



RESEARCH REPORT

Voices of the RARE DISEASE COMMUNITY



Rare Diseases
Ireland

LIVING WITH A RARE DISEASE IN IRELAND DURING THE COVID-19 PANDEMIC

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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 is committed to the identification, treatment, and cure of rare diseases and care for those living with a rare condition.

TERMS USED IN THIS REPORT

Health Authority refers to Department of Health (DoH), National Public Health Emergency Team (NPHE), Health Service Executive (HSE) & management structures therein, Health Information Quality Authority (HIQA), Hospital Managers, etc.

Health Care Provider (HCP) refers to Hospital Consultant, General Practitioner (GP), Nurse Practitioner, Public Health Nurse, Physiotherapist, Occupational Therapist, Speech & Language Therapist, Mental Health Practitioner, Counsellor, etc.

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INTRODUCTION

The novel coronavirus (SARS-CoV-2), and the disease that it causes, COVID-19, is causing significant challenges for the estimated 300,000 people living with rare diseases in Ireland. People with underlying conditions are at risk of more severe illness¹ from COVID-19, and rare disease patients, their family members and carers are seeing their lives and care disrupted. Rare Diseases Ireland (RDI) conducted a survey² over 2 weeks ending May 6th to help elucidate and understand these issues.

SNAPSHOT OF COMMUNITY RESPONSE

A total of 176 participants responded to the survey; 58% of respondents are living with a rare condition, and 27% are family members of and 15% are carers of those living with a rare condition. 23% of those living with a rare condition are under the age of 18³, 24% are aged 18-39, 44% are

aged 40-69 and 9% are aged 70 and above. Participants are living across the island of Ireland, with responses from all 9 HSE Community Health Organisation regions and Northern Ireland. Multiple disease categories are represented by participants including neurology/neuromuscular, immune system, endocrine (hormone), lung, metabolic, eye, musculoskeletal, rare intellectual disability, cardiovascular, haematological (blood) and other types of rare disorders. 31% of participants have 2 or more rare conditions. 84% of those with a rare condition are living with family, 12 % are living independently alone and the remaining respondents are living in residential settings, including the family of one individual living in an acute hospital (who died following COVID-19 infection).

1

<https://www2.hse.ie/conditions/coronavirus/people-at-higher-risk.html> Accessed 14/05/20

² RDI's COVID-19 Survey: *Living with a rare disease in Ireland during the COVID-19 pandemic* was promoted to the rare disease community through direct e-mail and RDI's website and social media channels. The survey was open to the public for 2 weeks, and closed on May 6, 2020. Responses were collected anonymously.

³ The survey was open to respondents 18 years and older. In some cases the survey was completed by a family member or a carer

with reference to a person under the age the age of 18.

KEY FINDINGS

It is clear from the survey results that the rare disease community is enormously concerned and impacted by COVID-19; 73% are worried about their personal health. Sources of concern and worry were varied, though a number of people raised concerns about deteriorating health, safely accessing health care if or when needed, accessing medicines/medical supplies, and worry about risk of exposure and the impact that COVID-19 may have on their underlying rare-condition.

56% of respondents are cocooning, with almost 50% of those cocooning choosing to do so because of personal concerns about their health and well-being. 53% of scheduled appointments across all healthcare settings have been cancelled, at a cost to the immediate and long-term health and well-being of those living with a rare condition. 26% are encountering difficulties accessing medicines and medical supplies. 62% believe that COVID-19 is having a negative impact on their mental health.

Many respondents have expressed frustration and disappointment with the lack of communication from their health care providers (HCPs). They have been left to cope with their rare condition in an enormously challenging environment and they are struggling to know how best to manage. This is further complicated by the almost complete cessation of all hospital care (most expertise in the care and management of rare conditions in

Ireland lies with hospital consultants). For some respondents closure of private hospital facilities is adding to the worry and confusion as individuals try to ascertain who is now managing their care. Hearing anecdotal stories of HCPs being admonished for providing 'non-emergency' consultations and advice to patients is compounding the issue. There is a belief that the health authorities are making the situation worse by exclusively focusing on COVID-19 and emergency care, to the exclusion of all on-going disease management and care.

