



Media Briefing

Isolated and abandoned: the hidden impact of rare conditions

“Most people just suffer in silence and find it hard to seek the help they need to deal with this devastating and life changing news.”

Pedro Fernandes whose son, Luca, has Duchenne muscular dystrophy



Shiv Thakrar was diagnosed with Duchenne muscular dystrophy aged three years old.

Duchenne muscular dystrophy is a life-shortening muscle-wasting condition, which currently affects around 2,500 people in the UK and around 100 babies are born with the condition each year. Usually diagnosed by the age of five, most children will use a wheelchair by the age of 12 and will face life-threatening health problems by their late teens as the muscles of the heart and lungs weaken. Current life expectancy for people with Duchenne muscular dystrophy is in the mid-twenties.

"The 8th of April 2014 is the day that will haunt us for the rest of our lives. As we made our way to the hospital, to collect Shiv's results we were full of mixed emotions of hope and fear. Wanting to see our consultant as soon as possible in order to end this nightmare and hear that the results were negative, yet at the same time, not wanting to go, as deep down we were still grappling with the realisation that Shiv could have Duchenne.

"We sat numb, our hearts broke and our world turned upside down. It was in black and white, Shiv has Duchenne muscular dystrophy. We knew there was no treatment or cure. We kept asking ourselves why this was happening to us to our beautiful little boy, but there are no answers.

"Coming to terms with Shiv's condition has been, and will continue to be, extremely difficult for us. The nights are endless, mostly crying ourselves to sleep, hours of feeling angry and frustrated, just holding Shiv in our arms, hugging him and wishing that we could do something to make it all go away.

"At the time of the diagnosis we felt very alone, as though we had nowhere to turn. Despite there being a wealth of information available to read about the condition, we felt that we needed to talk to someone, someone who "was in the same boat".

"We managed to get in touch with other parents and families affected in the same way, through Muscular Dystrophy UK. This was so important as it provided us not only with emotional support but practical everyday support which is key. This gave us strength. We realised it was possible to cope positive way, in time we too would be able to move beyond our current emotional state. It would take time but it was possible.

"Support, comfort and good friendship from others has become an important part of our lives, we are no longer dealing with issues alone. It is vital that support is available immediately at diagnosis. "

Sejal Thakrar, Shiv's Mum, London



Background:

There are currently between 6,000 and 8,000 known rare conditions in the UK, affecting approximately 3.5 million people.

Living with a rare condition can be extremely isolating particularly as families, friends and extended networks may never have heard of the condition or understand what the consequences will mean for friends, colleagues or loved ones. The diagnosis process can sometimes be lengthy and very distressing for individuals and families, particularly if the condition is life limiting.

The emotional support that is offered to patients during this confusing period is vital to managing their condition and emotional well-being for the rest of their life. All too often sufficient emotional support is not available to people, instead they are left out in the cold to support themselves or seek support from non professional sources including the internet.

If left unsupported, these negative experiences can have a detrimental effect on peoples' mental wellbeing and can ultimately lead to suicidal feelings.

Therefore, it is vital that professionals and charities work together to improve the support that is available at the point of diagnosis.

Muscular Dystrophy UK undertook a survey of people's experiences of the diagnosis process because we want to make sure families and individuals have the support they need at this crucial time.

Our nationwide survey brought together the experiences of 700 people living with muscle-wasting conditions. It revealed that many people feel that the services in place for people with rare conditions, at the time of diagnosis, are inadequate, exacerbate feelings of isolation and have led to mental health problems.

Key findings:

- ▶ One in five patients reported having experienced suicidal feelings at some point in the diagnostic process.
- ▶ Over half of patients experience feelings of depression when coming to terms with their diagnosis.
- ▶ One in four respondents were forced to wait more than three years for their diagnosis after first raising concerns with a health professional.

