As Ireland Enters New Phase of Global Pandemic, Rare Diseases Ireland Calls for Levelling of Health Playing Field & Urgent Resourcing of Genetic Services in Ireland

New Report Published by Rare Diseases Ireland Finds:

- ***Over a third have waited more than five years for a diagnosis***
- ***Almost one in five of those availing of public genetic testing wait more than two years for results***

To download a copy of "Rare Reality: Living with a Rare Disease in Ireland – Healthcare Experiences", visit www.rdi.ie

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More than a third (37%) of people with rare diseases, who responded to a recent survey by Rare Diseases Ireland (RDI), have had to wait more than five years for a diagnosis of their condition. Indeed, many have been investigated for multiple conditions and seen by multiple specialists along the way. Details of the survey are contained in a new report just published by RDI, *Rare Reality: Living with a Rare Disease in Ireland – Healthcare Experiences*.

For those surveyed who had undertaken genetic testing (64%), the disparity between going public and private was clear with private providers delivering the vast majority (75%) of results within three months, while almost one in five (18%) had to wait more than two years if availing of the public system.

A total of 111 people with rare diseases and family members took part in the survey via an online questionnaire during the month of October and the first week of November 2021[1]. Almost six in ten (59%) of the respondents are living with a rare condition, with just over a third (34%) being parents or guardians of people with rare conditions.

The survey comes following almost two years of the nation's health services dealing with the impact of COVID-19. While this has been clearly necessary to keep people safe, according to RDI, the time has now come to level the playing field across all conditions. Indeed, the Programme for Government made a number of pledges in relation to rare diseases which have yet to see any action, including work on publishing an updated National

Rare Diseases Plan, as well as commitments to establish a National Genetics and Genomics Medicine Network, as recommended in the Smith Report, 2015, and to support the medical genetics service in Children's Health Ireland at Crumlin. In particular, appointment of the HSE National Director of National Genetic and Genomic Medicine Network remains outstanding, while there continue to be vacant consultant geneticist roles at Children's Health Ireland at Crumlin which are contributing to increased genetics waiting lists.

The survey results provide clear evidence if ever it was needed of the necessity to radically overhaul genetic services in this country in order to provide timely access to a diagnosis so that people can be treated early and appropriately. The alliance warns that if the current situation prevails, people will continue to be subject to unnecessary consultations, tests and inappropriate and potentially harmful treatments, all the while their health is deteriorating and waiting lists are growing, adding more burden to an already overstretched healthcare system.

Survey Highlights

Among the key takeaways from the survey are:

- **Diagnosis:** while more than nine in 10 (92%) of respondents had received a diagnosis with a named rare condition, the time to final diagnosis (or time already spent seeking a diagnosis for those that remained undiagnosed at time of survey) is long – 21% had waited two to five years, 14% had waited between five and 10 years and 23% had waited over 10 years. Indeed, getting the final diagnosis of a rare disease first time is a relative rarity itself – more than half (53%) of those who answered have been investigated or treated for three or more conditions on their journey to diagnosis. Almost one in two (48%) reported seeing between three and five specialists on their journey to diagnosis, with one in four (25%) seeing six or more specialists before being diagnosed.
- **Genetic services:** more than three in five respondents (64%) had experience of genetic testing for their rare condition, and in a little over three-quarters (77%) genetic testing provided a definitive diagnosis. Three in four (75%) respondents received genetic test results within three months when accessed privately compared with 38% in the public system. Almost one in five (18%) respondents reported

waiting more than two years for genetic test results through the public system. In contrast, all test results were delivered within two years within private services. A little over half (54%) of genetic results were delivered in person and a quarter (25%) by phone. Just over a fifth (23%) received their potentially life-changing results via letter or email.

- **Personal impact:** reflecting the complex nature of these conditions, respondents reported, on average, that five areas of their health were affected, with neuromuscular/neurology (43%), ophthalmology (35%), rheumatology (30%), immunology (26%), cardiology (25%), gastroenterology (25%) and speech and language (25%) being the most common areas of health affected by their rare condition.
- **Treatment:** of the respondents that received treatment directly related to their rare condition (57%), the vast majority (84%) were provided treatments to relieve symptoms, and just under half (48%) had treatment to slow down or stop deterioration of their rare disease. A small number had a treatment to prevent (5%) or cure (11%) the condition. Almost one in five (19%) reported that treatment was not available in Ireland due to waiting lists, with 19% identifying HSE infrastructure/processes and 18% a lack of reimbursement as the principal reasons.
- **Specialist care:** just three in five (62%) have a specialist managing their care with expertise in their rare condition. Almost one-third (31%) indicate that there is no specialist in Ireland with the requisite expertise. Care is provided through an expert-led multi-disciplinary care centre for just 44% of respondents. A number of respondents had travelled internationally for diagnosis (13%), assessment (18%) and/or care (11%).
- **Medical visits:** one in three (34%) reported regularly attending four or more different types of specialist hospital clinics to manage their condition. One in four (27%) attend just one. Typically, a quarter (24%) of respondents engage with hospital/GP services 1-3 times per year, while a slightly higher number of respondents (30%) engage with those services more than 10 times annually. When it comes to other healthcare professionals and services in the community, almost half

(44%) of respondents engage with them 1-3 times per year, and almost one in four (23%) engage with them more than 10 times annually.

