



Living with Long Term Conditions – A Policy Framework

- 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 disease¹ 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 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.

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

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

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

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 – June 2011

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Content of the Policy Framework

Q1. Do you believe that implementation of this policy framework will help plan and develop more effective services to support people living with long term conditions and their carers?

A qualified yes.

We believe the systematic implementation of the long term policy framework would help to plan and develop more effective services to support people living with long term conditions and their carers. However, to ensure that the aims are realised for all people with chronic rare conditions and their carers, a strategy for rare diseases is necessary.

The maximum number of patients a single rare disease will affect in Northern Ireland will be around 850 people². In reality, the majority of rare diseases affect far fewer people, perhaps only a few, or even a single person in Northern Ireland. Due to the rarity of their condition, patients face a greater number of obstacles in comparison to those with more common long term conditions. Some of these obstacles include:

- a lack of information about their condition
- a lack of knowledge and understanding of their condition by health and social care professionals
- fewer, if any, appropriate sources of support
- a greater sense of isolation
- and fewer effective treatments and care pathways.

A targeted strategy for rare diseases is necessary to take account of the particular problems experienced by patients with rare diseases and their carers and to ensure effective and efficient use of resources. More general policy frameworks have, in the past, proven to be ineffective in meeting these needs and rare diseases often get overlooked as policy is geared towards common or well-known conditions.

Q2. Do you agree with the 6 principles underpinning the provision of quality care and support for people living with long term conditions and their carers?

Yes.

We agree with the six principles underpinning the provision of quality of care and support for people living with long term conditions and their carers. It is vital that each principle is not seen in isolation, but are taken together as a coordinated whole; for example, the first principle, working in partnership, needs to apply across each of the principles.

Q.3. Do you agree that implementation of these principles will result in improved care and support for adults in Northern Ireland living with long term conditions and their carers?

A qualified yes.

² Based on the maximum prevalence of a rare disease – 5 in 10,000 people.

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If these principles are systematically implemented, it would result in improved care and support. However, we believe a specific strategy for rare diseases is necessary to ensure the principles are realised for people living with chronic rare diseases.

The Health and Social Care Board and Local Commissioning Groups should report on how they are delivering the policy framework in practice and there should be an agreed set of outcomes to measure progress.

Chapter 1 – Working in Partnership

Q4. Should people with long term conditions, and where appropriate their carers, be recognised and involved as partners in how care is planned and delivered?

Yes.

It is imperative that people with long term conditions and their carers are recognised and involved as full partners in how care is planned and delivered. This is especially so in the case of rare diseases; in many instances the lack of expertise or knowledge of a condition means that patients or their carers have a greater knowledge of their condition than most health and social care professionals. This knowledge and expertise must be utilised to effectively plan and deliver care.

Q5. Should people with a long term condition have a personalised care plan, the content and detail of which reflects their own condition, needs and preferences for care and support?

Yes.

Patients with a long term condition should have a personalised care plan. This ties in with Recommendation 2.b of the Delivering Coordinated Care chapter of RDUK's recommendations for a strategy for rare diseases, which calls for a personal care plan for all patients with chronic rare conditions³.

Personalised care plans offer substantial benefits. Rare diseases often affect multiple systems of the body and, as a result, there are often many professionals across health and social care involved in a patient's care. Effective care plans play a key role in ensuring coordination of care by aiding communication between all of these professionals, ensuring that everyone involved in the care of a patient is aware of their role and how it fits into the wider care of the patient.

Patients with rare diseases often report feeling "lost in the system" due to fragmented care. A personalised care plan would prevent this by assuring a patient that there is a coherent care plan in place and allowing a patient to know which services and support they will be receiving and when. It also empowers a patient and/or their carers to take a more active role in the care they receive.

An effective care plan should be drawn up in partnership with patients and carers and regularly updated. It also needs to be drawn up in collaboration both with specialists and those providing more

³ See the introduction for more information about the report *Improving Lives, Optimising Resources: A Vision for the UK Rare Disease Strategy*

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day-to-day care locally. The care plan should set out drug, therapy and follow up regimes and ideally be linked to standard treatment guidelines for the condition.

The development of a care plan, and its implementation should be overseen by a designated care coordinator (see Recommendation 3b, Delivering Coordinated Care).

