



An Easy Guide to
Rare Diseases
in Ireland



A Resource for the Media
and the General Public

Introduction

This 'Easy Guide' has been produced to increase awareness about the challenges of living with a rare disease and to highlight the priorities that need to be included in the forthcoming National Plan for Rare Diseases (NPRD) in Ireland.

The aims of this Easy Guide are to:

- Explain what a rare disease is
- Highlight the challenge of living with a rare disease in Ireland through patient profiles
- Identify priorities that should be included in the National Plan for Rare Diseases in Ireland (to be completed by the Irish Government by 2013)
- Contribute to the development of a National Clinical Programme on rare diseases in Ireland

The Easy Guide has been structured as follows:

Section 1: An Overview of Rare Diseases in Ireland

Section 2: Living with a Rare Disease in Ireland

Section 3: The National Plan for Rare Diseases (NPRD): Priorities of Patient Groups

Section 4: Glossary of Key Terms

Section 5: Useful Resources

Section 6: Contact Details of Patient Networks and Patient Advocacy Organisations

For a brief introduction to rare diseases visit: www.rarediseaseday.org/solidarity

About those patient groups involved in this Easy Guide

This Easy Guide is published by the Task Force on Rare Diseases which brings together key patient networks and advocacy organisations concerned with rare diseases in Ireland, including:

Patient Networks

- GRDO (Genetic and Rare Disorders Organisation)
- IPPOSI (The Irish Platform for Patients' Organisations, Science & Industry)
- MRCG (Medical Research Charities Group)

Patient Advocacy Organisations

- 22q11.2 Deletion Syndrome
- Alpha One Foundation
- Ataxia Ireland
- Bee for Battens
- Cystic Fibrosis Ireland
- Cystinosis Foundation Ireland
- DEBRA Ireland
- Fabry Ireland
- Fighting Blindness
- Huntington's Disease Association of Ireland
- Irish Cancer Society
- Muscular Dystrophy Ireland
- Raynaud's & Scleroderma Ireland

Section 1: An Overview of Rare Diseases in Ireland

What is a Rare Disease?

Worldwide it is estimated that there are over 6,000 identified rare diseases. Many are ultra-rare while others are more common or more widely known, mainly because of the work of patient organisations and the media highlighting the challenges of patients and their families and the work of clinicians in treating rare diseases.

In the European Union, a disease is defined as rare when it affects fewer than 1 in 2,000 people. In Ireland it is estimated that 6-8% of the population has a rare disease (around 280-370,000 people). In other words, up to 1 person in 12 in Ireland may have a rare disease at some stage in their life.

The European Commission defines rare diseases as 'life-threatening or chronically debilitating diseases which are of such low prevalence that special combined efforts are needed to address them'. As stated, 'low prevalence' is defined as meaning fewer than 1 in 2,000 people.

Diseases that are statistically rare, but not also life-threatening, chronically debilitating, or inadequately treated, are excluded from the EU definition.

70-80% of rare diseases are genetic, and are present throughout the person's entire life, even if symptoms do not immediately appear. Many rare diseases appear early in life, and it has been estimated that 30% of children with rare diseases will die before reaching their fifth birthday.

Challenges for patients with rare diseases and their families in Ireland

There are many challenges for patients living with a rare disease in Ireland. These include for example:

- Medical practitioners may fail to identify symptoms that they rarely come across, resulting in significant potential for patients to experience lengthy delays in diagnosis and often experiencing misdiagnosis
- Even with a correct diagnosis, patients and their carers may have considerable difficulty sourcing appropriate information about the condition and identifying relevant/experienced specialists (should they exist in Ireland)
- The lack of a transparent process to evaluate, approve and commission innovative therapies (medicines and technologies for rare diseases) results in unnecessary and prolonged delays for patients trying to access available treatments
- Progressive disorders require pre-planning, fast track support including access to existing services, education and counselling services

Section 2: Living with a Rare Disease in Ireland

This section provides profiles of 15 people living with a rare disease in Ireland:

Profile 1	Lauren Shaw Friedreich's Ataxia
Profile 2	Jamie O'Brien Ehler-Danlos Syndrome Type 3
Profile 3	Josephine McGuirk Alpha-1 Antitrypsin Deficiency
Profile 4	Christy Murtagh Retinitis Pigmentosa
Profile 5	Clare Louise Creedon Ataxia Oculomotor Apraxia-1
Profile 6	Gerry Walker Cystic Fibrosis
Profile 7	Ciara Huntington's Disease
Profile 8	Emma Fogarty Epidermolysis Bullosa
Profile 9	Sarah Kehoe Rett Syndrome
Profile 10	Áine Lawlor 22q11.2 Deletion Syndrome
Profile 11	Meadhbh Cleary Scleroderma
Profile 12	Áine Hill Miller-Dieker syndrome
Profile 13	Fatiha & Manal Akka Merosin Negative Congenital Muscular Dystrophy
Profile 14	Kerry-Ann McLaughlin Thyroid Cancer
Profile 15	Liam & Saoirse Heffernan Batten disease

Profile 1 | Lauren Shaw | Friedreich's Ataxia

Lauren Shaw from Dublin is nine years old. When she was two, she was very unsteady on her feet. She couldn't feed herself, and was not reaching the milestones expected for her age. Staff at the local playschool she attended encouraged her mum, Sue, to get Lauren checked out. After physiotherapy and other treatments for a variety of suspected conditions, Lauren seemed to be getting worse instead of better. Finally, at the age of four, she attended a neurologist who diagnosed Friedreich's Ataxia.



“People tend to see the condition and not the person”

There was very little information about the condition available, Sue found, but she was given contact details for the Friedreich's Ataxia Society in Ireland (now Ataxia Ireland), who sent someone to visit the family. Lauren's case is unusual in that onset of the disease happened at a very young age. From then, Sue says, it has just been a case of getting on with things. Now Lauren uses a wheelchair when she's outside the home as her balance and co-ordination are very poor. “It's as if she wants to go one way and her body just goes the other,” according to Sue. But like any other nine year old, she likes to assert her independence when she can, and tries her best to stay out of the chair at home.

Lauren's school has arranged for a full-time assistant in the classroom to ensure her basic needs are met. As she cannot write or do most of the homework that the class gets, she is waiting for a new computer to help with this. The economic situation in Ireland at the moment means that the waiting time for any kind of assistance can be long. But the biggest challenge faced by Lauren and those like her is social acceptance.

“People tend to see the condition and not the person,” she says. Lauren is bright and bubbly, interested in all the things that her classmates like. Although she has physical challenges, in every other way she is like any other girl her age. “Lauren was thrilled to be asked to take part in the video for Rare Disease Day last year,” says Sue. “She thinks she's famous now!” Sue is only too happy to help spread the message about rare conditions like this. “I hope that it helps people everywhere to see that you can have a condition but you're still a normal person.”

About Friedreich's Ataxia

Friedreich's Ataxia is the most common type of hereditary ataxia and it is inherited in an autosomal recessive pattern. It is a slow progressive ataxia with the age of onset in the first and second decades of life – usually below age 25. Some individuals with Friedreich's Ataxia may not develop symptoms until very late in life. Weakness of the extremities and loss of sensation become more prominent as the disease progresses. Skeletal deformities, including scoliosis, are also common. Friedreich's Ataxia may also be associated with heart problems, including irregular heart beat and enlargement of the heart.

Most individuals with Friedreich's Ataxia have repeat expansions in both of their copies of the Friedreich's Ataxia gene. Some rare individuals may have a combination of a repeat expansion in one gene and a sequence change in their other copy of the gene. Carrier testing is available to family members. About 1-2% of the general population carries an abnormal gene for Friedreich's Ataxia, but these individuals do not have symptoms of Friedreich's Ataxia. It is only when both parents are carriers that the offspring will have a 25% chance of having the disease.

