

RevEal the burdeN on daily life for myotonic dyStrophy patients due to myotoniA: key symptom-related findings of ENSA



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Introduction

- The myotonic dystrophies (DM1 and DM2) are a heterogeneous group of hereditary, rare diseases.¹
- A common and defining symptom of both DM1 and DM2 is myotonia, which has diverse adverse impacts on patient quality of life (QoL).^{1,2}
- Current DM management relies on off-label symptomatic treatment, which may include mexiletine for myotonia:
 - Mexiletine has been used as an effective antimyotonic treatment for several decades.³
- The ENSA (revEal the burdeN on daily life for myotonic dyStrophy patients due to myotoniA) survey investigated the impact of myotonia for people living with DM 1 or 2.

Objective

- To understand what support is needed to reduce burden and improve QoL for people with DM by assessing patient-reported experiences of symptoms and associated issues.

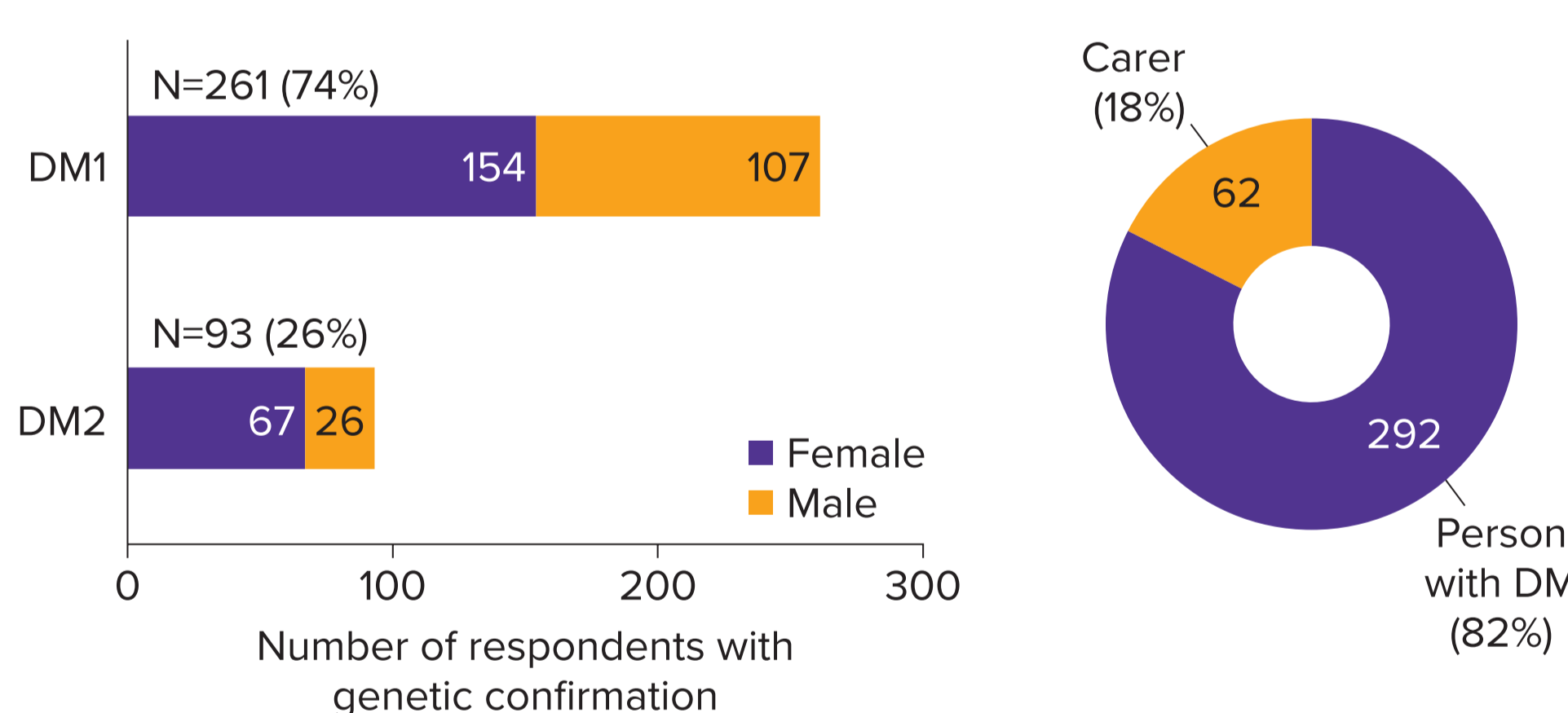
Methods

- ENSA was a global, anonymised, online survey (February–May 2023) of adults (≥18 years) living with DM1 or DM2, or their caregivers.
- Survey questions covered 32 topics: DM symptom onset/nature; diagnosis; myotonia frequency/location; disease management; impact on daily life.
- Respondents were asked if they had a genetic diagnosis confirmation; diagnoses were not verified by a medical professional.
- The patient-friendly questions (written to avoid medical terminology) provided multiple choice, free text or Likert scales (range 1–5: 1, Never; 5=Continuously [several times daily]).
- Descriptive analyses were undertaken (stratified by sex, country, and DM subtype).

