WELCOME READERS, TO THE SECOND 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 YOU, OUR READERS WITH SOME USEFUL INFORMATION, WHILE UPDATING YOU ON SOME OF THE WORK WE UNDERTOOK THIS YEAR.

INTERNATIONAL DAY FOR RARE DISORDERS

International Rare Disease Day took place on Saturday 28th February 2009, and to launch the week two young members, Josh Dillon (from Muscular Dystrophy Ireland) and Alison Hyland (from Debra Ireland) helped to get the week off to a great start with Joan Brosnan Walsh (who played Mags in Fair City). Joan was diagnosed with motor neurone disease last year.

GRDO together with the Irish Platform for Patients, Science and Industry (IPPOSI) and the Medical Research Charities Group (MRCG), held a conference on Wednesday 25th February: “Focus on Rare Diseases in Ireland; What is the National Plan?” The conference was attended by patients, medical representatives, scientists as well as policy makers and gave an insight into the different problems people with rare conditions in Ireland face, particularly in the area of access to information, diagnosis and treatment. The need for a national plan to address these difficulties was stressed at the meeting. Speakers included Dr John Devlin, Deputy Chief Medical Officer, Department of Health and Children, Christel Nourisser, General Secretary, European Rare Disease Organisation (EURORDIS) and representatives from patient groups, scientists and industry.

On Rare Disease Day itself, Saturday 28th February, GRDO held a successful family day ‘Patient Care: A Public Affair’ - from 11am-1pm in the Mansion House in Dublin. The Lord Mayor of Dublin, Councillor Eibhlin Byrne hosted the event, while Senator Deirdre de Burca launched “The Voice of 12,000 Patients”, a new publication from EURORDIS. This book presents the findings from surveys on the experience and expectations of 12,000 people across Europe with a selection of rare conditions regarding access to diagnosis and health services. The event was aimed at those who are affected by a rare disease themselves or through a family member or a friend. Over 20 organisations supporting people with rare disorders provided information on support services available in Ireland and work currently being undertaken by Irish groups at national and international level. Young people present enjoyed a musical workshop and face painting.

Similar events were held in Latin America, USA, Australia, Canada, China and throughout Europe. In Europe events were coordinated by the European Organisation for Rare Diseases (EURORDIS) of which GRDO is an active member. EURORDIS represents 300 organisations in 34 countries, covering more than 1,000 rare disorders.

THE EUROPEAN PROJECT FOR RARE DISEASES: NATIONAL PLANS DEVELOPMENT (EUROPAN)

The European Project for Rare Diseases National Plans Development (EUROPAN) is a three-year project of the Programme of Community Action in the field of Public Health (2003-2008), which began in April 2008. The main goal is to develop recommendations on how to define a strategic plan for rare diseases. The recommendations will provide information on the different steps to develop a strategic plan and, more important, it will include priority areas and actions of intervention in the field of rare diseases. The project will collect and disseminate information on EU member states’ national initiatives on rare diseases, on expectations on national plans for rare diseases and on best practices contributing to share experiences, data and effective strategies to address rare diseases.

The National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy, is the project’s lead organisation with 30 countries and EURORDIS participating in the project. The project aims to ensure an inclusive and wide engagement of stakeholders - Ministries, regional and local authorities, health care planners, programme managers, health care professionals, researchers and patients.

EUROPAN developed a recommendation on European action in the field of rare diseases to promote national plans and best practices for rare diseases within EU member states and to link national efforts with a common strategy at European level.

THE COUNCIL OF THE EUROPEAN UNION RECOMMENDATION ON ACTION IN THE FIELD OF RARE DISEASES

The Council recommendation on action in the field of rare diseases was adopted on June 9th 2009. The recommendation calls for concerted action at EU and national level in the following areas:

- Plans and strategies in the field of rare diseases
- Adequate definition, codification and inventorying of rare diseases
- Research on rare diseases
- Centres of Expertise and European reference networks for rare diseases
- Gathering the expertise on rare diseases at European level
- Empowerment of patient organisations
- Sustainability

This adoption marks the culmination of a series of legislative declaratory acts which have paved the way towards the identification of key issues they face, namely support for the social dimension of care and funding for research and patient-led activities.

