

the Ataxian

THE MAGAZINE FOR PEOPLE AFFECTED BY ATAXIA

Autumn 2011
Issue 175



Sharing the Journey

Getting together at the Branch Officers' Weekend

What's Inside

Issue 175, Autumn 2011

Update News	3
Conferences and events	4-5
Branch News	6
Services Update	7-9
Fundraising Thank you	10-11
Health Matters	12-13
Research News	14-18
Awareness Day	19-20



The Ataxian

We welcome your stories, pictures, letters and articles for the magazine. If you would like to submit anything to us or if we can help you in any way, please do contact us.

Ataxia UK

Lincoln House
Kennington Park
1-3 Brixton Road
London SW9 6DE

Office: 020 7582 1444
office@ataxia.org.uk
Fax: 020 7582 9444
www.ataxia.org.uk

Helpline opening times:

Mon-Thur 10.30am - 3.30pm
Friday 10.30am - 1.00pm

Helpline: 0845 644 0606
helpline@ataxia.org.uk
Text Connect 0779 7806436

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

Disclaimer

Information in the Ataxian is provided for information purposes only and while every care is taken to ensure its accuracy, no guarantee can be given. The mention of products, services, treatments or therapies in the Ataxian in no way constitutes an endorsement by the editor or by Ataxia UK. Individual medical advice should be sought before taking or refraining from taking any action based on the information contained in this publication. Nothing should be construed as medical advice given by Ataxia UK or any of its officers, Trustees, employees, advisors or volunteers. No person shall have any claim of any nature whatsoever arising out of or in connection with the contents of this publication against Ataxia UK or any of its officers, Trustees or employees.

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.

© 2011 All material is copyright and must not be reproduced without permission. Published quarterly by Ataxia UK.

Ataxia UK works across the whole of the UK and is a charity registered in Scotland (no SC040607) and in England and Wales (no 1102391) and a company limited by guarantee (4974832).

Take Action

Complete booking forms for the National Conference by **23 September**

Register your interest in the young persons' filming workshop

Submit nominations for the Anne Ford Cup and Ernie Heath Shield

Submit questions to Central Office for the Doctors' Q&A

Apply for tickets for the **Paradise Ball**

Plan your local **International Ataxia Awareness Day** activities around 25 September

Apply for Wheelchair Grants by **30 September**

Promote the **Radio 4 Appeal on Sunday 30 October** at 7.55am or 9.26pm to all friends and colleagues

Enter the pin badge competition by **16 December**

Trustee Board Elections

The election was uncontested as four valid nomination forms were received for the four available Trustee places.

They were from Alison Gregg; Peter Reeves; Howard Marshall and Alan Thomas who will all be proposed for membership of the Board at the AGM in October.

New Fundraising Packs

We have now launched the new fundraising packs, filled with great ideas and information to support your local activities.



You can request a copy from the Fundraising Team today.



Young Persons Contact Project

We have just launched a new volunteer-led project aimed at 18 to 25 year olds.

If you have ataxia and are aged between 18 and 25, and want get involved in youth activities with Ataxia UK, please write to Jaina Hirani, our volunteer, on jhirani@ataxia.org.uk

Christmas Cards

You will notice in the postal copy of this *Ataxian* an order form for this year's Childlife Christmas cards. Childlife is a charity raising money for a group of four children's charities, including Ataxia UK, so please do buy some today as it will benefit us directly. You can also order online: www.childlife.org.uk/support_us/christmas_2009.aspx

Case Studies

We are on the constant lookout for your stories so we can raise awareness of ataxia in local and national press. Please get in touch with the Central Office if you would be happy to be interviewed by a journalist.

Conferences and Events

Annual Conference, Leicester 8 October 2011

We are delighted with the positive response to this year's Annual Conference. You can still book although places are limited. The deadline is 23 September.

At the time of writing a few adapted rooms are available in nearby hotels and transport is organised to and from the Marriott Hotel. Please visit the Ataxia UK website to download a booking form or contact the office on 020 7582 1444.

The theme for this year is 'Sharing The Journey' and the agenda includes items that help and support the excellent work that carers undertake as well as people's personal stories about living with ataxia.



Sharing the Journey: Theme for 2011



The conference is a great opportunity to engage with other people affected by ataxia and to hear more about the latest developments in research. You can also take the opportunity to book 1-2-1 welfare and benefits advice as well as free massage therapy.

We are keen to encourage families to attend this year and will provide a crèche and activity centre in the morning for children aged three plus. There will be a coach trip to Bosworth Battlefield Heritage Centre in the afternoon. It will be very interactive and great fun as well as a chance to learn about our history and the last English monarch to be killed in battle!

