
# RESEARCH REPORT

Rare Diseases Ireland

# RARE REALITY

## LIVING WITH A RARE DISEASE IN IRELAND

## - HEALTHCARE EXPERIENCES -

**-31ST JANUARY 2022 -**

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

## TERMS & ACRONYMS USED IN THIS REPORT

**European Reference Networks –** virtual networks that bring together healthcare providers across Europe to tackle complex or rare medical conditions.

**Genetic testing –** where a person's DNA is tested for changes that could be linked to a rare disease, often used to help diagnose a condition.

**Genetic counselling –** where a trained expert helps an individual, and often their family, to understand how a genetic diagnosis may affect them and their family.

**HCP –** Healthcare Professional

**ICU –** Intensive Care Unit

## REPORT CONTENTS

# KEY TAKEAWAYS

This Rare Reality report reveals the shared experiences of those living with rare conditions and identifies solutions that would help.

- Rare disease **diagnosis must be improved**, including timely access to genetic services and the manner in which diagnoses are delivered; genetic services urgently need resources.
- A **lack of awareness** about rare conditions, particularly among GPs and emergency doctors, can lead to delayed diagnoses and treatments; education and appropriate referral pathways are needed for all healthcare professionals.
- People with rare conditions often experience **fragmented care**, and they or their guardians report needing to link up services and specialties themselves, adding to stress levels.
- The **lived experience is a valuable resource** that should be captured to feed into improved care for all with the same and similar conditions.
- There is a **strong appetite for involvement in research** among those living with rare conditions.

## INDIVIDUALLY RARE - COLLECTIVELY COMMON

## RARE DISEASES IN IRELAND

- A rare disease is a disease that affects less than 1 in 2,000 people.
- Around one in every 17 people lives with a rare disease. An estimated 300,000 people live with rare diseases in Ireland. [1]
- There are 6,000 to 8,000 defined rare diseases. 72% of rare diseases are genetic in origin. [2]
- A rare disease does not just impact the affected individual. A rare disease has implications for everyone in the immediate family unit and the wider family too - grandparents, aunts, uncles, cousins and second cousins.
- 70% of rare diseases start to show up in childhood. [2]
- At least 3.5% of children in Ireland are diagnosed with a rare disease, a life-long condition, by age 15. [1]
- Almost six in ten childhood deaths in Ireland are associated with rare diseases. [3]

[1] National Clinical Programme for Rare Diseases. Model of Care for Rare Diseases. https://www.hse.ie/eng/about/who/cspd/ncps/rare-diseases/resources/model-of-care-for-rare-diseases.pdf (Accessed Jan 2022)
[2] Nguengang Wakap S, Lambert DM, Olry A, *et al*.Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *Eur J Hum Genet.* **28** (2):165-173 (2020).
[3] Gunne, E., McGarvey, C., Hamilton, K. *et al*. A retrospective review of the contribution of rare diseases to paediatric mortality in Ireland. *Orphanet J Rare Dis* **15**, 311 (2020).

## THE 'RARE REALITY' – WHY IT MATTERS

Hundreds of thousands of people live with rare conditions in Ireland. Their needs are many and varied, including healthcare, education, employment and housing.

This is the first of RDI's "Rare Reality" survey series. The series will address different aspects of the challenges of living with a rare condition. Surveys are administered on-line, with people invited to complete the survey via RDI's website, social media channels and e-mail. Respondents also have the option to complete surveys via phone.

In this first Rare Reality survey, we wanted to take the pulse of how people living with rare conditions and their loved ones feel about the provision of healthcare for them in Ireland. Their healthcare needs are varied and, in many cases are frequent, intense and unmet. We wanted to find out what works, what doesn't work and how that affects the lived experience of people affected by rare conditions. By gathering this information now, we hope to identify ways to improve care and measure progress into the future.

## RARE DISEASES ARE COMMON

A rare disease is defined as a disease that affects fewer than 1 in 2,000 individuals in Europe. Between 6,000 and 8,000 individual rare diseases have been identified to date. While individuals living with each identified disease may be rare, collectively they are many. An estimated 300 million people worldwide live with a rare disease, including 300,000 people in Ireland.

Rare diseases are associated with multiple impairments including cognitive, developmental, intellectual, mental, physical and sensory, or some combination of these symptoms. Rare diseases are chronic, progressive, degenerative and often life-threatening. Rare diseases represent a significant burden on individuals who live with them, on families who are affected by them and on society, not just as a result of the direct costs associated with healthcare but also due to loss of economic activity.

