HOUSE OF COMMONS LIBRARY

DEBATE PACK

Number CDP 2017/0105, 24 March 2017

Implementing the UK Strategy for Rare Diseases

Sarah Barber
Jacqueline Baker

A debate is scheduled on Implementing the UK Strategy for Rare Diseases for Tuesday 28 March 2017 4.30-5.30pm. The Member in charge of the debate is Ben Howlett MP.

Contents

1. Summary

Implementation of the UK Strategy for Rare Diseases

A rare disease is generally considered as one affecting fewer than 5 people in 10,000. However, large numbers of people are affected by rare conditions. The *UK Strategy for Rare Diseases* estimated that in the UK alone, more than 3 million people will suffer from a rare disease at some point in their life.[1]

A significant proportion (around 80%) of rare diseases have a known genetic cause but other causes include infections, cancer and deficiencies or exposure to substances prior to birth. Due to the fact that relatively small numbers of people are affected by a particular rare disease, there are specific challenges in ensuring speedy diagnosis and access to appropriate services and treatments.

The UK Strategy for Rare Diseases was published in 2013. It aimed to raise awareness of rare diseases, and improve diagnosis, research and access to services for those affected.

This debate pack has been prepared for a Westminster Hall debate on Implementation of the UK Strategy for Rare Diseases. The debate has been tabled by Ben Howlett, Chair of the All Party Parliamentary Group (APPG) on Rare, Genetic and Undiagnosed Conditions.

1.1 The UK Strategy for Rare Diseases

In November 2013 the UK Department of Health and the devolved Governments published the UK Strategy for Rare Diseases.[2] This was the UK's first strategy to help build understanding of rare diseases and boost research to find effective treatments and therapies. It set out 51 commitments to be delivered by 2020, addressing 5 key areas:

- empowering those affected by rare diseases;
- developing better methods of identifying and preventing rare diseases;
- improving diagnosis and earlier intervention for those with a rare disease;
- developing better coordination of care for those with a rare disease, including joined up consultation and treatment schedules; and,

- building on research to improve personalised approaches to healthcare for those with a rare disease.

The strategy also highlighted the importance of collaborative working with patients, their families, industry, research and international partners.

Monitoring the progress of the strategy

The UK Rare Disease Policy Board was established in October 2016, and is responsible for the coordination of action to meet the commitments within the UK Strategy for Rare Diseases. The Board is made up of representatives of the UK departments of health, and rare disease charities such as Genetic Alliance.[3]

A larger group, the UK Rare Disease Policy Forum has membership from wider stakeholder groups. This group is responsible for providing insight and advice on the strategy and ensuring that those with a range of rare conditions are represented.

The first progress report of the Rare Disease Strategy, *Delivering for patients with rare diseases: Implementing a strategy*, was published by the UK Rare Disease Policy Forum in February 2016.[4] The report highlighted that there had been significant progress made since the publication of the strategy. Notable advances included:

- The establishment of the National Congenital Anomaly and Rare Disease Registration Service by Public Health England. This service records individuals with rare diseases across the UK and provides a valuable resource for clinicians and researchers.
- Improved research in rare diseases, for example the NIHR Rare Diseases Translational Research Collaboration (RD-TRC) which focuses on the causes, impacts and treatment of rare diseases.
- Improved approaches to diagnosis, for example greater access to genetic testing. The report highlighted the 100,000 Genomes Project and the establishment of regional genomics medicine centres.

Rare Disease UK (a national campaign for individuals with rare diseases) reported that there had been significant progress since the launch of the strategy, but called for improved collaboration across the UK:

> The report from the UK Rare Disease Forum recognises that there has been significant progress towards delivering the commitments in the UK Strategy. For example, the 100,000 Genomes Project will transform molecular diagnostics across the UK and the creation of a unified registry of rare disease patients by Public Health England will have a significant impact on assessing, planning and research for rare diseases.
>
> However, as 'rare diseases do not recognise national boundaries' the UK Rare Disease Forum is calling for the four nations of the UK to work together to meet the needs of patients. Collaboration is essential if the National Health Services in the UK are to deliver

[3] Department of Health, UK Rare Disease Policy Board and Forum
[4] UK Rare Disease Forum, Delivering for patients with rare diseases: Implementing a strategy, February 2016

