

Patient reported satisfaction in myotonic dystrophy management: ENSA international survey suggests benefits of Italian routine practice



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Introduction

- The myotonic dystrophies (DM1 and DM2) are hereditary rare diseases exhibiting heterogeneous symptoms.¹
- People with DM1 and DM2 often experience myotonia, which adversely impacts their quality of life (QoL).^{1,2}
- The management of DM currently relies on off-label symptomatic treatment, which may include mexiletine for myotonia:
 - Mexiletine has long been recognised as an effective antimyotonic treatment.³
- There is a paucity of patient-reported data in DM, particularly relating to possible country-specific differences in patient experiences.
- ENSA (revEal the burdeN on daily life for myotonic dyStrophy patients due to myotoniA) evaluated the impact of myotonia and its management on adults living with DM1 and DM2.

Objective

- This ENSA sub-analysis focuses on patient-reported outcomes regarding DM management; differences in experience reported by respondents treated at Italian DM centres, compared with those treated in other participating countries.

Methods

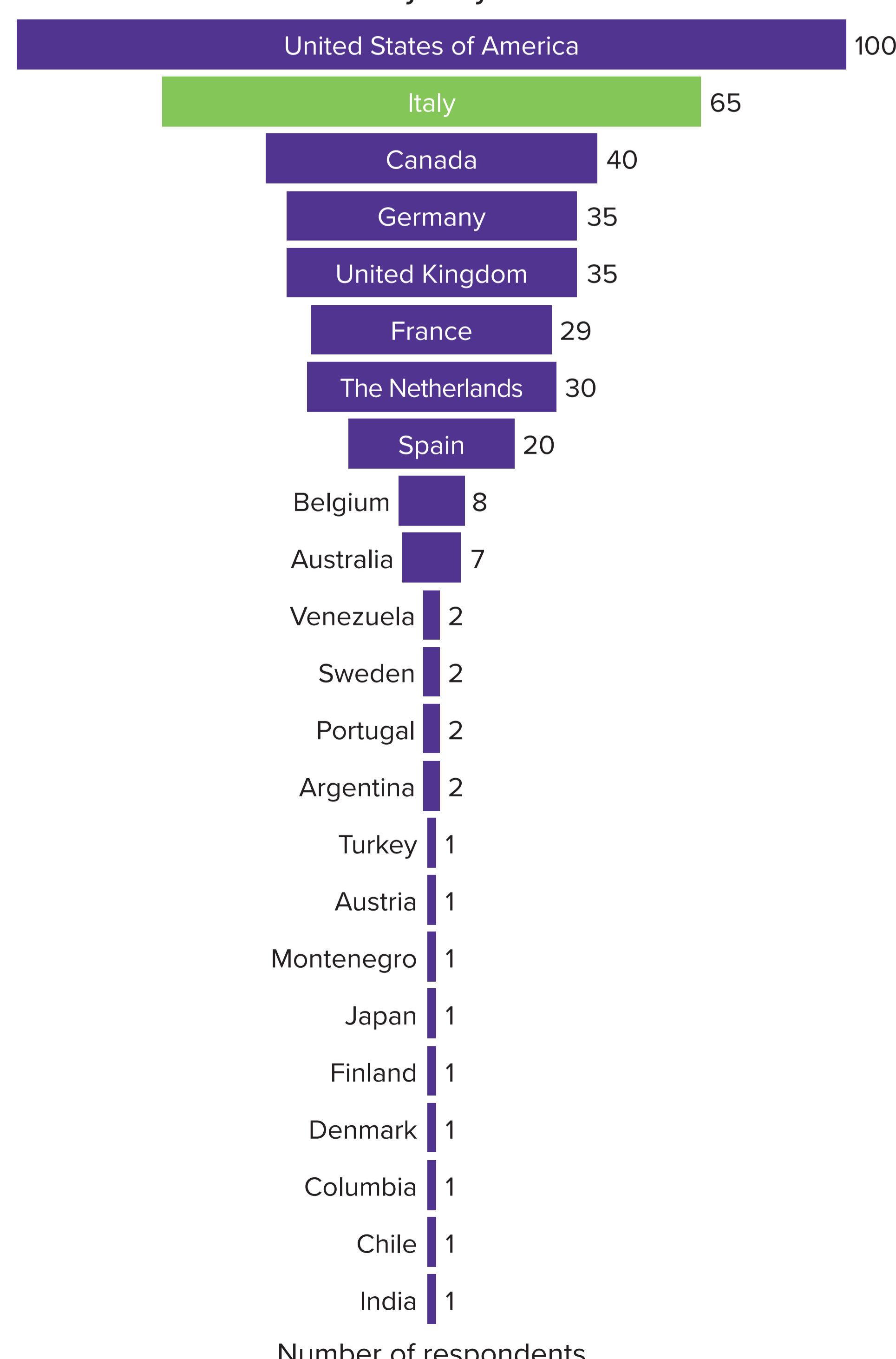
- ENSA was an online, international, patient-reported, anonymised survey, open February–May 2023, offered to people living with DM (types 1 or 2) or caregivers on their behalf.
- ENSA featured 32 questions on daily living with DM. Questions were structured in lay language, using free-text, multiple-choice, and Likert-scale responses, as appropriate.
- Respondents were not asked to name specific myotonia treatments they received.
- This narrative analysis presents clinical management/treatment satisfaction scores. Management satisfaction levels and burden were ranked on Likert scales (1, no improvement; 4 highest satisfaction) and presented as mean values.
- The statistical analysis was descriptive.

