



## Rare Disease UK response to Equity and Excellence: Liberating the NHS

### About Rare Disease UK

Rare Disease UK (RDUK) is the national alliance for people with rare diseases and all who support them. We have over 550 registered members including: over 130 patient organisations, clinicians, researchers, academics, industry and individuals with an interest in rare diseases.

**It is estimated that 1 in 17 people will be affected by a rare disease at some stage in their lives. This amounts to 3.5 million people across the UK. Collectively, rare diseases are not rare.**

RDUK was established by Genetic Alliance UK, the national charity of over 130 patient organisations supporting all those affected by genetic conditions, in conjunction with other key stakeholders in November 2008 following the European Commission's *Communication on Rare Diseases: Europe's Challenges*.

Subsequently, RDUK successfully campaigned for the adoption of the Council of the European Union's *Recommendation on an action in the field of rare diseases*. The Recommendation was adopted unanimously by each Member State of the EU (including the UK) in June 2009. The Recommendation calls on Member States to adopt plans or strategies for rare diseases by 2013.

RDUK is campaigning for a strategy for integrated service delivery for rare diseases<sup>1</sup>. This would coordinate:

- research
- prevention and diagnosis
- treatment and care
- information
- commissioning and planning

into one cohesive strategy for all patients affected by rare disease in the UK. As well as securing better outcomes for patients, a strategy would enable the most effective use of NHS resources.

RDUK calls for a National Clinical Director for Rare Diseases as recommended by the Chief Medical Officer in his 2009 Annual Report (March 2010) to ensure the implementation in practice of this strategy.

### Overview

RDUK aims to unite the rare disease community to work with the Department and to inform an effective strategy for rare diseases. We welcome the opportunity to comment on the White Paper.

We wish to show how a strategy for rare diseases would ensure that the vision set out in the White Paper also applies to patients and families with rare diseases, who are frequently forced to accept an inequitable service as they have the misfortune of their condition being rare.

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<sup>1</sup> A rare disease is defined as a condition affecting less than 5 in 10,000 of the general population.

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#### Rare Disease UK

Unit 4D, Leroy House, 436 Essex Road,  
London, N1 3QP

[www.raredisease.org.uk](http://www.raredisease.org.uk)

T: +44 (0)20 7704 3141 | F: +44 (0)20 7359 1447

E: [info@raredisease.org.uk](mailto:info@raredisease.org.uk)

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RDUK would like to emphasise that the scale and speed of the changes proposed by the White Paper could pose a threat to patients with rare diseases and their families unless there is a strong focus on safeguarding and improving NHS services for these patients. We believe that a strategy for rare diseases is essential to provide this focus.

## 1. Liberating the NHS

A strategy for rare diseases will help to ensure the most efficient use of NHS resources whilst providing better outcomes for patients. Patients with rare diseases are often already heavy consumers of NHS resources, but services are too frequently inefficient or poorly coordinated, and patients struggle to access the care, support and treatments they need. Not only does this lead to a reduction in patients' quality of life and potentially their life expectancy, it also creates waste through delays in diagnosis, duplication of services, unnecessary interventions, repeat visits to hospital, increased emergency admissions due to poor management of the condition, etc. Yet there are many examples of good practice in England and across the UK demonstrating that it is possible to provide innovative, cost-effective, high-quality services to patients with rare diseases.

A strategy for rare diseases should be seen not only as compatible, but a key element of ensuring that the vision of "fairness for everyone in society" and "equality" set out in the White Paper, also applies to those affected by rare diseases and also "cut waste" and to promote increased "efficiency and quality".

### Our values

We welcome the values set out in this section so long as active measures are taken to ensure that patients and families with rare diseases realise these values. A strategy for rare diseases would help ensure this.

There must be recognition that some (but by no means the majority) of patients with rare diseases require complex/expensive interventions if they are to achieve equality of outcomes from the health service.