Young persons' filming workshop

We will be running a young person's filming workshop during the conference as we are producing a video for young people affected by ataxia with support from Jeans For Genes.

We would like to invite young people (aged between 10 and 22) to help in the making of this video. This can either be by learning how to film on the day, helping edit or by telling your story. A professional production team will lead the filming and editing process and offer training and guidance throughout, allowing everyone to concentrate on having a great time creating their own video.

We are therefore on the lookout for interested young people to take part. If any young members of your family are interested in this project please get in touch with either Mike or Iza at the Ataxia UK office and we will be happy to explain more about it. The office number is 020 7582 1444.



Doctors' Q&A

We would welcome any questions you would like to ask to our panel of experts in advance of the conference. This will help us make the best use of their expertise in the time available. Please send your questions to: research@ataxia.org.uk or contact the office.

Ataxia UK Paradise Ball, Saturday 3 December 2011

Last year's Sparkling Winter Ball in the Royal Hospital Gardens Chelsea was absolutely fabulous and this year the Paradise Ball on the 3 December promises to be even more spectacular.

We have moved the venue to the prestigious Artillery Gardens of the Honourable Artillery Company in the City of London.



The evening will begin with a drinks reception followed by a delicious dinner from award winning *Create* and dancing in the exotic winter garden, evoking all the pleasure of paradise.

The auction and raffle provide a great fundraising medium for Ataxia UK and the "Win Your Weight in champagne" draw provides great excitement and fun for everyone.

Tickets are £85 if purchased before 3 October and thereafter they will be £100. Please contact the office if you would like to attend this great night out.

Anne Ford Cup and Ernie Heath Shield

Every year at the Annual Conference Ataxia UK awards prizes to people who have made an outstanding contribution to the charity. If you would like to nominate someone for either of the awards below, please contact the office on 020 7582 1444 or email: office@ataxia.org.uk

The **Anne Ford Cup** is awarded in recognition of a person with ataxia who has made an outstanding contribution to Ataxia UK and to people affected by ataxia. The **Ernie Heath Shield** celebrates an outstanding contribution made by someone who does not have ataxia.

Regional Conference, Glasgow 2 July 2011

It was great to see so many people at the Regional Conference held at the Beardmore Hotel in Scotland. Over 60 people attended and many stayed on for the dinner and quiz in the evening.

There were excellent interactive breakout sessions on Welfare Grants and Benefits, Fundraising and Communications, and working with the Neurological Alliance of Scotland in-between our regular updates.

Shona Heath shared her positive and energetic attitude which allowed her to travel and enjoy some fantastic experiences in South Africa. Nancy Collins gave an insight on the challenges she faced growing the Ayrshire Support Group into a Branch. A lively doctors' Q&A session helped build everyone's knowledge and understanding.

The weather was great all weekend and we had a chance to meet new and old friends. The feedback was clear that Ataxia UK should revisit Scotland soon. This we certainly plan to do.



Branch News

Tina Talk

West Country Branch

Our branch was started by Rebecca Downing, who has Friedreich's ataxia, in February 2006. She believed there was need for a support group in this part of the country.

Our aims are to meet and socialise with like-minded people, to support those who have or are affected by ataxia, to raise awareness and money, and most importantly, to have fun!

We are a small but friendly group, who meet four-five times a year for social activities. We produce newsletters several times a year so those who are interested can keep up-to-date with events.

The venue for our socials vary...so far we have had several boat trips from Exmouth up the River Exe, a visit to China Blue where you can paint your own pottery, a visit to Sidmouth Donkey Sanctuary and countless meals in pubs, always a popular option!

If you don't want to come in person, but wish to receive the newsletters, that's fine. If you are a bit shy, please be assured that we are a very friendly group who will welcome you and make you feel at home.

Give us a try — we would love to meet you.

Visit www.ataxia.org.uk for contact details



Tina Talk

It's that time of year when we look forward to annual conference and wonder where the year has gone since our last one.

This year's regional conference in Glasgow was well attended and we received lots of positive feedback. It's great to see so many of you taking up the opportunity to meet up with other people with and affected by ataxia and I really look forward to seeing lots of you in October in Leicester.

We will have a meeting on the Friday night for branches so we can discuss some important changes to the branch guidelines and a meeting on the Sunday morning for support groups to discuss any issues or concerns that you may have.

So get those booking forms in and I will see you all soon.

Tina

New Ataxia UK Pin Badge Competition

Calling on all creative Friends reading this issue of the magazine: we need your help in the design of a new pin badge for Ataxia UK!

Send in your drawings and ideas to the Central Office and win the chance to see your creation become a reality.

