**EAST OF SCOTLAND BRANCH OF ATAXIA UK**
**MEETING HELD AT LASSWADE HIGH SCHOOL CENTRE**
**ON SATURDAY 19 MARCH 2005**

**Present:** Penny Gardner, Andrew Hogg, John Hunter, Derek Main (Chairman), Andy & Lesley Pringle and John Reid.

**Apologies:** Liz & Pete Dalby, Anne Mitchell (West of Scotland Branch), the Quin family, Alan Smith and Frances Wright.

## 1. Introduction by Chairman

Derek explained that several members had been unable to attend: Liz and Pete because their daughter Sarah is moving, Frances and Anne are unwell, Alan Smith has not managed to arrange transport from Kirkcaldy yet and the Quin family are also moving house.

Everyone introduced themselves and Andy and Lesley Pringle were especially welcomed, as new members. Different neurologists and waiting times for first appointments with neurologists were discussed: these ranged from a few weeks in some cases to in excess of 1 year! Drug treatments and their benefits were also mentioned, including beta blockers for reducing tremor / counteracting depression and a new drug, Tizanidine. The effect of stress was found to make symptoms worse in some cases, and confused the initial diagnosis of the condition.

## 2. Report of Meeting on 22 January

This was approved, there was one error: 2005 is the 40th anniversary of Ataxia UK, not the 50th. Derek commented on the Burns supper and the good turnout at the last meeting, he hopes to get a good number of folk attending the AGM on May 14. Please let Derek know if you can / cannot attend on 0131 477 4371 or **CelticDerek@blueyonder.co.uk**

## 3. Matters Arising

Various fund raising events are planned: Derek's brother is doing a 10K run with several friends in May, Penny is organising people for the Great Scottish Walk and Derek plans to repeat his mile on the treadmill at Lasswade at the end of July / beginning of August – he hopes to improve on his best time of 55 minutes. Anyone is welcome to join him to raise funds. Penny is able to prepare **sponsor sheets** for anyone needing them, just pass the details on to her: telephone 0131 332 5218 or email **pennyjgardner@btinternet.com.**

Andy Hogg has a friend from work doing the Edinburgh marathon for a charity which is able to collect funds electronically: this suggestion will be passed on to Ataxia UK.

## 4. Professor Robert Will

Professor Will is attached to the Department of Clinical Neurosciences (DCN) in the Western General Hospital Edinburgh. He has many people with ataxia as patients at the DCN, Borders General Hospital and formerly at St Johns, Livingston. Professor Will trained in England with research connections to Ataxia UK, and latterly has been involved in CJD surveillance.

Professor Will is a neurologist and spoke about the issue of Ataxia in CJD, which is his current specialism. Although not directly related to FA and CA, he gave a most interesting talk on CJD connected with ataxia and other associated issues. **He emphasised that none of our members need to worry about CJD as the symptoms of CJD are easily recognised and diagnosis is made very quickly.**

### Professor Will's Talk:

Creutzfeldt-Jakob disease (CJD) and ataxia
Professor RG Will, National CJD Surveillance Unit
Western General Hospital, Edinburgh

**Creutzfeldt-Jakob disease (CJD)** has become well known to the general public because of concern that one form of this condition, variant CJD, may be caused by transmission of BSE in cattle (mad cow disease) to the human population. However, CJD has been known about for over 80 years and the commonest form, sporadic CJD, occurs all round the world and is not caused by transmission of infection from the environment. Sporadic CJD affects about one person in a million every year and other forms of CJD, such as variant CJD, accidentally transmitted cases and inherited cases are even rarer. CJD is a member of a group of diseases called **prion diseases**, which are unique because the causal infectious agent is not a virus or bacteria but is thought to consist only of a protein. These diseases are rapidly fatal after the illness begins but have a prolonged period between exposure to infection and the development of disease, which can range from years to decades.

Ataxia and other symptoms indicating malfunction of the cerebellum are common in human prion diseases and this is reflected in the criteria that have been established for the diagnosis of sporadic, variant and accidentally transmitted CJD. Historically the first demonstration that these diseases were due to infection was in **kuru**, which was a disease presenting with progressive ataxia in a tribe in New Guinea and which was caused by ritual cannibalism. Professor Will reassured members that this form of transmission was very unlikely to be related to their ataxias!

In **sporadic CJD** about 60% of cases present with ataxia particularly affecting walking but some patients also develop slurring of speech and clumsiness of the limbs. The diagnosis of CJD is made because the **symptoms progress very rapidly** over days or weeks and the individuals become helpless, develop jerking movements of the limbs and die with an average survival of only 4 months from the first symptom. Very rarely ataxia is the only symptom for some weeks and this is called the Brownell-Oppenheimer form of sporadic CJD. The speed of deterioration in sporadic CJD clearly distinguishes this condition from most other causes of ataxia, although it is always important to carry out tests such as a brain scan to ensure that no other diagnosis is missed.

Tragically CJD has been spread from person to person through treatments such as **human pituitary growth hormone**, human dura mater (brain covering) grafts and in a few cases through corneal grafts or neurosurgical instruments. Human growth hormone was used in children in the UK to boost growth. Some pituitary glands taken from people dying of CJD must have been included in the manufacture of the hormone and to date more than 30 people in the UK (and over 160 worldwide) have gone on to develop CJD, often at a young age, after this treatment. The illness always presents with a progressive cerebellar ataxia and unlike sporadic CJD there may be very little evidence of other neurological problems. Patients survive for about a year after the symptoms develop and the diagnosis is usually made because of the history of previous growth hormone treatment. Since 1985 short stature in children has been treated with a synthetic hormone.

