding/swallowing evaluation shows that one is indicated, keep the VFSS as short as possible while being certain to obtain the necessary information (your Speech-Language Pathologist and radiologist will guide such decisions), and repeat a VFSS procedure only when there are changes in swallowing that need to be defined or management decisions require the additional information.

General Guidelines for Eating and Drinking

Listed below & on the next page are general recommendations that have been helpful for many families:

How to Set Up Mealtimes or Snacks

- Eat 4 - 6 small meals versus 3 large meals daily to decrease fatigue
- Eat nutritious items rather than “empty calories.” It takes the same amount of time and energy to drink a milkshake or instant breakfast shake as it does to drink juice, punch, soda, or tea
- Sit upright:
  - Usually best for safe swallowing
  - Remain upright after eating
- Keep the head, neck, and trunk in a straight line
  - Try to have the chin pointed down
  - Avoid positions where the head is tilted back (eyes towards ceiling)

How to Eat and Drink

- Take small bites
  - Cut food into small pieces
  - Place small amounts of food or liquid in the mouth
  - Make sure each bite is swallowed before putting more into the mouth

- Alternate food and liquid swallows
  - o May help clear the mouth and throat.

- Use a straw carefully
  - o Encourage one sip at a time because the likelihood of aspiration is increased with rapid sips through a straw.
  - o Pinch the straw to slow the rate of drinking, if needed.

What to Eat

- Pureed or soft foods
  - o Foods that stay together in the mouth may be easier to swallow
  - o If pureed or soft foods seem easier for your child, you may be able to convert favorite foods into pureed or soft foods

- Examples of pureed foods: pudding, custard, yogurt or creamed cereals such as oatmeal

- Examples of soft foods: soft pasta with sauce (e.g., macaroni and cheese), canned, stewed or baked fruit, ground or stewed meat moistened with ketchup or gravy

What to Avoid

- Avoid foods that are difficult to chew:

  - o Some solid foods may require extra effort to chew and swallow. These foods may increase the time it takes to eat and may make it difficult for an individual to consume enough of the "right" types of foods.

  - o Examples of difficult to chew foods are: some meats (e.g. steak, pork chops), raw vegetables, and fruits (e.g., lettuce, apples, raw carrots)

- Avoid or limit very thin liquids (e.g., water, fruit juice, punch or soda)
  - o Very thin liquids have few calories per ounce and little or no nutritional value.
  - o Very thin liquids are most likely to be aspirated

- Offer thicker liquids (e.g., milkshakes, malts, fruit nectars and creamed soups)
  - o Whole milk is preferred over skim milk

- Add liquid supplements
  - o Introduce liquid or food supplements, instant breakfast products, and/or milkshakes to boost the diet as per instructions from a physician or dietitian.

Drooling Control

- Swallow often
  - o Provide reminders to swallow frequently
  - o May wear a terry cloth band on the wrist and to use to wipe mouth
- Ask about medications
  - o May be used to control excessive saliva
  - o Learn about the side effects associated with medications that control saliva production

Emergency Plan

o Anyone with swallowing difficulties should have an emergency plan in case the airway gets blocked and breathing is obstructed

o Individuals with A-T, all caregivers, a physician, and a rescue unit should develop this plan

o Post the plan for easy reference

Additional information on feeding and swallowing as well as mealtime recommendations are available on the A-T Children Project's website ***(http://www.communityatcp.org/Page.aspx?pid=1285)****.*

Maureen A. Lefton-Greif, Ph.D., CCC-SLP, BRS-S, ASHA Fellow, Associate Professor, Department of Pediatrics, Associate Professor of Otolaryngology- Head and Neck Surgery, Johns Hopkins University School of Medicine

The David M. Rubenstein Child Health Building 200 N. Wolfe Street, Eudowood Division of Pediatric Respiratory Sciences- Room 3017 Baltimore, MD 21287, Email: mlefton@jhmi.edu

We hear from other A-T support groups abroad

Poland

“Foundation Razem Zdażymy was established in 2007. The idea of Polish A-T families group was based on excellent model of A-T Society. Our British friends are for us great example of devotion, diligence, great ideas and support that my daughter and I experienced during Family Day in 2006. We hope that Foundation Razem Zdążymy will reach some day high standards set by A-T Society. Best regards from Poland.”