Muscular Dystrophy UK is calling for the following:

- ▶ The NHS must ensure that every individual or family, receiving a diagnoses at a muscle centre or clinic, should have access to support from a psychologist, ideally one with neuromuscular expertise;
- ▶ The UK Strategy for Rare Diseases calls to provide patients with 'clear and timely information about their condition and its development, treatment and therapy options and practical support'. Our survey reveals that, for many people, this is not standard practice. NHS England – and the devolved administrations - must ensure that high quality funded specialist care is provided so these aims can be met as quickly as possible;
- ▶ There needs to be a joined up approach between the UK Government's rare disease strategy and the mental health and suicide strategies; to make sure that those living with life-limiting, rare and progressive conditions do not fall through the emotional safety net;
- ▶ Health and social care professionals should signpost newly diagnosed families and individuals to Muscular Dystrophy UK in order to help overcome the feelings of isolation many have reported in the survey. Individuals and families can access our free phone helpline, information about specific conditions and research, as well as the chance to meet and connect with other people affected;

- ▶ An early diagnosis is vital for families whose children are affected by Duchenne muscular dystrophy. Being diagnosed early helps families access treatments - such as steroids – much sooner. This is why we are calling for the National Screening Committee to work towards implementing a UK-wide newborn screening programme for Duchenne muscular dystrophy, which would help avoid delayed diagnosis and misdiagnosis.
- ▶ NHS funded Neuromuscular Care Advisors and Nurse Specialists play a vital role in supporting people at the point of diagnosis. They can help to signpost to other services such as genetic counselling, if required. Many parts of the UK still lack these key roles and we want to see all newly diagnosed individuals and families able to access this support.
- ▶ More research is needed into rare, long term health conditions and mental health. Bodies such as the Association British Neurologists, the British Paediatric Neurology Association and the Royal College of Psychiatrists ought to work with organisation such as Muscular Dystrophy UK in order to drive forward research in this field.

About Muscular Dystrophy UK

Muscular Dystrophy UK is the charity bringing individuals, families and professionals together to beat muscle-wasting conditions.

- ▶ We support high quality research to find effective treatments and cures; and lead the drive for faster access to emerging treatments for UK families.
- ▶ We ensure everyone has the specialist NHS care and support they need, with the right help at the right time, wherever they live.
- ▶ We provide a range of services and opportunities to help individuals and their families live as independently as possible.
- ▶ We know we can beat muscle-wasting conditions more quickly by working together and hope you will join us.

Diagnosis:

The diagnosis of a muscle-wasting condition often comes out of the blue. It can be difficult to know where to turn. Muscular Dystrophy UK supports individuals and families living with a muscle-wasting condition.

Coming to terms with the news that a child has a muscle-wasting condition can be extraordinarily difficult. If the diagnosis is recent, they may feel overwhelmed and worried about what the future holds for their child, asking questions about how they can help their child to live as happy, fulfilled and independent a life as possible.

Many parents wish to hold off from telling their child about the diagnosis for as long as possible, they may often feel that by doing so they are protecting their child. This may particularly be the case if their child has been diagnosed with a condition which is likely to be life-limiting.

Muscle-wasting conditions:

There are over 60 types of muscle-wasting conditions. They cause muscles to weaken and waste over time, leading to increasing disability. The conditions may affect not only the muscles in the limbs, but also those of the heart and lungs, with many conditions significantly shortening life-expectancy.

All of the conditions are rare with a low incidence, and some are very rare. Muscle-wasting conditions can be genetic or acquired and, with few exceptions, there are currently no effective treatments or cures.

Clinical trials for some conditions are now underway and it is hoped that these may lead to the introduction of new treatments that can slow the progression of these devastating conditions.

Support:

We want to remind individuals and families that they are not alone in all of this. Muscular Dystrophy UK has a freephone helpline open between 8.30am and 6pm, Monday to Friday. Please contact: **0800 652 6352** or **info@muscular dystrophyuk.org**

Samaritans is available round the clock for anyone struggling to cope or feeling suicidal on **08457 909090** or email **jo@samaritans.org**

Mind infoline provides help for people living with mental health problems and are open 9am to 6pm, Monday to Friday (except for bank holidays). Please contact **0300 123 3393** or **info@mind.org.uk**

The emotional impact of being diagnosed with a muscle-wasting condition

Being diagnosed with a muscle-wasting condition can have a large and wide-ranging impact on a person and their family's life. These impacts most commonly lead to anxiety, feelings of depression and isolation, but also to suicidal feelings.

