Scottish Cross Party Group for Muscular Dystrophy UK

Zoom Meeting – Monday 13 June 2022 – Meeting Notes

Introductions

- Jackie Baillie MSP, Chair of the Scottish Cross Party Group, welcomed people to the meeting.

Context of discussion

- Michaela Regan, Head of Policy and Campaigns from Muscular Dystrophy UK gave an overview of the UK Rare Diseases Framework and the previous Action Plans that have been published by other nations.

Scottish Action Plan

- Next, Alan Burns from the Scottish Government updated on developments regarding the Scottish Action Plan.
- He said that the planned timelines for publication had changed since the last occasion that he came to speak to the group: the intention of a spring publication being delayed. Pandemic recovery across government was cited as one of the reasons for delay. He updated that the new timeline was going to be this Summer, and that a draft has been circulated to stakeholder groups for feedback.
- Running through the priorities of the plan and what developments the team was currently able to update on, the following insight was given:

Faster diagnosis

- A focus on the expansion of genomics to shorten the diagnostic odyssey is a big focus within the plan, making genomics more common place across Scotland. Additionally, improving understanding of new-born screening policy; and expanding Scotland's rare condition registry are also aimed at achieving this priority.

Raising awareness

- This is going to involve improving information that is available about rare diseases in partnership with NHS Education for Scotland, working with organisations to better signpost resources as early as possible along people's NHS careers.

Coordination of Care

- This priority will include identifying suitable models of care coordination for Scotland using a pilot process which will be starting shortly. Additionally, the plan will detail ways to make better use of existing tools such as care passports, inter-disciplinary care planning, and other methods which would be translated well to neuromuscular patients.

Access to specialist care, treatment and drugs

- The main insight given under this priority was that innovation will be more tailored for people with rare conditions, making sure that people in rare disease communities have their voices heard when unlocking barriers to research.

- Overall, the Group was told that most of these areas of focus are for short- and medium-term planning. Then, the plan will be updated with further annual iterations which will reflect on progress and can remain agile to new developments.
- In terms of the engagement, the group was updated that after the first iteration there will also be working groups on implementation and continued work with stakeholder organisations.

Genetic Alliance update on engagement

- Natalie Frankish, Policy and Engagement Manager for Scotland at Genetic Alliance UK gave the group an update on patient engagement events that have taken place to inform the action plan to date.
- She said that around 100 people have participated and have shared what their priorities are, with the outcome being reports from Genetic Alliance UK relating to the action plans. In particular, she drew attention to the recommendations on care coordination including a large focus on implementing a rare conditions care coordination service. Natalie also explained that one of the challenges of involving people well was that there are so many rare conditions and ensuring the actions reflect such wide ranging needs.
- The Cross-Party Group on Rare, Genetic and Undiagnosed Conditions - which is run by Genetic Alliance UK as secretariat - will be continuously reviewing the action plan and monitoring progress.
- Finally, it was noted that the Cross Party Group on Muscular Dystrophy and the CPG on Rare Genetic and Undiagnosed Conditions will be working together to engage on the action plan in a similar way that has been carried out on new-born screening.

Questions and discussion

<u>Timelines and monitoring</u>

- Jackie Baillie MSP asked for more clarification on when the group can expect to see a final itieration.
- AB said that the initial iteration will be in summer, but that the exact date is not certain, and that updates on that iteration will appear annually after that.

- JB asked, if the plans are being updated every year, and to what extent will there be detailed monitoring of what is happening at a health board level.
- AB responded that when the government moves to an implementation phase, the consideration will be on how this is going to be measured.
- AB said that this is part of the discussion that is going to be had at the Rare Disease Implementation Board this week.
- When asked about how this can be done through patient reported outcome and experience measures, and the best way to communicate these to boards, AB said that part of that will continue through the existing patient engagement programme. He also said that things like uptake of training and education resources can be measured and asking healthcare professionals about their awareness about what is available.

- NF reiterated that monitoring is critical to measuring success as, as learning from the most recent Rare Disease Strategy showed many people with a rare disease didn't feel much impact or change. She reflected that there are questions to answer on how this is done, including whether it is done across all different disease areas, and about

how NHS boards in Scotland will report back. She also said that a centralised service to better gather this information will be helpful.

Diagnosis - genomic testing and mental health support

- Turning specifically to genomics, JB said that she has been hearing that testing has been intermittent across Scotland.
- Mark McGregor, co-vice chair of the Scottish Medicines Consortium, highlighted the importance of laboratory capacity in this area, and that the Genomics Strategy Board has been established and recently reshaped, with one of their jobs being to look at testing.

- MR asked how diagnosis will be improved for people who don't have a genetic component to their condition.
- AB acknowledged that the government doesn't want to lose sight of those people in their plans that don't have a genetic basis to their condition.

- MR also asked if there will be a focus on mental health support to help address the impact of a diagnosis coming from screening.
- AB replied yes as this is something that has come up in engagement groups - both searching for a diagnosis, or post diagnosis.

Implementation and continued engagement

- JB asked if there was funding set aside to help deliver implementation of the plan
- AB explained that the team are waiting on health finance to clear and confirm funding.

- MR asked about the continued opportunities to engage on all iterations.
  - AB confirmed that the team will be reaching to ask stakeholders about the best way to carry out continued involvement.

Close and AOB

- JB closed the meeting by saying that she is encouraged by what has been said and is hopeful to see a plan in the summer as soon as possible, with a monitoring framework in place. She also noted that the ability to come back to the plan year after year and make improvements should be seen as a positive.

- NF added an item of any other business on the newborn screening pilot, stating it is progressing well and there should be an update soon with further information