TREAT-NMD DMD Core Dataset

Version 1.2

Please see the online version of this specification for up-to-date information and example forms.

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Introduction

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About

Scope of the dataset specifications

This dataset specification aims to leave registries as much flexibility as possible for their individual considerations and local requirements, whilst providing clear guidance and precise requirements wherever necessary. Several things are deliberately not restricted by this dataset:

- The dataset does **not** prevent registries from collecting further data. They are free to include any items not listed here.
- The dataset does **not** specify a data collection form. Although example forms are given to illustrate the dataset and provide a starting point for implementation, registries are free to choose the structure and wording for their forms as they see fit. There are often multiple ways to capture some information and registries should identify the best solution for their requirements.
- The dataset does **not** require registries to use a specific storage format or database structure. Although the items and records suggest a structure which is also used in the interactive example forms, registries will usually need to store further data. For example, many questions on a data collection form will have an Unknown option which is important for registries to track form completion, but which is not specified in the dataset. Similarly, it is often helpful to include further questions such as Was an anti-AAV9 antibody test performed? (yes/no) which lead to further questions being shown, even though the yes/no question does not correspond to an item in the dataset.

Conventions

This document describes the requirements for a registry conforming to the TREAT-NMD core datasets. TGDOC registries *must* inform TGDOC if they are not conforming to any mandatory requirements. Several keywords are used throughout the document to describe these requirements. When set in italics (e.g. *should*), the keywords are defined as follows:

- *must* or *required* or *shall*: specifies an absolute requirement for any registry conforming to this dataset
- *must not* or *shall not*: specifies an absolute prohibition for any registry conforming to this dataset

[1] <https://datasets.treat-nmd.org/introduction/videos#introduction>
[2] <https://datasets.treat-nmd.org/introduction/videos#items>
[3] <https://datasets.treat-nmd.org/introduction/videos#records>

- *should* or *recommended*: specifies a recommendation that conforming registries should generally adhere to, but which may be disregarded if there are valid reasons to do so
- *should not* or *not recommended*: specifies something that conforming registries should generally not do, but this advice may be disregarded if there are valid reasons.
- *may* or *optional*: specifies a possibility which is truly optional and which a registry may freely decide to follow or not

These definitions closely follow the internet standard RFC 2119[4], but use italics instead of upper case to ease reading.

- Individual refers to the registered person with the neuromuscular disease.
- Caregiver refers to the parent or legal guardian who is providing responses on behalf of the individual.
- Clinician refers to any healthcare professional working in clinic (for example physician, physiotherapist) who is providing data on behalf of individuals.
- Patient-reported (PR) registries: all data are collected directly from patients with no data provided by clinicians. Data should be checked/verified wherever possible by the Registry Curator or team (for example by reviewing clinical notes or reports if available).
- Clinician-reported (CR) registries: all data are provided via clinicians with no data collected directly from patients. Data could be entered into the registry directly by the clinicians, or by Registry Curators/other staff after review of clinical notes/reports or other health record systems.
- Dual-reported (DR) registries: Some data in the registry are collected directly from patients, and some data are provided by clinicians as described above.
- In this dataset specification, baseline or baseline entry always refers to the first data recorded for any given item or record, for an individual in the registry. Update refers to any subsequent data entries.

This dataset specification often uses the term collect or capture, for example registries must collect the surgery date. Collecting or capturing some information means including a question in a data collection form (or gathering it by other means) and having the possibility to store and use the information. It does not mean that this information *must* be present for all individuals. Certain things may be unknown to a data provider or may not be applicable in a certain context, and this should not interfere with the collection or usage of any other data.

A **group** is a collection of related items and records; previously called a Section in version 1 of the SMA dataset. Although this specification attempts to present the groups and their components in a logical order, neither the order nor the grouping itself is binding. A registry *may* use the groups and the order provided here as guidance for their data collection forms, but *may* also choose any other grouping or order. This also applies to the order of items within a record (see below for an explanation of items and records).

When this document refers to the ID of an item or record, the ID is set in a monospace font (e.g. `Scoliosis surgery`).

Data submission to TREAT-NMD

Schedule: Currently registries are asked to submit data to TREAT-NMD on an ad-hoc basis; when needed to respond to a 3rd party enquiry into the global registry.

Data: Currently registries are only asked to provide aggregate data (e.g. patient numbers, often stratified, against a specified number of data items).

[4] <https://tools.ietf.org/html/rfc2119>

The above conditions may change in the future if we are required to work with patient-level data as part of a specific postmarketing study, but this would not become a universal mandated requirement for all registries.

Method: Currently registries are asked to provide requested data by emailing it in an Excel spreadsheet. This will be made safer and more efficient in future with the development of the Global Registries Platform (GRP).

Identifiable personal data such as the name, address or contact details of registry participants will **never** be requested nor accepted by TREAT-NMD for central submission. If registries choose to collect these data locally they *must* ensure they are stored and processed according to relevant data protection legislation.

Items

An **item** (also called *data element* in other contexts) describes a single piece of information, for example the date of symptom onset. Often, an item will correspond to a single question in a data collection form. An item has a descriptive yet short name that is unique among all items in this dataset, for example `Symptom onset date`. Items are indicated with the symbol ☑ Note that the item name is not intended to serve as a complete definition of the item as some details may be omitted for brevity. Complete definitions are included underneath each item name.

