

Major Rare Disease Conference Hears Ireland Trails Europe in Provision of Genetic Services and of Critical Need for Government Action - Delays of more than 15 months for hundreds of families in accessing genetic services -

- February 29 is International Rare Disease Day with over 80 countries hosting events to highlight rare diseases and their impact on people's lives -

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A major conference took place in Dublin Castle yesterday (February 29) to mark International Rare Disease Day. Calls were made for the incoming Government to radically overhaul genetic services in Ireland and to ramp up implementation of the National Rare Disease Plan. The plan, launched in July 2014, has seen little progress in the development of genetic services in the almost two years since, with several hundred patients continuing to remain in limbo in accessing genetic services for periods of in excess of 15 months.

The conference was organised by the Genetic and Rare Disorders Organisation (GRDO), the Medical Charities Research Group (MRCG) and the Irish Platform for Patient Organisations, Science and Industry (IPPOSI). It heard first-hand contributions from speakers at the vanguard of rare disease care and research in Ireland.

It is estimated that there are over 6,000 rare diseases in existence in Ireland impacting an estimated 300,000-plus children and adults. Approximately 80 per cent of these patients are affected by just 350 rare diseases.

Unnecessary Hardship

Philip Watt, Chair of the Rare Disease Task Force, an umbrella body of patient networks and groups with a specific focus on rare diseases, highlighted how genetic services in Ireland are critically under-resourced:

"The National Rare Disease Plan was launched in July 2014, yet almost two years on, many of its recommendations remain to be implemented. The resourcing of clinical genetics services in Ireland is fundamental to the diagnosis, care and treatment of people with rare diseases. Current clinical genetics staffing levels are among the worst in Europe and between 70-80 per cent below those recommended by the Royal College of Physicians in London. By way of stark illustration, in a recent survey of 14 European countries, Ireland came last in terms of the number of genetic consultants per head of population, while Northern Ireland was ranked in sixth place.

"Right now, my understanding is that there are several hundred families sitting on critically-long genetics waiting lists for over 15 months in clear breach of HSE waiting times. Under European law, patients are entitled to a diagnosis within XX months (Avril to advise if possible, otherwise can be removed?). We need to see information on waiting lists for genetic services being made publicly available so that we can put in place the appropriate resourcing of this crucial speciality. Without access to genetic services, diagnosis is severely delayed preventing access to appropriate ongoing referrals for management and treatment, and vital support services. Meanwhile, a report on the

future of clinical genetics services—known as the Smyth Report—makes key proposals on the future of clinical genetic services in Ireland, yet it remains to be published which begs the question—why?"

Avril Daly, Chair of the Genetic and Rare Disorders Organisations, highlighted the impact the lack of genetic services has for patients in accessing promising new treatments:

"We know that progress is being made across a range of disease areas in the development of new treatments and therapies. Some of these will add years to a patient's life and many will improve the quality of life for people living with disabling conditions, allowing them to work and live independently and contribute to society, which is what all patients want. Yet people living with rare and genetic diseases, one of the biggest health constituencies in the country, are being deprived and delayed access to existing and emerging life-saving and quality-of-life-enhancing medicines because they do not have a proper genetic diagnosis. In real terms delays in diagnosis mean that a patient may go past the point where a treatment can be impactful for them and, if there is no genetic diagnosis at all, they are unaware of their treatment options.

"Every day of the week we are hearing about ground-breaking research, with the ongoing development of clinical trials and emerging gene therapies. And this brings so much hope. However, the rare disease community in Ireland is concerned that policy-makers think genetic therapies and innovative medicines are something to be considered way off in the future. For Irish patients to be able to benefit when these advances begin to be made widely available, we need people with the proper skill sets to be incorporated as part of healthcare delivery now. We welcome the development of a Genetics and Genomics Strategy in Ireland but as it is linked to the development of the National Paediatric Hospital, the completion of which is some years away, we are concerned as to how long it will take to be implemented. Irish people need genetic testing, counselling and care now. We need answers soon today from the incoming Government and not in the distant future. We cannot wait any longer."

