

Development and validation of the **MYOTONIA SYMPTOM CHECKER**

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Introduction

In the diagnostic odyssey¹ it may be challenging for undiagnosed symptomatic people to find useful information online and achieve prompt, accurate medical support:

- Diagnosis can be problematic if symptoms are non-specific, and are seen in both common and rare/ultra-rare conditions.²

In the ultra-rare disease non-dystrophic myotonia (NDM), lack of myotonia awareness is a key factor in the slow rate of disease recognition:

- Myotonia is the defining feature of NDM and is consistently associated with movement limitation, stiffness, weakness, pain and fatigue;^{3,4}
- However, NDM subtypes are genetically and physiologically diverse (developing at different ages; differing in anatomic location).⁵

There is no cure for NDM, and limited access to/awareness of symptomatic treatments:⁶

- People with NDM typically endure myotonia without support: if family members are affected, they may 'normalise' symptoms; even if no other family members are affected, people may still fail to seek medical help.³
- Coping silently may lead to a heavy daily burden because of symptom impact.³

Beyond specialist neuromuscular units, clinicians rarely see NDM;³ pathophysiologic heterogeneity further confounds its identification.^{4,7,8}

In 2022–2023, Lupin Neurosciences invited neuromuscular disease specialists and advocates (the Group) to discuss diagnostic barriers in NDM.

- The Group explored information-seeking behaviours in undiagnosed NDM, with the aim of developing an instrument, to help improve global public and clinical awareness of myotonia.

Methods



Scoping exercise:

After discussion, the Group investigated NDM-related internet search behaviours in patients/caregivers:

- The exercise involved people diagnosed with NDM 2–5 years previously (N=5) and NDM-naïve subjects (N=3);
- People with NDM retrospectively described symptoms they experienced pre-diagnosis. They then undertook online information searches based on these symptoms/descriptions, not their current knowledge of myotonia;
- NDM-naïve subjects received a scenario describing (not naming) myotonia, and searched for information as though they had these symptoms;
- Data on search strategies (i.e. terminology, devices used, time taken, information sources visited, actions, outcomes, search-engine features used) were collated; follow-up interviews were undertaken, to clarify responses.



Online screening tool:

The Checker featured 9 dichotomous (yes/no) and multiple-choice questions, optimised to suit different devices.

Users were always counselled to seek a medical opinion and were advised to download and save the report: no data were stored.



Checker review, validation and rollout:

The Checker was translated into English, French and Spanish; its format, usability and perceived value were tested in a prospective, real-world, international, validation using JotForm®:

The look, feel and content of the Myotonia Symptom Checker was investigated using 9 Likert-scale questions.



Free-text answers were also permitted.

After reviewing outcomes of the validation exercise, the Checker was refined further by the Group, and fully launched.

Results

'Symptoms' presented in the scenario, or recalled by patients, were perceived to be of concern to participants, but no search identified myotonia-relevant information (Table 1). Instead, searches typically directed participants to information associated with multiple sclerosis, amyotrophic lateral sclerosis or arthritis.

Search phrase	Phrase selection logic	Resources identified (i.e. possible causes)
Why do I have a tingling pain in muscles?	Symptom that caused great concern and triggered a search	Multiple sclerosis; ALS; Fibromyalgia
Why does my hand get stuck in place when opening a door?	Worrying symptom that often occurred and was noticed by others	Trigger finger; Carpal tunnel syndrome; Arthritis
What makes me fall over and my muscles lock?	Symptom that caused great concern and triggered a search	Multiple sclerosis; Muscle spasticity; Dystonia
My muscles feel stiff during cold weather	Worrying symptom that often occurred and was noticed by others	Diverse number of conditions suggested; this search term is too vague to be useful on its own
Difficult to walk upstairs	Worrying symptom that often occurred and was noticed by others; e.g. schoolteachers noticed that a patient avoided using the staircase in busy periods	Diverse number of conditions suggested; this search term is too vague to be useful on its own

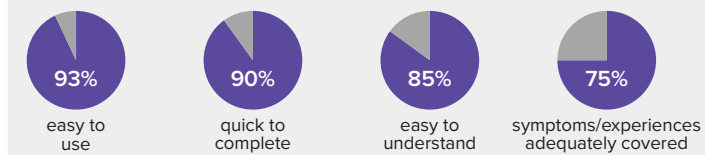
Table 1: The scoping exercise indicated difficulties accessing myotonia information when non-specific search terminology is used, as is typical for undiagnosed patients



Using data from the scoping exercise, the group developed the Myotonia Symptom Checker as a rules-based online form, targeted at families with history of NDM (to encourage them to seek medical help) and at symptomatic, myotonia-naïve people.

Participants (14 countries; N=32) used the Checker and completed the JotForm® survey.

Key responses to the validation exercise (N=32):



Few people had used a symptom checker before, but most were willing to share their results with clinicians.