Profile 2 | Jamie O'Brien | Ehler-Danlos Syndrome Type 3

For a long time, Jamie O'Brien's ambition has been to work in music production and website design. He lives in the North East of Ireland. He's 22 years old and last year he started a course in Creative Media at Dundalk Institute of Technology. A few months into the course, he realised that his condition would make it impossible for him to continue and so he had to take a break. Jamie has Ehler-Danlos Syndrome Type 3 (the hypermobility type).



Every day, several times a day, Jamie's joints dislocate. The slightest movement or exertion triggers dislocation – lifting something very small puts out his wrists, climbing a stairs will dislocate his knees or ankles. His symptoms first appeared when he was about eight years old as he was starting to get more physical and play sports. He could not write more than one sentence and his teachers at school thought he was just being lazy. Back then, the biggest challenge was getting people to believe that there was something wrong. "I didn't even believe it myself," he says. He did not understand then that his fingers were actually dislocating.

"The biggest challenge was getting people to believe that there was something wrong"

Some things got easier after he was diagnosed at 16. He found it less difficult to make friends once he could explain his condition. Disability services became available to help with certain aspects of life. Now his greatest challenge is his left shoulder. He has learned to deal with most of his joints, and to cope with constant pain. His attitude is "snap it in, keep smiling, keep walking, you have to get used to it." But when his left shoulder goes, he has no choice but to attend the hospital Emergency Department. And if the doctors are not familiar with him or his condition, it takes time to get the right treatment – more time than he can afford to be away from a busy college schedule.

He hopes to be able to study again though, and plans to return to college as soon as he can get the pain and dislocations under enough control to allow him 2-3 weeks between hospital visits. In the meantime, he has defied his diagnosis and come up with an ingenious solution to help him work as a DJ. Using a collection of splints and braces, some light-up wire and a lot of creative flair, he has invented a special suit that holds his joints in place and allows him to make the music that he loves. "It takes me 45 minutes to put it on but once I'm in, I'm safe. I'm getting gigs in Limerick, Cork and Belfast. It's something new and people really like it. And it shows you that, no matter what people say, I can do something I really want to do."

In the long term, Jamie would like more people, including doctors, to understand the issues around rare diseases. "There's no system for people with a rare disease," he says. But he hopes that a National Plan for Rare Diseases will go some way towards building awareness and improving the situation of all those affected. For now, as there is no specialist service for his condition in Ireland, he is fighting for the funding to allow him to access the appropriate specialist physiotherapy he needs at a centre in the UK.

About Ehler-Danlos Syndrome Type 3

Ehlers-Danlos Syndrome (EDS) is a genetic 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.

The most common sign of this condition is an unusually large range of joint movement, called hypermobility. Both large and small joints are unstable, and certain joints (such as the shoulder, knee and jaw) tend to dislocate frequently. Chronic joint and limb pain often begins early in life. People with this condition may have skin that is soft, velvety, or stretchy; however, skin symptoms vary among people.

The Hypermobility Type or EDS Type 3 is characterised by loose joints and chronic joint pain. This is the most common form of EDS. It is most often inherited in an autosomal dominant pattern, which means one copy of the altered gene is sufficient to cause the disorder.

Although it is difficult to estimate the overall occurrence of EDS, the combined incidence of all types of EDS may be about 1 in 5,000 individuals worldwide; however, research is showing that it may be more common.

Profile 3 | Josephine McGuirk | Alpha-1 Antitrypsin Deficiency

Josephine McGuirk has Alpha-1 Antitrypsin Deficiency, a genetic disease that affects the lungs and liver, and can cause life-threatening conditions. She was tested for the condition at age 53, and only because her brother had been diagnosed first. At that stage she had no symptoms but her brother sadly died. Two further siblings also tested positive and, while neither of them has yet developed any signs of the disease, Josephine, a long-term smoker, gradually started to feel the effects. At 65, she now has emphysema and needs oxygen for a couple of hours each day.



“Alpha-1 may not be rare but it is rarely diagnosed...so to be aware makes all the difference”

She has had to slow down her life a lot, and her physical activity is limited as she becomes breathless very easily. “The hardest thing is not being able to dance,” she jokes. But two things have dramatically improved her quality of life in recent years: moving to a single storey house at the seaside north of Dublin; and taking part in a drug trial. After two years of not knowing whether she was getting the protein replacement or a placebo, Josephine now knows she is getting Zamira and really feels the benefits. She would like to be sure, however, that the drug will be licenced and that the benefits will continue for her in the long-term, and not just for the duration of the trial.

What keeps her going is the support around her. She cannot speak highly enough of her medical and research team in Dublin. Josephine is keen to share her story, hoping that it will help other people. “Alpha-1 may not be rare but it is rarely diagnosed,” she continues, “so to be aware makes all the difference.” A simple finger prick blood test is all that is required and she wishes that she had known earlier about Alpha-1 so that she could have avoided smoking.

Josephine works with the Alpha-One Foundation Ireland, using her own experience and her positive outlook to benefit others with the disease and raise awareness among the general public.

About Alpha-1 Antitrypsin Deficiency

Alpha-1 antitrypsin deficiency (AATD) is a rare genetic disorder where the body does not produce enough of the antiprotease alpha-1 antitrypsin (AAT). The main site of action for AAT are the lungs where it provides a protective screen against cigarette smoke and bacterial infection. This lack of AAT in the lungs eventually leads to chronic obstructive pulmonary disease (COPD), and in some cases liver disease. Globally, COPD is a growing cause of mortality and will become the 3rd biggest killer by 2020 (World Health Organisation). The most common AAT gene variant associated with disease is the Z variant and the most severe form of the condition occurs when two copies of the Z variant (ZZ individuals) are inherited. Over 3,000 individuals on the island of Ireland suffer from the severe form of AATD, and 1 in 25 individuals are carriers for the condition.

Unfortunately, AATD is under-diagnosed and prolonged delays in diagnosis are common. World Health Organisation (WHO) guidelines advocate targeted screening of COPD, poorly-controlled asthma, and liver disease patients, as well as first degree relatives of known AATD individuals. In 2004, a national AATD targeted detection programme commenced with funding from the Department of Health and Children. The many advantages of an early diagnosis of AATD include increased lung and liver surveillance, family member testing, smoking cessation opportunities, as well as management of occupational and environmental exposures.

Profile 4 | Christy Murtagh | Retinitis Pigmentosa

As a child, Christy Murtagh was prescribed glasses. He never wore them for long though. Even then he realised that they weren't helping. At 19, while living in Hawaii, he found he was frequently driving in dark tunnels and the problem with his vision had become too bad to ignore. Retinitis Pigmentosa (RP) was suspected by doctors there, but it took over two years before Christy got an official diagnosis in Boston. He was told then that he would be blind by age 50. At 46, he's confident that this will not be the case, and dreams of some day visiting New Zealand, where he has heard that the light is great.



“Don't give up – keep going.”

RP is a hereditary eye disorder. It affects the retina, the light sensitive tissue at the back of the eye, in which the first stages of seeing take place. In RP, sight loss is gradual but progressive.

Christy used to work in the building trade but his condition made it impossible for him to continue. Now he is studying full-time to be a counsellor and looks on RP as a blessing. “It has made me a better person,” he says. In his new career, he feels that his own difficult experiences will help him to understand others better. His message to others affected is “Don't give up – keep going. Tap into what you have.”

Small things like his talking laptop computer enable Christy to pursue a normal life. He has to move more slowly now, and for safety reasons he will rely increasingly on a cane for walking. As he gets older, he struggles with the idea of being labelled as a blind person, but takes comfort from the fact that support is out there should he need it. Charities like Fighting Blindness run support groups for those affected by RP, and also for their partners and spouses.

