LEAVING NO ONE BEHIND:
Why England needs an implementation plan for the UK Strategy for Rare Diseases
The All Party Parliamentary Group (APPG) on Rare, Genetic and Undiagnosed Conditions aims to increase awareness of rare, genetic and undiagnosed conditions in Parliament and help to ensure that patients and families affected by these conditions have access to appropriate care and support.

Genetic Alliance UK is the national charity working to improve the lives of patients and families affected by all types of genetic conditions. We are an alliance of over 190 patient organisations. Genetic Alliance UK provides the secretariat for the APPG on Rare, Genetic and Undiagnosed Conditions.

Rare Disease UK is a multi-stakeholder campaign run by Genetic Alliance UK, working with the rare disease community and the UK’s health departments to effectively implement the UK Strategy for Rare Diseases.

SWAN UK (syndromes without a name) is a patient and family support service run by Genetic Alliance UK. SWAN UK offers support and information to families of children with undiagnosed genetic conditions.

Published by: Rare Disease UK
Level 3, Barclay House
37 Queen Square
London WC1N 3BH

Telephone: +44 (0)20 7831 0833
Email: appg@geneticalliance.org.uk
Website www.raredisease.org.uk

Registered charity numbers: 1114195 and SC039299
Registered company numbers: 05772999

Authors: Rosie Collington
Photo accreditation: Joshua Tucker (c)
Published: February 2017

Disclaimer
The views and opinions expressed in the quotes from patients are the respondents’ own, and do not necessarily reflect the views of members of the APPG, Genetic Alliance UK, Rare Disease UK or SWAN UK.

Any information provided that could be used to identify individual patients and their families, such as the names certain conditions, has been anonymised.
FOREWORD

I want to thank everyone that contributed to the inquiry conducted by the All Party Parliamentary Group on Rare, Genetic and Undiagnosed conditions that has led to this report published today. More than 300 patients, family members, patient organisations, clinicians and industry representatives submitted evidence as part of the enquiry into the implementation of the UK Strategy for Rare Diseases in England.

In November, I chaired three hearings with fellow members of the APPG in Westminster, which provided patients with the opportunity to share their experiences with Parliamentarians and representatives from the Department of Health, NHS England, the National Institute for Health and Care Excellence, and other bodies involved in rare disease patients’ care and treatment. These were thoroughly insightful meetings, and the APPG was grateful to hear directly about the challenges facing rare disease patients and their families.

It is clear that the current situation facing patients and families affected by rare diseases is not good enough. The UK Strategy for Rare Diseases has not been implemented in England, despite work behind the scenes. There are a number of commitments where no action has been taken at all:

- Too many people are struggling to access treatment and information about their condition and care that is coordinated between health and social care services
- Too many people do not feel involved in decisions about their care and treatment
- Too many people do not understand what is being done to improve their situation because these things are not being communicated properly.

Patients in Scotland, Wales and Northern Ireland have all benefited from having implementation plans for the Strategy and English residents deserve this too. An implementation plan would offer some direction to bodies involved in rare disease patients’ care, so they can take more action to improve the services patients need. It would also help patients to understand what progress is being made and more importantly to ask the right questions when it isn’t.

I look forward to hearing the Department of Health’s response to our recommendations of a comprehensive implementation plan that describes actions for its arm’s length bodies and anticipated dates for completion.

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

In the UK, it is estimated that 3.5 million people will be affected by a rare disease at some point in their lives. Despite the scale of the issue, a significant proportion of these patients do not have access to the right care and treatment.

The publication of the UK Strategy for Rare Diseases in 2013 should have heralded a new era in the treatment and care of rare disease patients in England, Scotland, Wales and Northern Ireland. Containing 51 commitments, the Strategy aims to ensure that health and social care systems across the four nations provide those living with rare conditions with the highest quality of evidence-based care and treatment, regardless of where they live in the UK.

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 four countries of the UK have until 2020 to implement the commitments outlined in the Strategy. While the departments of health in Scotland, Wales and Northern Ireland have all published country-specific implementation plans that reflect their respective health service structures and priorities, the Department of Health in England has not coordinated a plan for England.

Key features of the Strategy:

- A clear personal care plan for every patient that brings together health and social care services;
- Making sure patients, their families and carers have the information they need, are listened to and consulted;
- Developing better methods of identifying and preventing rare diseases;
- Improving diagnosis and earlier intervention for those with a rare disease;
- Better education and training for health and social care professionals;
- Building on research to improve personalised approaches to healthcare for those with a rare disease.

Scope of the inquiry

Between October 2016 and January 2017, the All Party Parliamentary Group (APPG) on Rare, Genetic and Undiagnosed Conditions held an inquiry on the implementation of the UK Strategy for Rare Diseases in England. Rare Disease UK collected written evidence on behalf of the APPG via e-mail and surveys, and conducted three hearings to hear oral evidence from patient representatives and other stakeholders involved in the Strategy, including the Department of Health and its arm’s length bodies (see annex 2). It also gathered information from existing publications to answer the following key questions:

- Why don’t we 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?
Key finding: the UK Strategy for Rare Diseases is not being implemented effectively in England. There are a number of commitments upon which no action has yet been taken.

– The Department of Health does not intend to develop an implementation plan, and believes NHS England should assume responsibility for doing so.

– NHS England does not intend to develop an implementation plan, and neither has the remit nor the capacity to influence all 51 commitments in the Strategy.

– There has been poor communication between organisations responsible for implementing individual commitments, and other stakeholders involved in the Strategy.

