

the Ataxian

THE MAGAZINE FOR PEOPLE AFFECTED BY ATAXIA

Spring 2011
Issue 174



Sharing the Journey

Supporting our runners at the Brighton Marathon, April 2011

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The Ataxian

We welcome your stories, pictures, letters and articles for the magazine. If you would like to submit anything please do contact us.

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The magazine is also available in large print, audio and email versions. To change your preference just contact us at the office.

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Contributions to the magazine are welcomed but the editor's decision is final and will depend on space and other factors. Pictures contributed will be returned on request and must be a minimum of 300 dpi for use.

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Take Action

Apply by **18 July** if you are interested in becoming a member of the Trustee Board, see the inserts.

Book now for the Regional Conference to be held in Glasgow on **2 July**, call the **Ataxia UK office**

Bookings for the National Conference open on **15 June**, see pages 4 & 5.

Submit questions to the office for the **doctors Q and A** at the National Conference

Nominate someone whose contribution you have valued for the **Anne Ford Cup** and the **Ernie Heath Shield**.

2011 Raffle Winners

Congratulations to our raffle winners and a big thank you to all who sold tickets! The winners:

1. James Ray Cobb 6456 - £250 Cash
2. Judith Turner 10456 - Picnic Hamper
3. Sheona Whatford 972 - Aspinal Diary
4. Margaret Malcolm 12580 - John Lewis Voucher
5. Francoise Snell 3863 - M&S Voucher
6. Gaz Harris 25865 - Paintball for 8
7. Mr & Mrs Humphrey 5166 - Teddy Bear
8. Sadie Hooley 12192 - Message To Space

We are sorry to say goodbye to two staff members at Ataxia UK. Claire McGowan has moved on to pastures new and Nina Roskamp has completed her Internship (see back page) and returned to Germany.

Claire has been with Ataxia UK for 2 years and was instrumental in developing The Ataxian, revamping the website and improving our communications.

As a result of Claire's departure there has been a re-organisation within the team. Izabela Seabrook has taken on the role of Fundraising and Communications Manager and Mike Garrett has become the Communications and Events Officer. We are delighted that Madison Bridgewater has now joined Ataxia UK on a full time basis as the Fundraising and Events Co-ordinator.

We are also pleased to welcome Sophie Smith who started in March as an Intern helping Tina Thatcher to raise awareness and develop our membership among black and ethnic minority communities.

Election of Trustees: call for nominations

Inside this issue you will find two inserts relevant to the nomination of new Trustees. The first details responsibilities, qualities and process involved, while the second is a form that needs to be completed and returned to Ataxia UK offices by 18 July 2011 (address on opposite page).

Ballot papers will be circulated in the next issue (175) and results of nominations will be published in the December edition (176).

Jerry Farr Travel Fellowship 2011 Winner

Our congratulations go to Geraint Williams who is the winner of the 2011 Jerry Farr Travel Fellowship.

Geraint says 'As you can imagine I am absolutely thrilled to receive the grant. The plan is a three city stop of the USA to include Washington, Atlanta and New York. I will be accompanied by my wife and two children. We will be visiting sites like the White House, Georgia Aquarium, Statute of Liberty and the Niagara Falls. The trip is likely to go ahead in August 2012'.



National Conference

Leicester 8 October 2011

Ataxia UK is pleased to announce the details for this year's Annual Conference to be held at the Marriott Leicester Hotel.

The event will take place on **Saturday 8 October 2011** and we hope it will be as popular and well received as the 2010 Conference held in Stansted, Essex.



Sharing the Journey: theme for 2011

Bookings for the conference and dinner will be taken from **15 June** either via the Ataxia UK website: www.ataxia.org.uk or by contacting the office directly on **0207 582 1444**.

The cost of the conference including lunch and refreshments is £20 for those with ataxia and £25 for those without. The cost of dinner on Saturday night is £30. Should you wish to join an informal buffet on Friday evening the cost would be £8. All these figures include VAT. Children under 12 will be able to attend for free.

We have taken into consideration the feedback we received from Stansted and are offering a range of accommodation options that should suit every budget. The main accommodation will be booked at the Marriott Hotel and additionally we have reserved rooms in some neighbouring hotels that offer more affordable accommodation. This year we are asking delegates to book their accommodation **directly** with the hotel of their choice. The list of hotels and prices are as detailed below.

It is always challenging to find a hotel with sufficient adapted room facilities to meet all our needs. We have reserved all the 12 adapted rooms at the Marriott as well as adapted rooms in the nearby Hilton Hotel. The Premier Lodge hotels also have adapted rooms but only with double beds. Should you wish to reserve an adapted room at any of the hotels please contact the Ataxia UK office who will help manage the allocations according to people's particular needs but with consideration to a 'first come first served' allocation. **The deadline for contacting the office is 15 July to reserve adapted rooms.** Thereafter adapted rooms will have to be booked on a strict availability only basis directly with the hotels.

