TREAT-NMD: an International Collaboration in Myotonic Dystrophy Type 2

Peric S1, Bennett N[2], Porter B[2], Walker H[3], Ashley E-J[4], Esparis B[5], Cumming K[6], Stevenson T[6], Nakamura H[7], Komaki H[7], Cosyns M[8], Jagut M[8], Marini-Bettolo C[3], Thiele S[9], Walter M[9], Rakocevic-Stojanovic V[10], Durmus H[11], Mergen S[11], Hodgkinson V[12], Korngut L[12], Lusakowska A[13], Kostera-Pruszczyk A[13], Voháňka S[14], Mokrá L[14], Rodrigues M[15], Forbes R[16], Yu E[16], Osredkar D[1], Golli T[1], Allison D[2], Campbell C[17], Guglieri M[18], Ambrosini A[19], Roxburgh R[15].

1 Registry of Slovenian Children with NMD, Slovenia; 2 Treat-NMD Services Ltd, UK; 3 UK National Registry for Myotonic Dystrophy, UK; 4 Cure-DM, UK; 5 University of Pennsylvania, USA, 6 Myotonic Dystrophy Family Registry, USA, 7 Remudy Registry, Japan, 8 Belgian Neuromuscular Diseases Registry, Belgium; 9 German NMD Registry, Germany; 10 Serbian Registry for DMs, Serbia; 11 Istanbul University, Turkey; 12 Canadian Neuromuscular Disease Registry, Canada; 13 Polish Registry of Patients with NMDs, Poland; 14ReaDY – FSHD & DM Registry, Czechia; 15 Pūnaha Io, the NZ NeuroGenetic Registry and Biobank, New Zealand; 16 Australian Neuromuscular Disease Registry, Australia; 17 Western University, London, ON, Canada; 18 Newcastle University and Newcastle Hospitals NHS Foundation Trust, UK; 19. Fondazione Telethon, Milan, Italy

Introduction:

Myotonic dystrophy type 2 (DM2) is a rare multi-system disease recognised in the last three decades.

One of the key infrastructures developed by the TREAT-NMD Alliance is the Global Registry Network, governed by the TREAT-NMD Data Systems Oversight Committee. The network is a federation of individual, independent, national (or regional) patient registries whose members collect agreed, disease-specific datasets.

Registries in the Global Registry Network collect data on patients living with a range of neuromuscular conditions DM2. The Myotonic Dystrophy Global Registry Network includes 22 registries, representing four continents (Figure 1A).

The aim is to assess the number of DM2 patients included in the network, and analyse socio-demographic and clinical features.