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

– The barriers to accessing information about a condition begin as soon as a diagnosis is made.

– Patient care continues to be poorly coordinated.

– Patients’ and families’ health and social care is poorly integrated.

– Patients and families do not feel involved in decisions about their care and treatment.

Key recommendation: 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 development of an implementation plan would enable stakeholders and bodies involved in the Strategy to coordinate actions for commitments they can influence. With this in mind, the APPG also recommends:

– NHS England should be more proactive in implementing the commitments it can influence, and dedicate more resources to improving coordination of care.

– The Department of Health should improve its processes to both engage and communicate with stakeholders in the Strategy.

– The Department of Health and the UK National Screening Committee should work together to establish robust programmes for identifying and preventing rare diseases.

– Training for front-line medical staff on rare diseases and their impact on patients should become widely available and incentivised.

– The departments of health in England, Scotland, Wales and Northern Ireland should work together more closely and collaboratively to ensure the Strategy becomes a reality.
A Strategy without a plan

In all devolved nations of the UK, the departments of health have taken responsibility for overseeing implementation of the UK Strategy for Rare Diseases. They have each developed a comprehensive plan that includes actions to be taken by health care services and other bodies, with expected dates for completion. The Scottish Government published “It’s Not Rare to Have a Rare Disease: The Implementation Plan for Rare Diseases in Scotland” in July 2014 (Scottish Government, 2014). The following February, the Welsh Government published the Welsh Implementation Plan for Rare Diseases (Welsh Government, 2015). In October 2015, the government of Northern Ireland published its Implementation Plan for Rare Diseases (Department of Health Northern Ireland 2015).

In England, there is currently no implementation plan for the Strategy. During its inquiry, the APPG learned that the Department of Health does not intend to develop an implementation plan, and maintains the position that implementation should rest with the organisations which deliver the services described in the document. Representatives from the Department of Health believe that its role is to facilitate bodies to implement individual commitments —without oversight or coordination.

In February 2014, the Department of Health published a table outlining which organisations would lead on actions to implement these individual commitments (Department of Health, 2014); this document is not an implementation plan. Unlike the plans developed in the devolved nations, the publication from the Department of Health in England did not include actions for delivery with proposed phasing and timing arrangements, and did not contain metrics for delivering or monitoring implementation.

The Department of Health believes that NHS England should assume responsibility for coordinating the implementation of the Strategy. NHS England, however, has stated that it does not intend to produce an implementation plan either. The APPG has learnt that NHS England neither has the remit nor the capacity to influence all 51 commitments in the Strategy. NHS England is not responsible for planning the work of other arm’s length bodies of the Department of Health.

Poor communication

In addition to issues with delivering the Strategy, the inquiry has revealed that there has been poor communication between the organisations responsible for implementing individual commitments and stakeholders involved in the Strategy. Patients, families, patient organisations, clinicians and industry
have not been provided with up-to-date information about the progress of implementing the Strategy. Nor were they informed or consulted with when a number of commitments were altogether abandoned.

The APPG has heard from over 300 patients with a wide range of diagnoses, their families and carers, clinicians, patient organisations and industry representatives. Although many patients and their families had initially felt optimistic when the Strategy was first published in 2013, the absence of an implementation plan and the failure to communicate progress effectively has led to widespread disillusionment and disappointment.

“Having a plan makes it seem as though it is a priority for the NHS. At the moment, it seems that rare diseases are not a priority.”

Parent of a child with a rare disease

Many stakeholders, including patients and patient representatives, do not think the Strategy has been implemented effectively in England. Respondents to the call for evidence do not understand why, when the departments of health in the devolved nations had developed implementation plans, the Department of Health in England had failed to do so.

This organogram shows the disparity between the existing processes for implementing the Strategy in England versus those in Scotland, Wales and Northern Ireland, and the resulting reduction of opportunity for collaboration, planning and input of patient voice in activities in England. (HTA: Health Technology Assessment)
“The UK Strategy for Rare Diseases seems to have become like a person with a rare disease, with no department seeming able or willing to decide who has overall responsibility. [...] Patients are used to the game of interdepartmental ping pong and now the UK Strategy for Rare Diseases is in the same situation.”

Patient with Mal de Debarquement Syndrome

Many patients and others involved in their care see an implementation plan as critical to improving services that support their needs because it would enable them to hold health and social care services to account.

“The 51 commitments in the UK Strategy for Rare Diseases are all excellent, although I realise that advances in technology may mean that some of them are no longer applicable in their original form. My main concern with them is that it is hard to know who is responsible for implementing them. If an implementation [plan] makes this clear – and encourages accountability – I’m all for one.”

Patient with Mal de Debarquement Syndrome

During the inquiry, the Department of Health stated that it does not believe it is responsible for communicating progress of the Strategy to stakeholders, because this role is already fulfilled by patient organisations. The majority of patient groups are under-resourced and run by volunteers and patients from the rare disease community. It is not clear how patient organisations would be able to access accurate and up-to-date information on implementation to disseminate.

Patient representatives at the APPG’s hearing on Tuesday 1 November 2016.
Progress: because of the strategy, alongside the strategy, and yet to happen

In the following section, the APPG has provided an overview of where progress has been made in implementing the Strategy, and where gaps remain.

Programmes developed in response to the Strategy

The developments listed below have evidence of both comprehensive plans for delivering commitments in the Strategy and outcomes that have been communicated to patients.