Results

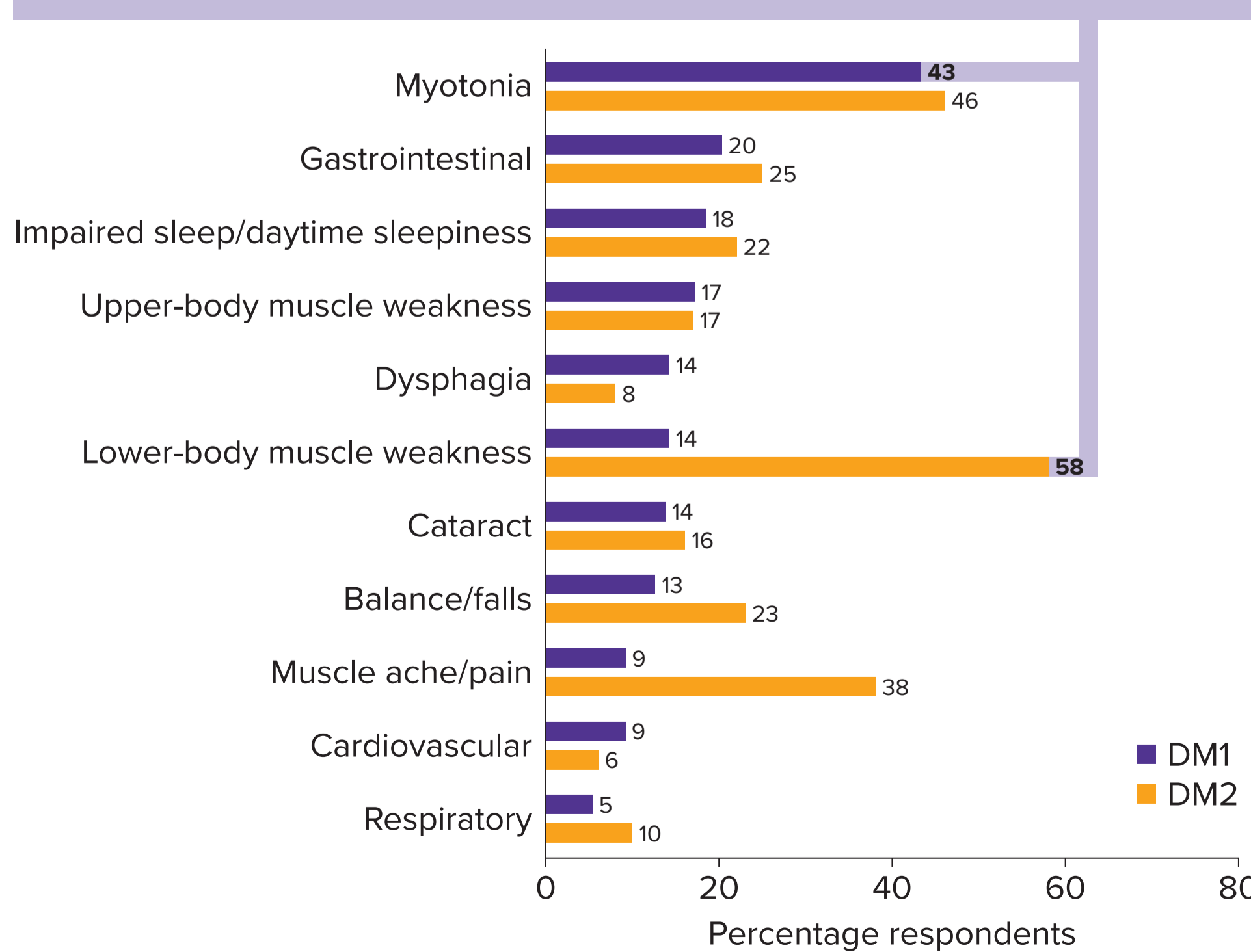
Demographics

386 respondents (aged 18–82 years, 61.7% female) completed the ENSA survey.
354 (92%) had, or were caring for, someone with, genetically confirmed DM.
 These results focus primarily on the genetically confirmed subset of respondents.