Chairperson of GRDO Ms. Avril Daly welcomed the recommendation as a positive step towards making rare diseases a public health priority in this country. There is a misconception that few people are affected by rare diseases in Ireland as you take a look at each condition individually the numbers are few. The reality is that 6-8% of the population is or will be affected by a rare disease in their lifetime. This gives us a figure of 140,000 people – which is a significant number by any standard. Most cancers, including all cancers affecting children, are rare diseases, 75% of rare diseases affect children and 30% of patients die before 5 years of age making access to a proper diagnosis and appropriate treatment a public health priority.

‘This recommendation has been a long time coming and The Genetic & Rare Disorders Organisation has represented the needs of Irish patients every step of the way. We hope the adoption of this recommendation will pave the way for the establishment of Centres of Excellence in the treatment of Rare Disorders and underpin the importance of sustaining the vital research needed to produce therapies for unmet medical needs. We are encouraged to see priority given to the empowerment of patients across the Member States to ensure they play a part in decision making that will ultimately affect their quality of life’.

GRDO together with all other interested parties, will follow-up on the implementation of the Council Recommendation at the European and national level.


GRDO SECURES COMMITMENT FROM MINISTER FOR HEALTH MARY HARNEY

GRDO representatives met with Minis- ter Harney on July 21st to discuss a na- tional plan for rare diseases in Ireland with reference to the EU Council recom- mendation on action in the field of rare diseases adopted on June 9th 2009.

GRDO highlighted the need for a gov- ernment representative to work with patient organisations, professionals and other stakeholders in progressing a na- tional plan. The minister was given an outline of GRDO’s history and objectives and a discussion regarding the need for patient input and for the identification of centres of expertise ensued.

Section IV of the Council Recommenda- tion recommends that centres of exper- tise be identified and supported by 2013 and that they are based on a multidisci- plinary approach to care when address- ing rare diseases. People with genetic and rare disorders require an integrated service with a multidisciplinary ap- proach which caters for the diagnosis, medical treatment/referrals, psycho- logical and social support of those af- fected. Given sufficient resources GRDO believes the National Centre for Medical Genetics (NCMG) is best placed to deliver such a service for people with genetic and rare disorders.

The NCMG had to cut back its laboratory and clinical services in 2008 because of budgetary restrictions at Our Lady’s Children’s Hospital. The NCMG has no defined budget, and has seen a drop in staff numbers of up to 25% to the non- filling of vacant posts, maternity leave etc. There are currently four Genetic Consultants in the Republic of Ireland in comparison to eight in Northern Ireland for a smaller population.

NCMG management has been in discus- sions with the National Hospitals Office (NHO) at the HSE about ensuring a prop- erly-structured and safe Medical Genet- ics service for Ireland. The NHO has agreed to initiate a needs assessment to determine the best way to achieve this. NCMG and GRDO are seeking the sup- port of the Minister for Health and Chil- dren to support and progress this vital needs assessment.

Many rare and genetic disorders are specific and require specialised support. The isolation and uncertainty that peo- ple diagnosed with a rare disease face was demonstrated in an interview in the Irish Examiner on 17th July, 2009, with a parent of a child with Angelman’s Syndrome. Remembering the numb- ness after receiving the diagnosis, he said that they did not know what to do: “What we could have done with was a cup of tea and someone to talk to while we tried to come to terms with it all”. This is a role that the voluntary sector can and does fill. Family Support Workers already operate within several of our groups to meet the individual or family from diagnosis or whenever they require it, to support them, to advocate for them when necessary, to be a link person between all the different services that people require and to assist people to live an independent life of their own chooseing.

The EU Council Recommendation on action in the field of rare diseases recom- mends that member states should consider supporting specific disease in- formation networks. Voluntary groups already have access to and disseminate a wide range of information about specific conditions, not only to people affected by those conditions but also to health professionals.

