# TREAT-NMD SMA Registries Core Dataset:

## Version 1 (31 August 2018)

## Suggested wording for patient-reported registries

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# 1. Introduction

TREAT-NMD is a network for the neuromuscular field that provides an infrastructure to ensure that the most promising new therapies reach patients as quickly as possible. Since its launch in January 2007 the network's focus has been on the development of tools that industry, clinicians and scientists need to bring novel therapeutic approaches through preclinical development and into the clinic, and on establishing best-practice care for neuromuscular patients worldwide.

The TREAT-NMD global network of SMA Registries feed into a central hub called the TREAT-NMD Global SMA Registry, which can receive requests from third parties in order to answer research questions. National registries that are part of the Global Registry must collect as a minimum, a standardized core dataset from their patients. The TREAT-NMD Global SMA Registry is governed by a Charter* and by the TREAT-NMD Global Database Oversight Committee (TGDOC). The TGDOC is responsible for reviewing the requests for data from the Global Registry and it votes on whether the request is in line with the Charter and is in patients' best interests.

The core dataset for TREAT-NMD registries collecting data on SMA patients was expanded in September 2018, following a pilot study. This work was done so that the TREAT-NMD Global SMA Registry can:

a. better inform on the natural history of SMA
b. provide context to understand the safety and effectiveness of new treatments
c. support post marketing surveillance (PMS) for those new treatments.

** The TGDOC Charter is currently under review*

## 2. This document

This document:

- provides suggested wording for all items in the expanded SMA dataset which are applicable to patient-reported registries.
- is in draft and we would very much value feedback or suggestions for improvements.
- supports version 1 of the dataset (full documents available on the project web page) which was released in September 2018.
- does not contain the full dataset – only sections/items which are relevant for patient-reported registries.

**Please note we do not intend to mandate that registries must use this wording, however it will be recommended for use in English language registries, and also as a basis for translations into other languages.**

If you have any feedback on the dataset itself (data items, sections, response options etc.) this would also be very welcome - but if you are suggesting new or significantly altered items, please also suggest the patient-facing wording to accompany them.

Please provide all feedback before Friday 13 March 2020, so it can feed into our first dataset revision (draft revision process attached). Version 2 of the dataset (including patient-facing wording) will be confirmed by the end of June 2020. Please note we will be making a strong effort to minimise or avoid significant changes, in order to support version compatibility, longitudinal data analysis, and to reduce burden on registries and patients.

# 3. The Dataset

**PLEASE NOTE:**

- For mandatory data items: TREAT-NMD SMA Registries are required to include these items in their case report forms, and make every effort to collect them (or agree actions to work towards their collection).
- However, the minimum data needed for an individual record to be accepted as valid for a global registry enquiry submission will be defined on a case-by-case basis.

- Registries should ensure that all data entries and updates are date-stamped (and time-stamped if possible)

KEY
Black text = mandatory items
Blue text = highly encouraged items
*Text in italics* = notes for Curators
[Text in square brackets] = System/coding notes

## Section 1: ENROLMENT

| Item no. | Data item description | Coding | Baseline | Follow-up |
|---|---|---|---|---|
| 1.03 | *(To be included in consent form)*<br>Do you give consent for your de-identified (anonymous) data to be shared with the TREAT-NMD Global SMA Registry, for purposes of research and clinical trial planning? | | | |

## Section 2: DEMOGRAPHICS

*Items 2.00-2.16: Registries are encouraged to collect the demographic items in blue for internal use, but only the mandatory items (in black) would ever be requested for central submission.*

^ = *items which support PPRL functionality (Privacy Preserving Record Linkage)*

| Item no. | Mandatory data items | Highly encouraged data items | Coding | Baseline | Follow-up |
|---|---|---|---|---|---|
| 2.00 | What is your date of birth? | | DD-MM-YYYY | X | |
| 2.01 | | What is your first name? | [Free text] | X | X |
| 2.02 | | What was your first name given to you at birth? | [Free text] | X | |
| 2.03 | | What is your last name? | [Free text] | X | X |
| 2.04 | | What was your last name given to you at birth? | [Free text] | X | |
| 2.05 | What is your gender? | | Male; Female; Unspecified | X | X |
| 2.06 | | Is this the same gender that you were assigned at birth? | Male; Female; Unspecified | X | |
| 2.10 | | What is your current address? | [Free text] | X | X |
| 2.11 | | What is your zip/post code? | [Free text] | X | X |
| 2.12 | Which country do you live in? | | [ISO 3 Standards] | X | X |
| 2.13 | | Which country were you born in? | [ISO 3 Standards] | X | |
| 2.14 | | Which city/town were you both in? | [Registry own picklist] | X | |