> the specialist and coordinated care required for patients affected by these multisystem conditions.
>
> The Chair of Rare Disease UK, Alastair Kent OBE said: "The publication of the first report of the UK Rare Disease Forum is a key stage in the implementation of the Strategy published by the UK Health Ministers in 2013. There has been solid progress in many areas, notably in research and the setting up of the National Congenital Anomaly and Rare Disease Registration Service. The needs of patients and families with rare diseases are now acknowledged in ways that would have been unimaginable only a few years ago, but much remains to be done to meet the ambitious commitment to improve care and support for rare disease patients in the original strategy by 2020. I am confident that this goal is an achievable one, given the commitment of the Government and the NHS to delivering on the promise made to the 3.5 million people living with rare diseases in the UK today."
>
> Life Sciences Minister, George Freeman MP said: "This report highlights the huge benefit that initiatives like our 100,000 Genomes Project and our £1bn per year National Institute for Health Research bring to people living with rare diseases and their families."[5]

Implementation plans

All 4 UK countries committed to producing implementation plans to provide more information about how the strategy will be put into action.

These implementation plans have been published in Scotland[6], Wales[7] and Northern Ireland.[8] Links to these implementation plans are included below:

- Scottish Government, It's not rare to have a rare disease:The Implementation Plan for Rare Diseases in Scotland
- Welsh Government, Rare diseases implementation plan
- Northern Ireland Government, The Northern Ireland Implementation Plan for Rare Diseases

In England, NHS England is responsible for implementing the rare disease strategy. It published a Statement of Intent in February 2014 that provides information on how its commitments will be achieved and working with partners to address the other commitments within the strategy[9].

A February 2017 report from the APPG for Rare, Genetic and Undiagnosed conditions, *Leaving no one behind: Why England needs an implementation plan for the UK Strategy for Rare Diseases,* has called

---

5 Rare Disease UK, Delivering for patients with rare diseases: Implementing a Strategy, February 2016
6 Scottish Government, *It's not rare to have a rare disease: The Implementation plan for rare diseases in Scotland* ,June 2014
7 Welsh Government, *Welsh Implementation plan for rare diseases*, February 2015
8 Department of Health, Health Minister announces new innovative era of treatment for rare diseases, October 2015
9 UK Strategy for rare disease, *NHS England statement of intent*, February 2014

on the UK Department of Health to publish an implementation plan.[10] The report states that NHS England is not in a position to act on a number of the commitments within the strategy and sets out how an implementation plan would improve the implementation of the strategy for patients with rare diseases:

> **The Department of Health should develop a comprehensive implementation plan that describes actions for its arm's length bodies and anticipated dates for completion.**
>
> The APPG recognises that although the bodies responsible for delivering actions have developed successful programmes in response to the Strategy, the absence of an implementation plan, with defined timeframes, objectives for actions and a forum for collaboration, has resulted in poor coordination among bodies and therefore limited progress in achieving the aims of the
>
> Strategy. An implementation plan is not just a piece of paper: it is a tool for delivery, coordination, collaboration, communication and monitoring.
>
> The development of an implementation plan for England would enable other stakeholders and bodies involved in the Strategy to coordinate activity on commitments where there is shared interest. It would send a clear message regarding the planned approach to delivering each aspect of the Strategy (which might include the decision not to proceed with a particular recommendation, due to a change in circumstance since the publication of the Strategy). This would enable stakeholder groups with an interest in progress towards implementation of specific commitments to understand which institutions they can work with to influence implementation, or which institutions they can expect to deliver implementation.
>
> As with the implementation plans produced by the other home nations, the plan should include a date for review and update.[11]

In a recent Parliamentary Question response, the Minister for Public Health and Innovation, Nicola Blackwood, stated that the strategy needed to be translated into an implementation plan and said that it was one of her personal commitments:

> Frist, let me pay tribute to my hon. Friend for his leadership of the APPG on rare diseases. I am sure he will join me in feeling proud that the UK is a recognised leader in research, treatment and care for rare diseases in particular. We are at the forefront of the genomics revolution. He is right that the UK strategy for rare diseases needs to be translated into an implementation plan, and that is one of my personal commitments.[12]

---

10 APPG for Rare, Genetic and Undiagnosed conditions, Leaving no one behind: Why England needs an implementation plan for the UK Strategy for Rare Diseases, February 2017

11 APPG for Rare, Genetic and Undiagnosed conditions, Leaving no one behind: Why England needs an implementation plan for the UK Strategy for Rare Diseases, February 2017

12 HC Deb 21 March 2017 c772

2. Parliamentary material

2.1 Debates

Access to medicines for people with cystic fibrosis and other rare diseases

HC Debate 8 December 2015 Vol 603 cc255-279WH

Drugs: Ultra-rare Diseases
HC Deb16 Jun 2015 | 597 cc29-55WH

Health: Rare Diseases
House of Lords Questions for short debate HL Deb 3 March 2015 | HL Deb 760 cc170-187