The deadline for entries is 16 December 2011.

Good luck!



Members of Ataxia East and Thames Valley enjoying time together at the MG Rally in May.

Did you know about... Individual Budgets - Resource Allocating System (RAS)

The idea of an Individual Budget was first developed and tested in Scotland by Simon Duffy in 1996. However it was the In Control programme that first persuaded local authorities to try using a formal Resource Allocation System for all social care budgets. The early success of this approach led to increasing levels of support from disabled people, local government and finally from central government. The initial model, developed by Simon Duffy was then amended by John Waters who proposed the application of a points-based system for calculating need. This enabled a whole range of further approaches which have enabled progressively more sophisticated and elaborate accounts of need and how such needs are related to funding. The purpose of the RAS is to tell people how much money they can reasonably expect to be made available through their personal budget. The RAS also specifies the individual's outcomes, based on need, that are to be achieved with that money.

By providing an allocation of resources and a set of key outcomes, the RAS provides a way of setting out the deal upfront using a simple and transparent set of rules.

So, what is the Resource Allocation System (RAS)?

The Resource Allocation System – or RAS - determines the value of a person's Personal Budget. It includes a Self-Assessment Questionnaire carried out by the allocated social worker to assess the need for social care support, for example for:

Access to the Community; everyday tasks; risk and safety; health risks; support needed; mobility; communication/sensory issues; mental health

The responses to each question are weighted to give a number of points. The greater the level of need, the more points a person will score.

Points are then converted into a Resource Allocation. As social care support is means tested, part of the process is to work out if someone should pay towards their own care. Once this has been done, the individual will be told how much their Personal Budget is.

How RAS works in practice

Using an assessment that provides a scored level of needs, and building up local intelligence about local costs, RAS (Version 5) provides a framework that can allocate appropriate levels of resources attuned to changing local market conditions.

RAS allows local authorities to use a simple, *scored assessment questionnaire* (SAQ), and link the scores to a set of locally-defined funding levels. This enables people who need support and the local authority to quickly and easily determine both the amount of money any individual needing support can reasonably expect in their Personal Budget, and what outcomes their support plan must address.

The SAQ measures the impact of a person's disability on their life in a number of key areas. Each area is scored and the total is adjusted according to the amount of support reasonably available to each person from their friends and family. In this way people with similar levels of needs in similar circumstances can be allocated the same level of funding, and guaranteed the same outcomes. Each level of funding is based on local intelligence about how much money has been needed for people in similar circumstances to meet their agreed outcomes in terms of the individual's amount of money awarded. This emphasis on agreed outcomes, with strong links to local costs and experiences of planning, ensures the system is robust and is seen to be fair and equal.

An overview of Universal Credit

Universal Credit is a new benefit that will be introduced in October 2013, replacing current means-tested benefits and tax credits for working-age people. From October 2013 onwards no *new* claims for existing out of work benefits will be accepted and will be treated instead as claims for Universal Credit.

People moving from out-of-work benefits into work will also transfer on to Universal Credit.

From April 2014 no new claims for tax credits will be accepted.

Between April 2014 and October 2014 all existing claimants of out of work benefits will be transferred to Universal Credit, but existing claimants will *not* lose out at the point of changing over. This means there will be transitional protection in the form of additional payments.

Who can claim?

Universal Credit will replace means-tested benefits and tax credits for working-age people up to pension credit age. It will be paid to people in or out of work. Many benefits will remain including child benefit, carer's allowance, contributory employment and support allowance, and contribution based job seeker's allowance. Although the later two will be aligned with universal credit. Listed below are the benefits that will be abolished:

Benefits that will be abolished:

- income support
- income-based jobseeker's allowance
- income-related employment and support allowance
- housing benefit
- council tax benefit
- child tax credit and working tax credit
- crisis loans and community care grants – responsibility for an equivalent will be passed to local authorities or devolved governments
- budgeting loans – to be replaced by payments on account (an advance of universal credit) in cases of need
- carer's allowance
- bereavement allowance, bereavement payment and widowed parent's allowance
- maternity allowance
- industrial injuries disablement benefit
- statutory maternity/adoption/paternity pay
- statutory sick pay
- maternity grants and cold weather payments – to be paid automatically when the qualifying criteria are met. According to current information available funeral payments will remain.



Conditions and Sanctions

There will be more conditions to satisfy and tougher sanctions for Universal Credit and for all existing benefits. All claimants will be required to accept a 'claimant commitment' as a condition of receiving Universal Credit. They will then be placed into one of four groups:

Claimants subject to no work-related requirements:

- people with limited capability for work-related activity because of health or disability
- lone parents or the lead carer in a couple with a child under one
- carers with regular and substantial caring responsibilities for a severely disabled person.