Results

Population

386 adults living with DM in 23 countries completed ENSA. 354 (92%) had, or cared for someone with, genetically-confirmed DM.

What country do you live in?



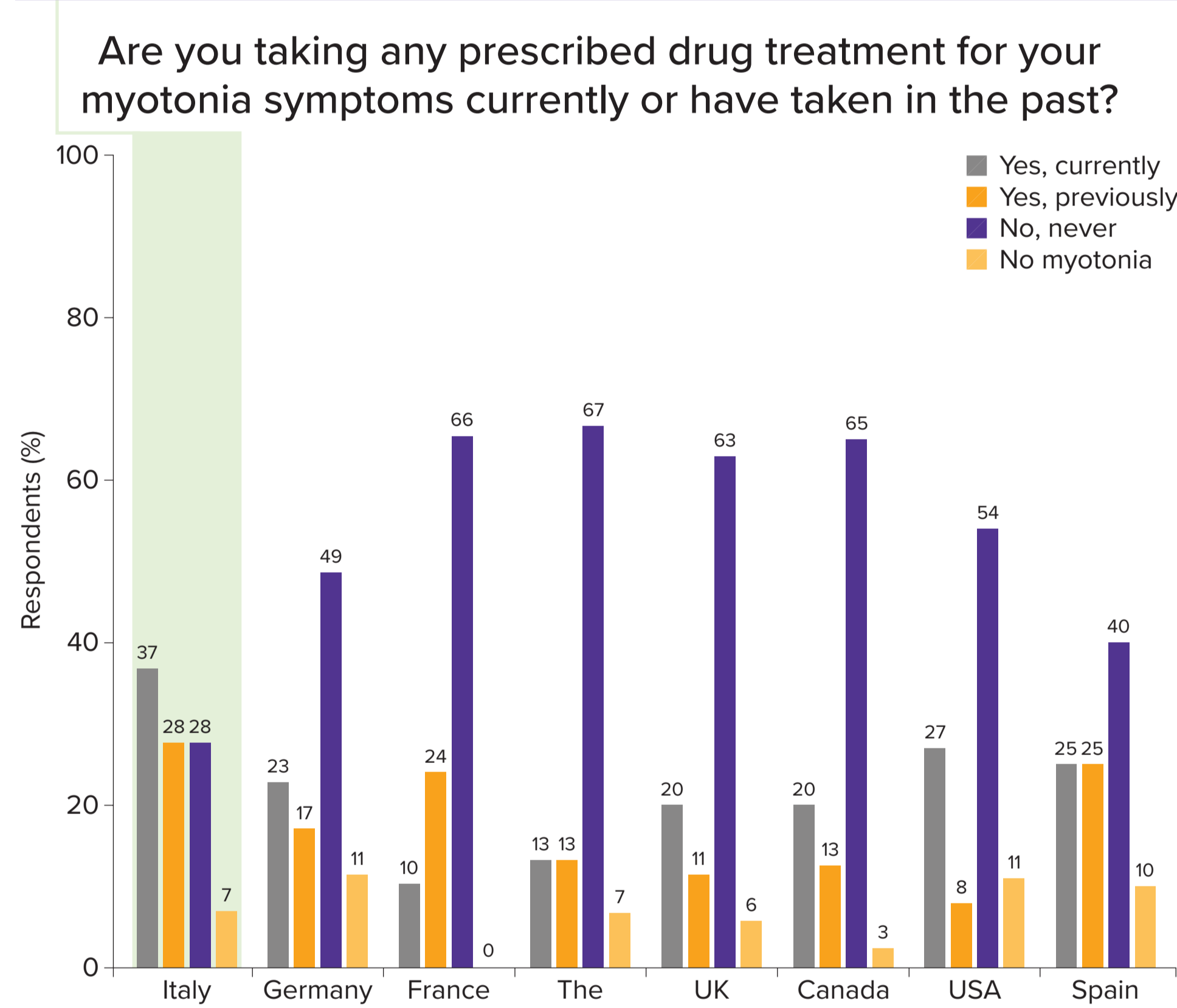
Data from the overall population (N=386)

