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Note of Meeting – Cross Party Group on Epilepsy, 26 January 2012

In Attendance:

Declan Ahern, ESN	Fiona Nicolson, Quarriers
Matt Barclay, Community Pharmacy Scotland	Caterina O'Connor, West of Scotland & Tayside Epilepsy MCN
Sheena Bevan, ESNA & NHS Grampian	Gemma O'Hare, Scottish Epilepsy Initiative
Tom Binns, Epilepsy West Lothian	Allana Parker, JEC Secretariat
Marilyn Bryce, Non MSP Individual Member	Derek Robertson, Pennywell Resource Centre
Gina Freeman, Epilepsy West Lothian	Susan Sheridan, Scottish Epilepsy Initiative
Gerry Gahagan, Quarriers	Jennifer Simpson, Special Products
Kenneth Gibson MSP	Richard Simpson, MSP
Lorraine Kennedy, Observer	Brian Stanage, GSK
Donald Mackintosh, ESN	Jacqui Telfer, Epilepsy Scotland
Jake McLeod, PA to R Simpson MSP	Anissa Tonberg, Epilepsy Scotland
Megan McTiernan, PA to Kenneth Gibson	Paul Wheelhouse, MSP
Pamela Martis, NHS Lothian	Nicolas White, Quarriers
Ann Maxwell, Muir Maxwell Trust	Lesslie Young, Epilepsy Scotland
Shirley Maxwell, Epilepsy Connections	Sameer Zuberi, Fraser of Allander Neurosciences Unit, Yorkhill

Apologies:

Jo Campbell, ESN	Angela Norman, ESN
Malcolm Chisholm, MSP	Pamela Parker, ESN
Murdo Fraser MSP	Lyndsay Ross, Student Learning Disability Nurse
Andy Gibb, Student Learning Disability Nurse	Debbie Service, Lanarkshire Epilepsy Service
Epilepsy Here, JEC	Ferne Swanson, Student Learning Disability Nurse
Jennifer Irvine, NHS Lanarkshire	John Park, MSP
Kevin Kelly, ESN	Sandra White, MSP
Alison McInnes, MSP	Sam Whitmore, Epilepsy Connections
Duncan McNeil, MSP	Sharon Wood, JEC
Helen MacDonald, Lanarkshire Epilepsy Service	Humza Yousaf, MSP
Nanette Milne MSP	

1. Convenor Kenneth Gibson MSP welcomed everyone to the Cross Party Group. Alison McInnes MSP who was to Chair today's meeting had been regrettably taken ill and was wished a speedy recovery. Mr Gibson welcomed guest speaker Dr Sameer Zuberi who was going to discuss epilepsy genetics.

2. Mr Gibson then gave a brief update since the last meeting:

   - Information on the numbers of Epilepsy Specialist Nurses in Scotland has been provided by Sheena Bevan (ESNA). This was circulated to group MSPs and spare copies were available. The draft note from the last meeting is with the speakers for their approval and will be distributed shortly
   - Bob Doris MSP has joined the Group which now has 25 MSP members
   - The Convener met with the Secretariat (Allana Parker) to discuss how the Group can progress various issues including those raised in the November meeting about the recent FAI and SUDEP. Mr Gibson will be writing to the Public Health Minister Michael Mathieson asking for details of how the government intends to take the FAI recommendations forward

- There is a proposed bill to reform Health and Social Care in Scotland. Some organisations present may put in their own submission and ask for amendments to this bill. The CPG on Epilepsy will be interested in how this bill might impact people with epilepsy. MSPs can also table amendments on the Group's behalf and some parliamentary committees like the finance committee may take evidence. As finance committee Chair, Kenneth Gibson agreed this will happen in terms of the financial memorandum. No new health bills will be tabled for the Parliament before the summer
- The Convener is keen to invite the Public Health Minister to a group meeting. He has advised the Secretariat to look at potential topics the Minister may address during the next session and ensure this CPG is in his diary
- The Convener also wants to arrange a group site visit to the new Scottish Epilepsy Centre once it opens. (He was officially involved in a recent reception about it with MSP Joanne Lamont). There may also be a possible joint meeting with the CPG on Mental Health in the next 12 months. He is looking to arrange another debate on epilepsy and the office bearers will consider a suitable topic that members of the group wish to put forward.