Other forms of **accidentally transmitted CJD** look more like sporadic CJD and the explanation for the cerebellar ataxia in human growth hormone recipients may be that the route of infection may determine the clinical presentation. Growth hormone was given by injection while dura mater grafts and neurosurgical instruments come into direct contact with the brain. Interestingly kuru which was transmitted through dietary exposure, another peripheral route, also presented with ataxia.

There are **very rare** forms of CJD in which the condition is inherited from generation to generation and in these forms the disease is linked to mutations in the gene that is involved in the production of prion protein. We all have this gene and large amounts of prion protein in our brains but in people with the mutation it is thought that the protein is unstable and eventually spontaneously turns into the type of protein causing disease. There are over 25 different mutations that cause hereditary CJD and in some the patients present with ataxia, notably a condition called Gerstmann-Straussler syndrome. This condition progresses slowly over many years and in the later stages there are usually other neurological

problems including dementia. The diagnosis is made through genetic analysis.

Finally, ataxia is an important symptom in **variant CJD**, the form of human prion disease that is caused by infection with BSE. This condition affects young people, with an average age of only 29 years, and usually starts with psychiatric symptoms such as depression or anxiety. After about 6 months the first neurological symptom is usually ataxia, which affects walking predominantly, although some patients also have slurring of speech or clumsiness of the limbs. After this many other neurological problems develop such as involuntary movements of the limbs and progressive dementia. The average survival from the first symptom is 14 months.

The diagnosis of variant CJD is usually made because of the clinical features in a young person and investigations such as MRI brain scan can be very helpful. To date 154 people have been affected by variant CJD in the UK (11 case have been found in other countries), but the numbers of deaths per year are declining and a large epidemic of variant CJD seems unlikely. However, concerns have recently increased because of evidence that variant CJD may be transmitted from person to person through blood transfusion.

Although ataxia is a frequent feature in human prion disease, it is important to stress that these conditions are extremely rare and are usually diagnosed promptly because of the rapid progression and the combination of other neurological features. One expert estimated that the chances of a GP seeing a case of CJD was once every 100 years. One matter for concern is the extremely low number of neurologists in the UK compared to other European countries. There are more job vacancies but not enough trained people to fill them. In the short term, maybe the solution is to import qualified people from elsewhere in Europe and further afield.

Professor Will was thanked for a most interesting talk and there were some questions and answers over the tea / coffee break.

## 5. Treasurer's report

Frances was ill but had passed information to Penny. Since the last meeting there has been little change: negligible income including a few members' subscriptions, a £10 donation from Mrs Evershed and the regular standing order from Mr & Mrs Smith. Penny and Frances will get together soon to prepare the accounts for 2004/05 for submission to Ataxia UK.

## 6. Publicity

We need a new poster to advertise the Branch in doctors' surgeries, clinics etc with the East of Scotland name and Derek as the contact phone no.

## 7. Website News

The existing domain name registration expires in May and would cost £60 to renew for a year or £100 for 2 years. John Reid stated that the existing provider gives poor service, so we may let it lapse and start afresh with a new company. As the Branch name has changed, we do not need the 'Elbataxia.com' name anyway. Penny is currently studying on a web design course and offered to redesign a site for her major assignment, this was agreed by the members. She hopes to get links to and from HO and Kate's website. The domain name will need some thought, in order to maximise 'hits' when people in Scotland are searching for information on Ataxia. Penny will ask Ataxia UK for advice and recommendations.

## 8. Fund Raising

Derek has a stock of **collecting cans** which can be placed in work, homes, shops, pubs etc. Please let him know if you would like one or more: 0131 477 4371. Mary Case of the **Blackhall Framing Gallery** is making a donation to Head Office of £1 for every picture framed this year. If you have any pictures that need framing, this is the place to go – it's near the old KwikFit on the Queensferry Road.

## 9. Social Events

Frances has got information on **Seagull Trust (Falkirk Wheel trip),** we need to agree a date – probably a Saturday - and ask them about it. This should be sometime in May or June. There is only one boat available, so the maximum number is twelve, including six wheelchairs and there must be two helpers. We would leave Edinburgh about 12.00 with a minibus, arrive 1pm, boat trip takes 1.5 hours, back by about 4pm. If using the Margaret Blackwood minibus, we would need to have Pete as he's a registered driver. They need to know the number of wheelchairs in advance. See the separate sheet to contact Frances and register for a place on this trip.

Penny is still investigating venues for a **BBQ at Dalkeith Park or Almondell.** These are usually popular family events. We hope members will tell us their suggestions for other **social events**, possibly including one or more joint events with the West of Scotland branch.

## 10. Other matters

Any member of Ataxia UK can apply for a **'Friends of Ataxia'** card, which explains the symptoms of Ataxia: ring Head Office on 0207 582 1444.

SKILL, the National Bureau for **students with disabilities**, has a free helpline: 0800 328 5050. Their web address is at the end of this report.

The **venue** for Branch meetings was discussed, however at the moment the advantages of Lasswade would be hard to beat: fully accessible, plenty of parking, very friendly and helpful staff and no charge made to the Branch. If equivalent (fully checked out) facilities are available elsewhere then alternating venues could be considered, however the depleted committee at present is not able to investigate themselves.

**11. Dates of Next Meetings**
The AGM is set for **Saturday 14 May** at Lasswade. If anyone has ideas for **future speakers**, please let us know. If anyone is willing to help out on the **committee**, please let Derek know.

**Branch Contacts are: Derek Main (Chairman) 0131 477 4371 or Penny Gardner (Secretary) on pennyjgardner@btinternet.com**

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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