Respondents told RDI:

- *Complete lack of contact by the respiratory team at [hospital] and by my GP.*
- *No advice received from consultant or hospital team re COVID-19 unlike NHS patients who received a letter detailing cocooning. Only advice was general HSE guidelines which GP just restated.*
- *I have had no contact from GP or Consultant about my rare condition.*
- *No contact from physio, e.g. phone call, for reassurance & safety.*
- *No call/ email from specialist nurse to check we're ok.*
- *No contact from community nurse.*
- *No phone call from day service.*

WHO IS COCOONING?

56% people living with a rare condition are **COCOONING**

People who are at very high risk (extremely medically vulnerable) from coronavirus (COVID-19), have been advised by the health authorities to protect themselves by 'cocooning'. Those cocooning are strongly advised to stay at home at all times, avoid face-to-face contact and minimise all non-essential contact with other members of the household. ⁴

A large percentage of the rare disease community in Ireland are cocooning. 24% of respondents are cocooning as a result of advice from the health authorities. 7% of respondents have been advised by their Health Care Provider (HCP) to cocoon. Additionally, 25% have determined that they should cocoon because of their concern about their rare condition and the risk of COVID-19 to their health. 7% of respondents should be cocooning, but are not due to personal circumstances.

⁴ During the period of this survey cocooning meant not leaving your home for any reason. Since May 5th people cocooning have been allowed to leave their home for a brief period to exercise.

Respondents told RDI:

- *Surprised that this is not recommended for people with my condition.*
- *We decided as his parents.*
- *Can't cocoon in acute hospital setting (probable cause of fatal viral infection).*
- *Following advice from (international) patient organisation.*
- *GP felt I am high-risk. Should I cocoon?*
- *If I could I would, but as a lone parent it is impossible.*
- *It's not just over 70s that need cocooning or community help. People with disabilities exist too*

SIGNIFICANT IMPACT ON ACCESS TO CARE

53% scheduled appointments **CANCELLED due to COVID-19**

Findings show that 53% of medical appointments have been cancelled. This rises to 60% when considering hospital appointments alone, where 61% of diagnostic/ monitoring procedures, 75% of 'elective' surgical procedures and 57% of in-/out-patient therapies have been cancelled. Many people with rare conditions spend months and years trying to pinpoint a diagnosis and find therapeutic relief. Cancellation of such a large number of appointments is extremely worrying for

all in the rare disease community. Everyone will be impacted by the knock-on effect of these cancellations and delays.

Community services are not faring much better with physio (65%), OT (57%), SLT (57%) and respite (67%) all seeing a majority of appointments cancelled. GPs (26%), mental health (43%) and counselling (40%) have lower rates of cancellation, and in fact have begun to adopt technology, which is being leveraged to provide service.

In all healthcare settings already overburdened HCPs now face the prospect of spiralling waiting lists. This is further compounded by the closure of the private hospital system, and is particularly frustrating when we hear of empty hospital beds and under-utilised resources.

Respondents told RDI:

- *We are experiencing cancellation of all visits.*
- *Surgery in UK cancelled. Need to get to the UK for surgery. UK better equipped per HCP.*
- *We are in extreme crisis now over homecare/community care for my parent.*
- *My parent's regular outpatient & community care supports & scheduled surgery have all been cancelled and respite was almost cancelled despite my emergency medical status. I had to engage TD's constituency officer's input to secure respite.*

- *Waiting LISTS will be crazy after this all ends - I am afraid of attending a clinic in the public system unless they change things as they are so crowded.*
- *Taken off my meds and awaiting surgery since. Symptoms are getting worse as is quality of life.*
- *Required surgeries that will become a bigger issue if not addressed.*
- *Postponed appointment was to review and schedule surgery for the summer.*
- *Concerned planned treatments and follow up appointments may be cancelled.*
- *Confusion over whether private patients are being seen by their Consultants or whether we are now on long public waiting lists despite having rare conditions. Rumours abounding that private patients only recently added to public lists will wait a long time for review etc.*
- *I have private medical insurance and I worry for future care - the ability to meet consultants and treatments has been vital to me.*
- *HSE productivity and management in the private hospitals is worryingly low, uniform healthcare will collapse if executed via current HSE model.*

31% appointments conducted REMOTELY using tech & at alternate sites

Technology and provision of care at alternative locations⁵ is being leveraged to provide services to some. There were 279 examples of care (197 scheduled and 82 other/emergency appointments) being provided via technology or outside of the normal setting. The experience has been described as positive or not different 75% of the time. 63% of GP appointments have been delivered remotely. Remote delivery of all other healthcare services is disappointingly low (e.g. only 33% of out-patient periodic reviews have been conducted remotely). These low figures are in part a result of the 'red-tape' surrounding adoption of technology. Additionally we are hearing anecdotal stories of HCPs being admonished for providing 'non-emergency' consultations, even via phone.

