



# Myotonic Dystrophy Family Registry: a patient report registry collecting demographic, symptomatic and quality of life measures

MYOTONIC  
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## Introduction

The Myotonic Dystrophy Family Registry (MDFR) is a patient report registry that was established in March of 2013 to collect the core DM data set identified at the 2009 meeting in Naarden on patient registries<sup>1</sup>. The Registry now contains data from 1461 individuals, with complete survey data from 1269 individuals. This self-report registry allows patients to enter and update their own data directly, allowing for fast accrual, but with the caveat that the data has not been reviewed by a clinician.

The Registry is divided into three sections: Demographics, Symptoms and Quality of Life:

- The **Demographic** section includes 49 fields and covers such items as name, address, age, gender, willingness to be contacted for trial opportunities, complete contact information and relationship to the registrant.
- The **Symptom** section includes 29 sections with sub-questions for each. The major items covered include functional, cardiac, respiratory, and GI status as well as information on myotonia, fatigue, cataracts, pain, psychiatric/behavioral issues and tumors.
- The **Quality of Life** section includes three parts, which explore the status of insurance coverage, employment, and disability impact on day to day living.

## Methods

Participants are recruited through the MDF website, email communication from the MDF, referrals from health professionals and word-of-mouth. For this report, the frequency of selected data elements reported to the Registry as of June 4, 2015 was compiled. Where selected data has been used to support individual figures, the parameters for those data are identified with the figure.

## Results and Discussion

Figure 1. Registry Growth

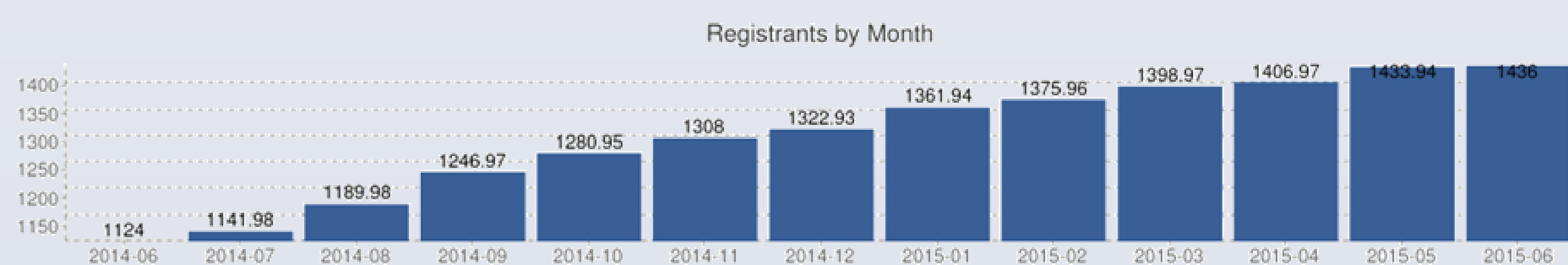


Figure 1. Since its launch, the number of registrants in the MDFR has been steadily increasing.

Figure 2. Geographic Distribution of Registrants

The majority of registrants reside in the United States, Canada, the United Kingdom and Australia, but 33 other nationalities are represented as well.

