



Beauty of Rare

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This project has been created and produced by photographer Ceridwen Hughes on behalf of Same but Different, an organisation that uses the arts to give people with rare diseases and disabilities a stronger voice in their communities.

We wanted to create something that encouraged people to want to know more and to find out about the people involved and their conditions. Despite 1 in 17 people being affected by a rare disease, awareness is limited.

You can find out more information about the people who have taken part and learn about their conditions in the online exhibition at www.samebutdifferentcic.org.uk.



Jonathan

“I have Huntington’s which is a really complex disease that affects you in different ways. I was diagnosed when I was 26, but to be honest, I’ve always believed that I’ve had it. Both my sister and brother have it and they live in a care home.

The worst part of the condition is the end probably. Seeing my sister and brother and the way it has affected them, but at this point I am just trying to be positive. That’s all I’ve ever done.

There is no treatment for Huntington's Disease at the moment. It is really important that as much research as possible is done to find a treatment because it is a terrible disease.

When you initially get tested and given the diagnosis, it is really hard going. It is a nightmare. I just realised that I had to keep my mind occupied and so that is why I started running. I used to run nearly every day and have completed marathons but I have had to give it up.

If I was able to speak directly to the researchers I’d say it is the worst illness of all time. They need to keep on fighting for the people like myself and my family because we’re all fighting for the research ourselves, every day.

If you start thinking about it, it’ll drive you crazy. You just have to crack on with your life.”





Lizzie

Lizzie's mother explained about the impact Huntington's Disease has had on their daughter.

"I was pregnant with Lizzie, she was my third baby, when we discovered my husband had the disease. One by one my children have been diagnosed with this terrible disease.

Lizzie is my youngest child, the baby, and she was diagnosed at 16. I just could not believe that my beautiful daughter had this terrible condition too. We went through really, really dark times with Lizzie after her diagnosis. She too took to self-harming and she really didn't want to be here.

She has deteriorated a lot quicker than her eldest brother and has gone from a bouncy, dancing, beautiful girl to being unable to speak, doubly incontinent and unable to walk. It is a terrible, terrible disease.

People ask me how I get out of bed in the morning but you have to, it's your kids. What would they do without me?

It would mean the world if they could find a cure or treatment for Huntington's Disease. No-one should have to go through what my three children are going through."





Vinnie

“We knew from about 4 weeks old that there was something not quite right as Vinnie wasn’t holding his head. I tried taking him to the hospital and to the doctors but no one would listen to me. When he was 8 weeks old I took him to the hospital again and refused to move until he was seen.

They carried out tests and Vinnie was diagnosed with Spinal Muscular Atrophy (SMA) Type 1. What this means for Vinnie is that his body only produces 10% of the protein because he’s missing the SMN1 gene. This means his motor neurones do not stay alive, they gradually die, leading to the muscle wastage around his body.

The weakness will tend to be worse in the lower region. A lot will have some arm, finger and hand movement but it will affect every muscle eventually, even swallowing and breathing. It can also affect the muscles in the eyes.

He is cheeky. His personality is out of this world and he’s just the funniest little boy. He’s so clever as well, oh my god, that boy is so bright.

Getting Vinnie accepted on a drug trial meant everything. It gave us hope. We don’t want anyone’s pity. Don’t feel sorry for him, he’s happy, he couldn’t be more loved and have a more supportive family.”





Toni

“I first heard of Niemann-Pick disease when my daughter, Lucy, was about 5 weeks old. We looked at our gorgeous little baby and just could not believe that she was so unwell. When we were given the initial diagnosis, we were given the wrong information. I would love to revisit that appointment with the consultant and have the strength to tell him how much an impact his words had upon us. He destroyed us completely within that appointment. Delivering news like that is very, very difficult but I feel that there must be more sensitive ways.

We went on to have two further children but sadly they too were born with the condition and we lost all three children within the space of four years.

For me, being involved in the patient group Niemann-Pick UK has been an absolute life saver.