| | | | | | |
|---|---|---|---|---|---|
| 2.15 | | Please provide an email address that we can use to contact you | [Free text] | X | X |
| 2.16 | | Please provide a telephone number that we can use to contact you | [Numerical value] | X | X |
| 2.20 | | Do you know of any other family member who is diagnosed with SMA? | Yes; No | X | X |
| 2.21 | | If you answered 'Yes' to the previous question, please tell us how they are related to you. | Mother; Father; Daughter; Son; Brother; Half Brother; Sister; Half Sister; Niece; Nephew; Maternal Uncle; Paternal Uncle; Maternal Aunt; Paternal Aunt; Maternal Cousin; Paternal Cousin; Maternal Grandfather; Paternal Grandfather; Maternal Grandmother; Paternal Grandmother; Granddaughter; Grandson [Can add multiple] | X | X |

## Section 4: GENETIC DIAGNOSIS

| Item no. | Mandatory data items | Highly encouraged data items | Coding | Baseline | Follow-up |
|---|---|---|---|---|---|
| 4.00 | Have you had a genetic test which confirmed your SMA diagnosis? | | Yes; No | X | X |
| 4.02 | If you have had a genetic test, please send/upload* a copy of your genetic report, if you have it.<br><br>** Delete as appropriate for your registry* | | [File upload option] | X | X |
| 4.03 | If you have had a genetic test, please tell us the name and location of the genetic testing centre. | | [Free text] | X | X |

## Section 5: CLINICAL OBSERVATIONS

| Item no. | Mandatory data items | Highly encouraged data items | Coding | Baseline | Follow-up |
|---|---|---|---|---|---|
| 5.00 | At what age did your symptoms first appear (or it was first suspected that something might be different)? | | During pregnancy; birth to 0.5 months; at age YY-MM | | |
| 5.01 | Which SMA type has your doctor diagnosed? | | Type 0; Type 1; Type 2; Type 3; Type 4 | | |
| 5.02 | | What is your height in cm? | [Numerical value] | X | X |
| 5.03 | | Method of height measurement | Standing height; Recumbent length; Arm span; Ulnar length; Other (please specify [free text]) | X | X |
| 5.04 | | What is your weight in kg? | [Numerical value] kg | X | X |
| 5.05 | | If you are registering an infant aged 0-23 months, please give their head circumference (in cm)*<br><br>** Registries should provide guidance (from the dataset manual) for parents/carers on how to measure this* | [Numerical value] cm | X | X |
| 5.06 | | If you are registering an infant aged 0-23 months, please give their chest circumference at full expiration (in cm)* | [Numerical value] cm | X | X |

| | | | | | |
|---|---|---|---|---|---|
| | | ** Registries should provide guidance (from the dataset manual) for parents/carers on how to measure this* | | | |
| 5.07 | | If you are registering an infant aged 0-23 months, please give their chest circumference at full inspiration (in cm)*<br><br>** Registries should provide guidance (from the dataset manual) for parents/carers on how to measure this* | [Numerical value] cm | X | X |
| | | Contractures<br>Patients with SMA are often affected by joint contractures, which can be described as limitations in the full range of motion of certain joints. Contractures are caused by immobility, reduced weight bearing, and muscle imbalance. | | | |
| 5.08 | | Do you suffer from shoulder contractures? | Yes; No | X | X |
| 5.09 | | Do you suffer from elbow contractures? | Yes; No | X | X |
| 5.10 | | Do you suffer from wrist contractures? | Yes; No | X | X |
| 5.11 | | Do you suffer from finger contractures? | Yes; No | X | X |
| 5.12 | | Do you suffer from hip contractures? | Yes; No | X | X |
| 5.13 | | Do you suffer from knee contractures? | Yes; No | X | X |
| 5.14 | | Do you suffer from ankle contractures? | Yes; No | X | X |
| 5.20 | Please tell us the name of the neuromuscular specialist or main doctor in charge of your care. | | [Free text] | X | X |
| 5.21 | Please tell us the name and location of your main healthcare centre. | | [Free text] | X | X |

