Poster EPO-392

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

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Introduction

- Myotonia is a symptom of myotonic dystrophy (DM) types 1 and 2.
- Myotonia can be a debilitating symptom that affects daily living, placing a substantial negative burden on quality of life (QoL) in people with non-dystrophic myotonia.[1]
- Although the impact of DM on patients' QoL has been evaluated,[2–4] the specific contribution of myotonia is unclear.

Objective

- The ENSA survey specifically assessed the impact of myotonia on QoL in patients with DM.

Methods

Development

- ENSA was developed following a number of online meetings between a multidisciplinary international group of experts involved in the care and management of patients with DM:
  - These included specialists in neuromuscular disorder management, patient advocacy, neurological research, statistics, and treatment development.

Study population characteristics

- Between March and May 2023, patients aged ≥18 years, with a confirmed diagnosis of DM1/DM2 (or their caregivers), were invited to complete an anonymized online survey, promoted through social media campaigns.
- The survey was open to all patients globally, but the predominant target regions were Europe, the UK, and North America.

ENSA survey structure

- ENSA explored patients' descriptions of DM symptom onset; time to medical consultation and diagnosis; past/present daily life with myotonia; impact of DM (and of myotonia) on work/study; nature/frequency/location of myotonia; levels of muscle weakness, fatigue, daytime sleepiness, gastrointestinal, and cardiorespiratory symptoms; disease management; and treatment history.
- ENSA could not ask about specific treatment experiences because there is no universal access to all treatments listed.

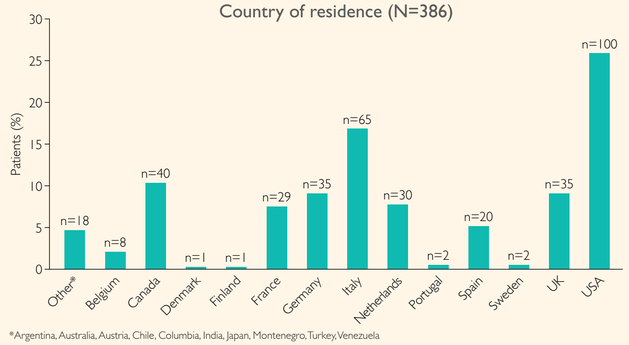
Results (Population characteristics)

- In total, 386 people completed the ENSA survey online. Initial data on some key variables (including demographics) are illustrated below.

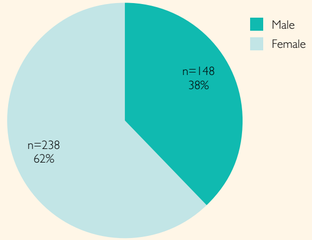
316 respondents (82%) were people diagnosed with DM

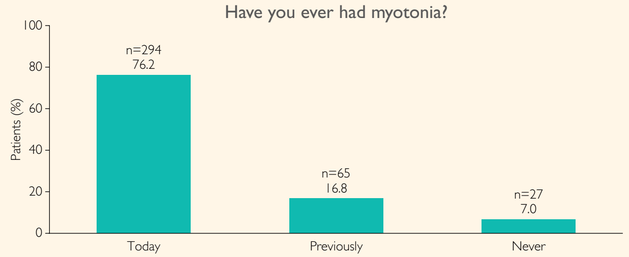
70 (18%) were carers responding on behalf of patients

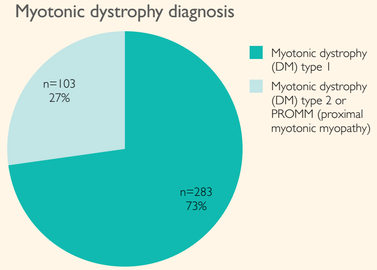
Mean (range) age of respondents: 48.3 (18–82) years

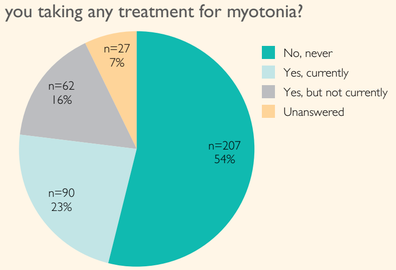


*Argentina, Australia, Austria, Chile, Columbia, India, Japan, Montenegro, Turkey, Venezuela









- Full study results of ENSA will be available in late 2023.

Conclusions

- ENSA is an anonymized survey exploring patient-reported descriptions of living with DM1 or DM2.
- 386 people completed the survey, which will be reported late 2023.
  - ENSA had a good uptake from patients with DM who have experienced myotonia currently or previously.
- ENSA will provide qualitative and quantitative information on the impact of myotonia on daily life and well-being of people with DM.
- Findings from ENSA aim to inform the need for appropriate myotonia management in patients with DM.
- Findings of ENSA aim to increase understanding of myotonia, and to support future clinical-trial outcome measures in DM studies.

References

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Disclosures

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