Myotonic Dystrophy Family Registry

Current Summary of Patient Demographics and Disease Characteristics

MYOTONIC DYSTROPHY FOUNDATION

Care and a Cure

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Myotonic Dystrophy Foundation

Introduction

The Myotonic Dystrophy Family Registry (MDFR) is one of the largest DM patient registries in the world. It was launched in February 2013 as a tool to help researchers and the myotonic dystrophy (DM) community learn more about the scope and impact of this disease and to organize the DM patient community for studies and trials.

The MDFR is an online, patient-reported survey that contains three sections. Section 1 focuses on diagnosis and demographic information. Section 2 contains questions relating to common symptoms and treatments utilized. Section 3 is focused on patient's quality of life (QoL) and includes questions pertaining to financial impact, employment status and living conditions. The registry is annually approved by the Chesapeake IRB.

Purpose

To provide a current summary of patients enrolled in the registry and to compare the symptom prevalence, device utilization, and QoL measurements between DM1 sub-types and DM2.

Methods

Analysis was performed on 1,768 patients that enrolled in the registry between February 2013 and July 2018 who gave full informed consent. Summary of symptoms and QoL data is presented for the groups "congenital DM1", "juvenile DM1", "adult onset DM1 with symptoms" and "DM2 with symptoms". Patients identifying as "adult onset DM1 without symptoms" or "DM2 without symptoms" were excluded from this analysis to enable the comparison to be focused on active disease characteristics.

Results

Figure 1: Number of Enrolled Patients in the Registry by Disease Type

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

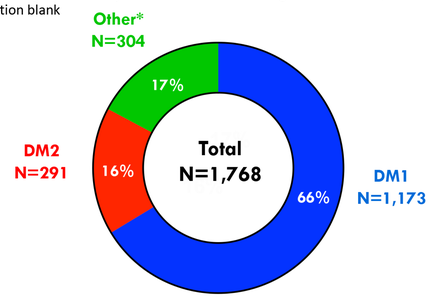
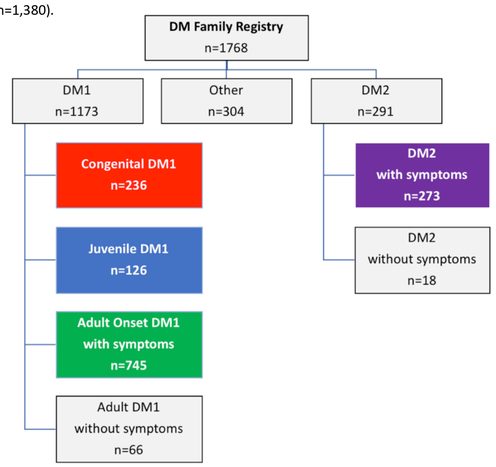


Figure 2: Flow Chart of Patients by Reported Clinical Diagnosis (Type and Sub-Type)

Subsequent analysis was performed on those patients reporting a disease diagnosis of congenital DM1, juvenile DM1, adult onset DM1 with symptoms and DM2 with symptoms (total n=1,380).



Results

Table 1: Demographics by Disease Sub-Type

	Congenital DM1 (n=236)	Juvenile DM1 (n=126)	Adult DM1 (n=745)	DM2 (n=273)	All Subjects (n=1,380)
Age (yr), mean (sd)	22.9 yr (17.6)	26.5 yr (11.4)	49.4 yr (13.4)	55.6 yr (15.2)	44.0 yr (18.8)
Male Female	51.7 % 48.3 %	53.2 % 46.8 %	47.8 % 52.2 %	46.9 % 53.1 %	48.8 % 51.2 %
White (%)	85.2 %	94.4 %	89.4 %	90.1 %	89.2 %
Relationship to affected person Self Parent Other	 17.0 % 62.7 % 20.3 %	 27.8 % 61.9 % 10.3 %	 74.5 % 9.4 % 16.1 %	 86.1 % 5.5 % 8.4 %	 62.7 % 22.5 % 14.8 %
Age at first medical problem*	Birth to 4 weeks (66% of answers)	8.8 yr (4.8)	30.2 yr (13.0)	34.4 yr (14.4)	n/d
Country of Birth USA Canada Europe Rest of World	 72.5 % 3.4 % 11.0 % 13.1 %	 84.1 % 4.0 % 3.2 % 8.7 %	 75.4 % 7.4 % 6.7 % 10.5 %	 79.1 % 6.6 % 8.4 % 5.9 %	 76.4 % 6.2 % 7.5 % 9.9 %
First person in family given diagnosis (%)	51.3 %	31.7 %	42.6 %	53.9 %	45.3 %
Genetically Confirmed Diagnosis (%)	83.9 %	84.9 %	82.1 %	86.1 %	83.5%