In response to the Strategy, the National Institute for Health Research (NIHR) launched the Rare Diseases Translational Research Collaboration (RD-TRC). The RD-TRC aims to empower those affected by rare diseases through patient engagement and involvement in research and research funding decisions. Through developing partnerships with a range of stakeholders, the RD-TRC contributes to research into the development of new treatments for rare diseases. For example, the RD-TRC works closely with industry partners to select rare disease therapeutic themes and projects for deep phenotyping. The RD-TRC also works with both clinicians and patients to assemble quality clinical databases and sample collections across a wide range of rare disease diagnoses.

Over the last four years, the NIHR has invested over £4 million in the RD-TRC, and the programme will continue for another five years with a £5 million investment. While the APPG is optimistic about the benefits this will yield for rare disease patients, it understands that the average total expenditure of the NIHR is £1 billion.

In March 2015, Public Health England in partnership with the Department of Health established the National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) to record information about people with congenital abnormalities and rare diseases across the whole of England. The programme has helped to progress a few key areas affecting diagnosis and early intervention of rare diseases, and the role of research. For example, NCARDRS has developed systems to record genetic and other relevant information accurately to identify the incidence and prevalence of diseases, and support service planning and international planning. It has also developed a patient portal, which enables patients to access their own data after it has been collected.

Jem Rashbass, National Director for Disease Registration at Public Health England at the APPG’s hearing on Monday 7 November 2016.

Following the publication of the Strategy, Health Education England (HEE) collaborated with the National School of Healthcare to produce two educational videos for health care professionals, to raise awareness of the problems faced by families with a child with an undiagnosed condition, and the importance of considering the possibility of a rare disease diagnosis. It is hoped that successful engagement with these videos could contribute to the training needs of GPs identified in the Strategy to improve diagnosis...
and early intervention of rare diseases. In 2015, the Department of Health commissioned a report through the London School of Tropical Medicine to assess the appropriateness of methodologies evaluating rare disease patients’ ‘diagnostic odyssey’.

**Programmes developed alongside the Strategy**

During the inquiry, the APPG was informed of a number of programmes that have not been developed in response to the Strategy, but complement its aims. While these developments should be taken into account in any evaluation of progress towards meeting the aims of the Strategy (and also in planning activity to implement the Strategy), they should not be considered in progress reports as an action resulting from the UK Strategy for Rare Diseases.

The APPG is encouraged by the work of Genomics England and the work of the 100,000 Genomes Project. When providing evidence to the APPG, the Department of Health and NHS England discussed the successes of the project in implementing the UK Strategy for Rare Diseases. The 100,000 Genomes Project was launched in 2012 as part of the Government’s Strategy for UK Life Sciences (Department for Business Innovation and Skills, 2011).

The APPG would like to acknowledge that the welcome progress in understanding the human genome does not benefit all affected by rare diseases. Approximately 20% of rare disease patients have a condition which is not of a genetic cause.

The Genomics Education Programme (GEP), developed by Health Education England, has helped to improve awareness among service providers of the effects that living with a rare disease can have on a patient’s life. This programme, has also been developed as part of the 100,000 Genomes Project.

Working with the Department of Health, NHS England has supported the development of European Reference Networks (ERNs). ERNs will be networks of expert centres that provide highly specialised healthcare, intended to improve diagnosis, treatment and high-quality care for patients with conditions requiring a particular concentration of resources or expertise (Hedley, 2016). In 2016, the European Commission received the first round of applications to establish ERNs. Of the 24 applications that were received, six were coordinated by UK Healthcare Providers. The APPG is impressed with the broad scope of UK ERNs and the support of the Department of Health in developing these. The initiative to create ERNs results from Directive 2011/24/EU on patients’ rights in cross-border healthcare.

A number of bodies involved in implementing the Strategy have continued to empower patients through engagement with their processes and structures. NHS England involves patients in decisions about its services through the Rare Diseases Advisory Group (RDAG) and multidisciplinary Clinical Reference Groups. The National Institute for Health and Care Excellence (NICE) involves patient and carer representatives in its Highly Specialised Evaluation Committee.

Patient representatives at the APPG’s hearing on Tuesday 1 November 2016.

In July 2014, the NIHR established a ‘Muskeeters’ Memorandum’, the UK Rare Genetic Disease Research Consortium Agreement, to help speed up the approval process for rare disease research (The British Society for Genetic Medicine, 2016). The
development of this agreement began in 2012, before the UK Strategy for Rare Diseases was published, through the efforts of representatives from NHS Trusts with regional genetic centres.

The Strategy states that specialist clinical centres should, as a minimum standard, coordinate care, ensure their expertise is available to families and their healthcare teams, and arrange for coordinated transition from children’s to adults’ services, amongst other actions. The APPG is concerned about the lack of evidence indicating that standards have been set or met nationally for specialist clinical centres, and that little has been done to encourage the excellent work of centres such as Birmingham Children’s Hospital to establish a rare disease centre.

It must be noted, nonetheless, that a number of specialist centres have independently developed programmes and staffing roles to facilitate successful coordination of rare disease patients’ care. For example, Birmingham Children’s Hospital, in partnership with Roald Dahl’s Marvellous Children’s Charity, employ a member of staff to coordinate patients’ care as they transition between children’s, adolescents’ and adults’ care settings. During the inquiry, a number of patient representatives praised the positive impact these efforts have had on the lives of patients and families under the care of Birmingham Children’s Hospital.

