



RARE DISEASE | UK

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



Rare Disease Day

Rare Disease Day 2012 – Key Messages

About Rare Disease Day

- Rare Disease Day takes place on the 29th February (i.e. a “rare day”). When it is not a leap year it is marked annually on the 28th February.
- This is the 5th Rare Disease Day. What began as a European initiative has now spread to become an international event.

Aims of Rare Disease Day

- To **raise awareness of rare diseases** – rare diseases have in the past not been considered a health priority. This is partly due to the mistaken belief that rare diseases affect a small number of people; that there is little that can be done to help people with rare diseases, or that what can be done would be unfeasibly expensive. We are using Rare Disease Day as an opportunity to highlight that there are over **6,000 rare diseases** affecting **3.5 million people across the UK (1 in 17 people)**. Collectively, rare diseases are not rare!
- To emphasise rare diseases as a **health priority**.
- To ensure that by **acting simultaneously and collaboratively**, nationally and internationally the voice of rare disease patients is heard by more people.
- To **highlight the issues** affecting people with rare diseases.
- To bring all of the **stakeholders involved in rare diseases together**. This includes patients, families, carers, policy makers, healthcare providers, clinicians, researchers, health workers, industry and patient organisations.
- To **coordinate policy actions** for rare diseases at each level – across the UK, at a European level and internationally.

Rare Disease UK

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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)



About rare diseases

- It is estimated that there are over 6,000 rare diseases affecting 3.5 million people in the UK at some point in their lives (1 in 17 people). Collectively, rare diseases are not rare!
- In Europe, a rare disease is defined as a disease affecting fewer than 5 in 10,000 of the general population.
- The term disease is used in a broad sense and includes cancers, conditions, syndromes etc.
- The maximum number of people a single condition can affect in the UK for a disease to be classed as rare is around 30,000 people.
- In Europe, there are an estimated 25-30 million people with a rare disease – approximately 6-8% of the population.
- Approximately 5 new rare diseases are described in medical literature each week.
- 80% of rare diseases have identified genetic origins.

The theme for Rare Disease Day 2012 – Solidarity

Solidarity has been selected as the theme of the 2012 campaign to focus on the importance and the need for collaboration and mutual support in the field of rare diseases.

The slogan of the 2012 campaign is:
“Rare but Strong Together”

This theme has been chosen to reflect people active in the field of rare diseases are not alone and should not act alone in order to meet their objectives. Even though they are few and far between, collectively they are many and if they act together they will be stronger.

This theme also reflects that despite the diverse range of rare diseases, each with a range of very different symptoms and prognoses, patients with rare diseases often face common problems for which there may be common solutions. These problems often include:

- Lack of access to correct diagnosis
- Delays in diagnosis
- Lack of information about the disease
- Lack of scientific knowledge of the disease
- No appropriate treatment for the condition
- Significant social consequences
- Lack of appropriate quality healthcare
- Inequities and difficulties in access to treatment and care

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A report [Experiences of Rare Diseases: An insight from patients and families](#) (available at www.raredisease.org.uk/documents/RDUK-Family-Report.pdf) produced by Rare Disease UK (RDUK), based on a survey of nearly 600 patients and family members affected by over 100 different diseases captures some of the problems faced by patients and families affected by rare diseases. Some of these findings are outlined below:

Delays in diagnosis

Almost half (46%) of patients with rare diseases had to wait over one year for a correct diagnosis. Of this:

- One in five (20%) waited over five years.
- More than 10% of patients had waited over ten years.

Close to half (46%) of patients were misdiagnosed before receiving a correct diagnosis. Of this:

- Almost one third of patients had received three or more misdiagnoses.

Over two-thirds (68%) of patients saw three or more doctors before their final diagnosis was made.

One in five (22%) saw six or more doctors.

Poor access to good quality care, information and support

Over half (52%) of patients aren't given enough information on their condition following diagnosis. Shockingly some patients are given no information at all on diagnosis.

64% of patients were not given details of the relevant patient support group on diagnosis.

37% of patients do not have someone they can approach with questions on their condition.

Only 33% patients receive sufficient support with their social needs.

Only 29% feel they receive sufficient psychological support.

Less than a quarter (24%) receive adequate support with financial concerns.

Fragmented and poorly coordinated care

75% of patients do not have a designated care coordinator/care advisor.

25% of patients have to attend three of four different clinics for their condition and 12% have to attend more than five.

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66% of patients have to travel for over an hour to get to their furthest clinic with one in three (32%) having to travel for more than two hours.

30% of patients reported experiencing problems in the transition from paediatric to adult services.

It is not all bad, however; there are many examples of excellent practice throughout the UK. These examples demonstrate that it is possible to provide high quality services and support to people with rare diseases. Many of these services have been shown to save money or are cost-neutral by delivering improved and more efficient care, leading to better health outcomes. Unfortunately, most patients with rare diseases cannot access such high quality services and those services in existence have generally developed ad hoc with little consideration of the overarching needs of patients with rare diseases. We believe that with a strategic approach to the development of services for rare diseases, we can move towards the situation where these services become the norm as opposed to the exceptions. The UK has the opportunity to lead in terms of research, the development of treatments and care guidelines and in the provision of high quality, innovative services for patients with rare diseases.

RDUK have been campaigning for a national plan for rare diseases to address the common problems faced by patients with rare diseases. We have developed comprehensive recommendations to inform this plan in our report [Improving Lives, Optimising Resources: A Vision for the UK Rare Disease Strategy](#) (available to download at: <http://www.raredisease.org.uk/documents/RD-UK-Strategy-Report.pdf>)

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