Conclusions

- This snapshot of patient-reported satisfaction relating to myotonia management suggests potential differences between satisfaction levels in Italy and elsewhere.
- Italy has a proactive system for managing myotonia, with a high level of therapeutic drug use that may not be available globally.
- One potential limitation of the survey is that the 4-point Likert scale for myotonia symptom improvement and activity of daily life may have been too subjective.
 - Therefore, respondents may have judged 'improved significantly', 'improved moderately', and 'improved occasionally' differently.
- Although there may be recall bias, quality-of-life improvements when on treatment suggesting that the Italian approach, with higher levels of therapeutic management for myotonia, may provide benefits in DM.

Myotonia treatment experience

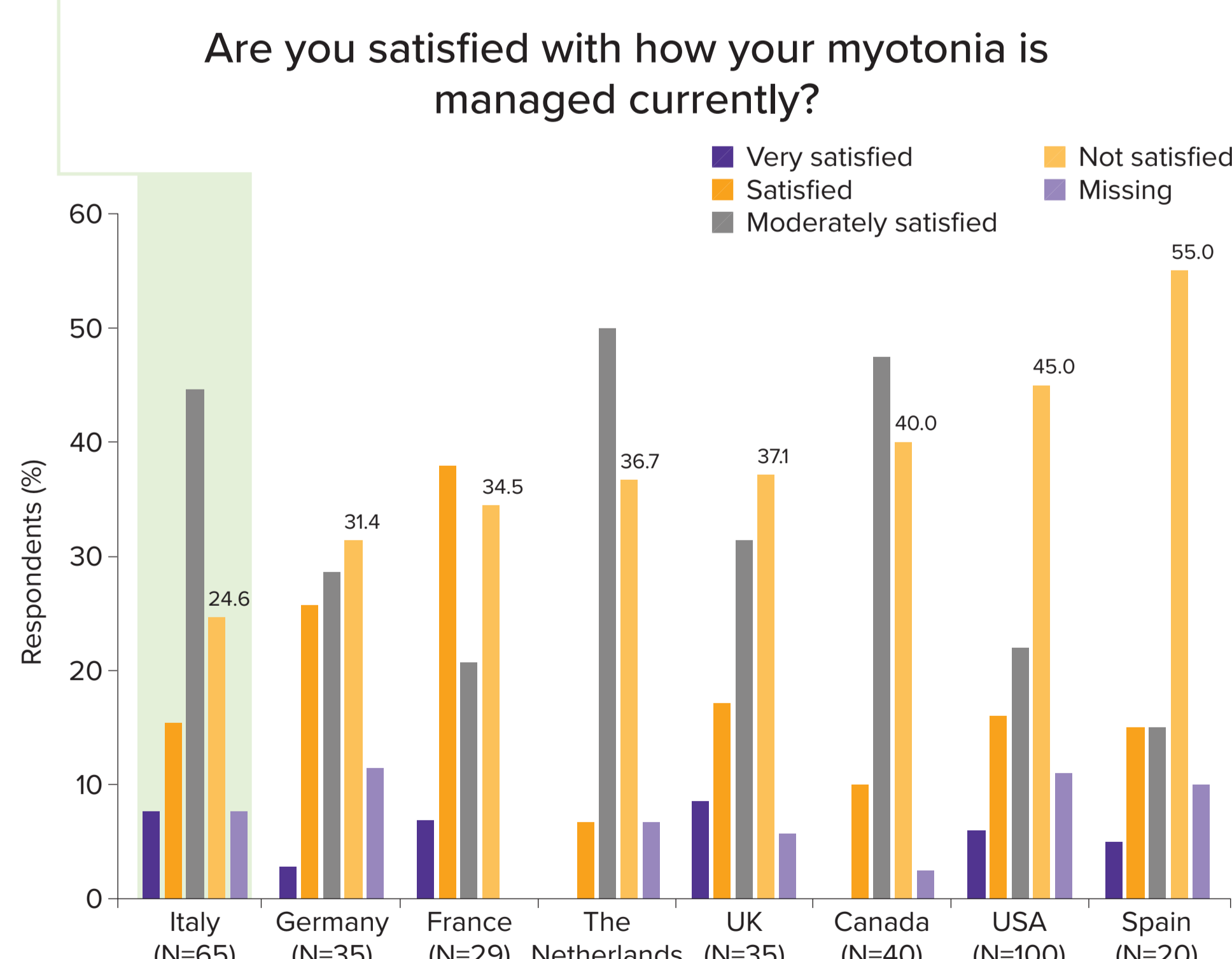
Respondents from Italy (N=65) reported the highest rate of current myotonia treatment (37%) and the lowest rate of never having received myotonia treatment (28%) compared to other countries with ≥20 respondents.



Data from 8 countries with highest number of respondents

Treatment satisfaction

Respondents from Italy reported the lowest levels of treatment dissatisfaction (24.6%) among the top 8 countries by respondent number.



Data from 8 countries with highest number of respondents

References

1. Landfeldt E, et al. J Neurol 2019;266:998–1006; 2. Heatwole C, et al. Neurology 2015;85:2136–46; 3. Díaz-Manera J, et al. Neuromuscul Disord 2023;33:208–217.