New Frontiers for Genetic Services

Dr Kevin Mitchell, Senior Lecturer in Genetics, Trinity College Dublin, and Wiring the Brain blogger, pointed to genetic diagnostics changing the future of patient care:

"We are soon likely to go from a situation where we have a few thousand human genomes sequenced to very many thousands, which will yield huge opportunities in identifying those gene mutations that give rise to disease. Currently, a referral for genetic testing comes at the end of a long and frustrating road for the patient and their family and is usually made as a last resort. The advances we are now seeing will place genetic testing at the front line of diagnostics—the question is how long are we prepared to wait for that to happen here. We know how important it is for patients and their families to have a diagnosis so that they can take the appropriate steps for their care, plan for the future, and so on. If the health service in Ireland is to keep pace with international developments and provide the best care for patients, the role of genetic services will have to be greatly expanded."

Richard Corbridge, Chief Information Officer with the Health Service Executive, referred to the development of electronic health records and how patient journeys are set to become much more seamless:

"eHealth and the introduction of the Electronic Health Record (EHR) across the Irish Health System will have a very positive impact on the treatment and care of all patients, including those with rare diseases. The potential of an EHR to provide a consistent and complete record of a person's clinical history, from cradle to grave, has obvious benefits. For people with rare diseases, where expertise may be spread across the globe, we will be able to link them with the leading clinical experts in their sphere, the latest research opportunities, and up-to-the-minute medical information, diagnostic tools and technology. This will mean faster diagnosis and more timely access to cutting-edge treatment and care. It also means that when people move from one part of the system to the next, that medical and health professionals will have full access to their notes, minimising the risk of information being lost in translation, and time previously spent gathering history being better used to provide treatment and support."

International Rare Disease Day is co-ordinated by EURORDIS, a European alliance of patient organisations, and takes place in more than 80 countries around the globe with the aim of raising awareness among the public, clinicians and policy-makers, of the huge impact of rare diseases on people's lives.

For specific information on rare diseases, emerging treatments and expert services, visit www.orpha.net/national/IE-EN www.grdo.ie or www.raredisease.ie, or for information on International Rare Disease Day, visit www.rarediseaseday.org

For social media updates, visit www.facebook.com/rarediseaseday and twitter.com/rarediseaseday

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Issued by: Don Delaney, d2 communications. Tel.: 087 7933249 / email don@d2communications.ie

About International Rare Disease Day

International Rare Disease Day takes place on the last day of February each year, with the aim of raising awareness amongst the general public and decision-makers about rare diseases and their impact on patients' lives. A disease is classified as rare if it affects fewer than one in 2,000. Four in five are thought to be genetic in origin, with the remainder a result of bacterial or viral infections, allergies and environmental causes, and so on. Since International Rare Disease Day was first launched by EURORDIS and its Council of National Alliances in 2008, it has contributed to the advancement of national plans and policies for rare diseases in a number of countries. For more information, visit www.rarediseaseday.org

About the Genetic and Rare Disorders Organisation (GRDO)

GRDO is a non-governmental organisation that 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, in order to achieve better support and services. It also acts as a watchdog in relation to legislation concerning disability to ensure that the rights of people with genetic or other rare conditions are protected. For more information, visit www.grdo.ie

About the Irish Platform for Patient Organisations, Science and Industry (IPPOSI)

IPPOSI is a patient-led organisation that works with patients, government, industry, science and academia to put patients at the heart of policy and medicines. It holds meetings, discussion groups and training days to promote patient involvement in the treatment and decision-making processes that affect them in areas such as diseases, clinical trials, health technology assessment, innovation, health information and connected health. It makes submissions to national and international public consultations, and prepares reports and briefing papers. A number of think-tanks have been created, conferences are held annually and lunchtime talks are organised with leading experts on issues of concern. For more information, visit www.ipposi.ie

About the Medical Research Charities Group (MRCG)

MRCG is an umbrella body of more than 30 medical research and patient support charities which represents the joint interests of charities specialising in restoring health through medical research, diagnosis and treatment and, where possible, the prevention of disease. Its mission is to generate a dynamic medical research environment in Ireland. For more information, visit www.mrcg.ie