“Fundacja Razem Zdążymy została założona w 2007 roku. Pomysł stowarzyszenia polskich rodzin dotkniętych problemem AT był oparty na wspaniałym przykładzie AT-Society. Nasi brytyjscy przyjaciele są dla nas wzorem poświęcenia, pracowitości, wspaniałych pomysłów i wsparcia, którego doświadczyłem wraz z córką w trakcie spotkania w 2006 roku. Mamy nadzieję, że Fundacja Razem Zdążymy osiągnie kiedyś wysoki poziom wyznaczony przez AT-Society.”

J. Nowak
President
Foundation Razem Zdazymy

USA

“The race for a cure for ataxia-telangiectasia (A-T) has been long and arduous. Although we have not yet reached the finish line, it has been an honor to have such a dedicated organization as the A-T Society by our side. The A-T Society does tremendous work. The commitment of the group is inspiring, and we are continually impressed by their efforts, including their orchestration of field-advancing clinical services and research workshops. We consider ourselves fortunate to partner with them to improve the lives of all affected by A-T. Together, we will one day reach the finish line.”

Cynthia Rothblum-Oviatt, PhD
Science Coordinator
A-T Children’s Project

Australia

“Good luck with your 20th Anniversary. Your foundation has helped so many families in the 20 years you have been going. You are all an inspiration to many.”
Krissy Roebig, Founder, BrashAT Australia

Germany

“Happy birthday, A-T Society!
Lucky twenty!
A-T is a very rare disease. So it is very important, that parents close together to fight for a correct diagnosis, a good treatment and perhaps for a cure in future.
Twenty years ago the location of the ATM-Gene was found, seven years later the gene was cloned. And always British people were involved in the development. This was the work of the A-T Society!
The A-T Clinic is a fantastic aid of the A-T Society. The treatment of the handicap is better then twenty years ago. But a cure is not to be seen. So much is done, but still more is to do.
I wish the A-T Society all the best for the future and much money to help the A-T families!”

Hermann Stimm
Deutsche Heredo-Ataxie-Gesellschaft
(German Society for Hereditary Ataxias)

20 Years On: What our families say about us

Left: Alecia celebrates her birthday at the Family Day with a special A-T Society 20th Anniversary cake

Marian Barber:
" I am sure that without the Society's help and interest over the years we would all be in a different and far less satisfactory, position in many ways.
The Society is successful at focusing on intensive medical research as well as assistance in every aspect of the effects of the condition on life of the individuals and their families. The research is crucial to alleviating the symptoms and the physiological impact of the condition as well as providing hope of a cure or reduction in severity. However, I think for me the greatest benefit has been the information exchange, the friendship and the sense of belonging. It has been extremely difficult for Emily to make and maintain friends over the years and the A-T Society has provided excellent links and opportunities for young people with A-T (who have just the same hopes and fears as any other set of young people) to get together. They are so disadvantaged in so many ways, but seeing charming, intelligent and witty young people having a whale of a time together is reason enough for me to support the Society!"

Helen Hart:
"When both my sons were diagnosed with A-T within a week of each other it was a very bleak time. In 1984 there was little information about the condition and what was available made very depressing reading - no treatment, no cure, a short life span, little research and no support around for families. The medical profession could not offer much in terms of best practice and ongoing support for a condition as rare as A-T. The setting up of the A-T Society which was started in 1989 by a few committed families immediately brought an end to the isolation that many of us felt. Over the 20 years of its existence - through hard work and a driving sense of wanting

to do the best for all children with A-T and their families, the progress made through funding research, raising awareness about the condition and improving the lives of those living with A-T through practical and emotional support have brought the Society to where it is today. Many of us are very grateful to the original group who had the foresight and determination to work for the best for children with A-T."