When he was asked to take part in the Rare Disease Day video, Christy did not hesitate. He wants the world to know that those with rare diseases are still normal human beings. He hopes that by telling his story he will help to make people more aware – not just of RP but of the issues that face all those with rare diseases. “Society is disabling us,” he says. “I could jog anywhere if there weren't so many obstacles out there.”

About Retinitis Pigmentosa

Retinitis pigmentosa (RP) is the name given to a group of hereditary disorders causing damage to the specialised cells in the retina known as the rods and cones. Rod cells handle vision in low light and in RP, these cells are usually affected first; therefore, night blindness and difficulty getting around in low lighting is one of the earliest symptoms. People with RP experience a gradual decline in their vision, as the disease is characterised by progressive damage and death of these light sensitive cells.

In Ireland, RP is the third major cause of blind registration, accounting for 7% of those on the register. If a family member is diagnosed with RP, it is strongly advised that other members of the family be examined by a specialist to detect any retinal degeneration. As of yet, there are no cures for RP, but retinal research is entering an exciting era where safe and effective therapies and treatments are realistic goals.

Profile 5 | Clare Louise Creedon | Ataxia Oculomotor Apraxia-1

Clare Louise Creedon uses a wheelchair all the time now. It really annoys her that there are so few wheelchair accessible toilets when she goes out in Dublin. On a trip to Orlando, Florida, she experienced how much easier life can be when disabled access is universal. Simple things like dips in the pavement can make all the difference to Clare Louise.



She was diagnosed with Friedreich's Ataxia when she was only 7 years old. In fact she has since discovered that hers is Ataxia Oculomotor Apraxia-1 (AOA1). Although she had no major symptoms as a small child, she had poor balance and her parents suspected something as her older brother had already had a diagnosis. For a long time Clare Louise got by holding onto walls to stay upright, then using a walking frame. In her 20s she was forced to start using a wheelchair some of the time for safety reasons and now, at 40, she needs it all the time.

"I do the same things as everyone else...I just have a lot more obstacles to overcome."

Her boyfriend, Ollie, is also a wheelchair user, and has helped her to accept a lot about her condition. "He is very supportive," she says, and that helps her to get on with things. "I do the same things as everyone else: I go to the gym, I go shopping, I get the bus to visit friends...I just have a lot more obstacles to overcome." Clare Louise would love to see attitudes to disability change. She hopes that initiatives like Rare Disease Day will help to make people more aware of Ataxia and other conditions like it. "My parents set up the Friedreich's Ataxia Society in Ireland (now Ataxia Ireland) when I was diagnosed. They had no-one to talk to about it," she says. But she thinks things are starting to get better on that front. She is glad that there is some help there for families now but what she finds hard about being involved with a support group is frequently meeting younger people with more severe forms of Ataxia than herself.

About Ataxia Oculomotor Apraxia-1 (AOA1)

Ataxia with oculomotor apraxia – recessive ataxia with oculomotor (muscles of eye movement) apraxia (difficulty in executing movement). AOA1 is often associated with other types of movement problems (chorea), neuropathy and, rarely, change in learning and memory. Symptoms of AOA1 usually develop early in childhood. Individuals with AOA1 may have high levels cholesterol and low levels of a substance called "albumin" in their blood. AOA1 is an autosomal recessive form of ataxia caused by sequence changes in a gene called "APT_X". In order to confirm a diagnosis of AOA1, sequence changes must be found in both copies of the gene. Carrier testing for family members may be available if sequence changes are identified in the individual with ataxia.

Profile 6 | Gerry Walker | Cystic Fibrosis

Gerry began hiking in 1985 and there began his love and obsession with mountains. "I was born with Cystic Fibrosis, so I was never going to break speed or endurance records."

Cystic fibrosis (CF) is an inherited chronic disease that primarily affects the lungs and digestive system. A defective gene and its protein product cause the body to produce unusually thick, sticky mucus that clogs the lungs and leads to life-threatening lung infections; and obstructs the pancreas and stops natural enzymes from helping the body break down and absorb food.



"I was capable of walking just 200 metres in a 6 minute test and my lung function was down to 25%, it was like living at over 8,000 metres"

Although he loved hiking, it brought on coughing fits and vomiting from the over exertion. He recalls that "things were beginning to crumble." Over the next 10 years, Gerry spent more and more time in hospital or on home intravenous antibiotics. CF was taking over his days and nights. In May 2003, after a bad winter he was prescribed oxygen, first to sleep, then to just walk.

In November 2003, Gerry was assessed for a double lung transplant at the Freeman Hospital in Newcastle. The verdict was "suitable for transplant but still too well". Now began the wait. Knowing the transplant might never happen, Gerry was determined to continue visiting the hills at home and abroad in between the infections. He recalls that "by October 2007, I was capable of walking just 200 metres in a 6 minute test and my lung function was down to 25%, it was like living at over 8,000 metres."

The Freeman team decided it was time to go on the active transplant list. After 10 months waiting for that life changing phone call, it finally came while he was on the shore of Lake Windermere on his 19th wedding anniversary. Margaret and Gerry drove to the Freeman. Shortly after midnight they were told the lungs were suitable and the transplant would go ahead. "I knew if I got on the operating table, the stamina built up on hills over the years would pay off and I would get through the ordeal," he recalled. Twenty four hours later Gerry was off the respirator, able to breathe freely again.

Since the transplant Gerry feels that his life has been transformed – "I'm back walking regularly. I have walked in the Dolomites, the Gredos, the Rockies and recently completed the Glover walk for the first time in over 15 years!"

Gerry feels that he wouldn't have been able to get through everything without his "best friend, supporter and wife Margaret Barron. Truth be known, the last few years have placed a heavy burden on her. Margaret has ridden the roller coaster to hell and back with me over the last few years – a true friendship."

About Cystic Fibrosis

Cystic fibrosis (CF) is an inherited chronic disease that primarily affects the lungs and digestive system of about 1,200 children and adults in Ireland (70,000 worldwide). A person with CF has inherited two altered genes, one from each parent, which together cause CF. Approximately 1 in 19 people in Ireland carry the altered gene that causes CF; where both parents carry an altered gene, their children have a 1 in 4 chance of having CF.

While CF is classified as a rare disease world-wide, it is 'borderline' as to whether CF should be classified as a rare disease in Ireland. Ireland has the highest incidence of CF in the world, three times the rate of the United States and the rest of the European Union. CF is the most common inherited disease in Ireland.

All people with CF need access to care in specialised centres staffed by multi-disciplinary teams. A network of such centres has developed in Ireland over the past decade, though the standard of infrastructure can vary considerably between one centre and another. In recent years new and innovative therapies have been developed to treat people with CF. In January 2013 the Irish government approved the ground-breaking drug Kalydeco, the first to treat the underlying causes of CF for those with at least one copy of the G551D gene alteration, which accounts for some 120 of the CF population in Ireland and up to 3,000 people with CF world-wide.

In 2011, CF was added to the national new-born screening programme for the first time.

Profile 7 | Ciara | Huntington's Disease

Ciara is in her mid-twenties. Her Dad is 67 years old. In his late 40s/early 50s his mood and aspects of his personality changed and he had problems with balance and movements. He and his family spent 7 years not knowing what was causing these changes. Eventually he was referred to a neurologist and he was diagnosed with Huntington's Disease (HD).



Each child of a parent with the HD gene has a 50/50 chance of inheriting the condition. People at risk of HD need genetic counselling and psychological support. They face discrimination in getting mortgages and life insurance and many fear discrimination in employment and promotion.

"It sent a shockwave through my family as we found out we were all at risk"

"It sent a shockwave through my family as we found out we were all at risk. We knew so little about the disease and we all had a tough individual choice to make."

Ciara has four siblings. Ciara and three of her siblings decided to take a predictive test for HD. In 2012 Ciara found out that she carries the HD gene. Her three siblings also received an unfavourable predictive test result.

