

# An Easyguide to **Rare Diseases** in Ireland **and Consensus for Action**

For Government, the General Public, Media and Political Parties



**February 2020**

Published by Patient Groups concerned with Rare Diseases



This guide is published in remembrance of Keelin Shanley. A very fine journalist and a good friend of Cystic Fibrosis Ireland.

Published by the Rare Disease Taskforce which brings together the national alliances HRCI; IPPOSI and RDI and rare disease patient organisations (see Annex 5).

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# Introduction

The aims of this c are to:

- Explain what a rare disease is and how it is defined in health policy
- Provide insights into living with a rare disease
- Identify priorities patient advocacy groups would urge the Irish Government to include in the Programme for Government, 2020 and beyond, including Sláintecare and the annual HSE Service Plans
- Provide an overview of policy and research developments at international and national policy levels
- Provide sources of information and support for rare diseases, including the National Rare Diseases Office (NRDO) and Rare Diseases Ireland (RDI).

## This Easyguide is aimed at:

- Government & Political Parties
- Department of Health & HSE
- Health Care Professionals
- Media organisations
- People concerned about rare diseases, including family members, carers and those living with a rare disease.

This Easyguide has been produced by the Rare Disease Taskforce, which brings together the three national networks of Health Research Charities Ireland (HRCI)<sup>1</sup>; Irish Platform for Patients, Science and Industry (IPPOSI)<sup>2</sup> and Rare Diseases Ireland (RDI)<sup>3</sup>. The Taskforce was formed as a platform for bringing together patient advocacy groups concerned with rare diseases in Ireland.

1. [www.hrci.ie](http://www.hrci.ie)

2. [www.ipposi.ie](http://www.ipposi.ie)

3. [www.rdi.ie](http://www.rdi.ie)

4. <https://www.ipposi.ie/images/An-Easy-Guide-to-Rare-Diseases.pdf>

5. <https://www.gov.ie/en/publication/a4ac1b-national-rare-disease-plan-for-ireland-2014-2018/>

## This Easyguide is structured as follows:

**Section 1:** An Overview of Rare Diseases in Ireland

**Section 2:** Living with a Rare Disease – The Challenge and the Determination

**Section 3:** Priorities for the Programme for Government (2020) and beyond

**Section 4:** Rare Disease Policy and Research Contexts

**Annex 1:** Glossary of Key Terms

**Annex 2:** The National Rare Diseases Office and Orphanet

**Annex 3:** The Rare Disease Policy Landscape: A Visual Overview

**Annex 4:** Further descriptions of Rare Diseases in Ireland

**Annex 5:** Patient Advocacy Organisations

## Acknowledgements and Thanks

This Easyguide to Rare Diseases in Ireland and Consensus for Action is a sequel to an earlier publication<sup>4</sup> drawn up by patient advocacy groups in 2013 to inform the National Rare Disease Plan (NRDP) for Ireland 2014 - 2018<sup>5</sup>.

Many thanks to all those who have contributed to this Easyguide. In particular those people living with a rare disease whose insights are featured in Section 2 of this publication. Many thanks to the three national networks: HRCI, IPPOSI and RDI which together published this Easyguide (led by CEO's Avril Kennan, Derick Mitchell and Vicky McGrath respectively).

Many thanks to the ongoing work of the National

Rare Disease Office and National Clinical Programme led by Professor Eileen Treacy (see Annex 2). Many thanks to the Department of Health, which liaises with the Rare Disease Taskforce on a regular basis since the NRDP was extended in 2019.

Many thanks to Cystic Fibrosis Ireland (CFI) which funded this Easyguide, in particular its Chairperson Patrica Duffy-Barber and its CEO Philip Watt, who is also Chairperson of the Rare Disease Taskforce and who edited and researched much of this Easyguide. Many thanks to Avril Kennan, HRCI who researched section 4.2 and Annex 3 and Sarah Techlenborg, CFI who researched section 4.1. and to Dr Amy McKnight and Dr Helen McAneney of Queen's University Belfast and the Northern Ireland Rare Disease Partnership for their inputs and expert advice.

Many thanks to the work of patient advocacy organisations in Ireland without whom this publication would not have been possible (see Annex 5).

Thanks to Pearse Cafferky, Design Focus, for the design and production of this Guide.

Finally, many thanks to our colleague and friend Avril Daly, former Chairperson of Rare Diseases Ireland and Vice President of EURORDIS - Rare Diseases Europe who continues to play a vital role in representing patient concerns on rare diseases at a national, European and global level, whilst living with Retinitis Pigmentosa, a rare eye disease (see Annex 4).

This publication is dedicated to Professor David Barton (recently retired) and Professor Andrew Green for their combined contribution to clinical genetic services and diagnostics over many years.

**The Rare Disease Taskforce**  
**February 2020**

### Disclaimer

Every effort has been made to make this Easyguide as comprehensive and up to date as possible. However with 6-8,000 rare diseases in Ireland it is only possible to provide examples of a few of these conditions in this publication (see Section 2 and Annex 4). We would hope to add to these insights in future editions and we would also refer readers to the Easyguide<sup>5</sup> published in 2013, which continues to have relevant information. Nothing in this publication constitutes medical advice. Health information changes over time and should be re-checked. When in doubt people concerned with rare diseases should consult their health care provider; their patient advocacy group (where one exists); Rare Diseases Ireland and the extensive information that is available from the National Rare Diseases Office<sup>6</sup> and the Orphanet websites (see Annex 2).

### How to Cite

Rare Disease Taskforce, 2020. 'An Easyguide to Rare Diseases in Ireland and Consensus for Action'. HRCI, IPPOSI and RDI.

*Rare diseases are characterised by their relatively low prevalence (less than 1 in 2,000 people in the EU).*

*Collectively rare diseases affect around 6% of the population in the Republic of Ireland, accounting for at least 300,000 individuals (HSE Model of Care)<sup>7</sup>.*

*There are between 18-30 million people with a rare disease in the EU (conservative estimate)<sup>8</sup>.*

*Although rare diseases by definition affect relatively small numbers of people, it is estimated that 350 million people globally have a rare disease, more than double the number of patients affected by AIDS and cancer combined (Nature Review, 2019)<sup>9</sup>.*

5. <https://www.ipposi.ie/images/An-Easy-Guide-to-Rare-Diseases.pdf>

6. <http://www.orpha.net/national/IE-EN/index/homepage/>

7. <https://www.hse.ie/eng/about/who/cspd/ncps/rare-diseases/resources/model-of-care-for-rare-diseases.pdf>

8. See also Wakap S, Lambert DM et al. 'Estimating Cumulative Point Prevalence of Rare Diseases. Feb 2020. [http://www.orpha.net/national/data/NO-NO/www/uploads/Orphanet\\_publication\\_pointprev\\_2019.pdf](http://www.orpha.net/national/data/NO-NO/www/uploads/Orphanet_publication_pointprev_2019.pdf)

9. Tambuyzer, E et al in 'Nature Review'. 13 December 2019. Therapies for Rare Diseases: Therapeutic Modalities, Progress and Challenges Ahead <https://www.nature.com/articles/s41573-019-0049-9>

## Section 1

# An Overview of **Rare** **Diseases in Ireland**



# An Overview of Rare Diseases in Ireland

Rare diseases are characterised by their relatively low prevalence (less than 1 in 2,000 people in the EU). To have a rare disease is to have a condition that often goes undiagnosed for years. Doctors may never have seen the condition before and hospital diagnostic services may struggle to find the rare disease presented by an individual and their family/carer.

Once accurately diagnosed, people with a rare disease and their carers may face further challenges to find a suitably defined care pathway for treatment and to access therapies, which can become even more frustrating if a condition is ultra-rare (less than 1 in 100,000).

However, there is hope. The outlook for some patients in Ireland with rare diseases has become steadily more positive in recent years in disease areas that have benefitted from significant investment in research, drug-therapies and services. These also tend to be disease areas that have an active patient advocacy group.

Sadly, however, this sense of hope and progress is less evident for many living with a rare or ultra-rare disease in Ireland where there has been less focus and investment in research and expertise and with no patient group. The reality for many people and their families living with a rare disease in Ireland today is that they still have to travel long distances to consult with the few experts who have experience in treating and studying their condition. There are also increasingly long waiting lists for key services such as clinical genetic diagnosis and counselling.

The impact on daily-life for both family and individuals living with a rare disease can be both

emotionally and financially profound, particularly where there are gaps and weaknesses in supports for health costs and allied social care. These supports can be as important as Centres of Expertise<sup>10</sup>.

When you talk to people living with a rare disease, there is often a quiet determination to live life to the fullest extent possible and not to be defined by their disease. People with a rare disease often dislike being referred to, or thought of, as 'victims' or 'sufferers' as it infers a sense of helplessness or resignation, though it is also understood that those using such terms do not usually intend such inferences. 'People (or sometimes patients) with a rare disease' is often the preferred self-ascription<sup>11</sup>.

Both children and adults in the general public can be unsure of how to respond to people with a profound or even a mild disability and very unkind, outdated or uneducated language persists and needs to be challenged.

For some people with a rare disease, as they grow older, they will often strive towards the ambition of some form of supported independent living with the help of carers and health professionals.

Further, many carers are active in a patient advocacy group and campaign or fundraise for better services, on top of their on-going commitments of supporting the care of loved ones living with a rare disease. Likewise people in later life may also find themselves caring for a spouse or loved one recently diagnosed with a rare disease.

Anne Lawlor, the founder of 22q11 Ireland, whose daughter Áine is featured in Section 2 of this

10. Orphanet provides information on centres of expertise or networks of centres of expertise dedicated to the medical management and/or genetic counselling for one particular rare disease or a group of rare diseases. [www.orpha.net](http://www.orpha.net)

11. See terminology used in HSE, Royal College of Physicians of Ireland. 'Model of Care for Rare Diseases'. The National Clinical Programme for Rare Diseases.

Easyguide and in the 2013 edition. Anne describes the impact of the patient advocacy group that she established in 2007, and where she has worked on a voluntary basis for 13 years,

*‘The impact that our charity has had on the Irish 22q community is phenomenal. When we began in 2007 with just 3 families very little was known about 22q deletion syndrome in Ireland. Now we have 150 families nationwide who are so much more clued-in about their children’s needs. Our parents are helping to educate doctors, dentists, consultants, nurses, therapists, teachers, social workers and a host of other professionals about 22q. A particular high point was when we hosted the European 22q Alliance meeting in Dublin in 2017. We connect, learn and share our most valuable resource –ourselves, and the kind of knowledge that can only come from the felt-lived-experience of loving and caring for a child with 22q’<sup>12</sup>.*

### 1.1 Why focus on Rare Diseases?

Although individual rare diseases by definition affect relatively small numbers of people, it is estimated that 350 million people globally have a rare disease, more than double the number of patients affected by AIDS and cancer combined<sup>13</sup>.

However it is only in relatively recent years that international bodies such as the United Nations have begun to focus significant attention on the multifaceted and somewhat neglected needs of those with rare diseases. In September 2019, UN member states adopted a Political Declaration on Universal Health Coverage, which includes rare diseases. This marks the first time that rare diseases have featured in a UN declaration adopted by all 193 member-states (see Section 4).

The National Rare Disease Plan (NRDP)<sup>14</sup>, published in 2014 and arising from an EU Directive<sup>15</sup>, was the first

major policy intervention on rare diseases in Ireland, which again reflects the relatively recent focus on rare diseases at a national level within Ireland (see section 4.1).

For those living with a rare disease and their carers, there can be a huge range of varied challenges including access to (or delays) in diagnosis; absence of (or remoteness from), a Centre of Expertise (CoE). There are also many uncertainties about the full clinical impact of a disease over the course of a lifetime, which can be progressive or sporadic and the prognosis may vary considerably from one individual to another, even within the same family.

Depending on the disease, survivability and quality of life can be shaped by a wide range of factors including at the most basic level genotype and phenotype (see Annex 1: Glossary of Key Terms). A recent international study shows that 72% of rare diseases are genetic in origin<sup>16</sup>, hence the importance of new and innovative research into the human genome and the need to improve clinical genetic services in Ireland, which are emphasised throughout this publication. There is growing hope among health researchers of the potential of new and innovative health research in addressing the underlying causes of diseases with a genetic origin (see Section 1.4).

Survivability and quality of life for those living with a rare disease can also be shaped by more macro factors such as income; environment; access to and quality of dedicated health and social services and access to new and innovative drugs and other therapies, which in itself is largely dependent on the investment of resources and commitment to research and clinical trials and which in turn is often dependent on the advocacy of a dedicated patient group that can effectively advocate on behalf of its members.

12. Anne Lawlor, 22q11 Ireland

13. Tambuyzer, E et al in ‘Nature Review’. 13 December 2019. Therapies for Rare Diseases: Therapeutic Modalities, Progress and Challenges Ahead <https://www.nature.com/articles/s41573-019-0049-9>

14. <https://www.gov.ie/en/publication/a4ac1b-national-rare-disease-plan-for-ireland-2014-2018/>

15. <https://www.sciencedirect.com/science/article/pii/S0925443915000599>

16. Wakap S, Lambert DM et al. ‘Estimating Cumulative Point Prevalence of Rare Diseases. Feb 2020. [http://www.orpha.net/national/data/NO-NO/www/uploads/Orphanet\\_publication\\_pointprev\\_2019.pdf](http://www.orpha.net/national/data/NO-NO/www/uploads/Orphanet_publication_pointprev_2019.pdf)

### 1.2 Definition of a Rare Disease

Rare diseases are characterised by their relatively low prevalence (less than 1 in 2,000 people in the EU). In the EU, a rare (or 'orphan' disease as it is sometimes known) is additionally defined as life-threatening and/or serious. In the United States a rare disease is defined as a disorder affecting less than 200,000 of the entire US population.

Rare diseases impact on both children and adults, although rare diseases are far more likely to be found in children and young people. 70% of rare diseases are exclusively paediatric onset and 12% are exclusively adult onset. 81.9% of rare disease are ultra rare (less than 1 per 100,000 persons). However, almost all people with a rare disease (more than 98%) have one of the 390 most prevalent diseases (more than 1 per 100,000 persons)<sup>17</sup>.

Life expectancy can vary considerably between one rare disease and another and also between people living with the same disease. With advances in genetics and research, the 2020's will be a period of renewed hope for some people living with a rare disease in particular those who have benefitted from a significant increase in focus and investment in research, including clinical trials. However, for most rare diseases there remains no cure and the management of symptoms can be significantly hampered by a continued lack of sufficient knowledge or a failure to translate knowledge into new and innovative therapies.

Collectively rare diseases affect around 6% of the population in the Republic of Ireland, accounting for at least 300,000 individuals, although the total disease burden for rare diseases is not precisely quantified, partly due to inadequate health surveillance coding<sup>18</sup>.

An analysis of the Orphanet database published in 2020 found that rare diseases affect at least 3.5–5.9% of the worldwide population. This translates into conservative figures of 18–30 million persons in the

EU, and 263–446 million persons affected worldwide by rare diseases at any point in time. As this analysis did not consider rare cancers, infectious diseases and poisonings, the number of people affected by rare diseases is likely considerably higher<sup>19</sup>.

