

Genetic and Rare Disorders Organisation

NEWSLETTER

2011 - 2012

GENETIC AND RARE DISORDERS ORGANISATION

Welcome readers to the fourth edition of the Genetic and Rare Disorders Organisation (GRDO) newsletter.

GRDO is an Irish alliance of voluntary groups supporting people with genetic and other rare disorders. We hope that this publication will provide our readers with some useful information, while updating you on some of our recent work.

INFORMATION & DEVELOPMENT OFFICER

GRDO acknowledges funding received from the Department of Environment, Community and Local Government scheme to support national organisations in the community and voluntary sector.



This funding enables the employment (part-time) of Information and Development Officer, Kathy Tynan. Further details inside.

INFORMATION & DEVELOPMENT OFFICER

In October 2011, GRDO appointed its first employee (part-time). Information & Development Officer, Kathy Tynan, is based at GRDO's new office in the headquarters of Muscular Dystrophy Ireland.

Since her appointment, Kathy has been busy representing GRDO at various meetings and conferences, including most recently the 6th European Conference on Rare Diseases and Orphan Drugs and the EURORDIS AGM (23-25 May 2012). Kathy has also initiated two surveys for GRDO, one of individual patient experience and one of patient organisations for those affected by rare conditions. Information gathered has been invaluable in assisting GRDO in its role on the National Rare Diseases Taskforce, and allowing GRDO to make recommendations for better support and services for people with rare conditions. Kathy has also been available to talk to members, keep the GRDO website up-to-date, promote GRDO in the media and co-ordinate activities for Rare Disease Day 2012, including the official European promotional video for the event, which was filmed in Dublin last December.

Contact details for Kathy Tynan (Mondays and Tuesdays):

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UPDATE ON NATIONAL RARE DISEASE PLAN

In April 2011, the Minister for Health established a National Steering Group to develop a policy framework for the diagnosis, prevention and treatment of rare diseases in Ireland, in line with an EU recommendation.

The National Steering Group identified a number of areas relating to rare diseases which it has been considering, including Centres of Expertise and European Reference Networks, Access to Orphan Drugs and Technologies, Rare Disease Research, and Patient Support and Empowerment.

A Rare Diseases Taskforce was established to help inform the work of the Steering Group. The Taskforce has brought together many patient advocacy organisations concerned with rare diseases in Ireland including GRDO, IPPOSI (Irish Platform for Patients' Organisations, Science & Industry) and MRCG (Medical Research Charities Group), as well as many of their member organisations.

Submissions to the Steering Group have been made by the Taskforce in each of the areas listed above.

An integral part of the National Rare Disease Plan will be the development of a dedicated clinical care programme. A proposal has been submitted to the HSE to establish a national clinical programme for rare diseases. The HSE has agreed to establish such a programme, the details of which remain to be worked through.

Update on National Rare Disease Plan (continued)

Minister for Health, Dr James Reilly TD, gave a commitment to developing this clinical care programme on 7 March this year.

Consultation

The Steering Group is planning this summer to consult patients and key stakeholders on the broad proposals and recommendations that will emerge. An event will be held on 11 June at Farmleigh in Dublin and, in addition, an online consultation process will be available to those not in attendance.

It is expected that the Steering Group will submit a plan to the Minister for Health during the latter half of this year.

RARE DISEASE DAY MARKED AT UCD



Three leading experts involved in the Rare Disease Module at University College Dublin's School of Medicine - Dr. Paula Byrne, Eibhlin Mulroe and Dr Tomas Carroll explained the significance of

International Rare Disease Day in an article to mark the occasion.

In partnership with the Irish Platform for Patients' Organisations, Science and Industry (IPPOSI), the UCD School of Medicine and Medical Sciences (SMMS) recently rolled out a new educational module on Rare Disorders and the Medical Healthcare Professional. IPPOSI patient group members are active participants in this important educational module which covers diseases such as Epidermolysis Bullosa, Emphysema and Alpha-1 antitrypsin deficiency, Sarcoidosis and hereditary metabolic disorders such as Fabry's Disease.

