

# Rare Diseases News

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## Introduction

Welcome to our second 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 Rare Disease 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.

## Letter from John Devlin

***Deputy Chief Medical Officer, Department of Health and Children,  
Chairman, DoHC Steering Group for Rare Disease National Plan***

In the past few years there has been a considerable amount of work done at European level in the area of rare diseases. Ireland has played its part in this initiative. The twin approach of developing a specific national plan together with European collaboration is the key element to improving the health and social care for people with rare disease.

Ireland has been supportive of the EU proposals on rare disease which concluded with a council recommendation in June 2009. The end point is that countries are recommended to develop plans or strategies preferably by the end of 2013. We are now well advanced in developing this work. In January of this year, Europlan organised a national conference bringing together patients, patient organisations and healthcare professionals to discuss what might feed into the development of a national strategy for rare diseases. The Minister for Health has now established a National Steering Group which is tasked with developing a five year national plan and which will deal with the diagnosis, prevention, management, treatment and research of rare diseases in Ireland. The key principals governing the plan will be those of patient centeredness, quality, care and equity of access to treatment.

It is recognised that the impact of a rare disease goes beyond the symptoms experienced by the person affected and involves the whole family, healthcare professionals and the broader health system. The Eurordis report 'The Voice of 12,000 Patients' clearly articulated the challenges of making an early diagnosis, the need for appropriate treatment and counselling as well as the ongoing requirement for family support. There are other challenges such as information systems that are required and we will need to consider the most appropriate registries and data bases which can be used to plan and manage services in Ireland.

The Europlan report which looked at services across Europe commented that in most countries, there are no designated centres of expertise and even where they exist that there is significant variation in their organisation and how they operate. The identification of appropriate centres of expertise for rare diseases is a key priority. Allied with this is the organisation of suitable healthcare pathways so that patients and professionals know where the most appropriate care can be provided. Another area is the development of a research infrastructure with appropriate support. This is where there is obvious added value in working with our European partners on programmes for rare disease research. Research is an integral part of overall care for rare diseases including access to clinical trials where appropriate. Access to appropriate medication and technology is also very important. In

Ireland the elements exist to have a transparent process to ensure equitable access to orphan drugs as well as to incentivise orphan drug development through our strong pharmaceutical and biotech industry. Another area is the empowerment of patient organisations. The Europlan report contains a lot of practical guidance of what can be done to empower patients and their families in a meaningful way.

The National Steering Group is working in all of these areas. It is also considering how best to consult with patients and key stakeholders on the broad proposals and recommendations that will emerge. This will be a key target for 2012.

Despite the challenging economic situation, we believe that significant progress can be made using the resources that are currently available. The uniqueness of rare diseases where there are a limited number of patients and a scarcity of professional knowledge and expertise can leave people isolated and vulnerable. The 'diagnostic odyssey' that patients sometimes have to undergo to avail of appropriate treatment is something that must be addressed. While a national plan will not change this overnight, nonetheless we can make progress on providing better information for patients, designating appropriate centres to provide high quality treatment, setting out a fair and transparent means to allow access to orphan drugs and promoting research for the benefit of patients with rare diseases. There is a strong commitment from all stakeholders which is key in our ongoing development of the Irish National Plan for Rare Diseases.

### **Letter from John McCormack**

***CEO, Irish Cancer Society***

***Chairman, MRCG (Medical Research Charities Group)***

***Chairman, Taskforce for Rare Disease National Plan***

I am delighted to see the commitment given by the Department of Health to the development of a National Plan for Rare Diseases. As a member of the Steering Group and Taskforce for this plan, it is good to see that the efforts of the many groups concerned with supporting the individuals and families affected by a Rare Disease in Ireland is starting to come to fruition. As is often cited, although rare diseases are individually rare, in totality they account for 6-8% of the overall population. We can do so much more to support these individuals - to provide better diagnosis and information, more co-ordinated care, access to available treatments and to prioritise research into rare disease treatments. Establishing and putting in place a National Plan for Rare Disease will take time, passion and commitment but will also start to address the marginalisation, frustration and often despair experienced by patients and families of rare diseases. Thank you to all who have worked to get us this far and we look forward to the challenge ahead.

## EUCERD Meeting Update Oct 25<sup>th</sup> and 26<sup>th</sup> 2011 / [www.eucerd.eu](http://www.eucerd.eu)

The European Union Committee of Experts on Rare Diseases (EUCERD) was formally established in November 2009. This committee is charged with aiding the European Commission with the preparation and implementation of Community activities in the field of rare diseases, in cooperation and consultation with the specialised bodies in Member States, the relevant European authorities in the fields of research and public health action and other relevant stakeholders acting in the field.

The EUCERD holds three meetings per year at the European Commission in Luxemburg. In addition, members regularly take part in working groups and conference calls on specific topics.