### 1.3 Some of the main characteristics of rare diseases:

- There are between 18-30 million people with a rare disease in the EU (conservative figure, 2020)
- Rare diseases are often chronic, progressive, degenerative, and often life-threatening
- Rare diseases are disabling: The quality of life of patients is often compromised by the lack or loss of autonomy/independence
- High level and complex levels of morbidity and care for the patient with a rare disease and his/ her family
- There are between 6-8000 rare diseases worldwide but this is increasingly looking like a significant underestimate
- 70% of rare diseases first appear in childhood and are life-long
- 64% of all deaths from the National Paediatric Mortality Register data for the years 2006-2016 were accounted for by rare diseases
- 72% 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 diseases can also be man-made, for example prescription or recreational drugs (including alcohol) and prescription drugs with unintended side effects that cause birth defects.

Many rare diseases have a serious, incapacitating and chronic nature often associated with multiple types of disabilities, including physical, sensory and

17. Wakap S, Lambert DM et al. 'Estimating Cumulative Point Prevalence of Rare Diseases. Feb 2020. [http://www.orpha.net/national/data/NO-NO/www/uploads/Orphanet\\_publication\\_pointprev\\_2019.pdf](http://www.orpha.net/national/data/NO-NO/www/uploads/Orphanet_publication_pointprev_2019.pdf)

18. HSE, Royal College of Physicians of Ireland. 'Model of Care for Rare Diseases'. The National Clinical Programme for Rare Diseases, p7

19. Wakap S, Lambert DM et al. 'Estimating Cumulative Point Prevalence of Rare Diseases. Feb 2020. [http://www.orpha.net/national/data/NO-NO/www/uploads/Orphanet\\_publication\\_pointprev\\_2019.pdf](http://www.orpha.net/national/data/NO-NO/www/uploads/Orphanet_publication_pointprev_2019.pdf)

cognitive. According to the Model of Care for Rare Diseases in Ireland,

*'Due to the rarity of these conditions and often lack of national expertise, the diagnosis of these conditions is often delayed for many years. This is associated with significant hardship for patients and families, unnecessary appointments, referrals and tests, loss of income and career prospects and causes delayed opportunities for intervention'<sup>20</sup>.*

There are both similarities and differences between rare diseases and rare cancers, and for the purpose of this Easyguide rare cancers are included. This is reflected in Section 2 and Annex 4 of this publication<sup>21</sup>.

#### **1.4 Therapies and the possibility of cures for those living with a Rare Disease**

Most rare diseases still lack approved treatments, never mind cures. Despite major advances in research in recent decades, it is increasingly recognised that there are massive gaps between translating advances in rare disease knowledge into potential medications<sup>22</sup>.

However there have been some remarkable breakthroughs in developing new and innovative orphan drug therapies in recent years. These have come about as the result of the study of a range of therapeutic modalities including; small molecules; monoclonal antibodies; protein replacement therapies; oligonucleotides and gene and cell therapies and gene editing/CRISPR<sup>23</sup>.

The reimbursement price of the drug therapies derived from these breakthroughs is often very high. These high costs can contribute to restricting or delaying the availability of new and innovative drugs worldwide, particularly in poorer countries, but also

increasingly in richer countries that are struggling to meet these very high costs. Availability can also be restricted or delayed by the way new and innovative drugs are assessed. Drug assessment/approval systems at a national level have often not kept pace with the remarkable breakthroughs in science and discovery. Further, drug approval systems in some instances do not take adequate account of the additional risks and costs associated with developing orphan drugs. In short, drug therapy reimbursement systems need to be reviewed and updated on a regular basis.

There are differing academic perspectives on whether the higher price of orphan drugs are justified. Some health economists have even questioned the 'value of rarity'<sup>24</sup>. Fortunately, this has not been the approach of the European Medicines Agency which plays a central role in facilitating the development and authorisation of orphan drugs. Applications for orphan designation are examined by the EMA's Committee for Orphan Medicinal Products and a full list of orphan designations are available in the Community Register of orphan Medicinal Products for Human Use<sup>25</sup>.

#### **1.5 Allied Health Supports and Services Needed for Rare Diseases**

The National Rare Disease Plan for Ireland 2014–2018 (extended in 2019) recognises that a holistic approach is needed to meeting the needs of people with rare diseases and their carers,

*'Specialised social services are instrumental for the empowerment of people living with rare conditions, as well as improving well-being and health. For people living with a rare, chronic and debilitating disease, care should not be restricted to medical and paramedical aspects, but should also take into*

20. <https://www.hse.ie/eng/about/who/cspd/ncps/rare-diseases/resources/model-of-care-for-rare-diseases.pdf>

21. The Irish Cancer Society has played a very positive and supporting role in helping to develop rare disease policy in Ireland, including for non-cancer diseases.

22. Tambuyzer, E et al in 'Nature Review'. 13 December 2019. Therapies for Rare Diseases: Therapeutic Modalities, Progress and Challenges Ahead <https://www.nature.com/articles/s41573-019-0049-9>

23. For an excellent introductory overview of the potential of gene editing/Crispr and safeguards needed, see <https://www.bbc.co.uk/iplayer/episode/m000dt7d/storyville-the-gene-revolution-changing-human-nature> Storyville: 'The Gene Revolution' broadcast 27 Jan, 2020.

24. McCabe, C. 'Orphan Drugs and the NHS: Should we Value Rarity?' BMJ, 29 October 2005.

25. <https://www.ema.europa.eu/en/human-regulatory/overview/orphan-designation-overview>

*account social inclusion and psychological and educational development.*<sup>26</sup>

The role and the needs of carers of people living with a rare disease have often been underestimated. While many carers choose to commit themselves to looking after a loved one with a rare disease, they are often dismayed by the limited supports that are available, including respite care, which can impact on the wellbeing of both carer and patient.

### 1.6 Universal Health Care and Rare Diseases - A Discussion and Points of Action

An inequitable and complex mixed public-private health system has persisted in Ireland despite much criticism and repeated reform attempts<sup>27</sup>. This inequitable and unfair system inevitably has the highest impact on those living with a chronic/long-term disease which by definition can be over decades.

The all party Oireachtas Committee's Final Report (Sláintecare), published in May 2017, highlighted the need to move toward equitable access to a high-quality, universal, single-tier health system for Ireland. The report recommended the introduction of universal GP and primary care, reducing or removing out-of-pocket fees, and substantially increasing public health care expenditure and capacity in a tax-funded system. It further recommended that the two-tier system in public hospitals should be addressed by eliminating the provision of private care in public hospitals. Implementation strategies for Sláintecare have been put in place and should be given the time and resources to do their work. The Rare Disease Taskforce has met with the Sláintecare office and has urged that rare diseases are given much greater visibility as the Sláintecare plan is implemented.

The Long Term Illness (LTI) Scheme was enacted as part of the Health Act (1970) and covers the costs of most medicines and hospital care without a means test for some chronic diseases, including a few rare diseases. However, the vast majority of patients with

rare diseases are not covered by the LTI Scheme and are therefore subjected to significant additional health costs.

There are 16 conditions covered by the LTI Scheme, including the rare diseases of Haemophilia, Cystic Fibrosis, Phenylketonuria, Muscular Dystrophies, Hydrocephalus, Acute leukaemia and conditions arising from use of Thalidomide. It is very unfair and inequitable that rare diseases that would meet objective criteria for inclusion in the LTI scheme are presently excluded. However it would be equally unfair and inequitable that those patients who presently benefit from the LTI scheme should be excluded through a scrapping of the LTI scheme, which would in effect be contrary to the commitment to provide access to universal health care in the Sláintecare Report .

In short, all those living with serious and chronic rare disease(s) should be given access to the LTI. This would be a significant step towards universal health care in Ireland. At the very least all those living with a serious and chronic rare disease should be given a Medical Card, without means testing.

### 1.7 Role of Patient Advocacy Groups and Patient Advocates

The role of patient advocacy groups in the development of policy and services for people living with a rare disease has often been glossed over or underestimated. Most rare diseases do not have a dedicated patient advocacy group and are represented instead by the voluntary advocates (mostly parents) with few, if any, resources other than a home computer and a printer and a determination 'to do something' not only for their own children but also the children of others with the same disease.

One such person is Les Martin. Les is the dad of two children, Cathal (6yrs) and Ciaran (4yrs) who were both diagnosed with the ultra-rare disease Metachromatic Leukodystrophy. Ciaran has recently benefitted from a new and innovative gene therapy. Les has mounted an increasingly successful

<sup>26</sup> <https://assets.gov.ie/37342/da70fc6fadd24425b98311e679f4406b.pdf> (Page 62).

<sup>27</sup> Connolly, S and Maev-Ann Wren (2019). 'Universal Health Care in Ireland – What are the Prospects for Reform?' In Health Systems and Reform, ESRI

campaign to urge the Government to widen the Newborn Screening Programme from the present 8 screened conditions (all of which are rare diseases) to 40 conditions, which is the best standard in the EU. There has been much research that indicates that where therapies exist, there is a much improved prognosis because treatment can commence as soon as possible after birth. Ireland had one of the first new born screening programmes in the world in 1966 and hopefully, with prioritisation by Government, we can rise to become a leading country worldwide once again.

There are many challenges to running a successful patient advocacy group in Ireland in 2020. These include compliance with the Charities Act, 2009 (effective since 2014) and the requirements of the Charities Regulatory Authority; the Companies Acts and the requirements of the Companies Registration Office and the many regulations that cover fundraising.

Those rare diseases that have been represented by a patient group over many years can often point to improvements in health care for their patients that are a direct response to successful advocacy. There is also a concomitant need for adequate State resources to voluntary bodies, including Rare Diseases Ireland which remains very underfunded.

### 1.8 Impact of Patient Groups on Rare Disease Policy and Services

Of increasing importance in recent years has been the emergence of 3 networks that work together as part of the Rare Disease Taskforce; HRCI, IPPOSI and RDI. As a result of the advocacy of this Taskforce and positive interaction from the Department of Health/HSE, there have been a number of important developments including the implementation of some important aspects of the National Rare Disease Plan such as:

- The establishment of the National Rare Disease Office
- The roll out of the National Clinical Programme for Rare Diseases and the interaction with ERNs
- The shaping of Government policy on access to

new and innovative medications

- Highlighting patient concerns and helping to shape policy responses ‘when drugs go wrong’ (see 1.11)

There is increasing recognition of the importance of Public and Patient Involvement (PPI) in health policy and research in Ireland. For this to be meaningful, it requires a vibrant, informed, evidence based, independent and resourced patient advocacy sector.

A compliant/silent patient groups sector is ultimately not in the interest of Government, as complaints or even observations about gaps and weaknesses in policy become unsaid or unheard, even when evidence is presented. Concomitantly the health sector is often forced to bounce from one crisis to the next and issues that could be resolved by mediation and early intervention are increasingly resolved through the courts, at huge public expense and often with much delay and related patient grief.

A different approach to PPI and health policy is now needed, building on the good examples that do exist. The regular meetings between the Rare Disease Taskforce and the Department of Health in respect of the implementation of the National Rare Disease Plan have been in general a positive experience, with important outcomes for patients and health providers, albeit the pace of implementation is never as fast as originally hoped and there remains many gaps in the implementation of the NRDP.

### 1.9 Patient Registries and Rare Diseases

The absence of a clear national strategy on patient registries for both common and rare diseases in Ireland is a significant weakness in the present health system in Ireland.

The existing ad-hoc Registry system in Ireland means that some disease areas, such as Cancer, Cystic Fibrosis and Skin conditions are comparatively well covered by effective Patient Registries, whereas many other conditions are lucky to have information on an excel sheet held by a patient advocacy group or on a consultant’s desk-top computer. In short, vital macro information on the extent and main characteristics and course of some diseases can either be non-

existent or partly available or not published through lack of resources. A further problem is that pharmaceutical companies are reluctant to carry out clinical trials for new and innovative medications in the absence of a Registry, thus denying patients potential early access to new drug therapies or at the very least the opportunity to participate in important health research.

A Patient Registry is defined as an organised system that uses observational study methods to collect uniform data (clinical and other) to evaluate specified outcomes for a population defined by a particular disease, condition, or exposure and that serves predetermined scientific, clinical, or policy purpose(s).

Generally, the few more developed Registries in Ireland publish an annual report and the board of governance of the most advanced involves a mix of both clinicians and patient group representatives. There is no strategic approach to fund or develop Registries in Ireland and little sign yet that it is a public policy priority. HRCI and IPPOSI have been calling for a national Patient Registry Strategy since 2011 with limited progress to date.

Patient Registries can:

- Capture disease demographics, clinical outcomes and survival rates
- Support patient recruitment for clinical research
- Support the undertaking of research studies and clinical trials
- Support pharmacovigilance
- Allow patients access to their own data, through patient portals
- Allow patients to submit their own data e.g. quality of life data or patient reported outcomes<sup>28</sup>.

A number of important stepping stones should be put in place to support the emergence of a Patient Registry Strategy. These include a Contact Registry that could offer the potential to:

- Enable patients in Ireland with a rare disease to receive better care
- Better understand specific rare diseases in Ireland
- Expand both clinical and scientific rare disease research in Ireland
- Make patients better aware of specific patient registries in Ireland.

The funding of a service to provide advice and practical tools for emerging patient registries would also be an important priority.

### 1.10 An Intercultural Dimension to Rare Diseases

Some minority ethnic groups in Ireland, both indigenous and more recent migrant groups, have a higher incidence of particular types of rare diseases. There is a growing body of evidence that these minority groups can face significantly higher barriers in accessing health services in Ireland, including a lack of prioritisation at a policy level and fewer resources for diagnosis and treatment.

This means that additional, targeted strategies are needed for 'at risk' minority ethnic groups, particularly those that might experience multiple forms of disadvantage and discrimination. Two such groups in Ireland are Travellers and people in Ireland with Sickle Cell Disease (SCD) who were originally from Africa or Asia and who are now Irish citizens or with refugee, asylum seeker or 'leave to remain' status.

Irish Travellers are a recognised minority ethnic group indigenous to Ireland with a population of around 40,000 people. They experience very severe health inequalities, as documented in the All Ireland Traveller Health Study, 2010<sup>29</sup>. It is also recognised that consanguinity contributes to inherited rare diseases within the Traveller community in Ireland. The 3 most common rare diseases within the Irish Traveller community are: Galactosaemia; Hurler Syndrome and I-cell disease. The higher carrier frequencies compared with the Irish population are as follows:

28. <https://hrci.ie/the-nuts-and-bolts-of-patient-registries-expert-presentations/>

29. UCD Study Team. 'The All Ireland Traveller Health Study'. 2010. University College Dublin.

**Published carrier frequencies of Travellers and Irish Population-wide recessive conditions<sup>30</sup>**

Disease	Carrier Frequency in Irish Travellers	Carrier Frequency in Irish Non Travellers
Galactosaemia	1/11	1/107
Hurler syndrome	1/10	1/81
I-cell disease	1/15	1/512

Early diagnosis of Sickle Cell Disease (SCD) through new-born bloodspot screening (the heel-prick test) significantly improves outcomes through a combination of both therapies and patient education. Despite the growing incidence of SCD in Ireland, the severity of the disease and the availability of therapies that are of optimised benefit when there is an early diagnosis, SCD is not one of the conditions presently screened in Ireland as part of the heel-prick test. A strong rationale for the inclusion of people living with SCD in the new born screening programme and additional resources for treatment was highlighted as far back as 2015 in a study published in the Irish Medical Journal,

*'We identified delays in referral and treatment, which reflect the lack of government funded support and policy. We suggest all maternity units commence screening for newborns at risk of SCD. It is a cost effective intervention with a number needed to screen of just 4 to prevent a potentially fatal crisis<sup>31</sup>.*

SCD is the commonest genetic condition worldwide but it is still a rare disease in Ireland. SCD has many symptoms including sudden and excessive pain. SCD is a chronic inherited autosomal recessive condition which has a significant impact on morbidity and

mortality, though it is difficult to predict the clinical course of the disease between one individual and another<sup>32</sup> (see Annex 4). SCD is also one of the diseases where gene editing/Crispr is being investigated as a potential therapy.

