



GRDO

Genetic and Rare Disorders Organisation NEWSLETTER

Summer 2010

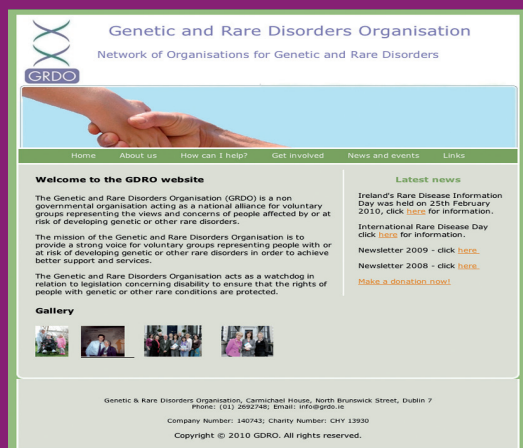
GENETIC AND RARE DISORDERS ORGANISATION

WELCOME READERS TO THE THIRD 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 THE WORK WE UNDERTOOK THIS YEAR.

GRDO'S UPDATED WEBSITE

OUR WEBSITE HAS BEEN UPDATED AND MODERNISED. PLEASE CHECK OUT OUR LATEST NEWS AT www.grdo.ie



THANKS TO YVONNE CUNNINGHAM AND SIOBHAN WINDLE FOR THEIR VALUABLE INPUT.

EUROPEAN CONFERENCE ON RARE DISEASES, KRAKOW 2010

Avril Daly of Fighting Blindness and Barbara Flynn of Friedrich's Ataxia Society of Ireland (FASI) represented GRDO at this year's European Conference on Rare Diseases in Krakow, Poland. There were over 600 delegates at the conference this year from 43 countries representing patients affected by a broad and diverse range of rare diseases.

Dr Andrzej Ry, Director for Public Health at the European Commission, opened the Conference. In his address he declared that the combination of factors involved – i.e. the overall number of patients suffering from rare diseases, the high European value due to the rarity of patients and experts for each rare disease together with the limited access to information of treatment options and drugs available – constitute a challenge that justifies action from the European Union. He highlighted that Member States have until 2013 to adopt rare disease plans or strategies in their own countries based on common policy recommendations.

According to Director of Orphanet, Dr Ségolène Aymé's presentation, National Plans have already been adopted in France, Portugal, Greece, Bulgaria and Spain, and are well under way in Germany, Romania and the UK. The first steps have been taken in other countries, such as Poland, host to the Conference.

"This ECRD 2010 Krakow has served to identify those areas that need better policies in order to fulfil the objectives of the Council Recommendation and to build momentum for national plans and strategies to be implemented across Europe," declared Yann Le Cam, CEO of the European Organisation for Rare Diseases (EURORDIS). "Indeed, the momentum applies to Poland as 20 Polish patient representatives, healthcare professionals and scientists met on the first day of the Conference to sign a Common Declaration to the government calling to establish a National Plan for Rare Diseases in Poland. They also suggested following the European guidelines to accomplish their goals."

Measures to improve accurate diagnosis and early treatment of many rare diseases were presented at the Conference including the coding and classifying of rare diseases and their integration into the World Health Organisation's International Classification of Diseases system. The identification and support of centers of expertise in all European countries and pooling existing expertise through European Reference Networks was also discussed.

The importance of sharing research infrastructures (e.g. databases, biobanks and registries) involving patient organisations in clinical trials is seen as vital to the development of treatments particularly for Rare Diseases. EU Committee of Experts on Rare Diseases (which will include around 50 representatives of all stakeholder groups acting as a sort of 'Parliament' of the rare disease community and following up on work to establish plans) was presented at the conference.

The ECRD series is a unique forum that sees patient representatives from all rare diseases groups from the majority of European countries and further afield gather with healthcare professionals, academics, researchers, policy makers and industry representatives to discuss the most recent rare disease initiatives in the fields of research, healthcare, information and social services. This important event reflected on the recent recommendation from the European Council of Ministers that all member states should have a National Plan for Rare Diseases in Place by 2013.

What was clear from the conference was that although Rare Diseases have become a public health priority at EU

level, this has not filtered down to the health ministries in some member states. The Irish delegation noted that in fact Ireland is lagging way behind on this issue.

