

# Rare Diseases News

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## Introduction

Hello and welcome to the first issue of Rare Diseases News!

This quarterly e-newsletter is published by the Rare Diseases Taskforce. The aims of the newsletter are to:

- Assist the Taskforce in acting as a catalyst for better health care services for people with a rare disease in Ireland.
- Highlight key developments in relation to rare diseases in Ireland and the EU context.
- Communicate issues related to the forthcoming National Plan for Rare Diseases in Ireland.

## What is a Rare Disease?

In the European Union, a disease is defined as rare when it affects less than 1 in 2000 people. In Ireland it is estimated that 6-8% of the population has a rare disease with (around 280-370,000 people). In other words up to 1 person in 12 may have a rare disease at some stage in their life in Ireland.

The European Commission defines rare diseases as 'life-threatening or chronically debilitating diseases which are of such low prevalence that special combined efforts are needed to address them'. As stated, 'low prevalence' is defined as meaning fewer than 1 in 2,000 people.

Diseases that are statistically rare, but not also life-threatening, chronically debilitating, or inadequately treated, are excluded from the EU definition.

70-80% of rare diseases are genetic, and are present throughout the person's entire life, even if symptoms do not immediately appear. Many rare diseases appear early in life, and it has been estimated that 30% of children with rare diseases will die before reaching their fifth birthday.

Worldwide it is estimated that there are over 7,000 *identified* rare diseases. Many are ultra-rare while others are more common or more widely known, mainly because of the work of patient advocacy organisations; the media highlighting the challenges of patients and their families and the work of clinicians in treating rare diseases.

The Taskforce will be publishing an 'Easy-Guide' which will provide accessible information on all aspects of rare diseases in Ireland and will include profiles of a number of people/families that are coping with a rare disease

## What is the Rare Diseases Taskforce?

The Rare Diseases Taskforce brings together many patient advocacy organisations concerned with rare diseases in Ireland. A list of organisations already involved is provided at the conclusion of this newsletter.

Many of these groups are already members of the GRDO (Genetic and Research Charities Group); IPPOSI (The Irish Platform for Patients' Organisations, Science & Industry) and the MRCG (Medical Research Charities Group).

## Towards 2013 – The National Plan for Rare Diseases (NPRD)

The European Union recommends all member states ensure that patients have access to high quality care, including diagnostics, and treatments and supports for those living with the disease and effective orphan drugs. The EU recommends that health policies are inclusive of rare diseases.

Consistent with this objective, the Irish Government, along with other EU member states, is required to draw up a National Plan for Rare Diseases (NPRD) by 2013. The NPRD has the potential to make a significant impact on the treatment of rare diseases in Ireland, provided the plan is robust, effective and resourced.

Also in line with EU policy, the active participation of patient groups is central to the process of developing a Plan. Further, the Taskforce seeks to work together with health care professionals, industry representatives and policy makers to assist in the planning, implementation and review of an effective NPRD.

## The Core Messages of the Taskforce

The following are the core messages of the Rare Diseases Taskforce which will inform our advocacy and public awareness in the development of the NPRD.

### 1. Planning

**Ireland needs an effective National Plan on Rare Diseases:** There is considerable fragmentation in current services and treatment for rare diseases. Ireland needs an effective NPRD to ensure that people with a rare, often life threatening disease, have equal and fair access to treatment and a better quality of life. There has been growing awareness in Ireland in recent years of the sometimes devastating impact of living with a rare disease, particular for families without access to adequate services and expertise.

### 2. Diagnosis

**Access to correct diagnosis:** Medical practitioners may fail to identify symptoms that they rarely come across, resulting in significant potential for patients to experience lengthy delays in diagnosis and often experiencing misdiagnosis.

The lack of a NPRD and the scarcity of medical expertise often result in patients experiencing prolonged delays in getting an accurate diagnosis. Frequently it involves years of repeated visits to different doctors. Misdiagnosis or non-diagnosis are significant hurdles to improving quality of life for thousands of patients, and often results in inadequate or even harmful treatments.

### 3. Information

**Access to information:** Once an accurate diagnosis has been made, additional hurdles must be overcome. Patients and their carers may have considerable difficulty sourcing appropriate information about the condition and identifying relevant/experienced specialists (should they exist in Ireland) can be particularly challenging. Delays and inappropriate treatment can aggravate existing symptoms and leave sufferers feeling helpless, vulnerable, neglected, misunderstood and uncertain about their future health.