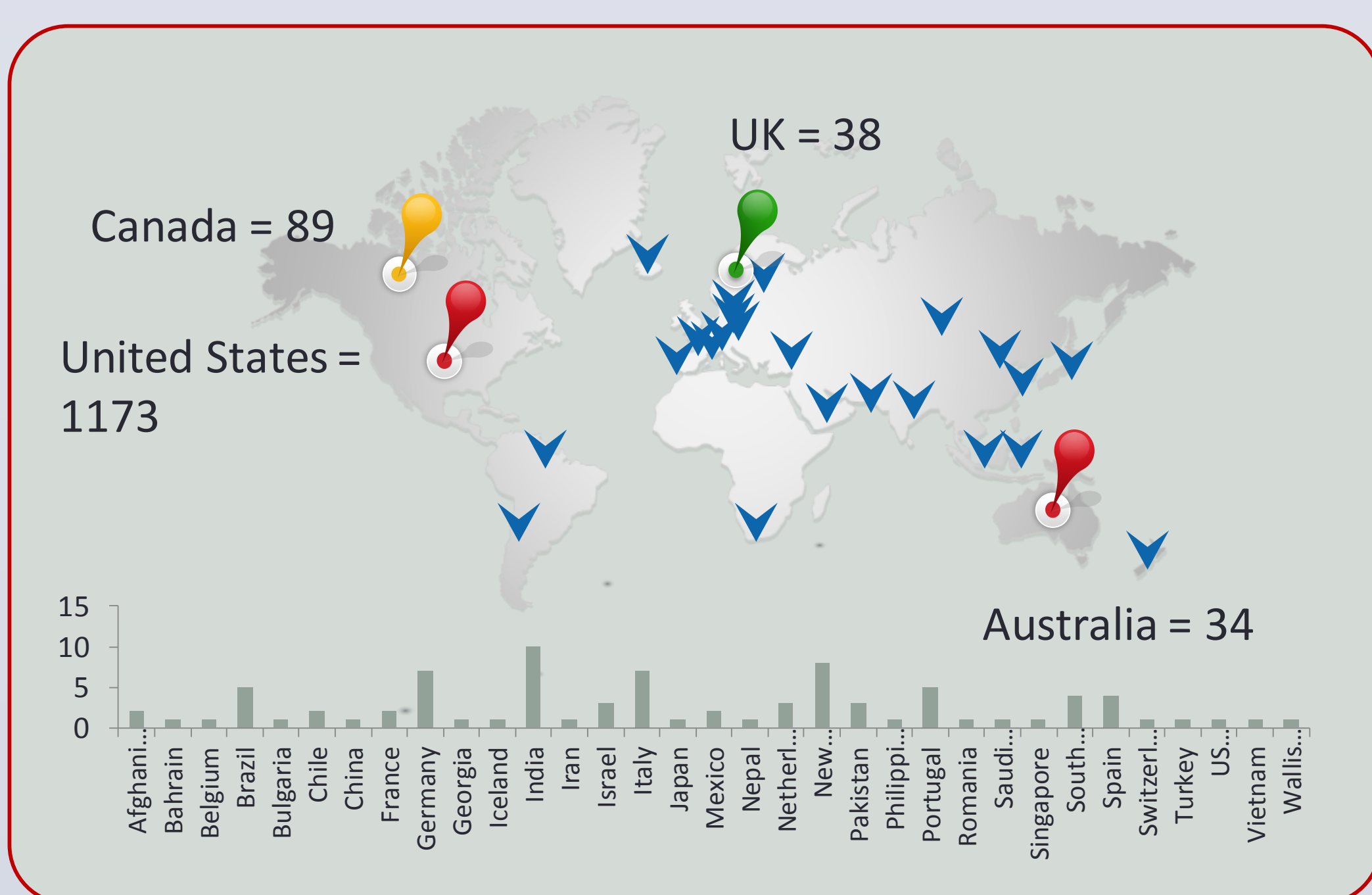
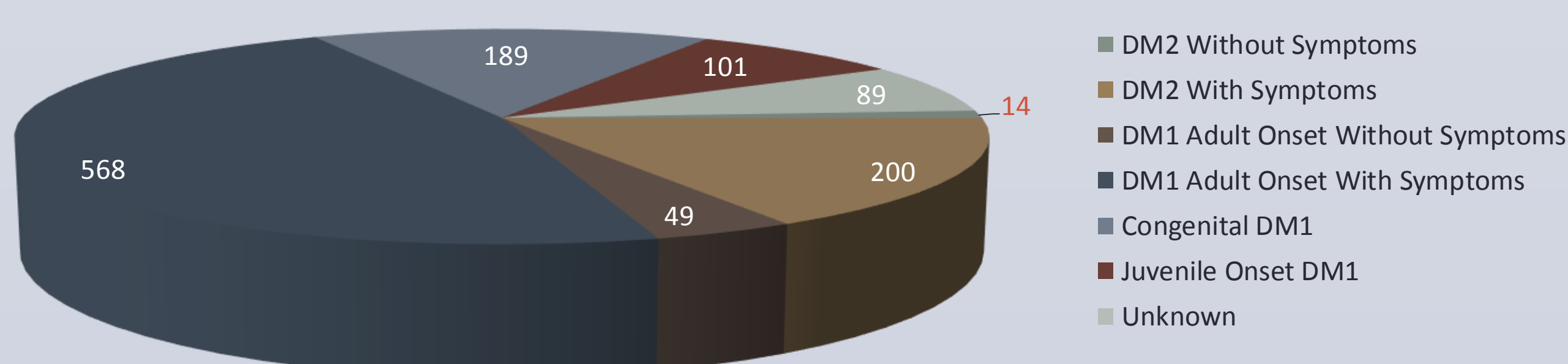
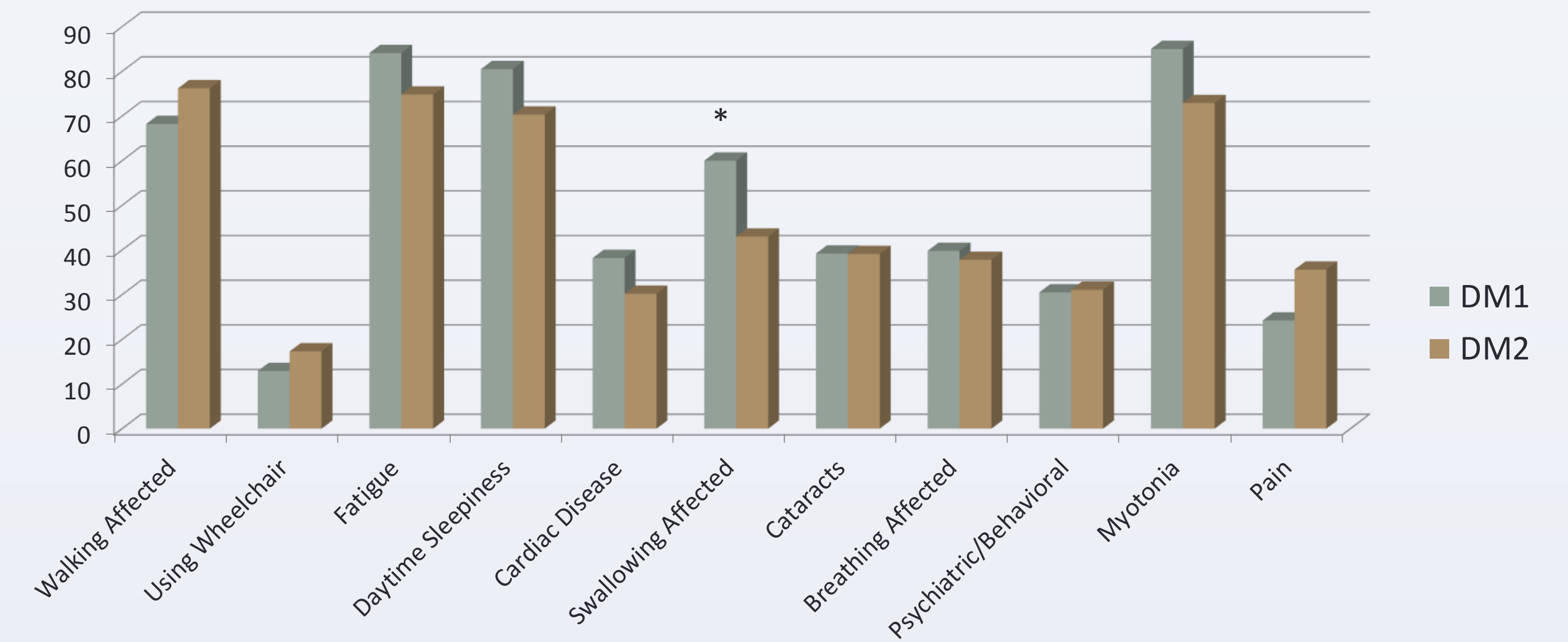