FIRST EURORDIS GALA FUNDRAISER AND AWARDS A SUCCESS

The first EURORDIS Black Pearl Gala Dinner took place on Rare Disease Day 2012 in Brussels, Belgium. Over €300,000 was raised, and the event incorporated the first EURORDIS Awards for excellence and leading work in the field of rare diseases.

Among those presenting the awards was GRDO Chairperson, Avril Daly, who is a member of the EURORDIS board of directors.

RARE DISEASE DAY 2012 | 29 February 2012

Rare Disease Day 2012 video and postcard campaign

In December 2011, GRDO collaborated with European Rare Disease Alliance, EURORDIS, in the production of a short awareness-raising video.

The video featured six people from Ireland, each with a different rare condition. As part of International Rare Disease Day 2012, the video was launched in 12 different languages and received over 100,000 views around the world. GRDO also produced a series of postcards based on the video, carrying basic information about rare disease issues.

These were sent to members of the media, and Dáil and Seanad representatives ahead of Rare Disease Day this year. GRDO would like to thank everyone involved in the campaign, especially the six participants. For more information and to watch the video see http://www.rarediseaseday.org/solidarity

MEET THE STARS



- **1. Lauren Shaw** is a bright and bubbly eight-year old. She lives with Friedreich's Ataxia, a rare condition that causes nerve damage and movement problems.
- **2. Jamie O'Brien's** ambition is to work in music production and website design. Jamie has Ehler-Danlos Syndrome, the hypermobility type that causes his joints to dislocate constantly.
- **3. Josephine McGuirk** moved to the seaside in recent years and feels healthier for it. She has Alpha-1 antitrypsin deficiency, a rare genetic condition that affects the lungs and liver.
- **4. Christy Murtagh** is embarking on a new career in counselling, having left behind the building trade. Christy is affected by Retinitis Pigmentosa, which causes gradual but progressive sight loss.
- **5. Clare Louise Creedon** has another form of Ataxia: Ataxia Oculomotor Apraxia-1 (AOA1), and is in a wheelchair full-time now. Despite this, she makes sure to do the things she enjoys like going to the gym, shopping, and getting the bus to visit friends.
- **6. Evan O'Gorman** is a keen actor and, though still at school, already has a few paid roles under his belt. Evan has Epidermolysis Bullosa, an extremely painful skin condition affecting his feet.

Special thanks also to the patient organisations involved: Ataxia Ireland; Alpha One Foundation; Fighting Blindness; and DEBRA Ireland.

GRDO ADDRESSES JOINT OIREACHTAS COMMITTEE ON RARE DISEASE DAY

On the afternoon of Rare Disease Day, GRDO Chairperson Avril Daly was among a delegation to address the Joint Oireachtas Committee on Health & Children about rare disease issues.

Margaret Webb, Chairperson of Irish Platform for Patients' Organisations, Science and Industry, IPPOSI; Andrew Kehoe, Chairperson of the Rett Syndrome Association of Ireland; and Michael Nolan of 1p36 Deletion Syndrome Ireland, also addressed the committee on behalf of all those affected.



'Join hands around the world for rare diseases' initiative

Caption: Representatives from GRDO, along with patient representatives from the Rett Syndrome Association of Ireland, participated in a worldwide show of solidarity on Rare Disease Day by joining together to raise hands for rare disease and submitting our picture to the official Rare Disease Day gallery.

Deputies and Senators were told of the many common issues facing those with rare conditions, including difficulties with diagnosis, difficulties accessing services and support, and a lack of information on rare conditions.

Speakers cited examples of countries such as France, Denmark, Spain, Portugal, Bulgaria and Romania, who have all developed national rare disease plans that have demonstrated benefit to patient outcomes, and, most significantly at this time, have also shown savings.

