



7 March 2012, Dublin | Commitment to develop a Clinical Care Programme for Rare Diseases welcomed by The Irish National Alliance on Rare Diseases – (GRDO)

The Genetic and Rare Disorders Organisation (GRDO) welcomes the announcement by Minister for Health, Dr. James Reilly TD of his intention to develop a Clinical Care Programme for Rare Diseases at Seanad Eireann (Irish Senate) on March 7.

A rare disease is defined in the EU as a condition that affects fewer than five in 10,000 people. Although precise data on rare disease prevalence has never been collected in Ireland, the Department of Health recognises that between 6% and 8% of the population could be affected by rare conditions (based on estimates by the European Committee of Experts on Rare Diseases, EUCERD)¹. This infers that as many as 350,000 citizens of Ireland² could be affected by a rare condition during their lifetime.

Currently in the Irish healthcare system, those affected by rare diseases can wait over a decade for the correct diagnosis, with a worrying number receiving an incorrect diagnosis initially. This has serious implications for both life expectancy and for quality of life. It leads to inefficient use of already overstretched resources.

A recent survey carried out by GRDO shows that 13.3% of people waited over ten years for a diagnosis and 37.2% received an initial incorrect diagnosis. Any delay in diagnosis or misdiagnosis can result in multiple and unnecessary appointments with doctors and consultants, inappropriate treatments and diagnostic tests, and significant distress.

Many patients worry about the level of awareness of rare conditions among healthcare professionals, including GPs and local community health services. More often than not in the case of paediatric conditions, it is the parents who are the experts informing the healthcare professionals.

A central point of information for patients and medical professionals in the field of rare diseases is an essential step towards alleviating the stress placed on families and pressure on the health service.

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 ongoing 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.

Chair of GRDO, Ms Avril Daly, said of the announcement: “Currently care pathways for patients affected by rare conditions in Ireland vary depending on the condition, the expertise available and where a patient lives in the country. Today’s announcement by the Minister for Health will go a long way towards addressing these issues and, we hope, the current information deficit among patients and healthcare professionals. 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.

¹ Ayme et al, 2011

² Census 2011

“The area of Rare Disease is a health priority throughout the EU. In line with an EU recommendation, Irish patients are working with the Department of Health, HSE, Health Research Board, Institute of Public Health and Health Information, the Irish Medicines Board and the Health Information & Quality Authority on the development of a National Plan for Rare Diseases by 2013. A public consultation will commence on this plan in May of this year. The issue of a National Centre for Rare Disease is fundamental to the development of such a plan and the efficient facilitation of a Clinical Care Programme and we are heartened by Minister Reilly’s comments at the Seanad today which have given hope of better care and support to what is the largest patient constituency in the country.”

ENDS

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About GRDO www.grdo.ie

The Genetic and Rare Disorders Organisation (GRDO) is a non governmental organisation acting as a national alliance for voluntary groups representing the views and concerns of people affected by or at risk of developing genetic and rare disorders.

The GRDO was incorporated in 1988 and its first achievement was to successfully lobby for a dedicated national centre for medical genetics which was established at Our Lady’s Hospital Crumlin in 1994.

The Genetic and Rare Disorders Organisation 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.

Characteristics of rare diseases:

Many rare diseases are complex and multi-systemic, with the possibility of physical, sensory, intellectual, social, emotional and genetic consequences. Rare disease affects the whole family. In addition to the symptomatic person, carers, siblings and other relatives at high genetic risk of the condition are all affected.

- There are between 6,000 and 8,000 rare diseases
- 75% of rare diseases affect children
- 30% of rare disease patients die before the age of 5
- 80% of rare diseases have identified genetic origins
- Other rare diseases are the result of infections (bacterial or viral), allergies and environmental causes, or are degenerative and proliferative

Rare disease patients face common problems:

- Lack of access to correct diagnosis and significant delays in diagnosis
- Lack of quality information and scientific knowledge of the disease
- Heavy social consequences for those affected
- Lack of appropriate, multi-disciplinary healthcare
- Inequities and difficulties in access to treatment and care

- Genetic discrimination

How can things change?

- By implementing a comprehensive approach to rare diseases
- By developing appropriate public health policies
- By increasing international cooperation in scientific research
- By gaining and sharing scientific knowledge about all rare diseases, not only the most “frequent” ones
- By developing new diagnostic and therapeutic procedures
- By raising public awareness
- By facilitating the networking of patient groups to share their experience and best practices
- By supporting the most isolated patients and their parents to create new patient communities or patient groups
- By providing comprehensive quality information to the rare disease community

European regulations and policies in place in favour of rare disease patients:

- EU Regulation on Orphan Medicinal Products (1999)
- EU Regulation on Paediatric Drugs (2006)
- Programme of Community Action in the Field of Public Health (2007-2013)
- EU 7th Framework Programme for Research (2007-2013)

Principles enshrined in the Convention on Human Rights and Biomedicine, the UN Convention on the Rights of Persons with Disabilities and the EU Charter of Fundamental Rights provide guidance on necessary anti-discrimination legislation.