### 1.11 Rare Diseases Caused by the Side Effects of Drugs

Rare diseases can also occur as the result of a man-made action, including both prescription and recreational drug use such as alcohol. The misuse of recreational drugs can have side effects that cause birth defects, including Foetal Alcohol Syndrome, a condition arising from alcohol exposure during the mother's pregnancy, which can cause brain damage and growth problems.

The subsequent inaction of health authorities, sometimes from fear of litigation or acknowledging mistakes, can sometimes compound these devastating occurrences. For example, in the wake of the Thalidomide scandal, there was an ineffective recall of the morning sickness drug in Ireland, after it had already been fully withdrawn in the UK and Germany<sup>33</sup>.

FVS - Foetal Valproate Syndrome (see Annex 4) is a range of devastating birth defects that can occur from the side effects of taking an anti-epilepsy or other drugs which contain valproic acid (VPA) during pregnancy. Despite studies dating back to the 1980's, both industry and national governments across the world (including Ireland), were slow to act on overwhelming evidence of the side effects of such drugs, including Epilim. Progress on addressing this issue is thanks to the advocacy of one Irish parent, Karen Keeley. Thanks to Karen and the patient advocacy group she founded, the Organisation for Anticonvulsant Syndrome (OACS) Ireland, combined with an European Medicines Agency judgement in February 2018, there are now considerably heightened warnings about the dangers of FVS<sup>34</sup>. The

30. Lynch, Sally Ann et al. 'Catalogue of Inherited Disorders found among the Irish Traveller Population.' J Med Genet, 22 Jan, 2018

31. Gibbons, C et al. Irish Medical Journal. 'Sickle Cell Disease: Time for a Targeted Neonatal Screening Programme'. February 2015 <https://www.lenus.ie/handle/10147/559231>

32. Geoghegan, R. RANP in Haemoglobinopathy Children's health Ireland, 2019 <http://www.cuh.hse.ie/Children-s-Services/News-Events/SCD-in-children-an-overview-09-04-2019.pdf>

33. <https://www.thejournal.ie/irish-government-thalidomide-prime-time-investigation-1226065-Dec2013/>

Health Products Regulatory Authority (HPRA) in Ireland has played an important role in ensuring there is heightened warnings about drugs containing valproic acid.

A FVS hospital Consultant has been appointed to improve services to affected children and adults. Karen's advocacy work was also given important support from groups involved in the Rare Disease Taskforce, Epilepsy Ireland, Cystic Fibrosis Ireland and the Disability Federation of Ireland through the FACS Forum<sup>35</sup>.

### **1.12 The shortage of expert medical staff in Clinical Genetic Services in Ireland is fast approaching a crisis point.**

Ireland employs just 4 consultant Clinical Geneticists. The Royal College of Physicians of Ireland (RCPI) endorses the need for 15 consultant Clinical Geneticists to be employed in Ireland, based on a recommended ratio of 0.3 per 100,000 of the population. The ratio in Ireland at present only 0.1. Of immediate concern to patient groups is the age profile of the existing consultants and the likelihood of depletion of this already scarce expert resource through retirement.<sup>36</sup>

34. [http://www.hpra.ie/homepage/medicines/special-topics/valproate-\(epilim\)](http://www.hpra.ie/homepage/medicines/special-topics/valproate-(epilim))

35. <https://www.epilepsy.ie/sites/www.epilepsy.ie/files/SV%20Rapid%20Assessment%20Report%20FINAL.pdf>

36. HSE and NDTP, 2019. Review of the Clinical Genetics Medical Workforce in Ireland.

## Section 2

# Living with a Rare Disease

# The Challenge and the Determination



## Section 2: Living with a Rare Disease – The Challenge and the Determination

This is the most important section of this Easyguide. A number of the patient advocacy groups involved in this publication approached one of their members to ask them to describe what it is like living with or caring for someone with a rare disease in Ireland in 2020. We thank those who have contributed. What comes across very strongly from their insights is ‘I might have a rare disease, but it does not have me’.

Of particular poignancy is that some of those featured in the first Easyguide (2013) are no longer with us. Gerry Walker for example. Gerry had Cystic Fibrosis and survived to the unexpected age of 50 years old, thanks in large part to an extraordinary commitment to fitness and adherence to therapies. Gerry received a double lung transplant in 2007,

which helped prolong his life. He was a senior statistician in the Central Statistics Office (CSO) for many years and a strong friend and advisor to Cystic Fibrosis Ireland.

With the kind permission of Tony and Mary Heffernan we have included a page from the 2013 Easyguide that features Liam and Saoirse Heffernan, both of whom died at 5 years of age from the ultra-rare Batten Disease. Many children with a rare disease do not survive into adolescence/adulthood. Their inclusion also shows the commitment of many parents to continue fighting for more research and better services through their patient group for the sake of other families, even when their own children have passed away.

### Profiles of people living with a rare disease featured:

PROFILE	Name	Rare Disease	Brief Description of Disease
1	Emma Fogarty	Epidermolysis Bullosa (EB)	Rare genetic disease. Causes skin layers and internal body linings to separate and blister at the slightest touch
2	Liam and Saoirse Heffernan	Batten Disease	Ultra rare genetic disease. Over time, affected children suffer mental impairment, worsening seizures, and progressive loss of sight and motor skills
3	Sharon Tracey	Stargardt Disease	Rare genetic disease. Causes a gradual decline in central vision. Side (peripheral) vision is usually preserved
4	David Comer	B-Cell Lymphoma	Rare cancer. A primary central nervous system lymphoma

PROFILE	Name	Rare Disease	Brief Description of Disease
5	Dara Woods	Hereditary Haemorrhagic Telangiectasia (HHT)	Rare genetic disease. Vascular disease causes severe recurrent, unexplained nosebleeds, often requiring transfusions. May also include shortness of breath, anaemia, haemorrhage, brain abscess and seizures.
6	Killian Fitzgerald	Phenylketonuria (PKU)	Rare genetic and metabolic disease. If undiagnosed/unmanaged can lead to profound irreversible intellectual and physical disability
7	Áine Lawlor	22q11.2 Deletion Syndrome	Rare genetic disease. Characteristic signs and symptoms include heart defects, palate defects resulting in feeding, speech and language difficulties and mild differences in facial features.
8	Katie Moore	Cystic Fibrosis (CF)	Rare (worldwide) genetic and metabolic disease. Impacts particularly on the lungs and digestive system but also many other organs of the body
9	Chloe Hayes	Juvenile Huntington's Disease (HD)	Rare <b>genetic neurodegenerative condition</b> . As brain cells die physical, cognitive and emotional symptoms appear and gradually worsen over time.
10	James Ennis	Cystinosis	Rare genetic and metabolic disease. Symptoms include excess thirst and urination, bone pain from rickets and loss of essential nutrients from the kidneys with kidney transplant inevitable, even with good treatment adherence
11	Dylan Finglas	Multiple Sulfatase Deficiency (MSD)	Ultra rare genetic disease. The condition is neurodegenerative and progressive. It is classified as a lysosomal storage disorder.

PROFILE	Name	Rare Disease	Brief Description of Disease
12	Sandra Phair	Cavernoma	Rare and ultra rare group of genetic diseases. A cavernoma is a cluster of abnormal blood vessels found predominantly in the brain and/or spinal cord. Symptoms can include strokes, seizures, partial/full paralysis, sight, speech and/or hearing problems.
13	Brendan Gallagher	Alpha-1 Antitrypsin Deficiency (AATD)	Rare genetic disease where the body does not produce enough alpha-1 antitrypsin (AAT). AAT is an important protein which protects the lungs against cigarette smoke and bacterial infection.
14	Sean O’Kelly	Spina Bifida	Spina bifida is the most common neural tube defect (NTD), which causes incomplete development of the spinal cord in the womb. Symptoms of spina bifida include hydrocephalus, varying degrees of paralysis, pressure sores, loss of sensation of the lower limbs, malformations, latex allergies, social and sexual issues, and bowel and bladder incontinence.
15	Joyce Stokes	Multiple System Atrophy (MSA)	Multiple system atrophy (MSA) is a progressive neurodegenerative disease. It is characterised by a combination of symptoms that affect both the part of the nervous system that controls involuntary action such as blood pressure or digestion and movement. The symptoms reflect the progressive loss of function and death of different types of nerve cells in the brain and spinal cord. It can sometimes be mistaken for Parkinson’s Disease which is also a progressive neurodegenerative disease.
16	Grace O'Malley	Spinal Muscular Atrophy (SMA)	Spinal Muscular Atrophy (SMA) is a rare neuromuscular condition causing extreme weakness and lifelong muscle wasting, affecting the arms and legs, as well as breathing, swallowing and head control.

See also Annex 4 of this publication for further descriptions of rare diseases, including those in the first Easyguide (2013).



PROFILE 1

# Emma Fogarty

## LIVING WITH EPIDERMOLYSIS BULLOSA (EB)

*“The one thing that gets me up every day and keeps me positive is knowing that there is something being done to fight the battle against EB.”*

My name is Emma Fogarty. I am 35 years of age and I live with Epidermolysis Bullosa (EB). Having EB means that my skin is as fragile and delicate as the wings of a butterfly and is just as easily damaged. The slightest knock or rub can cause my skin to blister and come off. 80% of my body is covered in bandages to protect open wounds and prevent further damage. It also affects me internally and externally and I have to take high levels of various medications throughout the day to try ease the pain.

But I never let it get me down. The one thing that gets me up every day and keeps me positive is knowing that there is something being done to fight the battle against EB. Research into better treatments and a possible cure for EB is happening worldwide and increasingly over the last few years there are more and more glimmers of hope.

This year I celebrated my 10th year as Patient Ambassador for DEBRA Ireland and I do everything I can to raise much needed awareness for the charity and the condition. I know

that I am very lucky to support and have the support of such a well-established organisation. It is so important to have an organisation to fight on your behalf...otherwise there would be no hope and I would have no reason to get up in the morning. I am happy to see something is being done for people with other rare conditions because I can only imagine how hard it would be to fight a separate battle alone. Living with a rare disease is enough of a battle for any family.

**A podcast of Emma talking about her life and EB is available:**

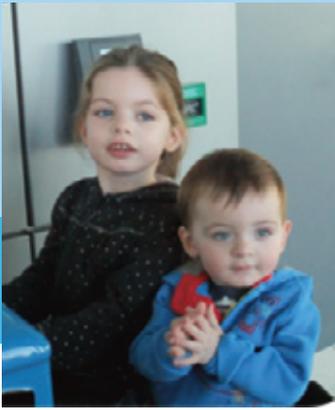
<https://podtail.com/en/podcast/the-other-side-of-perfect/emma-fogarty-talks-eb-life-outside-the-condition-d/>

## RARE DISEASE

### About Epidermolysis Bullosa (EB/DEBRA)

Epidermolysis Bullosa (EB) is a distressing and painful genetic condition causing skin layers and internal body linings to separate and blister at the slightest touch. It affects approximately 1 in 18,000 babies born, equating to approximately 300 people in Ireland, and can range from mild to severe. Severe forms can be fatal in infancy or lead to dramatically reduced life expectancy, due to a range of complications from the disease.

Patients with severe EB need wound care and bandaging for several hours a day and the condition becomes increasingly debilitating and disfiguring over time. Adult patients with severe forms are extremely susceptible to an aggressive form of skin cancer. There is currently no treatment or cure for EB. In Ireland, children with EB are cared for at the multidisciplinary clinic in Children's Health Ireland, Crumlin and adult patients at a multi-disciplinary clinic in St. James's Hospital. Care requires the involvement of many different clinical specialists to extend life span and improve quality of life.



## PROFILE 2

# Liam and Saoirse Heffernan

## BATTEN DISEASE

**“Children become totally disabled and eventually lose all bodily functions”**

*Two Children who were featured in the original Easyguide (2013) were Liam and Saoirse Heffernan who both had the ultra rare condition known as Batten Disease. Saoirse Heffernan died in 2011 aged 5 and Liam, her brother, died in 2014 also aged 5. The lives of their two children inspired their parents Tony and Mary Heffernan to establish the well known and respected patient advocacy group ‘Bee for Battens’. In tribute to their memory and to the work of Bee for Battens, The Saoirse Foundation and BUMBLEance, we reprint the profile that was in the 2013 Easyguide.*

Liam has an ultra-rare condition known as Batten disease, which is an inherited genetic disorder of the nervous system that usually manifests itself in childhood. Batten disease is named after the British paediatrician who first described it in 1903. It is one of a group of disorders called neuronal ceroid lipofuscinoses (or NCLs).

Early symptoms of Batten disease (or NCL) usually appear in childhood when parents or doctors may notice a child begin to develop vision problems or seizures. In some cases, the early signs are subtle, taking the form of personality and behaviour changes, delayed speech, slow learning, and clumsiness or stumbling. Over time, affected children suffer mental impairment, worsening seizures, and progressive loss of sight and motor skills. Children become totally disabled and eventually lose all bodily functions. In Liam’s case, he was diagnosed when he was just 18

months old, following the diagnosis of his sister, Saoirse. Batten disease is not, at this time, preventable. To date it has always been fatal. Saoirse passed away on Jan 18th 2011, aged 5 years, 7 months and 14 days.

On May 3rd 2011, Liam became the youngest ever child to undergo brain surgery as part of a treatment trial at Weill Cornell Hospital in New York. While the immediate months and first year after the procedure Liam showed significant signs of improvement, unfortunately, Liam has now started to deteriorate, and his parents face the fact that they will lose their remaining child to a rare disease.

## RARE DISEASE

### About Batten Disease

Batten's disease is an ultra rare, inherited disorder of the nervous system that usually manifests itself in childhood. It is one of a group of disorders called neuronal ceroid lipofuscinoses (NCLs). While it affects only a tiny number of children in Ireland, it is believed to be severely undiagnosed. The forms of NCL are classified by age of onset have the same basic cause, progression and outcome, but are all genetically different. The defective gene, inherited from both parents, causes malfunction at a cellular level. Early symptoms of Batten disease (or NCL) usually appear in childhood with vision problems or seizures. In some cases the early signs are subtle, taking the form of personality and behaviour changes, delayed speech, slow learning, clumsiness or stumbling. Autistic traits and dementia can also feature quite severely. Over time, affected children suffer mental impairment, worsening seizures, and progressive loss of sight and motor skills. Children become totally blind and disabled. To date it has always been fatal. Being an ultra rare condition, international collaboration in research is proving most successful in attempts to learn more and find a cure.