Q6. Do you agree that care and support for people with long term conditions and their carers can be improved through cooperation between statutory, voluntary and independent sector organisations?

Yes.

Care and support for people with long term conditions and their carers can be improved through cooperation between a range of organisations, especially patient organisations. Patient organisations are often the key (and sometimes only) source of information and support for patients with rare diseases. RDUK's survey of patients and families' experiences of rare diseases⁴ found that the relevant patient organisation was the main source of information for 52% of respondents. The expertise of patient organisations should be utilised in order to provide high quality care, however, many patient organisations for rare diseases are small or entirely volunteer-led and may need further resource from HSC to collaborate in the delivery of care.

One example of good practice is the collaboration between the Society for Mucopolysaccharide Diseases (MPS Society) and the Primary Immunodeficiency Association, in partnership with the Belfast Trust to provide an All-Ireland Advocacy Support Worker to provide support to patients and families affected by these rare conditions across Ireland.

Chapter 2 – Supporting Self Management

Q7. Should people be supported to develop their knowledge and skills to self manage their long term condition?

Yes.

People should be supported to develop their knowledge and skills to self manage their long term condition. We welcome the recognition that a better understanding of the condition and how to manage it effectively can result in improvements in the health, wellbeing and quality of life of the patient as well as providing better value for money for the health service.

Patients should be empowered to manage their condition effectively and information is crucial in this respect. However, good quality information on rare diseases can be hard to come by and health-professionals often struggle to support patients with their information needs.

We welcome the recognition of the role of a care plan in aiding self-management. A designated care coordinator would also help patients to manage their condition more effectively by providing a point of

⁴ *Experiences of Rare Diseases: An Insight from Patients and Families* (December 2010), available at: <http://www.rare-disease.org.uk/documents/RDUK-Family-Report.pdf>

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call for patients to ask questions about the management of their condition and a link between a patient and the health professionals involved in their care.

Q8. Should self management support be a core element of care pathways for long term conditions?

Yes.

Supporting self management should be a core element of care pathways so long as this approach is in fact tailored to the needs of the patient and the necessary support structures are in place.

In order for a patient to manage their condition effectively, there must be someone that they can approach with questions about their condition and their care. RDUK's survey of patients and families' experiences of rare diseases found that 37% of patients/carers do not have someone they can approach with questions about their condition. A designated care coordinator could provide this point of contact.

Chapter 3 – Information to Service Users and Carers

Q9. Do you agree that people with long term conditions, and their carers, should have access to information, education and training programmes in a format and medium that best suits their needs?

Yes.

Information and advice is essential in order to help patients and carers manage a condition effectively. Information is also crucial in terms of accessing the support and services needed. The consultation document correctly identifies that a variety of information is needed at different points in the trajectory of a condition.

Patients and carers often struggle to access good-quality information about their condition, how to manage it effectively and where to go for support. For many rare diseases good quality information is available but it may be difficult to find and health professionals may not know where to signpost their patients to. Patient organisations are often an important source of good quality information. For other rare diseases, especially when there is no relevant patient organisation, good quality information is scarce or non-existent. These gaps in information need to be addressed. We are concerned that the consultation document assumes that there is "a wealth of information available" for all long term conditions when this isn't the case for hundreds, if not thousands, of rare diseases.

A lack of information on diagnosis is especially problematic. RDUK has found that 52% of patients/carers feel that they are not given sufficient information on diagnosis. In the worst instances, patients are simply given the name of a condition and are then left to their own initiative to find information and sources of support.

'[We received a] phone call for diagnosis on New Year's Eve with the comment from GP 'I don't know anything about it go on a computer and look it up'. We didn't have a computer!!!'

- A relative of a patient with myasthenia gravis.

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‘We simply received the name of the condition, then Googled [it]’
- A relative/carer of a person with progressive supranuclear palsy⁵.

Testimonies such as this are not uncommon. To help improve this situation we strongly support the use of information prescriptions in line with Recommendation 1a (Patient Care, Information and Support) of RDUK’s recommendations for a strategy for rare diseases. We believe that each patient diagnosed with a rare disease in Northern Ireland should be offered an information prescription. Patients/carers should also be offered a face-to-face appointment with a relevant health professional at a designated time after diagnosis to discuss their information needs. We agree that this then needs to inform a personal care plan.