Hotel	Tel	Room Rate inc VAT	Distance to Marriott	Adapted rooms
Marriott	0116 282 0100	£90 double occupancy, £70 single, B&B		12
Hilton	0116 263 0066	£90 double occupancy, £70 single, B&B	1 mile	6
Premier Inn, Braunston	0870 197 7154	£45.50 room only	1.5 miles	3 doubles, no z beds, twins or interconnecting
Premier Inn, Braunston South	0870 850 6332	£45.50 room only	2.1 miles	4 doubles, no z beds, twins or interconnecting
Premier Inn, City Centre	0870 111 2844	£53 room only	4.9 miles	9 doubles, no z beds, twins or interconnecting

Theme and Agenda

Based on some of last year's feedback we will be including more items that recognise the important role that carers play, hence the theme 'Sharing The Journey'. There will be a full children's agenda including a crèche in the morning and a trip out to a local place of interest in the afternoon. For teenagers and younger people there will be an exciting opportunity to make a video during the conference. As well as the usual updates on the performance of the charity there will be reports on progress being made in research; the doctors Q and A; personal stories about how people live with ataxia; and benefits advice and therapies.

Come along, meet old friends and make new ones.

Bursaries

As usual Ataxia UK has set aside some funds to help those who may need some funding to help them attend the conference. Each bursary will be up to 50% of the total cost to attend the conference for people with ataxia, their parents or their carers. This will also include travel costs via public transport or equivalent and will be paid against receipts. Funds are limited so please do apply as soon as possible but **15 July** by the latest. Contact the office for an application form or download a copy from www.ataxia.org.uk.



The Children's crèche 2010

Roald Dahl Grants for families affected by ataxia

We have a grant from Roald Dahl's Marvellous Children's Charity which will provide limited funds this year to help families a child under 18 with ataxia to attend the Conference.

This Grant may cover up to 75% of travel, attendance and accommodation costs.

If you would like to be considered for a Grant please contact the Ataxia UK office by **1 July**. Funds are limited and priority will be given to those families who did not receive a Roald Dahl grant in 2010. *The Roald Dahl Grant can not be taken in addition to an Ataxia UK bursary.*

Regional Conference 2 July 2011

You can still book for this year's Regional Conference in Glasgow but hurry as it takes place on 2 July!

For last minute bookings please call the office on 0207 582 1444.



Dave with The Anne Ford Cup

Cup and Shield

Each year we present two awards to people who have made outstanding contributions to the charity and the lives of people affected by ataxia.

The Anne Ford Cup is awarded to someone who has ataxia. **The Ernie Heath Shield** is for someone who does not have ataxia. Last year's winners were Graham Kennedy for his fantastic Big Bad Bike Ride Fundraising, and Dave Stubley for his tremendous branch work and fundraising.

Who would you like to see honoured this year?

Please contact the office to nominate someone whose contribution you value.

Drs Q&A

We would welcome receiving any questions that you would like to ask to our panel of experts in advance of the conference. This will help us make best use of their expertise in the time available.

Please send your questions to: research@ataxia.org.uk or contact the office directly.

Service Update

Money Advice Service launched



The Money Advice Service, a new nationwide service to provide free unbiased advice to help people make the most of their money, was launched on 4 April 2011. The aim is to help individuals make the right financial decisions and act on them, by giving personalised online, telephone and face-to-face advice.

Find out more from:
www.moneyadvice.org.uk

- Are you having trouble claiming welfare benefits?
- Do you need information about other welfare benefits?
- Do you have ataxia and have problems with your employer?
- Do you have problems getting about in your home?
- Do you need some adaptations?
- Have you had your needs assessed?
- Are you having problems getting your voice heard?

If you have answered 'yes' to any of these questions our Advocacy Team may be able to help you.

If you have an enquiry or are experiencing any difficulties listed above, we may be able to help. Contact the Helpline: **0845 644 0606** or helpline@ataxia.org.uk who will put you in touch with our Advocacy Team.