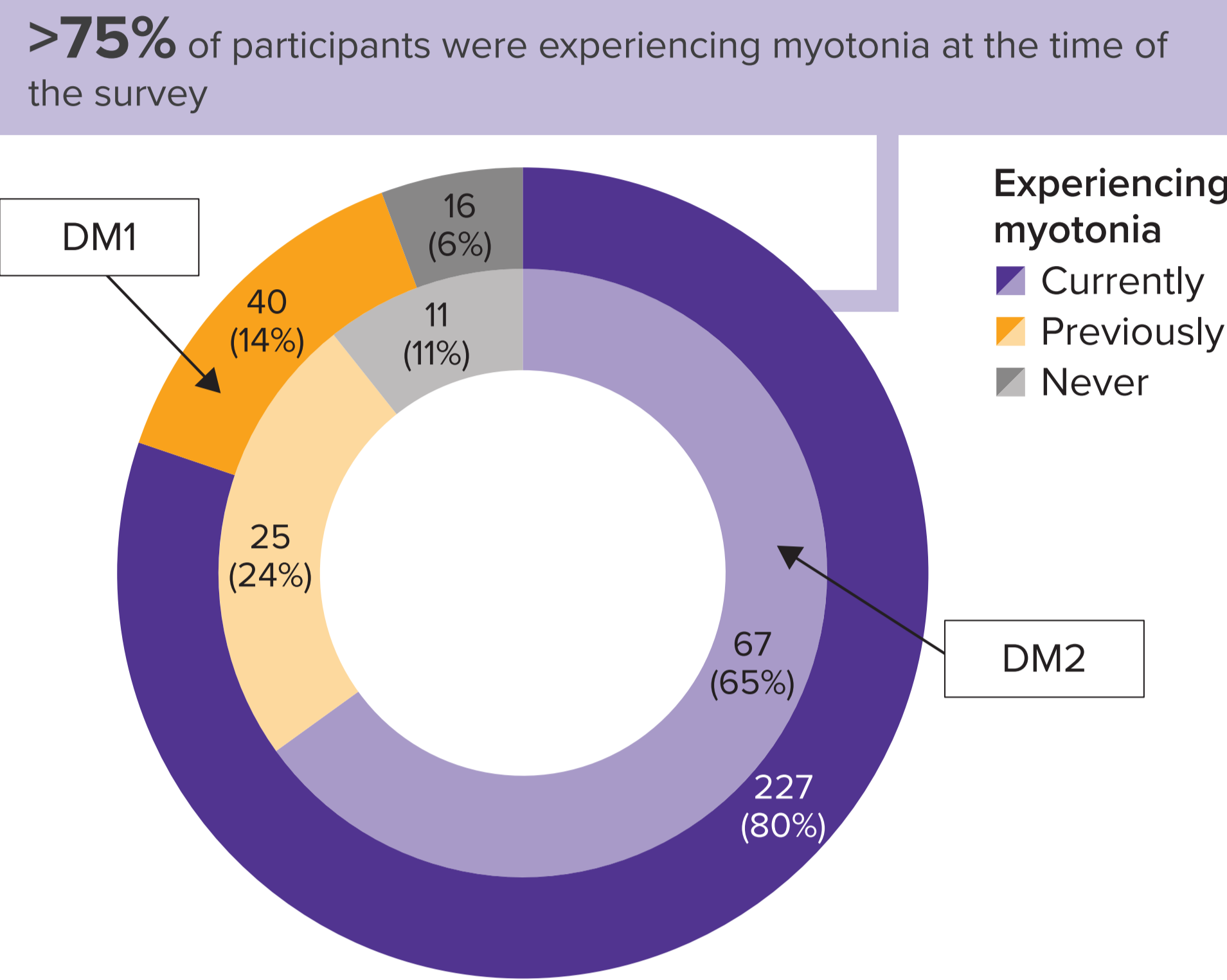
Symptoms prompting initial referral

For people with DM1, myotonia (43%), and for people with DM2, 'lower-body muscle