Scottish Parliament

Cross Party Group on Rare Diseases

Minutes of Meeting: October 1st 2013

1. Introduction:

Malcolm Chisholm welcomed all present and invited those in attendance to introduce themselves.

2. Update on status of Cross Party Group on Rare Diseases

Malcolm Chisholm explained that the CPG on Rare Diseases had been given formal recognition at a meeting of the Scottish Parliament's Standards, Procedures and Public Appointments Committee. It was noted that there had been some confusion over the financial information required by the Committee, however this has now been clarified and the necessary information has been provided and approved.

3. A Strategy for Rare Diseases

Malcolm Chisholm explained that a key issue for the CPG on rare diseases would be the work currently being undertaken to develop a strategy for rare diseases in the UK and the subsequent implementation plan that will be developed for Scotland. Invited speakers, Alastair Kent OBE, Chair of Rare Disease UK and Elizabeth Porterfied, Head of the Scottish Government's Planning and Quality Team were introduced and welcomed to the meeting.

Alastair Kent OBE: Chair, Rare Disease UK

Alastair Kent provided a short introduction to the work of Genetic Alliance UK and Rare Disease UK before providing background information on the European context of a Strategy for Rare Diseases. In 2009, the UK adopted the Council of the European Union's Recommendation on an action in the field of rare diseases, which requires member states to establish and implement plans or strategies by the end of 2013.

Mr Kent explained that in February 2013, Earl Howe established a stakeholder forum to pull together a UK strategy for rare disease. The target date for publication of the strategy is mid-late November 2013, ahead of the Council of Ministers meeting in early December where member states will be assessed on their work to date. It is expected that the stakeholder forum will reconvene to monitor the implementation of the strategy.

Mr Kent explained that in Scotland, approximately 300,000 people will be affected by a rare disease. A recent Rare Disease UK report highlighted that in Scotland just 30% of rare disease patients receive a quick diagnosis (under 3 months), with one in five patients having to wait longer than 5 years to obtain a correct diagnosis. Mr Kent highlighted that often patients with rare diseases find it difficult to find up to date and

accurate information on their condition and that, with most rare diseases being multi-system disorders, patients experience a lack of coordination of care. Mr Kent explained that better coordination of services for rare diseases would result in improved efficiencies within the NHS, with timely and appropriate interventions occurring in the right patients and the right time.

Elizabeth Porterfield, Scottish Government

Elizabeth Porterfield spoke of the work that the Scottish Government has been doing to develop the UK strategy for rare diseases. The policy context for rare diseases in Scotland was explained, including the 2010 Quality Strategy and the '20:20 Vision' which focus on safe, effective and personal care with a commitment to working closely with the third sector in Scotland. It was noted that there is currently a lot of work going on within the Scottish Government regarding The Public Bodies (Joint Working)(Scotland)Bill which will see the integration of health and social care in Scotland.

Elizabeth Porterfield spoke of the work that has been going on around access to medicines in Scotland, including the recent Health and Sport Committee Inquiry into access to new medicines and the reviews by Professor Swainson and Professor Routledge into the IPTR process and the SMC appraisal process. It was noted that an announcement from Alex Neil MSP should be expected in the coming weeks regarding the outcome of the reviews.

An overview of the NHS structures in Scotland was provided and Elizabeth Porterfield explained that there were a number of structures in place to support patients with rare diseases. For example, Scotland benefits from local, regional, cross-border and UK service delivery arrangements. There are four genetics centres in Scotland that deal with a great number of rare diseases and managed clinical networks exist for a number of rare diseases. There are also nationally commissioned molecular genetics and cytology and Molecular Pathology Laboratories which provide well developed laboratory provision and quality services.

Elizabeth Porterfield spoke of the close relationship between National Services Division(NSD)and England, explaining that NSD and the Scottish Government have been involved in developing the UK Strategy for Rare Diseases as part of the Stakeholder Forum established by the Department of Health. Whilst recognising that Scotland has a lot to be proud of, it was also noted that there are some things that Scotland cannot do alone. For example, the very small number of rare disease patients in Scotland makes the development of specialised commissioning of services for rare diseases extremely difficult. Patient registers were also cited as an area of difficulty due to IT infrastructure and data protection - IT architecture for an all condition register (or for many smaller, disease specific registers would need to be developed, financed and maintained at UK level. It was also noted that research on many rare conditions is likely to need multi-centre approaches between the 4 countries and the EU.

Elizabeth Porterfield explained that following the publication of the UK Strategy for Rare Diseases, there will be work to develop a Scottish specific implementation plan, which they expect to publish in time for Rare Disease Day (28th February)2014.

