ALL PARTY PARLIAMENTARY GROUP ON RARE, GENETIC AND UNDIAGNOSED CONDITIONS

Virtual reception - Rare Disease Day 2022

13.00-15.00, Wednesday 23 February 2022
Online

Speakers: Liz Twist MP, Chair of the APPG on Rare, Genetic and Undiagnosed Conditions
Nick Meade, Joint Chief Executive & Director of Policy, Genetic Alliance UK
Amisha Bagri, Parent of a child with a rare condition
Maria Caulfield, Minister for Patient Safety and Primary Care
Dexter Parker, Person living with a rare condition

Liz Twist MP hosted the Westminster reception on Wednesday 23 February. In her introductory remarks, she noted that though undoubtedly progress is being made, both in awareness of rare conditions and in terms of diagnosis, treatment and care, there remains an enormous amount of work to be done.

Liz Twist MP introduced diagnosis as the theme for the Rare Disease Day 2022 reception. She noted that it is encouraging that the UK Rare Disease Framework has prioritised helping people with a rare condition to get a diagnosis faster. However, progress must be measured by its impact on people with rare conditions.

Amisha Bagri spoke about her experience with her son who was diagnosed with dihydropteridine reductase deficiency, a form of Phenylketonuria (PKU), through the newborn screening programme. Amisha spoke on the impact of receiving a misdiagnosis and highlighted the importance of effective communication during the diagnostic journey; the healthcare professionals wanted to provide information and next steps before she did her own research which, in foresight, was a very good thing.

Amisha highlighted the value of early intervention and fast diagnosis as receiving early treatment has significantly limited the impact on his physical and cognitive abilities. A fast diagnosis and care from specialists at their hospital has allowed the family to adjust to living with a rare condition.

Maria Caulfied, Minister for Patient Safety and Primary Care addressed the rare condition community ahead of launching the England Rare Disease Action Plan on Rare Disease Day. She shared work happening in each of the priorities outlined by the UK Rare Diseases Framework. Maria Caulfield highlighted the importance of the voice of people with rare

conditions in the development of the UK Rare Disease Framework and the England Action Plan and looked forward to hearing feedback which can contribute to future iterations.

Despite the huge pressures of the pandemic, the work around rare diseases is happening at pace. The minister stated that the England Action Plan will have concrete actions and clearly owned outcomes and will be measurable and time-bound.

Dexter Parker shared his experience of being a student with Neurofibromatosis Type 1 via a prerecorded video. Dexter talked about the support he received from his family, the local hospital and from Nerve Tumours UK. Dexter stated that he was lucky to live close to the specialist centre and takes a positive approach to hospital visits.

He explained that his condition has not limited him in his aspirations to become a filmmaker due to the support he received at school and now at university. However, he recognises the challenges that he faces in his education due to the symptoms caused by NF1. He also recognises the impact NF1 has had on his personal life such as with making friends.

Nick Meade presented the Genetic Alliance UK Good Diagnosis report. Genetic Alliance UK recognises the value of diagnosis to someone with a rare condition. The project set out to better understand people's experience of diagnosis and to identify what matters most to people on their diagnosis journey through a series of workshops in November 2021. Key themes from the discussions, together with the findings of our Rare Resources (Scotland) and Rare Experience 2020 projects, were used to identify principles of a good diagnosis.

Good Diagnosis is:
Accurate and Timely
Informed and Supported
Collaborative and Coordinated
Acknowledged and Respected

The report uses these principles to make four recommendations to improve the experience of people at the three key stages of the diagnosis journey: the search for a diagnosis, receiving a diagnosis, and following diagnosis.