Drugs (Ultra-rare Diseases)
Commons Adjournment debates 20 January 2015 | Vol 591 cc22-31WH

NHS Specialised Services
Backbench Business Debate 15 January 2015 c335WH

2.2 Parliamentary Questions

Topical Questions
Ben Howlett (Bath) Conservative Party

Following the publication of the report of the all-party parliamentary group on rare, genetic and undiagnosed conditions on the UK rare diseases strategy, what plans does the Minister have to introduce an implementation plan for NHS England?

Nicola Blackwood | Conservative Party | Department: Health

Frist, let me pay tribute to my hon. Friend for his leadership of the APPG on rare diseases. I am sure he will join me in feeling proud that the UK is a recognised leader in research, treatment and care for rare diseases in particular. We are at the forefront of the genomics revolution. He is right that the UK strategy for rare diseases needs to be translated into an implementation plan, and that is one of my personal commitments.

21 Mar 2017 | | House of Commons chamber | 623 c771

Diseases

Jim Shannon: Democratic Unionist Party

To ask the Secretary of State for Health, what steps his Department is taking to ensure that patients do not face delays in obtaining access to treatments for rare and ultra-rare diseases.

Nicola Blackwood | Conservative Party | Department of Health

The UK Strategy for Rare Diseases, published in November 2013, contains 51 high level commitments which comprise a long term strategic vision for improving the lives of all those with rare diseases and conditions. The UK Strategy includes commitments to deliver effective interventions, treatments and support to patients quickly, equitably and sustainably. All four countries of the United Kingdom have agreed to implement the strategy by 2020.

We are committed to ensuring that patients with rare and ultra-rare diseases in England are able to access effective treatments.

This is why we launched the Accelerated Access Review to make recommendations to Government on speeding up access for National Health Service patients to innovative and cost effective new medicines, diagnostics and medical technologies. The Review was published on 24 October 2016 and further information is available at:

www.gov.uk/government/news/getting-patients-quicker-access-to-innovative-healthcare

The Government will respond to the review in due course.

The National Institute for Health and Care Excellence has recommended a number of treatments for rare and ultra-rare diseases through its Highly Specialised Technologies evaluation.

In addition, NHS England has made a number of treatments available for such conditions through its specialised commissioning procedure.

Written questions | House of Commons | 56703

Date answered: 16 December 2016

Diseases

David Hanson Labour Party

To ask the Secretary of State for Health, what consultation he expects to undertake on the implementation plan for England for the UK strategy on rare diseases.

George Freeman | Conservative Party | Department of Health

There are no plans to consult on the implementation plan for England on the UK Strategy for Rare Diseases. NHS England published a Statement of Intent to implement the UK Strategy in February 2014. Implementation of the Strategy is also included in NHS England's Five Year Forward View.

The UK Rare Disease Forum is responsible for monitoring progress on implementation of the Strategy across the United Kingdom. It published its first progress report on 29 February 2016. This recognises that significant progress has been made against majority of the 51 recommendations in the report. Further reports will be published in 2018 and 2020.

25 Apr 2016 | Written questions | House of Commons | 34391

Diseases

Julie Cooper Labour Party

To ask the Secretary of State for Health, if he will commission a review into the effectiveness of NHS England in discharging its duties in implementing the Rare Diseases Strategy.

George Freeman | Conservative Party | Department of Health

There are no plans to commission a review into NHS England's implementation of the UK Strategy for Rare Diseases. The UK Forum for Rare Diseases has been established to monitor implementation of the strategy across the United Kingdom. We have received a copy of the Forum's first progress report on implementation of the strategy and this will be published on 29 February 2016 - UK Rare Disease Day.

26 Feb 2016 | Written questions | House of Commons | 27373

Diseases: Health Education

Jim Shannon | Democratic Unionist Party

To ask the Secretary of State for Health, what steps his Department is taking to increase public awareness of rare diseases.

George Freeman | Conservative Party | Department of Health

The UK Strategy for Rare Diseases was published in 2013 and is currently being implemented across all four countries of the United Kingdom. The UK Rare Diseases Forum monitors the implementation of the Strategy and provides the Department with advice on rare disease policy development, including awareness raising. The 100,000 Genomes Project, a world-leading initiative launched by the Prime Minister in December 2012 has rare diseases as one of its key priority areas.