Robert Soper:
"The A-T Society has been helpful on several occasions, I wouldn't have been able to move to my current bungalow in Maidstone in 2005, which became an urgent necessity because of my deteriorating disability; I lived with carers in an adapted flat in Paddock Wood for ten years until things became impossible and I needed to move. The A-T Society arranged a meeting at my mother's house and a few weeks later I was in my bungalow. They also paid for me and my carer to go to Zurich to participate in a research project into A-T back in 2007".

Joan Bridger:
"When my son, Luke, was first diagnosed with A-T I felt totally isolated, very confused, and extremely frightened. After a lot of searching (this was 14 years ago) I eventually got to speak to Maureen who was the mother of 2 children with A-T. It was wonderful to speak to someone who knew exactly the position I was in, I didn't feel alone and words can't describe the relief I felt. My first visit to the A-T Family Day was traumatic but at the same time gave me hope that my beloved son did have a future. Sadly Luke died in 2001 but I became a Trustee of the A-T Society in the hope that in some small way I could bring hope to other families coping with this horrible condition."

Susan Simpkin:
"I think that what the Society has done for my family, words alone are not enough. When I first contacted the Society for help I had already fought for 10 years to get help with my daughter's deterioration in health and also for 4 1/2 yrs trying to get a social worker after the previous one had retired and having no success. Then Kay Atkins contacted us and she was absolutely vital to sorting out my family's needs at that time.
My husband was in denial and I had to deal with things on my own until Kay! She helped to contact Social Services and my local MP and things quickly moved along and I received the help I needed in the form of social workers.
Without her I would have no one to talk to about what is happening in our lives and my daughter's health problems which are slowly getting worse as explained to us by the A-T Society and all the paperwork they first sent me to read.
Although my daughter's condition is very serious I feel I have a lifeline to other families the same as ours when we meet at the annual Family Day where we also get to hear what the experts have to say about the condition and any other relevant information for the families.
As I stated earlier I could not have found better to help support us all."

Frances Prokofiev:
"We have been members of the A-T Society almost since its beginning; our son is now 24. Belonging to the A-T Society has given us the chance to meet others dealing with a similar condition which is hard to do when it is so rare and gives us courage about overcoming difficulties when we see that others cope courageously. As carers we get inspiration from other families who have faced the same difficulties in all sorts of different ways; for our son and his siblings it has given them the chance to know other patients and feel less isolated.
Another important benefit the A-T Society offers is that there is always someone at the other end of the phone in the office to give help and share the advice of others who have faced the same problems so that it is not a case of re-inventing the wheel each time.

The newsletter has given us invaluable access, for example, to knowledge about holidays or adapted tricycles. There is a strong sense of family at the annual gatherings which are always friendly and stimulating.

Parents and carers are brought up to date with new clinical findings and the entertainment provided for children and young people with A-T makes it a good day out for everyone.
The A-T Society's effort has meant that my son has been able to go to a specialist clinic throughout his life, receiving expert and accurate assessments and understanding about his condition which would not have been available through a general clinic which rarely comes across this difficult condition."

The Phillips Family:
"Initially, it was difficult for us to come to terms when both our sons were diagnosed with A-T, however with the help and support of family and friends and guidance of the A-T group we have successfully overcome most of life's difficulties. Once Travis and Dale accepted their disability they've grasped life and enjoyed travelling, raised money for A-T with parachute jumps, abseiling, etc. We have always enjoyed the support of the other families when we have our get togethers at the Family Days.

Our sons live at home with us and are happy to do so. We strive to make life good for them as much as possible and hope it will continue."

Jacqueline Armstrong:
"Although Pippa is no longer alive I keep up the contact because I feel that is what she would wish, also because I owe so much to the Society. I was able to meet people who understood-other parents and experts in various aspects of Pippa's problems."

Gwen & Albert Rowley:
"Although the A-T Society is relatively small it does wonderful work in the fields of research, providing information and support for families when difficulties occur."

Michele Keith:
"We first contacted the Society when George was diagnosed with A-T in May 1999, so ten years on, what a great help and support they have been! When we had the devastating news that George had A-T, we also had to cope with the added stress that I was already nine weeks pregnant with our second child but Maureen helped us straight away by putting us in touch with another family who had been in the same situation as us. We went to visit them a couple of weeks later and although the initial meeting was emotional as we had never met another child with A-T before, we gained so much hope and inspiration from this visit.