KEY POINTS FROM DISCUSSION

- It was noted that a debate on access to new medicines would be taking place in the Scottish Parliament on Wednesday 9th October.
- Alastair Kent noted that access to medicines is a hugely important issue and that, whilst it extremely important to have a process for assessing medicines that is robust, transparent and fair, it is necessary to look at the wider impact of a medicine. Alastair gave an example of a six minute test where a patient, as a result of taking a particular medicine, gained a six minute increase in the amount of time they could walk unaided – it was noted that although this may seem to be a small increase, the impact on the patient was significant with the patient perhaps being able to walk to the shops without a carer.
- Elizabeth Porterfield was asked whether the Rare Disease Fund had supported many rare disease patients, other than those originally funded for Ivacaftor (for Cystic Fibrosis). Elizabeth explained that it was her understanding that other patients, with other conditions, had been able to access the fund.
- Joan Fletcher from AGSD UK asked whether it must be a Scottish Clinician who submits an Individual Patient Treatment Request. Elizabeth Porterfield indicated that it did not have to be.
- It was noted that clarity of a referral pathway is key and that it was hoped the Strategy for rare diseases would have provision for this.
- Patricia Osborne (Brittle Bone Society), shared the success of the patient passport for patients with OI. Elizabeth Porterfield asked that details were emailed to her and that this sort of project would certainly be considered. Susan Warren (Haemophilia Scotland), explained that haemophilia patients were benefiting from national specification and a clear patient pathway. Elizabeth Porterfield said that she was very happy to hear of the positive work being done within the third sector and was keen to have third sector involvement.
- It was noted that there is a strong informal network of expertise in Scotland, particularly between the four genetics centres, but that it would be difficult to formalise such networks. It was agreed that technology, such as email, was key for sharing information.
- Scotland's Managed Clinical Networks were given as an example of good practice in Scotland and a model that should be developed further.
- It was noted that the UKGTN had published a very useful guide to centres with specialist expertise for rare diseases.
- A question was asked about how to raise awareness of rare disease. GPs may be likely to see very few rare diseases in their career and are taught to look for an unusual presentation of a common condition. It was noted that it would be very difficult for GPs and clinicians to know about every rare disease. A 'decision support programme' with a series of prompts may be a useful tool to improve diagnosis. It was noted that raising awareness of rare diseases was important amongst nurses, midwives etc and not just amongst GPs. Training days, such as a day focusing on congenital anomalies, were given as an example of a better way to get buy in from health specialities than training days for specific rare conditions.
- It was noted that there is nothing within the undergraduate nursing curriculum that focus specifically on rare diseases – however, a study day for 70 nurses

from different is due to take place this year, with the hope of rolling this out going forward.

- It was noted that patient registers were often held by clinicians and that perhaps this could be explored to assist the Scottish Government in developing plans for a patient register under the Strategy.
- A question was asked regarding newborn screening and whether more conditions would be added to the current list. It was noted that any changes to the current list of conditions would be determined by the UK Newborn Screening Committee.

4. Arrangements for next meeting

It was agreed that, due to the high level of attendance, future CPG meetings should be held at 5.15pm on Tuesday evenings. The proposed date for the next meeting, pending room availability, is Tuesday 3rd December.

Those in attendance were invited to suggest topics for future meetings. Suggestions included access to new medicines, planning and commissioning of services and research networks in Scotland.

With an announcement on the access to new medicines work expected, it was suggested that Alex Neil MSP be invited to the next meeting to discuss the Scottish Government's response to the recent access to new medicines review.

Natalie Frankish will explore the suggestions for topics for the next meeting and approach potential speakers for availability.

LIST OF ATTENDEES: CROSS PARTY GROUP ON RARE DISEASES, OCTOBER 1ST 2013

MSPs in attendance:

Malcolm Chisholm MSP (Co-convener and meeting Chair)

Nanette Milne MSP

Aileen McLeod MSP

Apologies were sent from Bob Doris MSP.

Invited Speakers:

Alastair Kent OBE: Director of Genetic Alliance UK and Chair of Rare Disease UK

Elizabeth Porterfield: Head, Planning Team, Scottish Government

In attendance:

Natalie Frankish:	Rare Disease UK (Secretariat)
Joan Fletcher:	AGSD UK
Patricia Osborne:	Brittle Bone Society
Lesley Loeliger:	PNH Scotland
John Eden:	Scottish Huntington's Association
Karen Ferguson:	Funny Lumps
Gail Currie:	Funny Lumps
Cathy Watt:	West of Scotland Genetics Service
Jonathan Berg:	Clinical Genetics, Dundee
Grace McLeod:	Single Gene Complex Needs Network
Karen Swan:	Genzyme Therapeutics
Paul Currie:	Scottish Government
Lynn Shields:	Tuberous Sclerosis Association
Kathryn Howieson:	Patient Representative
Rae McNairney:	Trustee, Genetic Alliance UK
Susan Warren:	Haemophilia Scotland
Heather Noller:	Carers Trust
Prof. Fiasal Ahmed:	Consultant in Paediatric Endocrinology, Glasgow
Michelle Conway:	Alexion
Jane Cox:	Genzyme Therapeutics

Apologies:

Arlene Smyth:	Turner Syndrome Support Society
Allan Clark:	Laurence Moon Bardet Biedl Society
Yvonne Hughes:	Cystic Fibrosis Trust