ALL PARTY PARLIAMENTARY GROUP ON RARE, GENETIC AND UNDIAGNOSED CONDITIONS

The UK Rare Diseases Framework

Joint Meeting of the APPG on Rare, Genetic and Undiagnosed Conditions and the APPG on Life Sciences
11:30-12:30, Tuesday 7 December 2021
Online

Parliamentarians in Attendance: Liz Twist MP (Co-chair)
Daniel Zeichner (Co-chair)
Christina Rees MP
Hilary Benn MP
Pauline Latham MP
Sally Ann Hart MP
Lord Rogan
George Howarth MP
Marion Fellows MP
Baroness Masham
Office of Caroline Ansell MP
Office of Chris Green MP

Speakers: Kerry Leeson Beavers, Alstrom Syndrome UK
Phillippa Farrant, Duchenne Family Support Group
Nick Meade, Genetic Alliance UK
Victoria Barrett, The Association of the British Pharmaceutical Industry
Jon Neal, Takeda

MINUTES

AGM for the APPG on Rare, Genetic and Undiagnosed Conditions

At the start of the meeting, the AGM for the APPG on Rare, Genetic and Undiagnosed Conditions was conducted. Liz Twist was reelected as Chair of the APPG. The following were put forward and elected as officers of the APPG in absentia:

- Lord Patel
- Lord Turnburg

- Baroness Neville-Jones
- Alex Sobel MP

Liz Twist MP, Chair, APPG on Rare, Genetic and Undiagnosed Conditions
Liz Twist MP welcomed attendees to the meeting. Liz introduced the topic of the UK Rare Diseases Framework and noted that the topic is relevant to both APPGs as a piece of policy which will influence the care for people with rare conditions. As such, this meeting is being held jointly so that we can hear a range of perspectives on the policy development in this area.

Liz Twist noted that, as the action plans to implement the UK Rare Diseases Framework are currently being written, this meeting is therefore particularly important as we are at a point of change where we can influence policy making to better serve people affected by rare conditions.

Phillippa Farrant, Development Officer at Duchenne Family Support Group and Kerry Leeson Beevers, Alstrom Syndrome UK
Philippa and Kerry shared some of the thoughts of the rare condition community on the development of the England Action Plan for Rare Diseases. There are ongoing concerns in the community around comments made by some of the regulatory bodies to introduce new actions or not to? change much due to the extra burden of dealing with the pandemic. With this in mind, the community are concerned that it is just a tick box exercise.

Phillippa noted the lack of content in Priority 3: better coordination of care. As representatives of the rare community in the England Rare Disease Action Plan Delivery Group, Phillipp and Kerry have guided the conversations to studies into care coordination such as CONCORD (CoOrdinated Care Of Rare Diseases) and better knowledge in the community by Medics 4 Rare Diseases. However it is unsure how much of this will be incorporated going forward.

Linked to the concerns above is the vagueness of the new commissioning structures with Integrated Care Systems. There is uncertainty around the impact on coordination of care for rare conditions.

The Department for Health and Social Care has expressed some level of enthusiasm by the delivery partners to support and listen to the patient voice. The deadline for February seems ambitious. The concerns of the patient community are being followed up with the representatives in between meetings but the community still feels that there are restrictions on what they are willing to change on the action plans.

Nick Meade, Joint Chief Executive and Director of Policy at Genetic Alliance UK

Nick Meade addressed a question raised in the last APPG meeting regarding cross border collaboration in the development of the action plans. Some key matters are reserved for the UK government but collaboration between nations is crucially important to get the delivery of the framework right. Cross Border collaboration is particularly important for rare conditions which affect a small population.

Timescales for the action plans do not appear to allow for much collaboration in the development of the plans. The UK Rare Disease Framework Board is the main forum to allow for collaboration butthis group only meets once every six months.
Delivery partners responsible for policy topics in the UK Rare Diseases Framework do communicate outside of the board meetings. However, the time pressures may limit this communication.

Nick expressed concerns that the timelines appear very tight, especially considering that a year of policy planning had been lost due to the pandemic. The rare condition community is eager for an action plan soon but not at the cost of its efficacy. Nick suggests that regular,annual, reviews of the action plans would allow for continuous policy development which could respond to the issues raised in this meeting and further issues. Wales has already adopted this approach and we hope other nations will commit to similar circumstances.

Victoria Barrett, Head of Health Technology Assessment & Market Access Policy, The Association of the British Pharmaceutical Industry
Victoria Barrett shared her perspective on priority four of the UK Rare Diseases Framework, improving access to specialists, care and treatment. Delivery of the priorities require an action oriented focus and while there are differences in delivery across the devolved nations, we must work to ensure consistency in delivery of services and not inadvertently introduce inequity.

Victoria noted that there is a lot to welcome under priority four in the draft actions of the England Action Plan. However, there are several actions under priority four which are already taking place so it would be beneficial? to see some differentiation between existing policy and those that will be implemented with this action plan.

Through the NICE Methods Review, NICE will be able to accept greater uncertainty in the evidence base. This is a key change for rare disease medicines. Additionally, it is important to note the shift in the type of evidence accepted by NICE to include examples such as real world evidence and quality of life for patients' carers as well as patients.

However, there are concerns about not reaching the level of change set out in the Life Sciences Review. The decision to not lower the discount rate and implement a severity modifier has reduced access to the Highly Specialised Technology Route in the appraisal process for rare disease medicines. This has resulted in a step backwards for rare conditions.

It is important that the England Action Plan sets metrics for the NICE methods review on access to rare disease medicines so it is clear what work needs to be done in the future.

Jon Neal, Managing Director, Takeda
Jon Neal gave a snapshot overview of the UK Rare Diseases Framework and shared some positive steps being made and some areas for improvement.

There are elements in the framework that will help to accelerate diagnostics and key to accessing treatment and care, in specific the revision of how decisions are made regarding newborn screening.

Priority two aims to put greater emphasis on rare conditions in medical education. Many healthcare professionals will rarely see these conditions, if ever, in their career but they are the gatekeepers in terms of having an awareness of rare conditions, being able to identify when a patient may have a rare condition and knowing the pathways to move the patients through the system. Jon points to the hard work of organisations such as Medics 4 Rare Diseases in this area and highlighted the ‘I am Number 17 campaign’ which aims to raise the voice of people with rare conditions

The NICE methods review is a great opportunity to improve the landscape and improve access to treatment. However, the fact that the government has made this a cost neutral review essentially neuters the ambition of that review. It is important that the ambition that has gone into developing that review isn’t withdrawn at the last moment.

Jon noted the lack of commitment around medicines regulation to opportunities that could bring regulation at the forefront and encourage licensing of rare conditions in a more aggressive way.

As a member of the Rare Diseases Stakeholder Forum, Jon has concerns that while there are metrics around each action, they lack specificity.

Discussion

In the discussion, the importance of good metrics was emphasised. Having the correct metrics to measure the meaningful success of the action plan will allow the collection of useful evidence which is lacking for rare conditions.

Deciding on the type of metric is essential for understanding the impact of the action plan. For example, the metrics could measure the action, delivery, outcomes or change in experience for people with rare conditions. By picking robust metrics, it will be easier to identify gaps in care for rare conditions.

Metrics are also useful for transparency: the rare condition community needs to see that progress is being made, if it is happening so that they know where to direct their work.

It was noted that while it is important that an accurate and timely diagnosis is a priority, the experience of diagnosis is equally important. The experience of diagnosis can be improved through the support, services and pathways by healthcare professionals and charities. There is considerable diversity in experience for care in rare conditions which needs to be addressed.