ALL PARTY PARLIAMENTARY GROUP ON RARE, GENETIC AND UNDIAGNOSED CONDITIONS

on Rare, Genetic and Undiagnosed Conditions

An update on the implementation of the UK Rare Diseases Framework and the Rare Disease Action Plans

Monday 20 February 15:30-17:00 (in person), 16:00-17:00 (online)

Parliamentarians in attendance: Liz Twist MP (Chair)

Baroness Pauline Neville-Jones MP

Christina Rees NMP

Pauline Latham MP

Alex Norris MP

Bambos Charalambs MP

Marion Fellows MP

MINUTES

At the start of the meeting, the Group conducted the AGM for the APPG on Rare, Genetic and Undiagnosed Conditions. Attendees reviewed the report and expenditure statement. Liz Twist MP was reelected as Chair of the APPG. The following were put forward and elected as officers:

- Baroness Pauline Neville-Jones MP
- Marion Fellows MP

The following were put forward and elected as officers in absentia:

- Lord Patel
- Catherine West MP

Liz Twist MP, Chair, APPG on Rare, Genetic and Undiagnosed Conditions

Liz Twist MP welcomed attendees to the meeting and noted that this meeting had been rescheduled from December due to the travel disruption caused by the snow and public transport strikes.

Liz Twist MP introduced the topic of the UK Rare Diseases Framework and the Rare Disease Action Plans. The Framework was launched in January 2021 and outlines four priorities and highlights five cross cutting themes which aims to improve the lives of those living with rare conditions.

Each nation committed to publishing a Rare Disease Action Plan to implement the Framework. As of December, all action plans have been launched. Since the original date of this meeting, there have been some considerable updates around the Rare Disease Action Plans.

Nick Meade, Genetic Alliance UK

Nick Meade gave an overview of the UK Rare Diseases Framework. The Framework is a UK-wide commitment signed by the four health ministers from the UK Parliaments in January 2021. The Framework identifies four key priorities and five underpinning themes to improve the lives of people with rare conditions. There is a five year timeline attached to the document with a deadline of 2025. The four nations took that first year to develop their action plans for implementation.

Nick Meade noted two points of concern that appeared in all of the action plans. Firstly, while each of the teams developing the action plans showed enthusiasm to implement the Framework, the interest was not matched by the funding provided alongside them. All of the nations rely on existing budgets to fund the delivery of these plans, meaning that rare conditions will be competing with other groups to secure funding.

All of the nations also indicate existing programmes in their plans. These programmes are crucial but should not be considered part of the plans. We are asking that the delivery partners responsible for implementing the plans engage with rare conditions beyond what already exists in the landscape.

Nick Meade noted some key elements to the implementation of the action plans. Notably, Scotland was the last to launch their action plan in December 2022, leaving only three years of a five year timeline to implement the plan. There are concerns about how much will be achieved in this time. Meanwhile, Wales has indicated in their plan that some of the actions will continue to the end of 2026, this is reassuring as it shows a level of commitment to the continuity of this work from the Welsh Government.

England will release their second action plan on Rare Disease Day 2023, during the development of this plan there has been an increase in patient and public involvement. It is encouraging that policy makers are seeing the value brought through the patient community.

In November 2022, Rare Disease UK Patient Empowerment Group published a paper which compared the three action plans which were published at the time to identify areas of learning and collaboration. These results have been summarised into a set of recommendations.

The key message in the paper is that all of the action plans contain positive actions, which would benefit individuals in the other nations as well. As such, cross pollination of action plans and collaboration between the nations would assist in implementing a more

consistent and comprehensive approach to the Framework.

Tony Thornburn, Behçets UK

Tony highlighted the importance of patient registries to conduct research into rare conditions. Behçets UK encouraged Behçet's Centres of Excellence to establish a patient registry which would be operational across the three specialist hospitals in order to build a more complete picture of the prevalence of Behçet's in the UK.

In response to Baroness Neville-Jones question on what is a patient registry, Tony explained that patient registries collect patient data to use in improving patient care pathways and specifically the development of drugs and other technologies. Patient registries can also help to build the understanding of the natural history of rare conditions to underpin research.

