



## Quality 2020: A 10-year quality strategy for health and social care in Northern Ireland

– A response from Rare Disease UK and the Northern Ireland Rare Disease Partnership.

### About us

It is estimated that 1 in 17 people will be affected by a rare disease<sup>1</sup> at some stage in their lives. This amounts to 100,000 people in Northern Ireland. Collectively, rare diseases are not rare.

Rare Disease UK (RDUK) is the UK alliance of people with rare diseases and all who support them. We have over 800 members including over 140 patient organisations, clinicians, healthcare professionals, professional bodies, researchers, academics, industry and individuals with an interest in rare diseases.

RDUK is an initiative of Genetic Alliance UK, the national charity of over 140 patient organisations supporting all those affected by genetic conditions, in conjunction with other key stakeholders.

RDUK is supported by an unrestricted educational grant from the Association of the British Pharmaceutical Industry's (ABPI) Orphan Medicines Industry Group and the Orphan Medicines Industry Group Partnership representing companies outside of the ABPI.

RDUK aims to work with policy makers and Health and Social Care to inform and aid the development and implementation of effective strategies for rare diseases in Northern Ireland and across the UK in accordance with the Council of the European Union's Recommendation on an action in the field of rare diseases. The Recommendation which calls for the development of plans or strategies for rare diseases by 2013 was adopted unanimously by each of the EU's Member States in June 2009.

A strategy for rare diseases should coordinate:

- Research
- Prevention, diagnosis and screening
- Commissioning and planning
- Access to treatment
- Multidisciplinary Care
- Access to information and support

As well as securing better outcomes for patients, a strategy for rare diseases would enable the most effective use of Health and Social Care resources.

This response has been drawn up together with the newly formed **Northern Ireland Rare Disease Partnership**. The partnership has been formed to give a stronger voice to patients and families affected

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

### 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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RARE DISEASE | UK

The National Alliance for people with rare diseases & all who support them

by rare diseases in Northern Ireland. The Partnership brings together rare disease patient organisations and individuals affected by rare disease to work collaboratively with key stakeholders.

RDUK published a report outlining comprehensive recommendations for a strategy for rare diseases in February 2011. The report, *Improving Lives, Optimising Resources: A Vision for the UK Rare Disease Strategy* is available here: [www.raredisease.org.uk/documents/RD-UK-Strategy-Report.pdf](http://www.raredisease.org.uk/documents/RD-UK-Strategy-Report.pdf)

The Northern Ireland Rare Disease Partnership supports the recommendations outlined in RDUK's report and believes they should form the basis of an effective strategy for rare diseases in Northern Ireland.

RDUK and the Northern Ireland Rare Disease Partnership welcome the opportunity to respond to this consultation.

### Rare Disease UK and the Northern Ireland Rare Disease Partnership – April 2011

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## Overview

RDUK and the Northern Ireland Rare Disease Partnership welcome the objectives set out in the consultation document. We are, however, concerned that without a strong focus on improving the quality of services for patients with rare diseases, they will continue to “fall through the net”. A strategy for rare diseases should be viewed as an integral part of ensuring that the vision outlined in the consultation document also applies to patients affected by rare diseases. This is particularly important as collectively, rare diseases affect 100,000 people (1 in 17 of the population) in Northern Ireland; a quality service must meet the needs of this substantial cohort of patients. Moreover, patients with rare diseases already make heavy demands on Health and Social Care resources, but these resources are often used inefficiently due to delays in diagnosis, misdiagnosis, fragmented care, a lack of information, few guidelines on the effective management of the condition and limited effective treatment options.

We also emphasise that success will depend on establishing effective implementation systems. Collaboration with service users is crucial in redesigning the delivery systems, and not just in measuring improvements.

### 1. Are the values, principles and assumptions underpinning this strategy appropriate?

We support the principles, values and assumptions underpinning the Quality Strategy. These align closely with RDUK’s recommendations for a strategy for rare diseases. We would particularly support the design principles that a high quality service should:

- Be holistic in nature
- Focus on the needs of individuals, families and communities
- Be accessible, responsive, integrated, flexible and innovative
- Surmount real and perceived boundaries
- Be informed by the active involvement of individuals, families and communities

A strategy for rare diseases would ensure that these principles are embedded in Health and Social Care in Northern Ireland. Recommendations as to how this can be achieved can be found in RDUK’s report *Improving Lives, Optimising Resources* (see ‘About us’).

We support the values identified. There are additional factors that need to be taken into account when considering some of these values in relation to rare diseases:

#### Empowerment

In order for patients to take greater responsibility for their own health and social well-being, they must be empowered with the information needed in order to understand and manage their condition effectively. However, good-quality information on rare diseases can often be scarce and difficult to find,

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or non-existent. RDUK's report, *Experiences of Rare Diseases: An Insight from Patients and Families*<sup>2</sup> found that 52% of patients are not given sufficient information on their condition on diagnosis and that 37% of patients do not have anyone they can approach with questions about their condition.

RDUK's *Improving Lives, Optimising Resources* report outlines a number of ways to empower patients with more information about their condition including, information prescriptions, follow-up appointments with a health professional, an information portal and a designated care co-ordinator. Patient organisations (where they exist) are often vital sources of information for patients and sources of information such as these should be utilised by Health and Social Care.