## LIVING WITH A RARE DISEASE IN IRELAND

This Rare Reality survey provides insights into the healthcare experiences of people living with rare conditions. 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. Many respondents were happy to share their experiences, and a number made suggestions about how to improve care and access.

Almost six in ten (59%) of the respondents are people living with a rare condition, while just over one third (34%) are parents or guardians of people with rare conditions. Other respondents included spouses and other family members. Nearly one in four of the people living with a rare condition (24%) are aged 15 or under, while almost half (46%) are aged 16 to 49 years.

We asked whether any of 26 areas of the person's health were affected by the rare condition. Brain/spinal-cord/ nerves were the most commonly affected. On average, survey respondents reported that five areas of health were affected, highlighting the complexity of rare diseases and their impacts on the person.

**5 the average number of areas of health affected by rare condition**

The complexity of rare diseases translates into complex demands on the healthcare system. Only one in four (27%) attend one specialist clinic for management of their condition. One-third (34%) of respondents regularly attend four or more different types of specialist hospital clinics for complete management of their rare condition.

**34% attend 4 or more different specialist hospital clinics for regular on-going care**

Typically, one in four (24%) respondents engage with hospital/GP services 1-3 times per year and almost half (44%) engage with services in the community 1-3 times per year, However, three in ten (30%) use hospital and/or GP services and almost one in four (23%) use community services more than 10 times per year.

**RARE DISEASES ARE COMPLEX**

| Aspects of health affected | |
|---|---|
| Heart (Cardiology) | 25% |
| Breathing/ lungs (Respiratory) | 24% |
| Hormones/ diabetes (Endocrinology) | 20% |
| Brain/ nerves/ spinal cord (Neurology & Neuromuscular) | 43% |
| Muscles/ ligaments/ joints/ inflammatory (Rheumatology) | 30% |
| Bones/ joints (Orthopaedics) | 23% |
| Skin (Dermatology) | 20% |
| Stomach/ Digestion/ Liver (Gastroenterology/ Hepatology) | 25% |
| Kidneys (Nephrology) | 12% |
| Cancer (Oncology) | 4% |
| Blood (Haematology) | 12% |
| Reproductive | 6% |
| Metabolic | 10% |
| Vision/Eye (Ophthalmology) | 35% |
| Hearing (Audiology) | 21% |
| Mental Health (Psychiatry) | 21% |
| Behavioural difficulties | 17% |
| Speech and Language | 25% |
| Sensory Processing | 20% |
| Intellectual disabilities | 19% |
| Vascular | 13% |
| Urological (uro-recto-genital) | 6% |
| Craniofacial & ENT | 16% |
| Birth defects/ Congenital malformations | 6% |
| Immune system (Immunology) | 26% |
| Dental | 19% |

Over the last 5-7 years most respondents report that the experience of care for their rare condition has remained about the same, and the majority feel that awareness of rare diseases has not increased in this period.

Participants reported a marked deterioration of health since March 2020, the start of the Covid-19 pandemic. Only 19% of respondents felt that physical health and health due to the rare condition remained unchanged, while only 12% and 9% felt their mental and emotional health respectively were unchanged.

Meanwhile, 28% of respondents felt that their rare condition deteriorated substantially during the pandemic and 33% felt their physical health deteriorated substantially. 43% felt mental health deteriorated substantially over the period, and 47% felt emotional health deteriorated substantially.

Despite hospitals being effectively ‘closed’ for routine/elective care for prolonged periods since the start of the pandemic, the survey showed greater dissatisfaction with community-based care than hospital-based care during this time. Respondents commented on the lack of supports during the Covid-19 pandemic and how they saw regression in that period, on how the emphasis on PCR testing led to delays when being seen, on the increased isolation during the pandemic and that while remote consultations can be helpful, they must not become the default.