An **item value** or simply **value** is a piece of information related to a certain individual. For example `2016-05` is a (partial) date which could describe the symptom onset date for an individual. Its precise meaning, for example that it describes the onset of the first symptoms related to the individual's neuromuscular condition, is described by the item it is associated with. An item value may have various formats or representations; for example, this date could be stored in a registrys database in the international format `2016-05`, but entered into their data collection form by selecting the month May and the year 2016 in drop-down menus.

Inclusion

A **mandatory** item means that designated registries (see below) *must* collect it (that is, include it in their data collection forms and be able to store and provide the data). They are marked within the document as follows:

- CR items are *mandatory* for clinician-reported registries
- PR items are *mandatory* for patient-reported registries
- Dual-reported registries should interpret this based on who reports each item in their registry.

Note that a mandatory item in the dataset does not always correspond to a mandatory field in a data collection form. It is always possible that the data provider does not know the value of a mandatory item. Although registries *should* make appropriate efforts to obtain the data for mandatory items, missing values generally do not preclude any individual's data from being submitted or considered in enquiries.

In addition to the appropriate mandatory items, each registry is encouraged to collect the other items in the dataset if they are relevant and feasible at the local level.

This dataset is not restrictive. Registries in the TREAT-NMD network are independent and as such are free to collect additional data according to their needs or priorities.

Item types

For each item, this dataset specifies an item type which is one of the following:

- **yes/no**: An item with this type has two possible values: a positive value, often denoted as yes or true, and a negative value, often denoted as no or false. A registry *may* choose any label for the positive/negative

values that are appropriate in the respective context. Depending on the context, a yes/no item may be implemented in a form using radio buttons, a dropdown menu, a checkbox. As noted below, a selection option for unknown values *may* be included.

- **decimal**: A number which may include a decimal point. A decimal item is generally implemented in forms using a text box together with the proper validation. Decimal items always specify a unit or scale, where the unit is generally an SI unit (International System of Units) such as kilograms or centimetres. A registry *may* collect values using different units, but *must* be prepared to convert them to the specified unit for data submission and analysis.
- **integer**: A whole number. An integer item is generally implemented using a text box with proper validation.
- **date**: A point in time that *must* be captured by registries with a minimum resolution of month and year. Note that TREAT-NMD will only ever request and accept partial dates consisting of a month and year for submission and enquiries. Depending on the item and context, registries *may* collect date items by asking for the individual's age at that time. In this case, either the age can be stored or the age converted to a date using the individual's date of birth. In any case, registries *must* be able to provide dates, but also be able to provide data for queries using ages. A data collection form could use an input box, dropdown menus or a calendar widget for inputting these data. If an input box is used, registries *may* use a localised date format depending on the language and location of the users, for example the format day/month/year for the UK. However, registries *must* be able to provide dates according to the international standard ISO 8601[5] in a format such as year-month-day. Whenever dates are exchanged with TREAT-NMD, this standard will be used.
- **single selection**: For each single selection item, all possible values are listed in this specification. Exactly one of those values *must* be collected (or no value at all). In a form, such items can be implemented using radio buttons or dropdown menus. Not all possible values listed here must be offered to users; registries *may* restrict the options depending on the item and the context. As noted below, a selection option for unknown values *may* be included. Registries *may* choose any order of the values in their data collection forms. Furthermore, the value of a single selection item *may* also be derived and not explicitly asked for, depending on the structure of the form. For example, a form may include separate sections for each type of disease-modifying therapy; then the value of the item `DMT` would not need to be explicitly asked for, but would be determined by which section of the form is filled out.
- **multiple selection**: For a multiple selection item, everything stated for single selection items also applies. However, multiple values may be collected. The order of the collected values is irrelevant, and each possible value may only be provided once. Multiple selection items can be implemented using checkboxes or listboxes.
- **free text**: A string of characters which has no restrictions on its format. Free text fields are usually implemented using text boxes.
- **restricted text**: A string of characters with some restrictions on the format, for example `Country of residence` where the value must be a two-letter country code. The implementation in a data collection form depends on the item and the technical possibilities.

Unknown and missing values

For any item, the value may be unknown to the data provider or missing in a registry for another reason (for example, because the form was not yet completed). Therefore, this specification acknowledges that in a TREAT-NMD enquiry or data submission, there may be no value for any item, even if it is mandatory. The precise way to indicate a missing value in a TREAT-NMD enquiry or data submission (for example by leaving

[5] <https://en.wikipedia.org/wiki/ISO_8601>

the cell in an Excel spreadsheet blank) will be specified for each submission.

Since this applies to all items, this dataset contains no explicit values to indicate unknown or otherwise missing values. For this reason, all `Unknown` values have been removed in version 2. However, a registry *may* add a selection option in their data collection forms for an unknown value wherever they deem appropriate. This allows a registry to track whether a data provider does not have a certain piece of information, or has merely not completed the form fully. This is important for data curation, but since curation is performed independently by the local registries, this distinction is not relevant for any analyses of data submitted to TREAT-NMD and is therefore outside the scope of this core dataset.

Wherever registries offer a selection option for an unknown value, any appropriate label may be chosen, for example Unknown or I don't know. To encourage registry users to return and complete any unknown values as soon as possible, registries may also choose to use To be confirmed where appropriate. Moreover, patient-reported registries may choose to include an option I do not wish to disclose for potentially sensitive questions in their data collection forms.