We welcome the recognition of the wide range of information needs; medical, social and financial. RDUK has identified:

- Only a third (33%) of patients/carers affected by rare diseases feel they receive sufficient support with their social needs
- Less than a third (29%) feel they receive sufficient psychological support
- Less than a quarter (24%) feel they receive adequate support with financial concerns

Improved information provision would help improve patient experience in these areas.

We support the need for information to be made available in a variety of media and formats to suit individual needs. Some patients will want more detailed information than others and it is important that information is tailored to the individual. A designated care coordinator can help patients/carers make sense of the information they receive as well as providing a point of contact to ask questions.

Q10. Should options for a central Health and Social Care information resource be explored?

Yes.

We strongly support the need for a central information resource. This is especially important for rare diseases where good quality information can be difficult to find. This corresponds to Recommendation 3a (Prevention and Diagnosis) of RDUK’s recommendations for a strategy for rare diseases, for a central portal to signpost user to good quality sources of information.

We have identified multiple benefits of such a resource:

To patients and carers

- Increasing accessibility of information on the condition, how it can be managed and treatment (where applicable) leading to greater empowerment, better management of the condition and better health outcomes.
- By providing information on sources of advice and support, reducing feelings of isolation.
- Information (often provided by patient organisations) has helped patients effectively guide their clinicians to a correct diagnosis. Information has supported patients and families to ask the right questions of clinicians and to seek the appropriate treatment or management of the condition once diagnosed.

⁵ Quotes taken from the *Experiences of Rare Diseases* report

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- Signposting patients to where they can find out about research being conducted. As there are no effective treatments for most rare diseases, information on new research is often particularly important to many families. RDUK has found that only 24% of patients feel they are given enough information on clinical trials being conducted and only 33% are given enough information on research in general.

To clinicians and care professionals

- It would help professionals fulfil their role as information providers by allowing them to direct patients and carers to good quality information more easily.
- Rare diseases often affect multiple parts of the body and require many health professionals to be involved in the care and treatment of a patient. Some of whom may know little, if anything, about a condition and may not know where to go for further information. A portal would help them access information both about the condition and where to go to for further information.
- Greater accessibility of information would facilitate the diagnostic process and make it easier to signpost patients to a relevant specialist once a diagnosis has been made.

A portal would also help identify where gaps in the availability of information exist. Efforts must be made to plug these gaps in information.

A central information resource for rare diseases could be developed in partnership with other health departments of the UK to avoid duplication of resources.

Chapter 4 – Managing Medicines

Q11. Should people with a long term condition have a personal medicines management plan?

Yes.

Medicines management should be included as part of a patient's care plan to ensure appropriate use and adherence to medicine regimes. Support should be available to patients to help them manage their medicines effectively.

Section 5 – Carers

Q12. Would the health and well-being of carers be improved by having access to information and services they need to support them in their caring role?

Yes.

We welcome the specific focus on the needs of carers as well as the needs of the person they're caring for. Caring for someone with a rare disease can be especially isolating as the person they are caring for may be one of few, or even the only person affected by that condition in Northern Ireland. In most instances there will be no specific support for carers of that condition in Northern Ireland due to the small patient numbers. As a result, efforts must be made to ensure carers of patients with rare diseases are adequately supported. Psychological support should be included as part of a carers care plan.

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Forums that bring carers of people affected by a particular disease together to discuss experiences and issues of common interest are often reported as being highly beneficial. Every effort should be made by Health and Social Care to support and facilitate the development of such services. This is especially important as these services are often provided by patient organisations, but as reported previously, a considerable proportion of rare diseases do not have relevant patient organisations in Northern Ireland.

The central portal for information discussed in Q.10 should also include details of support for carers.

Chapter 6 – Improving Care

Q13. Do you agree that the assessment of an individual's needs and their care plan should include consideration of their mental health and well-being?

Yes.