Victoria Jones from London whose daughter has Duchenne muscular dystrophy: "My world felt like it had ended. Every day was a low and a struggle we have many bad days coming to terms with the diagnosis."

Eleanor Easton from Blyth who has Becker muscular dystrophy: "The news was totally heartbreaking and unexpected"

Sharan Price from Milton Keynes who's son has Duchenne muscular dystrophy: "The whole experience is like a bomb exploding in front of you."

Laura Baldwin from Reigate, has Facioscapulohumeral muscular dystrophy: "I felt very lost and abandoned."

Hearing that your child, a family member or you yourself has been diagnosed with a muscle-wasting condition is overwhelming and confusing. For some people, it helps to talk to someone outside of their immediate family and friends, such as a Neuromuscular Care Advisor or a neuromuscular expert. Other people feel they need specialised professional emotional support from a mental health professional such as a counsellor or a psychologist. A referral to professional support can be made by a GP and/or for children, through a school or social worker.

However, our survey reveals that for many people this process is almost impossible.

What the survey reveals:

People have spoken about the emotional impact on their lives after either they or a family member was diagnosed with a muscle-wasting condition. These included mental health problems, with many experiencing depression and suicidal feelings.

Many people have also highlighted a number of problems with the diagnosis process. Most prominent was the length of time endured by people waiting for a confirmed diagnosis, as well as the distress experienced due to misdiagnosis.

- ▶ One in five respondents experienced suicidal feelings when learning of their diagnosis.
- ▶ Over half experienced feelings of depression when coming to terms with their diagnosis;
- ▶ More than 60 per cent of respondents experience feelings of isolation
- ▶ Four in five respondents experience feelings of anxiety when diagnosed.
- ▶ A quarter of people were left in limbo and forced to wait more than three years for their

diagnosis after first raising concerns

- ▶ A third of people have a misdiagnosis at some point during this process which can have detrimental effect on mental health.

What people have said:

Francesca Butler from Pevensey told us of her wait for the DNA test results which would eventually help diagnose her daughter with Becker muscular dystrophy led to her being medicated for “anxiety and depression during and after the diagnosis.”

Debra McLellan from Huntingdon has a son Dan who has Ullrich muscular dystrophy:

“Ullrich congenital muscular dystrophy affects less than one in a million children and it was a huge shock to us when Dan was diagnosed at 3 years old. We didn’t tell our family for weeks and weeks. We just didn’t want to believe it.

“Nothing can prepare you for the shock of diagnosis but having professional and experienced support for parents, siblings and children would definitely help during this incredibly challenging time.”

Eleanor Easton from Northumberland has a son with Becker muscular dystrophy (BMD) whose diagnosis had a traumatic effect on the entire family:

“The news was totally heart-breaking and unexpected, so scary. [I was] scared that my other two sons would also be affected, then the guilt kicked in I was found to be the carrier for BMD it ripped my family apart, my marriage ended, I still haven’t had the chance to grieve for the life my son won’t have, having to get my other two sons tested was just so stressful thankfully they tested negative, I haven’t had enough support for me and the rest of the family.”

Hayley Wood from North Yorkshire has struggled alongside her son, both in search of a specific diagnosis of their muscle-wasting conditions:

“It’s the worst thing waiting for a diagnosis as you feel uncertain about your symptoms, as doctors time and time again have misdiagnosed you or ignore symptoms. You just don’t talk to health professional anymore and start hiding the way you feel because you get the feeling that the doctors feel you are making it up and that you are depressed as you are tired all the time.

“When I was diagnosed by Oxford that I definitely had some form of mitochondrial myopathy (as yet unknown to science) it made me feel relieved and I felt I could pour out all my symptoms to doctors again and try to move forward. I felt I belonged to a community that Oxford introduced me to for the first time with group information evenings. That was a high point.”

Muscular Dystrophy UK is aware of only five specialist neuromuscular psychologists in post across the UK; with just three supporting families with children and two available to support adults diagnosed with the conditions.

This is an inadequate number of posts to support the 70,000 adults and children living with a muscle-wasting condition in the UK.