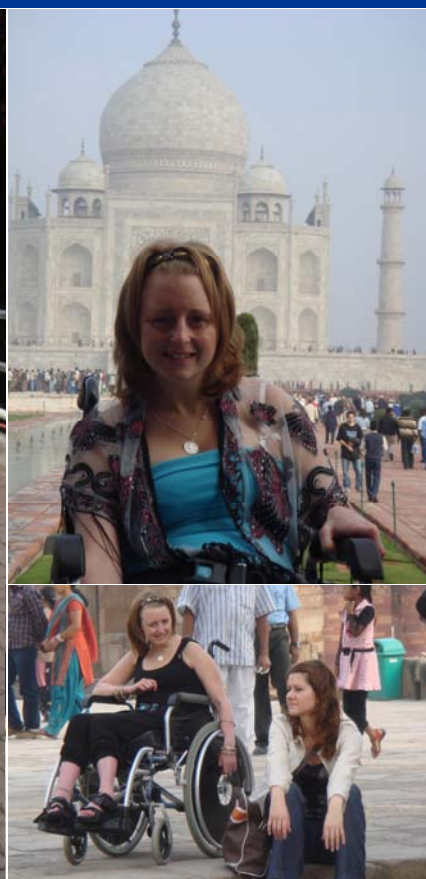
The Information Standard is a Department of Health accreditation scheme for health and social care information. It is a voluntary scheme and achieving the accreditation demonstrates that information is of the highest standard.

This certification helps people know information can be trusted - enabling better understanding of relevant issues and more informed choices about health and care. It requires organisations to produce information that is well researched and written, accurately sourced and referenced, balanced and impartial, and reviewed regularly to ensure continued relevance.

Organisations who have received the Information Standard accreditation are able to use the Information Standard 'kite mark' above.

In an effort to provide the best support to people affected by ataxia, Ataxia UK is seeking accreditation from the Information Standards and has begun the process to ensure all our practices are in-line with their requirements.

Jerry Farr Travel Scholarship 2010 Winner



The day I found out I had won the competition I was so excited and wanted to book my flights to New Zealand there and then. However with a bit more consideration and planning I found a tour that also included India, Thailand, Australia as well as New Zealand for only a couple of hundred pounds more.

This was fantastic news and I booked the tickets for myself and my carer Magda for my holiday of a lifetime.

I won't go into detail about all the wonderful places we visited as I am sure the editor would not be impressed with the length of the article.

However I'd like to share some of the real highlights with you:



My names is Millie Mae Ormsby, just call me Millie

I was born July 2001. I was fed up of crawling everywhere so walked at 10 months. At my first birthday party there was no way others were having MY spotlight so I danced all afternoon. I went to nursery from 12 months and learnt something new everyday, everywhere I went nobody could believe my age.

In February 2010 I was diagnosed with Friedreich's Ataxia. I do not really understand what that means but I do understand that I have to go to the hospital all the time. I have 2 surgeons, Mr Gardner for my back and Mr Glithero for my feet. I am going to meet a plastic surgeon for my hands this month. I have a Cardiologist for my heart, Dr Chickermane. He is lovely he always gives me the whole morning when he does my Heart Echo. I have Dr Wassmer for my nerves, she is lovely too and sponsored my mum £20 for a bungee jump. I also have Charlie my physiotherapist. I'm getting a special bed that will help me get up in the morning, a lift for the bath, and a special toilet.

I have to take 2 tablets everyday for my heart and 6 for my nerves, this is on a normal day, when I was in pain I was taking 24 tablets a day. In April 2010 I had spinal surgery; I have 2 metal rods in my back and it took Mr Gardner 7 hours to do this. I do feel a lot better now. I have to go back every six months and have them adjusted as I grow up.

All this does not stop me doing things, last week I went rock climbing with my outdoor activity group and I climbed higher than some of my friends. I do have a fantastic life; they know my name in Toys R Us! I have more pets, go more places, and do more things than anyone I know.

My family and friends are doing lots and lots of fundraising to support me and Ataxia UK. To find out more please visit: www.milliemaesworld.co.uk.

Toyah Wordsworth tells us about her journey...

Visiting the Taj Mahal in India was truly magical, it is such a majestic and spiritual place; eating curry for breakfast – naughty but nice; going into the rain forest in Cairns, Australia by cable car; snorkelling in the Great Barrier Reef and seeing Andy Murray in the Australian Open; having a BBQ on the beach in New Zealand for my 30th birthday: now that's not possible in England in February!

Everyone asks me what it was like for someone in a wheelchair to go on such a trip. It was fascinating to see how different countries react to and support a wheelchair user. If you are looking to find a disabled toilet in India then you are having a laugh. The disabled facilities there are very poor but everyone was so helpful even though at first they would stare at you in amazement; it is not usual to see a white person in a wheelchair. In Thailand the access was still not great but it was better than India. However, the local people were brilliant and couldn't do enough for you. It really is the land of smiles. In contrast Australia and New Zealand had much better access facilities but the attitude of local people varied so much from place to place. In Sydney for example people would sit and watch you struggle where as in Melbourne they couldn't be more helpful.

