13 February 2012 | Launch of Rare Disease Day 2012 Promotional Video

Featuring Six People from Ireland with Rare Conditions

Six people from Ireland, each with a different rare condition, feature in a fantastic new promotional video produced by Eurordis for Rare Disease Day 2012. Through co-operation with GRDO in Ireland (www.grdo.ie), the European rare disease alliance arranged to film the whole piece in Dublin recently. The message is simple – *It's not unusual to have a rare condition. Look out for us and you'll see how many we are!*

Read here about the people involved and watch the video at www.grdo.ie.

Stories of the participants

Lauren

"…you can have a condition but you're still a normal person."

Lauren Shaw from Dublin is eight 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 Friedreichs Ataxia.

There was very little information about the condition available, Sue found, but she was given contact details for the Friedreichs Ataxia Society in Ireland (www.ataxia.ie), 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 eight 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 an assistant in the classroom full-time, 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 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," says Sue. "She thinks she's going to be famous now!" And I'm 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."

Jamie

"You have to take life one laugh at a time."

Jamie O'Brien's ambition is to work in music production and website design. He lives in the North East of Ireland. He's 21 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 for now and so he has 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, starting to get 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.

Some things got easier after he was diagnosed at 16. He found it easier 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 Emergency Department of a hospital. 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. He would like more people, including doctors, to understand the issues around rare disease. "There's no system for people with a rare disease," he says. But he hopes that initiatives like Rare Disease Day will go some way towards building awareness and improving the situation of all those affected. In the meantime, he says "you have to take life one laugh at a time."

Josephine

"The hardest thing is not being able to dance."

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.

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 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 (www.alpha1.ie), using her own experience and her positive outlook to benefit others with the disease and raise awareness among the general public.

Christy

"Don't give up - keep going."

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.

RP is an 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 (www.fightingblindness.ie) 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

Clare Louise

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

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 Friedreichs 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.

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 Friedreichs Ataxia Society in Ireland (now Ataxia Ireland, www.ataxia.ie) 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.

Evan

"It's good to have someone close to talk to about my condition."

At 16, Evan O'Gorman can do everything his friends can do. But for Evan there can be serious consequences for even the simplest activity. To walk a short distance from his house for a game of football would cause Evan serious pain and leave him unable to walk at all the following day. Evan has Epidermolysis Bullosa (EB) Simplex.

With EB Simplex every step hurts for Evan. It started when he first learned to walk as a baby. His parents knew immediately that he was affected when they saw painful blisters starting to appear on his feet. His father also has the condition.

This has been a comfort to Evan throughout his life. "It's good to have someone close to talk to about my condition," he says. He has also met a lot of people with other kinds of EB through DEBRA Ireland (www.debraireland.org), the national charity. He gets around by cycling as much as possible instead of walking, and has become accustomed to the challenge of his condition after living with it for so long.

Occasionally the pain gets him down. But Evan has great hope that researchers will find better treatments in the future that will make things easier for him and other sufferers. He had surgery last November which has helped somewhat, even if only for the short term. Botox was put into his feet to reduce sweat production. This in turn reduces friction, which helps reduce the blistering effect.

Evan tends to focus on the positives in life to get him through. He has a great family and friends, and is a keen actor, with several paid roles already under his belt before he has even finished high school.

He was happy to volunteer for a part in the new film though, and says he enjoyed meeting the other participants. "It's great to meet others with different conditions and hear their experiences. Many of them, like me, look as if nothing is wrong. I hope that people watching will think twice the next time they notice me limping, and realise how many people out there could have a rare disease."

FACTS ABOUT RARE DISEASE

- A rare disease affects fewer than 1 in 2000 people.
- There are more than 6000 rare diseases.
- They are all different but those affected face the same problems.
- Together we make up an estimated 6% to 8% of the population – that's 270,000 + people in Ireland alone.
- 29 February 2012 is Rare Disease Day.

ABOUT GRDO

The Genetic and Rare Disorders Organisation (GRDO) is a non governmental organisation acting as a national alliance for voluntary groups representing the views and concerns of people affected by or at risk of developing genetic or other rare disorders.

The mission of the Genetic and Rare Disorders Organisation is to provide a strong voice for voluntary groups representing people with or at risk of developing genetic or other rare disorders in order to achieve better support and services.

The Genetic and Rare Disorders Organisation acts as a watchdog in relation to legislation concerning disability to ensure that the rights of people with genetic or other rare conditions are protected.

ENDS

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