Muscular Dystrophy UK’s call to action

- ▶ The NHS must ensure that every individual or family, receiving a diagnoses at a muscle centre or clinic, should have access to support from a psychologist, ideally one with neuromuscular expertise;
- ▶ An early diagnosis is vital for families whose children are affected by Duchenne muscular dystrophy. Being diagnosed early helps families access treatments - such as steroids – much sooner. This is why we are calling for the National Screening Committee to work towards implementing a UK-wide newborn screening programme for Duchenne muscular dystrophy, which would help avoid delayed diagnosis and misdiagnosis.

Information when being diagnosed

What the survey reveals:

Many participants in the survey felt there was a lack of information given to individuals and families when they are diagnosed with a muscle-wasting condition.

Some people were forced to cope with the huge blow after being informed of their diagnosis by post or over the phone, resulting in individuals and families feeling completely abandoned and vulnerable.

- ▶ Sixteen percent of patients were informed about their condition either by post (9%) or on the telephone (7%)
- ▶ Less than a third were offered emotional support either for themselves or their family members.
- ▶ Well over a third were left to fend for themselves by not being given any information or publications about their condition at diagnosis.
- ▶ Less than half of respondents are told about research into the condition and current clinical trials or told about the support available from charities such as Muscular Dystrophy UK.
- ▶ Only fifteen percent of people are offered assistance in connecting with another family living with the same or similar condition, which charities such as Muscular Dystrophy UK can help with.

What people have said:

Hayley Lloyd from Rugby has a son Tommy (9) who has limb girdle muscular dystrophy. He received a formal diagnosis last year:

"Last year, I finally learned that my son Tommy had limb girdle muscular dystrophy. I had waited so long for his diagnosis hoping for more information but Tommy's type was so rare they couldn't tell me anything. It felt like a grieving process. It's like having a crystal ball, seeing a glimpse into the future but you can't do anything about it. I had to put on a brave face for Tommy. He didn't need to see me falling apart. I'm a single mum so it was hard carrying on as normal. I did all of my crying when Tommy had gone to bed.

"I was given no emotional support when I was initially told Tommy had Becker muscular dystrophy. It was so hard to be told such life changing news and expected to go back to normal life. It was impossible. I would have happily accepted any support available. To know what I was going through was normal would have been the greatest help."

Muscular Dystrophy UK's call to action

- ▶ Ensure that every person with muscular dystrophy or a related neuromuscular condition is supported by a multi-disciplinary team which is led by a consultant neurologist who is supported by a physiotherapist when necessary.
- ▶ The UK Strategy for Rare Diseases calls to provide patients with 'clear and timely information about their condition and its development, treatment and therapy options and practical support'. Our survey reveals that, for many people, this is not standard practice. NHS England – and the devolved administrations – must update their response to the strategy and include the critical issue of investment in specialist care.
- ▶ We are concerned to hear about the number of people who were not given any information related to research upon learning about their diagnosis. We call on hospital trusts to ensure that each person or family diagnosed with a muscle-wasting condition is offered information about clinical trials, patient registries and databases. These are critical to recruiting patients in trials; as well as improving standards of care.

Ongoing emotional support from friends, family and charities

What the survey reveals:

Our survey highlights the importance of support given by friends, family and charities, as well as professionals. These groups all ranked very highly in the support given to people who have been diagnosed with a muscle-wasting condition.

- ▶ Support from family and friends ranked second in importance to people following their diagnosis with only expert Information; with support from consultants and senior specialists ranking at the top.
- ▶ Only a third of people had been in touch with a charity for emotional support following their diagnosis.
- ▶ Of that third, 75 percent felt that contacting a charity had helped them.

What people have said:

Lucy Stenbeck from North Yorkshire whose son has muscular dystrophy (suspected Becker). When asked who she turned to most after his diagnosis she said:

"Friends and family. They were simply there, when no other options were. While charities and support groups are out there, we were not made aware of them by the NHS and have had to do all the research ourselves."

Judith Wellby (pictured right) from Leicestershire is a keen advocate of the use of a support system, in particular, she found the support services offered by certain charities to be of help. Judith said of support during the diagnosis process:

"I was diagnosed with CMT type 2 over two years ago. I was totally unaware that I had a long term condition, so it was great shock to be told that I had an incurable, untreatable, progressive disease. My reaction was now what?"