Delegates referred to the forthcoming consultation process on Ireland's proposed national rare disease plan and called for the plan to be made a reality as quickly as possible. As a first step, the urgent need for a clinically-led national centre for the co-ordination of information and the establishment of care pathways for patients was also highlighted to the committee.

SEANAD DEBATE ON RARE DISEASES

Following the meeting of GRDO and other rare disease community representatives with the Joint Oireachtas Committee on Health and Children to mark Rare Disease Day, a Seanad debate was held on 7 March 2012.

During this session, Minister for Health, Dr James Reilly TD, announced his intention to develop a Clinical Care Programme for Rare Diseases. GRDO welcomed this commitment, which will go a long way towards addressing these issues and, we hope, the current information deficit among patients and healthcare professionals.

GRDO SURVEYS

Patient Experience

GRDO would like to thank members and all those who responded to our recent "patient experience" survey. In our role on the National Rare Diseases Taskforce, we have been able to use the information you have given us to help make recommendations for better support and services for people with rare conditions.

On the eve of Rare Disease Day, we issued a press release outlining some of the many challenges faced by those affected by rare conditions, which were highlighted in the survey, and calling for the establishment of a National Rare Disease Office.

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. 13.3% of respondents report waiting more than 10 years for diagnosis and 37.2% report receiving an incorrect diagnosis before the correct one. 73.1% of those surveyed report attending more than one hospital consultant or other medical professional for treatment yet 44.8% report not having a specific point of contact to go to with questions about their condition. 62.5% of respondents list the Internet as their main source of information.

The establishment of a National Rare Disease Office in Ireland and the development of co-ordinated healthcare pathways are the two changes to the healthcare system that respondents would most like to see (82%), followed by more information about their conditions (73%).

Patient Organisations

Between winter 2011 and spring 2012, GRDO also conducted a survey of rare disease patient organisations. Findings can be summarised as follows:

- Rare disease patient organisations in Ireland take many forms.
- The prevalence of most rare conditions can only be estimated in Ireland as patient registries do not exist here for the majority of them.
- 65% of organisations receive no statutory funding. Few are in
 a position to fund professional staff to provide specialist patient
 care services. Some fundraise to hold conferences and events
 to bring international expert knowledge to their patients here.
 Many use very modest resources to facilitate a voluntary
 network of patients and carers to provide peer support or to
 participate in research initiatives.
- Information services and family support are chief among the services offered by patient support groups in Ireland, followed by advocacy and research. This reflects both where demand lies in the community, and the limited funding available to most groups.
- Over half of the organisations surveyed have no paid employees. Links to international condition-specific networks are maintained by the vast majority (92.3%), however. This reflects both the lack of information and support structures in Ireland for rare conditions but also increasingly the dynamic nature of the patient body here, for whom the advent of the Internet age and social media has brought exciting new possibilities in communication.

Further details of both surveys can be found at www.grdo.ie

CRISIS IN MEDICAL GENETICS SERVICES

The provision of adequate medical genetics services is vital to the implementation of any national rare disease strategy. Around 80% of rare diseases are genetic in origin. This can mean that members of a family may be at risk of the same rare disease, or at risk of passing the rare disease on to any children they have. There is increasing concern in the rare disease community about the state of medical genetics services in Ireland. GRDO frequently hears from members of excessively long waiting times (more than one year) for specialist assessment, including genetic counselling. Testing for many rare conditions is simply not provided in Ireland.

The National Centre for Medical Genetics (NMCG), based at Our Lady's Children's Hospital, Crumlin, seeks to provide a comprehensive service for all patients and families in the Republic of Ireland affected by or at risk of a genetic disorder. Unfortunately, however, the NCMG simply does not have the resources to do so. It is currently 80% below the Royal College of Physicians recommended staffing levels. It cannot meet the required model of care to people affected by rare diseases due to insufficient geneticist and genetic counsellor: patient ratios. There are also insufficient administration and laboratory staff, and no specific disease registries. Although the NCMG labs are accredited and produce results of the highest quality, the scope of the service is restricted and reporting times do not meet international standards. At the moment there is no training programme for Clinical Geneticists or for Genetic Counsellors.