*"As someone at risk of respiratory infection and in need of rapid treatment for respiratory infection, delay to GP care was not good as could not access care until COVID respiratory illness had been ruled out.*

*"Phone consultations helped reduce unnecessary travel. But too many times COVID was used as an excuse not to be seen when I needed to be.*

*"We were previously linked with the early intervention team [….] prior to March 2020 but we have not seen anyone in person since then. They were only allowed see 'assessment of needs' and then Progressing Disability Services was the next excuse. Our daughter has regressed in many areas during this time and has not progressed as expected without access to physio/OT/SLT and psychology. We have been allocated a new CDNT but have still not seen anyone (Oct 2021 - 19 mths).*

## FAST AND ACCURATE DIAGNOSIS NEEDED

Obtaining a timely and accurate diagnosis is enormously important. Delays in diagnosis or receiving an incorrect diagnosis not only means the person may not get the care they need it may also lead to unnecessary and potentially harmful treatments. Delayed and inaccurate diagnoses also erode trust in the healthcare system and increase frustration and worry among those living with the condition and their families.

Of the respondents who answered, 42% reported initially consulting a doctor more than 10 years earlier, with the same percentage initially consulting their doctor within the previous 2 to 10 years. For some people symptoms start early in life, for others symptoms appear later. For most first symptoms mark the start of life with a life-long rare condition; cure is not the expected outcome.

The time to final diagnosis of a rare condition (or until now, if no definitive diagnosis has been made yet) is long - 21% waited two to five years, 14% waited between five and ten years and 23% waited more than 10 years. Some people reported having symptoms since birth or infancy, but the condition was not diagnosed until later in their childhood or even as an adult.

**37% waited over 5 yrs for diagnosis**

## THE DIAGNOSTIC JOURNEY IS LONG AND CONVOLUTED

*"I suffered from issues possibly from birth looking back at things. Was not getting answers for over 20 years prior to diagnosis.*

*"Symptoms from birth. Diagnosis after age 45 when syndrome finally described in medical publications.*

Getting the final diagnosis of a rare disease first time is a relative rarity itself - more than half (53%) of respondents have been investigated or treated for 3 or more conditions on their journey to diagnosis.

**53% assessed and/ or treated for 3 or more conditions on journey to diagnosis**

The vast majority who answered (more than 92%) had received a diagnosis with a named rare condition, but some were still awaiting confirmation. One respondent commented:

*"Unsure, diagnosed wrong twice. Personally still searching for confirmation.*

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.

**73% saw 3 or more consultants in different specialties on journey to diagnosis**

*"In spite of multiple hospital admissions, routinely seeing ENT, respiratory, asthma clinic, allergy specialist, amongst others, no one suggested an overall neurological exam (it was ultimately his SLT recommended it). Felt this should have been done much earlier.*

*"Receiving a diagnosis provides overwhelming relief for both the person and their family. It is a long process and a worrying time as you fear being unable to be treated without a diagnosis.*

## TIME FOR CHANGE

- We need greater awareness among GPs about referral and support pathways for people with possible rare diseases.
- Early referral for genetic testing could speed up diagnosis.
- Experts from different disciplines and areas of healthcare need to communicate and refer to each other more to improve diagnosis - look at the entire person, not just one aspect.

***"Better medical tools needed in Ireland. Awareness and more specialised doctors needed also."***

## GENETIC SERVICES REQUIRE RESOURCES URGENTLY

Genetic testing is a cornerstone of diagnosis and treatment for many rare conditions.

Genetic testing was the route to definitive diagnosis for many. The majority - 64% - of respondents had experience of genetic testing for their rare condition, and in a little over three-quarters (77%) of cases, genetic testing provided a definitive diagnosis.

**77% of those that accessed genetic testing received a definitive diagnosis**

Timeliness of genetic test results and consultations depends greatly upon their route of access to the services. Three quarters (75%) of respondents received genetic test results within three months when accessed privately compared to just 38% in the public system. Almost one in five (18%) 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.

**18% wait over 2 yrs for genetic test results in public system**

Perhaps unsurprisingly, people tended to be more satisfied with genetic services when they were delivered privately.

Receiving the results from genetic tests is a potentially life-changing moment for a person and their family. A little more than half (54%) of genetic results were delivered in person among our survey respondents and one-quarter (25%) by phone. Disappointingly, just over one fifth (23%) received results via a letter/email. Given the potential enormity of the diagnosis, or even disappointment when no definitive diagnosis is available, letters and email are not the best way of providing results.

The vast majority (79%) received their genetic test results from a consultant/specialist (split evenly between a geneticist and other specialties). Half of respondents have met a genetic counsellor or consultant, and another quarter would like a referral.