Ciara's dad is currently in a long term care facility appropriate to his needs. This was difficult to find and so he initially spent some time in a psychiatric hospital.

Physical and mental activity can improve quality of life and delay onset of symptoms. Ciara has completed a degree in social care and is now pursuing her career. She has a wide social support network and gets support from close friends and family. She also attends Huntington's Disease Association of Ireland support meetings. Initially Ciara found it difficult to absorb her predictive test result and those of her siblings but she is now ready to get on with her life.

About Huntington's Disease

HD is an inherited neurological disorder that causes brain cell degeneration. There are more than 500 people with HD in Ireland with over 2,500 people at risk. People affected can have physical, cognitive and psychiatric symptoms. Early symptoms might include slight involuntary movements and balance problems, changes to organisation, regulation and perception skills, depression and changes in mood and personality. In the later stages swallowing difficulties occur and involuntary movements increase. Decision making, reasoning, impulse and temper control, deteriorate significantly.

Profile 8 | Emma Fogarty | Epidermolysis Bullosa

My name is Emma Fogarty, I am 27 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 morphine twice a day to try and 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 cures for EB is happening worldwide and increasingly over the last few years there are more and more little glimmers of hope.

"My skin is as fragile and delicate as the wings of a butterfly and is just as easily damaged"

I am the 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 have such a well established organisation already in existence. 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 being done for people with other rare conditions because I can only imagine how hard it would be to fight a separate battle with the one you are already fighting against – a rare disease being so bloody hard!

About Epidermolysis Bullosa

Epidermolysis bullosa (EB) is a distressing and painful genetic condition causing the 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 up to several hours a day and the condition becomes increasingly debilitating and disfiguring over time. Adult patients with severe forms are also 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 multi-disciplinary "butterfly clinic" in Our Lady's Children's Hospital 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 9 | Sarah Kehoe | Rett Syndrome

Andrew and Julie Kehoe are parents to Sarah, age 15. They remember initially having concerns about Sarah when she was only 22 months old. Having developed apparently normally until then, Sarah began screaming for no obvious reason. Reluctant to accept that her behaviour was simply “the terrible twos,” her parents pushed to have her checked in Crumlin Hospital, where she remained under clinical assessment for about two years. During this time, Sarah’s motor and communications skills regressed significantly. Aged 4, she was diagnosed tentatively with autism and developmental delay. This, at least, allowed her parents to start applying for relevant therapies and supports for Sarah’s ongoing care and education.



“Her needs will only increase and the services just are not there”

Six months later, a blood test changed Sarah’s diagnosis to Rett Syndrome, a rare neurodevelopmental disorder. Today Sarah is totally dependent for all her needs. She has no formal means of communication, no hand function, can only barely walk a short distance with assistance, and has developed epilepsy. Her symptoms are typical of the condition, which affects almost exclusively girls, and becomes harder to manage as the child gets older. Many of those affected will, for example, eventually lose their swallow function and need to be tube fed. Those who can walk a little typically lose that ability as well, and require constant lifting. Most would require regular speech and language therapy, occupational therapy and physiotherapy.

Sarah attends a special school until mid-afternoon five days per week. Throughout the rest of the day and night, and during the school holidays, she has only her parents to see to her every need. Andrew says that obtaining support of any kind for a child like Sarah is “a constant battle, on top of the condition.” They get about six single nights of respite care throughout the year. Asked about the greatest challenge of this condition, Andrew says “When Sarah was younger, getting through the diagnostic process was harrowing. Now, our biggest worry is what will happen when we are gone and Sarah is left behind. Her needs will only increase and the services just are not there.”

After meeting other affected families at a conference eight years ago, Andrew and five other parents of girls with Rett Syndrome founded a support organisation, the Rett Syndrome Association of Ireland www.rettsyndrome.ie. The experience of having a child diagnosed with Rett Syndrome was, for all of us, “like a bereavement,” Andrew recalls. “We watched our apparently normal children regress to this state and we did not know what was happening. Families with a new diagnosis can now turn to the Rett Syndrome Association for information and support, as well as looking to the very hopeful advancements in Rett Syndrome research that are taking place internationally,” he continues. “It may be years before there is a cure but, in the meantime, Sarah herself keeps us going. We remain positive because she has brought a massive amount of joy into our lives.”

About Rett Syndrome

Rett Syndrome is a rare genetic neurodevelopmental disorder that primarily affects females (although there are some very rare cases of male diagnosis). It is caused by a gene alteration on the X Chromosome but is rarely inherited. The incidence is estimated at 1 in 12,000 female births (which translates to 3 potential diagnoses in Ireland annually). Although a rare disorder, Rett Syndrome is the most common cause of profound and multiple mental and physical disabilities in females.

Those affected are totally dependent for all of their needs for the duration of their lives and require around the clock care. Difficulties encountered include loss of communication skills, loss of hand function, general motor difficulties, epilepsy, curvature of the spine, breathing irregularities, feeding difficulties and gastrointestinal complications.

Profile 9 | Áine Lawlor | 22q11.2 Deletion Syndrome

Áine is 29 years of age. From the time she was very young she felt different to other children. She struggled with maths and spellings, and tired easily when exercising. She suffered with constipation all the time. In a mainstream school, 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.

Starting in St Michael's Special School aged 11 was a terrifying move for Áine, but it turned out to be the right one. "St Michael's was the best thing that ever happened to me," she recalls. There she started to enjoy school and was encouraged to try lots of things she had never tried before like running and cooking. She was a happy teenager, and is thankful to her parents for recognising the right environment for her, as well as to the school for helping her to develop into the bright young woman she has become.



"Everyone was ahead in work and I was always behind and nobody knew why"

It was not until she was 15 years old that Áine was finally diagnosed with 22q11.2 deletion syndrome. This chromosomal disorder, also known in various forms as VCFS or Di George syndrome, is not widely recognised but it has the potential to affect almost every system in the body and cause a wide range of health problems including varying degrees of: heart defects; palate differences; feeding and gastrointestinal difficulties; immune system deficits; growth delay; kidney problems; hearing loss; endocrine issues; developmental delay; behavioural, emotional and psychiatric differences.

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 was a keen Irish dancer for several years but, since developing osteoarthritis, has taken up the violin instead. These outlets keep her going. "It's not all that bad having 22q," she reports.

She is quick to acknowledge, however, 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 was one of the founding members of 22q11 Ireland, www.22q11ireland.org, a group which provides support – through education, research, outreach and advocacy – for individuals and families affected by the 22q11 deletion. In July 2013 the group will host the VCFS Educational Foundation 20th International Conference here in Ireland.

About 22q11.2 Deletion Syndrome

22q11.2 Deletion Syndrome is a disorder caused by the deletion of a small piece of chromosome 22. The condition affects an estimated 1 in 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 deletion syndrome often experience recurrent infections caused by problems with the immune system. Aside from the varied physical health issues, many children with 22q11.2 deletion syndrome 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 deletion syndrome 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. Currently, and unlike other countries including the UK, Ireland has no designated 22q clinic and no clinical nurse care co-ordinator. The 22q11 Ireland Group was set up in 2007 to support affected families.

Profile 11 | Meadhbh Cleary | Scleroderma

Meadhbh Cleary is a Religion and History teacher, currently living and working in Dublin city. When Meadhbh was in her late teens, she began to experience aches in her hands and wrists. She struggled to keep up with written schoolwork, but attributed her difficulty to the increasing pressure of the Leaving Certificate. Over the next few years, she noticed other changes to her hands – including swollen fingers and little cuts that took a month to heal properly – but she didn't make much of it. After all, she thought, hands are hands; it's not as though it's serious!



"It's the lack of stability that gets to me, she says. I just wish I could know what to expect."