## PROFILE 3

# Sharon Tracey

## LIVING WITH STARGARDT DISEASE

***“Visual impairment can be seen as a barrier to a full life, but it doesn’t have to be.”***

Sharon Tracey was 18 yrs when she was diagnosed with Stargardt disease. She wore glasses throughout her school years but could never see the blackboard clearly; she just assumed everyone had blurred vision. It wasn’t until she began having problems navigating that she visited her GP and insisted they refer her to a specialist.

Sharon was told that her eyesight would deteriorate and there was nothing they could do for her. At the time Sharon was still grieving her grandmother, who raised her, and it felt like her world had collapsed around her. She was in a dark place. Today, Sharon’s love for life is palpable, and contagious. “I made a decision a long time ago to think about my disability as a challenge to overcome, not a barrier. That mental shift changed my life.”

Sharon trained as a blind telephonist in Sligo and secured work experience with the Midland Health Board Tullamore. Since then her career has taken her to Guinness in Dublin, the Department of Education in Athlone and eventually back to Tullamore to the Department of Education and Skills where Sharon is an Executive Officer.

And that’s just the day job. “I love learning and meeting new people and have qualifications in Equality Studies, Counselling skills, Life Coaching, Reflexology, Access Consciousness, Integrated Energy

Therapy and Reiki.” explains Sharon.

“I also give motivational talks on the subject of disability. I’m a strong believer in everyone acknowledging their abilities and accepting their limitations and asking for help. We’re all human, we all need support – whether you have a disability or not. Visual impairment can be seen as a barrier to a full life, but it doesn’t have to be.”

Sharon is also a media ambassador for the charity Fighting Blindness and regularly speaks to radio stations and newspapers about the importance of research and support.

## RARE DISEASE

### About Stargardt Disease

Stargardt disease is the most common form of inherited juvenile macular degeneration, usually first identified in childhood or adolescence. It is a genetic condition that affects the central region of the retina, known as the macula. Light sensitive cells, called photoreceptors, are found here and are responsible for fine, detailed central and colour vision. In Stargardt disease, these crucial photoreceptors degenerate over time and people with this condition experience a gradual decline in their central vision. Side (peripheral) vision is usually preserved.

Stargardt disease has a prevalence estimated between one in 8,000 and one in 10,000 people. The carrier rate has been quoted as high as 1 in 20 to 1 in 50 people.

As of yet, there are no approved therapies or treatments, but retinal research is entering an exciting era where safe and effective interventions are realistic goals.



## PROFILE 4

# David Comer

## LIVING WITH CENTRAL NERVOUS SYSTEM (CNS) B-CELL LYMPHOMA

*“I was extremely lucky to survive”*

Prior to my illness I had been suffering from a headache in the left side of my forehead for about 3 weeks. This resulted in me suffering a seizure on April 4th 2018 at about 8pm in the evening. As a result of this I was taken to University Hospital Galway and subsequently moved to Beaumont hospital to begin the journey. A tumour was discovered in my brain and this was also discovered to be cancerous.

I was diagnosed with a rare form of cancer. CNS B-Cell Lymphoma was my diagnosis. It is a disease which affects the central nervous system. According to research carried out this form of cancer accounts for less than 1% of diagnosed forms of Lymphoma. The treatment for same is a very intensive cycle of chemotherapy to initially deal with the tumour, followed by a stem cell transplant at St James’s Hospital. The Chemotherapy process was in blocks of 6 days as an inpatient at University Hospital Limerick on 4 separate weeks with 2 weeks in between. During the 6 day cycle 7 doses of chemotherapy drugs would be administered. When this treatment was completed I was referred to St James’s hospital where I proceeded to undergo the transplant. This was a difficult process as I suffered from numerous infections and sickness. On the 11th November 2018 I was released after a successful transplant and have not looked back since.

Back in 2018 I was a 42 year male

employed as a Detective Garda in An Garda Siochana at Tuam Garda Station. I would have been of relatively good health and would have had healthy eating habits and exercised regularly by cycling approximately 200 kilometres on a weekly basis with friends. I had moved into my own house approximately 2 years prior to this. I lived quite a normal life and enjoyed socialising on a regular basis with friends and work colleagues. I would have been a keen golfer and held a full driving licence and drove on a daily basis. I would have been fairly independent in my lifestyle and had not been involved in a relationship with another person for just over three years. I enjoyed life but would have been hard on myself and negative towards life itself.

Since recovering from my illness, I have been diagnosed with Epilepsy and although this is far from suffering with cancer, it does inhibit my former lifestyle. I am unable to drive and have had to move home to

my parents' house, where I currently still reside. This occurred as a result of three surgeries I underwent to my brain prior to treatment beginning as a tumour had to be partially removed prior to my diagnosis

I am part of a close family with both of my parents still working and 2 sisters living close by. They have 7 children between them so I would have enjoyed spending time in their company when not working. All in all I was living a very comfortable life prior to getting unwell due to the illness I suffered from during 2018.

I now live a much different life. I have started to plan holidays, something I could not do for almost 18 months and had not done for the previous 3 years.

The disease I was diagnosed with, Central Nervous System B-Cell Lymphoma, which I was extremely lucky to survive, has probably opened my eyes up to life and realise how brittle life can be. I hope that due to the wonderful work carried out by the various medical professionals and nursing staff at University Hospital Galway, Beaumont Hospital, University Hospital Limerick and Saint James's Hospital in Dublin. Also I have received great support from both the Irish Cancer Society and Cancer Care West in Galway, both of whom provide phenomenal service to people in my precarious position. I also hope that I take better care of my life in the future.

#### RARE DISEASE

##### About B-Cell Lymphoma

A type of cancer that forms in B cells (a type of immune system cell). B-cell lymphomas may be either slow-growing or aggressive/fast-growing. Most B-cell lymphomas are non-Hodgkin lymphomas. There are many different types of B-cell non-Hodgkin lymphomas. Prognosis and treatment depend on type and stage of the cancer.



## PROFILE 5

# Dara Woods

## LIVING WITH HEREDITARY HAEMORRHAGIC TELANGIECTASIA (HHT)

Dara Woods is a married mother of 2 from Donegal, living and settled in Swords, Co Dublin who has a rare and hereditary genetic disorder of blood vessels called Hereditary Haemorrhagic Telangiectasia (HHT).

***“I came from a family of nose-bleeders.”***

As a child, Dara suffered unexplained and recurrent nosebleeds,

*‘I came from a family of nose-bleeders. Nothing unusual was suspected as 3 out of 4 of my siblings also suffered nosebleeds, as did our Dad.’*

However, as Dara grew older she spent most days dealing with her nosebleeds. Her pregnancies were difficult due to severe haemorrhaging daily through her nose. Nasal cautery was performed many times. Both of her children were also nose-bleeders but it wasn't until they became young teenagers they were diagnosed with HHT,

which is a blood-vessel disorder where unexplained nosebleeds are the outward sign.

*‘Genetic testing of both my children & myself confirmed we all had HHT’* explained Dara.

Sadly, her son Paul lost his life unexpectedly to HHT while on holidays in Paris at the young age of 22 years. HHT in his lungs had caused sudden rupture. His sister Katie continues to live an active and full life with her HHT. Constant monitoring & treatment of her HHT helps her to remain healthy.

### RARE DISEASE

#### **About Hereditary Haemorrhagic Telangiectasia:**

HHT - Hereditary Haemorrhagic Telangiectasia - is a genetic disorder of the blood vessels causing AVMs (Arterio Venous Malformations) in many vital organs eg: Brain, Liver, Lungs and Gastro-Intestinal Tract. Telangiectasias are small AVMs and appear like tiny red dots on face, skin, lips, fingertips and lining of nose. 90% patients suffer recurrent & unexplained nosebleeds, often requiring blood transfusions and iron infusions. Other symptoms may include shortness of breath, anaemia, haemorrhage, brain abscess & seizures. Each child born of a HHT parent has 50-50 chance of inheriting the disorder. Knowing the signs and symptoms of HHT can lead to a faster diagnosis.



## PROFILE 6

# Killian Fitzgerald

## LIVING WITH PHENYLKETONURIA (PKU)

*“85% of real food is off limits”*

Killian Fitzgerald from Limerick has Phenylketonuria (PKU). As an adult Killian finds weight management to be a very challenging aspect of the metabolic disorder as 85% of real food is off limits and the replacement medical and low protein foods are highly processed. Killian also found the transition from child to adult services difficult as the level of contact with the medical team is significantly reduced.

Killian regularly attends PKU Association of Ireland (PKUAI) meetings and receives tremendous support from those within the PKU online community. As a child he could never have imagined having friends that live with this rare metabolic disorder. It was not until he was fifteen years old that he first met another person with PKU and takes great comfort in knowing that he is not the only one living with the challenging metabolic disorder PKU,

‘We need more awareness. In my whole life I would say that five people knew what PKU was when I explained to them that I had it. I would love to see this change.’

### RARE DISEASE

#### About Phenylketonuria (PKU):

PKU is a rare genetic disease that affects a person’s ability to breakdown protein resulting in ‘restricted low protein dietary therapy’ for life. It is diagnosed through newborn blood-spot screening. If unmanaged PKU can lead to profound irreversible intellectual and physical disability. There is currently no cure for PKU. It is managed in Ireland through restrictive dietary therapy since the introduction of newborn screening in 1966. Research indicates that 60% of adults and 70% of adolescents do not strictly adhere to dietary therapy and it still leads to suboptimal outcomes signifying the real need for additional medical supports. Kuvan® a medicine benefiting a subgroup of PKU patients was recently approved for reimbursement in Ireland, ten years after EMA authorization, however the lack of HSE resources means it cannot be prescribed for eligible patients. Ireland has one of the highest incidences of PKU in the world at 1:4,500 compared with 1:10,000 worldwide.



## PROFILE 7

# Áine Lawlor

## LIVING WITH 22Q11.2 DELETION SYNDROME

***“Everyone was ahead in [school] work and I was always behind and nobody knew why”***

Áine is 36 years of age. From the time she was very young she felt different to other children. She was frequently ill and struggled to learn in school where she was bullied and unhappy and felt like a failure. ‘Everyone was ahead in work and I was always behind and nobody knew why,’ she remembers.

Áine was finally diagnosed aged 15 with 22q11.2 Deletion Syndrome. The diagnosis ‘explained a lot,’ says Áine,

*‘My life changed. People started to realise I was a bit different,’ she continued. ‘I’m not shy anymore. I love my life even though I have 22q. It can get hard but I truly believe in myself, that’s how I carry on through the days. Áine has a Fetac Level 5 qualification in Childcare and has a part time job in a creche. She is a ten-pin bowler with Special Olympics.*

These outlets keep her going. ‘It’s not all that bad having 22q,’ she reports. A 22qYEEP member (Young Experts by Experience Panel) Áine takes part in participatory action research and speaks to various audiences about her 22q lived experience.

Áine is quick to acknowledge that she has help carrying on,  
*‘My mother is my strength. She helps me to be strong. Without her I don’t know where I’d be’*

Áine’s mother (Anne) co-founded [www.22q11ireland.org](http://www.22q11ireland.org) which provides family peer support, information and regular family outings along with an annual conference. The group works in partnership with clinicians to develop integrated care pathways.

**RARE DISEASE****About 22q11.2 Deletion Syndrome**

22q11.2 Deletion Syndrome (DS) is caused by the deletion of a small piece of chromosome 22. The condition affects an estimated 1 in 2- 4,000 people worldwide. The features of this syndrome vary widely, even among affected members of the same family, and involve many parts of the body. Characteristic signs and symptoms include heart defects that are often present from birth, palate defects resulting in feeding, speech and language difficulties and mild differences in facial features. People with 22q11.2 DS often experience recurrent infections caused by problems with the immune system.

Aside from the varied physical health issues, many children with 22q11.2 DS have developmental delays and learning disabilities. Later in life, they are at an increased risk of developing mental illnesses such as schizophrenia, depression, anxiety, and bipolar disorder. Additionally, affected children are more likely than children without 22q11.2 to have attention deficit hyperactivity disorder (ADHD) and developmental disorders, such as autism, that affect communication and social interaction. Children with this disorder have complex care needs which require an appropriate care response tailored to each child's individual needs.



## PROFILE 8

# Katie Moore

## LIVING WITH CYSTIC FIBROSIS (CF)

***“Thanks to better medication and services and research (and determination), one quarter of the adult CF population in Ireland now have children”***

My baby boy Milo is 7 months old now - I still can't believe how lucky I am to be his mum, it's something I'll never take for granted. I'm 33 years old and was diagnosed with Cystic Fibrosis at 6 months of age. Noel and I are together almost 11 years now. We always talked about kids, and hoped we would be lucky enough to have some of our own. I'm very maternal and babysat for almost everyone in my town! When I turned 30 I decided to concentrate extra hard on my health and try prepare myself for getting pregnant, fingers crossed.

I was nervous at the thought of pregnancy and the possibility of becoming unwell, but it definitely wasn't going to stop me. I took all the extra vitamins and kept up my fitness to try my best at staying healthy and strong.

Although I live in Mayo, I was referred to the Coombe Maternity Hospital in Dublin, which deals in high-risk pregnancies and my doctor mentioned that due to my fitness (having run marathons), it would all stand to me during the pregnancy.

I'm a self-employed visual artist, I work mainly on commissions or bursaries. I had finished up a year-long project where I was awarded the New Work Award with Arts and Disability Ireland, and I had also just graduated from my Masters course.

Cystic Fibrosis is still a very tough disease to live with, in so many ways. However thanks to better medication and services and research (and determination), one quarter of the adult CF population in Ireland have children of their own now- something that would have been almost unheard of 20 years ago.

## RARE DISEASE

### About Cystic Fibrosis

Cystic fibrosis (CF) is a progressive, genetic multi organ disease that primarily impacts on the lungs and digestive system. CF causes persistent lung infections and limits the ability to breathe over time. At a basic level CF causes a build-up of thick and sticky mucus in various organs.

In the lungs, the mucus clogs the airways and traps germs, like bacteria, leading to infections, inflammation, respiratory failure, and other complications. For this reason, minimizing contact with germs is a top concern for people with CF, including from other people with CF.

In the pancreas, the build-up of mucus prevents the release of digestive enzymes that help the body absorb food and key nutrients, resulting in malnutrition and poor growth. In the liver, the thick mucus can block the bile duct, causing liver disease. CF also impacts on fertility in both men and women, Ireland has the highest level of CF in the world with 1 in 19 of the indigenous population carrying the altered CF gene. There have been significant improvements to CF care in recent years, though still more to do.



## PROFILE 9

# Chloe Hayes

## LIVING WITH JUVENILE HUNTINGTON'S DISEASE

*“Huntington’s is like having Alzheimer’s, Parkinsons’ and Motor Neuron Disease at the same time”*

Chloe Hayes lives with her mother Geraldine in Mullingar. 18 year-old Chloe has Juvenile Huntington’s Disease, a hereditary neurodegenerative condition. Geraldine remembers Chloe as a bright and happy schoolchild. At seven, Chloe started to experience rigidity of movement and uncharacteristic temper tantrums. Her teachers were concerned as her progress at school deteriorated. When her National Educational Psychological Service (NEPS) assessments indicated learning difficulties increasing over time Geraldine pushed for Chloe to have further tests and she was eventually referred to a Pediatrician.