GRDO in collaboration with organisations such as the Medical Research Charities Group (MRCG) and the Irish Platform for Patients' Organisations, Science and Industry (IPPOSI) have been liaising with representatives from the Department of Health to try to establish the current position on this issue in Ireland. The MRCG and GRDO are working together to lobby the government to work with us, the patients, in the development of a national plan.

It is hoped that the multi-stakeholder conference 'Europlan' which takes place on December 3 in Dublin Castle will give us all the opportunity to discuss the situation with regard to the recommendations laid out in the European Commission Communication on Rare Diseases on June 9 2009 through Plenary and Workshop presentations. All members are encouraged to attend this important event. For further information contact info@europlan.ie or see the Europlan website www.europlan.ie

RARE DISEASE DAY

In celebration of International Rare Disease Day, the Genetic and Rare Disorders Organisation (GRDO), together with the Irish Platform for Patients' Organisations, Science and Industry (IPPOSI) and the Medical Research Charities Group (MRCG), organised an Information morning on the theme: **"Patients and Researchers: Partners for Life."**



Professor Kieran Murphy (2nd right) with representatives of GRDO and IPPOSI at Rare Disease Day.

Áine Brady TD, Minister for Older People and Health Promotion launched the IPPOSI Report from the 2009 Rare Disease Day meeting entitled:

"Focus on Rare Diseases in Ireland - What is the National Plan?"

Minister Brady referred to the challenges associated with rare diseases due to their low prevalence, but accepted that because 6-8% of the population will be affected by a

Rare Disorder there is a real need for integrated planning in the management of rare diseases.

"I understand that there are between 5,000 and 8,000 distinct rare diseases which, in itself, is a challenge because of the complexity involved in diagnosing and managing all of these distinctive conditions. 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 and highlights the need for integrated planning to give effect to better diagnosis and treatment for these conditions"

The Minister acknowledged the value of patient organisations in providing support and information to individuals and families affected by rare disorders.

"For patients and their carers, sourcing of information can be a particular challenge and this is where the assistance provided by groups represented here today is particularly of value in facilitating this and in enhancing the concept of self-care which is a key to better overall healthcare. Lack of information can make it difficult for patients to access diagnosis and treatment".

She discussed the EU Council recommendation on action in the field of rare diseases adopted in June 2009 and referred to the main recommendations such as that Member States including Ireland develop plans and strategies for rare diseases, that research is promoted, that access to orphan drugs is facilitated, that rare disease patient organisations are supported and that individual Member States should cooperate with their European counterparts on rare diseases.

In conclusion the Minister stated:

"Last year we were talking about the need for a national action plan and I now believe we have the basis to progress this along the lines I outlined earlier. I am encouraged from the meeting last year and also today, that there is strong support from all stakeholders to begin this development. Today's meeting will add to this and I would like to acknowledge the advice and support from IPPOSI, the GRDO and the MRCG as well as the individual disease organisations and industry in helping to bring this to fruition. I am confident that by working together we can tackle the issues and make progress on the National Plan for Rare Diseases".



Young people help launch Rare Disease Day

Ms Jo-Ann Sheridan, a patient with a rare blood disorder, gave her perspective on living with a rare disorder and the difficulty in getting a diagnosis and accessing treatment. Representatives from the patient groups Friedreichs Ataxia and 22Q11 spoke about the value of patient organisations and the research been undertaken in the individual conditions they support.

"As an organization struggling to raise awareness of this recognized but very poorly understood condition, 22q 11 Ireland are appreciative of collaborative efforts between our group and professionals like Professor Murphy. As parents when faced with a rare disorder diagnosis we seek cures or at the very least to improve and enhance the quality of our children's lives. Research conducted today brings treatments in the future. We neither underestimate the value of Prof Murphy's research nor the value of the relationship that has grown between us" Anne Lawlor.