Enumerated values

For single selection and multiple selection items, all possible values are enumerated in the dataset specification.

Value ID

The value ID is a concise, stable and descriptive text that uniquely identifies a certain value for a certain item. Whenever providing data to TREAT-NMD, the value ID *must* be used. Like an item ID, a value ID is always in English and *must not* be translated into a local language. Furthermore, it is *recommended* that registries use the value IDs provided in the dataset specifications as internal identifiers in their registry platforms as well.

Description

The description of a value is sometimes the same as the value ID, but often it is longer in order to provide a precise and more comprehensible definition of the value. Registries *may* use the provided descriptions as labels in their data collection forms, but this is not required. In particular, registries which do not use English forms *should* of course use labels in their respective local language. Furthermore, the descriptions are generally worded for curators and clinicians, so patient-reported registries *should* adapt the wording where necessary to ensure that is easy to undertand for patients and their families.

Classification

For some values, the datasets provide a mapping to a classification such as the Human Phenotype Ontology[6] (HPO) or ORPHAcodes[7]. These mappings are provided as a convenience as the clasification may provide further information, synonyms that can be used as labels, and insight into why a certain definition was chosen in the dataset. Note that this mapping is only applies to one direction; i.e., if a certain value applies to a individual, then the mapped term applies, but not necessarily the other way around, because the value in the datset may be more specific than the term in the classification. For example, the item `LGMD type` in the LGMD dataset contains the values `LGMD D5` which denotes a dominant Bethlem myopathy as well as `LGMD R22` which denotes a recessive Bethlem myopathy. However, there exists only one ORPHAcode (ORPHA:610[8]) for Bethlem myopathy, so both values in the dataset are mapped to the same ORPHAcode. If one only knows that

[6] <https://hpo.jax.org/app/>
[7] <https://www.orpha.net/>
[8] <https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=610>

ORPHA:610 applies to a certain individual, the mapping in the dataset does not provide a unique value for the item LGMD type.

Deprecation

Some values are marked as deprecated since a certain version of the dataset. For example, version 1 of the SMA dataset specified Part-time as a possible value for ventilation duration, while version 2 provides the more fine-grained values Part-time awake and sleeping and Part-time sleeping. Registries who had implemented version 1 of the dataset will have the value Part-time recorded in many cases and this data should be used whenever required. But for collection of new data, it should no longer be provided as a possible option and is therefore marked as deprecated.

Longitudinal items

Longitudinal items are marked with the symbol Longitudinal For a given patient, many values for a longitudinal item can be collected over time. For each of these values, a datestamp which denotes the date the value refers to (for example, the date of a measurement) *must* be saved. For use in TREAT-NMD enquiries and data submission, it is sufficient that the datestamp has only the month and the year.

An example of a typical longitudinal item is Weight, which should be collected at each visit or registry update. When analysing the data, it is important to know when each weight measurement was made. When performing a registry update, either by the individual themselves or by a clinician after an examination, the date of the measurement will be equal to or approximately equal to the date of the registry update. In such cases, in particular when a data collection form contains questions such as What is your current weight?, a registry *may* automatically set the datestamp of a value to the entry date. If all values of a certain item are known to refer to the date of entry, a registry *may* not explicitly store any value date for this item at all and instead use the entry date whenever a value date is asked for.

However, when historical data is added, in particular at the baseline registration, the date of the measurement may be significantly earlier than the date of the entry. In such cases, registries *must* allow for the date to be entered explicitly.

In an example registry, the weight values may be stored in a table similar to the following one:

Patient ID	Date	Weight
1	2015-03	38.2
1	2016-04	43.8
2	2014-09	63.0
2	2016-09	61.5

For each patient, there exist multiple rows in this table where each row corresponds to one weight measurement.

Further examples of longitudinal items are Scoliosis diagnosis and Cobb angle. For Cobb angle the date of the measurement (that is, the radiology examination) will often be different from the entry date. In the example registry, the values for those items could be stored in the same table:

Patient ID	Date	Weight	Scoliosis diagnosis	Cobb angle
1	2015-03	38.2	no	
1	2016-01			8
1	2016-04	43.8	yes	
2	2014-09	63.0	no	
2	2016-09	61.5	no	

For patient 1, the positive scoliosis diagnosis was entered in April 2016. The radiology examination in which the Cobb angle was measured was performed in January 2016. Therefore, the row with the date 2016-01 only has the value of the Cobb angle, while the cells for the other items are blank; and similarly, the other rows have no value for the Cobb angle.

For a clinician-reported registry, the datestamp of a longitudinal value *should* usually be the date when the clinical examination on which the entry is based was performed. Exceptions are noted in the description of some items: For example, the datestamp of the Cobb angle *must* be the date of the radiology examination on which it is based. This is particularly important for baseline entries, when older data may be entered. If the precise date for a value is not available but the year is known, it *may* be specified using only the year. But if no information about a date is known, it *must* be omitted.

For patient-reported registries, the datestamp of a longitudinal value generally *should* be the date of the registry entry, with the same exceptions as for clinician-reported registries.

Datestamped items

A small number of items require datestamps, but no historical values; for example `Is family member affected`. Although this item should be collected on every update, only the latest value is relevant. However, it is still important to know when that value was last updated. Such items are called datestamped and marked with Datestamped For any such item, registries *must* save the date on which the value was last updated or marked as up-to-date. As with the datestamp for longitudinal items, only the month and the year are *required* for analysis.

