



## Response to Future Forum 2: education and training

### 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 raise professional awareness of rare diseases through education and training.

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' (February 2011, 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.

### What aspects of educating and training the health workforce needs improving? In particular, what are the skills and behaviour that need more development? How should these improvements be made?

Broadly, education and training about rare diseases can be improved by ensuring that resources are available to develop training courses; that professionals are aware of, and can access these opportunities; and that frameworks are put in place to evaluate whether training is fit for purpose.

There is a need to increase health professionals' knowledge and awareness of rare diseases. The lack of awareness and identification of rare diseases among health professionals often results in delays in diagnosis or misdiagnosis of rare disease patients.

---

#### 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)

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)



RDUK's research indicates that nearly half of patients (46%) with a rare disease wait over a year for a final diagnosis following the onset of disease symptoms. Of this, one in five (20%) wait over five years, and over one in ten (12%) wait over 10 years. Nearly half (46%) of patients with a rare disease are given an incorrect diagnosis before receiving their final diagnosis. Almost a third (30%) receive three or more misdiagnoses<sup>1</sup>.

Delays in diagnosis have multiple consequences including delays in accessing appropriate treatments or effective management of the condition as well as an inefficient use of NHS resources due to multiple avoidable appointments, diagnostic tests and interventions. A delay in diagnosis can cause a reduction in the patient's life expectancy and quality of life, while a misdiagnosis may result in a patient being managed for a condition – often a more common condition – that they do not have. Delays in diagnosis can result in missed opportunities for intervention, allowing conditions to become progressively worse and more difficult – sometimes impossible to treat. Early diagnosis, even for untreatable conditions, can also provide important information to guide future reproductive choices for the family.

Delays in diagnosis can be minimised by appropriate education, training and information resources to develop health professionals' skills to make appropriate referrals and to inform when to consider that a patient may have a rare disease. It is clearly not possible for all health professionals to know about each of the over 6,000 known rare diseases, however, there is a need to change the mindset that a health professional will never come across patients with rare diseases. It is estimated that 3.5 million people (1 in 17 people) in the UK will be affected by a rare disease at some point in their lives; most health professionals will come across patients with rare diseases on a regular basis.

Recommendations aimed at improving education and training that RDUK believe should underpin a plan for rare diseases include:

- inclusion of a module in the curriculum for medical students that covers general principles involved in diagnosing a patient with a rare disease;
- ongoing training through Continued Professional Development (CDP)/Continuing Medical Education (CME) modules;
- training of secondary care professionals in basic genetics;
- development of e-learning packages;
- ensuring that those who currently provide educational services to other health professionals have this time included as a formal part of their job plan.

How can we ensure that education and training in the new system is flexible and fit-for-purpose for the new way that care is delivered and enables training beyond the job, for example stimulating a culture of continuing professional development or academic and research development?

---

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

## 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)

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)



## Challenges imposed by financial pressures

RDUK is concerned that due to financial pressures, cuts will be made to education and training budgets and that health professionals will be limited to take only mandatory training. CPD and CME should be rightly recognised as an essential component in delivering good quality care and promoting innovative ways of working throughout the NHS. As a result, funding and resources for CPD should be ring fenced and available to all staff across all grades.

## Challenges imposed by developments in genomic medicine and technology

Advances in genomic medicine and technology offer significant opportunities in the field of rare diseases as well as common diseases. Equally these developments will also pose a significant number of challenges in terms of education and training of the workforce. Education planning tends naturally to focus on the development of new staff coming into a profession. In this situation, however, the development of the existing workforce too must be a priority. If it is not prepared, the potential contribution of genomic medicine to clinical care cannot be fully realised. There is a need to look critically at the existing infrastructure for doing this in a climate where providers and deliverers of service come under financial pressures. The Human Genome Strategy Group has been looking to develop a new framework for engagement and education in genetics and genomic medicine and RDUK believes that its forthcoming report should inform future provision of education and training in this area.

## How do we best ensure an effective partnership with health, education and research at a local level?

Building effective partnerships across boundaries and between all relevant stakeholders should be a key part of the process by which service providers are evaluated.

There should be the recognition that there is unlikely to be knowledge and expertise on rare diseases at a local level. Clinicians and health professionals in local services must be able to recognise the limitations of their own knowledge and that they need actively to seek help and guidance from specialised centres and experts. Equally the structures must be in place to enable the flow of information from experts to those working with patients locally. Ways this could be achieved include:

- hub and spoke service model of service delivery where specialised centres feed information to local services;
- staff from specialised services who perform an educational role, should have this included formally as part of their job plan;
- listed against each condition in the NHS Directory of Genetic Testing should be a named clinician who can act as a source of advice and information for that particular condition. This should be included as part of that clinician's job plan;
- staff exchanges, whereby staff from specialist centres undertake outreach work and educate staff at local services to share up-to-date and relevant information on best practice care.

RDUK advocates that every person with a chronic rare disease should have a designated care coordinator. As well as ensuring that a patient has access to all the relevant health professionals at the

---

### 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)

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)



right time, care coordinators play an important educational role by providing a source of information and support to health professionals working locally who may have little knowledge of the condition. Care coordinators can help build important linkages across institutional boundaries; between health and social services as well as between specialised and local services.

What would be the best way to feed the views and experiences of patients, service users and carers into the education and training process?  
How can we ensure appropriate and effective patient and public engagement in the new system?

Education and training should encourage and enable professionals to be responsive to patient perspectives and that patient perspectives may need to be derived using a wide range of methods. Patients and patient organisations must be adequately resourced to enable them to engage effectively in the new system (this includes reimbursement for time, travel costs etc).

An understanding of the impact diseases have on real life and its relevance to healthcare practice can effectively be derived from patient and carer testimonies. An example of good practice, which RDUK believes should receive continued funding, is the National Genetics Education Centre's "Telling Stories: Understanding Real Life Genetics" project (<http://www.tellingstories.nhs.uk/>). This resource enhances health professionals' appreciation of the implications of genetics for individuals, families and health professionals by providing interesting, powerful and often moving accounts of the implications of genetics for the daily lives of ordinary people, and the extraordinary way in which they often cope with this.

Providing funding for patient organisations to develop educational resources and CPD/CME training for professionals can also be an effective way to ensure that the education of professionals is influenced by patients' views and experiences. In the case of rare diseases, in the absence of a specialist service for most conditions, patient organisations are often the main source of information about a condition and this knowledge should be harnessed. Similarly, some patient organisations have effectively contributed to the education and training of professionals by employing education workers. For example, The Jennifer Trust for Spinal Muscular Atrophy has an Outreach Service (funded by the Big Lottery Fund) which allows them to employ two outreach workers. Their role is to provide a key service to newly diagnosed and bereaved families, but also to arrange local training sessions for health professionals so that families can be better supported by health professionals working in the local community.

Rare Disease UK – October 2011

---

## 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)

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)