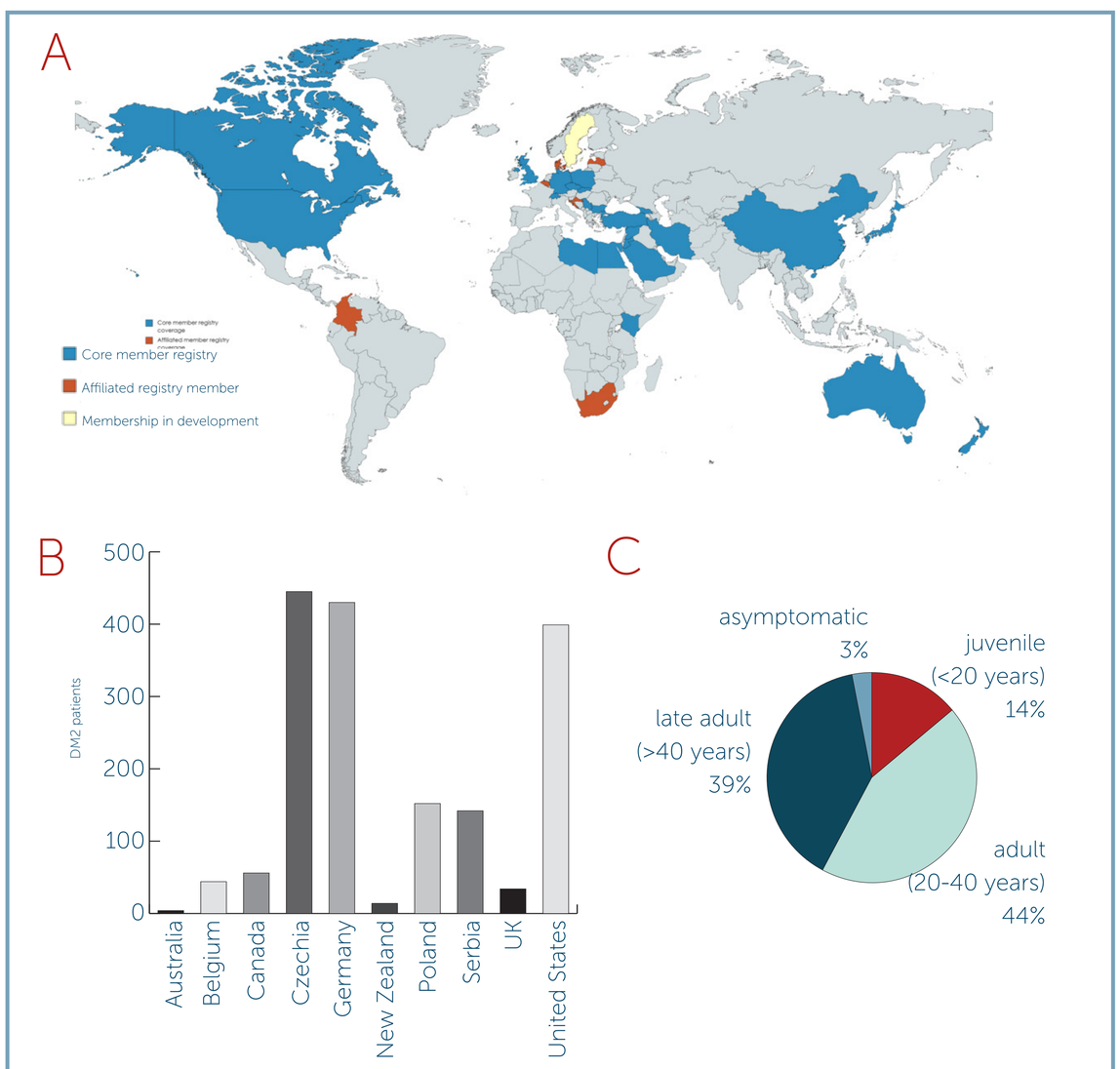

Figure 1

(A) The TREAT-NMD Myotonic Dystrophy Global Registry Network includes 22 registries across 5 continents. Image: mapchart.net. (B) In total, the responding registries collect data on on 1,720 DM2 patients. Median age on entering registry is 51 years and current median age is 56.5 years and 63% are female. (C) Age at disease onset.

Results:

Of the 13 DM registries that responded, ten enrolled DM2 patients. The total number of DM2 cases was 1,720, with the Czech/Slovakian, German and the USA (MDF) registries enrolling the most patients with 445, 430, and 339, respectively (Figure 2).

The highest number of registered cases per 100,000 population was seen in the Czech/Slovakia (4.2) and Serbia (2.0). The DM2:DM1 ratio was highest in the Central European countries. (Figure 2)

Registry enrolment occurred at a median age of 51 years with 63% being female. Onset of DM2 occurred before the age of 20 in 14% of cases. One fifth of DM2 patients used an assistive device to walk and 4% were non-ambulant. Pacemaker or implantable cardioverter-defibrillator was reported in 4% of DM2 subjects, while 7% used non-invasive ventilation.