01 Feb 2016 | Written questions | Answered | House of Commons | 24433

Diseases

Andrew Stephenson | Conservative Party

To ask the Secretary of State for Health, what steps his Department is taking to support cooperation between rare diseases organisations

through the EU Policy Action and Information for Rare Diseases in Europe.

George Freeman | Conservative Party | Department of Health

The Government published the UK Strategy for Rare Diseases in November 2013. The strategy includes 51 specific commitments designed to improve the lives of all those affected by rare diseases.

The Government is committed to improving the lives of all those affected by rare diseases and the United Kingdom is recognised as a leader in Europe in the research, care and treatment of rare diseases. The strategy includes commitments to contribute to European Union-wide action on rare diseases.

Through the University of Newcastle upon Tyne, the UK successfully led the most recent EU Joint Action on Rare Diseases which concluded in 2015.

14 Jan 2016 | Written questions | House of Commons | 21311

Diseases

Andrew Stephenson Conservative Party

To ask the Secretary of State for Health, what steps he is taking to support the NHS recruit, retain and help the professional development of consultants specialising in (a) rare and (b) ultra-rare diseases.

George Freeman | Conservative Party | Department of Health

The Government published the UK Strategy for Rare Diseases in November 2013. The strategy is a high-level framework which sets out a strategic vision for improving the lives of all those affected by rare diseases. The strategy includes specific commitments regarding the education and training of the National Health Service workforce in the context of genomics and rare diseases.

Health Education England (HEE) was established to deliver a better healthcare workforce for England and for ensuring a secure workforce supply for the future. In addition, the HEE Genomics Education Programme was established in 2014 and is intended to build capacity and capability in response to the genomic medicine revolution. The programme includes rare diseases within its scope and will support the professional development of both new and existing members of the NHS workforce.

HEE has also published a suite of awareness raising videos to inform healthcare workers and the public about rare diseases. One video is aimed at general practitioners and highlights the importance of considering the possibility that their patient may have a rare condition.

14 Jan 2016 | Written questions | Answered | House of Commons | 21310

Diseases: Research

Margaret Ritchie | Social Democratic and Labour Party

To ask the Secretary of State for Health, what steps he is taking to increase research into the causes and types of rare diseases in England.

George Freeman |Conservative Party | Department of Health

The National Institute for Health Research (NIHR) has invested significantly in world-class research infrastructure in the National Health Service, to provide the facilities and people to undertake and increase research into rare diseases.This includes significant funding through the NIHR Biomedical Research Centres and Units, the NIHR Bio Resource - Rare Diseases which provides opportunities for patients, their families and health volunteers to take part in early translational research on the basis of their genotype and phenotype, and the dedicated NIHR Rare Diseases Translational Collaboration that brings together the country's leading researchers in rare diseases and promotes collaboration including with the life sciences industry.

In 2012 the NIHR also launched a call for Applied Clinical Research on Very Rare Diseases as part of the researcher-led funding streams across six of the NIHR Research Programmes. This call was issued in support of the UK Strategy for Rare Diseases and in recognition of the need for increased high quality evidence on the organisation of super specialised services or their commissioning and to improve the health outcomes for sufferers of rare diseases.

14 Jan 2016 | Written questions | House of Commons | 21206

2.3 Statement on UK Strategy for Rare Diseases

The Parliamentary Under-Secretary of State for Health

(Dr Daniel Poulter):

The "UK Strategy for Rare Diseases" has been published today.

This strategy is the overarching framework document that sets out a shared strategic vision for improving the lives of all those with rare diseases.

In the UK, one in 17 people—or more than 3 million individuals—will be affected by a rare disease at some point in their life. Rare diseases are a major cause of illness and make considerable demands on the resources of the NHS and other care services.

The document commits each UK country to over 50 actions that will be taken to deliver the vision outlined in the strategy. These actions focus on five main areas:

empowering those affected by rare diseases;

identifying and preventing rare diseases;

diagnosis and early intervention;

co-ordination of care; and the role of research.

The UK is already at the forefront of research, treatment and care for rare diseases. This strategy will further embed and enhance this reputation to the benefit of patients and the economy.

The "UK Strategy for Rare Diseases" has been placed in the Library. Copies are available to hon. Members from the Vote Office and to noble Lords from the Printed Paper Office. It is also available at:

www.gov.uk/government/publications/rare-diseases-strategy.