weakness' (58%), were the most common reasons that respondents first visited a clinician about their symptoms.



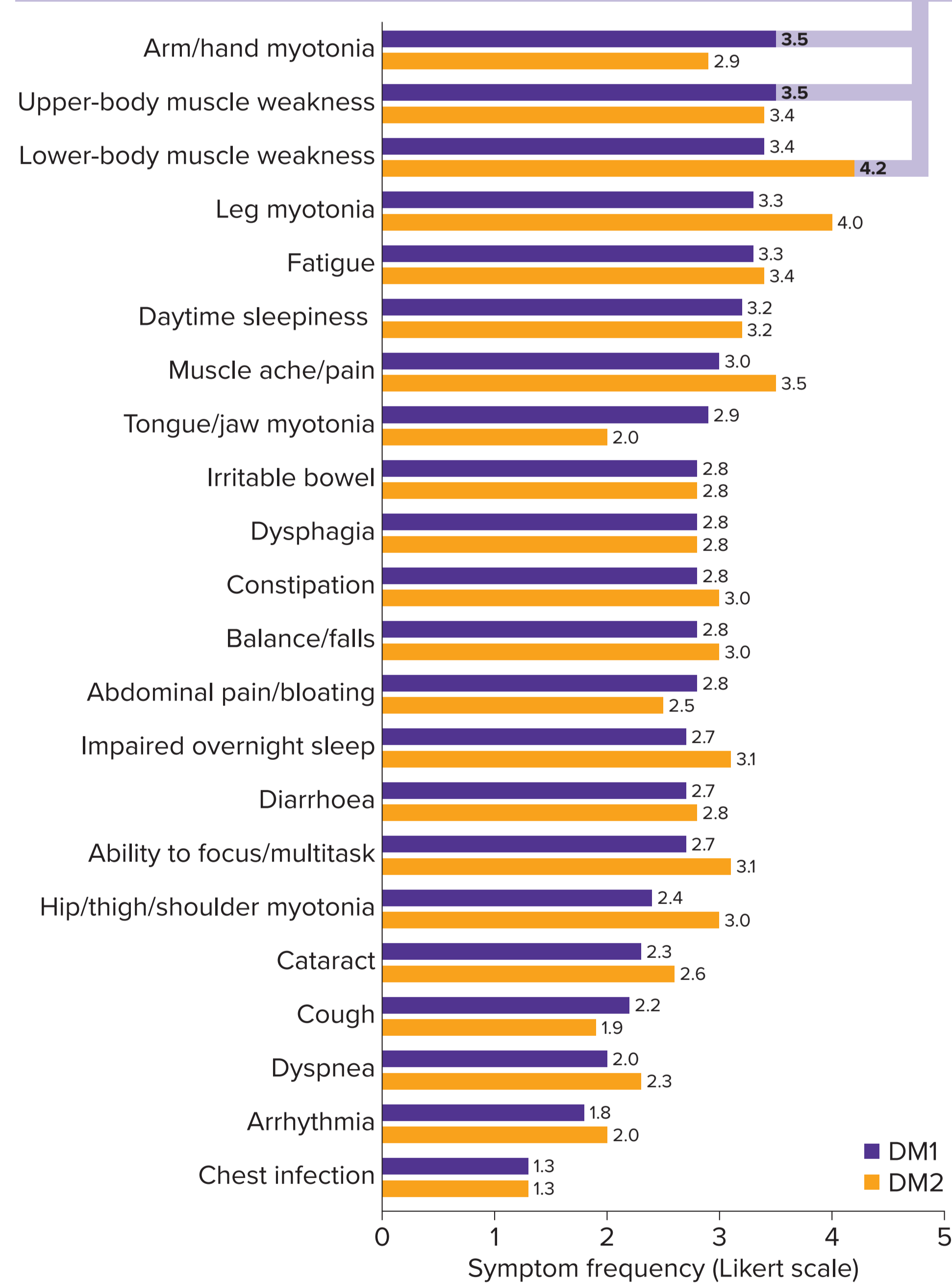
DM, Myotonic dystrophy.