One respondent commented on how they came to know their child's genetic diagnosis through overhearing a conversation in ICU, days before talking with the genetics consultant.

**GENETIC TESTING PROVIDES THE PATH TO DIAGNOSIS**

*"The genetics service is severely understaffed and underfunded but yet more and more individuals are being tested due to the advancement in genetic testing and patient outcomes. The service is in crisis I would go as far as to say. Our option was either wait 2-3 years to have the test results explained to us or see the one private geneticist in two weeks. I feel the aftercare following on from a diagnosis is non-existent. There is no counselling for the parents in respect of delivering often a life changing test result/diagnosis. We feel like there is so much more that could be done but feel completely abandoned by the system.*

*"If you could only get genetic counselling it would be great. The waiting list is unreal.*

*"My parents and siblings all had to be genetically tested - we had little information or support. We got most of our information off the internet.*

*"I was given very little information with diagnosis. I wasn't even fully aware genetic tests were still outstanding. Knew they had "ran bloods" and had gotten some results back, but wasn't aware more (genetics) were outstanding. Therefore the diagnosis was a complete bolt out of the blue. I was on my own with my young son present & demanding my attention whilst receiving this diagnosis. I was tested on the day and not informed when results came back. Found out incidentally when a different consultant mentioned it. In spite of many questions we still have not seen a genetic counsellor.*

## TIME FOR CHANGE

- Early referral for genetic testing could speed up diagnosis.
- Make it easier for people with a family history to be referred for genetic testing by their GP.
- Deliver genetic testing results in a manner that takes into account the psychological and emotional impact and the individual's need for more information.
- Provide more resources for genetic counselling to improve access.

***"Access should be quick as critical decisions that will impact the life of the individual with the condition and the family need to be made ASAP."***

## EDUCATION BUILDS TRUST AND SAVES LIVES

Being informed about a rare disease is important when living with one. Specialist clinics, patient organisations and online fora (such as closed Facebook groups and blogs) are key sources of information for many living with rare conditions and their family members.

The majority of respondents in our survey felt they themselves had sufficient information about the rare condition they are dealing with. Around one-third of respondents reported that they have (or the person with the rare condition has) an alert card or ID bracelet that provides information about their condition in case of a medical emergency.

However, there was a strong sense among respondents that healthcare professionals do not have sufficient information about rare conditions in general, and about their rare condition and specific symptoms and needs. This lack of information was particularly pronounced in primary care (e.g. GP clinic), emergency care (ambulance and EDs) and social care.

When asked about information provided by healthcare professionals, 29% expressed satisfaction with information provided pre-diagnosis. This figure increased at time of diagnosis to 38%, with a marginal drop to 37% following diagnosis.

**29-38% satisfied with information provided by healthcare professionals**

Interestingly, the perceived lack of awareness and knowledge among different groups of healthcare professionals correlated with lower trust in those groups of healthcare professionals. This suggests that increasing awareness and education among healthcare professionals about rare diseases could build greater trust with affected individuals and their families and care providers.

*"There is still a gap in knowledge especially at primary care (GP) relating to rare diseases and this can cause a delay in getting the appropriate diagnosis.*

*"Information and support is in the main provided by support group and linking in with European and international clinicians and researchers. Things could be improved if the rare disease group's expertise was utilised.*

*"When I have to attend a new medical professional they always have to Google my condition. It's very frustrating and I find they only want to treat what is relevant to their area and not my whole body. Anyone living with a condition knows it will affect different areas of your body so why professionals do not take this into account is beyond me.*

**KNOWLEDGE AND TRUST GO HAND IN HAND**

## TIME FOR CHANGE

- Include more education on rare diseases in the training of all healthcare professionals.
- Incorporate training from patient organisations and affected individuals into healthcare professional training curriculums.
- Provide information about rare conditions in dentists, opticians and allied health professionals.
- Increase support for patient-focused conferences and information groups.

***"Last year I asked for a letter from my consultant to carry on me with relevant details and care needed in an emergency situation."***

## INTEGRATED PERSON-CENTRED CARE

A major challenge for many people with rare diseases is obtaining care from an expert-led multi-disciplinary team that meets their needs.

