RevEal the burdeN on daily life for myotonic dyStrophy patients due to myotoniA: preliminary results of the ENSA survey

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Introduction/Objectives

- Myotonic dystrophy (DM) types 1 and 2 (DM1 and DM2) have many burdensome symptoms that negatively affect quality of life.[1–5]
- Although myotonia is a well-recognized symptom of DM,[4] its specific contribution to the daily burden on patients' lives is unclear.
- ENSA was a patient-reported, international, online survey to investigate the impact of myotonia on people with DM.
- ENSA also sought to uncover the first symptoms of DM that prompted a clinical consultation.
- This is a preliminary presentation of key data.

Methods

Study population

- Adults (≥18 years) with DM (or caregivers on their behalf) participated in ENSA, which was publicized via an outreach campaign:
  - ENSA was open globally between February and May 2023; North America, Europe and the UK were target regions.
- Participants were asked if they had undergone genetic testing to confirm their DM diagnosis, although genetic confirmation was not mandatory for inclusion.
- People did not require a history of myotonia in order to participate.

Survey structure

- Anonymized online survey that explored DM symptom onset; symptoms that prompted first DM-related consultation; time to medical consultation/diagnosis; myotonia frequency, severity and management.
- Symptom impact was measured on a 5-point scale (1, never/not at all; 5, continuously); treatment satisfaction was measured on a 3-point scale (1, satisfied, 3; dissatisfied).
- Myotonia management/treatment history was explored broadly:
  - ENSA did not investigate specific treatments; strategies for myotonia management differ considerably between countries.

Analysis

- Data presented as: n, %, median, median (range).

Results

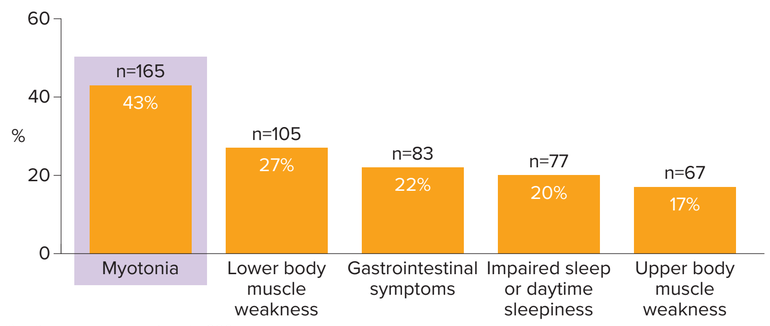
Demographics

- ENSA was completed by 386 people in 23 countries; most respondents were in the USA (n=100; 26%).
- N=238 respondents (62%) were women.
- Median (range) age of respondents, 48 (18–82) years.
- N=70 (18%) of surveys were completed by caregivers, on the patient's behalf.
- N=283 (73%) of respondents had DM1:[6]
  - n=140 (50%) adult-onset; n=54 (19%) juvenile onset; n=51 (18%) late-onset; n=12 (4%) infantile onset; n=15 (5%) congenital DM1; n=11 (4%) onset unknown.
- N=103 (27%) respondents had DM2 or proximal myotonic myopathy.
- N=354 (92%) respondents stated their DM diagnosis was confirmed by genetic testing.

Common symptoms

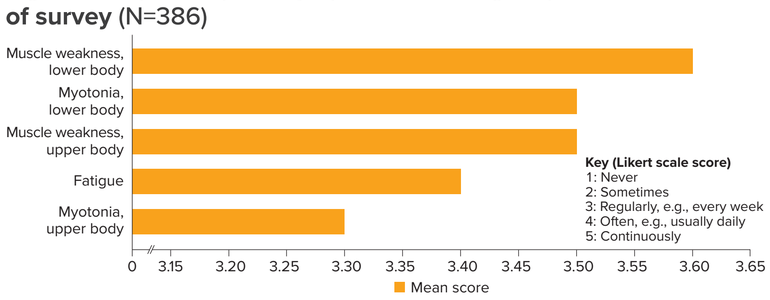
- Myotonia, muscle weakness (lower or upper body), gastro-intestinal symptoms and impaired sleep often prompted the first clinical consultation about DM (Figure 1).
- Muscle weakness and myotonia were the most common symptoms to affect patients at the time of the survey (Figure 2).

Figure 1: Myotonia was the most common symptom* to prompt a first clinical consultation about DM



*Multiple answers possible. N=386.

Figure 2: Top 5 frequent symptoms affecting respondents **at time of survey** (N=386)



Top 5 symptoms that respondents **wanted to improve** (N=386):

1. Muscle weakness (anywhere in the body)
2. Fatigue (extreme tiredness and inability to function due to lack of energy)
3. Myotonia (anywhere in the body)
4. Balance problems and/or falls
5. Muscle ache/pain

Myotonia presence

- 359/386 (93%) of patients had a current or previous history of myotonia. Figures 3 and 4 present key characteristics of myotonia frequency and location.