This trip really helped me get to know Magda better. It was challenging for her to move me around everywhere I wanted to go and I am so grateful for her support, I couldn't have done it without her and with the help of all the wonderful people we met. My only regret is that I didn't get to swim with the dolphins but other than that I had a marvellous time and would urge anyone thinking about such a trip to give it a go. I couldn't have done this though without the Jerry Farr Fellowship so my most sincere thanks go the Farr family for the best experience of my life!

Fundraising Thank you



Adam Leary (right) hands over the trophy for the Ataxia UK football tournament, held in aid of his London Marathon fundraising efforts. Congratulations to the winning team Real SoSoBad.



Marathon Achievements

Congratulations to all our runners: Adam, Hannah, Jason, Karen, Kate, Katherine, Nick, Sue and Verity in the London Marathon. along with Beth, Glen, Joanne, Kathryn, Sandra, Tyler in the Brighton Marathon. All completed the gruelling 26.2 miles in April this year! Their combined total raised is over £20,000 so far!

THANK YOU



If you would like to take part in a marathon or any other challenge or event for Ataxia UK contact the fundraising team today on
020 7582 1444
or email
fundraising@ataxia.org.uk



Chance to Win winners: £250 Hilary Huffell; £150 Judith Margolis; £100 Christopher Thomsett



Thank you to Steve and Aky who have just completed the Land's End to John O'Groats cycle ride.



Al Fox takes a leap at Tamworth Bungee Jump organised by the friends and family of Millie Ormsby. A lovely day was had by the 21 jumpers and spectators. Thank you Al & co!

Dates for your diary

Saturday 9 July **Swing Low Sweet Charity** www.sweetcharityonline.org.uk

Saturday 17 September **Big Bad Bike Ride** www.bigbadbikeride.com

Sunday 25 September International Ataxia Awareness Day

Sunday 30 October BBC Radio 4 Appeal for Ataxia UK

Details of all upcoming events can be found on our website and on our Facebook page. If you would like us to promote your event, let us know... and let us know how you get on!



Incorporatewear in Birmingham raised £2078 through a charity day and sponsored walk.



Merja and Kevin after completing the Reading Half Marathon.



Tami at the end of the Blackpool 10k.

We are reviewing our fundraising packs and gathering ideas for International Ataxia Awareness Day. Please get in touch to request a copy or share ideas!



Q: I have episodic ataxia type two (EA2), and have noticed that when I get hot, it can trigger the ataxia. What is known about this?

Dr Giunti: Research has shown that cells containing mutations in the same types of proteins that are mutated in episodic ataxias types 1 and 2 (EA1 and EA2) are much more susceptible to internal temperature changes. We also know that fever can trigger ataxic attacks in people with EA1 and EA2.

Less is known about the effects of outside temperature, but these are very rare conditions and we are still learning. Every day we discover something new and the information about what triggers attacks comes from the people who live with these conditions.

Dr Baxter: I agree; anecdotally many people have said that things like heat and stress and mild head injury can trigger attacks.

Dr Giunti: Caffeine and stress are also common triggers.

Q: Brain imaging and scanning - has the technology improved so that it can give more information than brain donation?

Alison Stevenson*: Some researchers are investigating brain imaging techniques in ataxia. MRS (magnetic resonance spectroscopy) is a non-invasive imaging technique that can be used to measure metabolites in the brain and thereby assess its function, and MRI (magnetic resonance imaging) measures the volumes of different brain regions, as possible ways of distinguishing between different types of ataxia and measuring progression of ataxia. Recent research indicates that it may be possible to use these techniques to distinguish between different types of ataxia and to measure the progression of ataxia. However, this is different from the sort of information that can be obtained from brain donation. Whereas imaging provides information about the structure and function of the brain and has the advantage of being able to analyse living material, brain donation can give more detailed structural and biochemical information. It is therefore likely that both methods will continue to be used to gain as much information as possible.

Dr Baxter: The microscopic structure of the brain can still only be seen through brain donation. Brain imaging is not yet able to look at the level of individual cells. As Alison says, the two techniques can partner each other and both contribute to knowledge.

Q: Are there any tests (eg MRI scans) that would detect the progression of the condition?

Dr Baxter: The best test is usually a clinical assessment by an expert in the field. Brain imaging can show changes in the cerebellum and other parts of the brain which correlate with changes in symptoms but the correlation is not always 100%.