Today, Meadhbh is 26 years old and knows that the pain she felt during her teens was the earliest sign of Scleroderma, a very serious autoimmune disease that primarily affects the skin and internal organs. The first obvious symptom – Raynaud's Phenomenon – was diagnosed in 2006 during her second year of university. Raynaud's is a circulatory condition triggered by cold temperatures and stress. It restricts blood flow to the extremities, including the fingers, toes, lips, ears and nose. For most people, Raynaud's Phenomenon is an isolated condition. In fact, statistics suggest that one-tenth of women in Ireland have some form of it. For this reason, Meadhbh was simply advised to wear gloves while outdoors and stay warm during the cold months.

That sounds easy, she thought, but it wasn't. Her first winter with Raynaud's Phenomenon was unbearably painful. Her hands became numb on a daily basis and the skin of her fingertips began to break down. It pushed her to find out more about the causes of the condition and she discovered that, in rare cases, Raynaud's is a symptom of a more dangerous disease. Four years and countless blood tests later, she was diagnosed with Diffuse Cutaneous Systemic Sclerosis, or 'Scleroderma'. Every case of Scleroderma is unique. The word means 'hard skin,' and describes how the disease causes the immune system to over-produce fibroblasts containing collagen in healthy areas of the body. The result is thick, hard and shiny skin. Meadhbh can no longer close her fists and still has trouble writing.

Due to the long wait for diagnosis, her college experience was tough. Without a known condition, she couldn't apply for special requirements during exams. By her fourth and final year, she received last-minute permission to use a computer, but the stress caused painful digital ulcers.

Today, Meadhbh takes medication to improve circulation in the extremities and prevent new ulcers. She also checks into hospital on an annual basis for rigorous investigations of her organs. Down the line, she knows she may have to move on to stronger medication, especially if her lungs and kidneys become damaged, as predicted. There is no cure for Scleroderma yet, but advances continue to be made in organ-specific treatments.

While her family have been continuously supportive, the shock of Meadhbh's diagnosis caused her to look for support amongst others with the disease. Six months after her Scleroderma was identified, she contacted Raynaud's & Scleroderma Ireland and started a support group for patients in her area, which she still runs today. She says that knowledge of her condition helps her to cope with it. With strangers, she often wonders how to explain her limitations. Scleroderma is not always visible, which makes it harder for people to empathise. During a meal out, for example, she has to avoid spicy foods, stay away from draughts and air conditioning, ask for water without ice and request a steak knife no matter what she orders. And that's before the meal even arrives!

In the long-term, Meadhbh worries about being able to afford to pay her health expenses, because she is already receiving help from her parents. As a creative person who needs her hands a lot at work, she is also concerned about sustaining irreparable damage to her fingers in the future, fearing that it may prevent her from doing what she loves. "It's the lack of stability that gets to me, she says. I just wish I could know what to expect."

About Scleroderma

The word 'Scleroderma' means hard skin. This is a chronic, autoimmune condition affecting connective tissue and blood vessels.

Scleroderma is a multi-system disease distinguished by overproduction of collagen, which is first deposited in the skin causing it to thicken and feel hard and tight. Someone described this as "feeling vacuum packed". Collagen may be deposited in the kidneys, lungs, heart, digestive system and muscles. Raynaud's is also usually present and may be the first symptom. Additional symptoms may include digital ulcers, dry eyes and mouth, acid reflux, indigestion and difficulty swallowing.

There are several forms; Diffuse Cutaneous Systemic Sclerosis, Limited Cutaneous Systemic Sclerosis, and localised scleroderma called Morphea; patches of dry, tight skin develop. Linear Morphea usually affects children. There is currently no cure for scleroderma but research is taking place. Early diagnosis and treatment with modern medicines can improve the outcome and slow the progress of the disease.

Profile 12 | Áine Hill | Scleroderma

Áine Hill is 6 years old. She was 10 months when she was diagnosed with Miller-Dieker syndrome, a condition characterised by a pattern of abnormal brain development known as lissencephaly.

Áine's mother Trudy knew that something was wrong from the beginning. While she clearly loved people and would smile a lot, Áine's eyes were turned and she had clicky hips. Her dad Ian spotted quickly that she would stare into space, holding her hands in fists. She wasn't sitting, rolling or clapping the way her sister had, or reaching any of her milestones. Trudy wasn't listened to when she tried to bring these things to the attention of medical professionals, both at the maternity hospital and with her local GP.



“...sometimes trying to figure out what is wrong with her is very hard due to her inability to communicate”

“Even in the family,” Trudy recalls, “we were in a sort of denial. When Áine got a bad dose of chicken pox, everyone said that was all that was affecting her...but at 9 months she was more like a newborn.” At that stage, a public health nurse visited the family's home and agreed that there was an issue and so Áine was eventually referred to Temple Street.

At the hospital, the ridge in Áine's forehead was recognised immediately as a characteristic sign of her condition. From the speed with which an EEG and then an MRI were organised, Trudy knew that “it was something catastrophic.” Within days epilepsy was confirmed. In little over a week, the family heard the rest of the diagnosis. “All of the information we received was very negative,” Trudy says. “There was nothing to give us any glimmer of hope.”

Now at six Áine is still like an 8-month-old, both physically and intellectually. She has kidney problems and a feeding tube, and often needs a lot of attention at night. She is very prone to chest infections too and sometimes trying to figure out what is wrong with her is very hard due to her inability to communicate.

Over the years, however, Áine has amazed her parents and learned to do much more than doctors ever expected. She is full of love and gets smiles everywhere she goes. She can now stand and walk a few steps if supported. She has a few words and shows signs of understanding much more. Her epilepsy had been quite well under control until last year but that is no longer the case.

“Keeping her strong and well is our greatest challenge,” her mum says. It takes time and a lot of mental energy. Now that Áine has started at St Vincent's Special School on the Navan Road there is some relief in the day for Trudy but, she admits, “it's not always easy to rest when you have a household to run.” She tries hard to keep things as normal as possible for Aoife, who is now ten years old, and worries about how Áine's needs will affect her sister.

"Family and friends step up when we need, though," Trudy adds. "And over time we have learned as parents how to manage better, often splitting up and taking a child each rather than trying to do everything together." Trudy and Ian also keep in contact with other parents of children with Miller-Dieker Syndrome through a Facebook group. As the condition is so rare, most of the other parents are from the United States but many of the issues they face are the same.

"We feel so lucky to have Áine," says Trudy. "We just see it as a privilege. It's hard work but, when you see her smiling face each morning, you can deal with anything."

About Miller-Dieker Syndrome

Miller-Dieker syndrome is caused by a deletion of genetic material near the end of the short (p) arm of chromosome 17. The size of the deletion varies among affected individuals. Most cases of Miller-Dieker syndrome are not inherited. When Miller-Dieker syndrome is inherited, its inheritance pattern is considered autosomal dominant because a deletion in one copy of chromosome 17 in each cell is sufficient to cause the condition. About 12% of people with Miller-Dieker syndrome inherit a chromosome abnormality from an unaffected parent. In these cases, the parent carries a chromosomal rearrangement called a balanced translocation, in which no genetic material is gained or lost. Miller-Dieker syndrome appears to be a rare disorder, and there is not much information about how many people have this condition worldwide.

The signs and symptoms of Miller Dieker include problems in brain development known as lissencephaly. These brain changes cause severe intellectual disability, delays in development, seizures, abnormal muscle stiffness (spasticity), weak muscle tone (hypotonia), and feeding difficulties. Some individuals with this condition also grow more slowly than other children. People with Miller-Dieker syndrome may also have life-threatening breathing problems. Most individuals with this condition have a shortened life expectancy and complex care needs.