Promoting the development of medical genetics services in Ireland is a key objective for GRDO and we appeal to all in the rare disease community to voice their concerns on this topic. Anyone affected by a rare disease who has direct experience of the negative impact of inadequate medical genetics services is welcome to get in touch by email with Kathy Tynan, kathy@grdo.ie

RARE DISEASE RESEARCH EVENT | 26 March 2012

On Monday 26 March, the Health Research Board (HRB), on behalf of the National Steering Group on a rare disease plan, hosted an exciting event entitled *Insights into Successful Rare Disease Research*. The workshop brought together national and international speakers and Irish researchers. Keynote presentations on the elements involved in successful research in the area of rare disease were followed by a panel discussion on the barriers, solutions and opportunities in rare disease research.

Speakers included: Dr Helen McAvoy, IPH, representing the National Rare Disease Steering Group; Professor Orla Hardiman, Trinity College Dublin; Dr José Maria Millan, Deputy Director of CIBERER, Valencia, Spain; Professor Brendan Buckley, University College Cork; and Céline Hubert, Operational Director, Rare Diseases Foundation, France. This meeting was intended to provide an opportunity to input to planning for the future of rare disease research in Ireland and inform deliberations in the area of research for the forthcoming Irish National Plan for Rare Diseases. This event outlined how Ireland can move forward in the area of rare disease research and participate at an international level.

EUROPEAN CONFERENCE ON RARE DISEASES & ORPHAN PRODUCTS | 22-25 May 2012

Ireland was well represented at the recent ECRD European Conference on Rare Diseases & Orphan Products, which took places in Brussels at the end of May. Avril Daly and Kathy Tynan represented GRDO. Among those also participating from Ireland were the Irish Platform for Patients' Organisations, Science & Industry (IPPOSI), the National Centre for Medical Genetics (NCMG), the Children's University Hospital, 22Q11 Ireland, Bee for Battens, Trinity College School of Nursing & Midwifery and UCD School of Medicine. Covering research, development of new treatments, health care, social care, information, public health and support at European and national levels, ECRD offers a unique platform across all rare diseases and all European countries, and brings together all stakeholders - academics, health care professionals, industry, policy makers, patients' representatives. For more information see www.rare-diseases.eu

GRDO CHAIRPERSON ELECTED VICE-PRESIDENT OF EURORDIS



The 2012 Eurordis General Assembly, which took place to coincide with the ECRD Conference, saw Chair of GRDO, Avril Daly, reelected to the Board of Eurordis and subsequently elected as Vice President of the organisation. Avril, who is CEO of Fighting Blindness, represents GRDO on the National Steering Committee established by the Irish Minister for Health in 2011 working on the

development of a framework for a National Plan for Rare Diseases. She is a member of both the Medical Research Charities Group (MRCG) and the Irish Platform for Patients' Organisations, Science and Industry (IPPOSI) and represents Retina International and Retina Europe on the Board of the European Patients Forum.

GENETIC AND RARE DISORDERS ORGANISATION

The Genetic and Rare Disorders Organisation acts as a national alliance for voluntary groups representing the views and concerns of people affected by, or at risk of, developing genetic or other rare disorders. A rare disorder is a disorder affecting fewer than 1 in 2000 people. The European Union Committee of Experts on Rare Diseases (EUCERD) estimates that between 6% and 8% of the population is affected by rare disorders, i.e. 270,000+ people in Ireland. 80% of rare disorders are of genetic origin, with many affecting children. No cure exists for the vast majority of rare disorders.

GRDO is represented on the boards of the following organisations:

- European Rare Disease Organisation, EURORDIS
- European Platform for Patients' Organisations, Science and Industry, EPPOSI
- European Patient Forum
- Irish Platform for Patients' Organisations, Science and Industry, IPPOSI
- Medical Research Charities Group

For more information on our work please contact us at:



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www.grdo.ie

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