* Dr Alison Stevenson (PhD) is Ataxia UK's Research Officer

Disclaimer

Please note that we publish this part of Ataxia UK's Conferences for information purposes only. Readers must seek their own medical advice before taking or refraining from taking any action based on the information contained in this document and nothing should be construed as medical advice given by the doctors, Ataxia UK or any of its officers, trustees or employees. No person shall have any claim of any nature whatsoever arising out of or in connection with the contents of this document against Ataxia UK or any of its officers, trustees or employees.

Raising Awareness of Ataxia

Help us disseminate the Ataxia UK Medical Guidelines and Ataxia UK leaflets to doctors. If you have an appointment with a neurologist or GP, they may welcome receiving a copy of the Ataxia Medical Guidelines. Why not take Ataxia UK leaflets to Neurology Outpatients Department and ask them for them to be displayed? (Copies of all our publications are available from the Ataxia UK office).



A tribute to David Gregory

We were deeply saddened by the loss of David Gregory at the age of 26. He ran the Sheffield support group.

David was a polite and wonderful young man who was so very keen to support people with ataxia. He was known by many and had made some very close friends from other branches and groups, and through attending annual conferences.

Lots of us had worked closely with David over the years and we have received many messages of condolence and praise for all his hard work and wonderful, positive, attitude to life. He will be sadly missed and we hope his legacy will live on in the Sheffield support group becoming a branch – which is what he wanted so much.

Our thoughts are with his family and many friends at such a sad time.



In April we held the 5th successful branch officers weekend attended by 24 delegates representing 13 branches. These events give branch officers the opportunity to meet one another and share ideas and best practice. They also give Ataxia UK an opportunity for consultation with the branches.

It was a very successful weekend with everyone participating with constructive ideas and suggestions. The results from the evaluation and feedback sheets were excellent, have a look at the results on the website!

The weekend covered a variety of topics, including Ataxia UK's business plan, fundraising ideas and applying for external funding, the young persons research project and reviewing the branch guide. Also throughout the weekend there was the opportunity for delegates to have training on how to download branch information onto the website.

If you missed it put it in your diary for next year!

Tina Thatcher

Ataxia UK Newcastle Branch by Jennifer Martin

I have lived in Newcastle all my life. I started my caring career at the age of six and by age eight had decided to become a Christian.

My son's partner has ataxia. Not having heard of this condition before, I quickly researched Ataxia UK and noticed there was no support group in my area. I made contact with Tina, Branch Development Manager and from then we kept in close contact, planning to set up a new group.

Our first meeting was on 11 April and I was delighted to see ten people turn up and witness the birth of the Ataxia UK Newcastle Branch. George Gibson took up the post of treasurer and is raring to go and I volunteered to be the Chair.



Our objectives for the Newcastle Branch are:

- To raise awareness of ataxia amongst health professionals and the general public.
- To disseminate Ataxia UK leaflets in the most prominent areas.
- To enlighten the community of the work of Ataxia UK and its Branches and Support Groups.
- To be here for newly diagnosed individuals.
- To help to alleviate the isolation and fear often felt by people with or affected by ataxia.
- To provide a friendly meeting place where people with or affected by ataxia can meet up.
- To create a stronger network for people with or affected by ataxia within the community.

To meet these aims we need to build-up funds in various exciting and fun ways.

Finding the best strategies, we will aim high, think big and pray for guidance that we will achieve our goals. Hopefully we will then be able to make a difference for those with and affected by ataxia now and in the future.

Items of Interest

Free guides

The Rough Guide to Accessible Britain Review by Alison Gregg

This is the 3rd edition of this book produced in association with Motability. It is free to Blue Badge holders and to Disabled Persons Railcard holders. Split into eleven regional chapters representing areas of Britain and Northern Ireland it details tourist attractions and days out in those areas.

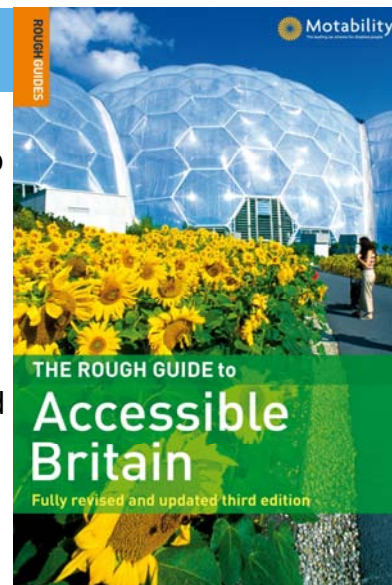
The Guide gives many ideas for days out suiting all interests and abilities. It is easy to use; each venue has been reviewed by disabled writers and has a key which allows the reader to see the range of accessibility features available at each site, including Blue Badge parking, toilet facilities, wheelchair access and audio-described tours and BSL interpreted talks and performances. There are contact details for each venue included in the guide.

