REPORT OF THE MEETING OF THE EAST OF SCOTLAND BRANCH OF ATAXIA UK
LASSWADE HIGH SCHOOL CENTRE,
SATURDAY 11 MARCH 2006
http://www.ataxia-east-scotland.org.uk

Present: Sheree & James Allan, Pete Case, Liz Dalby, Dr Paul de Sousa, Penny Gardner, Andy & Fay Hogg, Derek Main (Chairman), David Main, Maureen & Sandy Pender, Lesley & Andy Pringle and Peter Smeaton.

Apologies: Ronnie Browne, Andrea Bothwell, Rhona Brankin, Pete Dalby, Anne Green, Julie Greenfield, John Hunter, Alastair Macdougal, Susan McPheat, Jim, Essa and Lynn Quin, John Reid, Alan Smith, Eileen Steele, Anna & John Thomson Richard & Anne-Marie Thomson, Professor Bob Will and Frances Wright

1. Welcome and Minutes of last Meeting
The report of the meeting held on 21 January 2006 was approved – Penny apologised for the incorrect heading, which has been changed on the Branch website.

2. Secretary's Report – Branch Website News
The Branch website (http://www.ataxia-east-scotland.org.uk) has now had nearly 400 visitors. If you have ideas for any other content pages or useful links on our site, please let Penny know: info@ataxia-east-scotland.org.uk

3. Branch Christmas Lunch, Sunday 4 December 2005
As agreed at the last Branch meeting, due to the unsatisfactory meal and inadequate toilet facilities at this event, Penny wrote a letter of complaint to the manager. No reply to date, so a follow up letter is being sent.

4. Treasurer's Report
Frances sends her apologies; she has been off work all week with a viral infection. Apart from a visit to the doctor, she has not been out all week and doesn't want to risk passing the infection on. The report was presented to the meeting by Penny. The transfer to the CAF bank is almost complete, the final handover letter has been prepared and she hopes that the remaining funds will be sent by the end of March. She will also be getting together with Penny at the end of the month to sort out the books and final accounts for the year.

Bank balances at the last statement date (end of February) are:

- New CAF Account 1,085.00 (includes £1000 from HBOS)
- Old HBOS Account 88.90
- Total 1173.90

Since the last meeting, there have been the usual receipts and expenses as follows: **Receipts:** Donations: £30 from Rhona Brankin, £20 from Peter Smeaton, £15 from Andrea and £10 from Mrs Evershed (Derek's aunt). Anne-Marie's knitting raised £10. Winnie Walmsley also sent £5 which has not been paid in yet. Two collecting cans which Derek brought along last time totalled £51.23.

Expenses: Copying of £16.88 has been paid, plus a few other minor expenses in January which will be included in the annual accounts. Still to be paid are 6 monthly charges for the website £35.18, teas / coffees and the stock of stamps needs replenishing.

Other news
Frances had a trip to St Boswells last weekend and popped in to see Kathleen Allan. It was quite a while since she'd visited last and Kathleen's now very frail.

She has also had an information leaflet from the canal association who organise the Falkirk Wheel boat trips. These start up again in April, so we need to look at possible dates for an outing. She hopes to be able to attend the joint meeting with the West of Scotland Branch on Saturday 1 April.

5. Dr Paul de Sousa: Talk on Stem Cell Research
Paul explained that he joined the 'Dolly' team at the Roslin research institute team 8 years ago. Last summer he joined the University of Edinburgh where he is forming a new group. Beginning 3 years ago, Paul's lab began trying to isolate new embryo derived stem cell lines with the eventual aim of using them in various therapies. The area of research is called 'embryo biotechnology'.

In the 1950s and 1960s, scientists studied the developmental processes in animals and humans – how an egg develops into an early group of cells called a 'blastocyst'. They removed eggs from the ovary and cultured them, but found that every process applied to these had an adverse effect and less and less became viable.

In the 1980s, scientists working on mice found they could isolate primitive stem cells from a blastocyst in a dish. These cells were capable of developing into different types of cell eg skin cells, liver cells etc. These primitive cells could be genetically manipulated, and on being placed back in a blastocyst resulted in the creation of genetically modified animals. Unaltered and genetically modified primitive cells could also be preserved by freezing. Unfortunately, decades of research failed to translate this technology to other species.

