Cross Party Group on Rare, Genetic and Undiagnosed Conditions

Wednesday 3 March 2021 (10:00am - 11:30)

MINUTES

- **Welcome and introductions**

  Bob Doris MSP welcomes all in attendance. The meeting, taking place on Zoom, was the last meeting of the Cross Party Group (CPG) on Rare, Genetic and Undiagnosed Conditions in this Parliamentary session.

- **Minutes from last meeting**

  The minutes of the meeting held on 27 January 2021 were agreed.

  Natalie Frankish provided an update following the discussion around vaccination at the last meeting and the key messages from a Genetic Alliance UK meeting held with Scottish Government on 29 January 2021. This included a reassurance about the safety of the Covid-19 vaccines. Any concerns about allergies and underlying conditions should be discussed with GPs or specialist clinicians. Vaccines have not yet been approved for use in children but trials are ongoing. Invitations to vaccination are now being offered to adults in the JCVI group six. This priority group has been expanded to include individuals with mild learning disabilities. Anyone that has not received an invitation for a vaccination, but believes they should have, should contact their GP to discuss.

- **Presentation: The Future of Genomics in Scotland**

  Professor Miedzybrodzka gave an overview of the Scottish genetics services. Genetics services for NHS Scotland are based in four centres, with a team of genetic scientists, doctor and counsellors in each centre. Funding for services comes directly from territorial boards. Bridge funding was planned to enhance genetic testing, but plans were changed due to coronavirus.

  In Scotland, a lot of testing is done by mainstream services; more than in England despite having the same criteria. The current testing budget is approximately 15million pounds. NHS England recently tripled genomics investment, while Scotland is still making a plan to implement the recommendations of the SSAC Review: Informing the future of genomic medicine in Scotland.

  Professor Miedzybrodzka gave some background on genetics and genetic testing. Genomic testing for rare disease is focused on exons, portions of DNA that code how proteins work. 10,000 genes have been implicated in rare disease and looking at them together would be better to diagnose some patients. Exon sequencing is being done more because it focuses on relevant genes and is cheaper.

  Genetic testing is increasingly becoming part of mainstream healthcare and this needs to be part of how we move forward. Exome testing could become a standard service with rapid access, along with genome sequencing, if sufficient funding is in place.

Further bridge funding has been requested and a decision is expected soon. The future of the service would benefit from longer-term, sustainable funding to allow the service to grow in response to patient need.

- **Discussion: The Future of Genomics in Scotland**

  Sarah Ogilvie (Scottish Government) noted that a funding request for the service had been received and was under consideration. It was noted that the Scottish Government have accepted the recommendations of the Genome UK policy and work will be undertaken to consider how this policy is implemented in the Scotland context, with a goal of a long-term funding commitment.

  Tony Thornburn (Behcet's UK) raised the question of where to start investigating a disease that has an unknown cause. Professor Miedzybrodzka clarified that the 100,000 genomes project is improving diagnosis. Testing works well for diagnosing single-gene rare diseases such as immunodeficiency diseases. But it is difficult looking at multiple causing genes (variants) that increase predisposition. In the case of Behcet's, the environment may be more important as it is acquired at different points in life.

  Mike Cain (HSP Support Group) asked if findings in Scotland would be shared with other UK nations, as a bigger pool of knowledge can be useful. Professor Zosia clarified that it would depend on when Scottish patients would benefit from working with England.

  Natalie Frankish noted the perceived differences in investment between England and Scotland and asked whether this may result in Scotland falling behind. Professor Miedzybrodzka indicated that Scotland is doing the best it can with the money available. Sarah Ogilvie explained that Scotland uses a different approach from England using smaller investments. An approach with the four nations would also ensure there is consistency and Scotland does not fall behind.

  Bob Doris MSP noted that, whilst it was perfectly acceptable for Scottish services to be delivered differently than elsewhere in the UK and that funding did not have to necessarily match, it was important that outcomes for patients remain the same – for example, the range of tests available and the time taken to access them.

  ACTION: The CPG will write to the Cabinet Secretary for Health and Sport to ask for clarification on how money is spent and how to keep pace with developments elsewhere in the UK.

  A point was raised that diagnosis is just the start of the patient journey and support is required following diagnosis. Professor Miedzybrodzka contributed that there are also gaps during transition from paediatric to adult services and having a nominated person to support care would be beneficial.

  Harriette Campbell (Sickle Cell Support Group) agreed it is important to have a nominated person to take care of individuals. She also noted the value of newborn screening and how her organisation campaigned for newborn screening for sickle cell in Scotland. Bob Doris MSP thanked Sickle Cell Support Group for their work increasing awareness and influencing change.

  Ali Murphy (EDS UK) suggested the use of informed consent and counselling before genetic testing. Professor Miedzybrodzka responded that they use consent forms to discuss complex

tests in a standardised way. Informed consent is encouraged and enforced for particular conditions such as Huntington's Disease that do not have typical presentation. More awareness would be needed across health service.