Creation and modification timestamps

In addition to the datestamps explicitly required, registries *should* save the date and possibly time of each entry and value modification, ideally together with audit information such as which user performed the change. Registries *should* use a data collection platform which automatically provides this functionality. These timestamps support auditing data entries and ensuring data quality, but may also serve as a fallback if no explicit datestamp is available for a certain value. For example, if a certain value for the item `Cobb angle` has no date set, one at least knows that the date of the examination must be before the date this value was entered. However, this specification currently has no rules on when and how such dates would be used for TREAT-NMD enquiries and registries *must* aim to obtain explicit datestamps for any datestamped or longitudinal item whenever possible.

Past and present status

In many cases, both the past and present status is important. For example, registries should capture whether an individual is currently using a feeding tube, and also whether this has been the case in the past. While individuals will usually know both the present and previous status for a condition like this, clinicians may not always have the complete medical records. Furthermore, a registry may change a question from capturing

only the current status (e.g. Are you currently using a feeding tube?) to asking about the past as well (e.g. Have you ever used a feeding tube? with the responses Currently, Previously and No). It should then remain possible to handle the responses to the previous question in a uniform way.

To cover all possible ways to model such an item in a data collection form, several items in this dataset specify the values listed below. Please note, the full list of values for this item is not intended for use in a data collection form and is for data mapping purposes only. Instead, registries generally should only present the options Currently, Previously and Never with suitable user-friendly wording.

Value ID	Description
Currently	This is currently the case
Previously	This has previously been the case, but is not currently
Never	This has never been the case, neither previously nor currently
Sometime	This has been the case at some time, but it is unknown whether it is currently the case
Not currently	This is currently not the case, but it is unknown whether it has previously been the case

The following tables demonstrate how such an item can be implemented in a form and how the possible responses would be mapped to the values above.

In general, registries *should* use example A (with the wording adapted according to the context) for current data collection forms if possible, as it is concise and conveys the most information. Otherwise, examples B or C *should* be used as a basis. If neither of these variants is possible, example D *may* be used.

Example A: Has this ever been the case?

Response	Value
Currently	Currently
Previously	Previously
Never	Never
Unknown	[no value]

Example B: Has this ever been the case? and Is this currently the case?

In this variant, the item is collected using two separate yes/no questions which each have an unknown option.

Response 1 (ever?)	Response 2 (currently?)	Value	Remark
Yes	Yes	Currently	
Yes	No	Previously	
Yes	Unknown	Sometime	
No	Yes		invalid
No	No	Never	
No	Unknown	Never	implausible
Unknown	Yes	Currently	implausible
Unknown	No	Not currently	
Unknown	Unknown	[no value]	

Whenever No is selected for the first question, this *must* be mapped to the value `Never` and the second question *should not* be displayed, thus avoiding invalid or implausible entries. The combination of Unknown for and Yes is implausible as knowing that something is currently the case implies knowing that it has ever been the case; this *should* be avoided by hiding or disabling the option Yes for the second question whenever Unknown is selected for the first.

Example C: Is this currently the case? and Has this previously been the case?

This variant is essentially only a minor modification of example B in which ever is replaced by previously and the questions are swapped. The mapping is the same as above, but it is shown here in the modified order for convenience:

Response 1 (currently?)	Response 2 (previously?)	Value	Remark
Yes	Yes	Currently	
Yes	No	Currently	implausible
Yes	Unknown	Currently	implausible
No	Yes	Previously	
No	No	Never	
No	Unknown	Not currently	
Unknown	Yes	Sometime	
Unknown	No	Never	implausible
Unknown	Unknown	[no value]	

The combinations that are considered implausible or invalid in example B are considered implausible here as well (while considering the change in order): The combinations Yes and No as well as Yes and Unknown are marked as implausible because the current state at a given moment will be the past state just a moment later. The combination Unknown and No is deemed implausible because if someone knows that a certain condition never held in the past, then this assessment applies to all moments right up to the current moment and should therefore also apply to the current moment. In electronic data collection forms, these combinations *should* be avoided by only displaying the second question if the reply to the first is not Yes and by hiding the option No in the second question if the reply to the first is Unknown.

Example D: Has this ever been the case?

This alternative is similar to example B, but does not contain the question about the current status. As it does not capture as much data as the other variants, it *should* generally be avoided and used only to map previously collected data.

Response	Value
Yes	Sometime
No	Never
Unknown	[no value]

Consistency rules

The aim of the datasets to avoid redundancy wherever possible. That means that for any piece of information (such as whether the diagnosis of a person has been genetically confirmed) there should generally be only one

item (e.g. the item `Genetic confirmation`). But often, a certain degree of redundancy cannot be avoided. For instance, when details of a genetic confirmation are provided (e.g. in the record `Genetic report` in the SMA and DMD datasets or in the record `Variant` in the LGMD dataset), this already implies that there is a genetic confirmation. But the item `Genetic confirmation` is still important because it should be possible to state that a diagnosis is confirmed even when no details are known. However, this means that the values could contradict each other: Suppose that for a certain individual, a registry submits an instance of the record `Genetic report`, but also `No` as value of `Genetic confirmation`. Does this mean that the genetic report does not actually confirm the diagnosis? Or is the value of `Genetic confirmation` incorrect?