## Section 6: SCOLIOSIS

| Item no. | Mandatory data items | Highly encouraged data items | Coding | Baseline | Follow-up |
|---|---|---|---|---|---|
| | Scoliosis<br>Scoliosis is a common complication in people with SMA. It is characterised by a curvature of the spine (usually to one side) and is caused by a weakening of the muscles that support the spine, as the disease progresses. | | | | |
| 6.00 | Have you been diagnosed with scoliosis? | | Yes; No; Not known | X | X |
| 6.01 | | If you have been diagnosed with scoliosis, please tell us the Cobb angle recorded on the latest radiology results, if you know it. | [Numerical value] degrees | X | X |
| 6.02 | If you have been diagnosed with scoliosis, have you had any surgery for the scoliosis? | | Yes; No | X | X |
| 6.03 | | If you have had surgery for scoliosis, please tell us what kind of surgery you had, if you know. | Arthrodesis; Growing Rods; Other (please specify); Not known | X | X |
| 6.04 | | If you have had surgery for scoliosis, please tell us the date of your surgery (month & year). | MM-YYYY | X | X |

## Section 7: MOTOR FUNCTION

| Item no. | Mandatory data items | Highly encouraged data items | Coding | Baseline | Follow-up |
|---|---|---|---|---|---|
| | Motor function describes your ability to move your body.<br><br>For each of the abilities listed, please choose the appropriate answer.<br><br>*(Curators – definitions of each function are available in the Dataset Manual and should be made available to your registry participants when completing this section.* | | | | |
| 7.00 | Holding head up without support | | *For each motor function item, specify:*<br><br>Never able;<br>Gained [age YY-MM];<br>Gained & lost [age gained YY-MM] & [age lost YY-MM] | X | X |
| 7.01 | Rolling onto side | | | X | X |
| 7.02 | Sitting without support | | | X | X |
| 7.03 | Crawling on hands and knees | | | X | X |
| 7.04 | Standing with assistance | | | X | X |
| 7.05 | Standing alone (without assistance) | | | X | X |
| 7.06 | Walking with assistance | | | X | X |
| 7.07 | Walking alone (without assistance) | | | X | X |
| 7.08 | Able to walk 10 metres unaided | | | X | X |
| 7.09 | Climbing stairs | | | X | X |
| 7.10 | Useful function of hands | | | X | X |
| 7.11 | Reaching overhead in a sitting position | | | X | X |
| 7.12 | Raising hands to mouth in a sitting position | | | X | X |

## Section 8: WHEELCHAIR USE

| Item no. | Mandatory data items | Highly encouraged data items | Coding | Baseline | Follow-up |
|---|---|---|---|---|---|
| 8.00 | For patients 2 years old or older: Do you use a wheelchair? | | No - able to walk independently; Part-time [age began YY-MM]; Full-time [age began YY-MM] | X | X |

## Section 9: NUTRITION

| Item no. | Mandatory data items | Highly encouraged data items | Coding | Baseline | Follow-up |
|---|---|---|---|---|---|
| | SMA patients sometimes have trouble eating and swallowing food orally (by mouth) and therefore have to be fed through a feeding tube. A gastric feeding tube (also called a G-tube or a Peg) is one that goes directly into the stomach through an incision in the tummy. A nasal feeding tube (also called nasogastric tube) is one that goes through the nose and down into the stomach. | | | | |
| 9.00 | Have you ever used a gastric or nasal tube for feeding? | | Never; Previously fed only by tube [start and end date MM-YYYY]; Previously for supplementary feeding e.g. fluids [start and end date MM-YYYY]; Currently fed only by tube | X | X |

| | | | | | |
|---|---|---|---|---|---|
| | | | [start date MM-YYYY];<br>Currently for supplementary feeding e.g. fluids [start date MM-YYYY]; Not known<br>[Can select multiple] | | |

## Section 10: PULMONARY FUNCTION

| Item no. | Mandatory data items | Highly encouraged data items | Coding | Baseline | Follow-up |
|---|---|---|---|---|---|
| | Ventilation means breathing support from a mechanical ventilation device in the form of either **non-invasive ventilation** via a face or nose mask, or **invasive ventilation** via a tracheostomy (an operation to make an incision in the wind-pipe) or endotracheal tube (a breathing tube is inserted into the wind-pipe). Ventilatory support can be used all day or for just a few hours. | | | | |
| 10.00 | Have you ever used invasive ventilation? | | Never;<br>Previously [start date MM-YYYY] and [end date MM-YYYY];<br>Currently [start date MM-YYYY];<br>Not known | X | X |
| 10.01 | If you have used invasive ventilation, how frequently? | | Full-time; part-time; Not known | X | X |
| 10.02 | If you have used invasive ventilation, when did this start? | | MM-YYYY | X | X |