Claimants subject to the work-focused interview requirement only:

- lone parents or the lead carer in a couple with a child over one but below a prescribed age (yet to be advised)

Claimants subject to the work preparation requirement

- people with limited capability for work because of health or disability
- lone parents or the lead carer in a couple with a child aged three or four

Claimants subject to all work-related requirements

- everyone else – the default for claimants including lone parents and couples with children over the age of five.

Sanctions

Sanctions may be imposed on claimants subject to all work-related requirements, which includes failure for no good reason to comply with the requirement to prepare or apply for work, take up an offer of paid work, and ceasing work voluntarily or through misconduct. Other sanctions may be imposed on claimants subject to work preparation requirements for failure to undertake work-related activity, or in the work-focused interview group for failure to attend an interview. Hardship payments may be available for claimants who have been subject to sanctions and are, or will be in 'hardship'.

Right of appeal

It is expected that there will be a right of appeal against decisions concerning entitlement to Universal Credit, as with the current benefits and tax credits system. This is just an overview, but you can get the full factsheet on Universal Credit from our Helpline 0845 644 0606 or email helpline@ataxia.org.uk

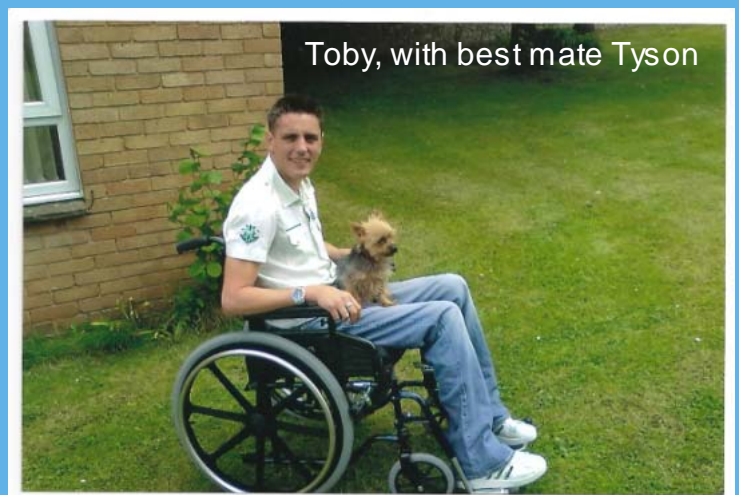
Wheelchair Grants: the next round of wheelchair grant applications for 2011 is open

Ataxia UK awards grants towards the purchase of wheelchairs for those who have not been able to secure funding elsewhere. The deadline for the next round of wheelchair grant applications is **30 September 2011**, so if you're interested and would like to apply, register your interest now by contacting the Helpline.

Maximum grant award is £1,000.

With help from our Advocacy team, Toby Keep was recently awarded an Ataxia UK Grant towards a new wheelchair.

Toby said *"I would love to say a big thank you for the grant for my new wheelchair, it has made a great difference to my life. Thank you, once again."*



Toby, with best mate Tyson

Fundraising Thank you

**A massive THANK YOU
to all our fantastic fundraisers up
and down the UK.**



The Bitteswell Over 40's team celebrating their successful 24-hour football marathon which has so far raised £1,500.

**We are currently taking
applications for the 2012
Virgin London Marathon and
Brighton Marathon.
Contact us today for a form.**



Jodi Gallagher (left) and friends climbed Snowden and Ben Nevis this summer raising £800.

Estelle Zauner-Maughan (right) before taking part in the British Gas Great North Swim.

Estelle donned her wetsuit for a mile long swim around Lake Windemere raising just over £1,000.



Julian, Andy, Dave & Ian completed the Lands End to John O'Groats Cycle raising a magnificent £9,358.



Simon, Neil, Chris & Dave took their car Bumblebee across Europe for the Italian Job Car Rally and raised over £400.

**Chance to Win winners: £250 Graham Dyer, £150 Debbie Poyser,
£100 Barry Luscombe**

Fundraising Thank you

Many thanks to Networkers International PLC for holding another amazing Golf Day in aid of Ataxia UK.

Former Chair of Trustees, Pit Rink, attended. She saw the raffle and auction, overseen by snooker legend Steve Davies (left) raise a fabulous £5,000.



Paul Howell was the only hand cycle participant in the 60 mile route from Blackpool to Manchester.

Dates for your diary

Sunday 25 September International Ataxia Awareness Day

Sunday 30 October BBC Radio 4 Appeal for Ataxia UK

Saturday 3 December Ataxia UK's Paradise Ball

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!