I didn't want anyone's pity and I still feel I don't need it, even though, yes, I get that reaction. If someone asks have you got children, and then I tell them our story, I can see the pity in their eyes. I don't want that, I want them to listen and understand that I am very grateful for the experiences I've had and the time that we had with our children. It's very special to me and I feel very lucky. I know that sounds ridiculous, but I do, I feel really lucky and I want people, if I talk to them about my experiences, to learn from me and pass it on and raise awareness and support and help. That's what I want from them and not their pity.”





Jack

“Jack was diagnosed with Duchenne Muscular Dystrophy in 2015 when he was just 2 years old. It’s a rare condition, that mainly affects boys. It’s a muscle wasting disease, so he’ll gradually begin to lose all of the skills that we’ve watched him gain.

When we were given the diagnosis it was done over the phone by a duty doctor and we feel we should not have found out in this way. It’s never going to be good news to hear, it completely throws your world upside down. It takes away all the hope that you have for your child and you start grieving for the life that they should have.

After the diagnosis we just threw ourselves into fundraising and finding out as much as we can in the hope that new drugs and treatments will come along and help to save his life. We try and keep him in the best condition that we can do so that when that treatment or cure comes along he’s going to have the best chances of extending his quality of life, and it’s all about quality of life.

We try not to think to the future. You have to live every day because looking ahead is just too scary, it’s too unbearable. You want to press a pause button, more so than any parent, because you dread what the future holds if this disease continues.”





Maddox

“Maddox has a rare genetic disorder, which is a perinatal form of hypophosphatasia (hpp).

When he was born, we all noticed different things about him that weren't what we were expecting. I noticed that his legs were bowed and my mum noticed his breathing. He was diagnosed at a week and a half old because of his failure to thrive.

Initially they were preparing us for bad news and they spoke about palliative care. We were fortunate that one of the consultants did lots of research and identified a drug trial for the condition in Birmingham.

It meant relocating for up to six months, which I had to do on my own, as Maddox's dad needed to continue to work. We were very lucky with the drug trial as we noticed changes in Maddox after a few weeks. He started to get stronger and continued to improve. He was late with sitting up and walking and crawling and that kind of thing but he caught up eventually.

We couldn't believe as a family that there was even a study going on about such a rare condition as hypophosphatasia. We feel really lucky that we were in the right place, at the right time and we had access to the drug for Maddox because without it, he wouldn't be here now.”





Lucy

“I have Ehlers-Danlos Syndrome Type 3 which means I am hypermobile. It is a soft tissue connective disorder which can affect anywhere in the body. Because it’s a collagen deficiency as well, it can affect anywhere in your body from your brain to veins or even your heart. It just has an effect everywhere.

As recent as 2017 I also lost all my hair, it just fell out in the space of just 48 hours. As a 17 year old girl, to lose all my hair was huge. I felt really ugly and just didn’t want anyone to see me.

Having a rare disease as a teenager has impacted on me massively. When I initially became ill, I was right at the start of my teenage years and building my first proper friendships.

I have only really got one proper friend who has stuck by me. There was a time when I missed a year and a half of school because of EDS and so I’ve basically had no friends or social life for a few years.

When you’re in hospital, you meet friends that are going through the same thing as you, but when you leave hospital you have no one that’s going through the same thing as you and it is hard.

If I could tell people one thing it would be that having a rare disease does not make you a weirdo. We’re just as normal as you, we just have a few quirky little bits about us. I just want people to treat me normally and accept our differences.”





Eddie

“Eddie has Infantile Spasms. This is why he is so slow to develop. It is like you’re constantly rebooting a computer, and so at nearly 3, he can’t sit, he can’t walk, he can’t talk and it’s because his brain is just firing away. That’s what makes it so catastrophic really.

He’s a really happy boy, he’s very sociable and loves hanging out with people. However, Eddie’s a boy who is seriously held back by his condition.

We’ve tried quite a few epilepsy medicines, there are still a couple we could try but the chances of them working are really slim now because he’s tried so many of them. The spasms are so catastrophic, the next option is to look at trying medical marijuana.

We started Eddie on CBD over a year ago and that has been what’s worked best for him. Because we’re operating within the law, we’re at the end of where we can go with this legally in the UK. One option we might have to consider is moving to Holland but this will mean uprooting our entire family and living somewhere else with none of the support structure we currently have.