Respondents told RDI:

- *Had one online combined appointment with SLT and OT.*
- *Completed a survey for [HCP] and was contacted by phone.*

⁵ Respondents were provided 4 examples of care being provided in a non-standard route, as follows: 1) monitoring of symptoms via phone or telemedicine, 2) nurse or other professional taking samples in your home, 3) GP/specialist consultation via phone, 4) therapy normally delivered in a hospital setting delivered in a satellite/off-site healthcare facility or in the home.

- *Nurse specialist linked in quickly and responded to concerns, linked with MDT.*
- *Actually very effective to have my regular 6 month check-up with my consultant by phone. Normally have to travel to Dublin and take a full day off college for it.*
- *Consultation by phone only. Would have been much better for Consultant to be able to see facial expressions etc.*
- *Nothing is better for examining a person than in person. Majority of communication is body language.*
- *Have not had any alternative to my cancelled hospital appointments offered.*
- *Telephone consultation to replace cancelled outpatient appointment.*

26% respondents encounter difficulty ACCESSING medicines and/or medical supplies

A significant number of respondents are having difficulty accessing their medications for their rare condition. Medications include pharmaceutical drugs, nutritional food products, bandages, PPE, etc. 17% have experienced medication shortages ('limited supply' and 'out of stock') and 3% can't get their prescription.

Respondents told RDI:

- *Medication was briefly out of stock which caused anxiety and*

breakthrough symptoms. Usual brand still unavailable.

- *Gloves for home care are difficult to get.*
- *Prescription harder/slower to get.*
- *Getting in touch with GP to renew prescription is extremely difficult.*
- *[Prescription] has to be written by Consultant, who I can't get to see at present.*
- *Where to get prescriptions which were previously done by hospitals at outpatient clinics.*
- *Chemist closed & reduced hours.*
- *Restrictions to paracetamol that can be purchased in one visit. Unable to go to shop. Partner trying to limit exposure to virus so not going to shop more than once a week. Cannot stock up due to restrictions on purchasing painkillers.*
- *The 5km restriction and Garda checkpoints. Not being able to access the wider range of low protein foods that a larger supermarket would have.*

31% respondents AVOIDED seeking care for complications related to their rare condition

A significant number of respondents are delaying accessing care for medical complications related to their rare condition that have arisen since COVID-19 restrictions were introduced. Respondents report a number of reasons for this including

concern about being exposed to COVID-19, confusion about who is the most appropriate HCP to provide care and strict adherence to 'cocooning' guidance.

Respondents told RDI:

- *I feel that my GP would not be able to help and would not appreciate being asked.*
- *It has made things more complicated. GP has restrictions which I encountered while having blood-test, no waiting indoors, stand outside until brought in therefore I would be slower to make contact.*
- *Our GP has been strictly by phone but very good since the restrictions.*
- *We had an ambulance last week for our daughter and once she was stable and well we made the decision to stay at home instead of being admitted as would normally happen due to the virus which we discussed with the paramedics*
- *I didn't think I'd be able to get an appointment last month for a non COVID-19 issue so instead of my GP I went to the specialist unit in the [hospital] who treat me.*
- *I don't want to get coronavirus. If I go to doctors could end up with virus.*
- *Avoiding GP contact re new onset chronic headaches.*
- *My vision has changed but I have not checked it out.*
- *Had bad pains in my lower right abdomen but was afraid to go to A&E.*
- *Recently admitted to hospital but not put under my consultant which*

is important in the care of rare diseases.