10 years ago Dolly the sheep was produced by somatic cell nuclear transfer, also known as cloning, which could now be used to create genetically modified large animal species. Ian Wilmut's team was able to do this by removing genetic material from an egg and replacing it with different genetic material from adult cells.

In 1998, a team in Wisconsin found that they could recover primitive stem cells from human embryos, which like the mouse could be coaxed to form a range of adult type tissues. However, due to ethical issues, no attempts have been or will be made to produce offspring from these cells or cells like them.

Human embryonic stem cells can be used for clinical evaluations – testing the effects of new drugs – and, potentially, for therapies. But before this can happen, there are several practical problems to be overcome:

- Firstly, they must be created in sufficient numbers to be useful.
- Secondly, they must be safe and free from contaminants.
- Thirdly, they must function in the same way they would normally.
- Lastly, they must be tolerated by the human body.

The problem is that there are very small numbers available in the first place and to grow them requires a culture medium and a growth stimulant – both of which could introduce problems. Traditionally, these have been derived from animal products – mouse, bovine and rabbits - but the Roslin team has been trying to devise chemical alternatives. They have also spent a lot of time creating a totally sterile environment to avoid contamination. This is more manufacturing than science.

The team had 50 donated embryos to start with and have been experimenting with different ways of encouraging the stem cells to grow. They have checked their composition, to see if they are stable, and have been making different types of cell. The aim is be totally independent of animal products, even in a remote way.

There are various sources of stem cells and ethical considerations involved. Some are from surplus eggs (fresh or frozen) from IVF (in-vitro fertilisation) which are donated for research purposes. There can also be surplus embryos – but the use of these could be more doubtful. Also, after fertilisation treatment, 30% of eggs are unusable and would be discarded anyway – material can be recovered from these. The team has also received some donated eggs from women undergoing voluntary sterilisation.

When embryos are cloned, sperm is not used; instead a stimulus is applied to start the process – chemical or electrical. 'Parthenogenesis' (virgin birth) uses the egg's own genetic material to advance the process of development. Although such embryos fail if implanted into the womb, there are actually some people in existence who contain cells with this condition.

Aside from the ethical issues, human cloning to make embryonic stem cells for therapy is practically challenging – but maybe could become practical in something like 20 years. Instead, at the present time, cloning could be used to study diseases where the genetic basis is not understood, eg motor neurone disease. Eventually it is hoped that we won't have to use eggs but will able to treat the cells in situ.

The University of Edinburgh is now forming a new Scottish Centre for Regenerative Medicine, being built at the New Royal Infirmary.

Questions then followed and Paul discussed the cloning scandal in Korea – this could not happen here as we have a strong regulatory environment with more safeguards in place. There's currently a funding gap, with money available from government for basic research and from industry for near market products that are almost ready – but not enough for the applied research needed in the middle. It could be 2 – 3 years more before we have proof that therapies are possible – the current permissive attitude to stem cell research in the UK will drop off if this does not emerge.

Paul thought that the first diseases that might be treated effectively could be Huntingdon's Chorea or Parkinson's - also spinal cord injury cases. The second area that might be treatable is acute liver damage – because the liver is slow to regenerate, people need help to survive the early stages. Liver-like function cells might be used in external blood circulation to enable this. Lastly, people with heart problems might be able to have stem cells injected to provide missing function.

Finally, Paul emphasised that research in the uses of embryonic stem cells is still in the early stages. He does not want to offer false hope, but if anyone has questions he is happy to answer or redirect them as needed (please send these through info@ataxia-east-scotland.org.uk). He was then thanked for his interesting and informative talk.

6. Research Update from Dr Julie Greenfield of Ataxia UK

6.1 Research funded by Ataxia UK

A new research project has just been approved for funding and should be starting soon: Its title is **Cognitive effects of cerebellar disorder**
Researchers: Prof Griffith, Prof Chinnery and Dr Welch (Newcastle Uni)
Funding: 3 years funding awarded for a PhD studentship.

