

# Myotonic Dystrophy Family Registry: Current Summary of Patient Demographics & Disease Characteristics



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## Introduction

The Myotonic Dystrophy Family Registry (MDFR) is a patient self-reported online database gathering data from individuals affected by myotonic dystrophy (DM).

The Registry launched in 2013 as a tool to:

- Help researchers and community members learn more about the scope and impact of myotonic dystrophy
- Help identify and organize the DM patient community for clinical trials and studies
- Speed up research and care in myotonic dystrophy by providing critically needed information to scientists and medical professionals pursuing treatments and a cure for DM

Currently one of the largest DM registries in the world, the MDFR\* collects information about:

- 1. Demographics:** This section includes 49 data entry points and covers items such as: name, address, age, gender, willingness to be contacted for trial opportunities, complete contact information and diagnosis.
- 2. Symptoms:** 46 questions about DM related symptoms are asked in this section. Items covered include: motor function, cardiac, respiratory, and GI status as well as information on myotonia, fatigue, cataracts, pain, psychiatric/behavioral issues and tumors.
- 3. Quality of Life:** This last section consists of 14 questions pertaining status of medical insurance coverage, employment, and disability impact on day-to-day living.

\*The MDFR is available in English and survey questions are multiple choice

Access the MDFR registration online at:  
<https://myotonicregistry.patientcrossroads.org/>

## Goal

The present study provides a summary of data collected in the MDFR from March 2013 to July 2023. Comparison between demographics, symptoms prevalence and quality of life reported by DM patients worldwide is shown.

## Results

Figure 1. Participants by reported clinical diagnoses

(\* Unknown category includes patients that selected a diagnosis of "other", "I don't know" or left the question blank.

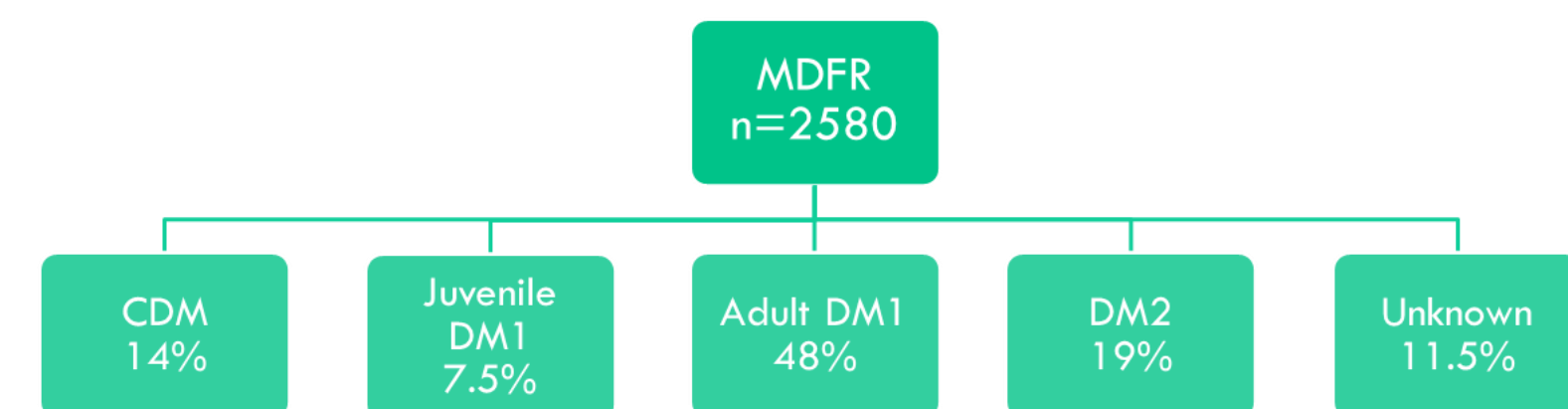


Figure 2. Geographic Representation of Registrants

55 countries are represented by 2580 registrants in the Registry. The majority of people reside in United States, Canada, and the United Kingdom.