- **COVID aftermath:** as a result of the pandemic, almost one in three (28%) felt that their rare condition deteriorated substantially, with 33% indicating similarly for their physical health, 43% for their mental health and 47% for their emotional health.

Unnecessary Delays

Vicky McGrath, CEO, Rare Diseases Ireland, comments:

"This report follows a period of the health system devoting, appropriately, significant resource and attention to the impact of COVID-19. However, we now need to get the rest of our health services back on track, and level the health playing field.

"Rare conditions are complex. Getting a timely and accurate diagnosis is far from easy and must be prioritised for action. Patients are waiting too long for a diagnosis and are adding to waiting list numbers as they are referred from consultant to consultant, undergoing a battery of investigations and tests. Delays mean that not only does the person not get the care that they need, but that they may also be provided with unnecessary and potentially harmful treatment. Care is fragmented with patients or their guardians having to link up services and specialties themselves.

"It is clear that resourcing of genetic services is key to addressing the tortuous diagnostic journey for many. Strategic development of genetic services must be a priority for leadership at the Department of Health and HSE. Their repeated failure to allocate adequate resources and appoint someone to the role of HSE National Director of National Genetic and Genomic Medicine Network is enormously frustrating.

"Likewise, the inability of the HSE to attract candidates to fill open permanent and locum consultant geneticist positions at Children's Health Ireland at Crumlin is increasing the burden on those currently working in genetic services. Only four of the six available consultant geneticist roles at the hospital are currently filled. The effect of these vacancies is clear with 4,029 on the clinical medical genetics waiting list as of December 23 last, up one third from 3,021 on December 19 2019 figures. [2]

“Equally, where expertise in a particular condition is not available in Ireland, patients should be given the opportunity to avail of expert care abroad. The recent development of European Reference Networks to provide access to expertise, research and education for rare conditions is a very welcome development. We now need to ensure that people in Ireland who need it can access this European expertise through the HSE.”

Prof Eileen Treacy, National Clinical Lead, National Rare Diseases Office, HSE comments:
“The ‘Rare Diseases Ireland’ Rare Reality survey reports the ‘real world experience’ of 111 individuals and families in Ireland living with rare diseases, during the COVID-19 pandemic.

“In parallel to this survey, improved care co-ordination between Health Care Providers, improved access to specialists and enhanced treatment opportunities were also highlighted as the top three highest unmet needs by the Rare Diseases community across EU Member States to be addressed by 2030 according to the Eurordis supported ‘Rare2030 Foresight Study’. The development of national care pathways for individuals with rare diseases were seen as a key recommendation of this foresight study.

“In response to these well recognised challenges and previous national consultations, the Rare Diseases Clinical Programme published a Model of Care for Rare Diseases in 2019. A number of the recommendations from this Model of Care have been implemented or in the process of implementation through the HSE, HRB and National Rare Diseases Office.

“A major recent development is that 5 major Irish academic HSE Hospitals were approved to lead Irish Rare Disease Networks to join 15 new (additional) European Reference Networks ERNs) effective since January 1, 2022. This is a very positive development for individuals and families affected with rare diseases for the enhanced cross border provision of highly specialised/rare disease services to facilitate the mobility of expertise, virtually or physically and develop and share information, knowledge and best practice and foster developments of the diagnosis and multidisciplinary treatment of rare diseases. Implementation and integration of these centres of expertise into the ERNs will be pivotal to and drive innovation, training and clinical research for highly specialised care in collaboration with the established momentum of the ERN model which incorporates the most talented clinicians and investigators in the European Community.

“In addition, a model for rare disease specific care pathways (including diagnostic pathways) has been developed by the National Rare Diseases Office team in collaboration with ‘Rare Diseases Ireland’ and national Rare Diseases speciality leads which are specifically aimed to enhance integrated care coordination.

Impacting Thousands in Ireland

An estimated 300,000 people in Ireland live with a rare disease, while the figure worldwide is 300 million people. 72% of rare diseases are genetic in origin. Around 70% of rare diseases start to show up in childhood and at least 3.5% of children in Ireland are diagnosed with a rare disease by age 15. Almost six in ten childhood deaths here are associated with rare diseases.

For more information on rare diseases in Ireland, visit www.rdi.ie

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Notes

1 The online survey was completed by 111 eligible participants (people aged 18 years or older living with a rare condition or family members or carers of people with rare conditions) from across every health region in the Republic of Ireland and from Northern Ireland (n=2). The survey was completed in October and early November 2021.

2 Figures from National Treatment Purchase Fund data - https://www.ntpf.ie/home/outpatient.htm

About Rare Diseases

A rare disease is defined as a disease that affects fewer than one in 2,000 individuals in Europe. Between 6,000 and 8,000 individual rare diseases have been identified to date, and while individual diseases and affected patients may be rare, collectively rare diseases are many. Rare diseases are chronic, progressive, degenerative and often life-threatening. They carry with them significant levels of morbidity and disability, including cognitive, developmental, intellectual, mental, physical and sensory challenges, and are a significant contributor to early foetal loss and perinatal mortality, as well as infant and child mortality.

About Rare Diseases Ireland

Rare Diseases Ireland (RDI) is the national alliance of voluntary groups representing people affected by, or at risk of developing, a rare condition. RDI's mission is equity for people with rare conditions in Ireland – this means equitable access to diagnosis, treatment, care and opportunity.