Lay summary
This work tests the hypothesis that disorders of the cerebellum affect not only movement but also the ability of patients to carry out certain types of computation. Specifically, there is evidence based on our pilot work that these disorders affect the perception of space and time and our ability to predict the way in which visual patterns and sound change over time. If the concept can be proven it will have important implications for patients who suffer from any cerebellar disorder. They propose to carry out the work on a group of patients with an inherited ataxia where the disease only affects the cerebellum (SCA6), to allow them to obtain clear information about the effect of cerebellar damage in the absence of other confusing factors.

The next Scientific Advisory Committee meeting is at the end of March. Three more research projects have been submitted and peer reviewed, and these will be evaluated at the meeting. If they are recommended for funding the Trustees will then decide if there are sufficient for the projects to go ahead.

For a full list of all the other projects currently funded go to the research page of Ataxia UK's website http://www.ataxia.org.uk This has been updated and has summaries of all projects now.

6.2 Research meetings:
Ataxia UK has offered to host the Euro-ataxia conference in the UK this year. It will be held in mid-September and we are hoping to also host a one-day conference on rehabilitation. In addition, in May Ataxia UK is organising the annual meeting of neurologists with expertise in the ataxias. The main focus of this meeting is the discussion of Clinical Guidelines for the care of people with ataxia. We aim to produce such Guidelines for healthcare professionals by the end of the year.

6.3 Raising awareness amongst medical professionals
In addition to the production of the Guidelines discussed above, in October 2005 we were pleased that an article we wrote on the ataxias was published in the British Medical Journal. The title is 'The Patient's journey: the progressive ataxias' and it focuses on the impact of the ataxias on people's lives and featured a number of examples of people with different ataxias at different stages. We felt that this was a good way of raising awareness amongst GPs, as this is the main journal for GPs in the UK.

This month we have been asked to write an article on the ataxias for the British Journal of Nursing, an ideal opportunity to reach a different audience. We are currently working on this.

6.4 Other research worldwide

New gene discovered

Researchers in the US have now found the gene responsible for spinocerebellar ataxia type 5 (SCA5), an article published in *Nature Genetics* in January 2006 reports. SCA5 is a form of ataxia that is inherited dominantly, meaning there is a 50% chance of passing on the ataxia to your children. This is an exciting discovery because it provides a new genetic test for people with ataxia of unknown cause and will lead to improved specific diagnoses. Researchers also now know more about what goes wrong in the cells of people with SCA5, which will help pave the way towards the development of treatments. SCA5 is caused by a mutation in a protein called spectrin. This protein normally plays an important role in maintaining the health of nerve cells.

Interestingly, the research has involved a long search over the last decade and the gene was actually identified in an 11-generation family descended from the US President Abraham Lincoln. This has also sparked interest in the US media about ataxia generally.

As this is very new research it will take some time before the diagnostic test will become routinely available across UK laboratories. However, when it does, it will be possible to test not only person with ataxia but also asymptomatic family members to see if they will develop the condition, and whether their children are at risk.

New clinical trial in the US for Friedreich's ataxia

In November the US charity FARA (Friedreich's Ataxia Research Alliance) announced the award of a large grant of $3 million dollars to the company Edison Pharamceuticals for the development of EPI-A0001 to treat Friedreich's ataxia.

How does A-0001 work?

People with Friedreich's ataxia have low levels of the protein frataxin in their mitochondria (compartments in the cell that are responsible for producing energy). Frataxin is thought to assemble a group of proteins known as iron-sulphur clusters that are involved in producing energy. When there are low levels of frataxin, there is a problem with assembling these iron-sulphur clusters and less energy is produced. In addition there is an accumulation of free radicals – damaging by-products. Researchers believe that A-0001 inserts itself into the mitochondria and assists in producing energy. It should result in significantly more energy being produced, and less damage caused by free radicals. **This treatment is essentially thought to bypass the fact that there is less frataxin in the cells of people with Friedreich's ataxia.**

FARA reports that planning is underway to progress this project and lead to a phase II clinical trial in people with Friedreich's ataxia in 2006. For more information on FARA go to: http://www.FAResearchAlliance.org.