| | | | | | |
|---|---|---|---|---|---|
| 10.03 | Have you ever used non-invasive ventilation? | | Never; Previously [start date MM-YYYY] and [end date MM-YYYY]; Currently [start date MM-YYYY]; Not known | X | X |
| 10.04 | If you have used non-invasive ventilation, how frequently? | | Full-time; part-time; Not known | X | X |
| 10.05 | If you have used non-invasive ventilation, when did this start? | | MM-YYYY | X | X |
| 10.06 | | Do you receive help with clearing out the secretions (e.g. mucus) that build up in your airways and lungs? This could be a manual technique, like chest physiotherapy, or a mechanical support such as a Cough Assist device. | Yes; No | X | X |
| | If you answered yes to the previous question, please tell us what kind of help you receive (you can select more than one). For each one, please tell us how often you receive it. | | | | |
| 10.07 | | Suction | Daily; Weekly; Occasionally | X | X |
| 10.08 | | Chest percussion | Daily; Weekly; Occasionally | X | X |
| 10.09 | | Cough Assist device | Daily; Weekly; Occasionally | X | X |
| 10.10 | | IPPV (Intermittent positive-pressure ventilation) | Daily; Weekly; Occasionally | X | X |
| 10.11 | | Other (Please Specify) | Daily; Weekly; Occasionally | X | X |
| 10.12 | To monitor your breathing, a Forced Vital Capacity (FVC) test may have been done. To test FVC, you | | Yes; No; Not known | X | X |

| | | | | | |
|---|---|---|---|---|---|
| | would have been asked to breathe in as far as you can, and then blow out into a machine that measures how much air is being exhaled. Have you had a Forced Vital Capacity (FVC) test? | | | | |
| 10.13 | If you have had a FVC test, when was your most recent one? | | DD-MM-YYYY | X | X |

## Section 11: THERAPIES AND MEDICATIONS

| Item no. | Mandatory data items | Highly encouraged data items | Coding | Baseline | Follow-up |
|---|---|---|---|---|---|
| 11.01 | Have you ever received Spinraza?<br>*(Question will be modified in future if additional therapies are approved)* | | Currently; Previously; Never | X | X |
| 11.11 | Have you taken any other prescribed drugs in the last 12 months *(first entry)* / since you last updated your data *(follow-up)*? | | Yes; No; Not known | X | X |
| 11.12 | If you have taken other prescribed drugs, please tell us which ones if you know (you can select more than one).<br><br>** Please note; the inclusion of a supplement in this list does not necessarily indicate TREAT-NMD endorsement.* | | Bone health: Vitamin D; Calcium; Biphosphonate; Gastro intestinal system: Drugs for gastroesophageal reflux; Drugs for constipation; Respiratory system: Antibiotics; Anticholinergic drugs; | | |

| | | | | | |
|---|---|---|---|---|---|
| | | | Immunisations: Annual influenza immunizations; Annual pneumococcal immunizations; Supplements*: Creatine; Acetyl-L-carnitine; Phenylbutyrate; Gabapentin; Thyrotropin-releasing hormone; Hydroxyurea; Valproate; Albuterol; Other (please specify [free text]) [Can add multiple] | | |
| 11.13 | | *For each drug selected in 11.12:* Start date (month & year) | [MM-YYYY] | X | X |
| 11.14 | | *For each drug named in 11.12:* Stop date (month & year) if not ongoing | [MM-YYYY] | X | X |
| 11.20 | Have you received any of these therapies in the last 12 months *(first entry)* / since you last updated your data *(follow-up)*? (You can select more than one) | | Physiotherapy sessions (e.g. stretches); Respiratory physiotherapy sessions; Massage; Home | X | X |

| | | | | | |
|---|---|---|---|---|---|
| | | | programme (e.g. stretches/exercises); Hydrotherapy/water-based activity; Management of contractures using orthotics (e.g. ankle foot orthoses); Brace; Occupational therapy sessions / input for home or equipment; Speech and language therapy sessions; Other (please specify [free text]) | | |

## Section 12: HOSPITALISATIONS AND COMORBIDITIES

| Item no. | Mandatory data items | Highly encouraged data items | Coding | Baseline | Follow-up |
|---|---|---|---|---|---|
| 12.00 | Have you been admitted to hospital in the last 12 months *(first entry)* / since you last updated your data *(follow-up)*? | | Yes; No; Not known | X | X |
| 12.01 | If you answered 'Yes' to the previous question, was it a planned hospital visit, or was it | | Planned; Unexpected | X | X |

| | | | | | |
|---|---|---|---|---|---|
| | unexpected? (If you have had more than 1 hospital visit, please add each one.) | | [Can add multiple] | | |
| 12.02 | *For each hospitalisation in 12.01*: When were you admitted for this visit? (month & year) | | [MM-YYYY] | X | X |
| 12.03 | *For each hospitalisation in 12.01*: How many days did you stay in hospital? | | [Numerical value] | X | X |
| 12.10 | In the last 12 months *(first entry)* / Since you last updated your data *(follow-up)*, have you been diagnosed with any other illnesses or conditions not related to your SMA? | | Yes; No; Not known | X | X |
| 12.11 | If you answered yes to the previous question, please tell us what you have been diagnosed with? You can add more than one if needed. | | [Free text]<br><br>[Can add multiple] | | |
| 12.12 | *For each Comorbidity:* Please tell us when this started | | [MM-YYYY] | X | X |
| 12.13 | *For each Comorbidity:* Please tell us when this ended (leave blank if you still have this condition) | | [MM-YYYY] | X | X |