SOURCES OF WORRY

73% respondents
more
CONCERNED about their
rare health condition
because of **COVID-19**

Faced with such significant changes to life as we have known it, it is no wonder that 73% of respondents are concerned about their rare health condition as a result of the COVID-19 outbreak. They are concerned about how their condition is deteriorating without access to their usual care programs, and how their complex health situation may be negatively impacted if they are infected and have COVID-19. Respondents are additionally concerned about how their rare condition may be perceived if they do contract COVID-19 and require ICU care.

Respondents told RDI:

- *Intubation not an option due to internal complications from [rare condition].*
- *ICU team had no experience of [rare disease] crisis management. [Consultant] didn't attend for 6 hours or guide over the phone. I lost speech and swallow so couldn't guide myself. [Drugs] administered on [rare condition] blackbox warning*

list. This resulted in 2 separate anaphylaxis attacks.

- *Condition is progressing*
- *What's the point!*
- *Blood test shows loss of muscle mass.*
- *Worried about contracting COVID-19 and unknown impact it may have.*
- *Concerned about worsening breathlessness.*
- *It's a long black hole with no light.*
- *It's hard; mom is front line health care worker; she was rejected for leave.*
- *Experiencing CHRONIC FATIGUE - Constantly!!!! Nervous of getting further strokes.*
- *Choking incident at home & several falls. Two wetting accidents. As a carer and wife just trying to do my best and keep my husband safe and out of hospital. We have 3 teenage daughters so trying to keep family life as normal as possible & everyone safe.*
- *I'm afraid my son will deteriorate without services*
- *Lack of understanding [of rare disease] by a lot of service providers, especially in acute hospital setting.*
- *Condition has deteriorated - once a skill isn't used consistently, it's forgotten, and can't be retaught.*
- *I would worry if I was to go to hospital that I would be just left there with staff not being able to understand the way I communicate through gestures and sounds. I would be afraid my incontinence pads would not be changed as frequently as my mother does. I would be afraid my toys would not be allowed because of risk of contamination - I function like a*

baby and just love my toys that have lights sounds and music - I have to have access to them - they keep me entertained - I love Barney/Thomas the Tank and all pre-school DVDs. I will be 22 years old in July 2020.

- *I am scared. If they have too many persons in intensive care would they make the decision not to treat me due to me having several comorbidities. As my condition is so rare most staff in hospitals do not know how to treat me.*
- *Not overly worried. But I do get anxious when I notice a slight deterioration in my sight.*
- *As her parent, I worry about her catching COVID-19. I also worry about her future health as she already has permanent damage to one of her lungs and she is only 9. There is no cure so we just have to do the best we can to slow the progress of the damage to her lungs.*
- *They don't know the affects COVID-19 would have on narcolepsy*
- *I'm afraid of being in a wheelchair eventually.*
- *I'm afraid of being in pain or a vegetable for the rest of my life."*
- *I wouldn't stand a chance if I got the virus (doc's words). Current treatment isn't working. Plan was to bring me into hospital to be monitored for new treatment which I have no idea could be done as the doctor who wanted to monitor me is treating COVID positive patients.*
- *[Rare condition] affects my lungs so I'm scared how my body will be if I get virus.*
- *Visually impaired - have to relearn familiar geography after cocooning - stress and concern going out now.*

- *Who will come to my house and take care of me if my parents become ill. I need 24/7 constant care.*

62% respondents say **COVID-19** has **NEGATIVE** impact on **mental well-being**

While employment and financial well-being are unchanged/positive for approximately 2/3rd of respondents, the impact of COVID-19 has been negative on both mental (62%) and physical (59%) well-being.

Respondents told RDI:

- *Missing family*
- *Extremely difficult caring for a child with complex special needs. Is impacting on everyone in the family.*
- *Can get a bit tearful watching information updates so we limit the news.*
- *I'm finding work mentally and emotionally very difficult*
- *Sadness and worry*
- *Stress level is beyond high.*
- *my job is an active job now I'm spending my days sitting at home and I miss my extended family*
- *Would normally go for long walks a few times a week*
- *World has turned 360 degrees*
- *I am not able to work at the moment*
- *I've lost my job and can't play [sport]. Two big parts of my life which have helped me greatly (mentally & physically) with caring for my young husband with [] and our three teenage daughters.*