Idebenone trial in Friedreich's ataxia – recruitment in the UK starting soon. As described in the Ataxian and at the 2005 Conference a company called Santhera are planning a clinical trial of the antioxidant idebenone for people with Friedreich's ataxia. This trial will take place in the UK as well as in other European countries. Santhera has already received regulatory approval from the UK Medicines and Healthcare products Regulatory Agency (MHRA) and is expecting final approval of the protocol from the Ethics Committee shortly (standard practice for all trials). The trial will be set up and start recruiting patients in the next month or so.

Why is the trial being done?
This trial will test the effect of idebenone in people with Friedreich's ataxia. Idebenone is a man-made drug similar to Coenzyme Q10. Idebenone has been tested in a number of small trials in the past eg in France, Italy, Canada and Spain (see *Ataxian* 142). The studies to date have shown some improvement in protecting the heart but there are mixed results on whether there are any improvements in the neurological symptoms (ie people's ataxia). These trials have been small and most not placebo-controlled. At the moment, there is not sufficient evidence that idebenone is of benefit to people with Friedreich's ataxia to satisfy the Regulatory Authorities that issue licences for drugs to be prescribed in the UK. This is the reason for Santhera funding and running the trial.

This study will be a larger trial (involving around 200 people) and will compare three doses of idebenone and placebo. It will be double-blind placebo controlled trial (ie the gold standard for clinical trials). This means that participants will be randomly assigned to either placebo or one of three doses of idebenone, and neither the participant nor the doctor will be aware of who is in which treatment group. The study will also tell us whether doses that are higher than those in previous published studies are of benefit.

What does the trial involve?
The trial will last about a year and participants will need to travel to one of three UK centres a number of times. The UK Centres are in London, Newcastle and Sheffield, where ataxia specialist neurologists are based. At each visit the doctors will take different measurements (eg test the heart, do neurological tests and use rating scales). One of the rating scales they plan to use is the Friedreich's ataxia impact rating scale (project funded by Ataxia UK described in *Ataxian* 152).

Taking part
As soon as we are given the go ahead we will be writing to all people with Friedreich's ataxia on our database offering the opportunity to take part in the trial. Details will be provided on the specific inclusion criteria required in order to participate.

7. AOCB
Joint meeting with West of Scotland Branch on **Saturday 1 April** at the Alona Hotel, Strathclyde Country Park from 2 – 5pm. **No charge for this.**

5 – 6 April is the **Scottish Disability Roadshow** at Ingliston, Edinburgh.

The usefulness of your **MSP** in getting problems addressed was mentioned – and a suggestion that if he/ she can't or won't help – go to the opposition! Pete Case said that his MSP had been very helpful in obtaining a suitable wheelchair service for their son.

Derek announced his intention of standing down as Chairman from the 2007 AGM.

8. Date of Next Meetings
The next Branch meeting is the AGM, which will be held at Lasswade on Saturday 6 May 2006 from 1.30pm to 4pm. Prior to that, we are holding a FREE joint meeting with the West of Scotland Branch at the Alona Hotel, Strathclyde Country Park 2 – 5pm on Saturday 1 April. Dates for September and November meetings have not yet been decided.

YOUR COMMITTEE

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USEFUL WEB LINKS

If there are any suggested additions to this list please let us know

www.ataxia-east-scotland.org.uk: our Branch website

www.ataxia.org.uk the Ataxia UK website, it has many good links.

www.ohbother.co.uk: by an Ataxian and full of very useful information.

www.bbc.co.uk/ouch for an inside view on disability news**.**

www.evoc.org.uk: for local disability information in Edinburgh.

www.digg.org.uk: Glasgow's online resource for disability information.

www.matchinghouses.com: re: accessible holiday house swaps.

www.skill.org.uk information & advice for disabled students

E MAILED REPORTS

If you would prefer an e mail instead of a hard copy, please let us know your e mail address:

Name _____ Telephone No. (optional) _____

E Mail address _____

Please post to the Secretary, Penny Gardner, at 3 Craigleith Gardens, Edinburgh EH4 3JW or e mail **penny@ataxia-east-scotland.org.uk**

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MEMBERS VOLUNTARY SUBSCRIPTIONS.

Please send a contribution if you can - £5 per household is suggested,

Please send a cheque, payable to East of Scotland Branch of Ataxia UK to: Frances Wright, Flat 8, 25 Queen Charlotte Street, Edinburgh EH6 6AX