REPORT OF THE MEETING OF THE EAST OF SCOTLAND BRANCH OF ATAXIA UK LASSWADE HIGH SCHOOL CENTRE, SATURDAY 13 MARCH 2010
http://www.ataxia-east-scotland.org.uk

Present: Derek Main (Chairman), Tony & Mary Callow, Liz & Pete Dalby,, Dr Richard Davenport (Speaker), Penny Gardner, Sally Gemmell, Doreen Greenwood, Andy Hogg, Sheree Logan & daughter, Gerry Pearson, Calum Pringle, John Reid, Tom & Doreen Vandepeear, Chris & Fiona Wragg and Frances Wright

Apologies: Andrea Bothwell, Rhona Brankin, Ronnie Browne, Anne Green, Stuart Greenwood, John Hunter, Andy & Lesley Pringle, Richard & Anne-Marie Thomson, Robert Will

1. Welcome and Minutes of last Meeting

Derek welcomed everyone to the meeting, especially those attending for the first time. The report of the meeting on 23 January 2010 was approved. Penny said that last year's reports will follow later.

Everyone introduced themselves:
Derek is the Chairman and has had CA since he was 14 months old.
John Reid from the Borders is on the committee and has FA
Tom Vandepeear who has had CA for 15 years & his wife Doreen
Gerry Pearson from Dalgety Bay has CA and retired 3 years ago
Doreen Greenwood's husband Stuart has Fragile X Tremor ataxia
Tony Callow has CA & his wife Mary.
Chris & Fiona Wragg – Fiona's Mum has CA - SCA 6
Frances Wright is the Branch Treasurer
Sheree Logan's son is 16 and has FA
Andy Hogg has symptoms of CA, but his diagnosis is not certain.
Calum Pringle's Mum has CA (SCA 2) and he is studying communication design, specialising in aids for people with speech impediments
Liz Dalby has CA and is also on the Branch committee & Pete who is our past chairman.
Sally Gemmell has FA and has come through on the train from Glasgow.
Dr Richard Davenport DCN at the Western General Hospital, Edinburgh.
Penny Gardner is the Branch Secretary.

2. Chairman's Remarks

Derek reminded members of the visit to the Scottish Parliament which has been organised for Weds 24 March at 10.00. He must have people's names in advance for security checks, **by Monday 15th if possible**.

An update from the January report:. Graham Kennedy from Ataxia UK has been awarded an MBE in the New Year Honours for his fundraising: www.bigbadbikeride.com The 2010 event is taking place in Pitlochry on 10 to 12 September – a 75 mile cycle ride.

3. Treasurer's Report

Frances reported that since the meeting on 23 January:
Donations have been received from the Greenwoods and the Wraggs of £20. Mr & Mrs Smith made their regular monthly standing order for £10.
£60.72 has been paid in from various collecting tins. Expenses since the January meeting were the haggis meal £45 plus stamps & photocopying £27.40. The balance at the bank stands at £920.

Frances' new address is 55 Carrick Knowe Road, Edinburgh EH12 7BN and has been updated on the website and documents.

A member from Kirkcaldy rang Frances to tell her that he has now been re-diagnosed and does not have SCA (Spinal Cerebellar ataxia) but has HSP (hereditary spastic paraplegia). He wishes to be taken off the mailing list. He's having a follow up appointment with a genealogist at the Western General. He said he's having 7 days a week care now and is doing fine.

4. Dr Davenport W General

Dr Davenport said that he would not be talking for an hour. Instead he planned an introductory talk, followed by a Q & A session. He's a neurologist working in Edinburgh – his 10th year of being a consultant, having trained in the old system (13 years training). He has a variety of interests and runs the movement disorder clinic. Last year AUK asked him to be medical advisor in Scotland, and thought he wasn't the right person, so emailed colleagues and no one replied. So he said ok. He's not a high flying scientist or academic, but has lots of working experience.

There is a divide between what doctors are mainly interested in (diagnosis, causes) and what patients & carers want to know (treatment). He plans to cover both of these in his talk: the 'middle ground'. He looked at the AUK website – a recurring theme is people reporting delays & uncertainty in diagnosis. They sometimes have to wait a long time, sometimes never getting a specific diagnosis.

So, why is diagnosing ataxia difficult? Firstly, it is not common. The best estimate is 10,000 people in the in UK with some form of progressive ataxia. In Scotland this is about 1,000 people out of a total 5 million. It's therefore on the periphery of most doctors' radar, if they are aware of it at all. Other specialities may not be familiar with it at all – which can be a problem if this is the first person you are referred to. Older onset people

can be difficult to diagnose. There are very few ataxia specialists in the UK, though there are now 3 centres registered in the UK, all in England.

