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 cures 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 myopathy, 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 surveys are multiple choice.

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

Table 1: Demographics by Disease Sub-Type

<table>
<thead>
<tr>
<th>Region</th>
<th>Diagnosis DM1</th>
<th>Juv DM1</th>
<th>Adult DM1</th>
<th>DM2</th>
</tr>
</thead>
<tbody>
<tr>
<td>Current age, mean ± sd</td>
<td>29.3 ± 8</td>
<td>26.7 ± 11</td>
<td>46.1 ± 14</td>
<td>59.4 ± 17</td>
</tr>
<tr>
<td>Male (%)</td>
<td>52.3</td>
<td>52.1</td>
<td>45.1</td>
<td>43.5</td>
</tr>
<tr>
<td>Female (%)</td>
<td>47.7</td>
<td>47.9</td>
<td>54.9</td>
<td>56.5</td>
</tr>
<tr>
<td>Ethnicity - White (%)</td>
<td>84.3</td>
<td>87.0</td>
<td>89.9</td>
<td>94.0</td>
</tr>
<tr>
<td>Place of Birth - USA</td>
<td>73.0</td>
<td>86.0</td>
<td>75.4</td>
<td>82.9</td>
</tr>
<tr>
<td>Canada</td>
<td>4.0</td>
<td>3.5</td>
<td>6.8</td>
<td>6.3</td>
</tr>
<tr>
<td>Rest of World</td>
<td>23.0</td>
<td>10.5</td>
<td>17.8</td>
<td>10.8</td>
</tr>
<tr>
<td>Age (yr ± mean ± sd at first medical problem)</td>
<td>Birth to 4 years (66% of answers)</td>
<td>8.8 ± 3.3</td>
<td>35.2 ± 34</td>
<td>35.4 ± 15</td>
</tr>
<tr>
<td>First person in family given diagnosis (%)</td>
<td>56.3</td>
<td>34.2</td>
<td>41.0</td>
<td>51.8</td>
</tr>
<tr>
<td>Sensitively Confirmed Diagnosis (%)</td>
<td>84.8</td>
<td>86.2</td>
<td>82.9</td>
<td>86.9</td>
</tr>
</tbody>
</table>

*For DM1 68% subjects responded “Birth to 4 weeks” age, 4% responded “1 to 1 month” age and 33% reported numerical answer on “3+”. 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

<table>
<thead>
<tr>
<th>Symptom</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>Myotonia</td>
<td>84.4%</td>
</tr>
<tr>
<td>Muscle weakness</td>
<td>93.8%</td>
</tr>
<tr>
<td>Fatigue</td>
<td>92.5%</td>
</tr>
<tr>
<td>Heart condition</td>
<td>54.3%</td>
</tr>
<tr>
<td>Sleep apnea</td>
<td>44.1%</td>
</tr>
<tr>
<td>Other sleep issues</td>
<td>30.2%</td>
</tr>
</tbody>
</table>

*Other sleep issues include troubles falling asleep, trouble staying asleep and off-nights sleep patterns.

“Heart condition diagnosed: For Myotonia and Fatigue, the survey asked if these symptoms had negative effects on normal daily activity. To calculate the prevalence, responses of “yes, severely,” “yes, mildly,” “no," “not at all” were combined. For Pain, the survey asked how much pain interfered with quality of life and prevalence was calculated by combining the responses of “very much,” “quite a bit,” “somewhat” and “a little.” For other symptoms diagnoses, the survey asked if the participant experienced and the symptom had negatively affected with usual activities; “yes,” “no” and “I don’t know.”

Figure 1. Participants by reported clinical diagnoses

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.

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 patients 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 (49%)
- Aggregate data show that 47% of respondents have difficulty walking, 79% have myotonia, 72% daytime sleepiness, 70% experience pain and 70% 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 DMFBR participants reported to have been financially affected by DM, with 30% being significantly 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 commonly 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/behavioral problems
- Registrants participate primarily from the United States with >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/