We have attended all the family weekends, we thoroughly enjoy these as do George and Francesca. They are able to interact with both A-T children and their siblings. We feel this is so important for them emotionally.

The Society's counsellor Helen helped us with a particularly bad time in George's school life. Her help and guidance was first class.

It is like having an extended family with lots of support.....basically they are our lifeline as being a parent of a child with A-T can be very lonely at times."

Ian McInnes*:*
"The good aspects of being a member of the Society:

a) You get to meet fellow suffererers and you strike up good friendships.
b) When I was a trustee I was able to put my point of view as someone with A-T which I think is very important for the Society.
c) I also enjoy giving my talks at the Family Days and for some reason they keep asking me back!"

International Ataxia Awareness Day

25 September 2009

Once again we're asking all our friends to do something to mark Ataxia Awareness Day, however small. Here are some easy ideas on how to get involved and raise awareness:

- Wear your A-T tshirt on Friday 25 September and tell people what it means.
- If you're shopping or searching online on 25 September, raise some free donations for the A-T Society by using www.days.org.uk and www.easyfundraising.org.uk and don't forget you can raise money for us simply by using www.everyclick.org.uk as your search engine.
- Hold a coffee morning
- Put flyers in your local supermarket, library, GP surgery, or hospital
- Wheely Wobbly Walk, Victoria Park – (see page 26 for more details)

We can support you with:

- Leaflets
- Sponsorship form
- Awareness day flyers
- T-shirts, balloons, and more
- Collecting tins and buckets
- Personalized local leaflet with your name and telephone on it

Join us on Facebook!

The A-T Society has set up a new Facebook group as a place to chat to others affected by A-T and keep up to date with all our news and events.

To join us go to www.facebook.com and search for AT Society. Don't forget to invite your friends! The more members we have the more awareness we raise.

We've currently got 286 members - let's make it to a thousand!

Spotlight on Fundraising

There have been many busy people raising money for the A-T Society this year. The next few pages show their fantastic efforts. Without their help, the Society would not be able to support families and fund research, so a big thank you to all fundraisers. Remember to log on to the Facebook group page for the latest fundraising news.

L to R – Sgt Charles (Charlie)-Driver, Sgt Revill (Rev), Sgt Mosedale (Mozz), Capt Williams (Steve), Sgt Smith (Gary), Sgt Preston (Presh) and SFC McDonald (Mike)-Driver complete the Three Peaks Challenge

Three Peaks Challenge

Almost £6,000 has been raised to date by a team in Dorset who have been raising funds jointly for the A-T Society and Help for Heroes. Rachel who is also co-ordinating the collection of sponsorship, kicked off the fundraising by holding a stall in the Dolphin Centre in Poole with David Owens (photo on top of next page). Both also attended an 'Evening of Games' held at the Barracks of the AFV Gunnery School in Lulworth. For the finale, a team of eight soldiers took part in the Three Peaks Challenge which they completed in 19 hours and 31 mins. Well done to all the team for their support.

Right - Rachel and David kicked off the Dorset fundraising in the Dolphin Centre, Poole

Support fund

The Society recently helped Beth purchase a new Hippocampe Chair, which can be used on all different terrains such as the beach and woodland – looks great fun!
http://www.hippocampe.co.uk/

Jo Betts Sainsbury's Bag Pack

£310 was raised at J Sainsbury's from a bag pack. Photo from left to right: Frances Marriott, Viv Betts, Sue Cook and Janet Stafford, who organised the event (photo below)

Swimathon

A swimathon took place in March at St Albans Swimming Pool in aid of the Mayor's three chosen charities of which the A-T Society is one.

Shown in the photo is The Mayor at the front plus four swimmers from Roundwood Park Secondary School in Harpenden.

Donation

£500 received from the Royal Bank of Scotland. Cheque presented to Maria who has A-T. Also in the photo is Maria's brother Ben who also has A-T and Kate, a friend of the family. Kate raised funds for the Society last summer and the RBS agreed to offer their support to Kate.