Other areas in development
During its hearings with the Department of Health, NHS England, the RD-TRC, NCARDRS and the NIHR, the APPG was informed about plans for actions in response to the Strategy that are currently at an early stage of development. The APPG looks forward to seeing evidence of implementation of these plans in the near future:

- NHS England is developing an electronic ‘prompt’ to encourage GPs to consider rare disease diagnoses. An effective prompt in primary care services would help doctors to identify a set of symptoms as a rare condition.
- The NIHR and its RD-TRC programme also use digital technologies to engage patients in research and increase the evidence base for rare disease research. The NIHR is currently redeveloping the UK Clinical Trials Gateway so that patients can make contact with researchers directly. A patient or family would be able to electronically indicate to a research group that they are interested in participating in a project. The RD-TRC is in the process of developing two experimental apps, which would enable patients to describe their experiences and symptom progression. This could then be mapped to clinical phenotypes as a direct measurement of, or proxy of, an underlying symptom.
- In 2014, NHS England developed a Rare Disease Annex (also known as the Rare Disease Insert) that could improve care coordination. Rare Disease UK’s Patient Empowerment Group (PEG) is a collective of expert patient representatives that has been following and supporting the implementation of the Strategy since its conception. PEG responded positively to the development of the Rare Disease Annex, and in September 2015, PEG provided feedback to NHS England on a draft version. The group was pleased that all suggested changes were accepted and in December 2015, the Rare Disease Annex was approved by RDAG. Over a year later, the Annex has not yet been incorporated into relevant NHS England service specifications.

Gaps in action
There are a number of areas in the Strategy where no action has been taken to implement commitments. The APPG did not find evidence of:
– Actions to identify and prevent rare diseases, including screening, carrier testing, preconception and antenatal care, antenatal and newborn screening and cascade testing for severe genetic conditions.

“There is no commitment by the Department of Health to implement this important commitment in the UK Strategy for Rare Diseases, which is creating delays in diagnosis for children with disorders that are treatable and that failure to treat at the earliest opportunity would lead to a life of disability or even death.”

Patient representative

– Improved processes for patients’ care coordination, such as the development of service specifications and a generic care pathway.

– A commitment to support and resource engagement with patients, clinical care teams, researchers and industry in a number of areas. In particular, the APPG is concerned about the potentially detrimental impact of the reconfiguration of the UK Rare Diseases Forum, which was originally charged with monitoring implementation and providing progress reports. In 2016, the Department of Health announced that it would meet just once, instead of four times, per year. The proposed UK Rare Disease Policy Group, which would meet in place of the current forum three times a year, did not initially include patient representation and external stakeholders with an interest in rare diseases. Questions about how patient engagement will be resourced and structured in the new UK Rare Diseases Policy Group’s Task and Finish Groups remain unanswered.

“There is little evidence of initiatives that contribute to the implementation of the UK Strategy for Rare Diseases. They really have no commitment to recognising patient groups as partners in implementing the Strategy.”

Patient representative

– Structures to facilitate collaboration in implementing Strategy between all four nations of the UK. With no central body to oversee implementation of the Strategy in England, the departments of health in Wales, Scotland and Northern Ireland are unable to coordinate effectively with health care and research bodies in England that are responsible for delivering individual commitments. The inability of all four nations to work collaboratively has a detrimental impact on patients in the devolved nations, who often struggle to access nationally funded specialised services in England.
How the absence of a plan is preventing effective delivery of rare disease services

The APPG has heard evidence from a wide range of stakeholders in the UK Strategy for Rare Diseases, including patients, their families, patient organisations clinicians, and industry representatives suggesting that rare disease patient experiences have not improved in a number of areas since the publication of the Strategy.

Many patients do not have access to appropriate treatment and information about their condition.

There are only a limited number of licensed medicines for rare diseases in England, and many patients are unable to access adequate support and treatment for their condition.

“Considering just 5% of rare diseases have a licensed treatment option [...] it is critical that the commitments outlined are taken forward to find a suitable appraisal system of orphan medicines, that fully accounts for rarity and supports the NHS in its mission to provide a comprehensive service, available to all.”

Industry representative

Even where specialist clinical centres exist, they are poorly utilised, although there are exceptions.

“We struggle to find anyone with knowledge of our son’s condition. We feel that we have to be the experts in the condition and pass this knowledge onto the professionals.”

Parent of a child with a rare disease

Referrals to specialist clinical centres are patchy, and many patients believe access to them varies significantly between regions.

“The service you get depends on where you live. The service in Newcastle was not good, and yet we had no other choice than to accept inappropriate advice and a poor standard of care. There was no access to any ongoing research. We have had an excellent service in Manchester.”

Parent of a child with a rare condition

“My experience of seeing the care for neuromuscular conditions (rare, genetic, highly specialised) is that the level of care and support that is provided by centres around the country is highly variable ranging from full service support and co-ordinated care to non-existent.”

Industry representative

There is a disparity between the standards of care patients are able to access depending on their diagnosis and whether a specialist centre exists to treat the condition. For example, the inquiry heard from a number of patients with Lymphangioleiomyomatosis (LAM) who were keen to highlight the excellent standard of care they receive at the National Centre for LAM at Nottingham University Hospitals NHS Trust.

“My treatment at the LAM centre at the Queen’s Medical Centre in Nottingham under Professor Johnson is excellent.”

Patient with LAM

However, many patients also find that even where specialist centres exist, patients are not referred to them.

“Even though there is a national alkaptonuria specialist centre, often patients don’t get referred. There is a lack of awareness about the centre.”

Patient representative

A number of patients and their families sought expertise and access to clinical trials abroad, where they have been unable to access appropriate treatment to manage their condition in England. Often, patients and their families were bearing the cost of this.