(*) For Congenital DM1 66% subjects reported the categorical answer of "Birth to 4 weeks", 6% reported categorical answers encompassing 1-11 mo and 23.4% reported numerical answers > 1yr. For Juvenile DM1, Adult Onset DM1, and DM2 averages were calculated using only numerical responses which accounted for 85%, 95% and 92% of answers, respectively

Figure 3: Disease Symptoms Collected in Registry

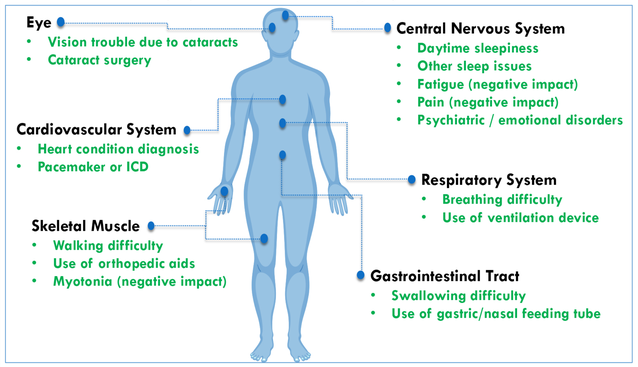
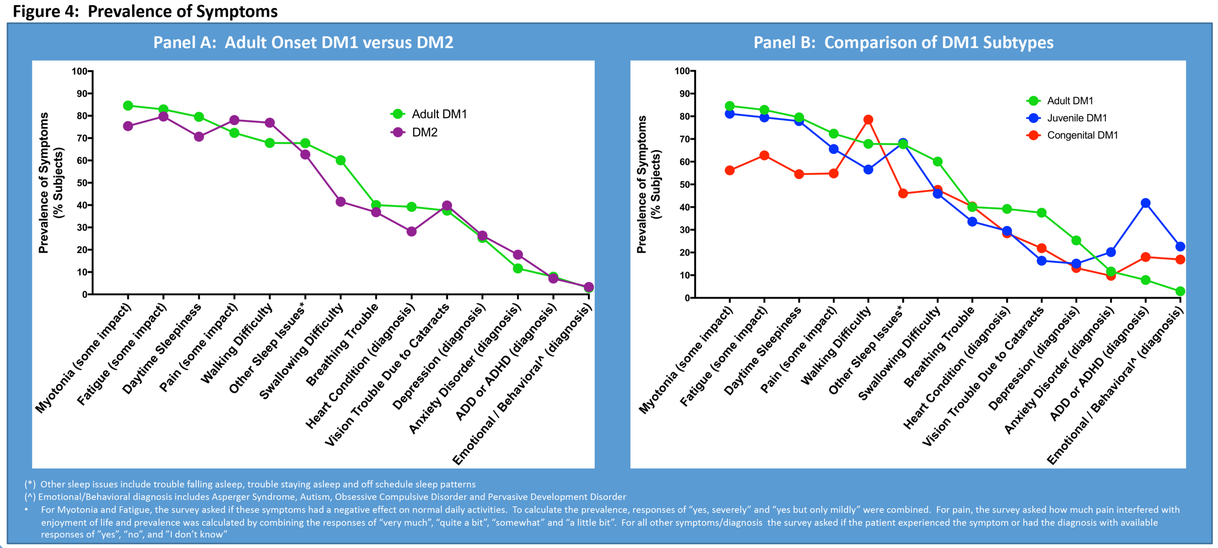


