<table>
<thead>
<tr>
<th>Time</th>
<th>Session Description</th>
<th>Presenter</th>
</tr>
</thead>
<tbody>
<tr>
<td>13.15</td>
<td>Welcome and meeting objectives</td>
<td>Julie / Jess</td>
</tr>
<tr>
<td>13.30</td>
<td>Ice – Breaker and curator introductions</td>
<td>Jess</td>
</tr>
<tr>
<td>14.00</td>
<td>TREAT-NMD 2022 update</td>
<td>Neil</td>
</tr>
<tr>
<td>14.15</td>
<td>Year 3 Annual Report - highlights</td>
<td>Julie</td>
</tr>
<tr>
<td>14.30</td>
<td>Registry Case Study – UK Patient Registry PROMS</td>
<td>Lindsay</td>
</tr>
<tr>
<td>14.40</td>
<td>Core Dataset – success and lessons learnt</td>
<td>All</td>
</tr>
<tr>
<td>15.30</td>
<td>Coffee Break</td>
<td></td>
</tr>
<tr>
<td>15.45</td>
<td>Group Activities – Group A, B, C, D (20 min)</td>
<td>Julie / Jess</td>
</tr>
<tr>
<td>16.05</td>
<td>Feedback session (10 mins per group)</td>
<td>All</td>
</tr>
<tr>
<td>16.45</td>
<td>Looking Ahead … priorities for 2023</td>
<td>Julie / Jess</td>
</tr>
<tr>
<td>17.10</td>
<td>What are the Take-Home Messages</td>
<td>Julie / Jess</td>
</tr>
<tr>
<td>17.30</td>
<td>Close Session</td>
<td></td>
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</table>
Welcome & Introductions

Julie Bohill
Project Manager

Jess Page
Project Coordinator

Victoria Hodgkinson
SMA Sub-group Lead,
Executive Director of NMD registry

Miriam Rodrigues
SMA Sub-group Lead,
Neurogenetic Research Lead,
New Zealand

Marcel Heidemann
IT Consultant
Our Aims ....

- **Update** on TREAT-NMD activities
- **Highlights** from Year 3 Annual Curator survey
- **Review** what’s going well with data collection
- **Discuss** the **key challenges** with dataset roll-out
- **Share** experiences and best practice
- **Agree** priorities for 2023 and next steps
Ice-breaker

• Please order yourself in line by birth month!

• Now introduce yourself to the person on either side of you
Registry Introductions

Please stand up and state:

• Your name

• Country

• Are you representing Clinician / Patient /Dual reported registry?

• SMA specific or general NMD Registry?

• What is your population? (i.e. paediatric, adult)

• What you hope to get out of today?
TREAT-NMD & Global Registry Network Update

NEIL BENNETT
TREAT-NMD GLOBAL REGISTRIES MANAGER
Consolidating TREAT-NMD’s not for profit status

TREAT-NMD Alliance Ltd is a registered charity with a wholly-owned business arm called TREAT-NMD Service Ltd. The companies are owned by the network, and work to drive forward the network’s aims.
Our vision

To accelerate the development of effective treatments and to establish best practice diagnosis and care for neuromuscular patients worldwide.

Our mission

To operate 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 neuromuscular patients globally.

Our goals

• Leverage and expand our global reach
• Provide the ‘go to’ tools and services to support each stage of translational research
• Provide educational tools to improve diagnosis, treatment and care
• Facilitate agreement and adoption of standardised care guidelines, pre-clinical models, outcome measures and disease-specific datasets
• Further de-risk and accelerate the development of therapies by extending our advisory committees and enquiries processes
• Raise our profile and that of the neuromuscular disease areas we serve
• Facilitate best practice in data collection and become the ‘go to’ provider of NMD data to support evaluation, approval and post authorisation requirements of new treatments
De-risking and accelerating drug development

TREAT-NMD has several complimentary work streams that support drug development and bring new treatments to patients as quickly as possible.

![Diagram showing work streams in drug development process]

- Preclinical
- Clinical Trials
- Regulatory approval
- Reimbursement
- Post-marketing surveillance
- Market development
- Consulting
- Global Registry Enquiries
- Patient-level data projects
- TREAT-NMD Education Programme
- Disease-specific data model development
- Patient Information Provision
- Horizon scanning
Key registry contacts at TREAT-NMD

TREAT-NMD has several complimentary work streams that support drug development and bring new treatments to patients as quickly as possible.