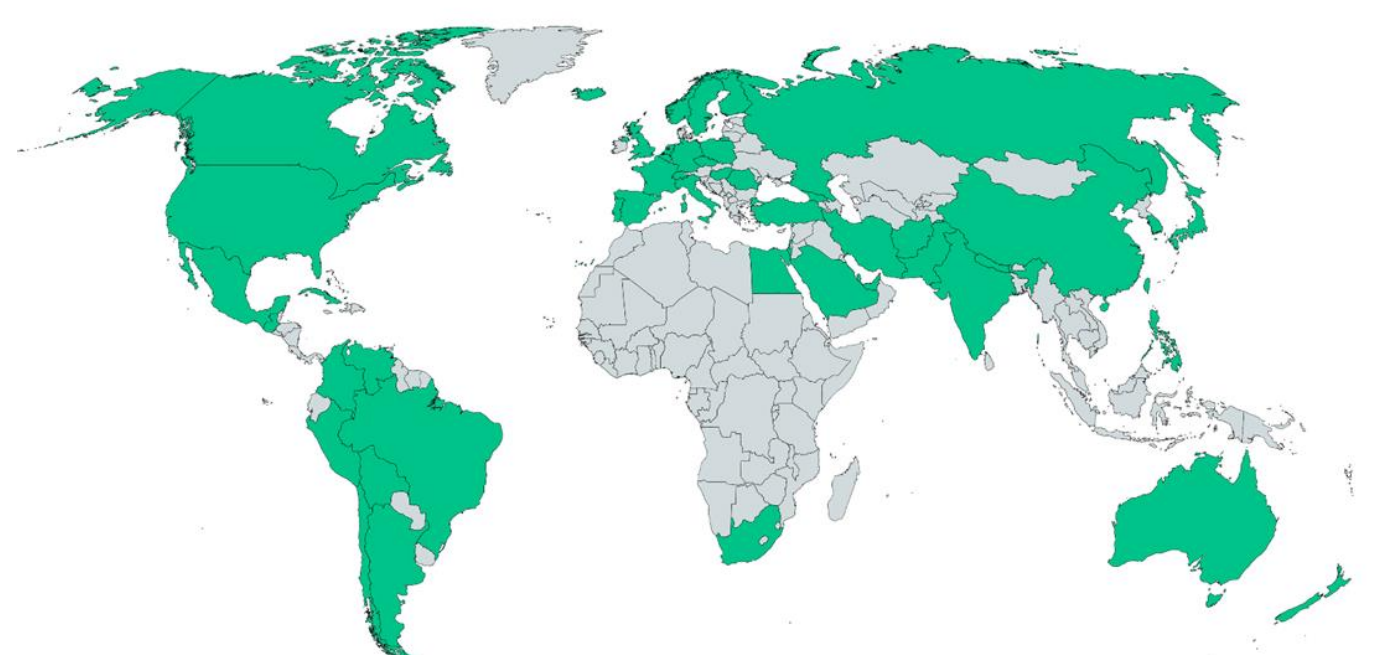


Table 1. Demographics by Disease Sub-Type

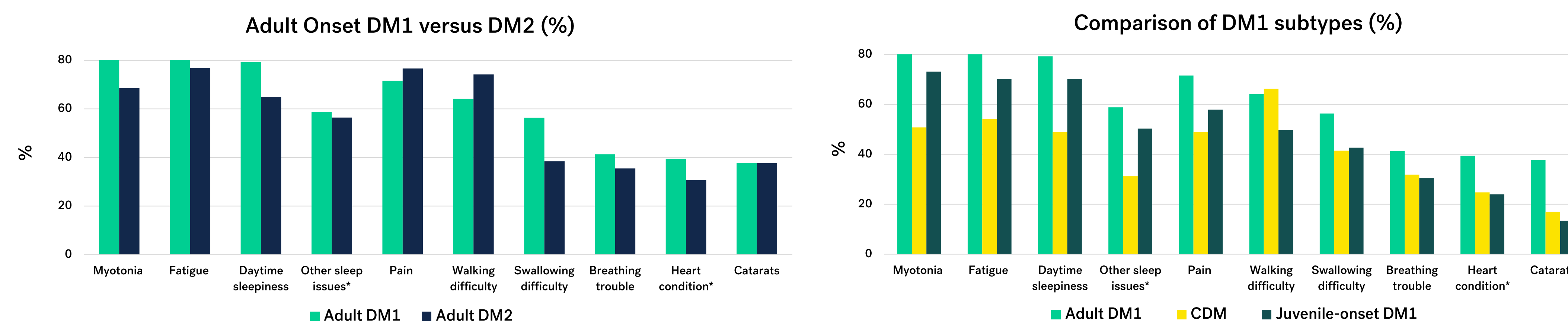
	Congenital (DM1)	Juvenile (DM1)	Adult (DM1)	DM2
Current age (yr), mean ± sd	29.3 ± 18	26.7 ± 11	46.1 ± 14	59.4 ± 17
Male (%)	52.3	52.1	45.1	41.5
Female (%)	45.6	46.7	54.6	57.8
Ethnicity - White (%)	84.3	87.0	89.9	94.0
Place of Birth (%)				
USA	73.0	86.0	75.4	82.9
Canada	4.0	3.5	6.8	6.3
Rest of World	23.0	10.5	17.8	10.8
Age (yr, mean ± sd) at first medical problem*	Birth to 4 weeks (66% of answers)	8.8 ± 6.3	35.2 ± 14	35.4 ± 15
First person in family given diagnosis (%)	56.3	34.2	41.0	51.8
Genetically Confirmed Diagnosis (%)	84.8	86.2	82.9	86.9