Stuart Meer ran 8.5 miles in the Great Midlands Fun Run and is getting ready to run the Great North Run in September.



Ataxia UK East Durham Support Group raised £707 with a stall at the Seaham Carnival. Their 'Lucky Straws' Raffle and Tombola proved to be a big success.

Ataxia training event for medical professionals

The first ataxia training day for healthcare professionals was held in Sheffield this May and was very successful. This event was funded and organised by Genetic Alliance UK in collaboration with Ataxia UK and the Sheffield Ataxia Centre. It was part of a Department of Health-funded research project conducted by Genetic Alliance UK. The project highlighted the need for increasing awareness and understanding of ataxia among healthcare professionals, which is also an ongoing aim of Ataxia UK.



The day featured talks by ataxia expert neurologists describing the intricacies of the different types of ataxia and their diagnostic pathways as well as guidance on ongoing care. The event was very well attended with 65 healthcare professionals of different disciplines participating (mostly physiotherapists, speech and language therapists, occupational therapists and neurologists). Feedback received on the event was very positive with 57 of 59 delegates who responded saying they found the event 'useful' or 'very useful'. See Ataxia UK website for more details of the day and selection of presentations.

Here is a selection of comments received from delegates :

'After listening to the patient's experience I have gained a better understanding of what the individual would like to be considered... We have learned a lot!'

'In addition to the excellent information presented, which I will benefit from, the clinical guidelines and other ataxia information will be very useful for colleagues and myself.'

London Ataxia Centre – recruiting volunteers

Patrick works in finance at Glaxo SmithKline and enjoys music concerts, photography and watching football (he is a dedicated Leyton Orient supporter) in his spare time. He has cerebellar ataxia and started volunteering as an Ataxia UK Representative at the London Ataxia Clinic last year; here he tells us about this experience.

'I have been a volunteer for Ataxia UK at the London Ataxia Centre for just over three months and I find the work enormously rewarding. My role involves seeing patients after their appointment with the neurologist and taking details to ensure they are receiving all help required. I also introduce them to Ataxia UK and the services offered. Alongside this however there is the sharing of a cup of coffee and a few biscuits and general chat to make sure everything is OK or just being the outlet for people to be able to discuss ataxia with a fellow sufferer. I think I am well positioned to help in this way – I believe it is important for the patient to be able to talk to someone either with ataxia, like me, or a strong connection to the condition. Empathy is very important. I am also inspired by the passion and dedication of the neurologist and staff. In short it is something I would not hesitate to recommend to anyone to undertake.'

We are looking for another volunteer to join our team of five. If you are interested please contact Julie Greenfield at the Central Office.



Report of 4th International Friedreich's ataxia Conference

Institute of Genetics and Molecular Cell Biology (IGBMC) in Strasbourg, France 5-7 May 2011

This conference was organised by the US ataxia charity, Friedreich's Ataxia Research Alliance (FARA) and researchers at the IGBMC. It was sponsored by many organisations including Ataxia UK. The annual meetings for the European FA Consortium for Translational Studies and *euro-ATAXIA* were also held at the same time.

Two hundred researchers attended this conference and a brief overview of the scientific news from the meeting is presented here; see the Ataxia UK website www.ataxia.org.uk and the FARA website www.curefa.org for more information.

Regulation of the frataxin gene and its silencing in FA

Since the initial discovery on the way the gene causing FA is switched off and the potential use of drugs called histone deacetylase inhibitors (HDACi) as a way of switching the gene back on, there has been much progress in this area of research.

Professor Festenstein's team updated us on the role of an HDACi in increasing frataxin levels which has now moved to a human trial stage. He also spoke about the role of another mechanism that could potentially be targeted for therapy. This is the novel use of a drug which increases the levels of frataxin by preventing the destruction of the enzyme, RNA polymerase II, which is essential for switching the gene on.

Another important discovery regarding gene regulation is that the GAA repeat expansion in the frataxin gene, which causes Friedreich's ataxia (FA), changes with time and researchers are looking at ways to prevent this increase in GAA repeat size. Mismatch repair genes (such as one called MSH2) have been studied as a potential target. For example, Dr Gottesfeld has been studying small molecule polyamides as a potential therapy, and these result in a decrease in GAA repeat expansions in Friedreich's ataxia cells and this could possibly be via displacement of MSH2 from the frataxin gene.

The US pharmaceutical company Repligen provided an update on their work on HDACi. They are starting a phase I human trial in Turin (Italy) to test for safety of their lead compound. Although mainly testing for safety, as this trial will actually be done on people with FA, they will also be able to test whether the drug is effective in increasing levels of frataxin. They are also working on new second and third generation HDACi with better properties in case the lead compound fails. Ataxia UK and other charities have partly supported this work at the Scripps Institute.