We found that only 62% of respondents receive care from a specialist consultant with expertise in their condition and only 44% of respondents said that this care is provided through an expert led multi-disciplinary care centre. Almost one third (31%) of respondents indicate that there is no specialist in Ireland that has any expertise in their rare condition. While not surprising, given the number of rare conditions and the ultra-rare nature of many of them (less than 1 per million population), what is concerning is that many of these have also not been given the opportunity to avail of expert care in locations outside of Ireland.

**31% no medical expert for the rare condition in Ireland**

Small numbers have travelled internationally for diagnosis (13%), assessment (18%) and/or care (11%). The EU's Cross Border directive is designed to address the needs of people requiring access to specialist expert care that is not available within their home country, but it is not delivering on the ground for individuals and families in Ireland, with some incurring significant out-of-pocket expenses that are subsequently not reimbursed.

*"Even though the neurologists in Ireland could not answer our questions or give a clear pathway for care we were deemed ineligible for payment/reimbursement of €18,500 when we took our neurologists advice and travelled to London to a specialist doctor and had specific tests done there.*

We asked participants about "treatments" that they have received, using a very wide definition, as follows: any type of medicine, therapy, surgery, or other medical treatments directly related to the rare condition. Just over half of respondents knew of one or more treatments for their rare condition and almost half of respondents reported being satisfied or very satisfied with the effects of any treatment they had received.

*"Ongoing it's tough to have to pay for GP visits, consultant visits and drug payment scheme. List of life long illnesses to narrow to qualify*

*"Specialist knowledge and therapies in Ireland would be life changing for our child*

*"The medication I take keeps me alive. Some of the medications are not covered by the HSE in Ireland and the costs are exorbitant.*

*"A dedicated multi-disciplinary centre is needed*

## ORGANISE CARE AROUND NEEDS OF THE INDIVIDUAL

Of the respondents that received a treatment (57%), the vast majority (84%) were provided treatments to relieve symptoms, and just under half (48%) had treatment to slow down or stop deterioration. A small number had a treatment to prevent (5%) or cure (11%) the condition.

Almost one in five (19%) reported that approved treatment was not available in Ireland due to waiting lists, HSE infrastructure/processes (19%) and lack of reimbursement (18%).

The vast majority (81%) of those that accessed treatment for their condition received it as part of their routine care provided by their specialist/GP. Only 6% accessed treatment through the treatment abroad scheme, 8% via a clinical trial and 4% via a managed access/compassionate use programme.

Only one in five (22%) of respondents said that there is a documented care pathway for their rare condition, with a smaller number (15%) indicating that they had a personalised care plan in place that describes the specific health services and supports that are needed for them.

**22%** **documented care pathway**

**personalised care plan** **15%**

*"At diagnosis, there was no clear pathway. Now in full time care there is a clear personalised care plan. Very satisfied with the level of care in the care facility.*

Almost half (48%) were dissatisfied or very dissatisfied with information and support received on the rare condition and plans for their future care.

**73%** **care is not coordinated**

A large majority of respondents (73%) do not feel that care is coordinated effectively. Almost half (47%) do not believe healthcare professionals work as a team. Only 41% feel that timing and frequency of appointments are convenient and 55% want more appointments to be provided locally.

**47%** **feel healthcare professionals do not work as a team**

*"Care is very much dependent on us arranging it & coordinating it between specialists. The majority of care is private as we could not secure all care required via the public system. It is an enormous financial strain.*

*"Seems as if nobody talks to each other and care gets mixed up as a result. If you are not able to be your own advocate, you will suffer from the lack of coordination around healthcare.*

Transitioning from paediatric to adult care is a particularly challenging time, as 'adult' children begin to manage their own care. Fewer than one in five (16%) of those that have transitioned expressed satisfaction with the experience. Respondents commented on the lack of co-ordination between children's hospital services and voluntary and therapy services, and that adult services were not equipped for some rare conditions.

**16%** **satisfied with transition care**

## TIME FOR CHANGE

- Healthcare professionals should approach individuals presenting with rare symptoms or diseases holistically. We need joined up thinking across all disciplines.
- Care should be co-ordinated between teams, between the hospital and the community and when transitioning from children's to adult services - it places a greater burden on the individual and their family and carers when they need to do this work.
- More must be done to ensure that those who do not have access to specialist expert care in Ireland have links to the expertise across Europe via European Reference Networks (ERNs).