<table>
<thead>
<tr>
<th>Registries Team</th>
<th>Projects Team</th>
<th>CEO</th>
</tr>
</thead>
<tbody>
<tr>
<td>Neil Bennett</td>
<td>Emma Watson</td>
<td>David Allison</td>
</tr>
<tr>
<td>Hsin Chieh Chua</td>
<td>Seung Lee</td>
<td></td>
</tr>
<tr>
<td>Farjana Ali</td>
<td>John McKenna</td>
<td></td>
</tr>
<tr>
<td>Registries Manager</td>
<td>Datasets &amp; GRP Coordinator</td>
<td>Registries Coordinator</td>
</tr>
</tbody>
</table>

TREAT-NMD Overview 2022
Any questions?
This year’s highlights – supporting registries

• Refreshed registry enquiries costing matrix

• Supported conference abstract writing and poster production

• Changed enquiry contract to better represent registries
This year’s highlights – registry activity

• Completed one SMA Registry Enquiry

• Agreed contract for first post-authorisation study

• Started one disease landscaping study

• Working on a hypothesis generation study for a post-authorisation study
This year’s highlights - behind the scenes

• Totally rebuilding the TREAT-NMD website

• Totally rebuilding the Monday.com boards

• Compliance, documentation and staff training etc
Next year...

• Registry summary publication

• Working with subgroups to address research questions

• Reviewing registry reimbursement

• Taking global registry platform to the next level
Any questions?
SMA dataset journey

**V0 (2009)**
- 29 data items

**V1 (2018)**
- 167 data items

**Current**
- **V2 (2020)**
  - 154 data items

**Future revision**
- **V3 (??)**

**2017:** Decision to expand
- Workshop 1
- Pilot (10 registries)

**2019:** Start of phased Implementation

**Phased Implementation**
- Year 1 (2019): n= 8 registries
- Year 2 (2020): n= 8 registries
- Year 3 (2021): n= 4 registries
- Year 4 (2022): n = tbc

May '23 Project End
SMA Core Dataset (V2)

154 Data Items …
Mandatory : 117 (CR), 91 (PR)
Non-Mandatory : 37

Mandatory Items
- PPRL items
- Genetic diagnosis
- SMA type & onset age
- SMA type & onset age
- Living status
- Wheelchair use
- Feeding tube use
- IV & NIV use
- Therapeutic interventions
- Clinical trial participation
- Date & cause of death
- Clinical observations incl. contractures
- Electrophysiology & biomarkers taken (Y/N)

Non-mandatory Items
- DOB, Sex, Country
- SMN2 copies
- Best & current motor function extended
- Disease-modifying therapies
- Medication and rehabilitation
- CGI according to clinician and patient
- Airway clearance Y/N
- Participation in other registries or NH studies

CR items are mandatory for clinician-reported registries
PR items are mandatory for patient-reported registries
SMA Core Dataset (V2)

Available as a technical data specification with sample forms and example data representations: sma.treat-nmd.org, hosted on the TNMD website.

10 min videos developed to support understanding.

- ≥ 1 validated motor outcome measure
- CGI according to clinician and patient
- Family history
- Screening programme & method of testing
- Airway clearance Y/N
- Clinical observations incl. contractures
- Electrophysiology & biomarkers taken (Y/N)
- Date & cause of death
- Participation in other registries or NH studies
- Participation in other registries or NH studies

sma.treat-nmd.org
Any questions?
Year 3 Annual Curators Report

Key Highlights
## Participating Registries

### SMA Dataset Project - Participating Registries (by year)

<table>
<thead>
<tr>
<th>Year</th>
<th>Registries</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pilot Year (2018) n=10</td>
<td>Australia, Belgium, Canada, Egypt NMD, Germany (Munich), India, New Zealand, Slovenia*, UK &amp; Ireland, Ukraine</td>
</tr>
<tr>
<td>Year 1 (2019) n=8</td>
<td>Czech Republic*, Hungary*, Latvia*, Poland, Serbia, Spain, Switzerland, Turkey (KUKAS)</td>
</tr>
<tr>
<td>Year 2 (2020) n=8</td>
<td>Armenia, Bulgaria*, Columbia, Croatia*, Egypt (PED NMD), Georgia, Malaysia, Sweden</td>
</tr>
<tr>
<td>Year 3 (2021) n=4</td>
<td>China, Lebanon*, South Africa*, Iran</td>
</tr>
<tr>
<td>Year 4 (2022) n= tbc</td>
<td>Argentina tbc, Chile tbc, Mexico*, Turkey (LUKAM)</td>
</tr>
</tbody>
</table>

**Key**
- Clinician Reported: 19
- Patient Reported: 9
- Dual Reported: 4
- GRP users*: 9
- Total Participating Registries: 32
Y3 Annual Report Update

Therapy availability*

- None: 17%
- Just one: 11%
- Just two: 24%
- All therapies: 48%

*Data presented is from a curator survey carried out in May 2022 and reflects what registry curators have reported regarding therapy access in their own country. Access is defined as any DMT availability at all irrespective of reimbursement restrictions or route of access.