- **Discussion: Schools returning**

  Natalie Frankish raised that families are concerned about returning to schools. For example, a family with the dad recently having a transplant and classified as extremely clinically vulnerable, had to decide whether to send their child to school or not.

  Bob Doris MSP indicated that in these cases, the general advice is that only the extremely vulnerable individual is asked to shield. Schools should do a risk assessment, but no information on the components of the risk assessment has been provided. Sarah Ogilvie agrees to take this concern back to the shielding team in the Scottish Government.

  Natalie Frankish adds that information from the education department would also be helpful to understand whether support for learning at home would be continued. Arlene Smyth (Turner Syndrome Support Society) explained that schools have cooperated and provided online learning.

  Mark McDonald MSP clarified that the Director of Education in his local authority has suggested that councils would take a sympathetic approach to the issue of school return, aware of the concerns families may have. Scotland does not operate a system of truancy fines and there will not be a hardline approach.

  ACTION: Natalie Frankish to write questions to Sarah Olgilvie to raise with the shielding team at Scottish Government.

- **Rare Disease Day 2021**

  Bob Doris MSP reflected on the Rare Disease Day virtual event held by Rare Disease UK and noted that it was useful to hear from CPG leads in other parts of the UK and that the event highlighted the importance of collaboration in rare conditions.

  Attendants also commented on their positive experience of the Rare Disease Day Parliamentary Event.

  The UK's official Rare Disease Day 2021 video was shared.

- **Cross Party Group Report – Final sign off**

  Bob Doris noted that the group had considered the CPG report 'Improving Care for Rare Conditions in Scotland' and its recommendations calling for an Action Plan to implement the UK Rare Diseases Framework to be in place by the end of 2021, the establishment of a Short Life Working Group to explore the delivery model of a Rare Conditions Coordination Service in Scotland, and the inclusion of a commitment to deliver a pilot of the Coordination Service in the Scottish Rare Disease Action Plan.

  MSP members agreed that the report was ready for publication and there was discussion about the promotion of the report, with Mark McDonald MSP suggesting a press release.

ACTION: Natalie Frankish to prepare a short briefing paper and press release and share with Bob Doris MSP and Mark McDonald MSP.

- **Scottish Rare Diseases Action Plan**

  Sarah Ogilvie (Scottish Government) explained that a progress report highlighting the progress of implementation of the former UK Strategy for Rare Diseases has been published. The new UK Rare Disease Framework was launched on 9 January 2021 and Scottish Government has committed to producing an Action Plan by end of 2021/22. There will be a programme of patient engagement to shape the plan and there will be a role for the CPG in this work.

  Bob Doris MSP spoke of the importance of maintaining momentum and having the CPG play a role in shaping the new plan. He suggested an MSP sponsored event be held in the new Parliament to discuss the issues – and that this meeting could be used as a means to gather support for re-establishing the Cross Party Group.

  ACTION: Following the election, Natalie will arrange an MSP sponsored meeting.

- **Next steps for the Cross Party Group on Rare, Genetic and Undiagnosed Conditions**

  Natalie Frankish explained that the CPG will cease to exist at midnight on 24th March 2021, at which time the Scottish Parliament will be dissolved for the election. The CPG can be re-established within 90 days of the first meeting of the new Parliament.

  Suggestions of topics for future CPG meetings should be sent to the Secretariat by email.

- **AOB**

  Bob Doris, Mark McDonald and Natalie Frankish thanked everyone involved for their work with the CPG and in ensuring the voices of people living with rare disease are heard.

  The final meeting of the CPG in this sitting of the Scottish Parliament came to an end.

Attendance – Cross Party Group on Rare, Genetic and Undiagnosed Conditions – 3 March 2021

Bob	**Doris MSP**	**Convener**
Mark	**McDonald MSP**	**Co-Convener**
Natalie	Frankish (Secretariat)	Genetic Alliance UK
Mike	Cain	HSP Support Group
Harriette	Campbell	Sickle Cell Support Group
Andrew	Deans	NHS Lothian
Gill	Dickson	PSP Association
Fiona	Watt	Primary Immunodeficiency UK
Tony	Thornburn	Behcet's UK
Arlene	Smyth	Turner Syndrome Support Society
Ali	Murphy	EDS UK
Russell	Ritchie	PANS PANDA UK
Karolay	Lorenty	Genetic Alliance UK
Rae	McNairney	Primary Immunodeficiency UK
Katherine	Behl	Alternating Hemiplegia of Childhood (AHC) UK
Zosia	Miedzybrodzka	NHS Grampian/University of Aberdeen
Lynne	Hocking - Mennie	Scottish Genomes Partnership
Joanne	Milne-Toner	National Specialist and Screening Directorate
Sarah	Ogilvie	Scottish Government
Carol-Anne	Redpath	Scottish Government
Kirsten	Patterson	NHS Tayside
Michelle	How	Amy and friends cockayne syndrome and trichothyodystrophy
Michelle	Erskine	The Aarskog Foundation
Amy	Comrie	EDS UK
Hazel	McLachlan	Behcet's UK