REPORT OF THE MEETING OF THE EAST OF SCOTLAND BRANCH OF ATAXIA UK LASSWADE HIGH SCHOOL CENTRE, SATURDAY 15 NOVEMBER 2008
http://www.ataxia-east-scotland.org.uk

Present: Derek Main (Chairman), Liz & Pete Dalby, Penny Gardner, Andy Hogg, Jim Shepherd, Richard & Anne-Marie Thomson, Tom & Doreen Vandepeear & Frances Wright, Jon Stone (speaker)
Apologies: Andrea Bothwell, Ronnie Brown, Rhona Brankin, John Reid

1. Welcome and Minutes of last Meeting

Derek welcomed everyone and we all made the usual introductions. The report of the AGM last May is on the Branch website (but not posted out yet). These were approved. Penny apologised that she has not yet typed the report of the branch meeting held on 6 September 2008. Derek introduced our speaker for the day – Dr Jon Stone, consultant neurologist at the Western General. Jon passed on a message from Charles Boyce who says hello.

2. Scout Post

This costs just 21p per card in the Edinburgh area. The deadline for collection is 8 December and they are being delivered from 12th to 21st December. Collection points Station Bar, Cadzow Place, Margaret Blackwood House Abbey Court, Greenside Church, Castle Rock Housing, Norton Place, Rossie Place.

3. Jon Stone

One of the consultant neurologists at the Western General and he covers West Lothian as well. All neurologists need to know about ataxia but very few have a specialist interest. It would be difficulty to run an ataxia clinic in Edinburgh as so few people. If there was an intervention (treatment) available, the position would be different.

One member asked if the cerebellum has a left and right side and Jon said definitely, “yes”. An injury of the left cerebellum would cause problems with the left arm and leg. With generalised (inherited) ataxia there is usually symmetrical damage.

3.1 What is Ataxia?

Jon showed an MRI scan showing the position of cerebellum. It has very fine details with fern like fronds and is quite beautiful. Severe ataxia causes atrophy (shrinkage), and the cerebellum becomes much smaller. Symptoms include unsteady walking, slurred speech (scanning cerebellar speech), lack of co-ordination in arms / legs, ‘jumpy’ eye movements

(nystagmus), possible slowness in cognition and subtle changes in emotional control in some patients.

Medical terms you might come across are:

- Slow saccades (eye movements L to R)
- Scanning speech
- Finger nose dysmetria
- Dysdiadochokinesis (unsteady gait)
- Heel shin test
- Broad based gait

Lack of co-ordination is key indicator.
Jon then showed us some short videos of nystagmus (horizontal, upbeat) and intention tremor - a man with injury to left side of cerebellum (finger nose test) having problem using his left arm.

3.2 What are the causes of Ataxia?

a) Genetic / congenital (most common)
b) Stroke (can be a bleed in the cerebellum)
c) Inflammation
d) Brain tumour
e) Head injury
f) Toxins (alcohol is no. 1)
g) Other

3.2 a) Genetic Types of ataxia

(i) Spinocerebellar ataxia – all cause ataxia but have other symptoms as well. Subtle problem with spinal cord as well as cerebellum.

- Type SCA 2 (patients often get cramp)
- Type SCA 3 (Machado Joseph disease
- Type SCA 6
- Type SCA 7
- Type SCA 8
- Friedreich's ataxia

These are mostly dominant ataxias (except FA) and it can be helpful to know which one you have as people can be offered genetic counselling. However, it is still up to individual choice on whether to be tested or not. Blood samples are routinely retested when new genes identified.

Some other SCA numbers (eg 30) occur in only one family.

(ii) Ataxia Telangectasia (recessive) visible blood vessels in the eyes
(iii) Hereditary ataxias with genes as yet unidentified. Runs in the family but cannot find the gene.

Inherited Ataxias can be caused by dominant or recessive genes.
Autosomal Dominant
Only one parent needs to have the gene. If one part of chromosome has it, it will be inherited (eg brown eyes dominant over blue). On average 2 children will have it and other 2 will not have it. There will be a clear family history of ataxia.

Autosomal Recessive
Both parents have to be carriers ie child must have 2 copies of the gene to get it (eg blue eyes). On average 1 child will have it, 2 will carry and 1 will not have it. Don't usually see a family history.

Genetic counselling – may help to know about problems in advance even if continuing with the pregnancy.

Repeat copies of bits of DNA are responsible for most hereditary ataxias. Bit of chromosome makes frataxin (don't know what it does!). Normally pattern 'GAA' is repeated 20 times. In FA the GAA code is repeated 500 times so patients makes less frataxin than they should – this must be important in the cerebellum.

Other SCA types eg SCA 8 have too many 'CTG' repeats and different proteins are probably involved.