Profile 13 | Fatiha & Manal Akka | Merosin Negative Congenital Muscular Dystrophy

Fatiha Akka is a bright and confident 12-year-old. She does well at school and has good friends, and looks forward some day to having a home and family of her own. Her younger sister, Manal, is five years old and their mother, Radia, says they are cheerful, happy girls.

When Fatiha was only a few months old, Radia noticed that she was not starting to move or support herself like other babies her age. Her GP took her concerns seriously and referred her to Temple Street Hospital where, after a series of tests and a muscle biopsy, they confirmed after another few months that Fatiha had Merosin Negative Congenital Muscular Dystrophy. Manal was also born with it.

The muscles of the whole body are affected by this condition. This means that both girls need 24-hour assistance with everything: feeding; washing; toileting; turning at night and every basic movement that others their age take for granted. They had each learned to shuffle on the floor, which gave them some independence around the home, but already Fatiha, due to her increasing age, has lost the muscle power needed for this, and even needs help to sit upright now.



“...both girls need 24-hour assistance with everything: feeding; washing; toileting; turning at night and every basic movement that others their age take for granted.”

Radia and her husband, Salim, care for their children full-time. They have both worked outside the home at different times since the girls were born but, as Fatiha and Manal grew and their care needs increased, it has proved impossible recently to find anything that would fit in with school hours. While they operate as a team, Salim principally looks after Fatiha, who wakes more than five times each night. Her blood sugar has to be watched constantly as she has diabetes, and she sleeps with oxygen and a BiPAP machine or portable ventilator. Radia is the principal carer for Manal.

Radia says “I do my best for them...I have always tried to give them confidence. I tell them ‘nobody can stop you.’ If they want to play like normal kids, I bring them to the playground. Salim can lift Fatiha, even if I can’t.” Like every parent, Radia wants the best for her children. She is currently fighting for a full-time Special Needs Assistant for Fatiha at school. As Fatiha is so weak, she cannot even pick up a pen for herself if she drops one. The family also need a car that can take two power chairs. They do not qualify for a grant until the children are much older but, in the meantime, without work, the banks will not loan to them either. “Sometimes I feel like we are stuck in one place,” Radia explains.

She admits that there have been times when it was very hard to cope. After Manal’s diagnosis, she attended a psychologist herself and learned how to stay positive.

"Sometimes I feel alone in all of this," she says, "but with the help I've had and my belief in God I put those thoughts aside and the feeling passes."

"When Fatiha was diagnosed, we had no internet access at home and I had very little information," she recalls. "I did not really want to believe that it was true anyway...But after a family support worker from Muscular Dystrophy Ireland (MDI), www.mdi.ie, visited and explained things to us, I started to accept it." Radia also found meeting other parents with children affected by the condition very helpful.

She can no longer take the children to Algeria to stay with family in the holidays because, now that Fatiha is older and cannot manage without her power chair, nothing is accessible there. Some respite care is available to the family through MDI but daily life without help at home is a struggle. Fatiha, especially, is an inspiration to her mother. She is big enough to tell Radia not to worry and Radia feels "Because they're happy, I'm happy."

About Muscular Dystrophy

Muscular dystrophy is the collective name for a range of genetic neuromuscular conditions which are characterised by the progressive weakening and wasting of the muscles. Some forms arise at birth or in childhood, others may not manifest themselves until later in life. The prevalence varies depending on the exact condition, for example from 1:3,500 male births for Duchenne muscular dystrophy (unlike other conditions which affect both sexes, Duchenne affects males almost exclusively), 1:6-10,000 births for spinal muscular atrophy and 1:20-50,000 births for congenital muscular dystrophy. There are over 640 individuals with neuromuscular conditions registered as members of Muscular Dystrophy Ireland but a full registry is needed to get more accurate statistics.

Although there is no cure for muscular dystrophy there are ways to manage the condition and enhance the quality of life. People with neuromuscular conditions require access to a timely diagnosis by a neurologist and access to genetic testing and counselling. They should continue to have neurological assessments at multidisciplinary muscle clinics throughout their lives with referral to relevant specialists when required, particularly as the condition can affect the heart and breathing.

Quality of life depends on the ability to access aids and appliances to address mobility problems, accessible housing and transport, respite care, personal assistants, family support and counselling. With these supports, people with muscular dystrophy can live an independent life of their own choosing, which is the ethos of Muscular Dystrophy Ireland.

Profile 14 | Kerry-Ann McLaughlin | Thyroid Cancer

At the age of 9, Kerry-Ann McLaughlin noticed that her jumpers and t-shirts were getting very tight and uncomfortable around her neck but her GP said it was nothing to worry about. Concerned that the swelling was gradually getting bigger, her parents took her to another doctor, who arranged a hospital appointment in Galway. After a series of tests, Kerry-Ann was told she had teenage goitre, that it was nothing to worry about and that she would grow out of it in time. That was in 1999.



“...a long journey to diagnosis was followed by an equally difficult road to treatment.”

For the next five years, she attended regular check-ups. As her neck continued to get bigger, scans were arranged in 2004 and again in 2006. Both times she was told that there was nothing to worry about. By 2007, her overall health had deteriorated; she was very tired and losing weight. On a further consultant visit, she was sent for a biopsy.

“At this stage my parents were beside themselves with worry,” remembers Kerry-Ann. “I was starting to get a little worried too as I was unable to do the things a normal 16-year old would be doing.”

A week before her Junior Cert began, she was told that a growth had shown up and would need to be operated on. Within days of finishing her exams, she was called for surgery. “My family waved me off and were told the operation would take forty five minutes to an hour,” she says. In what turned out to be a six and a half hour surgery, Kerry-Ann then had the right side of her thyroid removed “as a precautionary measure.” Less than two weeks later, her parents were shocked to receive a call to bring her in for a second surgery. This was the first time that thyroid cancer was mentioned. Cancer of the thyroid is rare anyway but even more so in a young person.

Kerry-Ann remembers now “I was 16, I was scared, but I didn’t realise what he was saying to me. We were shocked and disgusted that this wasn’t detected earlier with the previous scans that were done and that I had been attending the hospital for the past seven years with the same problem.”

Following the second surgery, Kerry-Ann was told that she would require radiotherapy within six weeks and was transferred to another consultant. But her long journey to diagnosis was followed by an equally difficult road to treatment. After months of unexplained delays, her parents tried everything to get treatment for her, including approaches to media and local politicians. Through a personal contact, they finally managed to get her an appointment in Belfast in June 2008.

After a year and a half off school Kerry-Ann came back fighting and passed her Leaving Cert. “I am now 21, in college and getting on with my life,” she says.

She points out that CanTeen has played a huge part in her recovery. CanTeen Ireland, www.canteen.ie, is a national support group for young people who have or have had cancer. It also provides vital support to their siblings and friends and is associated with the Irish Cancer Society.

About Thyroid Cancer

The thyroid is a gland at the base of your neck, just below your Adam's apple. It produces hormones that control your heart rate, blood pressure, body temperature and weight. Thyroid cancer occurs when the thyroid cells grow abnormally.

While thyroid cancer is rare, it is on the rise. In 2009, there were 154 people diagnosed with thyroid cancer in Ireland. It usually affects people who are middle-aged or older and it is more common in women than in men.

There are five main types of thyroid cancer. The most common type is papillary thyroid cancer. Symptoms may be vague at first because thyroid cancer grows very slowly. Symptoms can include:

- A painless lump in your neck, which gradually gets bigger
- Difficulty in swallowing or breathing
- Changes to your voice, including hoarseness

The exact cause of thyroid cancer in most people is unknown. Some risk factors which can increase your chance of getting thyroid cancer include:

- Certain benign (non-cancerous) thyroid diseases
- Diet
- Exposure to radiotherapy
- An inherited faulty gene

Surgery is usually the first line of treatment for thyroid cancer. Other treatments for thyroid cancer include hormone therapy, radioactive iodine and radiotherapy.