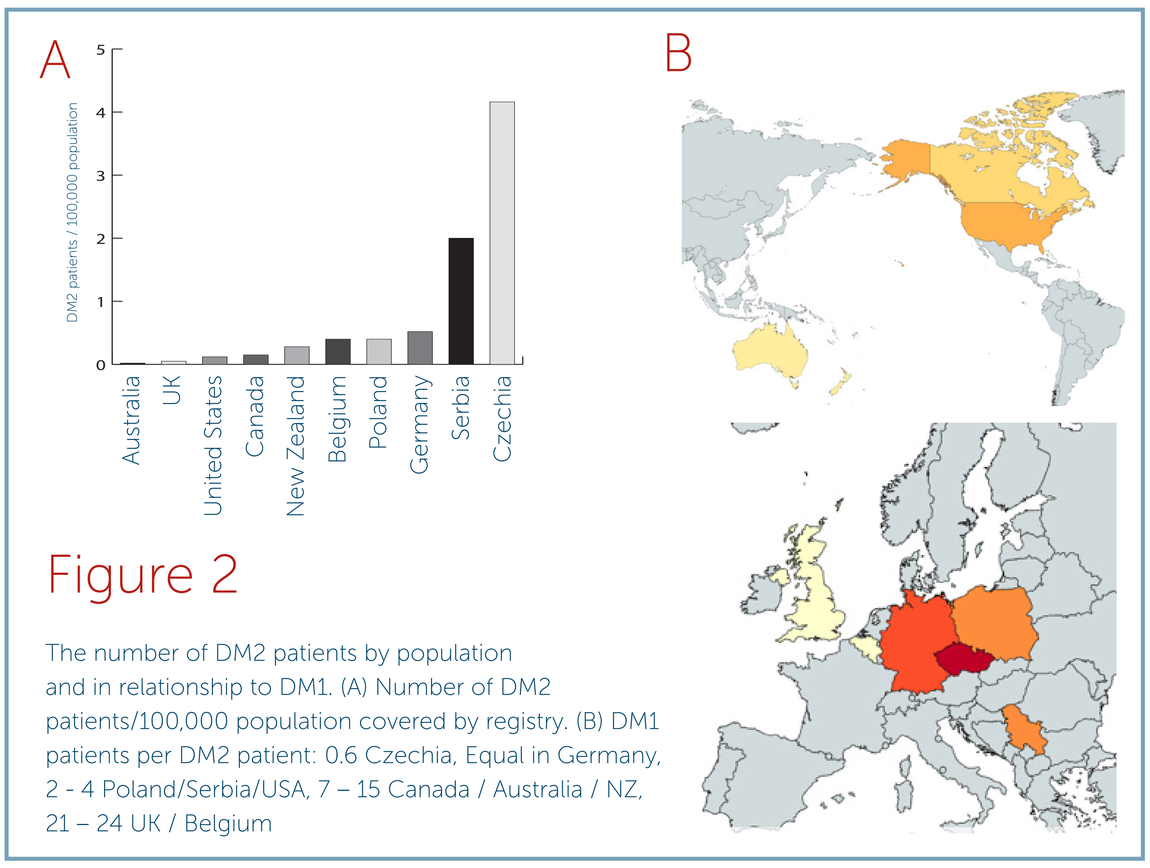

Figure 2

The number of DM2 patients by population and in relationship to DM1. (A) Number of DM2 patients/100,000 population covered by registry. (B) DM1 patients per DM2 patient: 0.6 Czechia, Equal in Germany, 2 - 4 Poland/Serbia/USA, 7 – 15 Canada / Australia / NZ, 21 – 24 UK / Belgium

Feature	*Number of pts with data*	Percentage
Ambulation status	*1601*	
normal walk		**76%**
assisted walk		**20%**
nonambulatory		**4%**
Hand grip myotonia	*1587*	**70%**
Cataracts and/or cataracts surgery	*966*	**58%**
Cardiac conduction defects and/or arrhythmia	*1176*	**26%**
Pacemaker or ICD	*1176*	**4%**
Daytime sleepiness	*855*	**40%**
Pulmonary restriction (FVC<90%)	*406*	**34%**
Respiratory device	*1210*	
None		**92%**
NIV		**7%**
IV		**1%**
Dysphagia	*1608*	**22%**
Gastrostomy or nasogastric tube	*1286*	**0.5%**

Table 1

The number and percentage of DM2 patients who report experiencing symptoms and the treatments and aids they report using.

TREAT-NMD DM Dataset

The TREAT-NMD Core Dataset for myotonic dystrophy was published in 2009 and is made freely available online.

The dataset is suitable for patient- or clinician-reported registries and compatible with Privacy Preserving Record Linkage (PPRL) tools.

Scan to view full details of the dataset

https://treat-nmd.org/downloads/file/registries_toolkit/DM1_core_dataset_August2009.pdf

Conclusions:

TREAT-NMD member registries were able to assemble the largest DM2 cohort to date with an international reach, providing meaningful clinical and demographic data. More DM registries should aim to capture DM2 data to contribute to important collaborations such as this one, which can support future research and clinical trial recruitment.

If you are interested in understanding more about the TREAT-NMD Global Registry Network and how interrogating this data could support your research or drug development efforts, please contact registries@treat-nmd.com.