An International Collaboration in Myotonic Dystrophy Type 2


On behalf of

TREAT-NMD Myotonic Dystrophy Subgroup & TREAT-NMD Global Registry Network

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**Our vision**
To accelerate the development of effective treatments and to establish best practice diagnosis and care for NMD patients worldwide.

**Our mission**
A collaborative, inclusive global network and organisational infrastructure that will overcome fragmentation, providing support services, information and data to advance treatment, diagnosis and care for NMD patients globally.
Global Registry Network

- >60 Independent patient registries.

- Members agree to TREAT-NMD Charter:
  - collecting the TREAT-NMD Core dataset for myotonic dystrophy (DM),
  - agreeing to sharing de-identified, aggregate data
  - completing a Confidentiality Disclosure Agreement

- Global Registry Enquiries
  - Pharma – scoping clinical trials***
  - External Researchers
  - Internal Researchers
    TREAT-NMD Myotonic Dystrophy working group

Subgroup co-leads
Dr Richard Roxburgh    Dr Stojan Peric

Subgroup patient representatives
Dr Belen Esparis (parent)    Emma-Jayne Ashley (parent)
TREAT-NMD Myotonic Dystrophy Global Registry Network

Core registries: 16
Affiliated registries: 8
Main focus of paper was on Registries

- **21 active registries:**
- **Majority national coverage**
- **Data quality**
  - 36% registries collected all mandatory and highly encouraged items
  - Data most often updated annually
  - 95% Data entered by clinician

- **Purpose and utility of the registries**
- **Resources and technical solutions**
- **Importance of agreed core dataset**

**Patient data**

- More than 10 000 DM pts
  - 86% DM1, 14% DM2
- No data on patients’ features
Main focus of registries
- 21 active registries
- Majority with national coverage
- Data quality
  - 36% registries collected all mandatory and highly encouraged items
  - Data most often updated annually
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- Purpose and utility of the registries
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Patient data
- More than 10 000 DM pts
  - 86% DM1, 14% DM2
- No data on patients’ features
• To assess the number of DM2 patients included in the TREAT-NMD Global Registry Network

• To analyse their
  ▪ Demographic features
    o Intercountry variation
    o Age structure
  ▪ Clinical features
    o Ambulation / Cardiac / Respiratory involvement
    o Cataracts
METHODS

• Questionnaire sent to all sixteen registries in the TREAT-NMD Global Registry Network

• Questionnaire in the form of an Excel spreadsheet

• Where results were incomplete follow up emails were sent to registered curators

• Data acquired over 8 week period in first half 2022
Registries
• 13 of 16 core registries responded
• 10 registries enrolled DM2 patients

Patients
• 1720 DM2 patients in the Network
• 63% female
• Disease onset
  juvenile (before 20) 14%
  adult (20-40) 44%
  late adult (above 40) 39%
  asymptomatic 3%
• Median age at entering registry 51y
• Current median age 56.5y
Number of DM2 patients / 100,000 population

- Australia: 0.02
- UK: 0.05
- United States*: 0.12
- Canada: 0.15
- New Zealand: 0.28
- Belgium: 0.40
- Poland: 0.40
- Germany: 0.52
- Serbia: 2.00
- Czechia: 4.16
Ratio of DM1 and DM2 patients

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
- **NO DM2 cases reported in Japan**
<table>
<thead>
<tr>
<th>Feature</th>
<th>Number of pts with data</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ambulation status</td>
<td>1601</td>
<td></td>
</tr>
<tr>
<td>normal walk</td>
<td></td>
<td>76%</td>
</tr>
<tr>
<td>assisted walk</td>
<td></td>
<td>20%</td>
</tr>
<tr>
<td>nonambulatory</td>
<td></td>
<td>4%</td>
</tr>
<tr>
<td>Hand grip myotonia</td>
<td>1587</td>
<td>70%</td>
</tr>
<tr>
<td>Cataracts and/or cataracts surgery</td>
<td>966</td>
<td>58%</td>
</tr>
</tbody>
</table>
### RESULTS - Cardiac

<table>
<thead>
<tr>
<th>Feature</th>
<th>Number of pts with data</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cardiac conduction defects and/or arrhythmia</td>
<td>1176</td>
<td>26%</td>
</tr>
<tr>
<td>Pacemaker or ICD</td>
<td>1176</td>
<td>4%</td>
</tr>
</tbody>
</table>
## RESULTS - Respiratory

<table>
<thead>
<tr>
<th>Feature</th>
<th>Number of pts with data</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Daytime sleepiness</td>
<td>855</td>
<td>40%</td>
</tr>
<tr>
<td>Pulmonary restriction (FVC&lt;90%)</td>
<td>406</td>
<td>34%</td>
</tr>
<tr>
<td>Respiratory device</td>
<td>1210</td>
<td></td>
</tr>
<tr>
<td>None</td>
<td></td>
<td>92%</td>
</tr>
<tr>
<td>NIV</td>
<td></td>
<td>7%</td>
</tr>
<tr>
<td>IV</td>
<td></td>
<td>1%</td>
</tr>
</tbody>
</table>
## RESULTS - Gastro

<table>
<thead>
<tr>
<th>Feature</th>
<th>Number of pts with data</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dysphagia</td>
<td>1608</td>
<td>22%</td>
</tr>
<tr>
<td>Gastrostomy or nasogastric tube</td>
<td>1286</td>
<td>0.5%</td>
</tr>
</tbody>
</table>
CONCLUSION

- DM2 uncommonly but not rarely occurs before 20
- DM2 is more common in Central/Eastern Europe and in countries where people from Central/Eastern Europe have emigrated
- DM2 very rare in Eastern Asia
CONCLUSION

• The vast majority of people with DM2 remain ambulant
• Respiratory and Cardiac involvement are common but therapeutic intervention is much lower than in DM1
• Gastro symptoms are also common seldom requiring PEG
CONCLUSION

• The *TREAT-NMD Global Registry Network* was able to assemble the largest DM2 cohort to date

• The data is useful for
  • Understanding the *Global Burden of Disease*
  • Planning pharmaceutical studies
  • Showing the availability of patients for research
  • Understanding the condition better

• Some data (esp. *respiratory*) which could be important for planning clinical studies should be collected more consistently across the network
Special thanks to...

Ben Porter
Global Registries and Market Development Manager
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