There are over 180 accessible days out across the UK featured ranging from museums and art galleries to nature reserves to theme parks and zoos.

Venues include The London Eye, Churchill Cabinet Rooms and War Museum in London, RHS Garden Wisley in Surrey, ZSL Whipsnade Zoo in Bedfordshire, Legoland Windsor, Rutland Sailability, Albert Dock in Liverpool, RSPB Conwy Nature Reserve in Wales, The Deep Aquarium in Hull, The Eden Project in Cornwall, Chatsworth House in Derbyshire, Cairngorm Sled-Dog Centre in the Highlands, House For An Art Lover in Glasgow and Armagh Planetarium.

I found that the guide has given me lots of ideas for places to visit across the UK; places which I previously thought would be completely inaccessible. I've had to update my wish list of places to go and things to do! There is also a new version of the Rough Guide to Accessible Britain specifically for family days out and this is available only as an online guide.

It is available from: www.accessibleguide.co.uk or by phoning: **0800 953 7070**. An audio guide, with a special introduction by Ben Fogle, is available to download free from: www.accessibleguide.co.uk/guide/audio_guide.html



UK Road Atlas

The Concise UK Road Atlas is also free for Blue Badge Holders.

The atlas normally costs £12.99 inc p&p but The Pie Guide website is offering it to Ataxian readers for the p&p only (£3).

To order online go to: www.thepieguide.com and add the atlas to your basket. At this point it says the price is £9.99. You'll need to continue to the checkout and enter your personal details, when you've clicked to continue you'll see a box to enter a coupon code. There, enter the code **bbuk11**

(The total cost will then be £0.00. The usual postage of £3 will be added after).

If you'd rather send a cheque then you need to make it payable for £3 to: The Pie Guide.

You'll need to include the reference **bbuk11** with your name, address and telephone number to: The PIE GUIDE, Caledonia House 223 Pentonville Road, London, N1 9NG.

Please call 020 7952 0459 if you have any questions about your order.

A similar publication is available from The National Trust for visitors to their properties and gardens and is free to Members of the National Trust: **The National Trust Access Guide**.

If you would like to order a hard copy of this book please contact the Access office: accessforall@nationaltrust.org.uk or call **01793 817634**.

Please quote ref no 7330909 for standard print, or 7331709 for large print.

The Access Guide is also available free on CD or tape.

Opportunity to take part in research

We've reported in previous issues of the *Ataxian* about a large Friedreich's ataxia research project funded by the European Commission, called EFACTS. The UK is part of the study and there are now opportunities for people with Friedreich's ataxia to take part and help the research effort. This project is important as it will provide information to be used in future clinical trials. If you have Friedreich's ataxia and are willing to travel to the Ataxia Centre in London (travel costs will be reimbursed) you may be able to take part. For more information read below. This project has a new Clinical Research Fellow, Dr Michael Parkinson, who is contactable via Dr Giunti's office at the Institute of Neurology at the address given below.



What is the purpose of the study?

The European Friedreich's ataxia Consortium for Translational Studies (EFACTS) is a 4 year study supported by the European Commission that will gather vital information about the progression of Friedreich's ataxia (FA) which can be delivered to patients to give them a better knowledge of their prognosis and the development of their condition. The purpose is to generate a large FA patient database, alongside an integrated clinical and natural history database; this will be linked to a biological samples repository. It also aims to define a panel of clinical assessment tools for use in future trials. The UCL Institute of Neurology is one of the 15 groups from 7 European countries participating in the EFACTS project.

What will happen to those who take part?

If you do decide to take part you will need to attend an initial consultation where your formal consent will be sought for participation in the study, which will either be carried out at the National Hospital for Neurology and Neurosurgery or on a home visit. In the same consultation, participants will undergo a medical check-up where their Scale for Assessment and Rating Ataxia (SARA) score will be determined, 9-Hole Pegboard (9HPB) and 8-metre Timed Walk (8mTW) Tests will be performed, blood will be taken to confirm the genetic diagnosis of FA, and questionnaires will be filled out.

What are the possible benefits?

It is hoped that by participating in this study and as a result having patient data stored on the registry, it will allow patients to be informed promptly as to any relevant new research trials or projects that may be commencing which they may be eligible to participate in.

This registry will be a large European data set, which will allow access to a great number of FA specialists when potentially recruiting to new trials.

