ALL PARTY PARLIAMENTARY GROUP ON RARE, GENETIC AND UNDIAGNOSED CONDITIONS

Update on the rare disease landscape

9:00 – 9:30, 20 October 2021 via Zoom

Parliamentarians in attendance:

Sir George Howarth MP
Christina Rees MP
Marion Fellows MP
Office member of Marion Fellows MP
Office Member of Sharon Hodgson MP
Office Member of Caroline Ansell MP

MINUTES

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

Liz Twist welcomed attendees to the meeting and introduced the session – giving an update to MPs on the two recent APPG meetings on access to medicines and newborn screening for rare conditions. Liz Twist pointed to the meeting on 7 December where these issues can be further discussed around the UK Rare Diseases Framework.

Liz Twist remembered Sir David Amess and recognised his involvement in the wider rare disease community and his attendance to Rare Disease Day events.

Following the final public consultation on the NICE methods and processes review, the process has resulted in less change than hoped for. Some proposals demonstrate steps forward for rare conditions. However, there are fears around inconsistent application of new proposals and conflicting criteria for the Highly Specialised Technology routing.

Currently, in the UK , the heel blood prick test for newborns screens for between five and nine conditions, depending on where you are in England, Scotland, Wales or Northern Ireland. However, no new conditions have been added to the heel prick blood test since 2017 in Scotland, 2015 in England and Wales and 2009 in Northern Ireland.

The UK Rare Diseases Framework provides the vision for rare disease policy for the next five years. The action plans to be released at the start of 2022 are currently in the process of being written and will

shape future policy. The next APPG meeting on 7 December will cover the UK Rare Diseases Framework in more detail.

Nick Meade, Joint Interim Chief Executive at Genetic Alliance UK

The NICE methods review public consultation has finished and we are now in a period waiting for the announcement of findings by NICE from the consultation.

All proposed changes are opportunity cost neutral so there is no intention of changing the resources put towards new medicines which puts a limit to the potential impact of the changes, most notably on the Highly Specialised Technologies pathway for rare condition medicines which would be for the most innovative and transformative medicines.

The cost neutrality of the changes undermines the proposed change to replace the end-of-life modifier (only used for oncology treatments) with a severity modifier, as the benefits would have to be shared across wider groups. It also impacts the discount rate which is the tool used to measure long term benefit into the future for the individual receiving a medicine, which is not being taken forward because of the decision to be cost neutral. The ultimate conclusion is that NICE are making some changes but the magnitude of these is too small to make a significant difference to people living with rare conditions.

NICE's decision to postpone their progress on health inequalities is frustrating for the community and will hopefully be a priority once this process has been completed.

The Innovative Medicines Fund has been delayed and highlights the point that NICE processes are not well linked with this new fund.

There is some good news as medicines are being approved for people with rare conditions but these changes are happening due to non-transparent processes at NHS England rather than because of the NICE process. This is therefore a sign that the system is not fixed and a concern is that medicines will become stuck in the NICE process as NICE delays making a decision.

Jess Hobart, The UK Mastocytosis Support Group (Masto UK)

Since the APPG hearing on access to medicines in April, the treatment for mastocytosis has received a positive recommendation from NICE. This followed an initial negative appraisal consultation document which set out the appraisal committee's preliminary recommendations to NICE on the basis of uncertainty. Their understanding is that the board made the positive recommendation to be nice and generous. However, the system does not support this decision-making.

Masto UK are cautiously hopeful about these changes in the NICE methods review but cost neutrality means that there is a fight for resources. They are pleased that there is increased openness to real world evidence. While there is also increased flexibility around uncertainty, the details are yet to be confirmed. Additionally, the severity modifier replacing the end of life modifier does apply some flexibility but is again restricted by resources.

There is a big space where many rare diseases exist between the standard technology appraisal pathway and the highly specialised technology pathway for very-rare conditions. This review has not been the big change that the community needed to see for rare diseases to have a chance in the NICE system. Mastocytosis has two new drugs that will be entering the NICE process soon and it is unclear whether they will receive a positive recommendation due to the uncertainty around rare conditions.

