



RARE DISEASE | UK

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

Response to Future Forum 2: how information can be improved.

About Rare Disease UK

Rare Disease UK (RDUK) is the national alliance for people with rare diseases and all who support them. We have nearly 1000 registered members including over 170 patient organisations, clinicians, researchers, academics, industry and individuals with an interest in rare diseases. RDUK was established by Genetic Alliance UK, the national charity of over 140 patient organisations supporting all those affected by genetic conditions.

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.

The Council of the European Union's Recommendation on an action in the field of rare diseases 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. A key priority of an effective rare disease plan should be to improve the provision of information to patients, families and health professionals.

RDUK has developed comprehensive recommendations to inform a rare disease plan in the UK. These recommendations were developed over the period of a year and a half in consultation with over 1000 stakeholders. These recommendations can be found in our report, 'Improving Lives, Optimising Resources: A Vision for the UK Rare Disease Strategy' (available at: <http://www.raredisease.org.uk/documents/RD-UK-Strategy-Report.pdf>). All of our recommendations below are key components of what RDUK would like to see in an effective plan for rare diseases.

Where relevant, we have answered both the questions aimed at patients and those aimed at professionals.

How can we ensure information is available that enables people to take more control of their own care and enable shared decision-making?

What kind of information would help you to take more control and have a bigger say?

Patient held records

RDUK advocates that patients should be given the option of holding their own medical records. As well as ensuring that the information is accurate and up-to-date and that health professionals have access to the most up-to-date records, it would also empower patients and encourage them to take more control over their care.

Rare Disease UK

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

www.raredisease.org.uk

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

E: info@raredisease.org.uk

An initiative of



Genetic Alliance UK
Supporting. Campaigning. Uniting.

A charity registered in England and Wales (no. 1114195)
in Scotland (no. SC039299).

A company Limited by Guarantee (Number 05772999)



Personalised care plans

Every patient with a rare chronic disease should have a personalised and regularly updated care plan which is drawn up in partnership with the patient and their carers (where appropriate). Having a care plan in place would prevent patients feeling “lost in the system” as they would be assured that there is a plan detailing what services and support they should be receiving and when. This would help empower patients to enforce their right to the high quality care they should be receiving and to take a greater role in planning their care.

Care coordinators

All patients with a rare disease should have a designated care coordinator. Care coordinators offer multiple benefits, including providing an important source of ongoing information and support to enable patients to take greater control in managing their own care effectively.

How can we ensure that information supports improved care and better integration of services (for example, commissioning, research, clinical audit, public health) whilst protecting patient confidentiality?

Systematic collection and use of accurate relevant information is crucial to support commissioning at different levels. Registries are a valuable tool in gathering a critical mass of clinical and other data about the natural history of rare diseases and the effect of any interventions in addressing symptoms and/or other aspects of the condition important to patients and families.

This information provides the opportunity to stimulate research and facilitates planning, allowing for sustainable services and the opportunity for the provision of appropriate Continued Medical Education/Continued Professional Development courses to be developed in ways that will appeal to clinicians and care professionals who are not specialised in the particular disease already.

Timely implementation of the International Classification of Diseases (ICD) 11, which offers greater granularity in rare disease classification, will provide opportunities for capturing data on the incidence and natural history of rare diseases.

Data collection and analysis requires resources and the political will to insist on it happening. Patients with rare diseases are often heavy users of NHS resources but without systematic gathering of good quality data, these resources are not being used in the most efficient way to achieve the best outcomes.

How can we open up access to information and support people to use it? What help do you need to make best use of the information out there?

Portal to information on rare diseases

In order to make better use of information in the most cost-effective way, consideration should be given to the utilisation and accessibility of good quality information that already exists. There is a wealth

Rare Disease UK

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

www.raredisease.org.uk

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

E: info@raredisease.org.uk

An initiative of



Genetic Alliance UK
Supporting. Campaigning. Uniting.

A charity registered in England and Wales (no. 1114195)
in Scotland (no. SC039299).