GRDO was pleased to secure a commit- ment from the Minister to progress this plan, and looks forward to working with the Government to ensure the needs of people with genetic and rare disorders are recognised.

DUBLIN BUS COMMUNITY SUPPORT PROGRAMME

The Dublin Bus Community Support Programme awards grants to local charities and voluntary groups based across the Dublin Bus service network. The grants are funded from long-term unclaimed change receipts. Ex-international footballer and Sunderland Chairman, Niall Quinn is patron of the initiative. He presented Judy Windle (GRDO secretary) with a grant of €1000 at a celebratory lunch in Croke Park on 29th September 2008.

CHARITIES ACT, 2009

The Charities Act, 2009 came into law in February 2009. Its provisions will become effective when the Minister for Community Rural and Gaeltacht Affairs introduces commencement orders.

The purpose of the Act is to establish a regulator for the charity sector, to reform the law of charities by ensuring greater accountability and to protect against fraud and the abuse of charitable status. A number of websites provide information on the Act:

- The Department of Community Rural and Gaeltacht Affairs: www.pobail.ie
- The Irish Charities Tax Reform Group: www.charitytaxreform.com

Former Fair City actress Joan Brosnan (Mags Kelly) who has Motor Neurone disease pictured at her home in Malahide with Alison Hyland (5) who has EB and Josh Dillon (8) who has Muscular Dystrophy.

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SAVE THE DATE

International Rare Disease Day, Sunday February 28, 2010

The theme of International Rare Disease Day 2010 is the importance of patient’s roles in the search for therapies and treatments for Rare Diseases.

Since 1999, the European Union has taken measures to fight against rare diseases and their impact on patients’ lives, and has made rare diseases a priority of its public health programmes.

Research represents hope for the millions of rare disease patients who are without cure today. In the last decade huge leaps have been made in genetics and medical technology, raising great hopes from scientific and therapeutic progress.

Rare disease patients are the best partners researchers can ask for – without their participation in clinical trials, bio banks, databases and registries, research would be brought to a standstill.

GRDO in association with the European Organisation for Rare Disorders (EUORDIS) and the National Organisation for Rare Disease US (NORD) are calling on patient groups to work closely with the research community to accelerate the search for treatments for conditions affecting more than 140,000 Irish people today.

Ella having her face painted at the Mansion House

TABLE QUIZ FUNDRAISER

GRDO is extremely grateful to Theatrical Cavaliers Cricket Club who once again designated GRDO as their chosen charity for their 2009 Annual Gala Charity Quiz in April. The Gala Quiz organisers put a huge amount of work into this event, seeking sponsorship, raffle prizes, publicising the event and organising the venue and refreshments. The event was very entertaining under the direction of the masters of ceremonies, Tara Quirke and Mark O’Reagan. Forty tables competed to win great prizes with the top four teams winning a selection of meals, wine, books and theatre tickets. The spirit of the quiz award went to one of our own GRDO teams for modestly coming in last. There were also raffle prizes which included a rugby ball signed by Grand Slam Captain Brian O Driscoll, a bicycle and Late Late Show tickets.

The night resulted in a fantastic €3,936 towards GRDO’s work. Special thanks to the committee of The Theatrical Cavaliers Cricket Club, in particular Fergus Linehan (president), Michael James Ford, Sean Duggan, Bridget Flynn and Luke Griffin for organising the event.

We would also like to thank all those who donated prizes and sponsorship.

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. It is estimated that there are 140,000 people in Ireland with a rare disorder. 80% of rare disorders are of genetic origin.

GRDO is represented on the boards of the following organizations:

European Rare Disease Organisation, EUORDIS

European Platform for Patient Organisations, Science and Industry, IPPOSI

European Patient Forum – Policy Advisory Board

Irish Platform for Patient Organisations, Science and Industry, IPPOSI

Medical Research Charities Group

Disability Federation of Ireland, DFI

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