IMPLEMENTATION OF THE UK STRATEGY FOR RARE DISEASES IN ENGLAND

Inquiry by the All Party Parliamentary Group for Rare, Genetic and Undiagnosed Conditions

The four countries of the UK have until 2020 to implement the 51 commitments in the UK Strategy for Rare Diseases (the Strategy), published in November 2013. In order to help put the Strategy into action, each country agreed to develop its own implementation plan by February 2014. The departments of health in Scotland, Wales and Northern Ireland have all published country specific plans that reflect their respective health service structures and priorities. The Department of Health in England has not developed or coordinated a plan for England.

Scope of this inquiry

At the inaugural meeting of the All Party Parliamentary Group (APPG) on Rare, Genetic and Undiagnosed Conditions members agreed to hold an APPG inquiry into the implementation of the UK Strategy for Rare Diseases in England. The APPG will take written evidence and conduct three hearings to collect oral evidence from patient representatives and all other stakeholders including the Department of Health and its arm's length bodies. The aim is to answer the key questions:

Why do we not have an implementation plan for the UK Strategy for Rare Diseases in England?

How does the absence of an implementation plan affect patients in England?

The APPG will also consider how this affects the rest of the UK. Rare Disease UK is working closely with the Chair, Ben Howlett MP, to gather information on behalf of the APPG on Rare, Genetic and Undiagnosed Conditions.

The UK Strategy for Rare Diseases

The Strategy represents a landmark for patients with rare diseases. It is the first time all four health departments of the UK have come together to respond to the needs of all those affected by rare diseases.

The aim of the UK Strategy for Rare Diseases is to 'ensure no one gets left behind just because they have a rare disease'.

The Strategy contains 51 commitments across five broad themes, to ensure that health and social care systems across the four nations provide those living with rare conditions with the highest

possible quality of evidence-based care and treatment, regardless of where they live in the UK. The five themes are:

1. Empowering those affected by rare diseases

The Strategy recognises that patients and families are often experts in their rare condition. It emphasises the need for patients to have access to reliable and correct information about their condition to make informed choices. It encourages health providers to ensure that patients and their families have a say in decisions about treatment and in the planning, evaluation and monitoring of services.

2. Identifying and preventing rare diseases

Developing better methods of identifying and preventing rare diseases will help to improve the lives of families affected by rare conditions. Screening (a way of finding out if people are at higher risk of a health problem) and carrier testing (a genetic test used to determine if a person is a carrier for specific disease) are both recognised as key tools in identifying and preventing rare diseases.

3. Diagnosis and early intervention

Rare diseases are by their nature difficult to diagnose. Many patients will receive at least one incorrect diagnosis and experience significant delays in their journey to secure an accurate diagnosis.

The Strategy calls on the four nations of the UK to ensure that every rare disease patient has a clear personal care plan that brings together health and care services. It notes that clinicians must recognise that many patients with access to reliable information can help them with decisions about referral and diagnosis, and that it is vital for patients with undiagnosed to have access to appropriate coordinated care.

4. Coordination of care

Rare diseases are often life-long and serious, affecting multiple systems of the body. Many of them are progressive resulting in the health and quality of life for affected individuals to deteriorate throughout their lives.

Rare disease patients are likely to have numerous professionals involved in their care, and as such it is essential that there is coordination and communication between them all. The Strategy states that care should be coordinated across health and social care services, so that care is effective, accessible and convenient to patients e.g. should not disrupt their work or education.

Problems can also occur if possible interactions between different treatments are not properly managed.

5. The role of research

Research is fundamental to improving the understanding of rare conditions. It can result in a variety of benefits, including: increasing awareness of a disease, developing a treatment, improving diagnosis opportunities, uncovering information that will increase medical knowledge, and even help improve the health of the general population.

For many patients and families research is their only hope for a more effective treatment or cure for their condition. Many are keen to participate in clinical trials that may eventually offer a cure or improve diagnosis.

Questions to help with your response

- In your opinion, do you think the UK Strategy for Rare Diseases has been effectively implemented in England?

  ☐ Yes

  ☐ No

  Please explain your answer:

- At the time of diagnosis were you provided with information on how to find and contact support groups, what treatment options (if any) were available, and whether there was relevant research being undertaken?

- Do you feel that you are listened to and consulted on to agree the best way forward for your (your child's) care and treatment?

- Do you have a clear personal care plan that brings together health and care services?

- Is the care that you receive (your child receives) coordinated?

To help us to better understand your response, please tell us if you are:

☐ A patient

☐ A family member or carer of someone affected by a rare disease

If you, or your affected family member, have received a diagnosis please state the year and the diagnosis:

Year _____ Diagnosis _____

The responses you provide will be crucial to helping us get a full, up-to-date picture of the implementation of the UK Strategy for Rare Diseases in England. All the information you provide will be treated as confidential. If, however, you have specific confidentiality concerns please mark this clearly in your submission. You do not have to provide us with any information that may identify you. The findings will be published in time for Rare Disease Day 2017 and they will be widely disseminated locally and nationally. By participating in this call for evidence you are agreeing that the information you provide can be shared anonymously with members of the All Party Parliamentary Group for Rare, Genetic and Undiagnosed Conditions and can be used for policy work by Rare Disease UK and Genetic Alliance UK

Proposed timeline	
25 October 2016	Launch call for evidence
1 November 2016	APPG hearing for patients and patient representatives
7 November 2016	APPG hearing
15 November2016	APPG hearing
2 January 2017	Deadline for call for evidence
24 January 2017	APPG workshop to develop recommendations/ APPG AGM
1 March 2017	Publication of findings and recommendations

The deadline for evidence submission is 2 January. Submissions should be made to appg@geneticalliance.org.uk with the subject line 'call for evidence'.

If you require further assistance please contact Rosie Collington, Public Affairs Assistant, at Genetic Alliance UK: rosie.collington@geneticalliance.org.uk