REPORT OF THE MEETING OF THE EAST OF SCOTLAND BRANCH OF ATAXIA UK LASSWADE HIGH SCHOOL CENTRE, SATURDAY 24 MARCH 2012
http://www.ataxia-east-scotland.org.uk

Present: Pete Dalby (Chair), Liz Dalby, John Hunter, John Reid, Kenny Hunter, Hazel Playfair, Tom & Doreen Vandepeear, Derek Main, Frances Wright, Penny Gardner & Dr Richard Davenport.

Apologies: Andrea Bothwell

Chair's Welcome
Pete welcomed everyone, especially Hazel (John & Kenny's sister) as it was her first meeting. Kenny said that he wad diagnosed with ataxia 7 years ago and fights it every day so it doesn't take over his life.

Matters Arising from last meeting (21 January)
We discussed a possible **meet up with the West of Scotland Branch** – Janice Heath is the contact – the Alona Hotel in Strathclyde Country Park is a good venue. Should we do something? Invite them over & bear the costs this time? Only 4 or 5 of us usually go; a shame as it is a very good get together.

Tina Thatcher at our last meeting suggested ideas for **increased involvement by members** & attendance at meetings. Sending out the Christmas cards worked very well and meant more folk attended our January meeting.

Other Matters which we discussed
Doreen commented that the **Branch website & page on the AUK** website badly need updating – Penny acknowledged this & suggested that more people be given the password to the AUK one, which is really easy to edit. The Branch website is more complicated, it requires knowledge of html and Dreamweaver – but she has a few contacts who might be able to help with it. Or if anyone in the Branch is really confident with using a computer she could give them some training.

NB Once Penny's retired next Jan / Feb she'll have more free time for Ataxia stuff! In particular, speakers at our meetings need to be organised in advance & put on the website. It would also be good to fix our proposed meeting dates a year in advance so people can plan for them.

Suggestions for speakers – maybe we could ask someone to talk about welfare benefits, as the situation is rapidly changing (for the worse).

Venue for Branch meetings: We discussed what would happen when the new school comes on line. Will they still be able to offer us a meeting room? Is there likely to be a charge? Do we need to be looking for somewhere else?

Pete hopes that as many people as possible will attend the May AGM. A meal is planned for the Polton Inn afterwards (NB May update – Polton Inn closed for refurbishment, so it's back to the Laird & Dog again!).

Pete has tried 3 times to get our **circulation booster** back from the new member to whom we lent it, to no avail. We might have to write it off ☹

Frances is planning to organise a **boat trip in August with the Seagull Trust** but has not rung them yet.

Treasurer's Report:
Since 21 January there have been £15 in donations and the 2 regular standing orders of £10 & £15 per month (2 months). Outgoings: cost of January haggis meal was £50 and the bank balance at 23 March was £1,236.21.

John H, John R & Hazel came using Gala Wheels & the Branch will cover the transport costs for them to attend meetings. Please note that the Branch will cover any reasonable costs incurred to enable members to attend meetings as this is a very important aspect of our existence.

Speaker – Dr Richard Davenport
Pete welcomed him and said how pleased we were to see him; Dr Davenport said thanks for asking him back – his last visit was 2 or 3 years ago.

He said he'd like to tell us about all the huge developments with Ataxia – there have been some but unfortunately not the tangible benefits that doctors, patients & their families are interested in ie treatments.

Many more indicators of genetic ataxias (SCAs) have been found – last time there were about 12, now over 30. This is helpful in some ways – knowing what the specific gene defect is – even if there are no treatments now, there may be some in the future. Also, having a specific diagnosis can be so important for the patient – this was discussed later in the meeting.

Dr Davenport said he planned to talk about reaching a diagnosis from the doctor's point of view. He discussed a case from some years ago (not local!) of a patient in their 60s who had been well until 5 years ago, a little asthma but nothing else. Smoked a bit, drank a bit (neither to excess) but had become progressively unsteady, difficulty in walking more than a few yards, slurring speech and problems in swallowing. There were no problems with memory, water, bowel, no tremor, pain, numbness or tingling. The patient felt weak

(though tests showed strength was OK) and walked with a broad gait. Brain scans had shown thinning of the cerebellum (atrophy). These symptoms suggested CA.

Dr Davenport had been asked to give a second opinion, hoping to identify a cause and (much less likely) a treatable cause.

There are several steps in diagnosis:
Firstly, identify the mechanism causing the symptoms: genetic or non-genetic. There is not necessarily always a family history with genetic ataxias. There are 3 types of genetic ataxias

1. Autosomal Dominant
2. Autosomal Recessive
3. Mitochondrial (this has been discovered fairly recently)

1. Genetic Ataxias – Autosomal Dominant
This type of ataxia is clearly handed down from a parent (mother or father) with a 50:50 chance of inheriting the gene.

For example, Huntington's disease is an example of autosomal dominant genetic inheritance with 100% penetrance – if the gene is inherited, the person gets the disease (if they live long enough, as Huntington's does not manifest itself until later in life).