3. Mr Gibson introduced Dr Sameer Zuberi, Consultant Paediatric Neurologist in the Fraser of Allander Unit at Yorkhill Hospital as well as part of the Glasgow Epilepsy Genetics Service. Dr Zuberi covered the major revolution in genetics over the last 10-20 years which have made a significant impact on paediatric and adult epilepsy care:
   - Modern genetics in medicine helps clinicians to make a definitive diagnosis for families and individuals. There are also wider public health benefits as genetic testing can actually save money and unnecessary tests, it can change clinical management by helping doctors decide on the correct medication and improve outcomes with a more accurate prognosis. These tests also help with genetic counselling and any recurrence risk for families
   - Genetics is the study of hereditary and variation in living organisms. DNA molecules are made up of "base pairs" which come together in strands to form the famous double helix. DNA molecules are basically the code for human genetic make-up. Sequences of DNA make up individual genes each of which code for the manufacture of different proteins which make up our bodies. With three billion "base pairs" the potential variation in every human being is enormous (even identical twins are completely individual). The brain has the majority (70 to 80%) of 30,000 genetic codes. (Interestingly, human beings have exactly the same number of genes as a rice grain). Human genes are very clever and can do all sorts of different jobs so a gene for epilepsy is not going to just cause someone to have epileptic seizures; it may impact on their learning and behaviour and other processes
   - The DNA code is very complicated and it is quite normal for little changes or mutations to occur. A beneficial change brings evolution and species change. Mutations which cause proteins not to be made properly or function in a wrong way can lead to a genetic disease. Several different types of epilepsy are caused by mutations which change the way proteins operate the electrical signalling of the brain, causing a disruption leading to seizures. Finding such a genetic mutation helps with diagnosis
   - Genetics may also be important in predicting whether a particular drug works or not as certain medications can improve and sometimes worsen seizures in particular genetic epilepsies. Medication in the body is carried by different transporters around the body and to the drugs target. Someone with a mutation in a gene that controls a drug transporter may also not respond as well to anti-epileptic medication. In the future, clinicians will be able to profile individuals for their particular genetic make- up and their responses to different types of drug. We are already able to test for the genetic susceptibility to certain side effects (e.g. bad rash) in specific ethnic groups
   - Dr Zuberi's particular research interest is in the severe epilepsies that begin in infancy. These make up about 10% of all of epilepsies but perhaps 70 – 80% in terms of his workload because these babies often have seizures in the first year of their life. Infantile epilepsies can so impact children that by their second decade of life they may have a significant learning disability, problems with behaviour and movement. Genetic research has found mutations in sodium channels that control

electrical signals in the brain are responsible for the epilepsy in a significant proportion of these infants. In the last decade tests have become available for this particular type of epilepsy

- Dr Zuberi's predecessor, Professor John Stephenson suggested that that those children thought to have whooping cough vaccine damage in fact had an epilepsy that began at the same time. This was subsequently shown to be Dravet's syndrome which begins in early infancy
- Genetic samples used to be sent for testing to Australia by Dr Zuberi and his colleagues. In 2005, the Muir Maxwell Trust funded the purchase of a Genetic Sequencer based at the Duncan Guthrie Institute of Medical Genetics at the Children's Hospital in Yorkhill. The Glasgow Epilepsy Genetic Service is now the primary genetic testing centre for the UK and Ireland. The service also does tests for Australia, New Zealand and other countries worldwide. Six genes are now tested for from 2,500 individuals living mostly in the UK. Today it is a National Services Division funded service and is linked to a research programme, initially funded for two years by the Muir Maxwell Trust, to study outcomes and other related problems in children
- Recent research papers (2011), including one by Dr Zuberi and his colleagues, have focused on the comorbidities of epilepsy for younger children. Doctors are being asked to concentrate on a child's behaviour, attention, concentration and learning problems as well as the seizures
- A service questionnaire to parents asked if getting a genetic diagnosis was helpful. Over 80% agreed it was. Genetic testing also helped to change treatments as examining the drug regime for a specific genetic diagnosis showed certain drugs will work better with a particular condition
- Interestingly, access to therapies and services increased once parents had a specific genetic diagnosis. This should not be the case as access to these should always be based on need
- Dr Zuberi showed video footage. A genetic diagnosis was made in a young child with cerebral palsy and epilepsy who was unsteady walking and took occasional seizures. A gene mutation had affected the passage of glucose into the brain. After a few months of being on a specific diet, called the ketogenic diet, (not drug) the child was walking normally and school work, mood and behaviour had all improved dramatically. Diagnosis by a simple blood test replaced having the child having to fast and then have the insertion of a needle into the base of the spine to check spinal fluid to make the diagnosis
- Looking ahead, the National Services Division plans to expand this service because of greater referrals from the UK and overseas. More tests will be offered that are unavailable anywhere else in the UK. A study is planned to offer children and adults genetic tests at diagnosis. If the child in the video had been genetically diagnosed as an infant, the learning and balance problems may not have happened because treatment could have started during infancy.