### The NHS today

We agree that there are many examples of world-class service delivery within the NHS. What works well should be built upon and used as models for other services as opposed to "reinventing the wheel". RDUK have collected many examples of good practice which we will be highlighting in a report which will be presented to the Department on Rare Disease Day next year (28<sup>th</sup> February).

We also strongly support the need for a patient-centred approach, not unnecessarily restricted by overly rigid service or territorial boundaries. If a service exists that would benefit the patient they should not, without good reason be prevented from being able to access it. One of our members summarised well the problems many people with rare diseases face:

"Being a rare condition there are no support or therapy services I can access locally...there is an excellent MS therapy centre nearby but only for MS patients, ditto with other conditions' therapy centres even though the symptoms and treatment would be ideal to my own condition. Being able to access these, and to have a determined care plan would be an immense improvement for me"

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Many rare diseases affect multiple systems of the body and as a result, effective integration and coordination of care is crucial. Much of the care can and should rightly be provided at a local level, but patients must also be able to access services on a regional or national basis. One solution to this is to ensure that patients with rare diseases are all appointed with a care coordinator.

RDUK's multi-stakeholder Working Group on Delivering Coordinated Care has come up with a number of recommendations as to how a strategy for rare diseases can improve integration of services and ensure patient-centred care.

The initial recommendations are available at:  
[http://www.raredisease.org.uk/documents/consultation\\_rduk.pdf](http://www.raredisease.org.uk/documents/consultation_rduk.pdf)

## 2. Putting patients and the public first

### Shared decision-making: nothing about me without me

RDUK strongly supports the need for involving patients fully in their own care, with decisions made in partnership with clinicians. This is especially true of patients and families with rare diseases; many are *the experts* in their condition in the absence of scientific or clinical understanding of their condition.

### An NHS information revolution

We strongly support that "information, combined with the right support, is the key to better care, better outcomes and reduced cost". Unfortunately, there is a lack of information on most rare diseases available both to patients and healthcare professionals or there is a lack of awareness about where to find information when available.

One member summarised the situation experienced by many, both with very rare and less rare conditions:

"Our son's condition is ultra rare so you HAVE to educate yourself on the topic, no doctor has time/inclination to research orphan syndromes for you."

RDUK's multi-stakeholder Working Group on Patient Care, Information and Support has come up with a number of recommendations as to how a strategy for rare diseases can improve the availability of information, both to patients and families and health professionals, including how information currently available can be better signposted and utilised.

The initial recommendations are available at:  
[http://www.raredisease.org.uk/documents/consultation\\_rduk.pdf](http://www.raredisease.org.uk/documents/consultation_rduk.pdf)

We strongly support patient held records; a recommendation of the Working Group. There is often a lack of communication between the different health professionals involved in the care of rare disease patients and patients' information is not always passed on when necessary. This leads to patients having to explain their condition multiple times to each professional involved in their care. Failure to pass on up-to-date information can result in inefficient care and in a waste of NHS resources and patients' time. This may be the result of human inefficiency, or may arise due to inappropriate protocols

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for the sharing of information. Either way, patient-held records would ensure that this information is available and also empowers the patient or their family.

The NHS is almost uniquely able to support clinical research as it is a single healthcare system for more than 50 million people. Central collection of data can facilitate research and development, and is essential for patients with rare genetic diseases if there is to be the possibility of creating a strategic response to their needs. In particular we recommend advance planning for the timely implementation of the International Classification of Diseases (ICD) 11 in 2015, which makes a greater granularity in rare disease classification possible. This will provide a crucial opportunity for capturing data on the incidence and natural history of rare diseases that are currently poorly understood. According to the Orphanet database, of the thousands of known rare diseases for which a clinical identification is possible, only 250 of them have a code in the current ICD 10.

### Increased choice and control

Due to the scarcity of expertise and knowledge and fewer (if any) effective treatments available, patients with rare diseases may face far more limited choice than those with common conditions. However, we welcome the commitments to:

- Introduce choice for diagnostic testing.
- Introduce choice in care for long-term conditions.
- Support preferences about how to have a good death.
- Give more information on research studies.