Figure 3. Diagnoses in the Registry



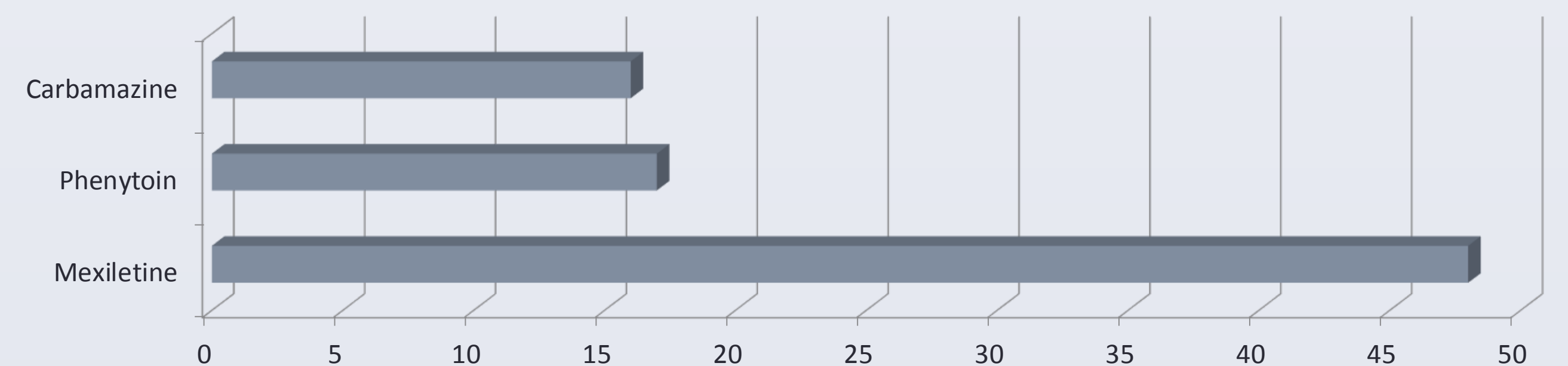
Question wording: "What is your diagnosis according to your doctor?" Of these, the majority have been diagnosed with DM1. Of those who have had genetic testing, 778 report being diagnosed with DM1 and 201 report being diagnosed with DM2 (1262 responses).