Presence of myotonia at the time of the survey



Most frequent DM symptoms

For DM1, hand/arm myotonia and 'upper-body muscle weakness' (mean score: 3.5) and for DM2 'lower-body muscle weakness' (mean score: 4.2) were the most common current symptoms reported. In both the DM1 and DM2 groups, the average respondent experienced muscle ache/pain, daytime sleepiness, fatigue, leg myotonia, lower/upper body muscle weakness, at least weekly.



Impact of age on DM symptom frequency

Despite the expected increase in lower body weakness with rising age (mean Likert scale scores: 2.7–3.6 for DM1 and 3.1–4.4 for DM2), leg myotonia was also reported more frequently in the older age groups (see highlighted columns below) for both DM1 and DM2 (mean Likert scale scores: 2.9/3.7, <40 years vs >3.3/>4.0, ≥40 years). Therefore, despite results indicating expected levels of muscle atrophy, a corresponding reduction in leg myotonia with age was not reported by respondents. The frequency of experiencing other myotonias (tongue/jaw, arm/hand) appeared to decrease with age for both DM1 and DM2.

Mean scores per age group and for all members of DM1 or DM2 subgroups. 1 = Never; 2 = Sometimes (e.g., a few times a year or month); 3 = Regularly (weekly); 4 = Often (daily); 5 = Continuously (several times daily).