<table>
<thead>
<tr>
<th>Year</th>
<th>Patients</th>
</tr>
</thead>
<tbody>
<tr>
<td>Y1</td>
<td>4402</td>
</tr>
<tr>
<td>Y2</td>
<td>4806</td>
</tr>
<tr>
<td>Y3</td>
<td>6492</td>
</tr>
</tbody>
</table>
Registry Type & Data collection methods

**Registry Types**

- **Clinician Reported 63%**
- **Patient Reported 17%**
- **Dual Reported 20%**

**Bursary Payments**

<table>
<thead>
<tr>
<th></th>
<th>Part A</th>
<th>Part B</th>
</tr>
</thead>
<tbody>
<tr>
<td>Number of Registries</td>
<td>24</td>
<td>9</td>
</tr>
</tbody>
</table>
Dataset Compliance Levels – Year 3

**Clinicin reported, n=19**

- Mandatory: 70%
- Non mandatory: 75%
- Total compliance: 80%

**Patient reported, n=5**

- Mandatory: 90%
- Non mandatory: 85%
- Total compliance: 90%

**Dual reported, n=6**

- Mandatory: 75%
- Non mandatory: 85%
- Total compliance: 90%
# Most common items reported NOT collected

## Mandatory

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<tr>
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## Non-Mandatory

<table>
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<tr>
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<tr>
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</tr>
<tr>
<td>Head circumference (n=30)</td>
<td>10 (33%)</td>
</tr>
</tbody>
</table>
Increased OM collection… building consensus!
What are the reasons for this?
• Increased therapy availability
• PMS studies
• Requirement for regulators & payers
Any questions?
UK SMA Patient Registry
PROMs data collection & sharing

TREAT-NMD SMA Dataset Workshop
7th December 2022

Registry manager: Lindsay Murphy
lindsay.murphy@newcastle.ac.uk
Date collection in the UK

These three data collection studies work collaboratively to form UK SMA network.
• Registration is patient-initiated.
• Collects *patient-reported* data.
• Active patient choice to participate and control use of their data; enables the patient voice to be captured.
• Available to all SMA patients in the UK & Ireland, independent of clinic.
• Affiliated to the TREAT-NMD SMA Global Registries Network; collects the TREAT-NMD SMA Expanded Core Dataset.
• Communication tool & ideal for collection of Patient Reported Outcome Measures (PROMs).

• Clinical databases for natural history.
• Collect *clinician-reported* data.
• To participate, patients must attend a Neuromuscular Clinic that is part of the SMA REACH networks.
• Apart from consent, patient participation is passive and occurs through their routine clinic visits.
• Networks are actively expanding, however cover is incomplete – therefore participation is not available to all UK SMA patients.
PROMs

• Capture the patient’s perspective on their condition & any changes, their quality of life and activities of daily living
• Inform the revision and improvement of Standards of Care
• Help improve understanding of the impact of SMA therapy, supplementing Clinical Data and Functional Assessments
• PROMs collection and reporting is a requirement for Nusinersen and Risdiplam Managed Access Agreements - inform and assist regulatory authorities determine the efficacy of treatments

- Collected via the UK SMA Patient Registry, implemented in April 2022
- Ask consent for the sharing of data with SMA REACH
- All SMA patients in the UK & Ireland are welcome to register and to complete PROMs questionnaires

www.sma-registry.org.uk/
PROMs Pilot Study

• Collection of PROMs for the Nusinersen & Risdiplam Managed Access Agreements:
  - 50 paediatric Nusinersen patients & 50 adult Nusinersen patients
  - 50 paediatric Risdiplam patients & 50 adult Risdiplam patients

 100 patients via SMA REACH UK
  100 patients via Adult SMA REACH

• PROMs must coincide with six monthly clinic visits & therefore with clinical data collected by SMA REACH

• PROMs Pilot Study was introduced in the first instance to enable coordination between SMA REACH sites & patient registry

• Clinics in PROMs Pilot Study will receive individual, patient PROMs reports

• SMA REACH coordination teams will align PROMs with clinic data, anonymise & report to Regulatory Authorities
PROMs data from the patient registry is shared with clinics in grouped patient reports:

- Sharing of identifiable, patient-level data with each patient’s own neuromuscular clinic to inform patient care (SMA REACH & patient registry consent in place), every ~6 months
- To enable this, sites first inform patient registry of patient ID
- Patient registry will provide all PROMs available for identified patients

