

Rare Diseases News

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Introduction

Welcome to issue 3 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.

Rare Diseases Day 2012

February 29, 2012 marked the fifth international Rare Disease Day. On this day hundreds of patient organisations from more than 50 countries worldwide organised awareness-raising activities converging around the slogan "Rare but strong together".

Eurordis launched a specially produced promotional video highlighting Rare Disease Day in Europe. The video was made in Dublin and featured real people affected by rare conditions.

Six people from Ireland, each with a different rare condition, featured in the video. The message is simple – *It's not unusual to have a rare condition. Look out for us and you'll see how many we are!*

To tie in with the video, GRDO created a postcard campaign to highlight Rare Disease Day in Ireland. Each postcard featured a character from the video and a short message about Rare Disease Day. The postcards were delivered to politicians and other public figures, as well as to media representatives across Ireland.

GRDO Patient Organisation Survey for Rare Disease National Plan

In February of this year, GRDO conducted a survey of Patient Support and Advocacy Organisations to gather information for the Rare Disease National Plan. The survey revealed significant difficulties in the areas of access to diagnosis, information and to treatment, as well as a lack of coordination in delivery of services. The results of this were very interesting and echo the backup the issues raised by the Rare Disease Taskforce.

Some of the results are listed here:

Respondent Profile

- 69 distinct rare conditions are listed by respondents
- The subjects of the survey (i.e. those affected, who either responded themselves or whose carers responded for them) have an average age of 24 years.
- Almost three quarters of them (72.4%) reported that their rare condition is genetic.
- 13.8% "don't know" whether their condition is genetic as well as rare.
- 13% say they are registered with the National Intellectual Disability Database (NIDD) and 15% with the National Physical and Sensory Disability Database (NPSDD).

- More than half of respondents say they are not registered with either, over one third “don’t know” whether or not they are registered.

Diagnosis and Referral

- Waiting time for diagnosis was reported as more than 10 years by 13.3% of respondents.
- 1.8% of those who took the survey said that they still do not have a definite diagnosis.
- 37.2% received an incorrect diagnosis before receiving the correct one.
- 70.4% of respondents report having to wait less than three months after diagnosis for referral to a consultant or specialist.
- 6.5% waited more than 18 months.
- Some patients received no referral as no specialist is available in Ireland in the relevant condition.

Treatment and Care

- Almost three quarters of respondents (73.1%) are receiving treatment from more than one hospital consultant or other medical professional for their condition. Over half of these (52.6%) report attending between 2 and 5 different medical professionals, and 5.1% report attending more than ten different medical professionals for their treatment.
- 17% of patients have accessed medical care for their condition in another country. 23% have accessed genetic testing abroad.
- The five most common services for which people are waiting to access are: occupational therapy; respite; speech and language therapy; benefits advice; and specialist clinics. Incidences of very long waiting times for essential treatment and care are common, and a small number of patients report paying privately for certain services, most frequently counselling, while waiting to receive them.

Information and Support

- 68.8% of respondents state that they do not feel that they were provided with sufficient information on their condition following diagnosis. Many comment that they did all the information gathering themselves, mainly using the Internet. Others report relying on patient support organisations for all information. Lack of genetic counselling or long waiting times for genetic counselling is also a major issue.
- 55.2% of respondents state either that there is no patient support group in Ireland for their condition or that they do not know of one.
- When asked who is their main source of information on their condition, respondents listed as follows:

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| Internet | 62.5% |
| Other individuals / families with the condition | 37.5% |
| Hospital Consultant | 35.4% |
| Patient organisation | 29.2% |
| Social media (e.g. Facebook, Twitter) | 26% |

- Fewer than 10% of respondents list a member of the genetics team, a GP or a nurse as being their main source of information on the condition.

Education and Employment

- 55.2% of respondents report that the rare condition affects their ability to participate in full-time education or employment.
- 35.4% of respondents say that lack of support / flexibility in relation to the rare condition excludes them from education and employment opportunities. 29.2% say lack of support or flexibility “somewhat” excludes them and a further 35.4% do not feel excluded in this field due to lack of support / flexibility relating to their condition.

Research into the Condition

- Only 12.6% of respondents are aware of a patient registry in Ireland for their condition. 10.5% are aware of a relevant patient registry in another country. 64% are not aware of any patient registry for their condition anywhere.
- 69.5% of respondents state that they are not informed of clinical trials or research relating to their condition. 75.8% have not participated in any research into their condition. A total of 21% say that they have participated in research, either in Ireland (14.7%) or elsewhere (6.3%).