Chloe’s parents split up before she was born and she grew up with little contact with her Dad. When Chloe was nine, Geraldine received the devastating news that Chloe's Dad had Huntington's Disease (HD) and was living in a nursing home. HD is hereditary. Each child of an affected parent has a 50% chance of inheritance. Symptoms usually appear between the ages of 35 and 50 years but about 5-10% of people with HD become symptomatic before age 20 (Juvenile HD).

An official diagnosis of Juvenile HD three years later confirmed Geraldine’s worst fears. Chloe had to leave her school after first year, so Geraldine decided to relocate from their home in County Meath to Mullingar. There, Chloe could attend St Brigid’s Special School and they had family support nearby. While at St Brigid’s, Chloe availed of speech and language therapy, physiotherapy and occupational

therapy and both she and Geraldine received excellent support from the school. Geraldine said,

‘Huntington’s is like having Alzheimer’s, Parkinsons’ and Motor Neuron Disease at the same time. As a mother I am often overwhelmed by the challenges Chloe endures but her magnificent smile keeps me going, her graduation last year was a very proud and happy time for us all’.

## RARE DISEASE

### About Huntington's Disease (HD)

Huntington's Disease is a life-limiting genetic neurodegenerative illness caused by a mutation in the 'Huntington' or 'HD' gene which leads to the destruction of certain brain cells. As brain cells die physical, cognitive and emotional symptoms appear and gradually worsen over time. In Juvenile HD the symptoms occur in childhood or adolescence and tend to follow a more rapid course.

In Ireland there are approximately 700 people living with HD and a further 3000 people at risk. Currently there is no cure however specialist multi-disciplinary treatment and care is essential to manage symptoms and enhance quality of life. Recent advances in research worldwide bring significant hope of gene therapies to target the root cause of HD.



## PROFILE 10

# James Ennis

## LIVING WITH CYSTINOSIS

***“The thing that gets me up...  
...is knowing that there is a lot of research being done into better treatments and cures for cystinosis worldwide”***

My name is James Ennis. I am 29 years old and I live with cystinosis. I have to take 30 tablets a day. The most important one is Cystagon which I must take every 6 hours to keep the cystine levels from building up. I also use Cysteamine eye-drops four times daily to prevent crystal build up in my eyes. I have had a kidney transplant.

I am a qualified joiner and worked in kitchen manufacturing. I had to give up work because of bad leg pain which severely affected my mobility. I have had surgery and physiotherapy but I am no better and now use a walking stick.

I try not to let it get me down. I became involved with Cystinosis Ireland around two years ago and became the Northern Ireland

Representative. I am also a member of the Cystinosis Community Advisory Board, which is part of Cystinosis Network Europe. The thing that gets me up every day and keeps me positive is knowing that there is a lot of research being done into better treatments and cures for cystinosis worldwide. I can see a glimmer of hope for the cystinosis community.

### RARE DISEASE

#### About Cystinosis

Cystinosis is caused by an inherited, recessive gene where the amino acid, cystine, accumulates in all organs, muscles and bones. Cystinosis is ultra-rare and has an incidence of 1:250,000. Children are usually diagnosed between 6 months and 3 years, but misdiagnosis is common. Patients present with excess thirst and urination, bone pain from rickets and loss of essential nutrients from the kidneys. The kidneys are the first organs affected, with kidney transplant inevitable, even with good treatment adherence. The sooner a child is diagnosed and the quicker they start treatment, the better the outcome.

The available current treatment slows deterioration but does not halt it. Two formulations of the treatment exists: Immediate-release, taken every six hours which means waking up every night and delayed-release taken every 12 hours, but is not yet widely available worldwide. Both formulations cause severe side effects and patients find it difficult to adhere to the regime. Gene therapy is at early stage clinical trial. Cystinosis Ireland ([www.cystinosis.ie](http://www.cystinosis.ie)) supports families, raises awareness and raises money to fund research.



## PROFILE 11

# Dylan Finglas

## LIVING WITH MULTIPLE SULFATASE DEFICIENCY (MSD)

*“Dylan started to gradually regress after 3 years of age, losing all gained skills.”*

Dylan Finglas, is 7 years old and was diagnosed with Multiple Sulfatase Deficiency (MSD) just after his 2nd birthday in late 2014. The diagnosis was very quick after a private consultation at Temple Street Hospital. Some of Dylan's best achieved skills were walking with assistance, being able to throw a ball with a reasonable amount of accuracy and being able to feed himself a biscuit.

Dylan started to gradually regress after 3 years of age, losing all gained skills. He lost the ability to walk and eventually to sit up unassisted. Any words he had were lost as was the ability to feed orally (now peg fed) and his eyesight has deteriorated to the point that he is now blind. Although Dylan is mentally and physically disabled now, he loves music and will laugh and smile most days with stimulation. He absolutely loves hydrotherapy and this is

greatly beneficial as he is non active. His family are saddened by the lack of hydrotherapy facilities that exist.

The early diagnosis, especially given that Dylan was so well at diagnosis led his family to establish the first ever charity to advocate for MSD research among other rare disease patient advocacy. They are called MSD Action Foundation/SavingDylan.com.

### RARE DISEASE

#### About Multiple Sulfatase Deficiency (MSD)

Multiple Sulfatase Deficiency, an ultra-rare condition, is a clinically devastating and fatal condition in children. Depending on age of onset, children affected by MSD, in most cases, do not live long enough to see their 10th birthday. The condition is neurodegenerative and progressive. It is classified as a lysosomal storage disorder.

The deficient gene is called the SUMF1 gene. Mutations or deletions on the gene result in a misfolded controlling enzyme called the Formylglycine Generating Enzyme. As a result, substrates build up in cells in the body including the brain and they become toxic. The myelin sheath does not form properly in the brain as a result. MSD is similar to Metachromatic Leukodystrophy and Sanfilippo Syndrome.



PROFILE 12

# Sandra Phair

## LIVING WITH CAVERNOMA

*“Sandra recognizes the importance of being connected to others that have the same condition”*

In 2005 at the age 26, Sandra had her first brain haemorrhage. She was diagnosed with a bleed from a cavernoma in the brain stem. She has the rare form of cavernoma, familial cerebral cavernous malformation (FCCM), with multiple cavernomas throughout the brain, and a small benign meningioma. She has subsequently been diagnosed with the CCM3 gene.

Before her first haemorrhage, Sandra had no symptoms. She was sent home from hospital with no information, just the name ‘cavernoma’. Sandra knew that she had a rare disease and had to become the expert in the disease. She was after all living with the disease every day. Sandra now knows more than most of her doctors and nurses about the disease.

Eight months after her first haemorrhage, Sandra had a second one, from the same cavernoma. She was at high risk of further haemorrhages, but surgery was not an option because of its location. Gamma knife radiosurgery was performed. A year later Sandra had brain surgery to remove two more cavernomas and a benign meningioma in the frontal/temporal lobe.

Sandra is not just living with the risk of further haemorrhage; she is also living with a range of invisible symptoms including chronic pain, migraine, neuro-fatigue and tinnitus. Sandra in fact has an acquired brain injury.

Sandra recognizes the importance of being connected to others that have the same condition. She has been supported by Cavernoma Alliance UK, and together with Kay McGrath, has recently launched the Cavernoma Ireland Support Group.

“Knowing you are not alone, on this path. We stand together, supporting, and searching for a cure.”

**RARE DISEASE****About Cavernoma**

A Cavernoma (also known as cavernous angioma, cavernous haemangioma or cerebral cavernous malformation) is a cluster of abnormal blood vessels found predominantly in the brain and/or spinal cord. It looks like a raspberry, and can measure from a few millimetres to several centimetres.

Symptoms, which depend on size and where in the brain or spinal cord the cavernoma occur, include strokes, seizures, partial/full paralysis, sight, speech and/or hearing problems. Cavernoma can bleed at random intervals, which can make symptoms worse, and this is the most feared complication of cavernoma.

Familial cerebral cavernous malformation (FCCM) has an estimated prevalence of 1/5,000 -1/10,000. Cavernoma causing symptoms are more rare (1/12,500). A study in Scotland found that each year 1 person out of 400,000 is diagnosed with a symptomatic brain cavernoma.

FCCM has a genetic origin, with one of three genes implicated (CCM1, CCM2 and CCM3). FCCM is transmitted from parent to child with a 50% risk of inheriting the mutated gene. In others the cavernoma is sporadic. The CCM3 gene is the rarest of the genetic mutations that cause cavernomas. There are only 30-50 people in Europe who are diagnosed with the CCM3 gene and an equivalent number in the United States.



PROFILE 13

# Brendan Gallagher

## LIVING WITH ALPHA-1 ANTITRYPSIN DEFICIENCY

***“Access to treatment would be a real life-line for me and others with Alpha-1”***

I received a diagnosis of Alpha-1, which is a genetic condition in 1996, and was referred to Beaumont Hospital Respiratory Consultant Professor Gerry McElvaney and his research team and have attended the Alpha-1 clinic since 2001. I have been affected to some degree by Alpha-1 for over 23 years but in the last 5 years have suffered diminished lung function.

I remain committed to the future - family, love for my wife, four children, their spouses and eight grandchildren. No other members of my family or siblings have symptoms of Alpha-1, COPD or Emphysema. I have been a non-smoker throughout my life as were my parents, but I had frequent exposure to passive smoking during teenage years and to environmental conditions such as diesel fumes in my working career.

I always loved cycling but I no longer cycle or use my car for even short commutes. I am fully dependent on portable oxygen and even walking short distances requires a break of approximately 2 minutes. Basic physical activities such as gardening, DIY, climbing the stairs are all reduced, and are carried out at a much slower pace. Through a DCU fitness research programme I joined a gym and do some basic exercises with a fitness coach. The gym combined with much-requested Grandad duties keep me busy. I have had to adjust to a different lifestyle

and gain support and understanding from my family and friends when having to equip myself with a portable oxygen backpack and nasal tubes to partake in social or family activities.

There is a treatment for patients with Alpha-1, the cost of which is not covered by the HSE in Ireland. Access to treatment would be a real life-line for me and others with Alpha-1.

## RARE DISEASE

### **About Alpha-1 Antitrypsin Deficiency (A-1)**

Alpha-1 antitrypsin deficiency (AATD) is a rare genetic disorder where the body does not produce enough alpha-1 antitrypsin (AAT). AAT is an important protein which protects the lungs against cigarette smoke and bacterial infection. AAT deficiency can lead to lung, liver and skin disease. Most people present with chronic obstructive pulmonary disease (COPD). Over 3,000 individuals on the island of Ireland suffer from a severe form of AATD, and 1 in 25 individuals are carriers of an abnormal AAT variant. Unfortunately, AATD is under-diagnosed and long delays in diagnosis are common. The WHO recommends screening of people with COPD, poorly-controlled asthma, and liver disease. Advantages of an early diagnosis of AATD include increased lung and liver surveillance, family member testing, smoking cessation opportunities, and reduction of occupational and environmental exposures.



PROFILE 14

# Sean O'Kelly

## LIVING WITH SPINA BIFIDA

*“...go reach your dreams because you will get there”*

My name is Sean O'Kelly, I am a 27-year-old disability activist living with spina bifida and hydrocephalus. I am a wheelchair user.

With regards to how spina bifida and hydrocephalus has affected my life - over the past number of years, I have found it difficult to gain employment. I am currently still on the job hunt! I don't know if this has been impacted directly because I have spina bifida and hydrocephalus but being a wheelchair user is definitely a factor.

I became a disability activist in 2016 out of sheer frustration, particularly in the area of public transport having experienced being stranded at Clontarf DART (Dublin Area Rapid Transit) Station in Dublin. I have co-founded a group called 'Access for All'. This is to highlight the fact that lifts at both DART and train stations are consistently 'out of order'.

Outside of activism, I am a DJ on Dublin South FM. My show is once a week from 4-5pm. I enjoy this as it helps me get the message of disability out in the media which is always important.

My advice for people living with lifelong medical conditions; feel the fear and do it anyway - go reach your dreams because you will get there!

## RARE DISEASE

### About Spina Bifida

Spina bifida is the most common neural tube defect (NTD), which causes incomplete development of the spinal cord in the womb. Translated, it literally means 'split spine'. The spine is made up of separate bones called vertebrae which normally cover and protect the spinal cord. With spina bifida, one or more of these vertebrae are not completely formed. Instead, they are split, and the spinal cord and its coverings usually protrude through a sac-like bulge on the back, covered with a thin membrane.

There are three main types of spina bifida: occulta, meningocele and myelomeningocele.

Myelomeningocele is the most severe and occurs when the spinal cord/neural elements are exposed through the opening in the spine. Effects of spina bifida include hydrocephalus, varying degrees of paralysis, pressure sores, loss of sensation of the lower limbs, malformations, latex allergies, social and sexual issues, and bowel and bladder incontinence. Spina bifida affects about one in every 1,000 children born per year in Ireland. Ireland has one of the highest incidences of spina bifida births in the world.



## PROFILE 15

# Joyce Stokes

## LIVING WITH MULTIPLE SYSTEM ATROPHY

*“Being mistaken for a drunk when you are trying to do some shopping and ask for something is a real trial”*

Joyce Stokes from Wicklow was diagnosed with the neurodegenerative disease, multiple system atrophy (MSA) in 2014, she was just 41, with a young family to look after, when her initial diagnosis of Parkinson’s Disease was reassessed and MSA was diagnosed.

She feels very lucky that her local doctor has been willing to research and find out about the disease to enable her to get the support she needs from speech and language therapists, physiotherapists and other services. So many professionals in the health and care sector have never heard of MSA and when Joyce was admitted to hospital recently because of a fall she found it hard having to explain what was wrong with her that caused her fall.

Affecting all autonomic functions, MSA causes a person to become trapped in their own body. As Joyce says: ‘being mistaken for a drunk when you are trying to do some shopping and ask for something is a real trial.’ She needs time to form her words and with her speech slowing down even further, Joyce wonders why everyone must rush so much.

The disease is slowly denying Joyce of all opportunities to mix with other people. She feels embarrassed about speaking or walking in social situations. She is becoming more confined to the house.

She finds Katie, the MSA Trust Nurse

Specialist who travels to Ireland for a week every quarter to meet with people who have MSA at support groups and MSA clinics, a lifeline. Her family has the opportunity to discuss any changes in symptoms with Katie and ask for advice when medication is changed, or dose altered. Katie also communicates via email when Joyce is too tired to speak.

One of the hardest things for Joyce is trying to support her children as they watch their mother deteriorate and they get upset by the lack of empathy from their peer group and unkind comments when they see their Mum struggle with her walking or speech. Lack of understanding about Joyce’s condition is incredibly hard on all the family.

## RARE DISEASE

### About Multiple System Atrophy (MSA)

MSA is an indiscriminate neurodegenerative disease affecting around 3,300 adults in the UK and Ireland. Roughly 4 people per 100,000 are affected. Its cause is unknown, and average lifespan from initial symptom onset is just 7 years. There is no cure.