A company Limited by Guarantee (Number 05772999)



of good quality information on some (but not all) rare diseases from a range of different sources. There is an issue, however, in that all of those who would benefit from that information do not know where to go to access this information. RDUK recommends that a rare disease internet portal is developed by the Department of Health to provide one easily accessible point to signpost users to good quality information about rare diseases. A portal should be used by both patients/carers and professionals.

Information prescriptions

An information prescription should be given to all patients when they are first diagnosed with a rare condition. The prescription should outline the information patients and families have identified as important, and define the way they should receive it, when they should receive it and from whom.

Potential headings within the prescription identified by RDUK include:

- medical information including, where appropriate, possible treatment options;
- when the patients will receive their care plan;
- information needed by carers and family;
- social information;
- financial information;
- educational information;
- welfare information;
- palliative care information;
- how to access information on research;
- any other relevant information or sources of further information.

The guidance offered in the information prescription should be strengthened by a face-to-face information appointment with a relevant healthcare professional at a designated time after diagnosis according to the nature of the condition.

PALS

Patient Advice and Liaison Services (PALS) should be improved to reach its full potential to open up access to information for patients with rare diseases. Whether or not PALS exist in their current form following the NHS reorganisation, there is still a need for help, advice and information services to be located within hospitals. There is a need for these services to be given the most relevant and up to date resources so that they are in a strong position to offer high quality information to patients and families. This should include resources such as an up-to-date 'Contact a Family directory - The essential guide to medical conditions, disabilities and support' (which is also available as a web-resource: <http://www.cafamily.org.uk/>). The basic infrastructure of this service is already in place, and as such could be easily developed and improved to make it more effective, without incurring a substantial cost. Ideally, these services should provide a designated computer and a knowledgeable assistant to help patients and families research their condition. The assistant would be able to give guidance on relevant, reliable websites, and be on hand to assist those who are not computer-literate. Access to these services should be open to anyone at any time and not limited to a time, for instance, when a patient is attending a clinic.

Rare Disease UK

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

www.raredisease.org.uk

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

E: info@raredisease.org.uk

An initiative of



Genetic Alliance UK
Supporting. Campaigning. Uniting.

A charity registered in England and Wales (no. 1114195)
in Scotland (no. SC039299).

A company Limited by Guarantee (Number 05772999)



Care coordinators

Having a knowledgeable point of contact for information would help alleviate the stress and anxiety experienced by many patients with rare diseases. RDUK found that 37% of patients with rare diseases do not have anyone they can approach with questions about their condition¹. A solution to this is providing a designated care-coordinator for people with rare diseases. A care coordinator provides a point of contact for patients, for queries about both their condition and their care. Care coordinators also act as a vital source of general support to the patient and carers. Alongside a patient-centred care plan, they provide a vital component in encouraging better management of a condition and shared decision making. A further benefit of a care coordinator is that they can create much-needed linkages to support the flow of information between health and social services, which is often hindered due to institutional barriers.

What information about health and care do you need – and in what form?

The information needed will vary depending on the preferences of patients and their family and information needs will vary at different ages and stages of disease progression. Information should be available to suit a range of different levels of understanding, in a variety of formats. Patients and family members should be supported by health professionals to access this information. Equally, professionals should be able to easily access good quality information and our previous recommendation for a portal for rare disease information can assist in this regard.

When RDUK asked patients and families what aspects of their condition they would like more information on responses included:

- the clinical management of the condition;
- disease progression and long-term prognosis;
- the cause of the condition;
- current research projects;
- treatment options;
- advice on how to explain the condition to schools, A&E staff and other medical professionals, and family members;
- information on non-medical issues including about access to social, psychological and financial support.²

How should services communicate with you? How do you want to communicate with them?

In our experience, face-to-face contact is generally the preferred option of patients. However, for many rare diseases there may be very few people with expertise in the condition and these experts may be

¹ 'Experiences of Rare Diseases: An Insight from Patients and Families' (December 2010, available at: <http://www.raredisease.org.uk/documents/RDUK-Family-Report.pdf>).

² As above.

Rare Disease UK

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

www.raredisease.org.uk

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

E: info@raredisease.org.uk

An initiative of



Genetic Alliance UK
Supporting. Campaigning. Uniting.

A charity registered in England and Wales (no. 1114195)
in Scotland (no. SC039299).