Figure 3: Myotonia history: Experiencing currently; Experienced in the past; Never experienced

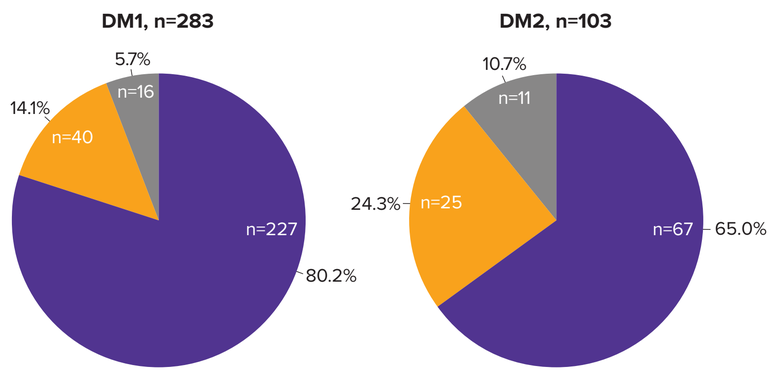
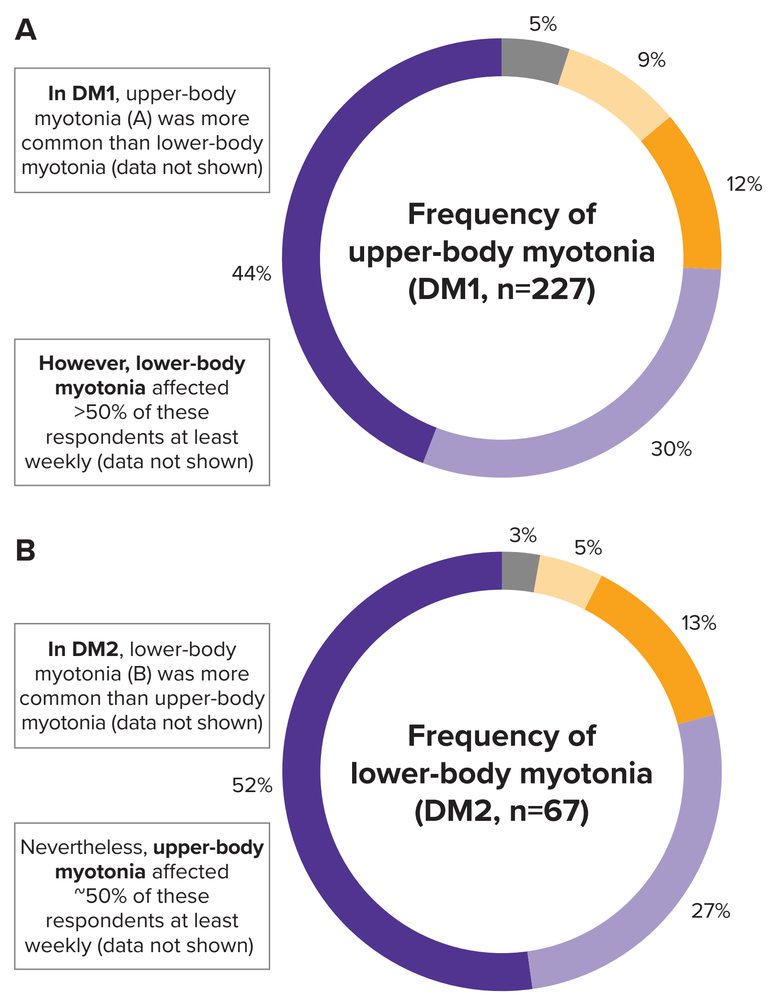


Figure 4: Frequency of (A) upper-body myotonia, DM1; (B) lower-body myotonia, DM2: Never/not at all; Sometimes (per month/year); Regularly (weekly); Often (usually daily); Continuously (several times daily)



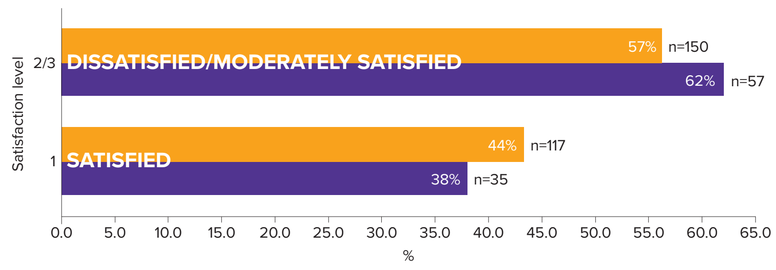
DM diagnosis and treatment satisfaction

Median age when patients experienced their first symptoms associated with DM
26 years

Median age for diagnosis
30 years for DM1
45 years for DM2

- Satisfaction with myotonia management was relatively poor across the cohort (Figure 5):

Figure 5: 207/359 (58%) respondents were either dissatisfied or only moderately satisfied with myotonia management:
DM1, n=267 DM2, n=92



- 207/359 (58%) of respondents who experienced myotonia had never taken drug treatment for symptomatic relief (Table 1):
  - Physicians not offering drug treatment was the most common reason, followed by participants' lack of awareness of myotonia treatments (Figure 6).
- Myotonia management is complex; not every efficacious treatment is suitable for every person with DM.

Table 1: Few respondents with myotonia said that they had received any drug treatment for this symptom

Respondents reporting myotonia, N=359 (100%)	DM1, n=267 (74%)	DM2, n=92 (26%)
Currently on prescribed myotonia treatment	n=61 (23%)	n=29 (32%)
Myotonia treatment prescribed previously but not currently	n=45 (17%)	n=17 (19%)
Never taken a prescribed treatment for myotonia	n=161 (60%)	n=46 (50%)

Figure 6: Most common reasons for not receiving drug treatment for myotonia (NB: more than one answer permitted; size of text reflects number of responses)

Conclusions

- ENSA survey findings show that myotonia is a debilitating symptom experienced by most people with DM
- Diagnostic delays ≥7 years affect people with DM1 and especially DM2;[4] such delays are typical for rare diseases, including myotonic disorders[5]
- Despite myotonia being one of the most common symptoms to trigger a clinical consultation (and frequently experienced), it is rarely treated
- Further planned analyses of ENSA data include stratifications by age, sex and genetic predisposition
- **ENSA patient-reported survey findings indicate that myotonia is a major symptom of DM and one of the top 5 that they want to improve; the burden – and potential treatment – of myotonia are under-recognized, both by physicians and by people affected by DM**

References

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