“Doing the trial in the USA meant that she would have to give up her work in the UK and leave behind her social life and friends.”

Parent of a patient with a rare disease

“I have to arrange everything or go overseas for treatment at my own cost [...] I fundraised and got to the USA and more recently, Germany. Things are too slow and not recognised here in the UK”

Patient with spontaneous intracranial hypotension

“There are three of my family affected. We have to pay for private overseas treatment.”

Patient with Lyme disease

Rupert Purchase, Chair of the Wilson’s Disease Support Group UK, at the APPG hearing on Monday 7 November 2016.
The barriers to accessing information about a condition begin as soon as a diagnosis is made.

Many patients experience delays in receiving a correct diagnosis, with many waiting years and receiving numerous misdiagnoses before being diagnosed with their rare disease. The overwhelming majority of patients reported that they had not received any information about opportunities to participate in research at the time of diagnosis, with many also indicating that they had not been provided with information about options for treatment or management of their condition.

“My parents were given no advice on what would happen with my disease and no real support. My mother researched the best that she could and pushed doctors and physicians to help as much as possible. It wasn’t until my twenties that finally a haemophilia centre agreed to have me under their care.”

Patient with a rare disease

“We were given a compliments slip with [the condition] written on it and told to ‘go and look it up on Google if we wanted more information’.”

Parent of a child with a rare disease

Often, patients and their families were referred to a patient organisation for support. The APPG recognises that in this regard, patient organisations often become responsible for providing information, support and services for patients and their families where public provision is inadequate. A number of patient organisations fund, for example, counselling and mental health services for both patients and families.

“We were given no information on receiving a diagnosis, just told to contact [the charity] Contact A Family.”

Parent of a child with a rare genetic disease

“My family was provided with information through Climb, a wonderful charity that supports children with metabolic diseases.”

Patient with a rare metabolic disease

Nonetheless, many stakeholders are uncomfortable with this model.

“I have ethical issues about the use of patient-run support groups and/or charities to deliver the UK Strategy for Rare Diseases. If patients do a lot of the work now, we’re setting up the next generation for more of the same, instead of making all the various departments responsible and accountable. They are publicly funded – we’re not, other than by charitable donations.”

Patient with Mal de Debarquement Syndrome

Patients’ care continues to be poorly coordinated.

Many patients still do not receive coordinated care. Research conducted by Rare Disease UK over the past seven years indicates that poorly coordinated care is an important issue for many rare disease patients and families (Rare Disease UK, 2010; 2013; 2016). Genetic Alliance UK’s Hidden Costs Feasibility Study shows further that receiving coordinated care remains a challenge for the rare disease patient community because it results in excessive and uncoordinated appointment scheduling; a lack of communication between providers; and a lack of resources and capacity within services (Genetic Alliance UK, 2016: 4).

In 2016, Rare Disease UK conducted a survey of rare disease patients and their families to get
James Palmer, Clinical Director of Specialised Services at NHS England, at the APPG’s hearing on Tuesday 15 November 2016.

an up-to-date picture of patients’ experiences of diagnosis, accessing information about their condition, receiving appropriate coordinated care, accessing treatments and finding out about research (Rare Disease UK, 2016). The report showed that 1 in 3 patients have to attend three or more clinics to attend appointments related to their condition (ibid: 22). Responses to the APPG’s call for evidence on the implementation of the Strategy in England indicate that there has not been an improvement in this area since then.

“My daughter is currently seen by consultants in medical disciplines at two hospitals, none of whom share records or communicate with the other. Every new referral requires us to explain the history all over again.”

Parent of a child with Kawasaki disease

An overwhelming number of respondents affected by conditions that affect multiple systems said that they do not receive coordinated care between clinicians treating individual symptoms.

“All doctors said they were only able to treat their part of my daughter’s anatomy. Doctors were unable to confer or share medical records. GPs said they ‘did not know who does what’.”

Parent of a patient with a rare disease

“When my son was found to have a malignant peripheral sheath sarcoma […] he was seeing a neurologist, a urologist, pain management, a general consultant for a double hernia, a nerve specialist plus other medical professionals. All this was during [a busy time in his life] so he also needed counselling. None of this was coordinated; he was offered treatments that were contra-indicated with other conditions he had.”

Parent of a patient with neurofibromatosis 1

Many patients and families feel that rare disease patients’ care becomes less coordinated when they transition to adult care. The disparity between treatment rare disease patients received under the care of children’s services for rare diseases and adults’ services was noted by a number of patients and their families. Rare Disease UK’s report on patient experiences of transition between care providers indicates that at times, adult care can be better coordinated as well (Rare Disease UK, 2014).

“When my daughter left tertiary paediatric service, she was told they could not refer to adult tertiary service, and to present at A&E ‘tactically’.”

Parent of a patient with a rare disease

“When my son was found to have a malignant peripheral sheath sarcoma […] he was seeing a neurologist, a urologist, pain management, a general consultant for a double hernia, a nerve specialist plus other medical professionals. All this was during [a busy time in his life] so he also needed counselling. None of this was coordinated; he was offered treatments that were contra-indicated with other conditions he had.”

Parent of a patient with neurofibromatosis 1

Many patients and families feel that rare disease patients’ care becomes less coordinated when they transition to adult care. The disparity between treatment rare disease patients received under the care of children’s services for rare diseases and adults’ services was noted by a number of patients and their families. Rare Disease UK’s report on patient experiences of transition between care providers indicates that at times, adult care can be better coordinated as well (Rare Disease UK, 2014).