A company Limited by Guarantee (Number 05772999)



located at a considerable distance from where the patient lives. Although generally patients with rare diseases are willing to travel, so long as they can access the expertise they require, at times this can place a considerable burden on the patient, their family or their ability to hold paid employment. This is especially so as the multi-factoral nature of many rare diseases, coupled with poor coordination of care, leads to multiple visits to different professionals, in different locations at different times. As a result there may be instances when a patient may prefer to contact someone knowledgeable about their condition online or by phone. However, this option should be the choice of the patient/carer and should not be seen as a replacement for face-to-face contact.

What works well already? What doesn't work?

Information when diagnosed

RDUK has identified the information given at the time of diagnosis as a significant problem area. Our research highlights that 52% of patients/carers do not feel that they are provided with sufficient information about all aspects of their condition following diagnosis of a rare disease³. Too often we hear from patients who are given no information at all about their condition and are left to their own initiative to find information, despite the fact that good quality information on rare diseases is not always easy to find without guidance.

Nearly two-thirds (64%) of patients were not given details of the relevant patient support group at the time of diagnosis⁴. Due to the scarce amount of widely available information on rare diseases, patient organisations (where they exist) can be a huge source of information for patients and families so it is vital that people are made aware of them as soon as possible. RDUK's research indicated that the relevant patient organisation is the main source of information to 52% of patients⁵.

Avoiding unnecessary obstacles to information

As elaborated earlier, patient organisations provide a vital source of information on rare diseases, both to patients and to clinicians. Rare disease patient organisations are often small or entirely volunteer led and generally lack the capacity to meet the requirements of the Department of Health Information Standard. The result is that much needed good quality information is barred from being included on important gateways to information such as NHS Evidence. RDUK acknowledges the need to accredit evidence, but there is also a need to balance this proportionally to the need for information and the ability of organisations to comply with the accreditation process. The rare disease portal proposed earlier is one way of gathering sources of good quality information and would allow the information to be seen and reviewed by the professional community. This allows both for the checking of accuracy of content and for professionals to learn about what patients find important and helpful.

RDUK has heard numerous examples of NHS Trusts blocking the use of information produced by patient organisations and other reputable sources due to unnecessary bureaucratic requirements, e.g. if the information does not carry the Trust's own logo. We have also heard from health professionals who

³ As above

⁴ As above

⁵ As Above

Rare Disease UK

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

www.raredisease.org.uk

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

E: info@raredisease.org.uk

An initiative of



Genetic Alliance UK
Supporting. Campaigning. Uniting.

A charity registered in England and Wales (no. 1114195)
in Scotland (no. SC039299).

A company Limited by Guarantee (Number 05772999)



are blocked from accessing and disseminating information by hospital firewalls. If a health professional thinks that a source of information would be useful to a patient or their family, NHS healthcare providers should work alongside them to make this information accessible.

Information on research

In the absence of effective treatments or cures for most rare diseases, research represents hope for many patients and families, if not for themselves, then the hope that the disease may be cured or prevented in future. As a result, there is often a strong desire among many patients affected by rare diseases to be informed about research and ways in which they can participate.

RDUK has found that only 24% of patients/family members feel they are given sufficient information about clinical trials and only a third (33%) are given sufficient information about research in general⁶.

As a result of the Deciphering Developmental Disorders (DDD) (<http://www.ddduk.org/>) study, each of the 17 regional genetics centres in England has a research nurse in place or is in the process of recruiting. The majority of these have been funded through the Comprehensive Local Research Networks. RDUK hopes that the appointment of these nurses will increase recruitment to all genetic studies and can provide a model of best practice for all rare diseases.

Care co-ordinators can also support the provision of information about research to patients. These should serve as examples of good practice for other services.

Rare Disease UK – October 2011

⁶ As above

Rare Disease UK

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

www.raredisease.org.uk

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

E: info@raredisease.org.uk

An initiative of



Genetic Alliance UK
Supporting. Campaigning. Uniting.

A charity registered in England and Wales (no. 1114195)
in Scotland (no. SC039299).

A company Limited by Guarantee (Number 05772999)