To avoid such ambiguities, the dataset specifications contain consistency rules to exclude invalid data. For example, in the SMA and DMD datasets, the item `Genetic confirmation` has the following rule: "Must be `Yes` in case an instance of the record `Genetic report` is provided.". In most cases, more than one item or record may be related to a rule (in this case, the item `Genetic confirmation` and the record `Genetic report`), but the rule is only specified in one of them. Registries *should* ensure that consistency rules are met at all times through form structure, conditional display rules and input validation (see below). When submitting data to TREAT-NMD, any data that violates any consistency rule may be rejected.

Note that consistency rules are different from the following other rules that are used in registries:

- **Conditional display rules** specify on what conditions certain input fields or parts of a data collection form should be displayed. The dataset specifications do not contain any such rules because they don't mandate any specific form structures. However, in most cases registries can use conditional display rules to enforce consistency rules. In the example above, the consistency rule would be met if the data provider is first asked whether or not there is genetic confirmation. Only if the reply is "Yes", the further input fields for the genetic report are displayed.
- **Input validation rules** specify under which conditions an input from a user is rejected. They are related to a specific data collection form within a specific data collection process. Therefore, the dataset specifications again contain no such rules, but input validation rules can be used to enforce consistency rules.
- **Completeness rules** can be used to determine whether a certain registry entry is complete. For example, when the genetic report details are mandatory (e.g. for a clinician-reported registry), they need to be collected if the diagnosis is genetically confirmed. An according completeness rule would be "If `Genetic confirmation` is `Yes`, then an instance of the record `Genetic report` *must* be provided." Note that this is the converse of the consistency rule. While consistency rules mandate the *exclusion* of data, completeness rules require the *inclusion* of data. The dataset specifications do not contain any such rules as absolute requirements, but rather use the mandatory/non-mandatory status of items and records to indicate which information must be collected by which registries on a best-effort basis. It is always possible that certain information is not available for any reason, but this will generally not lead to other data on the same individual to be rejected.

Related items in previous version

There exist previous versions of the SMA and DMD datasets which used item numbers (e.g. 15.10) instead of the descriptive item IDs used here. To aid the transition to the new datasets, all items and records contain references to the numbers of the items in the previous datasets. In many cases, there is a direct correspondence between the old and new items; i.e., they describe the same information. But in some cases, the structure has been changed, so one new item may be related to multiple previous items or vice versa.

The Excel spreadsheet which can be found in the downloads page of each dataset also contains a worksheet "Mapping of previous version". It lists all previous item numbers which have related items in the current dataset

together with their corresponding new item and record IDs.

Technical details on IDs

The ID of each item and record serves as a unique and stable identifier. It may only consist of ASCII letters, numbers, spaces and hyphens (to be precise, hyphen-minus signs). When implementing this dataset, registries *should* use the names of this dataset as identifiers in their datasets wherever possible. However, registries *may* also choose to use a variant of the names in which all letters are converted to lower case and spaces as well as hyphens are replaced with an underscore. For example, for the item `Anti-AAV9 antibody test date`, a registry *may* choose `anti_aav9_antibody_test_date` as a database column name or other identifier. Regarding potential future transfer of patient-level data, registries *may* expect that item names transformed in this way will be accepted just as the standard names used in this specification, as long as the transformation is consistent across all items for a given registry.

Records

For some items, there can be multiple values for one individual in a registry, without the item being longitudinal in nature. For example, the item `Affected family member relation` describes the relation of an affected family member to the individual, but should allow multiple values to be stored; one for each family member. Furthermore, there are other items that also refer to the affected family members: `Affected family member sex` and `Affected family member side`. It is important to capture which values of these three items belong together. Therefore, they are grouped in a **record**, which is simply a collection of related items. Records are indicated with the symbol ☑ For any record, there can be multiple **record instances**, which are collections of item values. In the example above, this would require a record instance for each affected family member. Each record instance would contain three item values; one for each of `Affected family member relation`, `Affected family member sex` and `Affected family member side`.

Just like an item, every record has a descriptive and stable textual ID that is unique among all records in this dataset. However, the IDs of records and items may overlap; i.e., there may exist a record and an item with the same ID.

Longitudinal records

While `Affected family member` is a record that captures information on multiple people, but all referring to the current situation, there are other types of records which are longitudinal in nature. For example, for the item `Height` it is important to know what measurement method was used for a height value. In order to make it explicit that the items `Height` and `Height measurement method` all refer to the same measurement, they are grouped in a longitudinal record, marked with Longitudinal Each instance of a longitudinal record *must* include a date which specifies the point in time when, depending on the nature of the record, a measurement was made, an event took place or a certain condition held. The same rules apply as for longitudinal items: Month and year are sufficient, and in many cases, the date of a longitudinal record may be assumed to be equal to the entry date (see above).

Episode records

While some records such as `Height` or `Scoliosis surgery` refer to measurements or events which are points in time, other records refer to conditions that hold true over a period of time. In this dataset, such time spans are called episodes and the records are marked with Episode One example is `Feeding tube usage episode`,

where registries should record when an individual started and, if applicable, stopped using a feeding tube in the past. In addition, if a condition currently holds true at the time of an entry, it is important to explicitly record this date as well, which is called the ongoing date in this dataset.