Little detail is given about these points in the White Paper and we welcome more detail on how these commitments will be achieved in practice. A strategy for rare diseases would help ensure that these choices do in fact extend to patients with rare diseases.

If choice of treatment is ever truly to become a reality for patients with rare diseases, further efforts need to be made to break down the barriers restricting R&D of therapies for rare diseases and more measures need to be taken to encourage the development and uptake of these therapies.

RDUK's multi-stakeholder Working Group on the Coordination of Research has come up with a number of recommendations as to how a strategy for rare diseases can facilitate R&D for rare diseases.

The initial recommendations are available at:

[http://www.raredisease.org.uk/documents/consultation\\_rduk.pdf](http://www.raredisease.org.uk/documents/consultation_rduk.pdf)

### 3. Improving healthcare outcomes

#### Developing and implementing quality standards

We are concerned that rare diseases will be excluded from the process of the NICE quality standards. With 150 standards to produce in a short amount of time, with topic selection based primarily on whether or not NICE feels that it has sufficient existing evidence, it seems highly unlikely that rare diseases will be included. Rare diseases in particular would benefit from care pathways to support the health professionals involved in caring for patients with rare diseases (many of which may not have any

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knowledge of that condition). These will enable a proactive approach to care, and may help to reduce unnecessary unplanned hospital admissions by enabling forward planning. The development of such pathways should form a key component of a strategy for rare diseases.

## Research

We welcome the recognition of the importance of research.

### 4. Autonomy, accountability and democratic legitimacy

Robust commissioning structures enabling the effective service delivery for patients with rare diseases should form a key element of a strategy for rare diseases. RDUK has responded to the proposals on commissioning structures in the “Commissioning for Patients” paper.

RDUK’s multi-stakeholder Working Group on Commissioning and Planning has come up with a number of principles that should underlie commissioning structures for rare diseases.

The initial recommendations are available at:

[http://www.raredisease.org.uk/documents/consultation\\_rduk.pdf](http://www.raredisease.org.uk/documents/consultation_rduk.pdf)

RDUK would like to reinforce here:

- Localism is not always the best means to understand the health needs of the patients we represent.
- Our strong support for the proposal that the National Commissioning Board be responsible for commissioning national and regional specialised services.
- An effective commissioning structure for rare diseases needs to ensure good communication and coordination between those commissioning services at the local level and those commissioning specialised services.
- The budget for specialist services that are currently commissioned must be allocated directly to the National Commissioning Board specifically for specialist services. This budget must take into account that, as the Specialised Commissioning Groups do not currently commission all the services in the Specialised Services National Definitions Set, their expenditure may not adequately reflect the budget that the National Commissioning Board will need to provide an equitable service across England. It must also be flexible to allow for the commissioning of new services at this level.

### Establishing the board and managing the transition

Care must be taken to ensure that those with expertise in commissioning specialised services are retained during the transition period and that an arbitrary approach to cutting “bureaucracy” is not taken, resulting in the loss of necessary expertise.

## Conclusion

Whilst RDUK welcomes many of the values outlined in the White Paper, and some of the developments have the potential to improve services for patients with rare diseases, a restructure of this scale and speed, set against a backdrop of cost-cutting and efficiency savings does also pose a great threat to services for patients with rare diseases. There are over 6000 rare diseases, most of which affect very

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small numbers of people; it is easy for patients with rare diseases to “slip through the net”. However, collectively it is estimated that 1 in 17 people will be affected by a rare disease at some stage in their lives. This amounts to 3.5 million people across the UK.

A strategy for rare diseases becomes even more necessary in this context. This is in order to ensure the efficient use of NHS resources and to provide the policy focus to protect and build upon high-quality services currently available to ensure that the vision of the White Paper applies equitably for patients with rare diseases who frequently have had to settle for an inadequate service as they have the misfortune that their condition is rare.

## Rare Disease UK – October 2010

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