Affecting all autonomic functions, MSA causes a person to become trapped in their own body. Over only a small number of years, they experience problems with swallowing ranging from uncontrollable drooling to choking on the smallest piece of food; vocal cord paralysis rendering communication with loved ones virtually impossible; impotence; and total incontinence. They become unable to walk, and eventually become entirely bedbound. Towards the end of their life they require around the clock care. Intellect, along with the ability to feel pain, does not diminish in any way.

Due to its appalling symptoms, it is unsurprising that in MSA Trust's recent survey, 80% of members with MSA reported feelings of depression, and 82% said that a reduced ability for them to carry out social activities had led to social isolation.



PROFILE 16

# Grace O'Malley

## LIVING WITH SPINAL MUSCULAR ATROPHY (SMA)

*"I started to miss key developmental milestones such as pulling myself up in the cot and taking my first steps."*

**My name is Grace O'Malley, and I have SMA type II. When I was born, there was no indication of the disease and my parents excitedly took me home from hospital just like any other mother and father. However, it soon became apparent that all was not right. I started to miss key developmental milestones such as pulling myself up in the cot and taking my first steps.**

In the years that have followed, I have had many trips to hospital with chest infections and other complications such as scoliosis for which I require ongoing surgery every year.

My body continues to deteriorate, and this has accelerated as I grow in physical size. For example, as a younger child I could lift my arms to answer a question at school and could write without difficulty. I am now ten years of age and I am in 4th class at school, and already I have lost these abilities.

My parents have been fantastic as I require help with everything, washing, brushing my teeth, dressing and even toileting. That being said, I count myself as one of the lucky ones. Many with more severe forms of the disease have to be suctioned, ventilated and even peg fed.

I have a great electric wheelchair which I am just about able to drive myself. This gives me some independence and makes a big difference. I try not to worry about the future and enjoy the most out of every day.

### RARE DISEASE

#### About Spinal Muscular Atrophy:

SMA is a rare genetically inherited neuromuscular condition affecting circa 1 in 10,000 worldwide. The SMA gene mutation causes extreme weakness and lifelong muscle wasting, affecting the arms and legs, but also breathing, swallowing and head control. People with SMA are grouped into one of four types based on severity. Historically, 95% of children born with type I SMA have died before their second birthday.

SMA Ireland represents the 25 children and 30 adults living with the condition in Ireland. In May 2017, "Spinraza" was the first drug to receive EMA approval for the treatment of SMA. The HSE have approved this drug for under 18's on a case-by-case basis. Research towards ever more effective treatments continues.



# Section 3: Priorities for the Programme for Government 2020 and beyond

This section outlines the 10 key priorities identified by patient groups for the Programme for Government, 2020 and beyond, including for inclusion in existing and future major health plans, such as Sláintecare and the annual HSE Service Plans. These 10 priorities seek to be strategic, evidence based and affordable rather than a detailed wishlist where the most important priorities can become invisible through excessive detail or too long a list.

An important feature of these priorities is that they are consistent with much of existing and forthcoming Government policy, including Sláintecare, which has the support of all the main political parties in Ireland. The personal testimonies in Section 2 of this Easyguide demonstrate the central importance of 'empowering, protecting and supporting rare disease patients and their carers'<sup>37</sup>. This includes the continued building of an effective, evidence based and resourced patient advocacy sector in Ireland.

Major advances in care for rare diseases in Ireland and worldwide can often be linked in part at least to the work of a patient advocacy group. This Easyguide seeks to ensure that we 'leave no one behind', as promised by the UN's 2030 Sustainable Development Goals, with an onus on everyone in both the statutory and voluntary sectors being mindful and supportive of emerging and under-resourced patient groups and individual advocates for improved care of those with rare diseases. The 10 priorities are as follows:

## Rare Diseases: 10 priorities for the next Programme for Government

1. **Full implementation of the National Rare Disease plan (NRDP).** The implementation of the National Rare Disease Plan has been extended since 2018 and there has been progress in some key areas, but much remains to be implemented, including on an Ireland North/South basis.
2. **Additional resources for prevention, diagnosis and care.** National Centres of Expertise (CoEs) and European Reference Networks (ERNs) for groupings of rare diseases is a key commitment in the NRDP. Increasing Ireland's participation in ERNs has a key role to play as does better cooperation between Ireland, North and South. A national Patient Registries Strategy for rare diseases should be prioritised. A Contact Registry and funding for a service to provide advice and support for emerging Registries is an important stepping stone.
3. **Additional resources and responsibilities for the National Rare Diseases Office.** Additional resources are needed to implement the National Clinical Programme for Rare Diseases's (NCPRD's) Model of Care (published in 2019) and Transitional Model Of Care (published in 2018) across rare diseases, and to develop the National Rare Diseases Office (see Annex 2). It is proposed that a business case is drawn up and additional responsibilities applied, including for example the oversight of a system of Patient Registries for

37. Department of Health. National Rare Disease Plan 2014-2018 (extended 2019). Section 6.

rare diseases in Ireland and expanding the ERN's as part of the National Clinical Programme (see also priority 2).

4. **To improve access to and reimbursement times for new and innovative medications for rare diseases.** These improvements should have the ambition of ensuring Ireland is regularly placed in the top 7 countries in the EU for orphan drugs according to the annual EFPIA/W.A.I.T Report<sup>38</sup>. The Technology Review Committee for Rare Diseases is presently dormant despite being only recently established and needs urgent reactivation including the appointment of a new Chairperson.
5. **Adequate funding of key clinical genetic services. Most rare diseases are genetic.** Clinical genetic services in Ireland are severely underfunded/under-prioritised. Current clinical genetics staffing levels are among the worst in Europe and between 70-80 per cent below those recommended by the Royal College of Physicians in London and in recent months staffing levels have deteriorated further. Adequate staffing levels for diagnostics and counselling need to be addressed as a priority to a standard that is at least equivalent to the UK.
6. **Leadership and infrastructure in Genetic and Genomic Medicine.** The HSE and UCD/TCD universities are currently progressing a lead post for Genetic and Genomic Medicine. This appointment is imminent at time of publication of this Easyguide and is strongly supported by the Rare Disease Taskforce. The new Professor will lead a proposed HSE National Genetic and Genomic Medicine Network which needs to be adequately resourced. A new commitment to recruit a National Clinical Laboratory Director needs to be completed as a matter of urgency in Q1 2020.
7. **Sláintecare mainstreaming and targeting.** To ensure that rare diseases are fully mainstreamed

into relevant national health reform strategies, in particular Sláintecare and the restructuring of regional healthcare in the implementation of the recently announced 6 regional healthcare structures in Ireland. To ensure marginalised groups including those on low incomes, Travellers and migrants to Ireland are included in targeted rare disease strategies.

8. **New Born Screening. Ireland is lagging behind other countries in respect of new born screening.** The forthcoming review of the existing new-born screening programme should seek to expand the number of rare diseases screened in Ireland at birth from 8 to at least 20 in the short-term which is the practice in many other EU countries. This expanded screening should be based on evidence, emerging good practice in Europe and through periodic review. Expansion to screening for 40 rare diseases would bring us in line with best-practice in Europe. We have an opportunity to be best-in-class and should strive to be a leader in this field of medical science.
9. **Health cost and social service supports.** Improving income and social service supports for people and families living with a rare disease, including for example broadening access to the Long Term Illness Scheme/Universal Health Care, and improved residential respite care for children and adults living with a rare disease.
10. **Independent Living.** To ensure that people with rare diseases have better access to supports for independent living, (where appropriate) access to disability friendly accommodation; education and training strategies and employment strategies and access to fertility treatments and related supports as appropriate, including IVF.

### An overarching, all-Island Priority

The full impact of the UK, including Northern Ireland leaving the European Union in 2020, has yet to be

38. W.A.I.T stands for Patients Waiting to Access Innovative Therapies, Report produced by IQVIA (IMS Health and Quintiles) based on availability and average time between marketing authorisation to post marketing authorisation.



*Alan Finglas (MSD Action), Linda McGrath (HCRI), Ken Rogan (IPPOSI), Vicky McGrath (RDI) and Caitriona Dunne (Fighting Blindness)*

determined at time of the publication of this Easyguide. The Rare Disease Taskforce will work in partnership with the Northern Ireland Rare Disease Partnership in 2020 and beyond to offset the potential negative impact of BREXIT on those living with a rare disease and will continue to optimise cooperation on the island of Ireland.

## Section 4

# Rare Disease **Policy and Research** Contexts



# Section 4: Contexts

There are three policy contexts covered in this part of the Easyguide. These are:

## 4.1. Key Policy Commitments on Rare Diseases at an EU and UN Level

## 4.2. Rare Disease Research in Ireland: The State of Play

## 4.3. Ireland North/South Collaboration

### 4.1 Key Policy Commitments on Rare Diseases at an EU and UN Level

The United Nations 2030 Agenda for Sustainable Development Goals<sup>39</sup>, pledges to ‘leave no one behind’ in the quest for physical and mental health and wellbeing. In September 2019 the UN member states, including Ireland, adopted a Political Declaration on Universal Health Coverage<sup>40</sup> which includes rare diseases. This marks the first time that rare diseases have featured in a UN declaration adopted by all 193 member states.

The World Health Organisation (WHO) supports countries on the path to Universal Health Coverage<sup>41</sup>, aiming to ensure all people have access to the health services they require. The Fair Pricing Forum<sup>42</sup> opens dialogue between regulators, pharmaceutical companies, insurance providers and patient groups to ensure sustainable access to medicines, including orphan drugs.

On an EU level, the European Commission is

increasingly supportive of collaborative initiatives to tackle rare diseases. The EU committee of experts on rare diseases (EUCERD 2010-2013)<sup>43</sup> and the European Commission Expert group on Rare Diseases (CEG-RD 2013-2016)<sup>44</sup> were established to provide expert guidance on the implementation of the Recommendation on action in the Field of Rare Diseases<sup>45</sup> adopted by the European Council in 2009. This recommendation supported the adoption of national strategies and plans to address the comprehensive needs of people living with rare diseases including diagnosis, care, support and treatment. The Irish Government supported this recommendation by publishing its own National Rare Disease Plan, 2014-2018 (subsequently extended in 2019).

The work of EUCERD and CEG-RD have been supported by two EU joint actions for rare diseases EUCERD Joint Action (2012-2015)<sup>46</sup> and RD-Action (2015-2018)<sup>47</sup>. EUCERD was established to develop and implement rare disease policies. RD-ACTION, co-founded via the 3rd EU Health Programme, elaborated upon these policies and improved codification of rare diseases.

In 2011, the European Commission adopted a Directive on Cross Border Health Care and Patient Rights<sup>48</sup> which paved the way for the establishment of the European Reference Networks (ERNs). The first ERNs were launched in 2017, this series of virtual networks aim to bring together the expertise of healthcare providers across Europe to concentrate

39. <https://www.un.org/sustainabledevelopment/development-agenda/>

40. <https://undocs.org/en/A/RES/74/2>

41. <https://www.who.int/mediacentre/news/statements/2018/rare-disease-day/en/>

42. [https://www.who.int/medicines/access/fair\\_pricing/fair\\_price\\_report\\_2019/en/](https://www.who.int/medicines/access/fair_pricing/fair_price_report_2019/en/)

43. <http://www.eucerd.eu/>

44. <https://ec.europa.eu/transparency/regexpert/index.cfm?do=groupDetail.groupDetail&groupID=3015&news=1>

45. <https://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF>

46. <http://www.eucerd.eu/>

47. <http://www.rd-action.eu/>

48. [https://ec.europa.eu/health/cross\\_border\\_care/overview\\_en](https://ec.europa.eu/health/cross_border_care/overview_en)

knowledge and resources for complex and rare diseases which require highly specialised treatment. Ireland's participation in ERN's is coordinated through the National Clinical Programme for Rare Diseases. Ireland is currently a member of 3 ERN's, with a further 16 applications currently under review at a European level. It is expected that we will learn the outcomes of these applications before the end of 2020.

EURORDIS- Rare Diseases Europe played an integral role in the adoption of rare disease and orphan medicine legislation at the EU level<sup>49</sup>. EURORDIS works to ensure that patients and patient advocate groups play a central role in driving the implementation and adoption of both national and international programmes and strategies for rare diseases.

EURORDIS are coordinating a foresight study, Rare 2030<sup>50</sup>, which seeks to propose policy recommendations that will improve the lives of people living with a rare disease in Europe over the next ten years and beyond. Rare 2030 brings together the views of a large number of patients, practitioners and key opinion leaders to achieve this aim.

In 2019, the EU launched a new five-year European Joint Programme on Rare Diseases<sup>51</sup> which brings together resources at a national and European level involving research funders, organisations and infrastructure; universities; hospitals and patient organisations to develop a 'comprehensive sustainable ecosystem' for rare disease research (see 4.2 for more information).

#### 4.2 Rare Disease Research in Ireland: The State of Play

For most rare diseases there is no cure and the management of symptoms can be very hampered by a lack of knowledge and by limited evidence. Rare disease research in Ireland is often driven by patients and their families or by clinical experts who see the immense needs first-hand. Most of our strongest research-focused rare disease charities are patient-

led and there are also pockets of research excellence among the rare disease clinical community. Regardless of who the research is initiated by, there tends to be very strong patient involvement and the rare disease community have long demonstrated many of the principals of what is now formally referred to as PPI (patient and public involvement).

While not solely a funding channel for rare diseases, the Joint Funding Scheme, run in partnership by Health Research Charities Ireland (HRCI), previously known as the Medical Research Charities Group (MRCG), and the Health Research Board (HRB), has proven to be an important funding avenue for rare disease research. **This scheme is open to all 40 members of HRCI, of which approximately one third support rare disease research. Government investment in this scheme is up to €1 million per year, which is matched by the charities, thereby facilitating the co-funding of competitive research projects. Since 2010, close to €10m has been invested by the HRB in rare disease research, the majority of which has been through the Joint Funding Scheme. This funding spans 12 rare diseases and the majority of the research falls into the category of applied biomedical, although any form of health research is fundable through the scheme.**

Irish involvement in the 24 European Reference Networks (ERNs) for rare diseases will increasingly become a very important driver of research, through facilitating networks, the inclusion of the Irish population in patient registries and, in all likelihood, by opening up future funding avenues. In addition, patient impact from research will be facilitated through the development and implementation of the evidence-based clinical practice guidelines that will be developed within each of the ERNs. Following considerable effort by the National Clinical Programme for Rare Diseases and the Rare Disease Office, we are on track to see official Irish involvement in the majority of the ERNs in 2020.

Through the HRB, and together with 22 other

49. <https://www.eurordis.org/>

50. <https://www.rare2030.eu/>

51. <http://www.ejprarediseases.org/>

## SECTION 4: CONTEXTS

countries, Ireland is participating in the European Joint Programme on Rare Diseases<sup>52</sup>, opening the possibility for Irish scientists to collaborate on transnational, patient-focused projects. Only one of the projects awarded funding in 2019 included an Irish partner but we hope that will increase in future calls. More locally, the UCD-led Rare Disease Research Partnership (RAinDRoP)<sup>53</sup> has recently brought together many stakeholders with an interest in rare diseases, to prioritize and progress research in Ireland.