“When my daughter left tertiary paediatric service, she was told they could not refer to adult tertiary service, and to present at A&E ‘tactically’.”

Parent of a patient with a rare disease

“When my son was found to have a malignant peripheral sheath sarcoma […] he was seeing a neurologist, a urologist, pain management, a general consultant for a double hernia, a nerve specialist plus other medical professionals. All this was during [a busy time in his life] so he also needed counselling. None of this was coordinated; he was offered treatments that were contra-indicated with other conditions he had.”

Parent of a patient with neurofibromatosis 1

Many patients and families feel that rare disease patients’ care becomes less coordinated when they transition to adult care. The disparity between treatment rare disease patients received under the care of children’s services for rare diseases and adults’ services was noted by a number of patients and their families. Rare Disease UK’s report on patient experiences of transition between care providers indicates that at times, adult care can be better coordinated as well (Rare Disease UK, 2014).

“Patchy and uncoordinated care will ultimately result in greater costs to the NHS than properly implementing the UK Strategy for Rare Diseases.”

**Patient with relapsing polychondritis**

“The savings are huge, potentially, for rare diseases that have treatments, that are already funded, that have centres of excellence. People should have a right to access these. With vasculitis, renal replacement therapy costs tens of thousands of pounds a year. If you treat someone right at the beginning of their disease, it won’t get to the point where they need dialysis. It’s cost effective.”

**Patient representative**

“It is important to note that patients without a diagnosis typically absorb significant amounts of medical resources, tests and consultations for no benefit. There is an opportunity, simply through better knowledge and coordination, for the NHS to deliver more effective treatment at a much reduced cost.”

**Patient representative**

**Patients’ and families’ health and social care is poorly integrated.**

Many patients experience poor coordination and communication between health and social care providers. This often is not only detrimental to the patient’s health, but has a negative psychosocial impact on the patient’s family.

“We have no help or support and every day can be a struggle. We have devoted all our time and energy into ensuring our child is as happy and supported as she can be with our limited means. I have given up my career to do this and we have no financial support.”

**Parent of a child with a rare disease**

“I don’t get care at the moment. I am told by the social services that they cannot get anybody to cover this area.”

**Patient with a rare disease**

During the inquiry, a number of patient organisations raised concerns about rare disease patients’ support for mental illness resulting from neurobiological symptoms that are a result of the condition, and/or the psychological impact of living with a rare disease. Many rare disease patients and their families struggle to access appropriate mental health services.

“I’m on my own and family have to care for me. [I was offered] a brief allotted time for Cognitive Behavioural Therapy for about six weeks when I was feeling suicidal”

**Patient with Behçet’s disease**

“Sheela Upadhyaya, Associate Director of Highly Specialised Technologies at NICE at the APPG’s hearing on Tuesday 15 November 2016.”
“I need more specialist input as certain treatments can cause issues with my rare disease. This isn’t available so I’ve had to struggle without medication for mental health issues as antidepressants cause vision problems in my PIC eye.”

Patient with punctate inner choroidopathy

Some existing specialist clinical centres for rare diseases provide specialist psychological support for patients, but this is rarely offered to patients.

“I was able to access an assessment of my son’s mental health with the specialist Deaf CAMHS [Child and Adolescent Mental Health Services], although I had to plead with my doctor to get a referral. The assessment carried out was fantastic because he understood the impact that sensory impairment has on mental health.”

Parent of a child with Alstrom Syndrome

Patients do not feel involved in decisions about their care and treatment.

Many patients feel their symptoms or conditions have been dismissed or not taken seriously by primary care clinicians who do not understand rare diseases. In some cases, this has resulted in misdiagnoses or complications during routine medical treatments. Patients, and sometimes their family members, reported that they had been accused of exaggerating or fabricating their symptoms. Few patients feel properly consulted on or involved in decisions about their care. This indicates not only a poor understanding among medical staff about rare diseases and diagnostic pathways for uncommon symptoms, but also a lack of understanding among front-line staff about the psychosocial impact of living with a rare disease. Misunderstanding about and mismanagement of a patient’s condition is psychosocially detrimental to the patient.

“I have been made to feel at times as if I am overreacting with some of my questions and prompting.”

Family member of a patient with a rare disease

“If we research things and discuss things on a medical level [...] I as the mum then get accused of fabrication of the illness and Munchausen’s by proxy.”

Parent of a child with suspected Mast Cell Activation Syndrome and related conditions

“The consultant assigned to us at our local hospital told us very clearly in our first meeting [that] he did not believe in her condition and basically accused her (at eight years old) of making it up.”

Parent of a child with a rare disease

Many patients wanted to highlight the efforts of individual clinicians and members of NHS staff who have supported them throughout their diagnosis, treatment and care. Nonetheless, patients frequently noted that they believe that the workload of clinicians and NHS staff often prevents them from providing adequate care.

“[The extent to which I feel I am listened to and consulted on to agree the best way forward for my care and treatment] all depends on the workload of the medical professional seeing me.”

Patient with Human T-Cell Lymphotrophic Virus 1

“There are four doctors who [...] have done their best in difficult circumstance and they have the best of intent. They are however working in a system that is broken.
These doctors are authentic individuals, with the patient at the core.”

**Parent of a child with a rare disease**

This was even the case at specialist clinical centres.

“At my specialist hospital [...] the staff are hugely skilled and compassionate but very over-worked.”