Acknowledgments

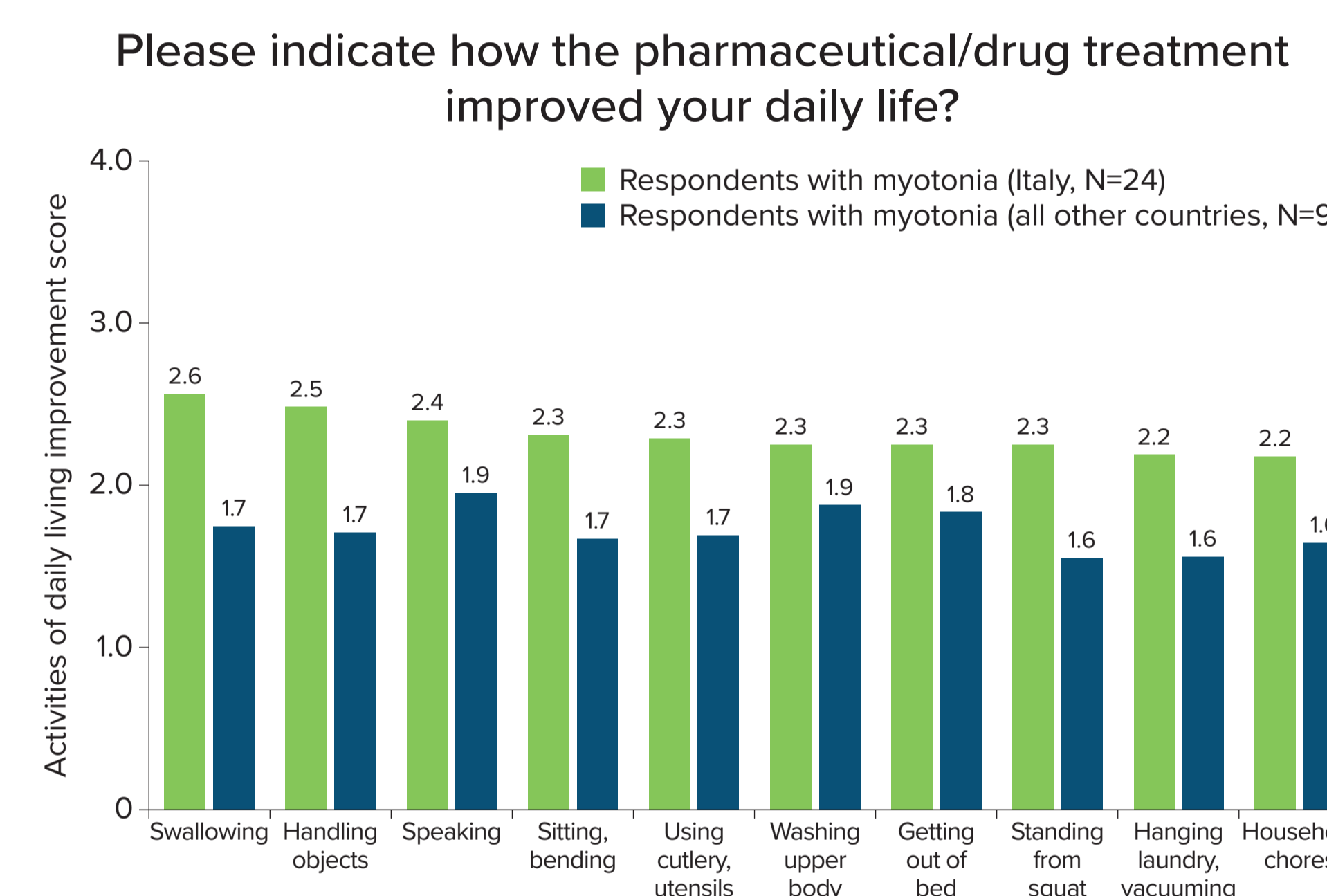
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Disclosures