4. The Convener thanked Dr Zuberi for his fascinating presentation and initiated questions:

   Kenneth Gibson MSP asked about the criteria for test referrals since demand may exceed supply. Dr Zuberi said that although 60% of childhood epilepsies are genetic in nature many of these do not have a genetic cause as yet. The most important group for testing at present are infants and young children who can have difficult to control seizures and develop learning problems. Genetic testing technology is moving on very rapidly so within six months 'next generation' sequencing is going to start in Glasgow both for breast cancer screening and epilepsy genetics screening. The limitation is going to be the interpretation of all this data by the team of professionals who work together closely. Epilepsy genetics is at the forefront compared to many other conditions. Genetic profiling will be important for all aspects of an individual's care and reflect how medicine is likely to change in 10- 30 years.

   Mr Gibson wondered if genetic testing would help determine whether someone actually has epilepsy given 23% of people in Scotland are misdiagnosed as having epilepsy and actually don't. Dr Zuberi said that deciding on whether and individual has epilepsy is based primarily on the history, particularly from the witness and the skills of the person taking the history. This can be supported by tests such as an EEG. He agreed that genetics may well help in deciding what

type of epilepsy people have. The gold standard test is an EEG while someone is having a seizure if that's possible.

Dr Richard Simpson MSP invited Dr Zuberi to describe Dravet's Syndrome in more detail. Dr Zuberi explained how Charlotte Dravet discovered a specific pattern of features in children with a learning disability and later a gene mutation was found. Dr Simpson asked how many of the six genes tested were linked to specific syndromes. Dr Zuberi said one of the complications with epilepsy and genetics is that a particular epilepsy syndrome may be caused by more than one gene. Several of the genes have more than one syndrome attached to them.

Dr Simpson remarked that this clearly had implications for the development of appropriate drugs based on a more genetic approach. He asked if the epilepsy genetics service, which was clearly cutting edge, had considered work with the pharmaceutical industry. Dr Zuberi said that there was no specific work directly with any particular pharmaceutical company but that genetics research is collaborative and other labs do work with pharma companies. Historically all the drugs used for many years often have been found by accident but one newer epilepsy drug specifically works with a mutated type of epilepsy, so there is a more focused approach. Dr Zuberi agreed with Richard Simpson's suggestion that promoting more pharmaceutical genetic work would take Scotland forward. Dr Zuberi said people within Scotland are beginning to see epilepsy is not the orphan condition of the past but important scientific discoveries can be made from its laboratories. Stem cell technology will also become an important area for collaboration.

Paul Wheelhouse MSP, who took phenobarbital for the first 12 years of his life, identified with Dr Zuberi's points. He mentioned that NHS Dumfries & Galloway do not have an epilepsy specialist nurse to support people with epilepsy. He questioned a reliance on GPs to spot epilepsy symptoms. Dr Zuberi explained that diagnosis should be made by someone with an expertise in epilepsy who has appropriate training. Specialist nurses are key in providing continuing care to individuals. This same point is echoed by parents.