- *Lack of stimulation, unable to leave home, terminal rare condition and not being able to see family and friends as time runs out*
- *Upsetting not being able to see family members, e.g. my nan*

COMMUNITY

EMPOWERMENT

36% respondents want **TRAINING** to better manage care of rare condition at home

Rare conditions are often complex and require continuous on-going care. With the reduction in provision of professional healthcare resources, many are turning to providing critical elements of care in the home themselves. 16% of respondents have received training to provide care in the home. Only 7% of those trained have expressed a preference for professional support, as opposed to 74% who are happy to continue to provide this care on a permanent basis. A further 36% of respondents would like to receive training to allow them to better manage care in the home.

Respondents told RDI:

- *Webex training [to allow me to provide] home care*
- *I provide physio for my child which she would normally get in school.*
- *Don't want my family involved in my care*

- *Can't monitor blood levels at home*
- *Would like to monitor [drug] at home just like they do in USA.*
- *I did training online [provided by patient organisation]*
- *Something that would help with managing chronic fatigue.*
- *My mother has completed numerous disability courses and is also trained to give specialised medicine if seizures out of control*
- *I was shown how to do chest physio with [my child]*

31% respondents have sufficient **INFORMATION on COVID-19** for people with rare condition

83% respondents feel they have sufficient general information on COVID-19, however only 31% felt that they have sufficient information that relates COVID-19 to their rare condition. The primary sources of general COVID-19 information are TV, health authority websites and national radio, whereas the primary sources of rare disease information is consultant, patient organisations and private on-line groups.

YOUR VOICE MATTERS

Recommendations from the rare disease community

RDI is listening to the patients and families in our community and we will continue to channel your collective voices into action. Here are some of the most common concerns and recommendation shared by survey respondents both with respect to care for their rare condition and how they should manage as current restrictions are lifted.

Respondents told RDI:

- *Respiratory care not related to COVID-19.*
- *Better communication, which at present is non-existent.*
- *Letter from treating hospital outlining future plans*
- *Access to services for people that have moved (back to family) as a result of COVID-19*
- *COVID-19 advice specific to specific rare conditions*
- *Clear guidelines for vulnerable people in respect to visiting GPs for non COVID issues.*

- *Rare disease patients diagnosed and undiagnosed should at least be contacted by their team to advise on how to return to work/college.*
- *Where to get prescriptions which were previously done by hospitals at outpatient clinics*
- *Counselling for the carer. Counselling to enable those in isolation to cope.*
- *Lack of respite for carers at present*
- *Dentist when they will open again?*
- *Schooling plans for those in "vulnerable" categories e.g. immunosuppressed.*
- *Schools, my son is regressing I believe*
- *Masks suitable for children*
- *PPE for Carers & nurses coming in not just mask*
- *Unrestricted access to larger supermarkets and priority queuing*
- *Communication campaign to educate others that being 'vulnerable' is not always easy to spot, e.g. immunocompromised.*
- *System to support vulnerable individuals if primary carers become unwell.*
- *Priority access to vaccines when available for individuals with rare conditions and/or their families and carers.*
- *My pharmacist gives better advice/resource referral than my current [HCP].*

WHAT IS A RARE DISEASE?

A rare disease is defined as a disease that affects less than 1 in 2,000 individuals in Europe. There are 6,000 to 8,000 individual rare diseases identified to date with more being described on a daily basis. While individual diseases and affected patients may be rare, collectively rare diseases are many. There are an estimated 300 million people worldwide, and 300,000 in Ireland, living with a rare disease.

Rare diseases are significant contributors to a number of poor health outcomes in terms of their high associated mortality, morbidity and disability. In particular, rare diseases are a significant contributor to early foetal loss and perinatal mortality, as well as infant and child mortality.

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 are a significant health burden.

KEY RARE DISEASE STATISTICS IN IRELAND

- There are an estimated 300,000 people living with rare diseases in Ireland.
- 72% of rare diseases are genetic in origin. 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, second cousins.
- 70% of rare diseases are exclusively paediatric onset.
- At least 4% of children are diagnosed with a rare disease, a life-long condition, by age 17 in Ireland.
- Approximately 2/3 of paediatric deaths in Ireland are associated with rare diseases.

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