Profile 15 | Liam & Saoirse Heffernan | Batten disease

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).

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



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.

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.

Section 3: The National Plan for Rare Diseases (NPRD): Priorities of Patient Groups

The European Union (EU) recommends all member states ensure that patients have access to high quality care, including diagnostics, treatments and supports and effective orphan drugs for those living with rare diseases. The EU recommends that health policies are inclusive of rare diseases.

Consistent with this objective, the Irish Government, along with other EU member states, is required to draw up a National Plan for Rare Diseases (NPRD) by 2013. The NPRD has the potential to make a significant impact on the treatment of rare diseases in Ireland, provided the plan is robust, effective and resourced.

Also in line with EU policy, the active participation of patient groups is central to the process of developing a Plan. Furthermore, the Task Force seeks to work together with health care professionals, industry representatives and policy makers to assist in the planning, implementation and review of an effective NPRD. The following are the key priorities that should be included in the NPRD as proposed by the patient groups involved in the Task Force on Rare Diseases:

1. Planning

Ireland needs an effective National Plan for Rare Diseases: There is considerable fragmentation in current services and treatment for rare diseases. Ireland needs an effective NPRD to ensure that people with a rare, often life threatening disease, have equal and fair access to treatment and a better quality of life. There has been growing awareness in Ireland in recent years of the sometimes devastating impact of living with a rare disease, particularly for families without access to adequate services and expertise.

2. Diagnosis

Access to correct diagnosis: Medical practitioners may fail to identify symptoms that they rarely come across, resulting in significant potential for patients to experience lengthy delays in diagnosis and often misdiagnosis.

The lack of a NPRD and the scarcity of medical expertise often results in patients experiencing prolonged delays in getting an accurate diagnosis. Frequently it involves years of repeated visits to different doctors. Misdiagnosis or non-diagnosis are significant hurdles to improving quality of life for thousands of patients, and often results in inadequate or even harmful treatments.

3. Information

Access to information: Once an accurate diagnosis has been made, additional hurdles must be overcome. Patients and their carers may have considerable difficulty sourcing appropriate information about the condition and identifying relevant/experienced specialists (should they exist in Ireland) can be particularly challenging.

Delays and inappropriate treatment can aggravate existing symptoms and leave those affected feeling helpless, vulnerable, neglected, misunderstood and uncertain about their future health.

4. Treatment and Development of a National Clinical Programme

Access to treatment: For some conditions where no medicine/drug therapy exists, symptoms can be managed effectively with a correct multi-disciplinary approach leading to improved health and a greater quality of life. Many rare diseases are multi-systemic; therefore timely referral to the range of appropriate consultants is essential with collaboration between specialists. Equality of access to treatment is a key issue regardless of condition, geographical location or income.

The lack of any national specialised assessment and commissioning process to evaluate, approve and reimburse orphan medicines results in unnecessary and prolonged delays for patients trying to access available treatments. We welcome the Treatment Abroad Scheme (TAS); however, every effort should be made to provide treatment within Ireland or as close to home as possible.

Progressive disorders require pre-planning, fast track support including access to existing services, education and counselling services.

Access to available orphan drug treatment is haphazard and due to the lack of a national policy, each region or hospital makes individual treatment decisions. This process results in geographic discrimination for patients, long delays in accessing treatment, lack of transparency and no identified appeals process. A commitment is required to ensure universal access to high quality healthcare on the basis of speedy access to good quality care, equality and solidarity. Treatment should be made available on the basis of need at the point of care closest to the patient.

It is to be welcomed that the Irish Government has recently agreed to develop a National Clinical Programme (NCP) for rare diseases which will be added to the existing programmes: www.hse.ie/eng/about/Who/clinical/natclinprog

NCPs have been established to improve and standardise patient care by bringing together clinical disciplines and enabling them to share innovative solutions to deliver greater benefits to every user of HSE services. As part of the NCP a lead clinician is appointed and a steering group bringing together key stakeholders is established.

The NCPs are based on three main objectives:

- To improve the quality of care delivered to all users of HSE services
- To improve access to all services
- To improve cost effectiveness

5. Access to and innovative therapies for rare diseases

New and innovative therapies, including exciting and effective gene modification drugs for rare diseases, are increasingly becoming available. It is important that such therapies should become available to patients in Ireland as soon as possible after assessment.

'Orphan drugs' for rare diseases are often expensive due to high research and development costs and because of a relatively small patient base from which drug companies can recoup costs. However, these new drugs can both significantly increase quality of life and save other public expenditure through, for example, reduced hospital admissions. In some cases, these new therapies can add years of life to a patient with a rare disease.

6. Expertise

The development of centres of expertise: Centres of expertise on rare diseases should be commenced or further developed in Ireland. This approach is recognised internationally as the most important way to address the multifaceted issues raised by rare diseases.

It is well recognised that the best way to improve access to high quality healthcare for rare diseases is to establish new centres and to develop existing centres of expertise. Centres of expertise may also facilitate the development of specialised social services which will improve the quality of life for those with rare conditions. In the absence of such centres in Ireland, patients and families search endlessly for funding and improved services, often without success. It is of vital importance that existing centres of expertise should be strengthened and resourced to serve people with rare diseases in Ireland. For example, the National Centre for Medical Genetics (NCMG) has a major role in helping Irish families with rare diseases, given that 70-80% of rare diseases are genetic in origin. However, the NCMG has lost over 20% of its staff due to the resource restrictions introduced by the HSE, leading to a diminution, rather than an improvement, in services (see Section 5: Useful Resources).

7. Co-ordination

Co-ordination of Policy at a national level: A clinical lead and a national office for rare diseases should be established to co-ordinate the planning, implementation and review of a NPRD.

8. Participation

Participation of patient groups: Patient groups are often the drivers for change in health policy in Ireland. Patient groups must be included in all aspects of the national action plan on rare diseases including the design, implementation and review of the plan. Recognition must be given to the many rare disease support groups which are sometimes the main specialist support and information provider on their particular rare condition.

The development of health policy and service delivery should be patient centred with the patient becoming an active participant in service design and implementation. True consultation must involve patients and representative organisations. Such stakeholder collaboration is essential and should not be confused with merely informing stakeholders of policy decisions after the event.

The co-ordination of care between paediatric and adult services including transition from one to the other should be managed effectively.

9. Research

Greater focus on research on rare disease at a national and international level: Funding for research into rare diseases should be given a considerably higher priority than at present. The more common diseases tend to attract the largest research budgets. Incentives/positive action should be introduced to stimulate research into rare diseases both within Ireland and through collaborative clinical research with other countries.

10. Data

Improved data to treat rare diseases: The importance of collecting and analysing data, including the development of electronic patient records, patient registers and patient registries is increasingly recognised as playing a vital role in the treatment of all diseases. National standards in collecting and analysing such data should also apply to rare diseases.

11. Mainstreaming, targeting and resources

The national action plan on rare diseases cannot be a 'virtual plan': All relevant health and social service strategies in Ireland should be inclusive of rare diseases, including the HSE Corporate Plan and all relevant current and forthcoming health strategies. Targeted additional resources to treat and manage rare diseases must be provided under the NPRD.