Figure 4. Reported Symptoms in Adult Onset DM1 and DM2



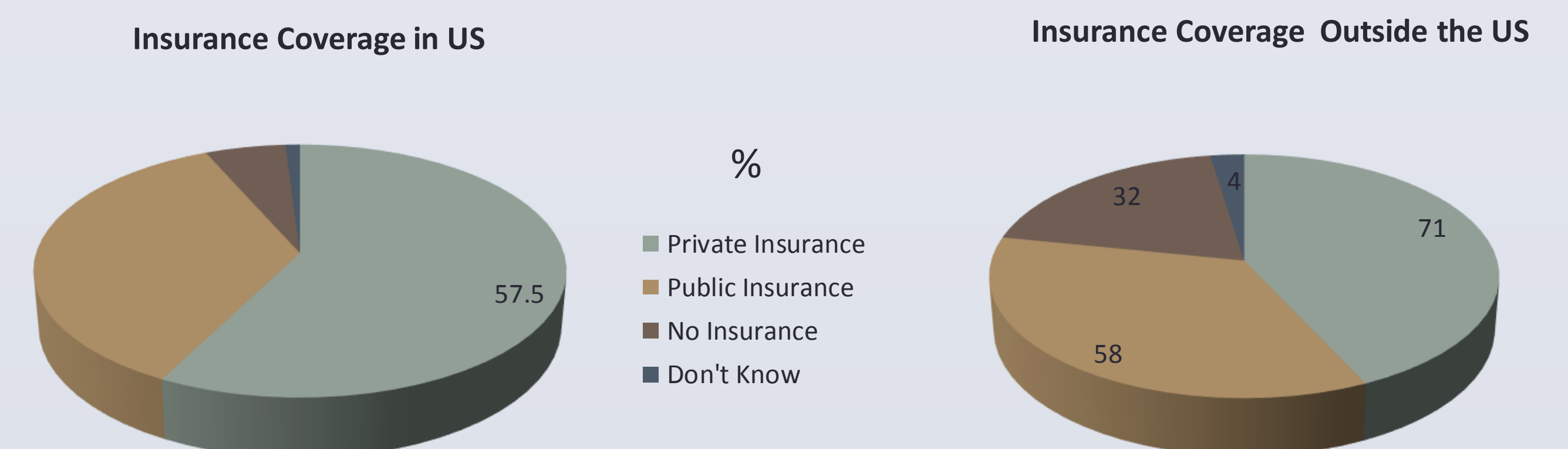
All responses were tabulated as "yes" or "no." Where degree of symptom was broken down, any degree of the presence of the symptom was considered a "yes." DM1 (average of 533 responses for each question). DM2 (average of 201.6 responses for each question). Swallowing was significantly more affected in DM1 than DM2 (Chi-square ; p=.002).

Figure 5. Medication for Myotonia



81 registrants reported on their use of medications for myotonia. The vast majority report using mexiletine. The Registry also collects data on medications used for daytime sleepiness.

Figure 6. Socio-Economic Data



- The majority of registrants say that DM has had little or no effect on their finances (1141 responses)
- 37% of registrants have completed college or technical school and 16% have completed graduate school (1149 responses)
- **67% of people with DM age 25-65 are unemployed or underemployed** (1149 responses)

## Conclusions

Registrants are primarily from the United States, Canada, the United Kingdom, and Australia, although 37 countries are represented. Adult onset DM1 is the most common diagnosis in the Registry. Fatigue, myotonia, daytime sleepiness and difficulty walking were the most frequently reported symptoms by registrants with a diagnosis of adult-onset DM1 or DM2. The prevalence of these symptoms in the MDFR is slightly lower than those reported by Heatwole and colleagues using the MDHI QOL instrument<sup>2</sup>, but the difference may be explained by variations in the questions and different selection bias for the study populations. The data also suggest that the majority of people in the Registry between ages 25 and 65 are unemployed or underemployed despite having a slightly higher than average education level for the US. Access to the MDFR is available by registration at <https://myotonic.patientcrossroads.org/> where every question is graphed for those registered as participants or researchers. Caveats for this dataset include a selection bias for those who are willing and able to complete surveys online and the potential for incorrect data to be entered by the registrant.

## References

1. Thompson R, Schoser B, Monckton D, Blonsky K, Lochmüller H. Patient Registries and Trial Readiness in Myotonic Dystrophy –TREAT-NMD/Marigold International Workshop Report. *Neuromuscular Disorders* 19 (2009) 860–866.
2. Heatwole C, et al. Myotonic Dystrophy Health Index: initial evaluation of a disease-specific outcome measure. *Muscle Nerve*. 2014 June;49(6): 906-14.

## Acknowledgements

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