Ann Maxwell praised Dr Zuberi for devising an effective questionnaire which helps identify potential candidates and obtain relevant samples for the DNA test. It is being translated and used worldwide. She also applauded his major breakthrough with Dravet's syndrome by utilising a third drug in conjunction with other medication which radically helped her own son five years ago. Dr Zuberi mentioned that genetic testing had stopped some drug use by matching the right medication for specific types of epilepsy. Ann Maxwell commented that five years ago, a test result done in an Australian research lab was available after 3 years. Some commercial labs charge $2500 for the test. Today it is free and takes 30 working days, which is phenomenal. Kenneth Gibson applauded Dr Zuberi and his team for their work, energy and enthusiasm and putting the Glasgow service at the forefront of these developments.

Sheena Bevan mentioned two new Epilepsy Specialist Nurses had been appointed since the last meeting (paediatric post in NHS Borders and adult post in NHS Forth Valley). As an adult ESN, she asked if consultant neurologists in Scotland could send blood tests to the service from adults that were not diagnosed as infants. Dr Zuberi agreed that they could and samples tended to come from Edinburgh, Dundee, Aberdeen and Glasgow as well as other parts of the UK. A recent research paper in the neurology journal 'Brain', written in collaboration with Queens Square colleagues in London, found improvements in adults with epilepsy in terms of their seizure control, behaviour, mood and quality of life, once they had a specific genetic diagnosis. Sheena Bevan asked about changes in medication for adults following a genetic diagnosis and how things improved. Dr Zuberi remarked that not only could seizures improve but where he changed medication for some teenagers they now talked more and engaged more in life.

Allana Parker asked about future funding of the service. Dr Zuberi said that the National Services Division, which funds genetic testing nationally, has monitored the referral rate and recognised the service was clearly helpful and beneficial and has agreed to future investment allowing the service to expand.

Paul Wheelhouse asked if genetics will become sophisticated enough to identify the specific type of epilepsy in an individual and deliver a specific treatment to stop it so the person might have the

ability to drive and have greater economic participation in life. Dr Zuberi said it may. He gave an example of benign epilepsies which can respond well to medication, children may grow out of them and this takes away a lot of the worry and anxiety for families.

Dr Simpson asked if there was any charge for the service. Dr Zuberi said there was no charge within Scotland because it is a specialist DNA service funded by the NHS National Services Division. Samples from England, Wales and Ireland are charged. Some compassionate testing was done for neurologists in third world countries. Dr Simpson suggested it would be useful to record these figures because it demonstrates Scotland is doing things on an international stage.

Ann Maxwell questioned if it would be possible to prevent a mutating gene happening during pregnancy. Dr Zuberi listed various factors that cause genetic change. In his opinion it would not be possible to completely prevent all mutation because this is a normal thing for evolution. In the future with new technology, there is a possibility of gene therapy in the brain, perhaps changing a mutated gene so it begins to function more normally.

Ann Maxwell mentioned her trust-funded research partnership using data collected with the epilepsy genetic service and Edinburgh University. It will look at areas of prevention and possible causes, including environmental and lifestyle factors, for some of these complex childhood epilepsies. Dr Zuberi said there is always an argument about whether something is genetic or environmental but in reality genetic and environmental factors work together to influence the human body. A new national study will look at all new cases of epilepsy (in collaboration with Dundee, Edinburgh and Aberdeen). It will compare the Scottish index of multiple deprivation in those cases as well as any social economic links. Findings will help with thinking how to target epilepsy services to children whose epilepsy is uncontrolled, perhaps with more outreach clinics in health centres.

5. Kenneth Gibson MSP moved onto parliamentary business and how to take things forward. The finance committee were considering fiscal sustainability. The 20% most deprived areas put an 80% demand on services. The introduction of preventative spending measures in some of these areas could reduce some of the health and equalities which also reduces crime. He suggested for National Epilepsy Week (20-26 May) having a member's debate on issues relating to epilepsy such as genetics, the number of ESNs and their impact, perhaps one area could be SUDEP or other topics. Group members will try to ensure that the profile of epilepsy remains high within the Scottish Parliament. Ideas are to be fed through the Secretariat.

6. The Convener thanked everyone for attending. The date of the next meeting is Thursday 26 April 2012 at 1pm in Committee Room 2. The speaker will be Dr Andy Elder who will discuss dementia and epilepsy in later life.