Rachel Poupard sky-dived for A-T on 13th April and raised over £800. She wishes to thank all the families who so very kindly sponsored her

Marathon success

Emma Ross completed the London Marathon in a time of 3 hours 38 mins and raised around £800 for the Society. Well done to Emma!!

Line Dancing

Thanks to Sheila who organised a Line Dance in January and raised £2,000.

Goaties!

Nick Keith sent in £260 raised by staff at Tesco Chatham by growing 'Goaty' beards!

School collections

£1,000 was raised from Campion School in Northampton in memory of Ryan Lecky. Another £1,000 was donated by Our Lady of Victories Primary School in Putney.

'Wool News'

Thank you to the 'Wool News' who regularly send in funds raised from the collection tins they have on the shop counter.

Raffles with a twist

Well done to Mrs Wills who decided to do something a little different and raised £30 with an Easter raffle and £25 with a Knicker raffle.

Coffee morning

Lorraine Birch raised £461 at her coffee morning which was held in November.

Evening of Entertainment

Lian (a trustee of the Society) held an 'Evening of Entertainment' last November, which included an Elvis tribute, a Saxophonist, an auction and a raffle. It raised an impressive £6,250 for the Society.

Shown left is Lian's husband Shane winning a gold thong in the auction!

Money in Lieu of Flowers

Recent donations made to the A-T Society in lieu of flowers, in memory of the following:

- **Mrs Peggy Alder**
- **Mr Bernie Spear**
- **Uncle Ken**

Wheely Wobbly Walk

Victoria Park, Sunday 27th September

This year we are collaborating with Ataxia UK for the 'Wheely Wobbly Walk' and we want as many people as possible to join us.

You can walk, wheel, or wobble for Ataxia Awareness. We will be kicking off at 12 noon. It is just £5.00 to enter but we will waiver this cost if you obtain sponsorship for us.

Give us a call and register today.

We really need your support!

If you can't join us at Victoria Park, why not set up your own walk. Setting up a walk is simple, and anyone can do it

Shopping for A-T: Fundraising at your local supermarket

By Lian
A recent fund-raising weekend at a supermarket by one of our families, raised several hundred pounds!

Here's how to do some easy fund-raising:

Contact a local supermarket store and organise a date for the collection.
(Most councils do not need to authorise such collections as they are on store property – but it is worth a phone call to check.)

Ask some volunteers to help you.
Make a rota so that the help is spread out during the day.
(2/3 volunteers at a time doing a few hours each.)

Put up posters by the trolley bays especially those nearest to the store, by the cash machine, on the entrance doors and other prominent positions.

Locate your collectors inside the foyer and outside too.
(A surprising number of people will make their donation once they have put their shopping and trolley away and had time to get some coins together.)

Apart from standing for a few hours, this is a really easy collection method. It raises awareness of the condition and valuable unrestricted funds for the A-T Society, much needed at the present time.

Contact Angie (01582 760733) for further information.

Cheque presentation

£150 was raised by the Bulwell branch of Nottingham Building Society, through a Christmas raffle and collection tins held at the branch. Receiving the cheque in the above photo is Kaid (who has A-T) with mum Jo and older brother Ben and staff from the Nottingham Building Society.

Radio 4 Appeal
On behalf of the A-T Society
Sunday 15 November
07.55 & 21.26
Thursday 19 November
15.27
Put the date in your diary now and tell your friends

Thinking of going on holiday? Unsure about finding somewhere wheelchair-friendly or having enough support? Here are some ideas recommended by families and the Society:

Caudwell Children
One of our families went to Florida with 'Caudwell Children'. The family had a wonderful time, everything was taken care of and paid for, including 24 hour medical assistance. They run holidays once a year and families can apply directly to them.
www.caudwellchildren.com

Scout Holiday Homes Trust
The Scout Holiday Homes Trust is a registered charity that provides affordable, self-catering holiday accommodation for families, carers or groups who have a member with a disability. They offer specially adapted and well equipped holiday homes.
For a free colour brochure phone 020 8433 7290 or visit **www.scouts.org.uk/holidayhomes** or email **scout.holiday.homes@scout.org.uk**