Models of FA

The conference highlighted how Dr Pook's mouse models funded by Ataxia UK have been used extensively by many researchers. Dr Pook has currently a grant jointly funded by Ataxia UK and Go-FAR to develop models with increased GAA repeats and showing more progressed stages of FA. As soon as they are developed they are immediately put to good use by many researchers due to their extensive collaborations with one another. Dr Puccio's team has also created more mouse models (see *euro-ATAXIA* conference report on page 16).

A large section focused on the development of induced pluripotent stem (IPS) cells as models of FA (see Dr Dottori's section in *euro-ATAXIA* conference report). Additionally, a FA Stem Cell Task Force meeting was also organised by ataxia charities, including Ataxia UK, to facilitate communication and collaboration between researchers working on these cells. It is important to ensure optimum access of these useful cell models to researchers worldwide, so mechanisms of achieving this were discussed.

Stem cells

The current status of stem cell research was described by a world expert from Australia, Professor Pera. He gave a keynote lecture describing the challenges faced in developing stem cells for the clinic; obtaining high numbers of pure cells, optimising delivery methods and integration into the host tissue. He was cautionary about the claims made by clinics offering stem cell therapy, saying their procedures are often costly, use a poorly defined product and are not based on positive clinical trial data. He recommended the Closer Look at Stem Cell Treatments website for more information www.closerlookatstemcells.org.

There were also two posters presenting results of FA stem cell research; one testing the effects of bone marrow stem cells in FA cell lines and one using stem cells derived from adipose (ie. fat) tissue in mouse models of FA. The former is being funded by Ataxia UK and is taking place in Bristol and the latter is from a team in Spain.

Screening for drugs

A number of researchers spoke about screening studies to find drugs that increase frataxin levels. This is done using different 'libraries' of drugs that act on different pathways within the cell. For example, Ataxia UK-funded researcher Dr Lufino from Oxford described his work in developing a cell model of FA and using this in a screening study with a library of drugs that are structurally similar to HDAC inhibitors. Two drugs from this library were found to increase frataxin levels in a specific manner and these will be therefore studied further.

A researcher from Australia presented results on a screening study that he did in which he discovered that resveratrol (an antioxidant found in grapes) increased frataxin levels in cells taken from people with FA. This has now led to the development of a phase I trial testing the effect of resveratrol in people with FA in Australia. Researchers in the US also did a screening study with a different library and identified some potential compounds of interest for FA.

Diagnosis

Researchers in the US reported the development of a new test to accurately measure frataxin levels in whole blood or dried blood spots that would be relevant for population screening. They mentioned an ongoing newborn screening pilot study in the US that is funded by the NIH. The aim of the study is to determine a suitable method of screening newborn babies for FA so that future therapeutic developments can be implemented from the earliest point possible and obtain maximum effect.

New drugs

A research team was working on developing synthetic CoQ10 analogues that work better than CoQ10 and idebenone.

BBC Radio 4 Appeal *Sunday 30 October 2011 at 7.55am* with Kim Wilde

It is time to spread the word. Ataxia UK will be featured on the Radio 4 Appeal. Our Patron, Kim Wilde, has kindly agreed to present the appeal, which will be broadcast on Sunday 30 October at 7.55am and then again at 9.26pm.

It is a great opportunity to raise awareness and funds, so we need your help. Add a note to the bottom of each email you send out, include a broadcast in your newsletter, tell all your friends, family and colleagues.

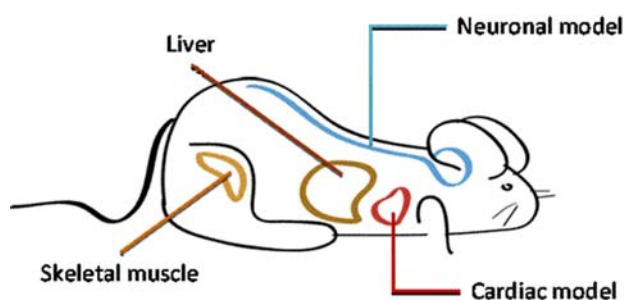


Scientific Research Report from *euro-ATAXIA*

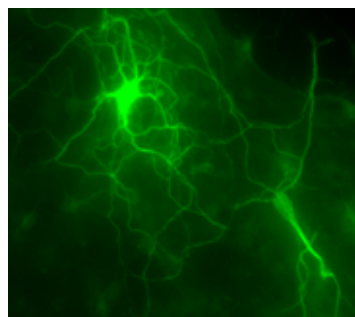
Ataxia UK is a member of *euro-ATAXIA*, a federation of ataxia charities from European countries. This year's annual meeting was attended by 14 member groups. We are very grateful to all researchers who took time out from their busy schedules to give research presentations at the *euro-ATAXIA* meeting. This is a summary of some of the presentations; a full report can be found on the Ataxia UK website.

