

# ALL PARTY PARLIAMENTARY GROUP ON RARE, GENETIC AND UNDIAGNOSED CONDITIONS



## Hearing on the implementation of the UK Strategy for Rare Diseases

Date: **1 November 2016**

Venue: **Room M, Portcullis House**

### Attendees

*Ben Howlett MP (Chair)*

*Mark Durkan MP*

*Nick Meade, Genetic Alliance UK (speaker)*

Farhana Ali, Genetic Alliance UK

Rosie Collington, Genetic Alliance UK

Lesley Harrison, Alkaptonuria Society

Lynn Laidlaw, Vasculitis UK

Eric Lange, National Society for Phenylketonuria

Kerry Leeson-Beevers, Alstrom UK

Brian Lovatt, patient

Jerome Ma, Department of Health

Bill Owen, Niemann-Pick UK

Rupert Purchase, Wilson's Disease Support Group

Beverly Searle, Unique

Jayne Spink, Tuberous Sclerosis Association

Jane Swainson, Rare Disease UK

Tania Tirraoro, Special Needs Jungle

Lorraine Yeomans, Macrocephaly-Capillary

Malformation UK

### Minutes

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

#### 2. Presentation from Nick Meade, Chair of the Patient Empowerment Group

Nick Meade discussed the history and aims of the UK Strategy for Rare Diseases, and the role of the Patient Empowerment Group (PEG) in providing a patient voice on the Strategy. He explained PEG's concerns with the Department of Health's reconfiguration of the UK Rare Disease Forum earlier this year.

#### 3. Discussion: implementation of the Strategy in England

Attendees felt that:

- There is a lack of coordination between health and social care services. Much of the improvement in patients care is a result of the efforts of clinicians and families who have taken responsibility for coordinating care.
- Families would be isolated if it weren't for rare disease support groups.
- Even where specialist centres exist, they often aren't utilised and patients are not referred.
- In regard to rare diseases, the Department of Health is under-resourced and under-staffed.
- England is among the least progressed European countries in terms of its approach to rare diseases. The absence of an implementation plan is embarrassing internationally.

#### 4. Discussion: implementing the Strategy in the devolved nations

Attendees noted that:

- It is senseless that Scotland, Northern Ireland and Wales have a plan but England doesn't.
- An implementation plan for England that identified the bodies in charge of taking forward each commitment would provide a good point of contact for the devolved nations.

- The devolved nations are also disadvantaged in that there are not enough patients to establish specialist centres and that these patients often struggle to access specialist centres based in England.

## **5. Discussion: the impact of Brexit on implementing the Strategy**

Attendees felt that:

- Leaving the EU is likely to impact research collaboration.
- The attractiveness of the UK as a place to do research and launch treatments has diminished.
- Departure from the European Medicines Agency (EMA) authorising rare disease medicines will be disastrous for UK rare disease patients.

## **6. Discussion: research, development and fundraising**

Attendees noted that:

- Pharmacological research is important for patients, but so is service research and provision.
- Many patient organisations directly fund both research into treatments and health and social care services for patients and families.
- Rare disease patients, families and carers would benefit from a designated care coordinator.
- Many patient groups fund and employ specialist staff, such as nurses. While attendees felt it was necessary, they did not think it was right that patient organisations had to fill the gaps resulting from the NHS's lack of service provision.

## **7. Discussion: mental health and rare disease**

It transpired that many patient organisations fund counselling services and employ mental health professionals. Attendees felt that:

- Patients face a number of mental health problems resulting from both neurological changes that occur with their condition, and the psychological impact of living with a rare disease.
- Many patient groups face similar psychological issues. There could be the potential to develop a specialist rare disease psychology service.
- Rare diseases not only affect the mental health of patients, but also their families.
- Cognitive and sensory impairments can be a further barrier to accessing mental health treatment.
- The lack of coordination of care experienced by so many patients affects their mental health.

## **8. Next steps**

Attendees broadly agreed on the following points:

- The Department of Health needs to take responsibility for implementing the Strategy.
- The Department of Health must develop an implementation plan and provide adequate resources to allow for effective coordination of such a plan.
- The development of the implementation plan would result in financial savings across healthcare services.
- There is poor communication between the Department of Health and patient representatives.
- The existing implementation plans in the devolved nations give the departments of health something to work towards, and the public a way of holding the respective governments to account.

Farhana Ali, Public Affairs Manager at Genetic Alliance UK, provided an overview of the next hearings and the inquiry. All rare disease patients, families, carers, patient representatives, clinicians, academics and industry representatives are invited to [submit evidence online](#). The inquiry will form a comprehensive review of implementation in England to date.

## **9. Close**