Willow Foundation
The Willow Foundation supports disabled young adults between the ages of 16 and 40 through the provision of special days. The objective of any special day is to help young people share quality time with family or friends, pursuing an activity they can all enjoy. Every special day is entirely individual and of the applicants choice. Past special days have included tickets to West End shows, pop concerts, city breaks, country retreats and health spa days.
To apply, and for more information, please contact the Willow Foundation on 01707 259777, or visit the website **www.willowfoundation.org.uk** to download an application form

Kids Direct Short Breaks
This organization offers short breaks for families with a child or young person with a disability, as well as 1 to 1 support for disabled children/ young people
www.directshortbreaks.org.uk

Dale's Holiday Diary

North Wales, April '09

Dale Phillips enjoying the sunshine at the side of the river in Llanrwst

My short break in the lovely village of Llanrwst, North Wales began on Saturday 25th April when at 11 o'clock my carer, a guy called Dalpat, picked me and our van up and we hit the road. We soon reached our destination; we couldn't go to our chalet until 3 o'clock so we explored the village, found a small supermarket and bought some supplies. We headed back to our chalet and once inside, my carer made us both a drink and a sandwich, then we had a lie down. At about 8 o'clock, after a couple of hours sleep, we headed for the pub for a few beers!! I bought a fish & chip supper and we ate it back at the chalet, then went to bed at 11 o'clock.

The following day, we decided to go to Llandudno. We walked along the pier and I bought some postcards & had an ice cream. It was a very good day. The weather was a mixture of sunshine and showers but it didn't spoil the day. We headed back at about 4 o'clock and had some chicken and mushroom pie, before going to the pub. We then went and got burger and chips takeaway!

On our last day, we decided to go to Colwyn Bay. There wasn't a lot to do so we just drove around the town and went along the sea front before heading home. I thoroughly enjoyed the weekend and might go again next year.

Contact a Family - Holidays, play & leisure - information for families

'Contact a Family' has created a guide which provides information about the play and leisure options available for disabled children, and suggests ways access can be improved. It offers information about arranging these holidays and gives details of organisations that provide grants. Download a copy here **www.cafamily.org.uk/pdfs/holidays.pdf** or call CaF direct on 0808 808 3555.

5th Polish A-T parents and children meeting (in May)

Wonderful wedding

Congratulations to Nuriye Cevik and James Kramer who recently married in London. Nuriye's brother Erdal has A-T and is pictured here in the middle, next to the happy bride and groom. Kay Atkins was very fortunate to attend the wedding and a good time was had by all!

In brief

Typing Assistance

Rupert Prokofiev has tracked down an item we think could help a lot of A-T young people who find using a keyboard (even a small one) very slow. Rupert's good at texting so it should make all the difference – Go to this link now & buy "cre8txt". It's only £50 **www.cre8txt.com**

Blue Badge safety

Link for a safe container for blue parking badges - some people might have had theirs stolen so this might help - **www.bluebadgeprotector.co.uk**

Exempt prescription charges

Professor Ian Gilmore (President of the Royal College of Physicians) is undertaking a review of prescription charges specifically looking at how to exempt people with long term conditions from prescription charges and how this exemption can be phased in. Exemption for people with long term conditions was announced by the PM last year – follow this link for more information **www.dh.gov.uk/prescriptionchargesreview**

Linedancing anyone?

Lian is holding a line dance in October and another in Jan '10. Would you or anyone you know like to join? Contact the office for more info.