Friedreich's ataxia (FA)

Work in Dr Puccio's lab in France focuses on improving cell models of Friedreich's ataxia (FA) and increasing understanding of the function of frataxin protein. Dr Martelli, who works in the lab, said that they have developed mouse models in which the frataxin gene can be deleted in certain parts of the animal's body (see diagram). Deletion of frataxin from the liver (important in regulation of iron levels) revealed that the primary role of frataxin is its involvement in the formation of iron sulphur cluster (ISC) proteins (complexes required for essential cell processes such as energy production, DNA and RNA metabolism, and iron metabolism).



Mouse models where frataxin can be deleted from the liver, skeletal muscle, neuronal cells or cardiac cells have been created by researchers in Dr Puccio's lab.



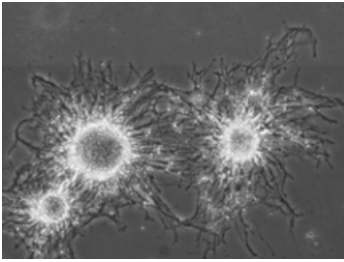
Neuronal cells grown in culture from Dr Puccio's mouse models.

FA stem cell therapy research

Dr Dottori from the University of Melbourne spoke about the production of and uses for induced pluripotent stem (iPS) cells. IPS cells are cells with stem-cell like properties that have been created in the lab from cells taken from adult tissue, such as skin cells. She described how the addition of a cocktail of specific proteins to these skin cells grown in a dish can reverse their programming and turn them into cells with stem cell-like features, that is the ability to replicate and to become other types of cells.

To date, Dr Dottori and her colleagues have established two iPS cell lines using skin cells taken from two people with FA. These cells have also been treated in the lab to coax them towards becoming nerve and cardiac cells, the cell types most affected in FA. These cells have been shown to have the same genetic characteristics as the original FA skin cells, meaning that they can be used to study the underlying mechanism of FA in these cell types and to screen for potential future drugs.

Researchers in France and the US are also working on the development of these useful cell models (see picture on page 17).



Induced pluripotent stem cell (iPS cells) from Dr Puccio's lab being treated to differentiate into neuronal cells.

Dr Dottori gave a cautionary word about stem cell treatment clinics that are available in some countries. She pointed out that the procedures carried out in these clinics are unregulated and the claims made are often exaggerated and unproven. She stated that though promising, stem cell research is still at the stage of basic scientific research to understand the mechanisms and processes involved and advised exercising extreme caution with respect to the clinics until the treatments had been proven in clinical trials to be safe and beneficial.

Cerebellar ataxia clinical trials

Dr Saccá has been involved with clinical trials for lithium, a commonly prescribed drug for bipolar disorder. A trial of lithium in people with SCA1 was started after improvements were seen with the drug in a mouse model of SCA1. The trial ended in February 2010, and the results are awaited.

The SCA2 trial that Dr Saccá will be running in Naples (Italy) will be a small placebo-controlled study (20 participants) looking at lithium treatment over a period of 48 weeks and will measure the safety and tolerability of the drug as well as looking at efficacy. The dose of lithium will be increased over the period of the trial in order to establish the optimum dose (as lithium can be toxic at high concentrations, any potential toxicity will be monitored carefully). Lithium is also being tested in a clinical trial for multiple systems atrophy (MSA).

Other drugs currently in clinical trials for SCA are sodium phenylbutyrate for SCA3, varenicline for SCA3 (an anti-smoking medication) and riluzole for hereditary cerebellar ataxias (a drug prescribed for motor neurone disease).

Researching the cerebellum

Cerebellar Cortical Control: Cells, Circuits, Computation and Clinic is a multinational group of researchers funded by the European Commission to study the function of the cerebellum and Professor Miall, from the University of Birmingham, UK, is part of this group.

He said that although the cerebellum is small in volume compared to the rest of the brain, it accounts for approximately 30% of the overall area and 50% of the total number of nerve cells in the brain. It has a unique structure and function and is similar in structure between different vertebrate animals, suggesting an important and conserved role. He described the group's imaging experiments that show the cerebellum is involved in hand/eye coordination and prediction of fine-tuned skilled movement. The results also suggest a role in the memory and learning processes associated with movement. Imaging studies suggest a role in emotional processing, working memory, language tasks and executive planning (the thinking processes involved in organizing thoughts, prioritising tasks, making decisions and time management).