## Involvement

We fully support the involvement of service users and carers in the design, delivery and review of services, however, active efforts must be made to facilitate this. Patients with rare diseases are often marginalised and isolated. There are no patient organisations representing the majority of patients with rare diseases in Northern Ireland. Where patient organisations do exist, they are often entirely volunteer-led. Responding to complex policy consultations which may have subtle long-term impacts for many rare disease patients is almost impossible for small representative bodies and for individuals. It must be possible to make strategy development and consultation processes user-friendly and publicly accessible and a concerted effort must be made to capture these voices to ensure that service design is not dictated by those who can "shout the loudest".

We welcome the Patient Client Council's focus on rare diseases as one of their priority areas in recognition of the need to support patients with rare diseases in decision-making.

## Respect

Patients affected by rare diseases often report feeling that they are not respected by health and social care professionals. Patients and families affected by rare diseases in many cases have to become the experts on their condition; their expertise must therefore be respected by health and social care professionals. The downside to this can be that patients have to educate all the professionals they come into contact with about their condition. This can be alleviated through better communication between professionals, education and information (see RDUK's recommendations in *Improving Lives, Optimising Resources*).

Patients report particular problems accessing social services. Due to a lack of awareness of most rare conditions there is no recognised "label" necessary for social services to be able to respond to a patient/family's needs.

## Partnership

We strongly support this value. Rare diseases often affect multiple systems of the body and the patient has to come into contact with a wide range of professionals across health and social care. Furthermore,

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<sup>2</sup> December 2010. Available at: <http://www.raredisease.org.uk/documents/RDUK-Family-Report.pdf>

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care needs to be accessed on many levels ranging from day to day services and care delivered at a local level to specialist care which a patient may need to access outside of Northern Ireland.

In order to effectively meet the needs of patients with rare diseases, the idea of partnership must be extended across national boundaries. No one nation can solve the needs of all rare disease patients alone. This is especially so in Northern Ireland, where the relatively small population means the maximum number of patients a specific rare disease can affect will be 850; the majority of rare diseases will affect far fewer than this, perhaps only one or two people in the country. Collaboration should be fostered with the rest of the UK, the Republic of Ireland and across Europe where appropriate.

## Excellence

There are many examples of excellent practice, delivering a high-quality, cost effective service to patients with rare diseases. These can be found in Northern Ireland and beyond and they should serve as models for future service development.

## Community

Although we recognise the importance of the community perspective and patients with rare diseases will rightly receive the majority of their day-to-day care in their local community, this must not be the only perspective. As we describe above, patients with rare diseases will often need to access care outside of their local area.

## Continuity

Coordination and integration of care is crucial for patients with rare diseases, most of which are multi-system requiring a range of professionals operating at different levels of the health and social care system. An integrated health and social care system in Northern Ireland provides significant opportunities, however, patients and professionals working with patients, continue to report difficulties due to bureaucratic hurdles created by, for example, different services coming from different pots of money. It can be extremely unclear who to approach for what service.

Continuity of services is crucial. When a service is proving to be effective it should not be constrained by short-term planning. Continuity of specialised services in Northern Ireland is a particular issue. Many services are reliant on one specialist. This places services under threat when a specialist retires, goes on maternity leave etc. This highlights the need for succession planning and for Northern Ireland to link into networks (see also 'Partnership').

We have identified transition as an area of particular concern with regards to continuity of services. This is relevant both in respect of transition from paediatric to adult services and from adult to elderly services. Our *Experiences of Rare Diseases: An Insight from Patients and Families* report identifies a number of medical, psychological, financial and social problems experienced at transition. Often patients and families feel like they "fall off a cliff" at transition as services that were previously available to them disappear and they have to deal with a new team of professionals with little or no knowledge of their condition. This is becoming increasingly problematic as medical advances see more patients with rare diseases living longer.

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### Value for Money

We support the recognition in the consultation document that ‘money is not the only determinant of high quality’ and that ‘low quality...can actually cost more’. As pointed out previously, patients with rare diseases are already heavy users of Health and Social Care resources, although this expenditure is often “hidden”. Resources are often used inefficiently due to delays in diagnosis, misdiagnosis, fragmented care, a lack of information, few guidelines on the effective management of the condition and limited effective treatment options. Failing to rapidly diagnose and effectively manage a rare disease in many instances result in more expensive interventions as a patient’s condition deteriorates.

### Equity

Equity necessitates that all patients should be able to access the high quality services needed for their condition. We believe that in order for Health and Social Care to provide an equitable service for *all* patients, including those with rare diseases, an effective strategy for rare diseases is essential in Northern Ireland.

#### **2. Are the vision and mission statements, as worded, appropriate for the purposes of this Quality Strategy?**

Whilst we support the ambitious vision, we would like to see a stronger emphasis on the involvement of patients and clients and their families in the design of services in order to achieve the strategic vision.

A focus on improved health and wellbeing for *all* must also include patients with rare diseases whose needs in the past have been overlooked.

#### **3. Are the strategy goals identified useful and meaningful? Have any important outcomes not been adequately covered by these goals?**

We support the strategic goals and believe they align closely with RDUK’s recommendations for a strategy for rare diseases.

#### **4. Are the detailed objectives for each goal useful and meaningful? Have any important issues not been adequately covered by these objectives?**

We support the detailed objectives for each goal. RDUK’s recommendations for a strategy for rare diseases align closely with these objectives and they can provide guidance as to how the strategic goals can be realised for patients with rare diseases in Northern Ireland.

#### **5. Are the delivery arrangements set out in the strategy meaningful and likely to be effective?**

We support the involvement of patients and service users as well as a wide range of stakeholders in the delivery arrangements. However, we would like to re-emphasise our point under Question 1; a concerted effort must be made to enable marginalised voices such as patients and families affected by rare diseases to meaningfully participate in the process.

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