ALL PARTY PARLIAMENTARY GROUP ON RARE, GENETIC AND UNDIAGNOSED CONDITIONS

Rare Disease Day reception and Annual General Meeting: minutes

Date: **26 February 2020**

Venue: **Terrace Pavilion, Houses of Parliament**

Parliamentarians

Nickie Aiken MP
Sir David Amess MP
Elliott Colburn MP
David Duguid MP
Julie Elliott MP
Katherine Fletcher MP
Sir Roger Gale MP
Kizzy Gardiner (Locum MP for Stella Creasy MP)
Sharon Hodgson MP
Sir George Howarth MP
Sir Greg Knight MP
Dame Eleanor Laing MP
Pauline Latham OBE MP
Kerry McCarthy MP
Karl McCartney MP
Baroness Pauline Neville-Jones
Gavin Newlands MP
Ian Paisley MP
Lord Naren Patel
Jim Shannon MP
Virendra Sharma MP
Cat Smith MP
Liz Twist MP
Catherine West MP
Matt Western MP
Munira Wilson MP

Guest Speakers

Dr Jayne Spink, Genetic Alliance UK
Dr Jenny Harries, Deputy Chief Medical Officer for England
Amanda Brodie
Kristy Blakeborough-Wesson

1. **Welcome from Liz Twist MP**

Annual General Meeting – election of the officers.
Election of chair: Liz Twist MP.
The election of the chair was seconded by Baroness Pauline Neville-Jones, Pauline Latham OBE MP, David Duguid MP.
Election of vice chairs: Catherine West MP, Baroness Pauline Neville-Jones, Lord Naren Patel, Lord Turnberg, Rt Hon Dame Cheryl Gillan MP and Pauline Latham OBE MP.
Election of officers: Alex Sobel MP and Matt Western MP.

Liz Twist MP opened up the Rare Disease Day reception by welcoming attendees and highlighted common challenges affecting rare disease patients. She noted that 2020 was particularly significant as Rare Disease Day would be taking place on 29 February, a rare day.

2. **Dr Jayne Spink, Chief Executive, Genetic Alliance UK, and Chair of Rare Disease UK**

Dr Jayne Spink noted that 2020 is the end date for the UK Strategy for Rare Diseases and talked about how the prospects for diagnosis, care and treatment have been transformed by significant advances in technology, science and medicine. Dr Jayne Spink reflected on the work of Genetic Alliance UK over the last year including work relating to newborn screening and access to medicines.

As we look towards the future, Dr Jayne Spink acknowledged the uncertainties of international research collaborations but also talked about the reasons to be hopeful for the future such as the role out of a Genomic Medicine Service.

Dr Jayne Spink also launched Rare Disease UK's report on the use of medical alert cards for rare disease patients, called 'Rare Alert', which demonstrates how medical alert cards can be highly effective and offer benefits to both patients and healthcare professionals. The report also sets out three reasons as to why it remains vital that NHS England deliver on the 2018 pledge to give every rare disease patient an alert card.

3. **Dr Jenny Harries OBE, Deputy Chief Medical Officer for England**

Dr Jenny Harries informed attendees of the publication of the second update to the UK Strategy for Rare Diseases from the Department of Health and Social Care. The update outlines the progress towards the delivery of the UK Strategy for Rare Diseases. She also announced that the publication of the Genomic Healthcare Strategy is upcoming in the following months.

Dr Jenny Harries also announced that the results from the National Conversation on Rare Diseases survey will be used to develop an ambitious post 2020 framework for rare diseases that will be followed by nation-specific implementation plans that lay out commitments in order to achieve the aims of the overarching framework.

Dr Jenny Harries acknowledged the importance of care coordination for rare disease patients and mentioned the use of whole exome sequencing currently being used for critically ill children in clinics.

Finally, Dr Jenny Harries talked about improvements to access to medicines under the new Innovative Medicines Fund that will allow doctors to use the most advanced, life-saving treatments for conditions including rare diseases.

4. **Amanda Brodie, patient speaker**

   Amanda shared her experience of living with Cushing's syndrome, a rare condition of the endocrine system that affects approximately two in a million people. Cushing's syndrome is characterised by high levels of cortisol in the blood and is usually caused by a benign tumour on the pituitary gland in the brain. Amanda told attendees about an emergency situation where a medical alert card could have helped her to get the care and treatment she urgently needed.

5. **Kristy Blakeborough-Wesson, patient speaker**

   Kristy informed attendees about how having a rare condition can sometimes be a lonely experience as many people don't know about her condition, HTLV-1, including healthcare professionals. She told attendees that carrying an alert card, holding specific information relating to her condition and potential complications (that is recognised by healthcare professionals), would be invaluable to enable correct care during times where she may not be able to communicate the complexities of her condition.

6. **Screening of a shortlisted film from the Rare Film Festival 2020 - '#FightingFailure' submitted by Sam Clarke and Alport UK**

7. **Close**