Cross Party Group on Rare Diseases

Tuesday 8th March 2016

Adam Smith Room, Scottish Parliament

1pm-2.15pm

Minutes

- **Welcome and Introductions**

  Bob Doris welcomed all in attendance and invited members to introduce themselves.

- **Rare Disease Day**

  Natalie Frankish provided feedback on the recent Rare Disease Day Parliamentary Reception. She explained that it had been the most successful to date, with over 130 in attendance.

  She noted thanks to Malcolm Chisholm for hosting the event and thanks to the Minister for Sport, Health Improvement and Mental Health for taking part in a successful 'Twitter Takeover' and for representing the Scottish Government at the Reception. It was noted that the reception was the forum at which the Minister announced that the Scottish Government would be providing further funding to expand genome sequencing technologies in Scotland, including the involvement of Scottish patients in the 100,000k Genomes Project.

  Members shared their feedback from the event. Positive remarks included the opportunity to meet other patient groups, feeling like a community, feeling as though the collective voice of the rare disease community had been heard and that it was excellent for the event to be used by the Scottish Government to make such a positive funding announcement. It was also noted that more chairs would be beneficial at future events!

- **Scottish Genomes Partnership**

  There was discussion on the announcement that there was to be a £6million investment in the Scottish Genomes Partnership (SGP). The SGP is a collaboration of Scottish Universities and the NHS capitalising on £15 million investment in whole genome sequencing technology by the Universities of Edinburgh and Glasgow. The Scottish Government is contributing £4 million and the Medical Research Council, £2 million.

  By combining knowledge of the whole genome sequence – or the entire genetic code – of patients and information from their health records, genetic diseases can be understood better and new ways to test, manage and treat these diseases devised. SGP will be using this technology for genomic research on rare diseases, cancers and Scottish populations, and to work with Genomics England on the diagnosis of patients in Scotland with rare genetic diseases.

  Members reported feeling encouraged by this news and that it was important for patients and patient groups to be involved in how this project develops. There was appetite for further information and it was agreed that the first CPG in the new session should focus on this topic, with invited speakers from the Scottish Genomes Partnership.

- **Genome Sequencing – What do cancer patient's think?**

  Natalie Frankish presented Genetic Alliance UK's Patient Charter on Genome Sequencing and shared copies of the Charter with members. It was noted by members that although the Charter focused specifically on Cancer, many of the findings resonated with other rare diseases. It was agreed that the Charter could be discussed further at the next CPG meeting which will focus on Genome Sequencing and the Scottish Genomes Partnership.

- **Access to New Medicines update**

  Bob Doris provided an overview of the work that the Health and Sport Committee has overtaken in relation to this topic and noted the continued interest of the Cross Party Group in this issue.

  There was agreement that SMC process change had been very welcome and that new processes, such as the Patient and Clinician Engagement Process, had been an improvement on the old system. However, concerns were raised about how PACE was being evaluated and it was noted that, at this stage, it wasn't possible to say for sure whether the PACE process has been the reason that more medicines had been accepted by SMC.

  Concern was also noted that the Peer Approved Clinical System was still not implemented and the lack of information about the transition including details of the pilot and operational guidance was a frustration to patient groups advising patients on this issue.

  It was noted that an independent review would be taking place in March, with Dr David Montgomery announced as the lead for this review.

  Natalie Frankish also shared details of Genetic Alliance UK's Patient Charter on Access to Medicines in Scotland which would be launched on March 23rd 2016.

  Thanks were noted for the MSP members of the CPG that had been involved in the Health and Sport Committee (Bob Doris, Malcolm Chisholm, Nanette Milne and Richard Lyle).

- **Specialist nursing in rare diseases**

  Natalie explained that the first stage of consultation on specialist nursing had been undertaken – this involved inviting comment from patient groups. It was noted that there had been a lot of interest in specialist nurses for rare diseases – a 'general' post that provides support to a variety of rare conditions, rather than being condition specific. The model of the Single Gene Complex Needs network was seen as a best practice model. It was noted however, that this coordination role, may not be limited to specialist nurses and there may be scope for third sector organisations to provide this kind of support.

  Natalie explained that the next stage was to gather clinical opinion and that work was already underway to map specialist nurses for rare diseases across Scotland.

  In the new session, the CPG will report it's findings and present recommendations.

- **Any other business**

Sue Rees thanked the CPG for it's support of the campaign for a consultant immunologist in NHS Lothian. Sue noted that an appointment to the post had been made.

Thanks were extended to Malcolm Chisholm for his ongoing support for rare diseases and the CPG members wished Malcolm all the best for his retirement.

Natalie explained that the CPG would be dissolved until to new session of Parliament, when the CPG will need to be registered again.