Meeting with the Joint Committee on Health

The Joint Committee on Health met with a group from GRDO (Genetic and Rare Disorders Organisation), the Rare Diseases Taskforce, Rett Syndrome Ireland and 1P36 Deletion syndrome on Rare Diseases Day, Wed 29th Feb. The meeting provided committee members with an in depth briefing on the issues faced in terms of diagnosis and treatment of rare diseases, access to appropriate medication and the identification of appropriate centres of expertise for rare diseases.

Clinical Care Programme for Rare Diseases

On March 7th, the Minister for Health, Dr. James Reilly announced his intention to develop a Clinical Care Programme for Rare Diseases.

It is hoped by including Rare Diseases in the National Clinical Care Programme a clearly defined pathway will be developed to enable medical professionals and patients to access the correct specialists and centres of expertise, up-to-date information regarding new treatments and management options and information regarding on-going clinical trials. Due to the complexity and number of rare conditions, expertise is not always available nationally therefore the programme must enable linkage with other European Reference Networks (ERNs) enabling experts to input into complex cases, speeding up the process and leading to correct and timely diagnosis and appropriate interventions.

Under a Clinical Care Programme for Rare Diseases we have the potential to utilise and better access national and international centres of expertise as well as European Reference Networks to enable the provision of safe care to all those affected by rare conditions in Ireland.

Rare Disease National Plan Consultation



John McCormack CEO Irish Cancer Society, Helen McAvoy Institute Public Health, Eibhlin Mulroe CEO IPPOSI, Minister James Reilly, Avril Daly CEO Fighting Blindness and Tony Heffernan CEO The Saoirse Foundation at the Rare Disease National Plan Consultation

On Monday the 11th of June, 2012 at 10am, the Department of Health's Consultation Day on a National Rare Disease Plan for Ireland took place at Farmleigh House in the Phoenix Park. This event followed the Minister for Health, Dr James Reilly's decision last April to appoint a National Rare Diseases Steering Group in order to develop a national plan for rare diseases. Similar plans are being drafted across the EU following a recommendation by the European Council of Health Ministers which requires Member States to implement national plans before the end of 2013.

Speaking in advance of the event Mr. John McCormack, Chairman of the Taskforce for Rare Diseases and of the Medical Research Charities Group (MRCG) said "Much work has been done to ensure this plan will meet the needs of people with rare diseases and I welcome the level of engagement which the Department has had with patients' organisations thus far. In particular the launch of the online consultation process offers people with a rare disease and their families the opportunity to contribute to the formation of the plan." The online consultation process will last for four weeks and can be accessed by visiting <http://www.hse.ie/eng/services/ysys/Consultation>

Eibhlin Mulroe a member of the Steering Group and CEO of The Irish Platform for Patients' Organisation, Science and Industry (IPPOSI) said "the move to include a patient's perspective in the process recognises the level of expertise which patients have and their willingness to engage with all the stakeholders. 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 and the EU is starting to come to fruition. Establishing and implementing a National Plan for Rare Disease will start to address the marginalisation, frustration and often despair experienced by patients and families of rare diseases."

Avril Daly, Chairperson of the Genetic and Rare Diseases Organisation (GRDO) commented that " Irish patient groups have engaged with stakeholders nationally and internationally for 25 years to advocate for the development of better systems leading to better health outcomes for patients affected by rare diseases. We welcome the commitment made in the Seanad last March by the Minister to develop a Clinical Care Program for rare diseases. We are hopeful that by including rare diseases in the National Clinical Care Programme and the establishment of a National Rare Disease Office a clearly defined pathway will be developed to enable patients and medical professionals access the correct specialists and centres of expertise, new treatments and management options and information regarding ongoing clinical trials."

Ms Daly who was recently elected Vice President of the European Organisation for Rare Diseases EURORDIS went on to say "Ireland will hold the European Presidency in 2013 which coincides with the deadline for member states to have a national rare diseases plan in place. It is therefore important that Ireland provides strong leadership on the rare diseases issue during its presidency."

Tony Heffernan, a Steering Group member and Founding CEO of Bee for Batters / The Saoirse Foundation said "In the past it has been very difficult for parents to find any information at all about rare diseases. The rarity of some of these disorders means that few doctors have an awareness and parents of course want to know, on a practical level, how they can best help their child. Hopefully this consultation process will give parents an opportunity to input directly into the plan".