22 November 2013 CH col 66WS.

3. News Items

Genomes project 'making huge difference'

BBC News

30 December 2016

http://www.bbc.co.uk/news/av/health-38466394/genomes-project-making-huge-difference

DNA database brings scientists closer to pinpointing genes for disease

Guardian

17 August 2016

https://www.theguardian.com/science/2016/aug/17/dna-database-brings-scientists-closer-to-pinpointing-genes-for-disease

UK shows progress on rare disease strategy

Pharma Times

1st March 2016

http://www.pharmatimes.com/news/uk_shows_progress_on_rare_disease_strategy_1020334

Government launches strategy for rare diseases

The Pharmaceutical Journal

22 November 2013

4. Press releases

Rare Disease UK:

APPG launches report on the implementation of the UK Strategy for Rare Diseases in England Last Reviewed 28/02/2017

http://www.raredisease.org.uk/news-events/news/appg-launches-report-on-the-implementation-of-the-uk-strategy-for-rare-diseases-in-england/

The All Party Parliamentary Group on Rare, Genetic and Undiagnosed Conditions has today published a report calling on the Department of Health to take action to improve services for patients and families affected by rare conditions.

The report will be officially launched in Parliament this afternoon to mark Rare Disease Day, an international event that aims to raise awareness about rare diseases and their impact on patients' lives.

Over 300 patients and other members of the rare disease community, including family members, clinicians, patient representatives and industry, submitted evidence as part of the APPG's inquiry into the implementation of the UK Strategy for Rare Diseases in England. If implemented, the Strategy will 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.

Key finding: the UK Strategy for Rare Diseases is not being effectively implemented in England.

The departments of health in Scotland, Wales and Northern Ireland have all developed plans to improve services for rare disease patients that reflect their respective health service systems. The Department of Health in England has not coordinated an implementation plan for England.

Many patients in England do not have access to appropriate treatment or information about their condition.

Patients' health and social care is often poorly coordinated, with many struggling to access mental health services needed to help them cope with managing their rare condition.

Key recommendation: the Department of Health should develop a comprehensive implementation plan to deliver the Strategy in England.

Ben Howlett MP, Chair of the All Party Parliamentary Group on Rare, Genetic and Undiagnosed Conditions said:

"The report published today shows that too many people are struggling to access the right treatment and support for their rare condition. After working with and speaking to hundreds of patients over the past few months, it is evident that health and social care services are not being coordinated effectively in England. Patients in Scotland, Wales and Northern Ireland all have implementation plans that help to improve the

services patients need. It is important for patients in England to have this as well.

"The APPG is optimistic that if the Department of Health develops a plan, the situation will improve for patients in England."

Alastair Kent OBE, Director of Genetic Alliance UK, the national charity working to improve the lives of patients and families affected by all types of genetic conditions, said:

"Since the publication of the UK Strategy for Rare Diseases there has been heartening progress for patients and families with rare diseases in a range of different areas. However, as this report shows, much remains to be done as many patients still struggle to get the services and support they need, especially in England, where the absence of a clear implementation plan has resulted in a disjointed and sometimes incoherent approach to service delivery. This affects all the bodies involved in delivering services for rare disease patients, including NHS England. The need for an implementation plan is urgent and there is a role for the Department of Health to use its influence and authority to bring this about."

Rare Disease UK Forum

Delivering for patients with rare diseases : Implementing a Strategy

29 February 2016

A report from the UK Rare Disease Forum

Today (Monday 29 February) on international Rare Disease Day, the UK Rare Disease Forum (the group charged with monitoring the implementation of the UK Strategy for Rare Diseases) has published its first progress report. The report recognises that there has been significant progress across the UK but much remains to be done to meet the needs of the millions of people affected by rare diseases.

The UK Strategy for Rare Diseases, launched in November 2013, is a shared vision for improving the lives of all those with rare diseases. There are 51 commitments in the UK Strategy that each UK country has agreed to take forward by 2020. Effective implementation of the UK Strategy for Rare Diseases will help to secure the best use of available resources, maximise the health gain for all those with rare conditions and create a framework for the research necessary to support the development of innovative therapies for unmet medical needs.

The report from the UK Rare Disease Forum recognises that there has been significant progress towards delivering the commitments in the UK Strategy. For example, the 100,000 Genomes Project will transform molecular diagnostics across the UK and the creation of a unified registry of rare disease patients by Public Health England will have a

significant impact on assessing, planning and research for rare diseases.

However, as 'rare diseases do not recognise national boundaries' the UK Rare Disease Forum is calling for the four nations of the UK to work together to meet the needs of patients. Collaboration is essential if the National Health Services in the UK are to deliver the specialist and coordinated care required for patients affected by these multisystem conditions.