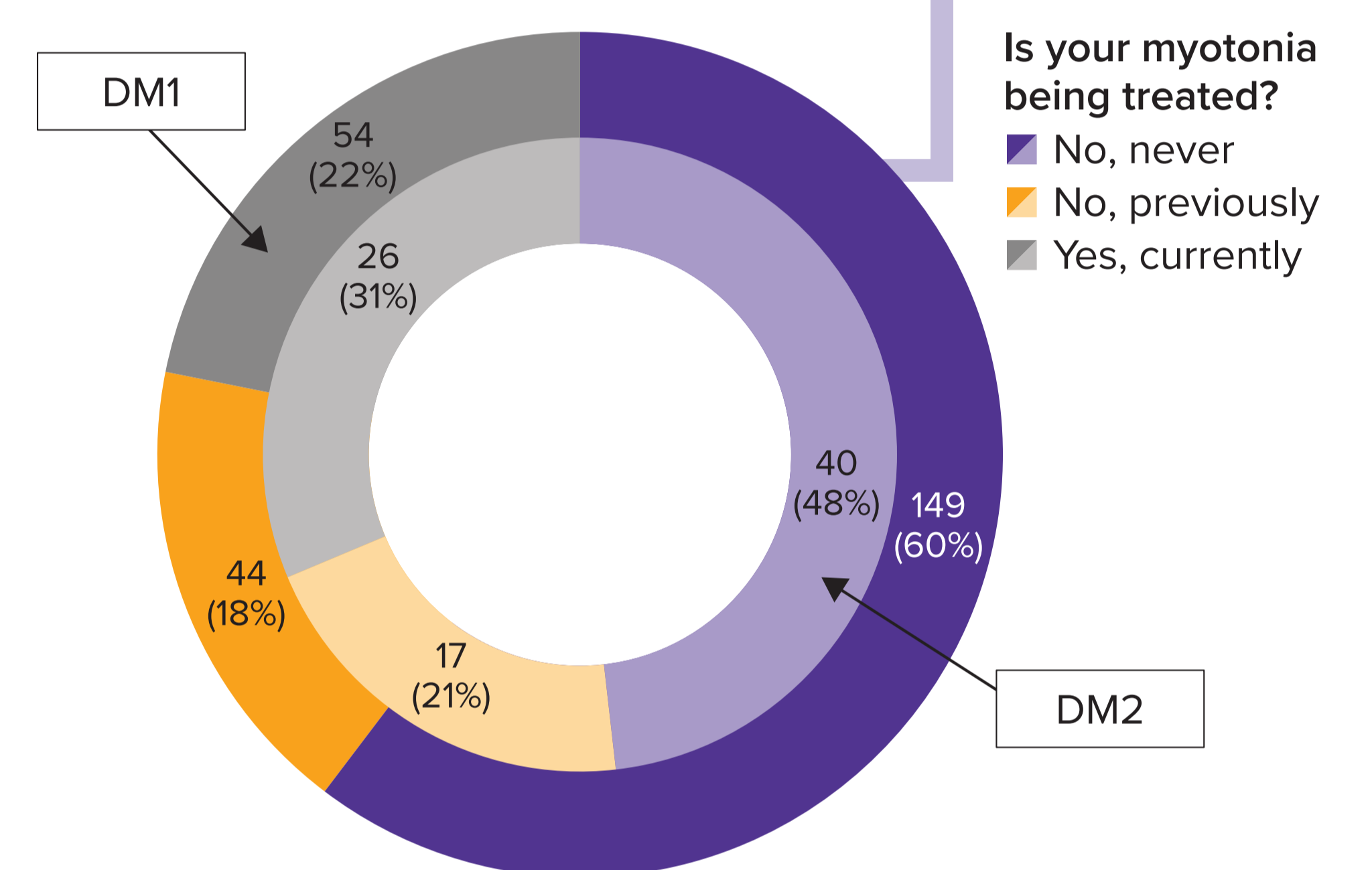
| Age | DM1 | | | | | | | | | | DM2 | | | | | | | | | | |
|-------------|--|--|------------------|----------------------------|----------------------------|------------------|-------------------------------|----------|--------------|-----------|-------------------------|-----------------|-----------|---------------|----------------------------|---------------------|------------|--------------------|-------------------------|---------|-------------------------------|
| | Myotonia in tongue and/or jaw or shoulders | Myotonia in hips, thighs arms and/or hands | Myotonia in legs | Upper body muscle weakness | Lower body muscle weakness | Muscle ache/pain | Balance problems and/or falls | Cataract | Constipation | Diarrhoea | Abdominal pain/bloating | Irritable bowel | Dysphagia | Chronic cough | Recurring chest infections | Shortness of breath | Arrhythmia | Daytime sleepiness | Impaired sleep at night | Fatigue | Ability to focus or multitask |
| <40 | 3.1 | 2.2 | 3.7 | 2.9 | 3.3 | 2.7 | 2.9 | 2.2 | 1.8 | 3.0 | 2.7 | 2.9 | 2.9 | 2.0 | 1.2 | 1.9 | 1.6 | 3.2 | 2.6 | 3.3 | 2.7 |
| >40 and <60 | 3.0 | 2.6 | 3.5 | 3.6 | 3.7 | 3.7 | 3.2 | 3.0 | 2.5 | 2.7 | 2.8 | 2.7 | 2.9 | 2.4 | 1.4 | 2.1 | 1.8 | 3.1 | 2.3 | 3.4 | 2.9 |
| ≥60 | 2.5 | 2.4 | 2.9 | 3.3 | 3.7 | 3.6 | 2.8 | 3.4 | 3.0 | 2.7 | 2.5 | 2.6 | 2.5 | 2.7 | 2.3 | 1.8 | 2.1 | 2.0 | 3.3 | 3.0 | 3.2 |
| All | 2.9 | 2.4 | 3.5 | 3.3 | 3.5 | 3.4 | 3.0 | 2.8 | 2.3 | 2.8 | 2.7 | 2.8 | 2.8 | 2.2 | 1.3 | 2.0 | 1.8 | 3.2 | 2.7 | 3.3 | 2.7 |
| <40 | 2.0 | 3.1 | 3.5 | 3.7 | 3.0 | 3.3 | 2.7 | 2.7 | 2.0 | 2.3 | 2.7 | 2.7 | 2.7 | 1.1 | 1.6 | 2.1 | 1.9 | 3.1 | 2.6 | 3.7 | 3.3 |