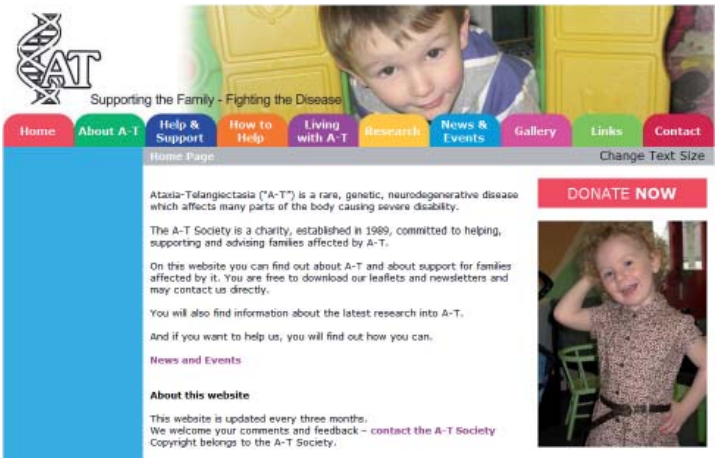
Turn 2 Us Benefit Advice

Would you like to find out more about benefits & grants? 'Turn2us' is a new independant charity that helps people access the money available to them – through welfare benefits, grants and other help. Their website has been created to help you find sources of financial support, quickly and easily, based on your particular needs and circumstances. **www.turn2us.org.uk**

Why make a Will?

Making a will gives you peace of mind - you can continue to care for your loved ones after you've gone and you decide exactly what happens to your money and belongings. Just knowing that you are helping to secure their future is of great comfort. If you also choose to leave a gift to the A-T Society you will be helping us to continue to improve the quality of life of children and young people with A-T. Your gift need not be huge, a modest sum of a few hundred pounds or just a small percentage of your estate would be a great help to the Society.

Every gift in every will can help us continue to support those affected by Ataxia-Telangiectasia. Please remember us too!



The new A-T Society website is live!
http://www.atsociety.org.uk/

Benefactors 2008

Many thanks to all our benefactors in 2008:

Aidis Trust
Maureen Ainsworth
Sylvia Aitken Charitable Trust
Mrs M Akerman
The Alchemy Foundation
Mrs MA Alexander
Eric Allen
Ms J Allison
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The Astor Foundation
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The Cotton Trust
Coutts Charitable Trust
Jane Crocombe
D & C Crosley
Mr JM Dean
Mrs K Dhirani
In memoriam - Anupam Dhirani
DM Charitable Trust
Mr & Mrs BB Dodge
Martin Doherty
Mr & Mrs LT Dorins
D Dorrell
Doreen Driscoll
Gwen Duff
The Eranda Foundation
Evangelistria Philoptohos
JE Evans
Mr L Evans
The Eveson Charitable Trust
Mr & Mrs I Farnsworth
Miss H Fenton
J & N Ferguson
Findel plc
The Fitton Trust
Mrs N Fitzhugh
DS Forsyth
Sister Francis
The Hugh Fraser Foundation
N & P Frost
Mrs PE Gaunt
Rev & Mrs DJ Gilchrist
Mr P Gilderoy
Penelope Gluckstein Charity
The Good Neighbours Trust
Ms Kate Gordon
Gordon Fraser Charitable Trust
The Gray Trust
Prof MHL Green
Mrs LG Green
Mrs Greenwood
Mr RF Griffin
Mr & Mrs PG Hammond
Mr & Mrs A Hammond
The Best of Harpenden
Mrs C Hartill
Z Hathaway
Havan Ladies
Mr & Mrs MS Hawkes
Hempshill Hall Primary School
Mr HW Henderson
Mrs A Hendrick
Mrs D Hewes
Mrs S Hewitt
Mrs E Hewson
M Higginson
Mr & Mrs G High
Louisa Hill
Mrs J Hill
Mrs D Hill
The Lady Hind Trust
The Jane Hodge Foundation
In memoriam - Joan Holgate
In memoriam - Vicky Holliday
Mr C Hollingdale
The Lady Hornby Trust
Mr & Mrs Horne
The Howard Charitable Trust
Howdens Joinery
Mr T Hughes
Mr & Mrs MJ Hunt
Samuel Hunt 2 Lodge
Icknield Scouts
Sir Robin Jacob
Mr RSD James
Rev MP James
Miss EM James
Lady Jarvis
Jarvis Harpenden Charitable Trust
Mrs Maureen Jenkins
Jivers '96 in memory of Gregory & Caroline
Mrs KE Keay
Mrs Maria Keily
Mr & Mrs H Kelliher
Mrs P Kemp
DM Kenny
RC Kingsley
The Christopher Laing Foundation
Ms Kelly Lammyman
In memoriam - Faye Larkin
A & S Lass Charities
Mr & Mrs S Last
Mrs Teresa Lawson
In memoriam - Ryan James Lecky