For each episode record, the following dates *must* be captured:

- `Start date`: The date when the condition described by the record started to hold, if known
- `Stop date`: The date when the condition ceased to hold, if applicable and known
- `Ongoing date`: The date on which the condition was known to hold, if applicable; this date generally *may* be assumed to be the date of entry or clinical examination on which the registry entry is based

The following consistency rules apply:

- All three dates *must not* be after the date of entry.
- `Start date` *must not* be after `Stop date` or `Ongoing date`.
- Only one of `Stop date` and `Ongoing date` may be provided; they *must not* both be specified.

Reference period records

Some items refer to a specific period of time. For example, a registry may ask about hospitalisations in a form using the question Have you been admitted to hospital in the last 12 months? or Have you been admitted to hospital since the last registry update?. Since the specific time frame may vary from one value or registry to another, and may not always be derivable from the entry date, it is important to explicitly record the time period. In this dataset, records which refer to a period of time are called reference period records and are marked with **Reference period** For every reference period record instance, the following dates *must* be provided:

- `Begin date` specifies the beginning of the reference period.
- `End date` specifies the end of the reference period.

These dates do not refer to any actual event or condition, but only the dates which the question on a form referred to. The terms `Begin` and `End` are deliberately chosen to avoid confusion with the terms `Start` and `Stop` used for episode records. Unless otherwise noted, registries *should* use the following periods:

- At baseline data collection (i.e., when the information this record applies to is first collected for this individual), the time period is the last 12 months. That is, `Begin date` is the date of entry minus 12 months, and `End date` is the date of entry.
- At data updates, the time period is since the last update of this item. That is, `Begin date` is equal to the `End date` of the previous reference period, and `End date` is the date of entry.

The aim is to have a collection of reference periods which are consecutive and non-overlapping.

Privacy-preserving record linkage

The items in this group are solely for the purpose of generating a privacy-preserving record linkage (PPRL)[9] code, which is a technology that TREAT-NMD aims to utilise in future. They should only be collected and stored locally, and will never be requested by the TREAT-NMD Global Registry for verbatim transfer.

Note that the specification of these items may be changed or refined when a particular system for privacy-preserving record linkage has been adopted by TREAT-NMD.

[9] <https://irdirc.org/activities/task-forces/privacy-preserving-record-linkage/>

First name at birth

CR PR

Item type: free text

Related items in previous version: 2.01

Last name at birth

CR PR

Item type: free text

Related items in previous version: 2.03

Full date of birth

CR PR

Full date of birth of the individual, including the day. The value *may* be taken from an official document such as a birth certificate, passport, identity card, or health insurance card, but *may* also be provided by the individual or their guardian. If the values from any of these sources differ, the value on the birth certificate, or alternatively a different government-issued document, *must* be used.

Item type: date

Related items in previous version: 2.05

Sex at birth

CR PR

The sex that was assigned to the individual at birth. If a birth certificate is available, this value *must* match the sex given on the certificate. If the sex at birth is not known, this value *must* be empty. The value `Unspecified` *must* only be used if a birth certificate exists, but has no sex recorded.

Item type: single selection

Related items in previous version: 2.13

Value ID	Description
Male	Male
Female	Female
Intersex	Intersex
Unspecified	There is no sex specified on the pertinent document (e.g. birth certificate, passport or identity card)

Country of birth

CR PR

ISO 3166-1 alpha-2[10] two-letter code (e.g. `GB` for the United Kingdom) of the country of birth of the individual.

Item type: restricted text

Related items in previous version: 2.06

Place of birth

CR PR

City or town of birth.

Item type: free text

Related items in previous version: 2.07

Demographics

Date of birth

CR PR

Date of birth of the individual, as given on birth certificate or as reported by the individual or their parent. Although registries *must* collect the full date of birth (see the item `Full date of birth`), only the year and month will be requested by the TREAT-NMD Global Registry.

Item type: date

Related items in previous version: 2.05

Sex

CR PR

Current biological sex of the individual.

Item type: single selection

Related items in previous version: 2.14

Value ID	Description
Male	Male
Female	Female
Intersex	Intersex
Unspecified	There is no sex specified on the pertinent document (e.g. birth certificate, passport or identity card)

Country of residence

CR PR

[10] <https://www.iso.org/iso-3166-country-codes.html>

ISO 3166-1 alpha-2[11] two-letter code (e.g. `GB` for the United Kingdom) of the most recent known primary country of residence of the individual.

Item type: restricted text

Related items in previous version: 2.08

Living status

In patient-reported registries, we do not suggest that the individual/caregiver should answer this question. Ideally this item will be managed/entered by the Registry Curator, following review of the registration or communication from individual's family.

Alive

CR PR datestamped

`Yes` means that the individual was known to be alive at the datestamp of this value. `No` means that the individual is known to be deceased; in this case, the datestamp of this value is irrelevant and *may* be omitted. If known, the date of death is to be provided in the item `Date of death`.

It should not be necessary to store an explicit datestamp for this item. Instead, a registry *may* use the date of the last update or contact with the individual as the datestamp.

Item type: yes/no

Related items in previous version: 5.01

Date of death

CR PR

Date of the death of the individual. The date of death *may* be provided as a year only, if the month is not known.

Item type: date

Consistency rules: May only be provided if the value of `Alive` is `No`.

Related items in previous version: 5.02, 5.03

Cause of death code

CR

Cause of death as code of the classification specified in `Cause of death classification`.

Item type: restricted text

Consistency rules: May only be provided if the value of `Alive` is `No`.