To improve their quality of life, people with mastocytosis depend on a mix of generic medicines to block symptoms. Supply of these treatments are dependent on wider demand which often results in supply issues when these drugs are no longer fashionable. DHSC tries to identify importale alternatives which are often too expensive. Masto UK are currently supporting a petition addressing price gouging where prices are forced up for no reason. They are also advocating for legislation to prevent market failure and ensure supply for these drugs in the same way they do new drugs. An NHS owned manufacturer of generic medicine could resolve this issue.

Nick Meade, Joint Interim Chief Executive at Genetic Alliance UK

There has been inertia in the community to take forward clear messaging and campaigning on newborn screening.

To improve the application process for newborn screening, a speciality team for rare conditions is needed as well as a better process for review of ongoing applications, as we heard in the previous APPG meeting. In the review, data from places which already test for these conditions need to be taken into account.

Both the Spinal Muscular Atrophy (SMA) community and the Adrenoleukodystrophy (ALD) community have launched petitions to add their respective conditions to the newborn screening programme. They have both received a response from the Government. These responses are concerning as the Government's strongly defends the current approach which it describes as 'cautious and rigorous'. This ever more divergent approach means 16 EU countries now screen more conditions than the UK.

Conversely, government funding has been allocated for a new pilot project on whole genome sequencing for newborn screening. These two policy positions are becoming increasingly divergent and leave us less well placed to succeed in both of these areas.

Karen Harrison, Alex - The Leukodystrophy Charity (Alex TLC)

Alex TLC received a response to their petition to add ALD to the newborn screening programme. It was disappointing to see the generic nature of the response to their petition and the similarity to that sent to the SMA community. The newborn screening committee agreed to a meeting with Prof Anne Mackie, Director of Screening for Public Health England, John Marshall, Evidence Lead for the NHS Screening Programmes, and Alex TLC. It was an open meeting and a good conversation.
Work needs to be done on the perceptions of early diagnosis, particularly for those conditions which are untreatable. Newborn screening is especially important to allow for reproductive options to prevent passing on the genetic condition to future generations.

The newborn screening committee put emphasis on limiting the harm caused without considering the significant good that can come from a diagnosis. A balance between the two needs to be addressed.

Newborn screening for ALD will bring significant good because, of the four phenotypes, the phenotypes that present in childhood are treatable if discovered prior to the onset of symptoms.

ALD is screened for in the US and the Netherlands so Alex TLC encourages the UK National Screening Committee (UKNSC) to take this evidence into consideration. The UK NSC acknowledge that they fail to recognise the fourth phenotype of ALD which is worrying for Alex TLC as 80% of males with this phenotype develop addison's diseases - a serious life threatening condition if left untreated but is easily treatable with replacement steroids. This information was clearly presented in the application but was not considered because the committee did not understand the context of the published paper. This information will now be considered in 2023.

Karen highlights the dependence on patient organisations to provide evidence which are beyond their remit and resources.

It was stated several times that the newborn screening committee is a small team with limited resources. This response is worrying as it puts into question the appropriateness of this decision-making body responsible for delivering vital recommendations for safeguarding the nation's health. There is now a campaign calling for a separate body for newborn screening from the wider screening committee.

Nick Meade, Joint Interim Chief Executive at Genetic Alliance UK

The UK Rare Diseases Framework is a new piece of policy published in January, on which Liz Twist hosted a Westminster Hall Debate in March. This framework creates the space for UK wide policy on rare conditions. The four priorities of the framework cover a wide variety of topics, including those covered today. However, there are still considerable gaps in areas of mental health, care pathways and transition between pediatric and adult care. These areas will be part of the discussion at the next meeting of the APPG which will be hosted in conjunction with the APPG for life sciences.

Discussion

Sir George Howarth MP looked forward to discussing these issues further in the next APPG meeting.

Christina Rees MP asked whether sepsis was represented in a registry. Sepsis fits under less specialised services so will be served by the NHS in the individual nations as it is not as rare. There are plans to rollout more registries across the UK to collect data. However, currently there is no registry for sepsis. While sepsis does not fit under this registry, Congenital Anomaly Register and Information Service (CARIS) is expanding its representation of rare conditions.

Liz Twist would like to address the differences across the nations in relation to the UK Rare Diseases Framework in the next APPG meeting.