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

Activities of daily living

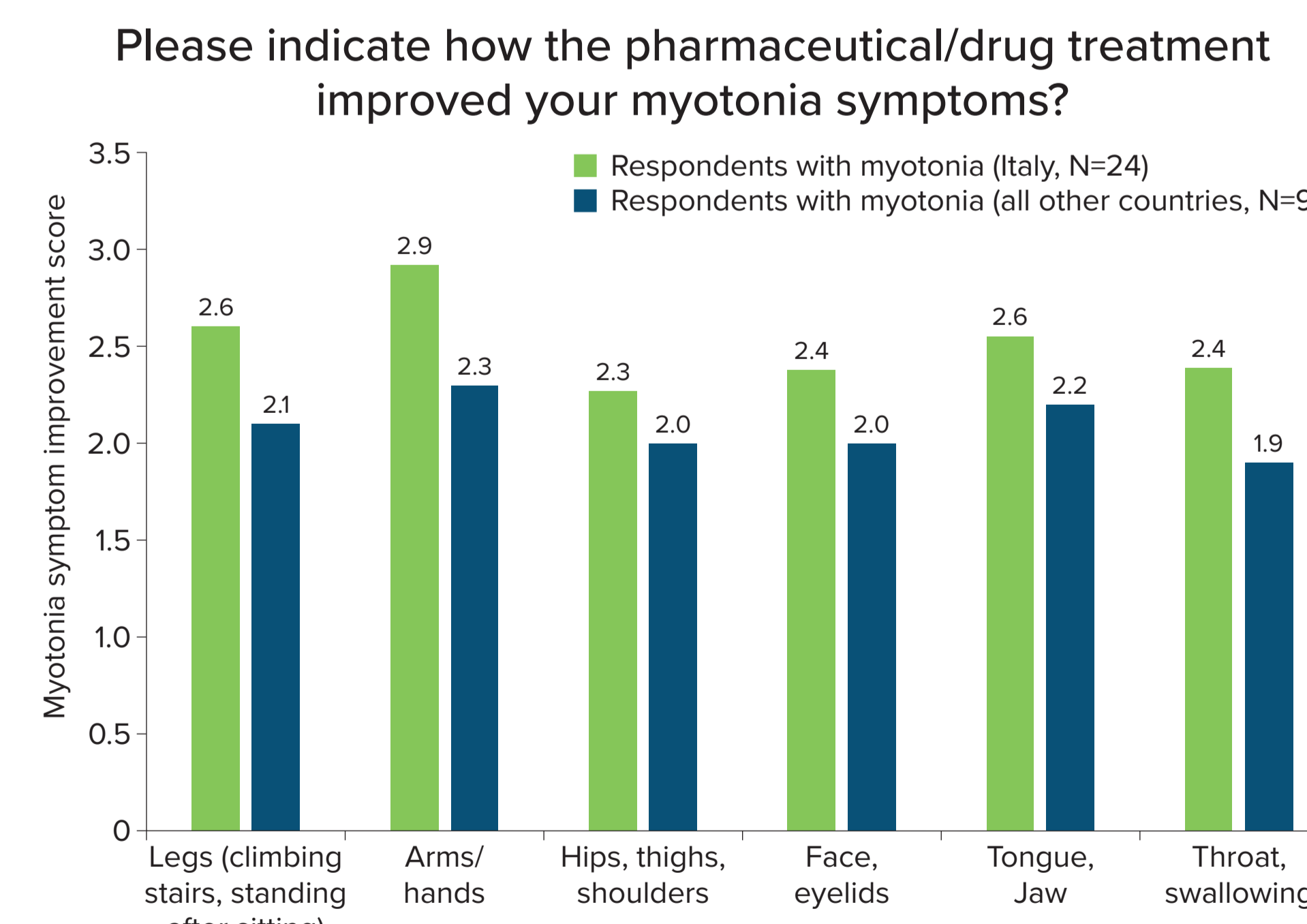
Respondents from Italy reported numerically higher treatment-related improvements in their ability to undertake activities of daily living, compared to respondents from other countries. The top 10 activities with the greatest reported improvements by Italian respondents are presented. Other activities that Italian respondents indicated improved at least occasionally included carrying a heavy object, walking without aid, brushing hair/teeth, ability to exercise, bodily hygiene, dressing, walking up 3 steps and standing for a long period (data not shown).



Improvement in activities of daily living due to myotonia treatment was scored on a Likert scale: 4=improved significantly; 3=improved moderately; 2=improved occasionally; 1=did not improve at all. 'Not-applicable' responses were excluded.

Myotonia symptom improvement scores

Generally, respondents with myotonia reported that treatment occasionally improved their myotonia symptoms. Respondents from Italy with myotonia reported numerically higher treatment-related improvements in myotonia symptoms compared with respondents from other countries.



Improvement in myotonia symptoms was scored on a Likert scale: 4=improved significantly; 3=improved moderately; 2=improved occasionally; 1=did not improve at all. 'Not-applicable' responses were excluded.