## Section 13: CLINICAL RESEARCH

| Item no. | Mandatory data items | Highly encouraged data items | Coding | Baseline | Follow-up |
|---|---|---|---|---|---|
| 13.00 | Have you ever participated in a clinical trial? | | Yes I am currently participating in a clinical trial; Yes I have previously participated in a clinical trial; No I have never participated in a | X | X |

| | | | | | |
|---|---|---|---|---|---|
| | | | clinical trial; Not known | | |
| 13.01 | Please tell us the name of the trial/trials, if you can remember. You can add more than one if needed. | | [Free text] [Can add multiple] | X | X |
| 13.02 | *For each trial named in 13.01:* Please tell us the name of the drug that you received during this trial, if you can remember. | | [Free text] | X | X |
| 13.10 | | Are you currently part of any other registry and/or natural history study? | Yes; No; Not known | X | X |
| 13.11 | | *If 'Yes' to 13.10:* Please tell us which one (you can add more than one if needed) | [Free text] [Can add multiple] | X | X |

## Section 15: PATIENT-REPORTED OUTCOMES (PRO)

| Item no. | Mandatory data items | Highly encouraged data items | Coding | Baseline | Follow-up |
|---|---|---|---|---|---|
| 15.02 | How do you feel that your condition has changed in the last 6 months? | | 1=Very much improved; 2=Much improved; 3=Minimally improved; 4=No change; 5=Minimally worse; 6=Much worse; 7=Very much worse | X | X |
| 15.03 | *Curator should ensure that the date of the above score is recorded* | | DD-MM-YYYY | X | X |

| | | | | | |
|---|---|---|---|---|---|
| | Patient-reported registries may also choose to incorporate a patient-reported outcome measure (PROM) into their questionnaires. Validated PROMs known to TREAT-NMD are listed below, and registries would be asked to share with the Global Registry the score and date for each patient who has reported the PROM.<br><br>Selection of an appropriate PROM is at the discretion of the individual registry. Further information about each one will be available in the Outcome Measures Toolkit due for completion in May 2020. | | | | |
| 15.11 | | **PedsQL**<br>*(Paediatric Quality of Life Inventory)* | | X | X |
| 15.13 | | **PEDI-CAT**<br>*(Pediatric Evaluation of Disability Inventory - Computer Adaptive Test)* | | X | X |
| 15.15 | | **SMA FRS**<br>*(Spinal Muscular Atrophy Functional Rating Scale)* | | X | X |
| 15.17 | | **ACEND**<br>*(Assessment of Caregiver Experience with Neuromuscular Disease)* | | X | X |
| 15.19 | | **ACTIVLIM**<br>*(ACTIVLIM measure of activity limitations)* | | X | X |
| 15.21 | | **DISABKIDS**<br>*(DISABKIDS measurement of quality of life and level of distress)* | | X | X |

# 4. Feedback, Harmonisation & Revisions

The TREAT-NMD Global SMA Registry is one of several notable data collection initiatives in operation across the world. Harmonisation and comparability across these different data repositories is vital to ensure that the collected data meets the current and future needs of the SMA community.

Considerable work has gone into ensuring that the data collected by the TREAT-NMD registries, through this first iteration of the expanded core dataset, will be comparable with the data collected by other initiatives. However, many of these initiatives are still in development, and in addition to this, global consensus on the most appropriate and relevant data to collect is expected to continue to evolve over the coming years.

The TGDOC are committed to the continued harmonisation of the TREAT-NMD SMA Core Dataset with other data collection initiatives, and to the evolution of the dataset in response to the needs of the SMA community. Therefore, an **formal revision process** has been developed, which will allow suggested revisions to be accepted throughout the year, and considered and implemented (if appropriate) on a streamlined basis.

The first revision is planned to commence in March 2020 resulting in version 2 of the dataset in June 2020. Please note, we do not anticipate that these changes will be significant, and we are equally keen to limit the burden on the registries who will need to implement any changes made.

If you have any questions or feedback on the dataset, please contact joanne.bullivant@newcastle.ac.uk.