How do we approach a diagnosis? Ataxia is a symptom not a disease. Think of being drunk. People with ataxia are often mistaken for being drunk with unsteadiness, speech, visual problems etc. May well also have other symptoms from other conditions eg MS.

The first task is to take a history. This baffles patients – why do we spend so long chatting? Doctors place enormous importance on taking the history, and surprisingly the examination is less important. The age at onset of symptoms is very important. Genetic causes usually present earlier but not always. The evolution of the ataxia also important; genetic causes tend to develop symptoms over the lifetime, whereas ataxia due to cancer can be over days / weeks / months. Other symptoms can help diagnosis. Drugs you take can be a factor, both prescribed & otherwise eg recreational drugs. Excessive alcohol use can lead to ataxia & smoking cigarettes can lead to cancer. Many people do not have a family history especially with FA as it's caused by a recessive gene. Other medical problems; diabetes gastro intestinal diseases may be associated with ataxia. A patient's social background is also important. All these questions are very important as the answers help to build up a picture.

Secondly, the doctor examines you. This is looking for specific features; if you can walk they look at your gait - is it unsteady, broad based? Test tandem gait – walking heel to toe. Ataxia patients cannot do this. Test co-ordination finger to nose, touching something in space eg a finger. Tend to pass the target (pass point) and wobble on approach. The heel shin test is also very difficult for people with ataxia. They look at the way your eyes move and listen to your speech, which often exhibits dysarthria (slurring). The doctor will look out for features of other conditions eg Parkinson's or liver disease (Wilsons disease)

Thirdly the doctor will fuse the history and the examination and search the database – narrowing it down to a 'list of suspects'. WHERE is the problem? In nervous system? Where in the nervous system? Cerebellum, spinal cord. Think ataxia, cerebellum, now WHAT is it? The two questions - WHERE and WHAT? There are three categories of ataxia:

- Genetic
- Acquired
- Unknown / idiopathic

All 3 groups can be expanded, and Dr Davenport showed us a 2004 article listing the **genetic** causes of ataxia. There were 7 or 8. Now 6 years later there are 29 SCA types recognised. Doctors find it difficult to keep pace

with these developments. Some ataxias are very rare eg only found in one family in the Mediterranean, others eg SCA 6 and FA are relatively common. Sometimes strange connections are found eg DRPLA was thought to be unique to the Japanese but has been found in South Wales.

Dr Davenport showed us cut down lists of **acquired** causes (a long list) including toxic agents, vascular diseases (strokes) inflammation eg MS, endocrine causes eg Thyroid, infections eg chicken pox, structural tumors, degenerative conditions, metabolic causes and vitamin deficiency (a shortage of vitamin E is one of the rare, treatable forms of CA)

Idiopathic means that we don't know what caused it: the term comes from the Greek words 'ones own' and 'suffering' ie it is a disease 'of its own kind'. This diagnosis is especially likely in late onset ataxia. There may be a cause, but we haven't found it yet.

There are lots of different causes, and up to 50% of cases will defy a specific diagnosis. Various tests can be carried out. Sometimes these can be very accurate & pinpoint it exactly eg FA requires one blood test.

A recently published document (pdf download) on the AUK website is very useful for doctors & also patients. It lists the tests that typically will be carried out. Doctors have to be specific when asking for blood tests, and this document helps them with that.
http://www.ataxia.org.uk/data/files/ataxia_guidelines_web.pdf

Some of the causes of ataxia are incredibly rare and even experienced neurologists find ataxia a challenging area.

Q&A Session with Dr Davenport
Derek explained that he has idiopathic SCA; his father blamed it on an infection from milk in 1958. Dr Davenport said that there may still be treatments available, also advances in science can come along and give a diagnosis. Due to waiting list pressures, neurologists may be discouraged from following up patients with chronic conditions where no treatment is possible However, it is very important to press for regular appointments as diagnosis can change and new treatments can come along.

Liz said that she had 42 blood tests taken in 1995. Just recently they have identified SCA 8 and are very excited about it - only 2 people in the UK have it. She goes to the Western General to help with the medical examinations (as a test patient). Dr Davenport said that having a rare & unusual condition can attract a lot of interest from doctors, especially those in training. However it is so important to see real patients during medical training.

Pete said that the first stop when someone starts developing symptoms is that they go to their GP, whose specialist knowledge will be limited. GPs are very important since they act as gatekeeper to other specialities. They have a much harder job than a specialist as they are faced with everything. Most things they see are trivial or self limiting. Occasionally a patient will come along with a more serious condition eg ataxia, cancer or a heart condition. 12 – 15 years ago there could be a 2 – 3 year waiting list to see a neurologist. Things are much better now, people are getting appointments and MRI scans quickly. However, people's expectations have also risen, and some may be unrealistic.

Dr Davenport explained that cultures where marrying relatives is common may be more likely to produce genetic abnormalities. When new SCA types are found they don't know at first if they are going to turn out to be rare.