While there are many positives, challenges remain for research into rare diseases in Ireland. As highlighted, at a meeting focused on rare disease clinical research<sup>54</sup>, held by the Rare Disease Clinical Programme in 2018, protected research time and practical supports for clinicians to undertake clinical trials and other forms of clinical research remain a huge barrier. Clinical trials can give patients early, and sometimes the only, access to new and innovative therapies and so the Irish population are disadvantaged by this. There have been recent

positive developments in this regard, with a first Action Plan for Health Research<sup>55</sup> launched by the HSE in 2019, but it will remain a challenge to ensure a policy and practice focus on research, in the face of many immediate care needs.

The lack of a national strategy for genomics in Ireland, along with lack of support for patient registries are other major factors hampering Irish progress in rare disease research. A recent position paper by HRCI, entitled Research for a Healthier Ireland<sup>56</sup>, highlights the need for progress in these areas, along with other important aspects of health research in Ireland relevant to rare diseases.

As we look to the future, more will be required to create formal and resourced rare disease research networks in Ireland and to facilitate our increased involvement in international networks. It is also important that we focus on research as a critical part of the solution for improved rare disease care, rather than treating it as a separate and less urgent endeavour.



*Photo: Most of the Irish contingent at the European Conference for Rare Disease, Vienna 2018. The group includes many rare disease research advocates, including representatives from the Rare Disease Office, Cystinosis Ireland, Rare Diseases Ireland, Retina International, 22q11 Ireland, RAinDRoP, Fighting Blindness, HRCI, PTC Therapeutics, the National Clinical Programme for Rare Diseases, DEBRA Ireland, Friedreich's Ataxia Research Alliance Ireland and Muscular Dystrophy Ireland. Photo credit: Dr Suja Somanadhan.*

52. <https://www.hrb.ie/funding/funding-schemes/all-funding-schemes/grant/european-joint-programme-on-rare-diseases-ejp-rd-joint-transnational-call-2020/>

53. <https://www.nmhs.ucd.ie/news-and-events/raindrop-rare-disease-research-partnership>

54. <https://www.hse.ie/eng/about/who/cspd/ncps/rare-diseases/resources/ncprd-clinical-research-workshop-report-2018.pdf>

55. <https://hseresearch.ie/wp-content/uploads/2019/12/10-Year-Action-Plan.pdf>

56. <https://hrci.ie/wp-content/uploads/2019/09/HRCI-Position-Paper-2019-1.pdf>

### 4.3. Ireland North/South Collaboration on Rare Disease Policy and Related Networking

An important dimension to this Guide is the focus on collaboration between Northern Ireland and the Republic of Ireland at a number of different levels.

The strongest element of this collaboration is between patient groups, north and south. Every year for the past 10 years there has been an all island conference held to mark international rare disease day, which is typically the last day of February every year- a rare date in the calendar in a leap year. These are important opportunities for cooperation between the Rare Disease Taskforce and the Northern Ireland Rare Disease Partnership (NIRDP).



*Dr Helen McAneney (QUB); Dr Suja Somanadhan (UCD) and Dr Amy McKnight QUB At the Centre for Public Health, QUB.*

The NIRDP is a 'Not for Profit' Company and a NI Registered Charity. They bring together those living with a rare disease and organisations representing them; clinicians and other health professionals; researchers and producers of specialist medicines and equipment; health policymakers and academics.

The aim of the NIRDP is that no one is disadvantaged because of the rarity of their condition. The NIRDP

aims to work constructively with stakeholders and service delivery organisations to find practical ways of improving the quality of life, treatment and care for those with rare diseases in Northern Ireland.

To do this, the NIRDP works closely with the Northern Ireland Department of Health; the Health and Social Care Board, Public Health Agency and the Patient Client Council; and with Patient Representative Organisations in the UK (Genetic Alliance UK; Unique) Republic of Ireland (HRCI, IPPOSI and RDI- the Rare Disease Taskforce) and Europe (EURORDIS), as well as with a wide range of condition-specific groups and organisations.

There is already some collaboration on the delivery of health services that impacts positively on people with a rare disease in Ireland, north and south. If you are entitled to public health services in Ireland, you may opt to access those services in another member state of the European Union (EU) or European Economic Area (EEA), which also includes Iceland, Liechtenstein and Norway, and be repaid the cost in your own home country if you meet the requirements. This is provided for by the Cross-Border Healthcare Directive (EU Directive 2011/24/EU).<sup>57</sup>

The amount that will be repaid is the cost of the public healthcare treatment in Ireland, or the cost of your treatment abroad, if that is less. It does not include other costs such as travel.

Treatments that qualify for the Treatment Abroad Scheme are not covered under the Cross-Border Healthcare Directive. In general, the Treatment Abroad Scheme covers treatments that are not available in Ireland while the Cross-Border Healthcare Directive only covers treatments that are publicly funded and available in Ireland (for further information search online Citizens Information Centre/NI Citizens Advice Bureaux).

The National Rare Disease Office (see Annex 2) can provide information on the processing for accessing specialised care via the Treatment Abroad Scheme and the Cross Border Directive, helping to locate relevant specialists.

57. [www.hse.ie/eng/services/list1/schemes/cross-borer-directive/about/](http://www.hse.ie/eng/services/list1/schemes/cross-borer-directive/about/)

Some examples of north south cooperation (which all have a strong rare disease dimension, but are not restricted to rare diseases) include:

- **North-West Cancer Centre in Altnagelvin Hospital** in Derry which commenced in 2016 and which works closely with Letterkenny hospital on prostate, breast, lung, bowel, bladder and head and neck and lymphoma.
- **Crumlin Children's Hospital** in Dublin which is the site for the childrens' cross-border cardiac surgery programme.
- **Belfast City Hospital and Beaumont Hospital** in Dublin cooperate on kidney transplants, particularly from living donors.
- **Mater Hospital Dublin** which is the site of the National Lung Transplant Programme and which is currently seeking increased cooperation on lung transplants on a north/south basis, but with limited progress to date because of resources.
- **North West Rare Disease Forum** launched in 2016 in Derry, the Forum provides support and advocacy for those with rare diseases, including on a north/south regional basis.

The full impact of the UK, including Northern Ireland leaving the European Union in 2020, has yet to be determined at the time of publication of this Easyguide. However if 'no one left behind' is to be realised, it is incumbent on the authorities in Belfast and Dublin to ensure that people with rare diseases and their families are not disadvantaged by BREXIT in Ireland north and south. Patient advocacy groups will make this a priority in 2020 (see also Section 3).

# Annexes



# ANNEX 1: Glossary of Key Terms

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## **Autosomal dominant genetic conditions**

These are conditions whereby a person needs only to inherit one changed copy (alteration) of the gene in order to be affected by the condition, or become affected by the condition later in life. The changed gene is dominant over the normal gene.

---

## **Autoimmune Disorders**

More than 80 diseases occur as a result of the immune system attacking the body's own organs, tissues, and cells. Some of the more common autoimmune diseases include type 1 diabetes, rheumatoid arthritis, systemic lupus erythematosus, and inflammatory bowel disease.

Although the causes of many autoimmune diseases remain unknown, a person's genes in combination with infections and other environmental exposures are likely to play a significant role in disease development.

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## **Autosomal recessive genetic conditions**

These are conditions whereby a person has to inherit two changed copies (alterations) of the gene (a changed copy from each parent) to be affected by the condition. A person who has only one copy of the changed gene will be an unaffected carrier.

---

## **Autosomes**

We have 23 pairs of chromosomes. Pairs number 1 to 22 are called autosomes and look the same in men and women. Pair number 23 are different in men and women and are called the sex chromosomes.

---

## **Carrier of a changed gene**

A person who is generally not affected with the condition (at that moment), but carries one copy of a changed gene. In the case of recessive conditions, the person will not usually be affected; in the case of dominant conditions, the person may become affected at a later stage.

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## **Cell**

The human body is made up of millions of cells, which act like building blocks. Cells in different parts of the body look different and do different things. Every cell (except for eggs in women and sperm in men) contains two copies of each gene.

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## **Chromosomes**

Thread-like structures which can be seen under the microscope and contain the genes. The usual number of chromosomes in humans is 46. One set of 23 chromosomes we inherit from our mother and one set of 23 chromosomes we inherit from our father.

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<b>Chromosome disorder</b>	These are conditions that affect the structure of a chromosome, where a piece of chromosome material can be missing or extra or a whole chromosome is missing or extra.
<b>Chromosome testing</b>	This is called karyotyping and involves looking at the overall structure of the chromosomes to check for large pieces of missing or extra chromosome material. This test cannot show small or subtle changes in chromosomes.
<b>DNA</b>	A chemical substance which makes up the genes, and which contains the information needed for the body to work.
<b>Family tree</b>	A diagram to show the people in your family who do and do not have the genetic condition, and how they are related to you and to each other.
<b>Gene</b>	Information needed for the body to work, stored in a chemical form (DNA) on chromosomes.
<b>Gene alteration or mutation</b>	A change in a gene that is sometimes also known as a gene alteration. Sometimes when a gene is changed, its information is altered so it does not work properly. This may cause a genetic condition.
<b>CRISPR gene editing</b>	CRISPR gene editing is a genetic engineering technique in molecular biology by which the genomes of living organisms may be modified. It is based on a simplified version of the bacterial CRISPR-Cas9 antiviral defence system. It is increasingly recognised as having considerable potential in treating genetic diseases, including many rare diseases but the technology is still at an early stage. <sup>58</sup>
<b>Genetic condition</b>	A condition or disease caused by an abnormality in a gene or chromosome.
<b>Genetic counselling</b>	Provides individuals or family with information and support regarding health concerns which run in their families. It may involve the diagnosis of a genetic condition, the provision of information and support. Genetic counselling does not involve therapeutic counselling.
<b>Genetic counsellor</b>	Graduate non-medical health professionals with specialist training in genetics and counselling. They provide information and support to help families adjust and understand new and complex information.
<b>Genetic test</b>	A test which can help identify if there is a change in a particular gene or chromosome. It is usually a blood or tissue test.

58. For an excellent introductory overview of the potential of gene editing and safeguards needed, see <https://www.bbc.co.uk/iplayer/episode/m000dt7d/storyville-the-gene-revolution-changing-human-nature> Storyville: 'The Gene Revolution' broadcast 27 Jan, 2020.

<b>Genotype</b>	Genotype is one of three factors that determine phenotype, along with inherited factors, epigenetic factors and non-inherited environmental factors.
<b>Hereditary condition</b>	One that is inherited (passed down through families).
<b>Metabolism</b>	The process your body uses to get or make energy from the food you eat. Food is made up of proteins, carbohydrates and fats. Chemicals in your digestive system break the food parts down into sugars and acids, your body's fuel. Your body can use this fuel right away, or it can store the energy in your body tissues, such as your liver, muscles and body fat.
<b>Metabolic disorder</b>	Occurs when abnormal chemical reactions in your body disrupt the metabolic process. When this happens, you might have too much of some substances or too little of other ones that you need to stay healthy. A metabolic disorder can either be inherited or acquired and can affect major organs of the body.
<b>National Rare Disease Plan</b>	Irish Government Plan on Rare Diseases commenced in 2014 and extended in 2019 arising from EU Council Recommendation (2009) and covering all EU countries.
<b>National Rare Diseases Office</b>	Based in the Mater Hospital (see Annex 2 of this Easyguide).
<b>Orphan Disease and Drugs</b>	An orphan drug is a pharmaceutical agent that has been developed specifically to treat a rare medical condition, the condition itself being referred to as an orphan disease. The reimbursement costs are often higher than other drugs because of high research and development costs and the relatively limited number of patients that can benefit from an 'orphan drug'.
<b>Phenotype</b>	In genetics, the phenotype of an organism is the composite of the organism's observable characteristics.
<b>Predictive testing</b>	A genetic test for a condition that may or will occur later in life. This testing is available to healthy individuals who are pre-symptomatic (no signs and symptoms of condition) but who are at risk of the condition due to their family history.
<b>Sporadic</b>	This means that a genetic condition can happen for the very first time in the person who has the condition and is not usually inherited from a parent.
<b>Teratogens/Teratogenic</b>	Drugs that cause birth defects – these can be prescribed drugs with unintended side effects or the misuse of recreational drugs, including alcohol.
<b>Ultra-Rare Disease</b>	A disease is generally considered to be ultra-rare if it affects less than 1 in 100,000.

## ANNEX 2:

# The National Rare Diseases Office

**NRDO**  
National Rare  
Diseases Office



The Rare Diseases Office provides reliable information about rare diseases in Ireland by phone and email. More information is available on our website.

**Telephone:** (01) 854 5065  
Monday to Thursday 9.30am - 1.30pm

**Email:** [rare.diseases@mater.ie](mailto:rare.diseases@mater.ie)

**Website:** [www.rarediseases.ie](http://www.rarediseases.ie)

The National Rare Diseases Office provides up-to-date and reliable information about rare diseases to patients, their families, researchers and healthcare professionals. The Health Service Executive established the Office in 2015, as recommended in the first Irish National Plan for Rare Diseases.

It is sometimes difficult to find accurate and useful information about rare diseases. We aim to bridge this information gap and to answer your rare disease-related questions on:

- Rare diseases
- Expert clinics in Ireland and across Europe
- Patient support groups
- Research projects and clinical trials
- European Reference Networks and cross-border care
- Social care supports

More information is available on our website [www.rarediseases.ie](http://www.rarediseases.ie). Here you can find how to access support services and cross-border care as well as educational materials.