If you would like to participate in this study or know of anyone who may be interested, and would like some more information, please don't hesitate to get in touch:

Principal Investigator:

Dr Paola Giunti

Principal Clinical Research Associate and Honorary Consultant Neurologist

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Queen Square House

Queen Square

London WC1N 3BG

E: P.Giunti@ion.ucl.ac.uk

T: 02078373611 Ext: 3100

New research project funded by Ataxia UK

Researchers at Brunel University have been awarded a grant to investigate gene therapy as a possible treatment for Friedreich's ataxia.

Friedreich's ataxia (FRDA) is caused by a mutation in the frataxin (*FXN*) gene that leads to reduced levels of an essential protein, called frataxin, within cells of the body. This loss of frataxin is particularly damaging to cells in certain regions of the brain and spinal cord, heart muscle cells and pancreatic cells. Currently there is no suitable treatment for FRDA, particularly one that addresses the early stages of dysfunction.



Dr Michael Themis, Mr Hassan Knosari and Dr Mark Pook

FRDA researchers Dr Mark Pook and Dr Michael Themis will address this by examining gene therapy as a potential treatment. Gene therapy involves artificially introducing extra frataxin genes into the body with the help of viral vectors to supplement the defective gene, thereby increasing the frataxin protein levels in cells and correcting one of the earliest phases of the pathogenic process. Although this is still a relatively new technique in terms of the clinic, promising results have recently been seen in gene therapy clinical trials for other inherited disorders, such as a form of blindness known as macular degeneration.

However, before gene therapy clinical trials can begin for FRDA, important preclinical work needs to be done to investigate the safest and most effective method of delivery of the *FXN* gene. This project will use an FRDA mouse model, developed by Dr Pook and his team, to undertake this crucial pre-clinical work. The aim is to establish a gene therapy method that will create a stable, healthy level of frataxin protein in affected tissues in the mouse. The investigations will firstly use cells from the FRDA mouse model to assess the efficiency and potential side effects of the gene therapy. Then, the gene therapy will be tested in the FRDA mice and the safety, efficacy and longevity of increases in frataxin protein level will be measured. Improvements in FRDA-like symptoms of the mice will also be assessed. This will provide valuable information for considering potential future gene therapy clinical trials.

The principal applicants for the project, Dr Pook and Dr Themis, are based at Brunel University and are both well-respected in their fields of research. Dr Pook is a long-standing FRDA researcher whose group generated a mouse model that resembles FRDA, and Dr Themis' expertise is developing lentiviral vectors for gene therapy and testing their safety. Their combined experience will provide a sound knowledge base and valuable expertise for the project.

The work will form a PhD studentship which is being undertaken by a promising student, Hassan Khonsari, who recently completed his MSc in Dr Pook's lab and is keen to pursue this area of research. The project is being co-funded with The Friedreich's Ataxia Research Alliance in the US and the Friedreich's Ataxia Research Association Australasia; a total of £82,355 is being awarded over the three year period.

Further details can be found on the Ataxia UK website: www.ataxia.org.uk.

Many thanks to all those who responded to the request for volunteers in *Ataxian 173* and are taking part in the physiotherapy research project at Salford University.

Anita Watson, the principal investigator was really grateful for all the responses she received. If anyone is still interested in participating please get in touch with the researcher as she is still keen to include more participants: a.watson@salford.ac.uk or visit www.ataxia.org.uk.

Study looking at the prevalence of childhood progressive ataxias in the UK

A study investigating childhood ataxias in the UK and Ireland is underway. How many children have ataxia is currently unknown, but this project will collect data to give an estimate of the number in the UK and Ireland who have it.

The study is being led by Dr Peter Baxter, Consultant Paediatric Neurologist at Sheffield Children's Hospital. It is being funded by Ataxia UK & FASI, and supported by a grant from Takeda UK. The surveillance study will run for two years and will be conducted by the British Paediatric Neurology Surveillance Unit (BPNSU), an established research subsidiary of the British Paediatric Neurology Association (BPNA). Every month, the BPNSU asks BPNA members to report cases of certain neurological conditions via a secure website and anyone reporting cases of ataxia is asked to complete a short (one page) questionnaire requesting further clinical details. Responses are sent to Dr Baxter.

The data collected since February shows approximately 4 cases per month are being reported. We anticipate this figure will rise as more neurologists are encouraged to reply. To verify the accuracy of the data collected and eliminate 'double counting', it is hoped that results from the study can be cross referenced with other sources, eg genetic labs, which could provide numbers of diagnoses made in a specified time period. You can help by mentioning the study to your child's paediatric neurologist and encouraging them to take part.

Report of Ataxia UK's Medical Advisory Panel Meeting

The Medical Advisory Panel meets once a year to exchange research developments, discuss clinical practice and assist Ataxia UK in the work we do with improving the healthcare for people with ataxia. The 2011 meeting was held in March with 7 of the Medical Advisors. Updates were given on the paediatric study (above) and plans for a European paediatric ataxia database. The group discussed a recent paper on deep brain stimulation in people with essential tremor and its potential implications for people with ataxia.