3.2 b) Stroke
2 halves of cerebellum joined in the brain stem

3.2 c) Inflammation
- Multiple Sclerosis (MS) is the most common condition caused by inflammation
- Gluten sensitivity eg coeliac disease of the brain (controversial area) but cause and effect not proven yet.
- Paranoplastic ataxia (very tiny cancer somewhere else in the body)

3.2 d) Brain Tumour
- Can be coming from the cerebellum
- From nearby structures eg acoustic neuroma
- Spread from elsewhere in the body eg lung

3.2 e) Head Injury
Can result in direct damage to the cerebellum.

3.2 f) Toxins
- Alcohol

- Drugs eg phenytoin can have long term effect on cerebellum
- Hormonal eg underactive thyroid
- CJD, MSA and other degenerative conditions
- Episodic ataxia

If you have ataxia and drink a lot it might make you feel worse. But it could also make you more steady by calming your nerves!

3.3 Is Ataxia associated with changes in thoughts & emotions?

In some patients only (especially strokes in cerebellum) there may be:

- Slowness of thought
- Lack of emotional ability: laughing outwardly instead of inwardly, crying when watching films or when happy. Also called 'pathological laughter and crying' or 'emotional incontinence'
- Occurs also in stroke, Motor Neurone Disease and MS
- Rarely – there may be disinhibited behaviour.

There can be thought changes (again, some patients only):

- Impairment of executive functions eg planning, verbal fluency, judgement, insight, abstract reasoning
- Working memory difficulties
- Visual - spatial cognition & memory language deficits.
- Problems with thinking about more than one thing at once (disco-ordination)

A neural circuit in brain may be responsible for this.

Some of these symptoms may be treatable
Information may help patients & carers understand what is happening.

3.4 Are there any promising drug treatments?

Unfortunately, there is nothing very dramatic as yet.
Co-enzyme Q10 – showed some promise on FA – but changes not dramatic, eg heart becoming abnormal a bit less slowly.
Vitamin E / Vitamin C - no evidence of these helping, nor N Acetylcysteine
(Information from Nature Clinical Practice Neurology)
No other promising treatments at present

With Co Q10 there was a follow up study over 5 years: 84 treated, 15 not.
Only a trend to slower deterioration for some patients.
Heart looked different but cardiac function was the same.
Co Q10 is now moving to bigger trial – it's promising in Friedreich's Ataxia but not a wonder treatment.
Possible anti-oxidants Vit E and Vit C may be a help but nothing certain yet
So many false hopes are offered eg on internet – this is irresponsible.

The cost of Co Q in the much higher doses needed is £41,000 per year per patient (mega doses).

Dr Stone was asked, if he had ataxia what would he take? He said if he had CA he would eat healthy food to ensure plenty of vitamins in his diet. If he didn't think his diet was healthy he would take multivitamins to include Vitamin C, E and A.

3.5 Other treatments
Many of these are available at the Astley Ainslie Hospital in Edinburgh.

- Physiotherapy
- Occupational Therapy
- Speech therapy
- Dietition advice
- Psychology
- Disabled Living Centre
- Symptom management

4. Treasurer's Report
Since 6 September, there has not been a lot of change. Income £12.61 interest and the usual standing orders from Mr & Mrs Smith and Claire Shepherd, collecting cans from Lasswade and members' subscriptions £33. Elaine Scott wants to set up a standing order and will be sending a large cheque from her Halloween fund raising (to come). Anne-Marie has raised £117 from the sale of knitting. The collecting can from the Laird & Dog and Tom & Doreen's wee box from their house have not been counted yet.

Expenditure: copying charges to come once Penny has finished typing up the meeting reports. Members' treatments from Susan McVicker the Lasswade therapist who will be coming in January will be billed to the Branch (about £15 each) and we will be taking bookings for this.

The Bank statement balance was £1268.90 at 15 November.

If anyone wants to set up a direct debit for donations please contact Frances on 0131 555 5665.

5. Social Events
There will be a Meal at the Laird and Dog following the meeting (4pm). If anyone has other ideas for social events ring Derek on 0131 477 4371.

6. Any Other Business
Penny mentioned Ataxia South Africa who send round an information filled weekly newsletter by email. http://groups.msn.com/ATAXIASOUTHAFRICA

7. Dates of Next Meetings

The next Branch meetings will be held on Saturday 24 January 2009 (with Haggis, neeps and tatties) and Saturday 14 March 2009 at Lasswade.

YOUR BRANCH COMMITTEE

Chairman: Derek Main		0131 477 4371 derek@ataxia-east-scotland.org.uk
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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.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://groups.msn.com/ATAXIASOUTHAFRICA you need an MSN ID for this (eg hotmail address) and have to apply for membership by emailing ATAXIASOUTHAFRICA@groups.msn.com . They are moving to Multiply in February 2009: http://multiply.com/ Worth joining for the excellent information and news every week.

ADDRESS FOR MAILING:

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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

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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