12. The importance of a cross-border and European dimension to treating rare diseases

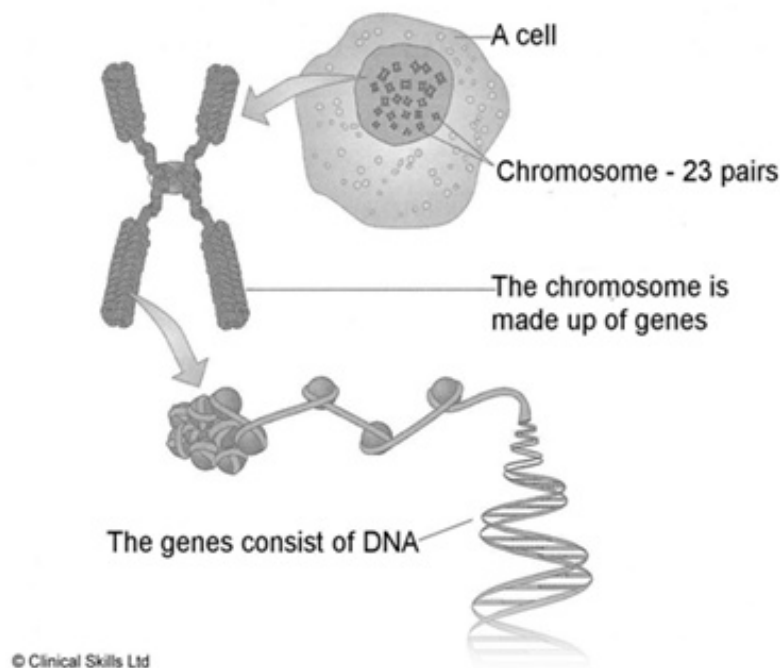
A vital part of the forthcoming NPRD should be a commitment and indicative actions to promote collaboration. There should be innovative ways for clinicians and patient groups working in the field of rare diseases in Ireland (north and south) and Britain to work collectively. Public policy on rare diseases is an increasing concern for the EU and a pan European dimension should also be reflected in the NPRD.

Section 4: Glossary of Key Terms

The following section seeks to provide an introduction to frequently used terms in relation to genetic rare diseases.

Genes are located on small thread-like structures called chromosomes. Usually we have 46 chromosomes in most cells. One set of 23 chromosomes we inherit from our mother and one set of 23 chromosomes we inherit from our father. So we have two sets of 23 chromosomes, or 23 pairs. Sometimes, there is a change (alteration) in one copy of a gene or chromosome which stops it from working properly. This change can cause a genetic condition because the gene is not communicating the correct instructions to the body. Some examples of genetic conditions include alpha-1 antitrypsin deficiency, cystic fibrosis and muscular dystrophy.

Genes, Chromosomes and DNA



Genetic glossary

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.

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

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

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.

Chromosome disorder

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.

Chromosome testing

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.

DNA

A chemical substance which makes up the genes, and which contains the information needed for the body to work.

Family tree

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

Gene

Information needed for the body to work, stored in a chemical form (DNA) on chromosomes

Gene alteration

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.

Genetic condition

A condition or disease caused by an abnormality in a gene or chromosome.

Genetic counselling

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.

Genetic counsellor

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.

Genetic test

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.

Hereditary condition

One that is inherited (passed down through families).

Metabolism

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.

Metabolic disorder

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.

Orphan Disease and Drugs

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'.

Predictive testing

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.

Sporadic

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.

X-linked genetic conditions

These are conditions that mainly affect males as the changed gene is on the X chromosome. Males only have one X chromosome and are affected as they don't have another copy of the gene like females do, as females have 2 X chromosomes. Females are generally not affected but this can vary depending on the genetic condition.

Section 5: Useful Resources

Further Resources – Ireland

- Clinical Trials Information for Patients | www.clinicaltrials.ie
- Department of Health and Children | www.dohc.ie
- Disability Federation of Ireland (DFI) | www.disability-federation.ie
- Europlan Information | www.europlan.ie
- Inclusion Ireland | www.inclusionireland.ie
- Irish Patients' Association | www.irishpatients.ie
- Irish Stem Cell Foundation (ISCF) | www.irishstemcellfoundation.org
- National Centre for Medical Genetics, 5 year report, 2007-2011 | www.genetics.ie/documents/5-year-NCMG-report-2007-2011.pdf
- Staffing of Medical Genetics Centers across Europe | www.eshg.org/111.0.html

Further Resources – Transnational

- European Organisation for Rare Diseases (Eurordis) | www.eurordis.org
- Genetic Alliance UK | www.geneticalliance.org.uk
- National Organisation for Rare Disorders (NORD) USA | www.rarediseases.org
- Northern Ireland Rare Disease Partnership | www.nirdp.org.uk
- Orphanet Report Series – Disease Registries in Europe – January 2013 | www.orpha.net/orphacom/cahiers/docs/GB/Registries.pdf
- Rare Connect | www.rareconnect.org
- Rare Disease Day | www.rarediseaseday.org

Section 6: Contact Details of Patient Networks and Patient Advocacy Organisations

Patient Networks



GRDO (Genetic & Rare Disorders Organisation)
C/O MDI,
75 Lucan Road
Chapelizod
Dublin 20
t: 01 6269774 / 086 0229262
e: info@grdo.ie
w: www.grdo.ie



IPPOSI (The Irish Platform for Patients' Organisations, Science & Industry)
1 Grove Road
Rathmines
Dublin 6
t: 01 407 1629
e: info@ipposi.ie
w: www.ipposi.ie



MRCG (Medical Research Charities Group)
La Touche House
1 Grove Road
Rathmines
Dublin 6
t: 01 491 2044
f: 01 633 5104
e: info@mrcg.ie
w: www.mrcg.ie

Patient Advocacy Organisations



22q11 Ireland
Carmichael House
North Brunswick Street
Dublin 7
t: 087 7412856
e: tech.22q11ireland@gmail.com
w: www.22q11ireland.org



Alpha One Foundation
RCSI Building
Beaumont Hospital
Dublin 9
t: 01 809 3871
e: alpha1@rcsi.ie
w: www.alpha1.ie



Ataxia Ireland
(formerly Friedreich's Ataxia Society Ireland)
4 Leopardstown Business Centre
Ballyogan Avenue
Dublin 18
t: 01 29 99 033
f: 01 29 99 055
e: info@ataxia.ie
w: www.ataxia.ie



Bee for Battens
Castledrum
Castlemaine
Co. Kerry
t: 083 0044 444
e: buzz@beeforbattens.org
w: www.beeforbattens.org



Cystic Fibrosis Ireland
(formerly the Cystic Fibrosis Association of Ireland)
24 Lower Rathmines Road
Dublin 6
t: 01 496 2433
f: 01 496 2201
e: info@cfireland.ie
w: www.cfireland.ie



Cystinosis Foundation Ireland

1 Terenure Place
Terenure
Dublin 6W
e: mail@cystinosis.ie
w: www.cystinosis.ie



DEBRA Ireland

La Touche House
1 Grove Road
Rathmines
Dublin 6
t: 01 412 6924
f: 01 633 5104
e: info@debraireland.org
w: www.debraireland.org



Fabry Ireland

w: www.fabryireland.ie



Fighting Blindness

1 Christchurch Hall
High Street
Dublin 8
t: 01 709 3050
f: 01 709 3010
e: info@fightingblindness.ie
w: www.fightingblindness.ie



Huntington's Disease Association of Ireland

Carmichael Centre
North Brunswick Street
Dublin 7
t: 01 872 1303
e: hdai@indigo.ie
w: www.huntingtons.ie



Irish Cancer Society

43/45 Northumberland Road
Dublin 4
t: 01 2310 500
f: 01 2310 555
e: reception@irishcancer.ie
w: www.cancer.ie



Muscular Dystrophy Ireland

75 Lucan Road

Chapelizod

Dublin 20

t: 01 623 6414 or 623 6415

f: 01 6208663

e: info@mdi.ie

w: www.mdi.ie



Raynaud's & Scleroderma Ireland

Paradigm House

Dundrum Office Park

Dundrum

Dublin 14

t: 0818 363 999

f: 01 215 7945

e: info@irishraynauds.com

w: www.irishraynauds.com

*Published by The Task Force on Rare Diseases
which brings together key patient networks
and advocacy organisations concerned with
rare diseases in Ireland.*