The Chair of Rare Disease UK, Alastair Kent OBE said: "The publication of the first report of the UK Rare Disease Forum is a key stage in the implementation of the Strategy published by the UK Health Ministers in 2013. There has been solid progress in many areas, notably in research and the setting up of the National Congenital Anomaly and Rare Disease Registration Service. The needs of patients and families with rare diseases are now acknowledged in ways that would have been unimaginable only a few years ago, but much remains to be done to meet the ambitious commitment to improve care and support for rare disease patients in the original strategy by 2020. I am confident that this goal is an achievable one, given the commitment of the Government and the NHS to delivering on the promise made to the 3.5 million people living with rare diseases in the UK today."

Life Sciences Minister, George Freeman MP said: "This report highlights the huge benefit that initiatives like our 100,000 Genomes Project and our £1bn per year National Institute for Health Research bring to people living with rare diseases and their families."

European Reference Networks: 900 medical teams to connect across Europe for the benefit of patients

Brussels, 28 February 2017

http://europa.eu/rapid/press-release_IP-17-323_en.htm

On 1 March the newly established European Reference Networks (ERNs) will start their work.

ERNs are unique and innovative cross-border cooperation platforms between specialists for the diagnosis and treatment of rare or low prevalence complex diseases.

Speaking to doctors, patients and the media at the University hospital in Leuven Belgium, Vytenis **Andriukaitis**, European Commissioner for Health and Food Safety, said:

"Today, on European Rare Disease Day, I am particularly pleased to launch the European Reference Networks. As a medical doctor, I have too often been witness to tragic stories from patients with rare or complex diseases who were left in the dark, sometimes unable to find an accurate diagnosis and receive a treatment. I have also seen my colleagues struggling to help because they lack information and

opportunities to network. These Networks will connect the considerable EU knowledge and expertise that is currently scattered between countries, making this initiative a tangible illustration of the added value of EU-collaboration. I am very confident that ERNs can light the way for rare disease patients, leading them to potentially life-saving and life-changing breakthroughs".

24 thematic ERNs, gathering over 900 highly specialised healthcare units from 26 countries, will begin working together on a wide range of issues, from bone disorders to haematological diseases, from paediatric cancer to immunodeficiency. Joining up of EU's best expertise on this scale should benefit every year thousands of patients with diseases requiring a particular concentration of highly specialised healthcare in medical domains where the expertise is rare.

The challenge of rare diseases

Rare diseases are those that affect no more than 5 in 10 000 people. Taken together, between 6 000 and 8 000 rare diseases affect the daily lives of around 30 million people in the EU - many of whom are children. Rare and complex diseases can cause chronic health problems and many of them are life-threatening. For example, there are almost 200 different types of rare cancers alone and each year more than half a million people in Europe are diagnosed with one.

The impact of such diseases on sufferers, their families and carers is substantial and patients often go undiagnosed due to lack of scientific and medical knowledge or to difficulty in accessing expertise. The fragmentation of knowledge about rare diseases and the small numbers of patients affected by a single disease marks it out as an area of particular EU-added value.

ERNs in brief

European Reference Networks (ERNs) are virtual networks bringing together healthcare providers across Europe to tackle complex or rare medical conditions that require highly specialised treatment and a concentration of knowledge and resources. They are being set up under the EU Directive on Patients' Rights in Healthcare (2011/24/EU), which also makes it easier for patients to access information on healthcare and thus increase their treatment options.

In practice, ERNs will develop new innovative care models, eHealth tools, medical solutions and devices. They will boost research through large clinical studies and contribute to the development of new pharmaceuticals, and they will lead to economies of scale and ensure a more efficient use of costly resources, which will have a positive impact on the sustainability of national healthcare systems, and for tens of thousands of patients in the EU suffering from rare and/or complex diseases and conditions.

The ERNs will be supported by European cross-border telemedicine tools, and can benefit from a range of EU funding mechanisms such as the "Health Programme", the "Connecting Europe Facility" and the EU research programme "Horizon 2020".

For more information

MEMO: Questions and Answers about European Reference Networks

Follow us on Twitter: @EU_Health_ IP/17/323 Press contacts:

- Enrico BRIVIO (+32 2 295 61 72)
- Aikaterini APOSTOLA (+32 2 298 76 24)

General public inquiries: Europe Direct by phone 00 800 67 89 10 11 or by email

Scottish Government:

Rare Diseases Action Plan

16 July 2014

More than 300,000 people affected in Scotland

The Scottish Government has set out a new strategy to combat rare diseases, which are estimated to affect more than 300,000 people in Scotland.

