ALL PARTY PARLIAMENTARY GROUP ON RARE, GENETIC AND UNDIAGNOSED CONDITIONS

RARE DISEASE UK

Hearing on the implementation of the UK Strategy for Rare Diseases

Date: **7 November 2016**

Venue: **Room M, Portcullis House**

Attendees

Ben Howlett MP, Chair	Jerome Ma, Department of Health
Farhana Ali, Genetic Alliance UK	Ian McKay, Department of Health
Rosie Collington, Genetic Alliance UK	Nick Meade, Genetic Alliance UK
Neil Dugdale, Sobi	Colin Pavelin, Department of Health
Dorothy Francis, Department of Health	Rupert Purchase, Wilson's Disease Support Group UK
Chris Goard, Genetic Alliance UK	Jem Rashbass, Public Health England
Kathryn Irons, NIHR	Beverly Searle, Unique
Paul Jeff, Department of Health	Dr Tony Soteriou, NIHR
Jo Johnson, NIHR	Jayne Spink, Tuberous Sclerosis Association
Alastair Kent OBE, Genetic Alliance UK	Verity Thomas, Shire

Minutes

1. **Welcome and introductions from Ben Howlett MP**

2. **Presentation from Dr Tony Soteriou, the National Institute for Health Research (NIHR)**

NIHR research infrastructure for rare diseases consists of biomedical centres, units, collaborations and other infrastructure within the NHS that helps support research into rare diseases. The NIHR's Rare Diseases Translational Research Collaboration (RD-TRC) is a new initiative to bring a number of its investments together. NIHR also funds BioResource, a cohort of people – patients, their families and healthy volunteers – that might be called for experimental studies on rare disease medicines. It has also funded research projects and programmes for very rare diseases. NIHR has developed new processes to speed up the approval process for new rare disease research, as well as ethics approvals and NHS permissions. NIHR encourages patient involvement in all areas of its work through platforms such as INVOLVE. Dr Soteriou was optimistic that digitalisation of NIHR research would improve its accessibility to patients.

3. **Presentation from Kathryn Irons, NIHR Rare Diseases – Translational Research Collaboration (RD-TRC)**

RD-TRC sees public involvement as a means to achieve better research, with a greater focus on the needs of patients, and fast translation of research findings into real-life patient benefits. It has recruited two lay members to its Strategic Oversight Group, as well as fourteen lay members to be part of its Patient Advisory Group. The RD-TRC strives to involve patients and carers in showcasing its work. To date, it has

hosted two research symposia, held in September 2015 and 2016. It funds around 50 rare disease studies across England, and requires applicants to provide details of patient and public involvement during both the application process and documentation. The RD-TRC endeavours to involve patients and carers in data collection. It aims to work with clinicians and patients to assemble quality clinical data sets across the rare disease community as a platform for translational research. This year, the RD-TRC has been developing two experimental apps to enable patients to input data directly. To support and develop the next generation of rare disease translational researchers, the RD-TRC has nearly 17 Clinical Trainees and Fellows (at PhD or postdoctoral level). It uses large-scale events, such as Rare Disease Day, to promote the research it is supporting and to encourage greater involvement.

4. Presentation from Jem Rashbass, the National Congenital Anomalies and Rare Disease Registration Service (NCARDRS)

NCARDRS is a population-level register; this will make it possible to identify incidence of all rare diseases in England. NCARDRS has worked with patient groups to address issues concerning data collection, and has developed strong information governance. NCARDRS aims to collect existing data from primary sources within the NHS into its systems. To ensure this does not put a time and resource strain on the NHS, NCARDRS has a team of Data Liaison Officers, who specialise in extracting data and will accept data in any form from the NHS.

5. Presentation from Colin Pavelin, the Department of Health

Colin Pavelin said that the Department of Health is not responsible for implementing the Strategy, but facilitating arm's length bodies to implement it. The Department of Health has nonetheless worked to establish European Reference Networks across the EU. The Department has also been involved in developing the 100,000 Genomes Project, led by Genomics England. The Department has commissioned a report to identify the issues in diagnosing rare diseases. The Department is working with analysts to develop measures and a framework to monitor progress across the 51 commitments, which will be published in the next biennial report on the Strategy's implementation in 2018. Not all recommendations will be implemented by 2020, as many of them will be outdated due to developments in genomics technology.

6. Discussion

- The Department of Health does not feel that England is at a disadvantage compared to the devolved nations because it does not have an implementation plan.
- NCARDRS is not able to collect data on patients who do not have a diagnosis listed on Orphanet, which patient representatives felt disadvantages rare disease patients with multi-system disorders and patients with very rare conditions.
- The Department of Health recognised the importance of coordinated care pathways for patients. It stated that the genomic medicine centres developed through the 100,000 Genomes Project could contribute to coordinated care pathways due to their potential for embedding precision medicine.
- Patient representatives noted that the charity sector often coordinates rare disease patients' care where there are gaps in provision by the state. Problems with coordination of patients' care are the result of poor communication between the clinical and social care systems.
- Patient representatives believe an implementation plan would act as a point of reference for patients who want to understand what is happening with the implementation of the Strategy in England, and when.
- Colin Pavelin said that the Department of Health had decided to reconfigure the UK Rare Disease Forum because it did not feel it was an appropriate platform to develop policy.