Behçets UK is one example of a patient organisation filling the gap where there is no patient registry for a rare condition. There are several concerns with this approach.

- Funding can be unstable.
- The variation in methods of collecting and storing the data makes it difficult for researchers, health professionals and other stakeholders to use this information.

The national patient registries for rare conditions in each nation represent a solution for standardised and comprehensive capturing of rare populations. The four UK patient registries work together to ensure that they are complementary approaches to capture patients so that data can be easily accessed on a UK-wide scale. However, these registries are not sufficiently funded and resourced to provide a full service.

To remove the responsibility of providing patient registries from the patient organisations and effectively support the delivery of this service by the national registries, it is essential that enough funding is allocated to the registries.

Llion Davies from the Congenital Anomaly Register and Information Service (CARIS) in Wales joined the discussion and gave insight on the progress being made by CARIS on the expansion of the number of conditions captured in the registry.

Proposed action: it was suggested that the members of the APPG could write to Helen Whateley, Minister for Social Care, on the importance of patient registries for rare conditions and need to ensure appropriate, significant funding is set aside for an enhanced rare disease registry, and ring fenced; both resource Departmental expenditure limits but also capital resources.

Alice Fabre, SMA Newborn Screening Alliance

Alice Fabre is the Project Manager at UK SMA Newborn Screening Alliance. The Alliance's mission is to achieve the earliest possible introduction of newborn screening for SMA in the UK which will deliver the best outcomes from treatment and reduce future healthcare costs.

Every five days in the UK, a baby is born with Spinal Muscular Atrophy (SMA). SMA is a rare and devastating genetic condition. Without treatment, it is a leading genetic cause of infant death. Earlier treatment (before symptom onset) is critical for babies' outcomes and

newborn screening is the fastest route to a diagnosis of SMA. Many European countries already have newborn screening (NBS) for SMA, but the UK is not currently one of them.

SMA NBS Alliance have three main asks:

- **Timescales** – we ask the UK NSC to commit to complete its review of newborn bloodspot screening for SMA at the earliest opportunity, with clarity on the key evaluation milestones needed to reach that deadline.
- **Engagement** – we ask the UK NSC to take a more transparent and collaborative approach to engagement with the SMA expert clinical and patient community.
- **Evidence –** We ask the UK NSC to adopt a streamlined and pragmatic approach to evidence assessment for SMA, to reduce the chance of further unnecessary delays.

Proposed action: Alice asked for support to secure a Westminster Hall Debate to raise awareness of the issues surrounding the Newborn Screening Programme and put pressure on the Government to deliver changes which will modernise the approval process. Suggested title: 'UK progress on the newborn screening heel prick test'.

Professor Adrian Williams, University Hospitals Birmingham NHS Foundation Trust

Prof Adrian Williams presented on the optimum clinical pathway for adults with neurological conditions and highlighted the value of care pathways for rare conditions.

Prof Williams gave an overview of the Genomic Medicine Service, established in 2018. The overarching aim of the service is to enable the NHS to harness the power of genomic technology and scientific advances to improve population health and patient outcomes. Aim to mainstream genomic testing into routine clinical practice and ensure equity of access for patients.

The National Neuroscience Advisory Group (NNAG) Core Group developed an optimum clinical pathway which looked at a bottom up approach to the delivery of clinical care alongside genomic testing for neurology patients. The optimum clinical pathway for neurogenetics cross-cuts the vertical condition specific pathways for neurology and neurosurgery in development. The pathway outlines the overarching principles and priorities across the pathways which changes emphasis depending on the patient's needs.

Rare conditions often require multidisciplinary care meaning that care pathways must cross over departments and vertical condition specific pathways. The Rare Disease Action Plans have a focus on mainstreaming the provision of genomics, as do other health policy documents. It is important that we are also providing the care that comes with a diagnosis of a genetic condition to deliver better health outcomes.

Next step: During discussion, the value of care pathways for rare conditions were invaluable in getting access to appropriate care due to their multidisciplinary nature. It was suggested that each nation should include an action in their plans which support the development and recognition of care pathways for rare conditions. An action was not agreed and will be followed up after the meeting.