### 3. Treatment

**Access to treatment:** For some conditions where no medicine/drug therapy exists, symptoms can be managed effectively with a correct multi-disciplinary approach leading to

improved health and greater quality of life. Many rare diseases are multi-systemic, therefore timely referral to the range of appropriate consultants is essential with collaboration between specialists. Equality of access to treatment is a key issue regardless of condition, geographical location or income.

The lack of any established process to evaluate, approve and commission orphan medicines results in unnecessary and prolonged delays for patients trying to access available treatments. We welcome the Treatment Abroad Scheme (TAS), however every effort should be made to provide treatment within Ireland or as close to home as possible.

Progressive disorders require pre planning, fast track support including access to existing services, education and counselling services.

Access to available orphan drug treatment is haphazard and due to the lack of a national policy, each region or hospital makes individual treatment decisions. This process results in geographic discrimination for patients, long delays in accessing treatment, lack of transparency and no identified appeals process. A commitment is required to ensure universal access to high quality healthcare on the basis of speedy access to good quality care, equality and solidarity. Treatment should be made available on the basis of need at the point of care closest to the patient.

#### 4. Expertise

**The development of centres of expertise:** Centres of expertise on rare diseases should be commenced or further developed in Ireland. This approach is recognised internationally as the most important way to address the multifaceted issues raised by rare diseases.

It is well recognised that the best way to improve access to high quality healthcare for rare diseases is to establish new and to develop existing centres of expertise. Centres of expertise may also facilitate the development of specialised social services which will improve the quality of life for those with rare conditions. In the absence of such centres in Ireland, patients and families search endlessly for funding and improved services, often without success. It is of vital importance that existing centres of expertise should be strengthened and resourced to serve people with rare diseases in Ireland. (For example, the National Centre for Medical Genetics has a major role in helping Irish families with rare diseases, given that 70- 80% of rare diseases are genetic in origin. However, the NCMG has lost over 20% of its staff due to the resource restrictions introduced by the HSE, leading to a diminution, rather than an improvement, in services).

#### 5. Co-ordination

**Co-ordination of Policy at a national level:** A clinical lead and a national office for rare diseases should be established to coordinate the planning, implementation and review of a NPRD.

#### 6. Participation

**Participation of patient groups:** Patient groups are often the drivers for change in health policy in Ireland. Patient groups must be included in all aspects of the national action plan on rare diseases including the design, implementation and review of the plan. Recognition must be given to the many rare disease support groups which are sometimes the main specialist support and information provider on their particular rare condition.

The development of Health policy and service delivery should be patient centred with the patient becoming an active participant in service design and implementation. True consultation must involve patients and representative organisations. Such stakeholder collaboration is essential and should not be confused with merely informing stakeholders of policy decisions after the event.

The co-ordination of care between paediatric and adult services including transition from one to the other should be managed effectively.

## 7. Research

**Greater focus on research on rare disease at a national and international level:** Funding for research into rare disease should be given a considerably higher priority than at present. The more common diseases tend to attract the largest research budgets. Incentives/positive action should be introduced to stimulate research into rare diseases both within Ireland and through collaborative clinical research with other countries.

## 8. Data

**Improved data to treat rare diseases:** The importance of collecting and analysing data, including the development of electronic patient records, patient registers and patient registries is increasingly recognised as playing a vital role in the treatment of all diseases. National standards in collecting and analysing such data should also apply to rare diseases.

## 9. Mainstreaming, targeting and resources

**The national action plan on rare diseases cannot be a 'virtual plan':** All relevant health and social service strategies in Ireland should be inclusive of rare diseases, including the HSE Corporate Plan and all relevant current and forthcoming health strategies. Targeted additional resources to treat and manage rare diseases must be provided under the NPRD.

## Get Involved: Rare Diseases Day (RDDay), February 29<sup>th</sup> 2012

On Wednesday February 29, 2012 millions of people around the world will observe Rare Disease Day for the 5th year, including Ireland.

Besides dealing with their specific medical problems, people with rare diseases struggle to get a proper diagnosis, find information, and get treatment. The rarity of their conditions makes medical research more difficult.

2012 is a leap year so Rare Diseases Day (RDDay) will fall on a comparatively rare day, Wednesday, 29<sup>th</sup> of February.

