TREAT-NMD SMA Registries Core Dataset: Overview

- This is a high-level overview of the TREAT-NMD SMA Registries Core Dataset. It is supported by, and should be used in the context of, the following documents:
    - "**TREAT-NMD SMA Registries Core Dataset**": Contains the full detailed dataset, including guidance on question structure and multiple choice answers.
    - "**TREAT-NMD SMA Registries Core Dataset Manual**": Contains full definitions and guidance on data collection.

- **Toolkits** are in development to support affiliated registries in the selection and collection of motor measures and patient-reported outcomes.

- Any anticipated **training or support requirements** should be reported to joanne.bullivant@newcastle.ac.uk.

PLEASE NOTE:

- TREAT-NMD SMA Registries are required to include the mandatory 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 each global registry enquiry 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
Items in black text are mandatory
Items in blue are highly encouraged
^{CR} = mandatory only for clinician-reported registries
^{PR} = mandatory only for patient-reported registries

Contents

Black = mandatory
Blue = highly encouraged

[CR] = mandatory only for clinician-reported registries
[PR] = mandatory only for patient-reported registries

	MANDATORY ITEMS	HIGHLY ENCOURAGED ITEMS
	1. Enrolment	
1.00	Date of enrolment	
1.01	Date of consent (if different from enrolment)	
1.02	Date of any re-consents	
1.03	Consented to TNMD global registry?	
1.04	Local registry ID	
	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 Protecting Record Linkage)*	
2.00	Date of birth	
2.01		First name
2.02		First name at birth (if different) ^
2.03		Last name
2.04		Last name at birth (if different) ^
2.05	Sex	
2.06		Sex at birth (if different) ^
2.10		Address
2.11		Zip/post code
2.12	Country of residence	
2.13		Country of birth ^
2.14		City/town of birth ^
2.15		Email address
2.16		Telephone number
2.20		Any other family member affected?
2.21		If Yes; state kinship
	3. Living status	
3.00	Patient alive?	
3.01		Date of death
3.02		Cause of death
	4. Genetic Diagnosis	
	[◊] = *In patient-reported registries, items marked with [◊] should be reported by the Registry Curator/Coordinator, following review of the patient's genetic report.*	
4.00	[◊] Genetic confirmation of SMA?	
4.01		If yes, was it through screening?
4.02	[PR] Send/upload copy of genetic report	

4.03	Name/location of genetic testing centre	
4.04	◊ Date of genetic diagnosis	
4.05	◊ Mutation name in SMN1 gene	
4.06		◊ Method of SMN1 testing
4.07	◊ Was SMN2 Copy number tested?	
4.08		◊ Method of SMN2 testing
4.09	◊ SMN2 copy number	
	5. Clinical Observations	
5.00	Age of symptom onset	
5.01	Spinal Muscular Atrophy type	
5.02		Height/length (cm)
5.03		Method of height measurement
5.04		Weight
5.05		Head circumference Δ
5.06		Chest circumference at full expiration Δ
5.07		Chest circumference at full inspiration Δ
		Δ *= for infants <24 months old*
		Contractures:
5.08		Shoulder
5.09		Elbow
5.10		Wrist
5.11		Finger
5.12		Hip
5.13		Knee
5.14		Ankle
5.20	PR Name of NM specialist or main doctor in charge of care	
5.21	PR Name/location of main healthcare centre	
	6. Scoliosis	
6.00	Scoliosis diagnosis?	
6.01		If 'Yes': Cobb angle
6.02	If Yes; has had Scoliosis surgery?	
6.03		If Yes; Surgery technique
6.04		If Yes; Date of first surgery
	7. Motor Function	
	Best and Current Motor Function: For each motor function, specify: -Never able -Gained (age gained) -Gained & lost (ages gained & lost)	

	-Observed in clinic; Reported by patient/Caregiver	
7.00	Holding head up without support	
7.01	Rolling onto side	
7.02	Sitting without support [WHO]	
7.03	Crawling on hands and knees [WHO]	
7.04	Standing with assistance [WHO]	
7.05	Standing alone (without assistance) [WHO]	
7.06	Walking with assistance [WHO]	
7.07	Walking alone (without assistance) [WHO]	
7.08	Able to walk 10 metres unaided	
7.09	Climbing stairs	
7.10	Useful function of hands	
7.11	Reaching overhead in a sitting position	
7.12	Raising hands to mouth in a sitting position	
	8. Wheelchair use	
8.00	Wheelchair use (for patients ≥ 2 years old)	
	9. Nutrition	
9.00	Gastric or nasal feeding tube use	
	10. Pulmonary function	
10.00	Invasive ventilation use	
10.01	Invasive ventilation frequency	
10.02	Invasive ventilation start date	
10.03	Non-invasive ventilation use	
10.04	Non-invasive ventilation frequency	
10.05	Non-invasive ventilation start date	
10.06		Assistance in airway clearance and secretion mobilisation?
		If 'Yes'; Types of assistance
10.07		Suction
10.08		Chest percussion
10.09		Cough Assist device
10.10		IPPV (Intermittent positive-pressure ventilation)
10.11		Other (Please Specify)
10.12	Forced Vital Capacity (FVC) test done?	
10.13	FVC date	
10.14	[CR] FVC litre	
10.15	[CR] FVC predicted %	

