Cross Party Group on Rare Diseases

Scottish Parliament, Tuesday 29th April 2014, 5.30pm-7.00pm

Agenda

1. Introductions and apologies

Malcolm Chisholm MSP welcomed all in attendance and noted apologies from Jackie Baillie, Nanette Milne and Bob Doris. Apologies had also been received from Arlene Smyth, Lindsay Lockhart, Allan Clark, Jonathan Berg and Joan Fletcher.

2. Scottish plan for rare diseases – Elizabeth Porterfield, Scottish Government

Elizabeth Porterfield provided an update on the progress of the Scottish Plan for Rare Diseases. A draft discussion document has been circulated to most members of the CPG and the Scottish Government are engaging with a number of stakeholders to gain feedback. In collaboration with Rare Disease UK, two consultation meetings with patient groups have been scheduled to provide the Scottish Government with feedback on the draft plan. The meetings will take place on May 6th and May 12th.

Elizabeth Porterfield explained that the other home nations were working to produce their plans – Wales are undertaking a three month consultation on their draft plan and England have released a statement of intent. It is hoped that, pending Ministers agreement, the Scottish Plan will be complete before Parliamentary recess begins in June.

ACTION: Natalie will circulate the Scottish Government's discussion document and details of the consultation meetings to Cross Party Group members.

3. Rare disease research in Scotland – Alan McNair, Chief Scientist Office

Alan McNair, Senior Research Manager from the Chief Scientist Office provided an overview of the work that the CSO carries out in relation to rare diseases. The CSO is part of the Scottish Government Health and Social Care Directorates and supports high quality research aimed at improving the quality and cost-effectiveness of services offered by the NHS in Scotland. The CSO funds project grants, research and clinical academic fellowships. CSO also contributes financially to OSCHR, to allow Scottish researchers to bid for funding awarded by NIHR.

The CSO has a budget of approximately £69 million and decisions for funding research projects are subject to external peer review. Funding is not targeted to particular disease areas. Allan estimated that around 10% of funding goes to rare disease research projects, but there is not a given amount allocated to rare disease research. The CSO engages with the Third Sector in identifying and developing research opportunities- for example, the CSO has worked closely with Action Duchenne and the Muscular Dystrophy Campaign to support a research fellowship. The CSO also funds genetic research nurses in Scotland's clinical genetics centres.

The CSO regularly reviews its research strategy and will be doing so this year. The CSO will be meeting with representatives of Genetic Alliance UK in the near future to discuss research into rare diseases. Alan McNair explained that the CSO have had significant involvement in the development of the Scottish Plan for Rare Diseases.

4. NIHR Clinical Research Network: Genetics – Dr Gillian Borthwick

Dr Borthwick provided an overview of the work of the NIHR UK Rare Genetic Disease Research Consortium Agreement 'Musketeer's Memorandum'. The Musketeer's Memorandum is an agreement between the UK's 23 genetic centres to link as one consortium to benefit rare genetic disease research by reducing the length of time taken to receive approval for a research proposal.

To be covered by the Musketeer's Memorandum, a research proposal must fulfil certain criteria. The research must involve a rare disease (1 in 2000 incidence), be low risk and low cost and be a non-clinical trial of investigational medicinal product.

The approval processes is considerably quicker under the Musketeer's Memorandum, with studies likely to be approved within four weeks. This is a considerable improvement as prior to the Memorandum it could take months, or even years, for research a research proposal to be approved. Further details of the Musketeers Memorandum and the process can be seen in Dr Borthwick's presentation (attached).

Further information can be found at the British Society for Genetic Medicine website.

5. Patients and research– Patricia Osborne, Brittle Bone Society

Patricia Osborne, CEO of the Brittle Bone Society provided an insight into the charity's experience of research. The Brittle Bone Society supports patients with Osteogenesis Imperfecta, a genetic bone disorder characterised by fragile bones that break easily. Patricia spoke of the BBS' ambitious educational campaign to raise awareness of the condition in schools across Scotland's 32 education departments.

Patricia gave an overview of the NIHR funded RUDY study (a study in rare diseases of the bones, joints and blood vessels) that the BBS have been involved with. Patricia emphasised the importance of involving patients at each stage of research – including developing research programmes and ensuring patients receive feedback on the results of their participation. In the case of the RUDY Study, the BBS was a conduit to circulate information regarding the study, to begin the steps to get the project of the ground and to monitor the development of the study. Patricia's presentation is attached for further information.

Patricia welcomed Penny Clapcott, a BBS member, to the meeting and invited her to share her experience of the RUDY project and her experience of taking part in the 2012 Paralympics opening ceremony. Penny's video can be viewed here.

6. Cystic Fibrosis Transplant Campaign – Yvonne Hughes, Cystic Fibrosis Trust

Yvonne Hughes shared details of the Cystic Fibrosis Trust's 50th anniversary campaign. Yvonne explained that the Trust had been founded in 1964 to promote research and improved clinical care. In Scotland, there are approximately 1000 families living with cystic fibrosis but approximately 1 in 24 people carry the cystic fibrosis gene. As it is rare for a patient with cystic fibrosis to reach the age of 50, the Cystic Fibrosis are celebrating 50 years with a 'no party, party' and are inviting supporters to 'blow' up a virtual balloon – this can be done here. Attendees at the CPG were invited to put on a campaign t-shirt, blow up a balloon and have their photo taken for the campaign.

7. Matters arising, member's updates & AOB

Registries: Following the presentations on research, there was some discussion on the use of registries for rare diseases. Allan McNair explained that the CSO did not directly fund the establishment of registries, but that the SHARE register was an exa mple of a national register in Scotland. SHARE is a NHS Research Scotland initiative to establish a register of people willing to take part in health research. A number of members around the table spoke of specific disease registries being supported by the third sector, an example being the Cystic Fibrosis register. Liz Porterfield spoke of the difficulties in funding a national rare disease register and consideration was given to the merits of coordinating the current registries that are in place.

My Condition, My DNA: Natalie Frankish shared details of Genetic Alliance UK's 'My Condition, My DNA – Shaping genomic sequencing in UK healthcare' project. The project requires patient and families to take part in a series of engagement sessions involving interactive tutorials, answering survey-type questions and live chats. The project is looking to recruit Scottish families. Natalie circulated a project brief to members and invited anyone interested in the project to contact Alice Hazelton (alice@geneticalliance.org.uk)

8. Date of next meeting

The next meeting of the Cross Party Group on Rare Diseases will be **5.30pm on Tuesday 17th June**. The topic for the meeting is coordination of care. Confirmed speakers for the next meeting are Deirdre Evans (Director of National Services Division) and Lauren Roberts (Coordinator, SWAN UK).

9. In attendance:

Name	Organisation
Gill Borthwick	NIHR Genetics Speciality Group
Rebecca Bramhall	Brittle Bone Society
Penny Clapcott	Brittle Bone Society
Dan Farthing	Haemophilia Scotland
Natalie Frankish	Rare Disease UK
Steve Harris	Fragile X Society
Yvonne Hughes	Cystic Fibrosis Trust
Marie McGill	Single Gene Complex Needs Service
Alan McNair	Chief Scientist Office
Rae McNeirney	Genetic Alliance UK Trustee
David Mills	PSP Association
Patricia Osborne	Brittle Bone Society
Richard Petty	NHS Greater Glasgow & Clyde
Liz Porterfield	Scottish Government
Yvonne Robb	Molecular Medicine Centre, Edinburgh
Euan Scott	Fragile X Society
Lynn Shields	Tuberous Sclerosis Association
Sandra Thoms	Fragile X Society