(\* For CDM 66% subjects responded "Birth to 4 weeks of age", 4% responded "1 to 11 months of age" and 30% reported numerical answers >1yr. For Juvenile DM1, Adult DM1 and Adult DM2, averages were calculated using numerical responses reported by participants.

Table 2. Disease Symptoms Collected in the Registry

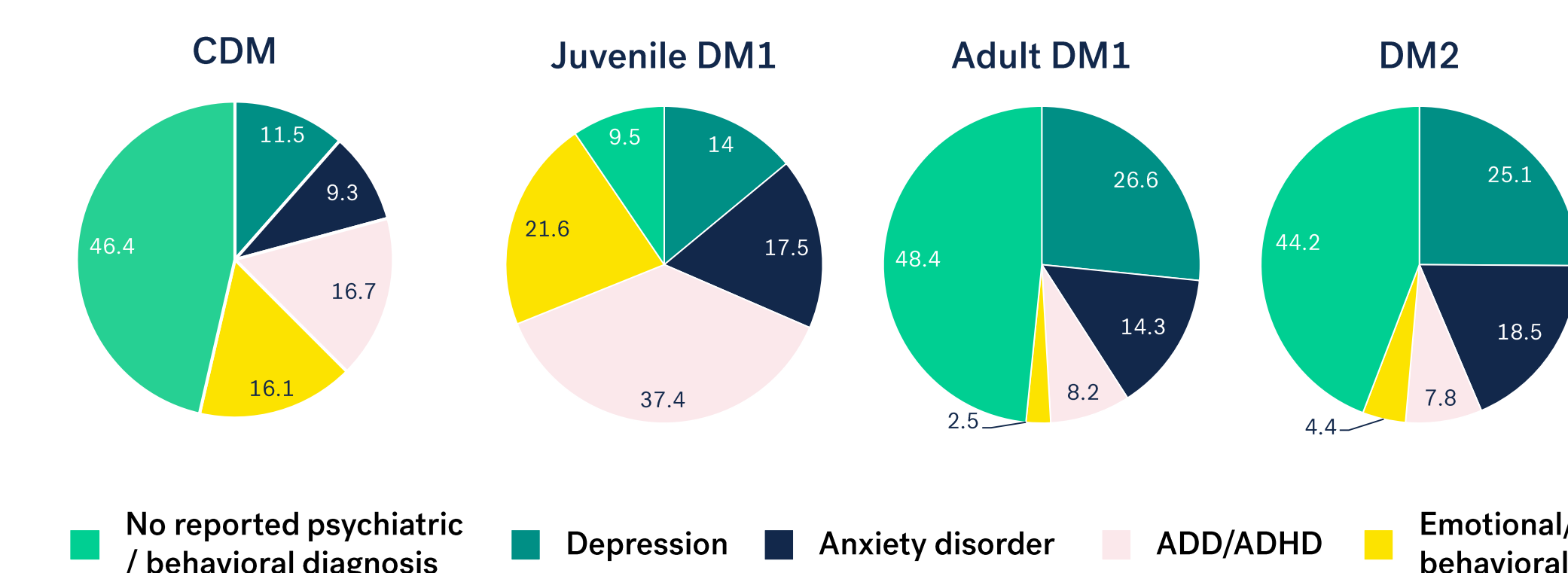
SKELETAL MUSCLE	CARDIOVASCULAR	RESPIRATORY	GASTROINTESTINAL	VISION	BRAIN
<ul style="list-style-type: none"> <li>• Walking difficulty</li> <li>• Use of orthopedic aids</li> <li>• Myotonia</li> </ul>	<ul style="list-style-type: none"> <li>• Heart condition diagnosis</li> <li>• Pacemaker or ICD</li> </ul>	<ul style="list-style-type: none"> <li>• Breathing difficulty</li> <li>• Use of ventilation device</li> </ul>	<ul style="list-style-type: none"> <li>• Swallowing difficulty</li> <li>• Use of gastric/nasal feeding tube</li> </ul>	<ul style="list-style-type: none"> <li>• Vision trouble due to cataracts</li> <li>• Cataract surgery</li> </ul>	<ul style="list-style-type: none"> <li>• Daytime sleepiness</li> <li>• Other sleep issues</li> <li>• Fatigue</li> <li>• Pain</li> <li>• Psychiatric/emotional disorders</li> </ul>