I just want something that’s going to make our son better. It scares the living daylights out of me. I want the authorities to recognise that there’s something hugely unfair that there are other countries that are willing to take this product on board, that they have evidence for that works, and I want them to do the same over here.”





Barbara

“Thirty five years ago when my two children were small, and I was a single parent, I was diagnosed with progressive Multiple Sclerosis. To say it changed my life is possibly no understatement.

Like most people, I love sunny days and spending time with my family and friends. I also love my weekly singing sessions with the Tenovus choir. The odd tittle of red wine is also welcome and some may say that my singing even improves afterwards!

If I had to list things I do not like, I would have to include the rain, miserable people, being tired and not being able to pursue what I want to do.

As no two days are the same I have to plan my life on a weekly/daily basis. I try not to plan too many events in a given time, which is frustrating, but the severe fatigue from doing too much is unbelievable.

MS manifests itself differently from person to person and mine affects my balance. I sometimes have a major balance malfunction and appear drunk and it restricts how far I can walk now. Basically, my head and heart want to do so much but my body has other ideas!”





Meliz

“I was diagnosed with Beta Thalassaemia Major at a relatively late age as I was three. I have six siblings and some of them have the less severe form. The condition itself is a blood disorder that requires me to have a blood transfusion every three weeks. This takes up to 6 hours each time as I am given three litres of blood. The side affect of this is that it can lead to a build up of iron which means I have to take tablets to reduce it and be careful what I eat. It is surprising how many fruit, vegetables and meat contain high levels of iron. When I eat a meal I often have a cup of black tea as that reduces the iron absorption.

I can get very tired and irritable in the time leading up to my transfusion. Growing up with the condition meant I missed lots of school and it really affected my friendships. Not everyone understood why I would be tired or needed to go to hospital so often.

Awareness is really important because having a condition like this affects everything from education to getting a job. Not all employers are understanding or tolerant of my need to take time off to get a transfusion. I am really lucky with the company I work for now but it has been difficult in the past.”





Rare Disease Facts

A rare disease is defined by the European Union as one that affects less than 5 in 10,000 of the general population. There are between 6,000 and 8,000 known rare diseases.

1 in 17 people, or 7% of the population, will be affected by a rare disease at some point in their lives. This equates to approximately 3.5 million people in the UK and 35 million people across Europe.

In the UK, a single rare disease may affect up to about 30,000 people. The vast majority of rare diseases will affect far fewer than this – some will affect only a handful, or even a single person in the whole of the UK.

75% of rare diseases affect children. Rare diseases include rare cancers such as childhood cancers and some other well-known conditions, such as cystic fibrosis.

It is also estimated that over 200 new forms are identified each year.

1 in 100 babies are affected by rare diseases.

1/3 of infant deaths are caused by rare diseases.

Only 200 rare diseases have a treatment.

Rare Disease Day takes place on 28 February.



How can you help?

Many people and their families who are affected by rare disease say isolation is one of the biggest issues. Here are some ways you can help:

Have a conversation about rare diseases - tell someone else, share information. You can even share or invite people to like our Facebook page (@samebutdifferentcic)

Become a #RareAware Champion - use your own experiences to help others.

Help raise funds - in order to support our activities we need to continually raise funds. Can you help with this?

Host an exhibition - do you work at or run a business that could host one of our exhibitions? Rare diseases have a wide impact throughout the community and so the more people who know about the impact, the better.

Take part - we are always looking for people to take part in our projects.

Social media awareness - can you help us reach a wide audience by sharing rare disease awareness posts. Please use #RareAware so we can follow. On Facebook find us at @samebutdifferentcic and twitter @SBDRareProject.

Same but Different

We are incredibly grateful to everyone who took the time to take part in this project as they travelled from all parts of the UK. Our thanks also go to Soughton Hall Hotel near Mold and Gladstone Library in Hawarden for allowing us to use their wonderful venues for the photography.

We are very grateful for the support of the Big Lottery Fund Wales and F. Hoffmann-La Roche Ltd who have enabled us to create this project.

If you would like to host one of our exhibitions or to find out more about our work, please visit our website at www.samebutdifferentcic.org.uk or email us on enquiries@samebutdifferentcic.org.uk.