Black = mandatory
Blue = highly encouraged

[CR] = mandatory only for clinician-reported registries
[PR] = mandatory only for patient-reported registries

	11. Therapies & medications	
11.00	[CR] Disease-modifying therapy for SMA?	
11.01	[PR] Receiving Spinraza?	
11.02	[CR] Name of drug	
11.03	[CR] Start date	
11.04	[CR] Stop date if not ongoing	
11.05	[CR] Reason for stopping	
11.06	[CR] Dosage	
11.07	[CR] Frequency	
11.08	[CR] Route of administration	
11.09	[CR] Following current recommended dosing schedule?	
11.10	[CR] If 'No': Reason	
11.11	Prescribed allopathic drugs?	
11.12	Name of drug	
11.13		Start date of drug
11.14		Stop date of drug if not ongoing
11.20	Therapeutic interventions	
	12. Hospitalisations & comorbidities	
12.00	Any hospitalisations?	
12.01	Initial type of hospitalisation (planned/acute)	
12.02	Admission date (for each hospitalisation)	
12.03	Number of days in hospital (for each hospitalisation)	
12.04	[CR] Reason for each acute hospitalisation	
12.05	[CR] Reason for each planned hospitalisation	
12.06	[CR] For each acute hospitalisation: also reported as an SAE?	
12.07	[CR] If 'Yes' to 12.06; for which medication?	
12.10	Other co-morbidities?	
12.11	Comorbidity details	
12.12	Comorbidity start date	
12.13	Comorbidity end date if not ongoing	
12.14	[CR] For each comorbidity: also reported as SAE?	
12.15	[CR] If 'Yes' to 12.14; for which medication?	
12.20	[CR] In addition to hospitalisations, co-morbidities or death already recorded: any other SAEs reported?	
12.21	[CR] If 'Yes' to 12.20; for which medication?	

	13. Clinical Research	
13.00	Ever participated in clinical trial?	
13.01	Name of trial	
13.02	Name of drug	
13.10		Part of other registry and/or natural history study?
13.11		If 'Yes'; Please specify
	14. Motor Measures	
	*Clinician-reported registries only. Registries are required to collect a minimum of **1 validated motor measure**, in addition to the previous mandatory motor function question (section 7).* *Selection of appropriate motor measure(s) is at the discretion of the clinician and/or preference of the patient. Where there is no pre-existing preference, the measures marked with * are suggested by TREAT-NMD, based on current Standards of Care and prior use in Clinical Trials.*	
14.00	[CR] Validated motor measure(s) taken?	
14.01	[CR] If 'No'; give reason	
	[CR] If 'Yes'; provide relevant details:	
		Infantile onset SMA:
14.10		* CHOP-INTEND score
14.11		* CHOP-INTEND date
14.12		* HFMS score
14.13		* HFMS date
14.14		* HFMS-E score
14.15		* HFMS-E date
14.16		HINE Section 2 score
14.17		HINE Section 2 date
14.18		Observed WHO score
14.19		Observed WHO date
14.20		Other validated measure (specify)
14.21		Other validated measure score
14.22		Other validated measure date
		Later onset SMA:
14.30		* HFMS-E score
14.31		* HFMS-E date
14.32		* RULM score

14.33		* RULM date
14.34		Brooke score
14.35		Brooke date
14.36		Revised Brooke score
14.37		Revised Brooke date
14.38		MFM score
14.39		MFM date
14.40		6MWT score
14.41		6MWT date
14.42		10MWT score
14.43		10MWT date
14.44		TUG score
14.45		TUG date
14.46		Egen Klassification score
14.47		Egen Klassification date
14.48		Observed WHO score
14.49		Observed WHO date
14.50		CHOP-ATEND score
14.51		CHOP-ATEND date
14.52		Other validated measure (specify)
14.53		Other validated measure score
14.54		Other validated measure date
	15. Patient-Reported Outcomes (PRO)	
15.00	[CR] Clinical Global Impression of Severity (CGI-S) - baseline only	
15.01	Date of CGI-S score	
15.02	Total Global Impression (TGI) according to patient/parent	
15.03	Date of patient/parent TGI score	
15.04		TGI according to clinician – follow up only
15.05		Date of clinician TGI score
15.10	Other validated PRO taken?	
	If 'Yes'; provide relevant details:	
15.11		PedsQL (NM & fatigue scales) score
15.12		PedsQL (NM & fatigue scales) date

Black = mandatory
Blue = highly encouraged

[CR] = mandatory only for clinician-reported registries
[PR] = mandatory only for patient-reported registries

15.13		PEDI-CAT score
15.14		PEDI-CAT date
15.15		SMA FRS score
15.16		SMA FRS date
15.17		ACEND score
15.18		ACEND date
15.19		ACTIVLIM score
15.20		ACTIVLIM date
15.21		DISABKIDS score
15.22		DISABKIDS date
15.23		Other validated PRO (specify)
15.24		Other validated PRO score
15.25		Other validated PRO date
	16. Electrophysiology and biomarkers	
16.00		CMAP done?
16.01		DEXA done?
16.02		Muscle imaging done?

The TREAT-NMD Global Database Oversight Committee (TGDOC) intend to review this dataset annually and feedback is welcomed. Please send suggestions to joanne.bullivant@newcastle.ac.uk.