PROMs collected

- Quality of Life EQ-5D
- Patient Global Impression of Change (Severity, Improvement)
- SMA Independence Scale (SMAIS) – non-ambulant
- Free-text box

Patient-reported & caregiver-reported versions
PROMs communications

- Regular & frequent communications with Pilot Study sites
- Increased frequency of patient registry automated reminder email from annual to every six months
- Emphasise the importance of reporting of PROMs at the same time as clinical data
- Patient registry PROMs promotional material
- Postcard & business card to be distributed to all sites
- Short animation posted to websites & shared with patient organisations
PROMs – UK SMA Patient Registry case study

https://www.youtube.com/watch?v=9EsZsBb5wb8&t=5s
Registry thoughts on the UK PROMS case study

- Do you think this is a good initiative?
- Would this be useful for your registry?
- Have you delivered any similar communications to patients / clinicians?
- Sharing best practice initiatives – how can improve on this in the future?
SMA Expanded Core Dataset

Group Work (50 mins)
Whova Poll – Results

Poll question
Please rate how easy/difficult your registry found it to adopt V2 of the SMA Dataset?

My response

- Easy: 50%
- Difficult: 25%
- Still not adopted: 25%
- Very Easy: 0%

Only displaying the top results. There were 1 other poll option.
Whova Poll – Results

Poll question: Which elements of the Dataset Specification would you like further clarification/training on?

My response:

- **PROMS/ Outcome Measures**: 70%
- **Mandatory/ Non Mandatory**: 50%
- **Longitudinal**: 40%
- **PPRL Privacy Preserving Record Linkage**: 20%

Only displaying the top results. There were 2 other poll options.
Main dataset challenges

...... feedback from Annual Survey
## Most common items reported NOT collected

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</table>
Feedback from groups

- Identify reasons why specific data items are proving difficult to collect?
- Should some items be re-classified as non-mandatory?
- How can we learn from one another?
What are the benefits/advantages of adopting the V2 dataset (word cloud - Whova poll)
**Group Breakout Session**

**Group A**
What are the key research questions the SMA community want answers to?
How can this lead to publications / poster?

**Group B**
How can the global SMA registry network work better together?

**Group C**
Working with industry/ regulators - share registry experience of what has worked well and not so well

**Group D**
Sharing best practice on patient recruitment strategies and how to make registries more patient centric
Break-out Group feedback
Next Steps (Dec 22 – May 23)

SMA Project Timeline

Dec ‘22
- SMA workshop
- Annual Curators Meeting
- Analyse workshop feedback

Jan ‘23
- Prepare & circulate workshop Report
- Monthly drop-in Sessions
- Continue supporting new/ existing registries
- Review TNMD dataset revision process

Feb ‘23
- Prepare & Issue Curator Surveys
- Confirm adoption of V2 by remaining registries
- Bursary processing
- Support new TNMD registries
- Circulate TNMD revision process for comment to TGDOC

Mar - April 23
- Analyse survey responses
- Registry 1-2-1 calls
- Process Final Bursary payments
- Start final report preparation

May ‘23
- Early May deliver report to Biogen.
- Upload to website
- Close project!
TREAT-NMD Dataset Revision Process

- Need a single co-ordinated strategy for all NMD datasets – implement ‘23

- Dataset harmonisation across NMD's datasets

  ✓ Who should be involved?
  ✓ When should it take place?
  ✓ How long will registries be given to adopt the dataset to retain membership status?

- Does the One size fit’s all model work?

- Multi stakeholder involvement in discussions
Key priorities for 2023

- Outline what support TREAT-NMD can provide to assist with registry data collection
- What future networking events do SMA registry curators want
- Identify the key research questions that need to be asked
- Support for publications / posters for conferences
- Sub-Group Lead Role – seeking nominations (2023)
Whova Poll – Results

Poll question
What support could we provide which would be of most benefit to registries/curators?

My response

- Educational materials on value of data... 33%
- Help to prepare research publications 33%
- More networking events 11%
- Financial 11%

Only displaying the top results. There were 2 other poll options.
Take-home messages!

- Collection of robust, standardised data is essential to building real world evidence to support PMS studies

- Need to balance needs of various stakeholders alongside registry maintenance

- How do we educate patients and clinicians on the value data collection

- Improve networking going forward - work collaboratively improve knowledge sharing and best practice

- Keep a patient focus to registry data collection. Patients focus on patient outcomes – registries should focus on the clinical need AND the patient outcomes and QoL
Thanks for the valuable support from our project sponsor

Please scan to complete the survey!