A rare disease is defined as a life-threatening or debilitating condition that affects fewer than five people in 10,000. There are currently around 8,000 such recognised conditions.

'It's not rare to have a rare disease' is the Scottish Government's first plan of action to combat rare diseases. The report, published today, was drawn up following a consultation with patients and clinical experts who were asked for their views on the treatment of rare diseases in Scotland. It sets out a series of suggested actions to bring about improvements in services and to empower people affected by rare diseases. The report also suggests ways to improve prevention, diagnosis and early intervention.

Each UK nation is developing their own plan of action as part of the UK Rare Disease Strategy. In turn this forms part of a Europe-wide drive to improve rare disease services.

Michael Matheson, Minister for Public Health, said: "What this report demonstrates is that although these diseases are individually rare, the sheer number of different conditions means that the total number of people affected is surprisingly large. "That is why we have drawn up this implementation plan, so we can really focus our efforts on improving services for people who are living with rare diseases.

"As with any disease, early and correct diagnosis is important. However, in the case of rare diseases, this can present particular challenges.

"I am very keen that patients will be closely involved in deciding how services are provided, and how treatments can be improved."

It's not rare to have a rare disease can be viewed in full here: http://www.scotland.gov.uk/Publications/2014/07/4751

Better support, treatment and research for millions of patients with rare diseases

Part of: Rare diseases and Research and innovation in health and social care

Department of Health,

22 November 2013

This was published under the 2010 to 2015 Conservative and Liberal Democrat coalition government

The first UK Rare Diseases Strategy was launched by Health Minister Lord Howe today.

Better support, treatment and research for millions of patients with rare diseases

The UK's first strategy to help build understanding of rare diseases and boost research to find effective treatments and therapies was launched by Health Minister Lord Howe today.

The UK Rare Diseases Strategy aims to ensure that none of the three million people in the UK who are affected by rare diseases are left behind.

For the first time, it sets out a UK-wide vision for building on our reputation as a world leader in rare disease research, including revolutionary genomic research to help transform diagnosis and treatment.

Key elements of the strategy include:

- a clear personal care plan for every patient that brings together health and care services, with more support for them and their families
- support for specialised clinical centres to offer the best care and support
- better education and training for health and social care professionals to help ensure earlier diagnosis and access to treatment
- promoting the UK as a world leader in research and development to improve the understanding and treatment of rare diseases

Health Minister Lord Howe said:

Millions of people in the UK are affected by the thousands of different kinds of chronically debilitating illnesses that are defined as rare diseases. So when looked at as a whole, rare diseases are not rare and our focus should be on making sure that no one who suffers from one is left behind.

For the first time, we are strengthening the links between research and the treatment and care of patients with rare diseases. This is about

putting those patients first, with better diagnosis, treatment and support for them and their families.

The UK already leads the way with ground-breaking research to better understand and treat these illnesses and this strategy will help cement our reputation as the driving force in this field.

A rare disease is a life-threatening or chronically debilitating disease that affects five people or fewer in 10,000, and requires special, combined efforts to enable patients to be treated effectively.

There are already more than 5,000 rare diseases identified and the total is steadily rising as new genetic discoveries explain previously unexplained disease patterns. It is estimated that one in 17 people will suffer from a rare disease in the course of their lifetime. In the UK, this means more than three million people will have a rare disease.

Therefore, when looked at as a whole, rare diseases are not rare. They represent a significant cause of illness, making considerable demands on the resources and capacity of the NHS and other care services.

The United Kingdom is a recognised leader in the field of rare disease research, treatment and care. The genomics revolution has the potential to radically transform the diagnosis and treatment of patients with rare diseases over the next ten years. Last year, the Prime Minister, David Cameron, announced that the personal DNA code — known as a genome — of up to 100,000 patients or infections in patients will be sequenced over the next five years. This will improve understanding, leading to better and earlier diagnosis and personalised care.

The project is being led by Genomics England, a company entirely owned by the Department of Health, and rare diseases is one of its key priorities.

In parallel with the launch of the UK Rare Diseases Strategy, the Government announced that the National Institute for Health Research (NIHR) is establishing a Rare Diseases Translational Research Collaboration (TRC). With investment of £20 million over four years, the NIHR Rare Diseases TRC will help to increase research collaboration that will lead to improved diagnosis, treatment and care for people with rare diseases.