"Muscular Dystrophy UK and CMT UK was and is my lifeline. Having someone who understands you have problems without limiting your abilities, and encourages you to get involved in helping others in life, therefore helping yourself and having a purpose in life."

Sheonad MacFarlane, Scotland has a daughter, Eilidh has spinal muscular atrophy (type 2):

"On the day we started on our journey with SMA I was heartbroken and without hope. I was lost. I remember feeling instant grief; I felt as though my heart had fractured. For me, there was anger, disbelief, tears. I couldn't reach out to tell our friends but I needed to, so I emailed them – it was the only thing I could do."

"My friends and family were so supportive and not only helped me to cope, but gave me hope. I needed comfort and a sense of peace while my husband wanted information and support. Eilidh kept me going with her smile and joy, and I started my blog as a way to tell my story, to give me hope and connect to others in a similar situation."

Muscular Dystrophy UK can help individuals and families at the point of diagnosis and at every stage thereafter. We can:

- ▶ provide accurate and up-to-date information and progress in research into these conditions
- ▶ give people tips and advice about day-to-day life, written by people who know exactly what it's like to live with a muscle-wasting condition
- ▶ put people in touch with other parents and adults living with the same condition, who can share their experiences

Meeting and connecting with people who have already been diagnosed can be a lifeline

Gemma Rose lives in Essex. She was diagnosed with limb girdle muscular dystrophy.

“I have had a connection with Muscular Dystrophy UK since I was diagnosed and turned to them for information and advice. Talking to someone with a first hand understanding of living with a neuromuscular condition can make a difference. It shows there are other people who know what you are going through.”

Muscular Dystrophy UK’s call to action:

- ▶ Health and social care professionals should signpost newly diagnosed families and individuals to Muscular Dystrophy UK in order to help overcome the feelings of isolation many reported in the survey. Individuals and families can access to our free phone helpline, information about specific conditions and research, as well as the chance to meet and connect with other people affected.

On-going professional support focussing on mental health professionals and hospitals

What the survey reveals:

Our findings suggest that professional support is very important to people with muscle-wasting conditions and has a huge impact on their emotional well-being. However, the provision of this support is not always routinely available to individuals and families with muscle-wasting conditions.

- ▶ Expert information and support from professionals was ranked as the most important factor when coming to terms with a diagnosis.
- ▶ A further one in four respondents rarely received emotional support during the diagnosis process.
- ▶ Only fourteen percent felt they received sufficient emotional support at this time.

What people have said:

Professor Katie Bushby, an Honorary Consultant Geneticist at Newcastle upon Tyne Hospitals NHS Foundation Trust, and one of the world’s leading neuromuscular experts:

“When someone has received a diagnosis of a muscle-wasting condition, psychological and emotional support can really help individuals find a way of living well; supporting them to achieve their goals. For parents whose child has been recently diagnosed with a muscle-wasting condition, they can feel “lost”; and having access to psychological support at this time can make a huge difference in how families are able to cope and adjust.

“Although many families affected by muscle-wasting conditions would benefit from the support of a clinical psychologist at the point of diagnosis, very few people actually have access to this. The NHS must recognise the huge importance of the psychological care of people with muscle-wasting conditions at the point of diagnosis - and to make this an urgent priority.”

Dr Sadie Thomas-Unsworth, Specialist Clinical Psychologist at Bristol Children’s Hospital:

“When someone finds out that they or a loved one has a muscle wasting condition they typically experience a range of emotions. Many will feel “lost” in this new uncertain world they are entering. This is often a world in which many of their hopes and dreams for the future have suddenly been called into question.

“The challenge of finding a path through this new landscape is for many huge and having access to psychological support at this time can make a huge difference in how able they are to cope and adjust. Timely psychological support then can help individuals to find a way of living a full life again

in spite of their condition by providing a safe space to talk through difficult feelings whilst also supporting them to identify what really matters to them and to be able to think about how they can still achieve their goals”.