Related items in previous version: 5.04, 5.05

[11] <https://www.iso.org/iso-3166-country-codes.html>

Cause of death classification

CR

Classification used in the item `Cause of death code`.

Item type: single selection

Consistency rules: *Must* be provided if `Cause of death code` is provided.

Value ID	Description
ICD-10	ICD-10
ICD-11	ICD-11

Diagnosis

Please also see the online examples.

Diagnosis date

Date on which the individual received the diagnosis.

This item refers to a diagnosis of DMD or BMD.

Item type: date

Related items in previous version: 6.01, 6.02

Diagnosis method

Method(s) used to establish the initial diagnosis.

This item refers to the *initial* diagnosis of DMD or BMD.

Item type: multiple selection

Related items in previous version: 6.03, 6.04

Value ID	Description
Genetic	Genetic/molecular test
Physical examination	Physical examination
Muscle biopsy	Muscle biopsy
Biomarkers	Biomarkers

Genetic confirmation

CR PR

Specifies whether the diagnosis has been genetically confirmed. If the genetic test results are pending, the value *must* be `No`.

This item refers to a diagnosis of DMD or BMD. If the value is `Yes`, the results *must* be captured in one or more

instances of the record `Genetic report`.

Item type: yes/no

Consistency rules: *Must* be `Yes` in case an instance of the record `Genetic report` is provided.

Related items in previous version: 1.05

Genetic diagnosis

CR PR

Item type: single selection

Related items in previous version: 1.05

Value ID	Description
DMD	Duchenne muscular dystrophy
BMD	Becker muscular dystrophy

Screening

CR

Specifies whether the diagnosis was made as a result of screening.

This item refers to a diagnosis of DMD or BMD.

Item type: single selection

Related items in previous version: 6.11

Value ID	Description
Family screening	The diagnosis was made as a result of family screening
Newborn screening	The diagnosis was made as a result of a newborn screening programme
Prenatal screening	The diagnosis was made as a result of prenatal screening
No screening	The diagnosis was not made as a result of screening

Genetic report

CR PR

All information collected in this record *should* be extracted from the genetic report and confirmed with a geneticist if necessary.

Genetic report date

CR PR

Item type: date

Related items in previous version: 6.05, 6.06

DMD variant CNV

CR

Exon copy number variations (CNV) in the DMD gene specified using the syntax described in the following examples:

- a deletion from exon 45 to exon 49 is written as `del ex45-49`
- a duplication from exon 2 to exon 7 is written as `dup ex2-7`
- a triplication of exon 51 is written as `tri ex51`
- non-contiguous CNVs are written as `dup ex2-7 and dup ex45-49`

Please see the guidelines for curators on mutation entries in DMD registries[12] for more information.

Item type: restricted text

DMD variant HGVS

CR

Description of the variant according to HGVS nomenclature[13].

The variant described here *must* be located in the DMD gene.

Item type: restricted text

Related items in previous version: 6.07

DMD testing method

CR

Testing method used to obtain the genetic result.

Registries *may* add an additional free-text field in their data collections forms to capture possible methods other than the ones provided in this item; values in the free-text field *should* be checked by a curator and mapped to the provided values wherever possible. New methods may be added to the dataset by TREAT-NMD whenever appropriate.

This item refers to the testing method used to obtain the result provided in `DMD variant HGVS`.

Item type: single selection

Related items in previous version: 6.08

Value ID	Description
RFLP	RFLP (Restriction Fragment Length Polymorphism)
HRM	HRM (High Resolution Melting)
MLPA	MLPA (Multiplex Ligation-dependent Probe Amplification)
qrtPCR	qrtPCR (Quantitative Real-Time PCR)
ddPCR	ddPCR (Droplet Digital PCR)

[12] <https://treat-nmd.org/wp-content/uploads/2021/05/uncategorized-New-guidelines-for-curators-of-DMD-registries.pdf>

[13] <https://varnomen.hgvs.org/>

Value ID	Description
Exon sequencing	Exon sequencing
RNA analysis	RNA analysis
Southern blotting	Southern blotting
FISH	FISH (Fluorescence in situ hybridization)

Clinical observations

Symptom onset

datestamped

Period in which the first symptoms manifested, as reported by the individual or their family. Any symptoms which were not considered abnormal at the time, but can retrospectively be attributed to the disease, *should* be considered as well. If the value is `Asymptomatic`, this item *must* be collected at each update until the individual shows first symptoms. If the value is not `Asymptomatic`, the datestamp is irrelevant and therefore not *required*.

This item refers to symptoms of DMD or BMD. If the value is not `Asymptomatic`, the date *must* be specified in `Symptom onset date`, if known.

Item type: single selection

Consistency rules: If values for both this item and `Symptom onset date` are provided, they *must* be consistent.

Background: The possible values for this item are based on the Human Phenotype Ontology[14] and include the broad classes `Pediatric onset` and `Adult onset` to cover cases in which a more specific range (e.g. `Childhood onset`) is not known.

Related items in previous version: 7.01

Value ID	Description
Neonatal onset	Neonatal onset (within the first 28 days of life)
Pediatric onset	Pediatric onset (between 28 days and 15 years)
Infantile onset	Infantile onset (between 28 days and 1 year)
Childhood onset	Childhood onset (between 1 and 5 years)
Juvenile onset	Juvenile onset (between 5 and 15 years)
Adult onset	Adult onset (16 years or later)
Young adult onset	Young adult onset (between 16 and 40 years)
Middle age onset	Middle age onset (between 40 and 60 years)
Late onset	Late onset (60 years or later)
Asymptomatic	Asymptomatic

[14] <https://hpo.jax.org/app/>

Symptom onset date

Date of the onset of the first symptoms as defined in the item `Symptom onset`. Registries *may* ask for the onset age in their data collection form and calculate the date from the date of birth.