**Patient with a rare connective tissue disease**
CASE STUDY: SUCCESSFUL PATIENT INVOLVEMENT AND ENGAGEMENT IN RESEARCH

The Rare Diseases Transnational Research Collaboration (RD-TRC) was established by the National Institute for Health Research (NIHR). Since 2013, the RD-TRC has developed a number of “mechanisms and opportunities for meaningful and sustained patient involvement in rare disease service provision and research” (Department of Health, 2013). The RD-TRC has recruited two patient and public representatives to be part of its Strategic Oversight Group, and a further fourteen to be part of its Patient Advisory Group. These groups help to direct the overall activity of the RD-TRC and establish its fundamental values and ethical principles. One patient, who has been involved with the Patient Advisory Group described her motivations for getting involved:

“I felt it was important to be able to positively make a difference to those people who are still to be diagnosed and are still being pushed from doctor to doctor with no success, or even worse given the wrong diagnosis.”

The RD-TRC strives to involve patients and carers in its work. To date, it has hosted two research symposia, held in September 2015 and 2016. In the most recent event, patients spoke about their involvement in research where they were able to contribute to the design and success of the projects.

The RD-TRC has also sought to achieve its aim through training activities. The Think Research! Rare Diseases Patient Day was a collaboration between the RD-TRC and NIHR BioResource – Rare Diseases, and was held on 12 October 2016 at the Barbican Centre in London. The event aimed to offer patients free training in various aspects of being part of rare disease research, as well as provide an opportunity to network with other patients and groups. 135 people attended the event, over 100 of whom were patients. Becks Breslin, founder of charity Beat SCAD (spontaneous coronary artery dissection), co-ran a training session on running a successful patient group at the event:

“The Think Research! Rare Diseases Patient Day was a fantastic experience – it is wonderful that the NIHR are hosting these events as they are enabling patients and families affected by rare disease to share experiences, learn more about the industry and have a platform to ask questions.”

The RD-TRC has also developed a number of structures to ensure the studies it funds prioritise patient involvement through the award process. For example, its funding application form specifically asks applicants to provide details about the involvement of patients and the public in identifying the research question and preparing the proposal; and future plans for the involvement of patients and the public in the proposed research.
DISCUSSION AND RECOMMENDATIONS

The rarity of a person’s condition should not determine what their future looks like. The All Party Parliamentary Group on Rare, Genetic and Undiagnosed Conditions is optimistic that implementation of the UK Strategy for Rare Diseases in England will significantly improve the lives of rare disease patients and their families. It is disappointed, however, by the critical lack of evidence indicating progress to overcome a number of issues identified in the Strategy. This adversely affects not only the rare disease community, but also our wider understanding about the causes, treatment and care of more common conditions as well.

The findings presented in this report are indicative of a failure to implement the UK Strategy for Rare Diseases in England effectively. In response to the issues identified during the inquiry, the APPG recommends that:

**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

---

Lesley Harrison, Patient Support Manager from the AKU Society, at the APPG’s hearing on Tuesday 1 November 2016.
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.

NHS England neither has the remit nor the capacity to influence all 51 commitments in the Strategy. NHS England is not responsible for planning the work of other arm’s length bodies of the Department of Health and therefore, cannot be responsible for developing an implementation plan for England. However, the APPG recognises that NHS England could do more to implement the Strategy and makes the following recommendations to that end.

**NHS England should be more proactive in implementing the commitments it can influence and dedicate more resources to improving patients’ care coordination.**

During the APPG inquiry, coordination of care was raised as a particularly important issue to patients, yet this is an area that is significantly underdeveloped. The APPG recommends that NHS England delivers on its responsibility to develop service specifications, care pathways and other processes to improve rare disease patients’ care coordination. This includes the development of minimum standards for specialist clinical centres. Improved care coordination would also help to reduce the heavy workload of clinicians working in rare diseases, which many patients regard as harmful to their condition. It would help to overcome patients’ concerns about patchy referrals. Specialist clinical centres are ideally placed to provide psychological support to patients affected by rare diseases.

Health and social care do not exist independently from each other; processes should also be put in place to improve coordination between health and social care services.

The APPG hopes that the Department of Health would involve patient organisations that have successfully delivered care coordination pilots in its plans for implementation. Patient organisations play an indispensable role in providing services and support, but many of what they currently fund cannot be delivered nationally without NHS integration and support.

**The Department of Health should improve its processes to both engage and communicate with stakeholders in the Strategy.**

The APPG believes that communication and engagement with the rare disease patient community and other stakeholders is a necessary and valuable task. It also agrees this is vital to ensuring patient involvement in the development of an implementation plan. The Department of Health should not only communicate progress in implementing the Strategy in England, but also the reasons why there remain gaps in certain areas.

Kerry Leeson-Beevers, National Development Manager from Alström Syndrome UK, at the APPG’s hearing on Tuesday 1 November 2016.
In recognition of patients’ expert knowledge of the reality of living with a rare disease and their invaluable contribution to the development of the Strategy, the APPG hopes the Department of Health will ensure that patient involvement in oversight of the Strategy’s implementation is both substantive and supported with appropriate resources.

The Department of Health and the UK National Screening Committee should work together to establish robust programmes for identifying and preventing rare diseases.

The APPG is concerned about the lack of coordinated action to improve the identification and prevention of rare diseases through screening and carrier testing. Developments in this area have the potential to change the lives of hundreds of patients with genetic conditions and families affected by fetal abnormalities. Given the extent of existing and ongoing research in this area that complements the aims of the Strategy, the APPG would like to see concrete plans to implement commitments for identifying and preventing rare diseases.

