MUSCULAR DYSTROPHY UK | OUR MUSCLES MATTER

Key community issue: awareness of muscle wasting and weakening conditions among non-neuromuscular specialist health professionals

Note: this document primarily applies to England, but has relevance for our work throughout the UK more broadly.

Background

There are over 110,000 people in the UK living with one of over 60 muscle wasting and weakening conditions. These are usually progressive, multi-system conditions that can lead to lifelong disability. They can impact the way we walk, move and breathe. As a result, people living with muscle wasting and weakening conditions need complex, long-term and multidisciplinary care to help manage their condition, to maintain independence and quality of life.

Early access to treatment, care and support is essential for people living with a muscle wasting and weakening condition. But getting a timely diagnosis for these rare conditions is often a struggle. On average, it can take five years to receive a rare disease diagnosis in Europe.[1,2] This can be because of various factors like multiple referrals, inconclusive tests, misdiagnoses, and limited health professional awareness.[3,4] Sometimes, patients may then need to become experts in their own condition, if professionals are unable to help.[5] This all can lead to a lengthy and troublesome diagnostic process, sometimes called a 'diagnostic odyssey'. In response, the UK Rare Diseases Framework identified getting a faster diagnosis and increasing healthcare professionals' awareness as two of its four priorities.[6]

People living with muscle wasting and weakening conditions also face similar challenges. Our last *Community Survey* found that more than half (55%) had four or more meetings with healthcare professionals to get a diagnosis.[7] Under half (47%) faced being misdiagnosed at some point and a significant number (69%) had to wait more than a year to receive a diagnosis. This helps explain why

[1] Eurordis, "*Earlier, faster and more accurate diagnosis*".
[2] Eurordis, "*The diagnosis odyssey of people living with a rare disease*".
[3] European Journal of Human Genetics, "*Time to diagnosis and determinants of diagnostic delays of people living with a rare disease: results of a Rare Barometer retrospective patient survey*", May 2024.
[4] Eurordis, "*The diagnostic odyssey of people living with a rare disease*", May 2024.
[5] Rare Disease UK, "*The Rare Reality – an insight into the patient and family experience of rare disease*", January 2016.
[6] Department of Health and Social Care, "*The UK Rare Diseases Framework*", January 2021.
[7] Muscular Dystrophy UK, "*We're calling for better understanding of neuromuscular conditions to speed up referral and improve vital support*", May 2024.

84% of our community identified increasing the understanding and awareness of muscle wasting and weakening conditions as a key priority.[8]

Increasing health professional awareness will therefore support people receiving a timely diagnosis. People in our community need this to access a wide range of health, social care and other support services. Taken together, this will enable people living with muscle wasting and weakening conditions to get the earliest access to the right treatment, care and support, ultimately improving health outcomes.

What are the challenges?

Our community has often raised how a lack of awareness of muscle wasting and weakening conditions by non-neuromuscular healthcare specialists is a significant issue.[9]

This can lead to:

- Delayed diagnoses, because of slow referrals into neuromuscular specialist centres and patient support organisations.
- Undergoing inappropriate tests or incorrect treatments, which can cause an unwanted effect on their condition. It may also impact them financially if they must stay in the hospital longer and miss work.
- A negative impact on their mental health, for example from being transferred between several different health departments to receive a definite diagnosis.
- Feeling invalidated as healthcare professionals are unable to recognise the symptoms they present with.
- Having to face these challenges while lacking clarity on what their condition may be or learning to live with a rare condition without the right support.
- Unplanned admissions in hospitals, which increase the risk of incorrect and harmful treatments, which can lead to the need for more care, thereby raising costs to the system.[10]

David Hick, 35, was told for over 20 years that there was nothing wrong with him. "I got diagnosed with Becker at 35, but I started noticing a decline in my health in high school. My parents took me to several doctors who told me there was nothing wrong with me and I was just being lazy. After a while, I started believing them and ignoring what my body was trying to tell me. I felt like I just wasn't good at certain things which really affected my confidence in my early 20s."

[8] Muscular Dystrophy UK, "*How you're shaping our work*", 2023.
[9] For example, non-neuromuscular specialists can include General Practitioners, Accident and Emergency staff and some Allied Health Professionals.
[10] Muscular Dystrophy UK, "*Ambulance action: improving emergency care for people with muscle-wasting conditions*".

