Rare Diseases Ireland address to Joint Committee on Health Wednesday, May 26th 2021

Good Morning - My name is Vicky McGrath. I am the CEO of Rare Diseases Ireland. We are the national alliance for voluntary patient-led organisations for people affected by or at risk of developing rare diseases. Our vision at RDI is **equitable access to diagnosis, treatment, health, social care and opportunity**.

Thank you very much for the opportunity to speak with you this morning on the topic:

'Improving the lives of those affected by rare diseases and their families.'

Today I am joined by Dr Sally Ann Lynch, Consultant Clinical Geneticist from Children's Health Ireland (CHI) at Crumlin and Avril Daly, CEO at Retina International and VP at EURORDIS-Rare Diseases Europe. Avril also has first-hand experience of living with a rare condition.

The challenges of living with a rare condition are acknowledged in the Programme for Government published in June 2020, ***'many rare conditions are complex, and their impacts are severe on the patients living with them. At times, it can be difficult to access appropriate medications and technology.'***

We are asking for your support today to press for delivery on the promises made to the rare disease community in the Programme for Government, in particular those promises around genetics and genomics and an updated national rare disease plan. Almost 1 year on from publication of the Programme for Government, there is no progress with these promises. As a priority we must see allocation of resources at the Department of Health to bring leadership and accountability to these commitments.

Before proceeding any further however, let me provide some background data on rare diseases in Ireland.

- A rare disease is defined as a condition that affects less than one person in every two thousand people. This is the definition used in Europe.
- There are over six thousand individual rare diseases that we know of, with more being described on a daily basis.
- It is estimated that there are **three hundred thousand** people living with rare conditions in Ireland. This compares with one hundred and seventy thousand people living with or recovered from cancer today in Ireland.

INDIVIDUALLY RARE; COLLECTIVELY COMMON.

- Three hundred thousand people, that is a little over 6% of the population that are living with a rare condition. More than one in every twenty people is living with a rare condition in Ireland today.

- 4% of children have received a diagnosis of a rare condition by the time they reach 16 years of age. Consider this, of the 61,500 18 year olds in their leaving cert year this year, 2,500 have already been diagnosed with a rare condition.
- Paediatric rare conditions have an enormous impact - 58.6% of deaths in those under the age of 16 are due to an underlying rare condition.

Rare conditions are characterised by a wide diversity of symptoms and signs that vary not only from condition to condition but also from person to person with the same condition. Rare conditions are chronic, progressive, degenerative and often life-threatening.

While many people with rare conditions live with disabilities such as motor, sensory or intellectual impairments, for others there are no visible external signs that they are living with a rare condition. In many cases however the burden of living with an invisible rare condition is as challenging as the burden faced by those with visible disabilities. Planning and managing care and medical appointments, managing household chores, accessing education and employment opportunities, social interaction and building relationships are all challenges for people living with rare conditions.

People living with rare conditions are often described as having complex medical needs. Complex medical needs are not an excuse for poor and disorganised provision of care and support. Complex medical needs are the challenges that are driving enormous innovation in this space today - be it whole genome sequencing, orphan medicines, gene therapies, digital living spaces or artificial intelligence – much innovative work is being undertaken today across the globe to address the needs of the rare community. One might think that Ireland should in fact be a global leader in this space – after all we have a huge industrial footprint in pharma, medtech and ICT.

Innovation in the rare disease space is not just about therapies and treatments. Innovation is also about how and where care is delivered, in particular pooling the resources across Europe to ensure the best outcomes for all citizens of the EU living with rare conditions. As citizens of not just Ireland but the EU is it unreasonable for us to expect to have access to the same care, treatment and opportunities as our fellow EU citizens in countries such as France, Germany & Denmark.

The National Rare Disease Plan, originally published in 2014 must be updated and overhauled to account for the transformations that have taken place in rare disease care and delivery over the last 8-10 years. The revised National Rare Disease Plan must also take account of Slánitecare, a transformation program that was not even under consideration in 2014.