Figure 3. Prevalence of Symptoms



\*Other sleep issues include trouble falling asleep, trouble staying asleep and off-schedule sleep patterns. \*\*Heart Condition: diagnosed. For Myotonia and Fatigue, the survey asked if these symptoms had a negative effect on normal daily activities. To calculate the prevalence, responses of "yes, severely," and "yes but only mildly" were combined. For Pain, the survey asked how much pain interfered with enjoyment of life and prevalence was calculated by combining the responses of "very much," "quite a bit," "somewhat" and "a little bit." For all other symptoms/diagnoses, the survey asked if the patient experienced the symptom or had the diagnosis with available responses of "yes," "no" and "I don't know."

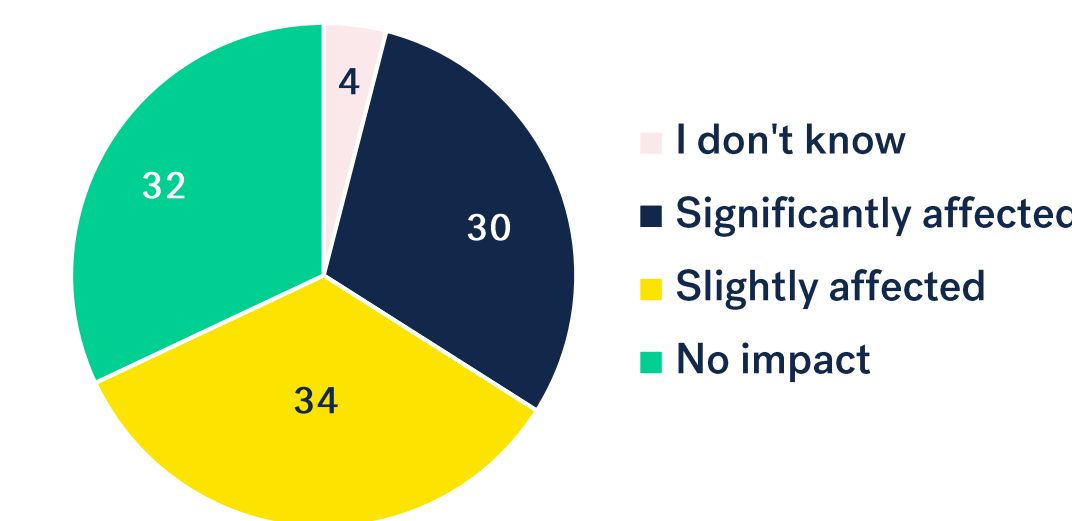
Figure 4. Effect of DM on Mental/Behavioral Health (%)



\*Emotional/Behavioral diagnosis includes Asperger Syndrome, Autism, Obsessive Compulsive Disorder and Pervasive Development Disorder

Figure 6. Socio-Economic Data

Impact of DM on Families Finances (%)



\*To calculate impact of DM on participants finances survey responses reporting "significantly affected" and "severely affected" were combined.