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General practitioners (GPs)

For many people suspected of living with a muscle wasting or weakening condition, their first point of contact tends to be their GP. However, when GPs are faced with unusual symptoms associated with rare conditions like muscle wasting and weakening conditions, many don't have the appropriate knowledge to identify these symptoms for what they are.[11]

When Stewart was diagnosed with inclusion body myositis (IBM), all he received was a letter in the post. "I'd never heard of the condition, so I called my GP to get some information. The response I received was 'Oh, that's interesting. At least you know what it is now. I don't know anything about it.' And that was that."

Accident & Emergency (A&E)

Likewise, many A&E staff are unlikely to have an awareness of rare diseases, unless they have a personal interest in them. Our *"Ambulance action: Improving emergency care for people with muscle-wasting conditions"* (2019) report showed that more than 70% of respondents thought A&E staff didn't have a basic understanding of their condition and emergency needs.[12] Further research also supports the concern that emergency care staff are inexperienced in managing complications of muscle wasting and weakening conditions if faced with them.[13]

John Foxwell struggled with his breathing for years before being diagnosed with Pompe disease. "When doctors listened to my breathing, they had always dismissed me and said my lungs sounded fine. But when I was eventually referred to a specialist, they discovered I couldn't breathe out carbon dioxide. The problem was with my diaphragm and not my lungs. I was given a Bipap, which is a breathing machine, and the improvement was incredible and instant, but it took a long time to get this relief."

Allied Health Professionals (AHPs)

Although there are examples of excellent community physiotherapy provision in the UK, our "*Overstretched: Improving access to physiotherapy for people with muscle-wasting conditions*"

[11] Department of Health and Social Care, "*The UK Rare Diseases Framework*", January 2021.
[12] Muscular Dystrophy UK, "*Ambulance action: improving emergency care for people with muscle-wasting conditions*".
[13] The Lancet Neurology, "*Diagnosis and management of Duchenne muscular dystrophy, part 3: primary care, emergency management, psychosocial care, and transitions of care across the lifespan*", May 2018.

(2016) report also highlighted how some community physiotherapists may have limited experience working with people with muscle wasting and weakening conditions.[14]

What we're calling for and what we're doing

Addressing these challenges could result in more symptoms being recognised by the first point of contact, a quicker referral into neuromuscular specialist centres, an improved diagnostic journey, and faster access to treatments. This would also enable more people with muscle wasting and weakening conditions being referred/signposted to appropriate post-diagnostic support, such as to patient support organisations like us.

What we're calling for:

- We support calls from the wider rare disease sector for the UK Government to commit to building on the current UK Rare Diseases Framework and set new goals for the next Framework after the current one ends in 2025.[15]
- We're asking for greater investment in workforce education to provide non-neuromuscular specialist healthcare professionals with more time, capacity and incentive to complete rare disease training modules, including on muscle wasting and weakening conditions.
- We're asking for greater collaboration with the NHS on rare condition alert cards. By working together, we can increase their visibility and give more healthcare professionals the confidence to act on their critical insights, especially in emergencies.

What we're doing:

- We have been working hard to engage and upskill health professionals to better care for people with muscle wasting and weakening conditions. This includes reaching out to non-neuromuscular healthcare specialists to raise awareness of these conditions so even more health professionals can support our community. To do this, we provide a range of resources and events.
- We have created E-learning modules for GPs and other AHPs. We also run upskilling webinars in collaboration with neuromuscular specialists. Some examples of the webinars we have run in the past few years have targeted the specific needs of A&E staff, A&E respiratory care staff, and Speech and Language Therapists.
- We have run events and conferences to increase connections between neuromuscular specialists and non-neuromuscular specialists, for example with our Care Advisor and AHP conferences.

[14] Muscular Dystrophy UK, "*Overstretched: Improving access to physiotherapy for people with muscle-wasting conditions*", July 2016.
[15] Genetic Alliance UK, "*A Manifesto for rare diseases ahead of coming General Election*", March 2024.

- Our event at the RHS Chelsea Flower Show in 2024 allowed us to connect with more than 100 non-specialist professionals, to learn more about the over 60 conditions we support our community with.[16,17]
- Our alert cards enable anyone with a muscle wasting and weakening condition, and their families, to easily inform healthcare professionals of their specific care needs. These credit-card sized booklets cover a wide range of condition-specific symptoms and treatment needs, and important contact information for the person's specialist neuromuscular and respiratory teams.

For questions about this key community issue or campaigning, email campaigns@musculardystrophyuk.org.

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[16] Muscular Dystrophy UK, "*We're delighted to be at the RHS Chelsea flower show 2024*".
[17] Muscular Dystrophy UK, "*Raising awareness of muscle wasting conditions amongst health professionals*", May 2024.