| >40 and <60 | 2.2 | 3.3 | 3.2 | 4.1 | 3.5 | 4.2 | 3.7 | 2.9 | 2.7 | 3.0 | 2.8 | 2.7 | 2.9 | 1.9 | 1.4 | 2.5 | 2.0 | 3.2 | 3.3 | 3.4 | 3.2 |
| ≥60 | 1.6 | 2.3 | 2.4 | 4.0 | 3.2 | 4.4 | 3.4 | 3.4 | 2.8 | 3.0 | 2.8 | 2.3 | 2.8 | 2.7 | 1.2 | 2.1 | 1.9 | 3.2 | 2.8 | 3.5 | 2.9 |
| All | 2.0 | 3.0 | 2.9 | 4.0 | 3.4 | 4.2 | 3.5 | 3.0 | 2.6 | 3.0 | 2.8 | 2.5 | 2.8 | 1.9 | 1.3 | 2.3 | 2.0 | 3.2 | 3.1 | 3.4 | 3.1 |

Conclusions

- The ENSA survey indicated that people self-reporting a genetic diagnosis of DM consider myotonia to be a frequent symptom that somewhat interferes with daily living/QoL.
- Myotonia is particularly burdensome; research indicates it is often associated with stiffness and pain, especially in people with DM2.⁴
- ENSA aims to improve understanding of DM symptoms, particularly myotonia, and to emphasise the ongoing need for effective treatment, ultimately to improve daily life for those living with DM.

Most respondents were not currently receiving myotonia treatment

78% of respondents with DM1 and 69% with DM2 who answered this question were not currently receiving myotonia treatment.



Symptoms that respondents most wanted to improve

Muscle weakness was the symptom that the majority of respondents with DM1 who were not currently on myotonia treatment most wanted to improve (n=134), followed by fatigue (n=111) and myotonia (n=104). In respondents within the DM2 cohort who were not currently on myotonia treatment, muscle weakness (n=48), muscle ache/pain (n=30) and fatigue (n=29) were the symptoms that people most wanted to improve.

References

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Disclosures

Zozulya-Weidenfeller is employed by Lupin. Other authors received honoraria from Lupin Neurosciences as consultants during the ENSA creation.