As a matter of urgency we are seeking a commitment from the Department of Health to prioritise people living with rare conditions as we emerge from the coronavirus pandemic. People living with rare conditions deserve to benefit from the enhancements in community care spoken of in Sláintecare. It is no longer acceptable to think that the rare community can wait. There are 300,000 people living with rare conditions that must

be prioritised. If we can build a healthcare system that meets the needs of the most vulnerable in our society it will be a healthcare system that meets the needs of everyone in the country.

One of the greatest challenges for people living with a rare condition in Ireland today is the diagnostic journey. Diagnosis is often delayed for many years and involves multiple specialists. The diagnostic journey is associated with significant hardship for patients and families alike, with unnecessary appointments, referrals, tests and interventions, loss of income and education or career prospects and delayed opportunities for the correct treatment. Would we accept delayed diagnosis and treatment in other specialties? We all know what a delayed diagnosis for cancer patients means, yet it is accepted as 'normal' for a rare diagnosis to take several years.

72% of rare conditions are genetic in origin. Genetic testing, genetic consultation and genetic counselling are thus the fundamental building block for diagnosis of rare conditions. In spite of this in Ireland we have a hugely under-resourced genetics infrastructure. The Clinical Genetics service in CHI at Crumlin provides a diagnostic, counselling and clinical genetic testing service for children and adults affected by or at risk of a genetic condition. The service is the sole provider of general genetic service to the population of Ireland. The service cares for and manages families with genetic conditions.

The HSE's Review of the Clinical Genetics Medical Workforce in Ireland, published in 2019, reveals that there is a lack of a co-ordinated genetic testing service in Ireland, due primarily to funding issues. There has been no significant investment in the service in the intervening period. Lack of a co-ordinated genetic testing service has led to poor practice in terms of testing requests and also poor quality foreign laboratories handling Irish samples. There is an inherent risk in this. Additionally some people, including those without symptoms, have taken it upon themselves to have testing conducted at private lab facilities, sometimes without even a referral from their own doctors. They are desperate to understand what the future may hold for them.

With normal attrition due to retirement and sick-leave and difficulties attracting and hiring qualified personnel there are currently just 3 Genetic Consultants in position in CHI at Crumlin. The HSE's 2019 Review indicates that there should be fifteen. The most visible knock on effect is growing waiting lists. As of March 2021 there were 3,999 people on the waiting lists for Clinical (Medical) Genetics, up from 3,052 just one year earlier. 1,392 of these are children under the age of 16. Typically the priority waiting list is between 15-18 months and routine referrals wait over 2 years to be seen. As of March there were 941 people on the waiting list for over 18 months, of which 657 were under the age of 16. Our health service is failing the children of Ireland.

Genetic Counsellors see patients with known genetic disorders, under the direction of a Genetic Consultant. The British Clinical Genetics society recommends 8 Genetic Counsellors per million of population. The Department of Clinical Genetics in CHI at

Crumlin currently has about 6 WTE Genetic Counsellors for the entire population of Ireland.

We cannot over emphasise the critical need for genetic services in Ireland. Day-to-day management of people living with rare conditions, and the health system as a whole, would benefit from early accurate diagnosis. There are huge risks in the current system. People are sitting on waiting lists unnecessarily as they seek a diagnosis. There is a potential for unqualified personnel to provide diagnosis and treatment based upon difficult to interpret results, and patients taking matters into their own hands seeking testing and diagnosis outside of the normal healthcare environment. There are also enormous opportunities being missed – people living with rare conditions are missing out on early access to new therapies and potentially enrolment in clinical trials for novel therapies.

In the short term we must think creatively about addressing some of the bottle-necks that exist in genetic services today with a view to building a best-in-class robust genetics service for future generations. Genetics are not just the future of healthcare, they are here now. We need genetic services today that will allow the Irish people to benefit from innovations that are with us now, and prepare us for those that will be with us in the years ahead.

In summary, we are asking for your support today to ensure that the government delivers on its promises in the Programme for Government. The very first step we believe is allocation of resources at the Department of Health to bring leadership and accountability to update and development of a new National Rare Disease Plan for Ireland and delivery of world class genetic and genomic services. In the past we have seen the Department of Health provide the leadership necessary to deliver on cancer strategies and plans for the country. It is not unreasonable to expect a similar commitment to the 300,000 people living with rare conditions and their families and friends.