Future work will focus on how the cerebellum interacts with the cerebral cortex, its involvement in cognitive processes (such as awareness, reasoning, judging and learning) and how these processes are affected by cerebellar damage, eg in ataxia.

Continued

Scientific Research Report from *euro-ATAXIA (continued)*

Update on idebenone trials in FA

Dr Will Andrews, Vice President, Medical Affairs, Santhera Pharmaceuticals explained there have been three trials sponsored by Santhera testing the effect of idebenone in FA (two in the US and one in Europe; see *the Ataxian* 171). While the first trial showed a statistically significant improvement in moderately affected FA patients taking idebenone compared to those taking placebo, these results were not replicated in the following two studies.

To gain more information, Santhera is pursuing this further by extending the two most recent trials in the US and Europe by 12 months, with all participants knowingly taking idebenone. Results from the 12 month US extension study combined with the six month original trial (ie a total of 18 months) were presented and showed that at the highest dose the ataxia rating scale values did not get worse, which is encouraging given FA is a progressive condition. This data is currently under review by Health Canada, where regulatory authorities conditionally approved Santhera's idebenone product (Catena®) in July 2008 for use in FA, pending future trial results. About half of FA patients in Canada are taking idebenone.

A new, two month randomised withdrawal study, known as the PROTI study has also been recently started by Santhera. It involves people in the European extension study being temporarily removed from it and enrolled in the PROTI study where they are randomly assigned to either continue idebenone or receive placebo in a blinded fashion. Their fatigue levels and speech will be assessed (anecdotally, patients have reported benefits in these symptoms), in addition to other neurological endpoints. Patients will also guess whether they think they have been taking idebenone or placebo, as a way of measuring if they feel the benefit. Santhera hopes that information gained from this study will help them to better understand FA and how idebenone helps FA patients feel and function better. This information will help Santhera in the design of new, larger clinical studies evaluating the potential benefit of idebenone in FA patients.

eBay sales

We would like to say thank you to Patsy Riggs, Chair of the Ipswich and Colchester Support Group for her continuing crochetsales through eBay. It is a wonderfully innovative and personal way to support Ataxia UK. Well done!

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

Myra Williams, Doris Bear, Roy Woombell, Stanley Potter, Philip Moore, Thomasina McLellan, Richard Frith, Cheryl Cooling, Raymond Broad, Bridget Hough, Martin Wells, Martyn Tubb, Lilian Nash and Thomas Edgumbe.

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. 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.

'Living with ataxia' Exhibition The Delfina, 18 September 2011

This wonderful exhibition will go on show for two weeks at The Delfina on 50 Bermondsey Street, London SE1 3UD from Sunday 18 September to Sunday 2 October.

Please do come and have a look! If you have a venue that would like to showcase our photographs later in the year, let us know on fundraising@ataxia.org.uk.



International Ataxia Awareness Day (IAAD) Website

We will be launching a new website www.ataxiaawareness.org for IAAD on **18 September 2011**. Packed with information, pictures and resources it will feature an ever growing gallery of photographs and last minute details of local events. The more information you provide, the better it will become.

Please send in details of events local to you to fundraising@ataxia.org.uk and include any photographs you would wish to share. Thank you.

Details of the London Wheely Wobbly Bridges Walk can be found on the back page of the magazine.



**TEXT "ATAX11£5" TO 70070
TO DONATE £5 TO ATAXIA UK**

Ataxia is a neurological condition that affects over 10,000 people in the UK

There is currently no cure


It attacks speech, movement, sight and hearing

Ataxia UK searches for a cure and helps people live with ataxia everyday

We have a selection of awareness raising posters available in electronic format to go with the Awareness Raising Pack (one pictured here).

Look out for the poster campaign in Leicester from the 25 September 2011 till the start of our National Conference on the 8 October 2011.

You can register today by calling
020 7582 1444 or by email:
fundraising@ataxia.org.uk
Quote **IAAD 2011** to get an
Ataxia UK t-shirt on the day!
We look forward to seeing you at
the start line!



Bridges

Join Our Wheely Wobbly Walk

Sunday 25 September 2011 at 11am

Registration on the day opens at **10am** at the **Riverside Gardens**, Milbank, next to Vauxhall Bridge.

Lunch and Half-Way Start Point at **12.30pm** at the **Victoria Embankment Gardens**, next to Embankment tube.

Finish by the **Tower of London**.

Come join us for a fabulous day out raising awareness!

Ataxia
UK
Caring today,
researching for tomorrow