Lindsay Davidson from Hexham has two sons with Duchenne muscular dystrophy. Angus 6 and Robert 4:

“We took Angus to the doctor after we noticed Angus was having problems running, jumping and climbing up the stairs. Angus was referred to a physiotherapist who noticed something wasn’t quite right and she contacted my GP who then arranged an urgent blood test. The GP rang me with the results of the test, and I was told over the phone that Angus had Duchenne muscular dystrophy.

“When we got the news of the diagnosis me and my husband both felt totally numb. It’s like falling off a cliff. Our world had been turned totally upside down and we weren’t really sure how to start rebuilding it.

“We were told that our youngest boy Robert needed to be tested too. We were then dealt the devastating blow that he also had the condition. At the time, I could barely find the strength to tell people. It’s almost as though you are going through a process of grieving. I could have really benefitted from having someone to speak to.

“Thankfully, we’ve come a long way since then. It is so important for families like ours to have someone to talk to and have counselling services available to us as this is a very difficult time.”

Muscular Dystrophy UK’s call to action:

- ▶ NHS funded Neuromuscular Care Advisors and nurse specialists can play a vital role in supporting people at the point of diagnosis. They can help to signpost to other services such as genetic counselling, if required. Many parts of the UK still lack these key roles and we want to see all newly diagnosed individuals and families able to access this support.

Research into the impact of long-term conditions and mental health

Research has shown that people with long-term physical health conditions commonly experience mental health problems such as depression and anxiety. Not only can this have an impact on the prognosis of someone’s long-term condition but also on the quality of life they experience. In addition, the costs of providing care to this group of people are increased as a result.

Evidence collected by the King’s Fund and Centre for Mental Health shows a clear link between long term conditions and mental health. They have calculated that:

- ▶ Long term conditions affect 30% of population of England, which is approximately 15.4 million people
- ▶ 30% of people with a long-term condition have a mental health problem, which is approximately 4.6 million people
- ▶ 46% of people with a mental health problem have a long-term condition

Well respected medical journals, such as Quality in Primary Care, have argued that the growing rate of mental health issues among individuals with disability attributed to other chronic conditions calls for renewed efforts to make sure mental health interventions are available and accessible in general medical settings.

Another publication, The Journal of Rehabilitation, argues that good mental health practices are essential to rehabilitation (Chen and Crewe 2009). However, they suggest that the lack of research into mental health impacts of muscle-wasting conditions in particular has meant that little progress has been made in assessing how big a problem this actually is.

Muscular Dystrophy UK's call to action:

- ▶ More research is needed into rare, long term health conditions and mental health. Bodies such as the Association British Neurologists, the British Paediatric Neurology Association and the Royal College of Psychiatrists ought to work with organisation such as Muscular Dystrophy UK in order to drive forward research in this field.

Luca Fernandes was diagnosed with Duchenne muscular dystrophy aged three years old.

"Our son Luca was diagnosed with Duchenne muscular dystrophy in November 2013. He was just three years old.

"Utterly devastated, it was like a death sentence had been handed down for our son.

"We were left to spend long periods with no information, and we were in no state to chase for it. On top of this, Luca needed an invasive muscle biopsy and full genetic testing to confirm the diagnosis of Duchenne.

"We were offered no emotional support and instead, had to rely completely on family and friends, who had never heard of the condition and were in the dark as much as us.

"I believe that most people just suffer in silence and find it hard to seek the help they need to deal with this devastating and life changing news.

"Practical support for my family has been available from a variety of places including Muscular Dystrophy UK. It's very hard to discuss the condition but we've forced ourselves to do so, so that we could raise awareness of it.

"We have no other option but to put our efforts into something positive. Research into Duchenne muscular dystrophy is giving us great hope, and Luca is one of the boys eligible for a new drug, Translarna. We are pushing hard for the drug to be funded in this country and continue to be heavily involved in fundraising for research that could lead to further treatments for Luca's condition.

"You make yourself strong for your child, you have no other choice. However, no family should go without emotional support like we did when Luca was diagnosed.

"There is nothing more to say, families need more support at diagnosis."

Pedro Fernandes' son, Luca, aged 5 from Poole, has Duchenne muscular dystrophy