Prof. Kieran Murphy, Professor and Chairman, Department of Psychiatry, Royal College of Surgeons in Ireland and President of Medical Council discussed his research into behavioural aspects of individuals affected by 22Q11 and the value of working with the patient support group and family members. He referred to the Medical Council's new guide on research standards, "Guide to Professional Conduct and Ethics for Registered Medical Practitioners" which is available to view at www.medicalcouncil.ie. He discussed the benefit of European support in sharing information and knowledge about a rare disorder and the value of shared conferences with professionals and family members in attendance.

Representatives from science Prof. Jane Farrar, Trinity College and industry Dr Jon Beauchamp, Alexion discussed the progress of research and the need for continued funding in this area. Ms Avril Daly, Chairperson of GRDO discussed the EURORDIS European Project for Rare Diseases and National Plans Development. She called on the Government to appoint a dedicated representative to progress Ireland's National Plan.

SCREENING

GRDO in association with Muscular Dystrophy Ireland (MDI) and the Irish Platform for Patients Organisations Science and Industry IPPOSI) held a special screening of CBS Films - **Extraordinary Measures** - starring Harrison Ford and Brendan Fraser at Movies@Dundrum on Thursday February 25th. The film is inspired by the true story of John Crowley who tirelessly pursued a cure for Pompe Disease, a rare condition which affected his two young children.

A welcome reception in the lobby gave people the opportunity to chat prior to the movie.



GRDO and MDI representatives at the screening of the movie "Extraordinary Measures" to mark Rare Disease Day.

PRESIDENT McALEESE'S VISIT TO THE CARMICHAEL CENTRE

Our patron President McAleese recently visited the Carmichael Centre. She complemented the ethos of the Centre and the value of groups working together and sharing resources.

To view the Presidents speech in Carmichael Centre see: <http://www.youtube.com/watch?v=QLR1AQVZg8Y>

The Carmichael Centre Autumn 2010 Training and Development Programme may be of interest to your organisation, see www.carmichaelcentre.ie



RARE DISEASE TESTING IS THREATENED BY PROPOSAL TO REVISE REGULATIONS

An EU proposal to revise the regulations governing diagnostic devices could threaten rare disease testing.

Laboratory diagnostic test kits (known as in vitro diagnostic devices or IVDs) are regulated at EU level under a 1998 directive. The IVD Directive is up for revision, and the European Commission has published a public consultation document. One of the proposals in this document would do away with the current exemption for tests developed and used within a single laboratory. This proposal, if accepted, would wipe out testing for almost all rare disorders overnight. So why would the EU propose such a change? Well, IVD manufacturers are concerned that the current scope of this "in-house exemption" is too broad. Manufacturers of IVDs are subject to stringent standards for the design and manufacture of their reagents and devices, while laboratories are free to assemble in-house test kits from inexpensive laboratory reagents. Commercial manufacturers cannot compete with such in-house tests on cost, and complain that there is not a "level playing field". Meanwhile, the lack of oversight of the quality of design and manufacture of in-house tests gives

rise to concerns about the quality of the results produced by these tests. Specialist laboratories, on the other hand, argue that the in-house exemption is essential to enable them to develop highly specialised tests which would never be commercially viable. They also point out that the availability of alternative assays provides an important "second opinion" in cases where the commercial IVD gives an unexpected result.

EuroGentest, a network for promoting quality in genetic testing, maintains that the only level playing field that matters is a level playing field for patients, whether their disease is rare or common. EuroGentest has argued that the in-house exemption must be preserved but that, in the interests of patients, it should be limited to laboratories which are accredited to an appropriate international standard. Accreditation standards require that all in-house tests are properly validated, and ensures that a robust system for quality assurance, quality improvement and continuing external review is in place. If this proposal was accepted, it would be a major step forward for the quality of rare disease testing. I am leading the EuroGentest response to the EU consultation; I will be working closely with GRDO in the coming weeks to produce a robust response on behalf of its members as well

- David Barton, National Centre for Medical Genetics.

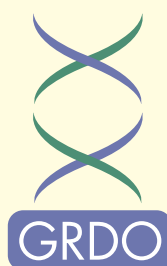
GENETIC AND RARE DISORDERS ORGANISATION

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 2,000 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 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:



Genetic and Rare Disorders Organisation,
Carmichael Centre,
North Brunswick Street,
Dublin 7.

Tel: (01) 269 2748.

www.grdo.ie

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