Mr & Mrs S Lloyd
The Lloyd Fund
London Law Trust
Mrs KS Luke
Mr S Lynn
Sophie Lynn
Mrs P Lynn
The Lynn Foundation
Mace Ltd
In memoriam - Billy Manley
Marsh Christian Trust
Mr & Mrs GW Marshall
Mrs M Matthews
Matthews Wrightson Charitable Trust
The Maunsell Family
Mayor of St Albans' Charity
Mrs A McCann
Ms Vera McDonald
Mr M McGrath
SA Meadows
Tony Metherell Charitable Trust
Hans & Elizabeth Meyer
Michaelis Charitable Trust
Mid Kent College
AJ Midwood
Miss V Moore
Jim Moore
Mrs SF Morel
Morgan Crucible Charitable Trust
Morrish & Partners
In memoriam - Aaron Murch
Mr P Murphy
Murphy-Neumann Charity
LA Nesbit
Nottingham West Music & Drama
The Oak Trust
Mr & Mrs Oates
Mr & Mrs K O'Brien
The Oppenheimer Charitable Trust
Carl O'Sullivan
Our Lady of Victories School
PR Owens
RC & JM Owens
Mr & Mrs D Owens
T Parker & J Shand
RH Parsons
Sara Pasalodos-Sanchez
Mr TW Paterson
Constance Paterson Charitable Foundation
Mrs VM Pearce
Mrs MJ Penelrick
Mrs S Perry
Mrs C Peters
Pettit Charitable Trust
Dale Phillips
Mr & Mrs T Phillips
Points of Light
Mr & Mrs ML Poupard
Mr & Mrs PB Poupard
Mr & Mrs PV Poupard
Ms K Preston
Mrs FA Prokofiev
Quorum singers
Redbourn Methodist Church
Redrock Financial
Reuben Foundation
DL Rhys
Miss S Ridley
The Rind Foundation
The Roan Charitable Trust
The Sir James Roll Charitable Trust
The Rowan Charitable Trust
AE Rowley
Royal Union Public House
JD & HS Sabel
J Sainsbury plc
Mrs LK Saunders
MH Senanayake
Patricia & Donald Shepherd Charitable Trust
Mr K Sherry
SK Shuttleworth
Mr & Mrs AAL Silver
G Simpkin
In memoriam - Alan Slater
Mr R Smith
Sobell Foundation
JI Soper
The Souter Charitable Trust
South West Charitable Giving
Southdown Methodist Church
FM Spalding
Richard Speller
St George's Vaults
St John's Club
BJ Standing
JSE Staughton
The Steel Charitable Trust
A Stewart
Mr C Stewart
Ms S Stewart
Sure Start Barkerend Children's Centre
Mr RW Tennuci
Staff of Tesco Chatham
Glenday Thomas "A Measure of Scotch"
Tivoli Stores
Staff of Steve Turzynski
Mrs Zeenat Ulnisa
Ulster Bank
United Reformed Church Fordingbridge
Mr M Wainwright
Mr & Mrs Walmsley
Mr & Mrs JB Warren
Mr & Mrs C Waters
Mr DCE Watson
Mary Webb Trust
Mr R Wells
Mrs JE Wells
Kay Wells
Wesleyan Assurance Society
Mr WJ Wheeler
Mrs Helen White
Mr Whitehead
Mrs Teresa Whittle
Mrs JM Wilkins
William Hill
Mrs A Wills
In memoriam - Barbara Jeanette Wills
Ms Marion Wilson
Mr JB Wingrove
The Anona Winn Charitable Trust
Pam Woodford Smith
Mr JG Woodward
Miss K Wright
Mrs Pam Wright
The Xerox (UK) Trust
Mrs Lian Yarlett