Training for front-line medical staff about rare diseases and their impact on patients and families should become widely available and incentivised.

Training can help to overcome the various barriers patients face when trying to reach a correct diagnosis and access to treatment.

The APPG hopes that raising awareness about rare diseases among front-line medical staff would help to improve the relationship between patients and their families and the professionals who treat them. Successful education would equip clinicians with the knowledge to provide clear and timely information about practical and psychological support available to patients.

The departments of health in England, Scotland, Wales and Northern Ireland should work together to ensure the Strategy becomes a reality.

Effective implementation of the UK Strategy for Rare Diseases will not be possible without concerted collaboration between all four nations of the UK.
REFERENCES


Rare Disease UK, 2010. Experiences of Rare Diseases: An Insight from Patients and Families. [pdf]. London: Rare Disease UK. Available at: <https://www.raredisease.org.uk/media/1594/rduk-family-report.pdf> [Accessed 13 January 2017].

Rare Disease UK, 2013. Rare Disease Coordination: Delivering value, improving services. [pdf]. London: Rare Disease UK. Available at: <https://www.raredisease.org.uk/media/1639/rduk-care-coordinator-report.pdf> [Accessed 13 January 2017].


ANNEX 1: METHODOLOGY

The APPG on Rare, Genetic and Undiagnosed Conditions conducted an inquiry between October 2016 and January 2017 on the implementation of the UK Strategy for Rare Diseases in England. Stakeholders in the Strategy were invited to submit evidence.

In November 2016, the APPG hosted a series of hearings in Portcullis House, Westminster. These events aimed to gather qualitative evidence on the reasons why there is not currently an implementation plan for England, and what impact the absence of an implementation plan is having on patients in England. Minutes for all these events and a full list of attendees can be found on the Genetic Alliance UK website.

- On Tuesday 1 November, rare disease patients, family members and patient representatives were invited to share their experiences on accessing rare disease services, and their understanding of the implementation of the Strategy.
- On Monday 7 November, representatives from the Department of Health, Public Health England and the National Institute for Health Research provided evidence and shared views on the reasons for the absence of an implementation plan and its impact.
- On Tuesday 15 November, representatives from NHS England and the National Institute for Health and Care Excellence provided evidence and shared views on the reasons for the absence of an implementation plan and its impact.

Alongside these events, Rare Disease UK ran a Call for Evidence between November 2016 and January 2017. Patients, family members, carers, patient representatives, clinicians, academics and industry representatives were also invited to submit evidence online via a survey or e-mail. Further information about the questions asked as part of the Call for Evidence can be found on the Genetic Alliance UK website.

In total, the APPG heard 317 responses from patients, family members, patient representatives, clinicians and industry representatives through the hearings and the Call for Evidence. 153 rare diseases were represented in those responses:

<table>
<thead>
<tr>
<th></th>
<th>Survey</th>
<th>E-mail</th>
<th>Hearing 1</th>
<th>Hearing 2</th>
<th>Hearing 3</th>
<th>TOTAL</th>
</tr>
</thead>
<tbody>
<tr>
<td>Patient</td>
<td>187</td>
<td>11</td>
<td>1</td>
<td>n/a</td>
<td>0</td>
<td>199</td>
</tr>
<tr>
<td>Family</td>
<td>89</td>
<td>2</td>
<td>0</td>
<td>n/a</td>
<td>0</td>
<td>91</td>
</tr>
<tr>
<td>Patient</td>
<td></td>
<td></td>
<td>6</td>
<td>4</td>
<td>3</td>
<td>18</td>
</tr>
<tr>
<td>representative</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Clinician</td>
<td></td>
<td>1</td>
<td>n/a</td>
<td>n/a</td>
<td>n/a</td>
<td>1</td>
</tr>
<tr>
<td>Industry</td>
<td></td>
<td></td>
<td>0</td>
<td>2</td>
<td>3</td>
<td>8</td>
</tr>
<tr>
<td>TOTAL</td>
<td>276</td>
<td>22</td>
<td>9</td>
<td>6</td>
<td>6</td>
<td>317</td>
</tr>
</tbody>
</table>

*Where patient representatives attended more than one hearing, their response has counted once.
ANNEX 2:
GLOSSARY OF TERMS

APPG
All Party Parliamentary Groups (APPGs) are informal, cross-party groups of MPs and Peers interested in a particular issue. APPGs do not have any power to make laws and are not funded by Parliament. There is a great number of APPGs, covering many and diverse fields such as health, education, transport, defence, finance, the media and sports.

APPG on Rare, Genetic and Undiagnosed Conditions
The APPG on Rare, Genetic and Undiagnosed Conditions held its inaugural meeting in February 2016. The aims of the group, agreed by members, are to increase awareness of rare, genetic and undiagnosed conditions in parliament and help ensure that patients and families affected by these conditions have access to appropriate care and support.

The APPG provides a useful forum for parliamentarians to meet patients and families, who are often vital to converting complicated and abstract debates to the everyday reality of people’s lives. It also provides the opportunities for MPs to learn about the many similar issues facing patients and their families. The APPG helps to give patients affected by rare, genetic and undiagnosed conditions a voice in parliament.

Arm’s length bodies of the Department of Health
Arm’s length bodies are partner organisations and agencies of a ministerial department. They help to develop and deliver policies and services with the ministerial department. The Department of Health has fifteen arm’s length bodies, including NHS England, the National Institute for Health and Care Excellence (NICE), Public Health England (PHE) and Health Education England (HEE).