Alastair Kent OBE, Chair of Rare Disease UK, which was part of the stakeholder forum that helped shaped the strategy, said**:**

Patients and families affected by life limiting rare diseases have often struggled to get a diagnosis and to access services that meet their needs. While the NHS has demonstrated that it is capable of delivering a world class service, the experience of patients has been that this is by no means a universal experience.

With the development of the UK Strategy for Rare Diseases patients and families can have a clear expectation of what to expect from the NHS, wherever they live in the UK. For the first time the four nations of the UK have come together to recognise and respond to the needs of rare disease patients systematically, structurally and in accordance with the

founding principle of the NHS that treatment should be a response to need.

Turning the UK Strategy's aims into practical benefits for patients will require hard work and detailed planning. Now, at least, patients can have a clear expectation of what the NHS aspires to provide for them, wherever they live in the UK. This is a huge step forward, and the publication of the Strategy will give hope and comfort to the 3.5 million people affected by rare diseases across the UK.

To mark the launch of the strategy, Lord Howe visited Great Ormond Street Hospital, which hosts a large number of nationally commissioned services for rare diseases.

Professor David Goldblatt, Director of Clinical Research and Development at Great Ormond Street Hospital and Director of the NIHR Biomedical Research Centre, said:

Many of the children attending Great Ormond Street Hospital suffer from rare diseases. Their families' experience of delays in the recognition of their syndrome or the absence of a clear diagnosis can be profoundly affecting. We look forward to continuing our work, in partnership with local NHS Trust and Primary Care physicians, in improving the experience of families with rare diseases.

Rare diseases are under-researched at present and in response, the NIHR-funded Biomedical Research Centre at GOSH/UCL is now focussing almost exclusively on rare disease experimental research. While Great Ormond Street Hospital has made some great strides, for example in pioneering gene therapy, much more work needs to be done.

Our ambition is to invest in rare disease research and advance understanding and treatments for the benefit of patients across the world. The hospital's charity is building a new Centre for Children's Rare Disease Research for the hospital and UCL Institute of Child Health, which will play a key role in developing therapies for many disorders for which no treatment or cure currently exists.

Background information

Click here to view the UK Rare Diseases Strategy.

Rare Disease UK (RDUK) is the UK alliance for people with rare diseases and all who support them. RDUK has over 1,400 members including over 240 patient organisations, individual patients and carers, clinicians, health professionals, researchers and industry.

RDUK is an initiative of Genetic Alliance UK, the national charity of over 160 patient organisations supporting all those affected by genetic conditions.

5. Further reading and useful links

UK Strategy for Rare Diseases Policy Paper

Information about the UK Strategy for Rare Diseases and how it will be implemented.

Department of Health

Published: 28 February 2014, Updated: 17 February 2017

UK Strategy for Rare Diseases Department of Health 2013

https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/260562/UK_Strategy_for_Rare_Diseases.pdf

UK Rare Disease Policy Board and Forum

https://www.gov.uk/government/groups/uk-rare-disease-forum

Rare Disease Forum Progress Report February 2016

http://www.shca.info/blog/post.php?s=2016-02-29-uk-rare-disease-forum-publishes-first-report

Rare Disease UK

https://www.raredisease.org.uk/about-us/

All-Party Parliamentary Group on Rare, Genetic and Undiagnosed Conditions

http://www.raredisease.org.uk/the-westminster-all-party-parliamentary-group-on-rare-genetic-and-undiagnosed-conditions/

Welsh Government - Rare diseases implementation plan

http://gov.wales/topics/health/nhswales/plans/rare/?lang=en

Northern Ireland Rare Disease Partnership

http://www.nirdp.org.uk/

Scottish Government Implementation Plan for Rare Diseases in Scotland

http://www.gov.scot/Topics/Health/Services/RareDiseases

.

DEBATE PACK
Number CDP 2017/0105
22 March 2017

About the Library

The House of Commons Library research service provides MPs and their staff with the impartial briefing and evidence base they need to do their work in scrutinising Government, proposing legislation, and supporting constituents.

As well as providing MPs with a confidential service we publish open briefing papers, which are available on the Parliament website.

Every effort is made to ensure that the information contained in these publically available research briefings is correct at the time of publication. Readers should be aware however that briefings are not necessarily updated or otherwise amended to reflect subsequent changes.

If you have any comments on our briefings please email papers@parliament.uk. Authors are available to discuss the content of this briefing only with Members and their staff.

If you have any general questions about the work of the House of Commons you can email hcinfo@parliament.uk.

Disclaimer