The event in October started with a Press Conference and was followed the next day by a cluster meeting on **“European Actions to improve the life of patients living with rare diseases”**. Avril Daly, CEO of Fighting Blindness was invited to speak as a patient at the Press Conference of this event

The purpose of this event was:

- To justify the usefulness of the EU Public Health Programme (and its budget of €300m in total for the period 2007-2013)
- To further increase investment in and maintain Rare Diseases as a top priority based on the demonstration of its high community added value and real impact on people's lives

The Public Health Programme (PHP) is essential to provide support, empowerment and a better quality of life to those living with rare diseases across the EU. It is the instrument funding the Operating Grant of Eurordis, rare diseases conferences, essential projects such as EUROPLAN, and projects which we hope to benefit from in the future such as EPIRARE - which is an initiative to develop registers for rare diseases across the EU. It also supports the Joint Action of the European Committee of Experts in Rare Diseases (EUCERD), ORPHANET and all projects to develop European Reference Networks in the future.

The European Commission (EC) is currently preparing the budget for the period 2014-2020. Due to current EU budget constraints, the Commission needs to cut its budget in specific areas - while also clearly demonstrating its benefit to citizens living in the EU. There is a risk that the Public Health Program (PHP) will simply disappear in the budget cuts, in areas which are not a priority in the European Treaties or it may indeed be reinforced based on the demonstration that the Public Health Program (PHP) brings value for money, that European citizens / civil society are requesting it and that the Public Health Program (PHP) has a real impact on health outcomes of the European population.

The Health Commissioner John Dalli (Malta) has announced and reiterated publicly that rare diseases is one of the top priorities for the future. Also, President Barosso in his declaration of the outline of 2014-2020 has maintained the Public Health Program (PHP) and has explicitly mentioned rare diseases as a priority. However, despite all of this perceived support, the track record of successful projects on Rare Diseases and despite proposals from the Commission, the final say on budget lies with the Council of Ministers (all Member States) and the European Parliament.

The press conference was organized to make the position clear and to try to protect this important initiative for the sake of the 30million people affected by Rare Diseases throughout the EU. It included speeches from:

Mrs. Paola Testori-Coggi, the Director General of DG SanCo, Mrs. Despina Spanou, and Special Advisor to Commissioner Dalli and Mr. Mars di Bartolomeo, Minister of Health for Luxemburg. There were also testimonials from four patients/family members on the concrete impact of EU funding/policies in everyday life.

The four patient representatives were asked to speak about their personal situation and how they got involved in rare disease advocacy and also to speak on the impact of EU policy.

The patient representatives were:

- Yann Le Cam / Cystic Fibrosis / the Rare Disease movement in the EU / CEO of EURORDIS
- Lesley Green / Cystinosis & Metabolic Diseases / COMP & Orphan Drugs
- Kay Parkinson / Alstrom Syndrome / Role of EU funded projects for a very rare disease
- Avril Daly / Rare Retinal Diseases / Patient involvement in the development of National Plans on Rare Diseases in Ireland

The audience consisted of representatives from 17 health ministries, industry, scientists, patient representatives and 20 journalists from across the EU. Joanne Hunt from the Irish Times accompanied Avril from Ireland.

### **GRDO Patient Organisation Survey for Rare Disease National Plan**

The Genetic and Rare Disorders Organisation (GRDO) is currently gathering information relating to patient support and advocacy organisations operating in Ireland for people with rare conditions.

GRDO, together with the Medical Research Charities Group (MRCG) and the Irish Platform for Patients' Organisations, Science and Industry (IPPOSI) and their patient representatives, will use this information to assist the National Rare Diseases Taskforce ('Towards 2013 - the National Plan for Rare Diseases') to engage with the Rare Diseases Steering Committee of the Department of Health.

If your organisation has not already completed the patient organisation survey please do so now. The survey can be completed online at the following link:  
<http://www.surveymonkey.com/s/K82YY8Y>

### **Patient/Family Survey for Rare Disease National Plan**

The Patient Support & Empowerment Sub Group, of the Dept. Of Health Steering Group for Rare Disease is seeking inputs from anyone who wishes to contribute to the submission. You can email your contribution to Tony Heffernan of Bee For Battens, Patient Representative & Chairman of the Patient Support & Empowerment Subgroup, at [info@BeeForBattens.org](mailto:info@BeeForBattens.org).

If your comments contain references to existing publications or papers, please ensure that an appropriate hyperlink and document title is provided.

## Get Involved: Rare Diseases Day (RDDay), February 29th 2012

We want to remind you again about Rare Disease Day 2012.

On Wednesday February 29, 2012 millions of people around the world will observe Rare Disease Day for the 5th year. 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, 29th 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 is 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 (focusing 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 an 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 29th 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).