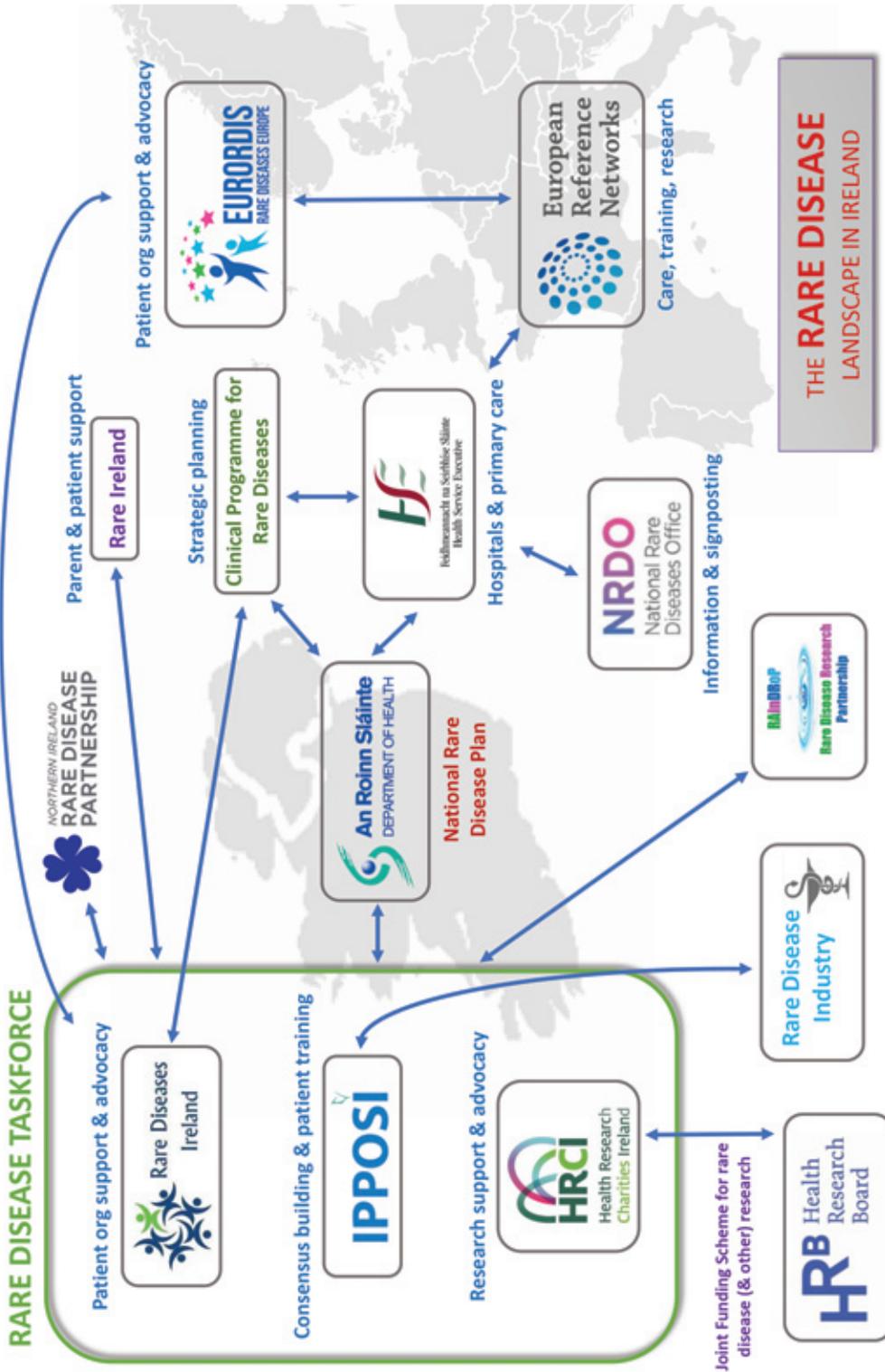
Our office staff are healthcare professionals with experience working with people with rare diseases. We do not operate a clinic and cannot diagnose, treat or coordinate patient care. We will do our best to help you get the information you need.

**orphanet**

We also collect information about Irish rare disease resources for Orphanet ([www.orpha.net](http://www.orpha.net)). Orphanet is a reliable international database of rare disease resources. Orphanet is an excellent starting point for disease summaries, guidelines and links to Irish expert clinics and patient organisations.

A display of rare disease information leaflets can be found at the Eccles Street entrance to the Mater Hospital. All the information leaflets are from charity-registered patient organisations.

# ANNEX 3: The Rare Disease Policy Landscape: A Visual Overview



# ANNEX 4:

## Further Examples of Rare Diseases<sup>59</sup>

RARE DISEASE	DESCRIPTION
1. <b>Duchenne Muscular Dystrophy (DMD)</b>	Rare genetic disease. Duchenne Muscular Dystrophy (DMD) is a progressive and disabling neuromuscular condition that primarily affects boys. Muscle weakness can eventually lead to acute respiratory failure. Early diagnosis means early access to treatment, improved outcomes, and possible access to clinical trials for new and innovative medications.
2. <b>Fabry Disease (FD)</b>	Rare genetic disease. Causes a build-up of a particular fat, globotriaosylceramide in the body's cells. Symptoms include episodes of pain, clusters of spots on the skin, problems with the gastrointestinal system and later life-threatening complications such as kidney damage, heart attack and stroke.
3. <b>Spinal Muscular Atrophy (SMA)</b>	Group of Rare genetic diseases. SMA (Spinal Muscular Atrophy) is a group of neuromuscular disorders that varies in severity and onset but primarily involves the wasting (atrophy) of muscles involved in walking, sitting, head control and in some instances breathing and swallowing.
4. <b>Friedrich's Ataxia (FA)*</b>	Rare genetic disease. Causes progressive nervous system damage and movement problems. It usually begins in childhood and leads to impaired muscle coordination (ataxia) that worsens over time. This damage results in awkward, unsteady movements and impaired sensory functions. It may also cause problems in the heart and spine.
5. <b>Ehler-Danlos Syndrome Type 3 (EDS)*</b>	Group of rare genetic diseases. EDS is a connective tissue condition characterised by skin extensibility, joint hypermobility and tissue fragility. There are six different types of EDS and they are classified according to signs and symptoms. EDS is caused by changes in genes that produce a protein called collagen, which is the main building block of the body and provides strength and support in ligaments, tendons and organs.
6. <b>Retinitis Pigmentosa (RP)*</b>	Group of rare genetic diseases. RP affects the retina in the eye causing cells to die and progressive vision loss. As the condition progresses individuals develop tunnel vision (loss of peripheral vision) and eventual loss of central vision.

\*if featured in the 2013 edition

59. With apologies to those disease that are omitted. As there are 6-8000 rare diseases in Ireland only a few can be mentioned in this Easyguide. Further on-line editions will seek to broaden the number included.

RARE DISEASE	DESCRIPTION
7. <b>Rett Syndrome*</b>	Group of rare genetic diseases. Rett syndrome is a brain disorder that occurs almost exclusively in girls. After birth, girls with classic Rett syndrome have 6 to 18 months of apparently normal development before developing severe problems with language and communication, learning, coordination, and other brain functions.
8. <b>Scleroderma*</b>	Group of rare autoimmune disorders. Varies from person to person but may involve changes in the skin, blood vessels, muscles and internal organs and can include localized or systemic variations. No known cause.
9. <b>Miller-Dieker Syndrome*</b>	Rare genetic disease. Condition characterized by pattern of abnormal brain development causes severe intellectual disability, developmental delay seizures and feeding difficulties with children rarely surviving into adulthood.
10. <b>Merosin Negative Congenital Muscular Dystrophy*</b>	Group of rare neuromuscular disorders. Associated with muscle weakness and wasting, feeding disorders and seizures may also occur with many children not progressing to adolescence.
11. <b>Thyroid Cancer*</b>	Group of rare cancers. There are four main types of thyroid cancer. These are papillary, follicular, medullary, and anaplastic. Papillary is the most common type. The four types differ in how aggressive they are. Thyroid cancer that is found at an early stage can often be treated successfully.
12. <b>Lyme Disease</b>	Rare bacterial infection. Lyme disease is a bacterial infection that is spread to human by infected ticks. It can cause a high temperature, muscle pain, joint pain and swelling, neurological symptoms, such as temporary paralysis of the facial muscles. The actual incidence in Ireland is unknown but hospital referrals appear to be increasing and it may well be no longer a rare disease.
13. <b>Sickle Cell Disease (SCD)</b>	Rare genetic disease in Ireland but one of the most common worldwide. Unpredictable with crises in severity and individuals can present as acute or chronic. Symptoms include a disorder in which red blood cells are destroyed faster than made and 'stuck' sickle cells slow or even totally block blood flow. Some parts of the body do not get the oxygen they need as a consequence. That can cause intense pain that lasts anywhere from a few hours to a few weeks.
14. <b>Fetal Valproate Syndrome (FVS)</b>	Rare disease caused by side effects of a prescribed drug therapy. FVS may occur if a developing baby is exposed to valproic acid during pregnancy. Valproic acid, also known as valproate, is a medication that is often used to treat epilepsy, bipolar disorder, and migraines. Epilim is the most commonly prescribed drug with valproate in Ireland. High risk of a wide range of physical and developmental defects if pregnant mothers have been exposed to valproate.
15. <b>Motor Neuron Disease (MND)</b>	A group of rare diseases. MND are a group of progressive neurological disorders that destroy motor neurons, the cells that control essential voluntary muscle activity such as speaking, walking, breathing, and swallowing. Over time, the ability to control voluntary movement can be lost.

# ANNEX 5: Patient Advocacy Organisations and Networks

## 5.1 Organisations



### **22q11 Ireland**

Carmichael House  
North Brunswick Street  
Dublin 7  
t: 087 7412856  
e: tech.22q11ireland@gmail.com  
w: www.22q11ireland.org



### **Alpha-1 Foundation Ireland**

RCSI Education & Research Centre  
Beaumont Hospital  
Dublin 9  
t: 01 809 3871  
e: alpha1@rcsi.ie  
w: www.alpha1.ie



### **Ataxia Foundation Ireland**

Gorteen  
Inch  
Gorey  
Co. Wexford  
t. 087 361 6616  
e. info@afi.ie  
w. https://afi.ie/



**BeeforBatters.org**

### **Bee for Batters**

Castledrum  
Castlemaine  
Co. Kerry  
t: 083 0044 444  
e: buzz@beeforbatters.org  
w: www.beeforbatters.org



**Cavernoma Ireland**

e: [cavernomaireland@gmail.com](mailto:cavernomaireland@gmail.com)  
f: [www.facebook.com/CavernomalrelandSupport/](https://www.facebook.com/CavernomalrelandSupport/)  
w: [www.cavernoma.org.uk/](http://www.cavernoma.org.uk/)



**Cystic Fibrosis Ireland**

(formerly the Cystic Fibrosis Association of Ireland)  
24 Lower Rathmines Road  
Dublin 6  
t: 01 496 2433  
e: [info@cfireland.ie](mailto:info@cfireland.ie)  
w: [www.cfireland.ie](http://www.cfireland.ie)



**Cystinosis Foundation Ireland**

1 Terenure Place  
Terenure  
Dublin 6W  
e: [mail@cystinosis.ie](mailto:mail@cystinosis.ie)  
w: [www.cystinosis.ie](http://www.cystinosis.ie)



**DEBRA Ireland**

La Touche House  
1 Grove Road  
Rathmines  
Dublin 6  
t: 01 412 6924  
e: [info@debraireland.org](mailto:info@debraireland.org)  
w: [www.debraireland.org](http://www.debraireland.org)



**Fabry Ireland**

w: [www.fabryireland.ie](http://www.fabryireland.ie)



**Fighting Blindness**

3rd Floor  
7 Ely Place  
Dublin 2  
DO2 TW98  
t: 01 678 9004  
e: [info@fightingblindness.ie](mailto:info@fightingblindness.ie)  
w: [www.fightingblindness.ie](http://www.fightingblindness.ie)



**HHT Ireland**

17 Drynam Court. Swords. Co Dublin  
 e: [dara@hhtireland.org](mailto:dara@hhtireland.org)  
 w: [www.hhtireland.org](http://www.hhtireland.org)



Huntington's Disease  
 Association of Ireland

**Huntington's Disease Association of Ireland**

Carmichael Centre  
 North Brunswick Street  
 Dublin 7  
 D07 RHA8  
 t: 01 872 1303  
 e: [hdai@indigo.ie](mailto:hdai@indigo.ie)  
 w: [www.huntingtons.ie](http://www.huntingtons.ie)



**iCAN Irish Children's Arthritis Network**

Ballydavid  
 Bansha  
 Co Tipperary  
 t: 086 828 9817  
 e: [icanireland@gmail.com](mailto:icanireland@gmail.com)  
[www.icanireland.ie](http://www.icanireland.ie)



**Irish Cancer Society**

43/45 Northumberland Road  
 Dublin 4  
 t: 01 2310 500  
 e: [reception@irishcancer.ie](mailto:reception@irishcancer.ie)  
 w: [www.cancer.ie](http://www.cancer.ie)



Irish Lung Fibrosis Association

**Irish Lung Fibrosis Association**

ILFA, PO Box 10456, Blackrock, Co. Dublin.  
 t: 086 871 5264  
 e: [info@ilfa.ie](mailto:info@ilfa.ie)  
 w: [www.ilfa.ie](http://www.ilfa.ie)



**Multiple System Atrophy Trust**

51 St Olav's Court  
 Lower Road  
 London, SE16 2XB  
 t: 0333 323 4591  
 e: [support@msatrust.org.uk](mailto:support@msatrust.org.uk)  
 w: [www.msatrust.org.uk](http://www.msatrust.org.uk)



**Muscular Dystrophy Ireland**

75 Lucan Road  
Chapelizod  
Dublin 20  
t: 01 623 6414 or 623 6415  
e: info@mdi.ie  
w: www.mdi.ie



**MSD Action Foundation**

Grattan Lodge  
Balgriffin  
Dublin 13  
e: info@msdactionfoundation.org  
e: info@savingdylan.com  
w: www.savingdylan.com



**Raynaud's & Scleroderma Ireland**

Paradigm House  
Dundrum Office Park  
Dublin 14  
t: 0818 363 999  
f: 01 215 7945  
e: info@irishraynauds.com  
w: www.irishraynauds.com



**Sickle Cell and Thalassemia Ireland**

19 Belvedere place  
Dublin 1  
T: 0870656807  
W: www.sicklecellireland.ie



**SMA Ireland**

e: info@smaireland.com  
w: www.smaireland.com



**SWAN Ireland**  
SYNDROMES WITHOUT A NAME

**SWAN Ireland - Syndromes Without a Name**

e: syndromeswithoutanameireland@gmail.com  
f: www.facebook.com/swan.ireland.77

## 5.2 Networks in Ireland, North and South



**HRCI** - Health Research Charities Ireland, previously known as the Medical Research Charities Group (MRCG), is the national umbrella organisation of charities active in medical and health research. Through support and advocacy, we represent the joint interests of our 40 member organisations, working to improve health and prevent illness through research. HRCI also hosts and supports the Irish Health Research Forum (IHRF), bringing together all stakeholders to improve health research in Ireland.

### **HRCI Contact Details:**

Digital Office Centre  
12 Camden Row, Dublin 8,  
D08 R9CN  
t: 01 479 3234  
e: [info@hrci.ie](mailto:info@hrci.ie)  
w: [www.hrci.ie](http://www.hrci.ie)



**IPPOSI** – The Irish Platform for Patient Organisations, Science and Industry is a patient-led organisation that works with patients, government, industry, science and academia to put patients at the heart of health policy and innovation. IPPOSI is funded as a public-private partnership that works to ensure early, equitable access to Health Innovation for improved patient outcomes.

### **IPPOSI Contact Details:**

Irish Platform for Patients' Organisations, Science & Industry  
77 Camden Street, Dublin 2,  
D02XE80  
t: Tel: 01 556 8269  
e: [info@ipposi.ie](mailto:info@ipposi.ie)  
w: [www.ipposi.ie](http://www.ipposi.ie)



**RDI** – Rare Diseases Ireland is the national alliance for voluntary patient-led groups representing people affected by or at risk of developing genetic or other rare diseases in Ireland. RDI works to advocate for the rare disease community, including:

- provide a voice for member organisations on issues of common concern;
- build awareness of rare diseases;
- promote development of genetic and health/social care services;
- facilitate dialogue and exchange of information;
- liaise with national/international experts; and,
- collaborate with national/international stakeholders on policy and regulation.

**RDI Contact Details:**

Digital Office Centre  
12 Camden Row, Dublin 8  
D08 R9CN  
t: 01 4793234  
e: [advocacy@rdi.ie](mailto:advocacy@rdi.ie)  
w: [www.rdi.ie](http://www.rdi.ie)



**NIRDP** – See Section 4.3 in this Easyguide for further details.

**NIRDP Contact Details:**

2 William Street  
Newtownards  
BT23 4AH  
County Down  
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