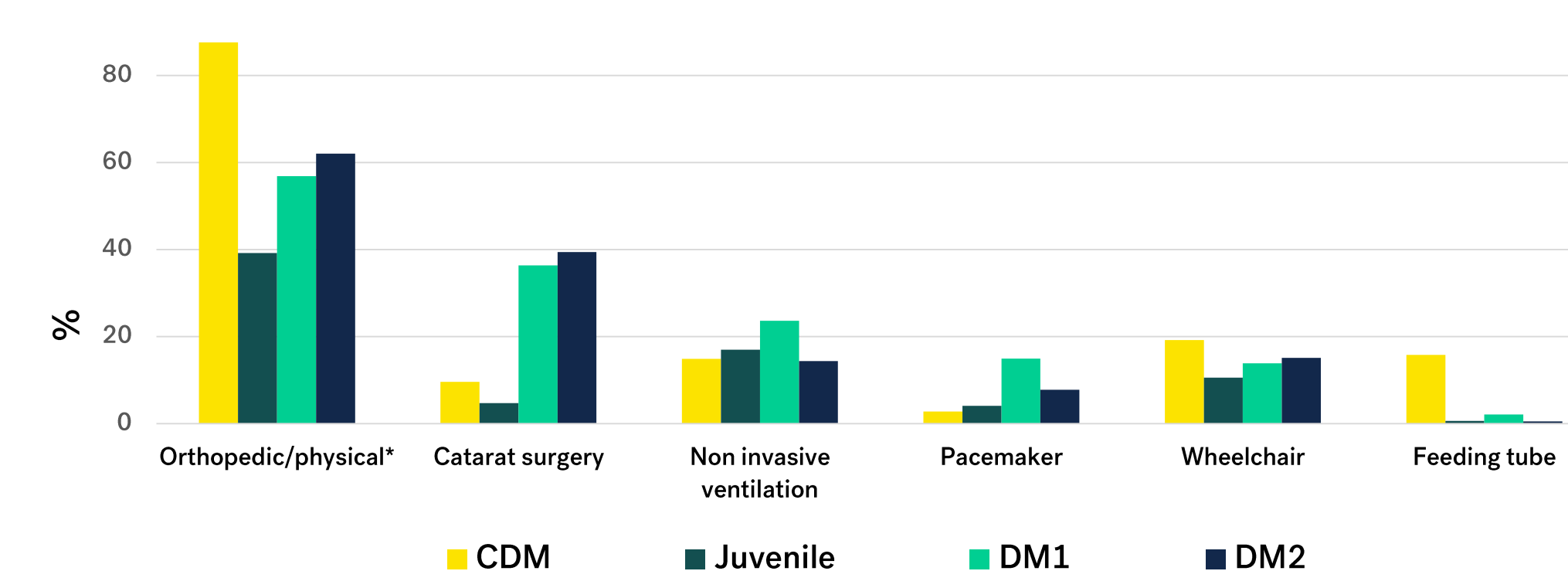
Employment Status

	DM1 n=1007	DM2 n=416
Employed - Full Time	27.7 %	32.7 %
Employed - Part Time	11.2%	7.7 %
Unemployed	11.7 %	5.3 %
Disabled / Unable to Walk	25.7 %	21.2 %
Retired	10.5 %	21.2 %
Unknown	13.2 %	11.9 %

How Has DM Affected Your Employment?

	DM1 n=907	DM2 n=368
Lost My Job	14.4 %	12.5 %
I Took Early Retirement	10.6%	16.5 %
Job Changed to Accommodate Physical Limitations	10.5 %	13.3 %
DM did not Affect my Job	36.3 %	35.1 %
I Went on Disability	28.2 %	22.6 %

Figure 5. Percentage of Patients Using Specified Devices



\* For Orthopedic Devices responses reporting use of "ankle or leg bracelets", "cane", "orthotics", "scooter", "stroller" and/or "walker" were combined.

## Summary

- >2500 participants from >50 countries enrolled in the MDFR from March 2013 to July 2023
- ~ 80% of participants are from United States
- On average, 45.8% of participants are the first in their families to receive the diagnosis and 85.2% have had their diagnosis genetically confirmed
- Adult onset DM1 is the most common diagnosis (48%)
- Aggregate data show that 67% of respondents have difficulty walking, 75% have myotonia, 72% daytime sleepiness, 70% experience pain and 76% report some degree of fatigue
- Over 35% Juvenile DM1 have been diagnosed with ADD or ADHD and show higher incidence of emotional/behavioral diagnosed conditions than patients with other DM types
- More than half (64%) of total MDFR participants reported to have been financially affected by DM, with 30% being significantly to severely impacted

## Conclusions

- The Myotonic Dystrophy Family Registry is an important tool to collect information on the impact and scope of myotonic dystrophy from the perspective of patients and their families
- Adult onset DM1 is the most common diagnosis in the registry
- Analysis of symptom prevalence, device utilization and quality of life measurements show a substantial burden of disease in a significant proportion of patients. Fatigue, myotonia, daytime sleepiness, pain and difficulty walking are the most common frequently reported symptoms
- Adult onset DM1 and DM2 show very similar symptom prevalence
- Juvenile DM1 patients show a higher percentage of diagnosis for ADD, ADHD, emotional or health problems
- Registrant participants are primarily from the United States with > than 80% of all participants self identify as White

## Future

The Myotonic Dystrophy Foundation works with the DM community in an effort to keep the Registry relevant and up to date, asking Registry participants to update their information (symptoms prevalence; quality of life) on a yearly basis. Having consecutive data entry points per participant will allow us in the future to perform longitudinal analysis of the data gathered over time and learn more about the impact and progression of myotonic dystrophy.

Learn more at <https://myotonicregistry.patientcrossroads.org/>