Ataxia Centres were discussed and views were sought on the value of different models for these Specialised Centres. Ataxia UK is planning a Review of Ataxia Centres, eight years after the initial consultations that lead to their development. Dr Marios Hadjivassiliou presented some interesting data on diagnosis from his ataxia clinic in Sheffield. Out of 115 referrals to him from other neurologists without a specific diagnosis, he was able to give a specific diagnosis for 70% of them. A small number of these were treatable conditions.

Ataxia UK-funded researcher Dr Iain Hargreaves and his PhD student, Kate Duberley, presented their work on Coenzyme Q10 (CoQ10) deficiency. Dr Hargreaves talked of the potential implications of his research for clinical practice, including their work developing of a more sensitive diagnostic test to detect Coenzyme Q10 deficiency. He also talked about current arrangements at the Institute of Neurology for testing for CoQ10 deficiency. Doctors can contact him if they want testing for patients with ataxia with suspected CoQ10 deficiency.

We are very grateful for our dedicated Medical Advisory Panel and all the support they give us.

Spreading the word about ataxia research...

To help spread the word about ataxia research, Ataxia UK has awarded three small grants to support the dissemination of ataxia research among healthcare professionals and scientific researchers.

Betty Cassidy, a physiotherapist from Brunel University, was awarded £510 to attend the World Congress of Physiotherapy in June. Betty is studying for a PhD, having completed an Ataxia UK-funded research project on contemporary physiotherapy practice for people with ataxia. At the Congress, she will present and discuss a poster on the physiotherapy section of the Ataxia UK clinical guidelines (to which she was a major contributor) bringing the information to a wider, international audience.

Rachel Brookes, a final year medical student from Imperial College London, was awarded £500 towards her attendance at the American Academy of Neurology Conference in April. Rachel spent time in Japan working on a research project for spinocerebellar ataxia type 6 (SCA6) and developed an interest in neurology. At the conference she presented a poster of the SCA6 research. We look forward to hearing what Rachel does next.

£1000 was awarded to support an 'Iron-Sulphur Protein Biogenesis' meeting in August at Cambridge University. A session on frataxin, the protein that is mutated in Friedreich's ataxia, will look at frataxin mouse models and interactions of the frataxin protein to try and better understand its function. A number of Friedreich's ataxia researchers are expected to attend the meeting so it will be a good opportunity for them to share and discuss.

Photographic Project by Nina Roskamp

Here is a sneak preview of some of the photographs taken during our series of interviews with people living with ataxia.

They will be exhibited at the Romford Hospital in Essex, during the National Conference in Leicester and at the end of our International Ataxia Awareness Day celebrations in London.

Look out for details in the next issue!



Raising awareness amongst healthcare professionals

Ataxia UK has raised awareness amongst healthcare professionals by promoting an ataxia physiotherapy training day that took place in Bristol in April. The event, which was the first ataxia training day for physiotherapists, was organised by Dr Lisa Bunn. Dr Bunn recently completed her PhD (funded by Ataxia UK) looking at balance control in people with SCA6.

The training day was held at the same time as the AGM for the South West Branch of the Association of Chartered Physiotherapists Interested in Neurology (ACPIN). The talks gave an overview of cerebellar ataxia, reviewed the current assessment and management of balance problems in people with cerebellar ataxia, and looked at how research can be used to improve management of balance problems.

Graham Fickling, a former Trustee of Ataxia UK, attended the event and thought it was a very informative day that helped to raise awareness of Ataxia UK amongst physiotherapists. Graham was manning an Ataxia UK stall with leaflets and information for the delegates; he said the copies of the clinical guidelines went 'like hot cakes!' Thank you to Graham for his support on the day.

We would like to thank all who sent in donations in memory of the following people:

Mr B.Welch; Brenda Hall; Doreen Rimmer; Paul Rashbrook; Frank Wisbey; Kenneth Downen; Muriel Briggs; Diana Millington; Raymond Curtis; Michael Pimblott; Jack Lighten; Audrey Lovett; Dane Baekdal; Jean Titterington; David Gregory; Hans Roskamp; Leonard Walker; Lee Kennedy; Dennis Carmichael.

Leaving a legacy in a Will is one of the most enduring ways to make an impact on fighting ataxia. In the last ten years much of our research funding has come through the foresight and generosity of our Friends and supporters in leaving us a legacy. We currently have over 20 projects running, all of which are at least part-funded by gifts left in Wills.

Your legacy can mean hope for the future.