Figure 4: Prevalence of Symptoms



(*) Other sleep issues include trouble falling asleep, trouble staying asleep and off schedule sleep patterns

(^) Emotional/Behavioral diagnosis includes Asperger Syndrome, Autism, Obsessive Compulsive Disorder and Pervasive Development Disorder

- 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/diagnosis the survey asked if the patient experienced the symptom or had the diagnosis with available responses of "yes", "no", and "I don't know"

Results

Figure 5: Percent of Patients Using Specified Devices or with a History of Cataract or Pacemaker Surgery by Disease Sub-Type

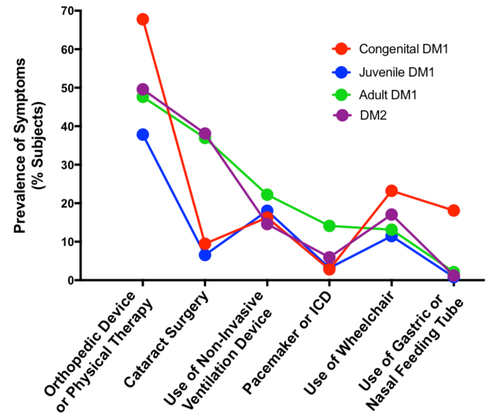


Table 2: Current Living Condition

Only congenital DM1 and juvenile DM1 patients older than 18 yrs were included in this analysis. The semi-independent category combines the responses "semi-independent (I live with a relative or parent)" and "I live in a group home". The dependent category combines the responses "dependent-I live with a relative or parent" and "dependent-I live in an assisted or other skilled living facility"

Current Living Condition	Congenital DM1 (>18 yrs) N=98	Juvenile DM1 (>18 yrs) N=89	Adult Onset DM1 N=695	DM2 N=251
Independent	5.1 %	11.2 %	15.2 %	23.5 %
With Spouse or Significant Other	26.5 %	12.4 %	67.6 %	66.5 %
Semi-Independent	16.3 %	20.2 %	7.2 %	2.4 %
Dependent	51.0 %	55.1 %	9.2 %	7.2 %
I don't know	1.0 %	1.1 %	0.7 %	0.4 %

Results

Table 3: Employment Status of Adult Onset DM1 versus DM2

What is Your Employment Status?

	Adult DM1 N=700	DM2 N=252
Employed – Full Time	28.7 %	33.7 %
Employed – Part Time	12.9 %	7.9 %
Unemployed	14.1 %	7.9 %
Disabled / Unable to Walk	28.7 %	25.4 %
Retired	11.3 %	21.0 %
Student	3.1 %	3.6 %

How Has Myotonic Dystrophy Affected Your Employment?

	Adult DM1 N=692	DM2 N=248
Lost My Job	16.7 %	12.5 %
I Took Early Retirement	10.8 %	16.9 %
Job Changed to Accommodate Physical Limitations	9.7 %	13.3 %

Summary

SUMMARY of REGISTRY PARTICIPANTS BY THE NUMBERS

- Approximately equal distribution of female and male
- ~75% were born in the US
- 45% were the first in their family to receive the diagnosis
- 83% had their diagnosis genetically confirmed
- 15-22% use a non-invasive ventilation device
- 18% congenital DM1 use a feeding tube
- 23% congenital DM1 and 17% DM2 use a wheelchair
- 42% juvenile DM1 have been diagnosed with ADD or ADHD
- >50% congenital DM1 and juvenile DM1 >18 yrs have dependent living conditions
- >25% adult onset DM1 and DM2 are disabled / unable to work

Conclusions

- 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
- Analysis of symptom prevalence, device utilization and QoL measurements show a substantial burden of disease in a significant proportion of patients
- Adult onset DM1 and DM2 show very similar symptom prevalence and impact on QoL measurements
- Congenital DM1 and juvenile DM1 patients show a higher percentage of diagnosis for ADD, ADHD, emotional or behavioral health problems