John asked about difficulties swallowing. It is unusual in ataxias for swallowing to be very severe, but there other ways of feeding which can be used eg for motor neurone disease, there are physical techniques which help or feeding directly into stomach. Experienced physiotherapists can be very useful, but one who doesn't specialize in neurological disorders can be worse than useless. Speech & language therapists are a bit better. The UK is traditionally under-doctored and under-nursed. Finland with a similar population to Scotland has 250 neurologists to Scotland's 50. It has improved - in 1992 there were 4 neurologists in Edinburgh now there are 20 or so. But we are still a long way behind the rest of Europe

Doreen asked about Tom's hospital notes having been destroyed. Lothian & most UK hospital trusts have a policy - if notes are not active for 5 – 7 years they destroy them; to avoid this the neurologists have to physically files them themselves. GP notes on the other hand are entire & follow you around. Hospitals used to microfiche the records, now they don't even do that. Resources (or a lack of them) is at the bottom of this (people to file the records or transfer to computer, space to store them etc).

Dr Davenport was thanked for his interesting and informative talk.

5. Update from Calum Pringle

Calum has been adapting speech recognition software for his Mum. At the moment there is nothing on the market between using your own voice (which gets progressively worse with Ataxia) and using a product that speaks for you (like Stephen Hawking). Calum's aim is to develop an intermediate device between the two.

His Mum tried using Dragon Naturally Speaking (Nuance) but it didn't really work – even with training it didn't recognize her speech. Instead Calum started her on building up her own dictionary from nothing, a word at a time and it did work. He has designed a scarf with a combined iPod application which displays the key words as his Mum is speaking. It will be an enormous help, especially when she speaks with strangers. The training to use the software can be done on her own and it's helping her with speech too. Until she heard Calum's recording she didn't know what her speech sounded like – training the software has enables her to focus on pronunciation and the improvement in this can be heard in this video: Video of Calum's Mum using the iPod app: http://vimeo.com/9408396

The software is set up to exclude small words eg 'a' and 'the' so it just shows the keywords on the screen; the ones that help a listener to follow the conversation. Work still needs to be done on the speed with which the screen shows the words and the exclusion of the small ones eg you don't want 'the' missing from a word like 'mother'!

There are 3 menu choices for the type of conversation for example a more formal conversation has a more formal font and a friendly chat uses an informal font (this is why we were looking at fonts in our January meeting!). Calum has designed a font using his own handwriting for the chatty one. The words are displayed in a light colour on a dark screen which is easier to see. When the programme is off the iPod screen shows a display to match the colour and weave of the scarf so is more unobtrusive.

We were all very interested and excited to see Calum's prototype and could envisage many uses for it – basically anyone who has difficulty speaking, not just ataxians. He said that a big advantage of an iPod application is its relative cheapness compared to the other products currently available.

Calum's website is at: www.freecalum.com & info on the 'Subtle Subtitles' project is at: http://cargocollective.com/freecalum#250203/subtle-subtitles

5. Social Events

The West of Scotland Branch of Ataxia UK have invited us to join them on **Saturday 10 April** at the Alona Hotel Strathclyde County Park. Several of us went last year and it was a very enjoyable occasion. Please contact Derek if you'd like to go and he will pass the information on.

6. Other Business

Penny apologised for the lack of reports from the 2009 meetings. She has not forgotten them but decided it was better to at least have the most recent ones done and hopes to catch up later.

7. Dates of Next Meetings

The next Branch meeting will be held on Saturday 8 May (the AGM) at Lasswade. Our speaker Alison Stevenson from AUK will be giving us an update on the latest news about research into ataxia.

YOUR BRANCH COMMITTEE

Chairman: Derek Main		0131 477 4371 derek@ataxia-east-scotland.org.uk
Secretary and Vice-Chair: Penny Gardner		0131 332 5218 penny@ataxia-east-scotland.org.uk
Treasurer: Frances Wright		0131 467 9023 frances@ataxia-east-scotland.org.uk
Committee Member: John Reid		johnreid@ataxia-east-scotland.org.uk
Committee Member: Liz Dalby		liz@ataxia-east-scotland.org.uk
Email committee Member: Susan McPheat		Contact through: susan@ataxia-east-scotland.org.uk

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

www.ataxia.org.uk the Ataxia UK (AUK) 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.gig.org.uk Genetic Interest Group

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

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

www.simr.org.uk/pages/news/index.html seriously ill for medical research

http://www.vocal.org.uk VOCAL

ADDRESS FOR MAILING:
East of Scotland Branch of Ataxia UK,
Penny Gardner, Branch Secretary
3 Craigleith Gardens, Edinburgh EH4 3JW

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
……

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, 55 Carrick Knowe Road, Edinburgh EH12 7BN.