Figure 1: The Checker was launched in 2024 and is available at www.myotoniasymptomchecker.com

Welcome to the Myotonia Symptom Checker.
Please take a few minutes to answer these questions, which may help to describe your symptoms or risk of myotonia. You can download a report listing your responses.

Are your muscles sometimes stiff, or do they "lock up", or feel tense?
Choose the answer that applies most to you

No | **No, but some people in my family have these symptoms** | **Yes**

Tailored thank you message.
Respondents are excluded from completing the symptom checker

Do your muscle problems (e.g. stiffness, locking, pain) disappear after you use your muscles (e.g. after a few minutes of walking)?

Are your symptoms worse when the weather is cold, and/or when you hold or eat something cold?

Are your symptoms worse when the weather is hot, and/or when you are sweating (e.g. in summer or when exercising)?

Are your symptoms worse when you are being active (e.g. going for a walk, exercising, or doing a physical job)?

Do your muscles lose their strength, quickly getting tired and achy?

Do you experience any of the following situations regularly (e.g. more than once per month)? Choose all that apply

My muscles lock up if I suddenly move (e.g. after resting, sitting still or standing for a while)

Difficulty getting up and moving smoothly after sitting

Difficulty starting to move, e.g. when crossing a busy road or waiting for public transport

Moving slowly because of my muscle symptoms

Difficulty climbing or going downstairs because of muscle symptoms

Falling if I start to move quickly

Difficulty doing sporting activities (e.g. starting to run a race)

Clumsiness as a child (e.g. difficulty performing physical activities, running)

Weakness or tiredness in my muscles

Cramp

Muscle pain

Difficulty lifting things (e.g. picking up a child), because of my muscle locking/stiffness

Difficulty brushing my teeth, or my hair, or getting dressed/undressed

Difficulty chewing or swallowing

Starting to eat something (e.g. bite into a sandwich)

Difficulty opening a jar

Difficulty releasing a handshake or clenched fist

Difficulty using my hands (e.g. when writing)

Difficulty opening eyes (e.g. after sneezing or crying)

My muscle symptoms feel worse when I am stressed/anxious

Difficulty speaking clearly (e.g. speaking slowly, slurring or mumbling)

What do your muscle symptoms feel like? Tick all that apply

Numbness

Dull ache that is always there

Throbbing (like toothache)

Sudden tightness

I don't get much pain, I just get a physical symptom (e.g. locking)

Tingling

Do you have any family members who have the same problems with their muscles? Choose one

Yes | **No** | **Don't know**

How do you think the size of your muscles compares to other people's muscles? Choose one

I have always had big muscles. I don't need to exercise to build muscle

I have well-developed muscles because I exercise a lot and eat healthy

I am average build

I am less bulky than other people

When did your muscle symptoms develop? Choose one

I've always had them

When I was a child

When I was a teenager

In adulthood



The Checker was expanded to 12 questions (Figure 1), encompassing many aspects of myotonia symptomatology and NDM presentation within a simple framework. When difficulties in understanding the questionnaire were checked, errors were corrected in the French translation.



The Checker development also focused on search-engine optimisation strategies, to help direct people to the page when their initial queries are not myotonia specific. Language used in the scoping exercise interviews and searching has proved to be extremely useful for search-engine optimisation.

Conclusions



- **unMASC NDM Myotonia Symptom Checker** is a simple tool that helps people to characterise symptoms suggestive of myotonia, and encourage them to seek medical advice; its results aim to guide timely and appropriate specialist referral.
- Ultimately, it is hoped that the Checker may help to reduce the time taken for people with myotonia symptoms to reach appropriate medical support, regardless of their final diagnosis.
- The Checker does not replace a clinical consultation or provide a diagnosis: it fulfils an unmet need for symptom awareness building during the diagnostic odyssey:
 - o Delays in recognising ultra-rare diseases such as NDM negatively affect those long burdened by unexplained symptoms^{5,7}
- The Checker used a simple, rules-based algorithm, a basic validation exercise and a consideration of search-engine optimisation to attract users, thereby generating a cost-effective tool relatively quickly:
 - o This method is easily replicated, and may be considered by other groups who seek to build disease-awareness tools.

References

1. Bauskis A, et al. Orphanet J Rare Dis 17, 233 (2022).
2. Chazal PE, et al. Front Pharmacol. 2021 Oct 22;12:734601.
3. Díaz-Manera J, et al. EMJ 2021;6:37–46.
4. Trivedi JR, et al. Brain 2013;136:2189–200.
5. Vereb N, et al. J Neurol 2021;268:1708–20.
6. Díaz-Manera J, et al. Neuromuscul Disord. 2023;33:208-217.
7. Trip J, et al. J Neurol Neurosurg Psychiatry 2009;80:647–52.
8. Matthews E, et al. Brain 2010;133:9–22.

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