As reported previously, only 29% of patients/carers affected by rare diseases feel that they receive sufficient psychological support. RDUK's recommendations for a strategy for rare diseases include:

- The delivery of psychological and emotional support for both the patient and their family or carer should be considered from the outset of the patient's care and should be included as part of a patient's care plan. Those with more severe distress should be given access to counsellors, psychologists and psychiatrists. (Recommendation 3a, Patient Care, Information and Support).
- Patients should be made aware of the various sources of support available to them and encouraged to make use of this support. These sources should be included as part of an information prescription and there should be an advice service in all hospitals to direct people to sources of support. (Recommendation 3b, Patient Care, Information and Support).
- The provision of a designated care coordinator would also help to avoid feelings of isolation by providing a specific point of contact and support. (Recommendation 3c, Patient Care, Information and Support).

For many rare diseases, little can be done to alter the natural course of the condition. Peer-to-peer support through patient organisations can have a significant effect on patients and carers' mental health and well-being. Systematic support for patient organisations can be cost effective as well as meeting an otherwise unmet need in a user-friendly manner.

Q14. Should appropriate use be made of technology, adaptations and equipment to support people with long term conditions and their carers?

Yes.

Appropriate technology, adaptations and equipment can help to support patients with rare diseases live an independent life in some instances, or assist carers to effectively support the person they're caring for in others. As a result, patients and carers should be able to access the equipment and adaptations they need. Despite this, many patients/carers report having to battle through a complex maze of bureaucracy to get what they need. This is often aggravated by a lack of understanding of rare diseases; patients feel that as they do not have a recognisable "label" which results in providers not being able to respond to their needs.

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As discussed below in Q15 the majority of patients with rare diseases will need to access specialists, but a large proportion of their care day-to-day will be delivered in the community or locally. Technology could play a role in improving the interface both between specialists and a patient with a rare disease, but also in improving linkages between specialists and those involved more locally in providing care for a patient. For most rare diseases there are no specialists based in Northern Ireland. Technology such as video conferencing could remove the need for patients to have to travel as frequently to see specialists based on mainland UK and elsewhere. However, the use of such methods should be based on the preferences of the patient and their carers and not seen as an alternative to face-to-face appointments.

Steps should be taken to reduce, as much as possible, the time it takes for requests for support equipment to be fulfilled. The progression of some rare conditions, such as amyotrophic lateral sclerosis, is so quick that patients' needs have increased by the time requests are fulfilled, and the equipment is no longer adequate to provide support. For example, a request for hand rails to be installed can be fulfilled so late that they are no longer useful to the patient who by then is in need of a motorised wheel-chair.

Q15. Do you agree that a skilled, competent and confident workforce should be in place to enable a more community-based model of care to be provided?

Yes.

The majority of chronic rare diseases affect multiple systems of the body and there will be many health professionals involved in the care of a patient. Some of this care will be delivered by specialists at a tertiary level but the majority of day-to-day care will be delivered by primary or community care providers. Those who deliver care more locally often have little knowledge of rare diseases. They struggle to find sources of information and advice on how they can best discharge their caring duty effectively. This can lead to feelings of disempowerment among those providing this care locally.

We strongly support personalised care plans, drawn up holistically with all those involved in the care of a patient, overseen by a designated care coordinator as a way to ensure that care is rightly and most effectively provided as locally as possible. Improved information provision would also help to support those working in the community.

RDUK's recommendations for a strategy for rare diseases identifies a number of additional ways to help improve coordination of care between specialist and community setting including:

- Ongoing CPD/CME training in rare diseases (Recommendation 1b, Prevention and Diagnosis)
- Development of e-learning packages to increase overall awareness of rare diseases by qualified professionals (Recommendation 1d, Prevention and Diagnosis)
- Staff exchanges to share information and knowledge (Recommendation 2a, Prevention and Diagnosis)
- Hub and spoke models between specialised and local services (Recommendation 2c, Prevention and Diagnosis)
- There should be a named expert listed for each of the tests on the NHS Directory of Genetic Testing to act as a source of advice on that particular condition. This role of providing information should be included in the clinician's job plan. (Recommendation 3d, Prevention and Diagnosis)

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- An online portal for information about rare diseases (see response to Q10.)
- Ensuring linkages between what is commissioned centrally and what is commissioned at a local level (Recommendation 2, Commissioning and Planning)
- Regular meetings between all the professionals involved in the care of a patient (Recommendation 2c, Delivering Coordinated Care)
- Patient held medical records (Recommendation 2f, Delivering Coordinated Care)

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