At a European level the goal of the RDDay is to ensure Rare Diseases are a higher priority in public health and research agendas, and to increase the budget for Rare Diseases. The theme is 'solidarity'.

The particular focus of RDDay in 2012 is to identify the priorities that need to be included in Ireland's National Plan on Rare Diseases (NPRD), which to be completed before 2013.

### What you can do for RDDAY 2012

We are asking all organisations concerned with rare diseases to do the following:

1. If you publish a newsletter, the front cover/banner headline of your February edition (or nearest edition) could usefully highlight RDDay 2012, including:
  - a. Why we have a RDDay
  - b. The need for an effective NPRD
  - c. Personal stories of those affected by a rare disease (focussing on issues such as late diagnosis, access/cost of treatment)
  - d. A forthcoming 'easy guide to rare diseases' will assist you in this process and the wide distribution of this guide to your members would also greatly assist
2. If you are organising event around this time, why not plan to coincide it close to RDDay to raise the awareness of the day among your members (while avoiding holding it on the 29<sup>th</sup> of February itself because of the national event)
3. Join our 'write in for rare diseases' when all organisations concerned with rare diseases will be joining together to write to all TD's and Senators highlighting the importance of an effective NPRD (a template to guide the main points that could usefully be included will be provided along with list of Oireachtas members).

More information to RDDAY 2012 will follow in future issues of this Newsletter.

### Background: The European Council (EU) Recommendation

The European Council adopted in June 2009 the 'Recommendation on an action in the field of rare diseases'. The Recommendation supports the adoption of national plans and strategies for responding to rare diseases (RD) before 2013.

The Council recommends that Member States should establish and implement plans or strategies for rare diseases or explore measures for rare diseases in other public health strategies, to ensure that patients have access to high quality care, including diagnostics, treatments and supports for those living with the disease and, if possible, effective orphan drugs.

## Taskforce Members

The following are the patient advocacy members of the Rare Diseases Taskforce

### Networks

- GRDO (Genetic and Research Charities Group)
- IPPOSI (The Irish Platform for Patients' Organisations, Science & Industry)
- MRCG (Medical Research Charities Group).

### Patient Advocacy Organisations

- Irish Cancer Society
- Fighting Blindness
- Cystic Fibrosis Association of Ireland
- Pompe Disease
- Alpha One Foundation
- Huntington's Disease Association of Ireland
- Bee for Battens
- Cystinosis Foundation Ireland.
- 22Q11
- Fabry Ireland
- Muscular Dystrophy Ireland
- Friedreich's Ataxia Society Ireland
- Debra Ireland
- Irish Raynaud's and Scleroderma Society

If your organisation wishes to become involved, or if you are an individual who wants to find out more about our work, please contact Marie Downes [mdownes@mrcg.ie](mailto:mdownes@mrcg.ie)

We would welcome information items for the next issue of Rare Diseases News. If you wish to contact us about this or the next issue of **Rare Diseases News** please contact Philip Watt, CEO Cystic Fibrosis Association [pwatt@cfireland.ie](mailto:pwatt@cfireland.ie). Copy Date is 9 December 2011.

## Other News

### Survey on the experiences of People and Families affected by a Rare Disease

On behalf of the Rare Disease Taskforce GRDO will be conducting a survey in the coming months to gain information on the experiences of individuals and families affected by rare diseases.

This survey will help to identify common issues and problems. We will seek the support of patient organisations and other stakeholders in order to distribute the survey to as wide an audience as possible

### Addition to newborn screening

An important tool in diagnostics for some rare diseases is the use of newborn blood spot screening. Newborn screening for Cystic Fibrosis (CF) commenced in July 2011.

The National Newborn Bloodspot Screening Programme identifies babies who may have rare but serious inherited conditions, which are treatable if detected early in life. Early treatment can improve their health and prevent severe disability or even death. The screening test is offered to all parents when the baby is about 4 days old.

The conditions that are presently tested for are:

- Phenylketonuria (PKU)
- Homocystinuria
- Maple Syrup Urine Disease
- Classical Galactosaemia
- Cystic Fibrosis
- Congenital Hypothyroidism

For more information see the following recently developed HSE web page  
<http://www.hse.ie/eng/services/healthpromotion/newbornscreening/>