***"We as parents have to coordinate multiple professionals across multiple sites with no help from a coordinator/key worker. It would help with decision making and planning if we could have meeting (virtual or in person) with all members of the team across hospital/community/preschool, etc. to help with planning."***

## LISTEN AND LEARN - THE LIVED EXPERIENCE

Levels of trust and confidence and a feeling of being listened to tended to be higher when dealing with hospital and GP staff, and lower with emergency and other community services and social care.

Fewer than half of respondents (46%) felt listened to by hospital/GP based healthcare professionals, dropping to only one in five (20%) respondents feeling listened to in emergency and social care settings.

**46% hospital & GP HCPs listen**

**Emergency & social care listen 15%**

*It's like banging your head against a brick wall. Not being heard or listened to.*

*Medics need to listen to the people living with the condition.*

*We are told as opposed to consulted.*

Listening and sharing information and experience is a two-way process. Healthcare providers need to empower those living with rare conditions and enable them to manage and recommend care for themselves.

*"My child has a diagnosis. I am still waiting for her mutation to understand mode of treatment.*

39 respondents reported engaging with research in the last seven years, mostly through sharing information with or joining a condition-specific registry, taking part in research for health or social care and completing surveys around the lived experience. Only small numbers have had involvement in interventional drug (13%) and/or device (3%) trials. Sadly there is also very low involvement in PPI initiatives, where patients' views are included in research prioritisation, design and oversight.

More than 80% of respondents would like more opportunities to take part in research. Priorities for research are therapeutics (drugs/devices), disease mechanisms and diagnosis.

**80% want more opportunity to get involved in research**

*"Because it's a rare disease it seems pharmaceutical companies are not interested in providing medication because it's not profitable.*

*"Think it's important to take part in research to raise awareness and help profile a rare diagnosis*

*"Research is very poor in Ireland. I have contacted America and UK for research programmes.*

**EXPERT BY EXPERIENCE**

# RDI'S CALLS TO ACTION

### RAPID DIAGNOSIS AND COMPREHENSIVE GENETIC SERVICES

The first step on the rare journey is diagnosis – this must be expedited. People living with rare conditions are waiting too long for a diagnosis and are adding to waiting list numbers as they move from specialty to specialty and undergo a battery of tests. First steps are easier expedited access to genetic testing and comprehensive newborn screening.

### INTEGRATED CARE & TREATING THE WHOLE PERSON AS OPPOSED TO THE SYMPTOM

It should not be up to the individual to manage their care alone. Cross-functional multi-disciplinary teams representing all of their care providers in the hospital and the community would enable more holistic care to be provided. Technology should be exploited to help manage their care and their condition. Where appropriate, services should be community based, near to where people live.

### EDUCATION OF HEALTHCARE PROFESSIONALS, BOTH WITHIN THEIR SPECIALTY, AS CROSS-FUNCTIONAL TEAMS WORKING TOGETHER AND AS SENSITIVE/EMPATHETIC PROVIDERS OF CARE

Education on rare conditions in general is required across all care professions at all points in their training. HCPs need to learn how to work together as a team and share with colleagues in all settings. They need to learn how to listen to the experiences of those living with rare conditions. People living with rare conditions should be invited to tell their stories as part of the formal HCP education and training.

### LISTEN TO OUR VOICES – WE ARE THE EXPERTS WITH LIVED EXPERIENCE; WE ARE FOCUSED ON OUR CONDITIONS AND HAVE MUCH TO CONTRIBUTE

Many individuals spend hours every day researching their conditions, joining the dots and often engaging with others around the world on a similar health journey. These individuals have a huge amount of learnings that must be considered in their care and treatment. The person is at the centre only if they are listened to. This is an inexpensive behavioural change that must be delivered.

### RESEARCH & TREATMENTS THAT GET TO THE ROOT CAUSE OF THE CONDITION (PREVENTION & CURE) AS OPPOSED TO SLOWING PROGRESSION AND TREATING SYMPTOMS

Most treatment of rare conditions is about managing symptoms and slowing progression. It is not often that prevention and cure is an option. More needs to be done to enable people with rare conditions to access trials for innovative novel products that may transform their lives. The willingness of those living with rare conditions to get involved in research should be harnessed – there is ample opportunity for innovation in Irish universities to address some of the complex challenges and needs of the rare community.

# ACKNOWLEDGEMENTS

Thank you to all of those that participated in this survey, freely giving both their time and their expertise - we very much value and appreciate their experience and insights.

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Proudly supported by:





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