Ataxias tend to have less than 100% penetrance, so if the gene is inherited they may not get the disorder but may pass it on to their children.

2. Genetic Ataxias – Autosomal Recessive
Typically with this type of inheritance there is no family history. For example with Friedreich's Ataxia there may be no family history, or if there is, it is remote.

These genetic ataxias result from a sex-linked gene. Females have chromosomes XX and males have XY. Some genetic defects on the X chromosome, inherited by females are not challenged by a Y – so the patient will get the disorder. But if a faulty X is inherited by a male it may be challenged by the Y and the disorder will not appear.

Some genetic disorders are more likely in boys eg Duchennes Muscular Dystrophy – boys tend to develop the full symptoms whereas females carrying it may only have a subtle muscle weakness. Haemophilia is another well known example of a sex-linked genetic disorder; carried by females & manifesting in males.

3. Genetic Ataxias – Mitochondrial

These have been discovered only fairly recently. The mitochondria are part of every cell in our bodies and they have their own DNA. This can go wrong and can result in some ataxic disorders. In general, inherited disorders can be young onset (under 20) or late onset, manifesting themselves later in life.

4. Non-Genetic Ataxias

Dr Davenport showed us an article by Judith van Gaalen and Dr Bart van de Warrenburg in Practical Neurology "A Practical Approach to Late Onset Cerebellar Ataxia" which outlines the steps taken in reaching a diagnosis: http://pn.bmj.com/content/12/1/14.abstract

There are many tests to be carried out, including blood tests and MRI scans – Ataxia UK issued a useful guide to these for doctors in November 2009. Firstly people see their primary care team (GP) then a hospital specialist (secondary care team) and if there is still no diagnosis then someone like Dr Davenport might be asked to help. It's possible that there will still be no diagnosis when all avenues have been explored. This is then called 'sporadic late onset idiopathic cerebellar ataxia' formerly just 'idiopathic CA' which means the doctors do not know what has caused it. Unfortunately, even if a cause could be identified, it's unlikely that there would be any treatment.

Treatable Ataxias

Very few ataxias are currently treatable, and mostly they are the childhood ones:

- Vitamin E deficiency;
- Wilson's Disease, in which copper accumulates in tissues;
- Ataxia caused by a CoQ10 deficiency.

Question & answer session

Several questions were asked.

SCA numbering – each new gene mutation gets a number. Some are incredibly rare. Intermarrying (eg by cousins) in some cultures tends to increase the manifestations. SCA6 produces a 'pure' CA. Liz has SCA8 which is a very rare one mostly found in Dumfries – she doesn't have any family there as far as she knows.

Waiting Lists

When Dr Davenport first came to Edinburgh in 1992, patients had to go to Newcastle for scans and there was a 12 – 18 month wait. There were only 4 neurologists in Edinburgh and it could be a 2 year wait for an appointment.

Now there are 20 people with 'consultant neurologist' in their job title (though they may have other duties as well and be concerned mainly with research

rather than clinical work). There can still be delays in diagnosis but it's a much reduced 12-14 week wait for an appointment and scans are much more readily available.

Gene Therapy
Unfortunately this has not yet delivered the benefits that were hoped for. Some people are being duped into spending £000s on dubious, useless and sometimes downright dangerous gene therapies abroad. Science has advanced but this has not yet translated into useful treatments.

The Importance of Diagnosis
One major point of discussion was the importance of a diagnosis to the patient. From the doctor's point of view, diagnosis of a cause when there is no cure or treatment is depressing – but for the patient, having a 'name' to attach to their symptoms is very helpful. Everyone at the meeting agreed on this. Knowing which gene / SCA type is responsible is important.

Pain
Should you experience pain with ataxia? Dr Davenport said that people could have pain but usually not as a direct result of their ataxia. Immobility causes pain and neuropathy can result from damage to nerve endings. Pain might be helped by physiotherapy, analgesics or other drugs. However it can be difficult to manage.

Genetic History
Penny asked if our genetic history is always what we think it is – ie there might appear to be no family history of ataxia but perhaps we don't know the full story. Dr Davenport said that genetic studies have indicated that biological fathers are not always the same as "dad" in real life.

Onset of Symptoms
Studies have been carried out on people with Parkinson's – the age at which the disease shows itself. I in 20 will manifest under the age of 40, the average is mid to late 60s and 2% or 3% over the age of 80.

Danger of Google
If you enter any symptoms at all (randomly) you will always get MS, MND and Parkinsons at the top of your search results. This is because of the way Google works, and these 3 are the most high profile neurological disorders which ranks them at the top of any searches. It does not mean that a person has them! Our access to knowledge has gone through massive changes over the last 20 years and this is a mixed blessing, with advantages & disadvantages.

Pete thanked Dr Davenport for his fascinating talk – we hope to see him again in a few years' time!

Date of Next Meeting (the AGM)
This was agreed for Saturday 5 May, followed by a meal at the Polton Inn.

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

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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, 9 Colinton Mains Terrace, Edinburgh EH13 9AT