Item type: date

Related items in previous version: 7.02

Height

CR longitudinal

Height

CR

Height or length of the individual, as directly measured or calculated using the method specified in `Height measurement method`.

Item type: decimal

Unit: centimetres

Related items in previous version: 8.03

Height measurement method

CR

Method used to obtain the value of `Height`.

Please note: Standing and Recumbent methods will not give accurate results where contractures and/or significant scoliosis exist. Arm span method will not give accurate results where arm contractures exist. In these cases, the Ulnar length method should be used.

- **Standing height**: Person length (height) is measured using a vertical length scale. The person would stand with footwear removed over a fixed platform or the floor and an unfixed headboard would be adjusted to the top of the head. Record the measurement to the nearest cm mark.
- **Recumbent length**: Employment of a horizontal length scale (or bench with steel ruler or tape). The person is placed flat on the horizontal measuring board, with footwear removed. The head should be placed against the fixed headboard, and the footboard adjusted so that it is against the base of the feet. Record the measurement to the nearest 1/2 inch / 1 cm.
- **Arm span**: Measure using a flexible tape, from the tip of the middle finger of one hand to the tip of the middle finger of the other hand. The person stands with their back to the wall, with both arms abducted to 90ř, the elbows and wrists extended and the palms facing directly forward.
- **Ulnar length**: Measure between the point of the elbow (olecranon process) and the midpoint of the prominent bone of the wrist (styloid process) (left side if possible). If using this method, the body height calculated from the measurement is to be provided.

Item type: single selection

Related items in previous version: 8.04

Value ID	Description
Standing height	Standing height
Recumbent length	Recumbent length
Arm span	Arm span
Ulnar length	Ulnar length

Weight

CR PR longitudinal

Weight of the individual.

Item type: decimal

Unit: kilograms

Related items in previous version: 8.08

Bone

Scoliosis diagnosis

CR PR longitudinal

Specifies whether the Cobb angle is above or below 30ř, or alternatively whether scoliosis has been diagnosed without reference to the Cobb angle. Whenever the Cobb angle is known to be above 30ř, registries *should* provide `Cobb angle above 30` as value. If the Cobb angle is unknown, but scoliosis was diagnosed, the value `Scoliosis diagnosed` *should* be provided instead. Whenever no scoliosis has been diagnosed or the Cobb angle is known to be 30ř or less, the value `Scoliosis not diagnosed` *should* be provided. Registries, in particular patient-reported registries, *may* ask about a scoliosis diagnosis without reference to the Cobb angle in a yes/no question where the replies are mapped to `Scoliosis diagnosed` and `Scoliosis not diagnosed`, respectively.

If the value is `Scoliosis not diagnosed` at the baseline entry, but the individual has had surgery that corrected a previous scoliosis, a backdated value for this item *should* be provided that indicates the scoliosis status before the surgery.

Item type: single selection

Related items in previous version: 9.01

Value ID	Description
Cobb angle above 30	Cobb angle greater than 30ř
Scoliosis diagnosed	Scoliosis diagnosed, Cobb angle unknown
Scoliosis not diagnosed	Scoliosis not diagnosed or Cobb angle lower than or equal to 30ř

Scoliosis surgery performed

CR PR longitudinal

Specifies whether the individual has ever had surgery specifically to try and correct scoliosis.

Item type: yes/no

Related items in previous version: 9.02

Scoliosis surgery

CR

Surgery for invasive lengthening of growing rods *must not* be included in this record.

Scoliosis surgery date

CR

Date of the scoliosis surgery.

Item type: date

Related items in previous version: 9.06

Scoliosis surgery type

CR

Item type: single selection

Related items in previous version: 9.03

Value ID	Description
Arthrodesis	Arthrodesis
Growing rods	Growing rods

Spinal fractures diagnosed

CR PR datestamped

Specifies whether the individual has ever had any spinal fractures confirmed by imaging. A datestamp is only *required* if the value is `No`.

Item type: yes/no

Related items in previous version: 9.07

Spinal fracture date

CR

Date of the first confirmed spinal fracture.

Item type: date

Related items in previous version: 9.09

Non-spinal fractures period

CR PR reference period

The first reference period collected at baseline *should* range from the date of birth of the individual to the date of the baseline entry so that the item `Non-spinal fractures diagnosed` specifies whether a non-spinal fracture has ever been diagnosed up to that point. For registry updates, the period *should* range from the previous entry to the date of the current entry, so that `Non-spinal fractures diagnosed` specifies whether a fracture has been diagnosed since the last update.

Using reference periods allows registries to explicitly state that no fractures have been diagnosed in a certain period.

Non-spinal fractures diagnosed

CR PR

Specifies whether any fractures other than spinal have been diagnosed.

If the value is `Yes`, the dates and locations of the fractures *must* be collected in instances of the record `Non-spinal fracture`.

Item type: yes/no

Consistency rules: *Must* be `Yes` if any instance of the record `Non-spinal fracture` with a datestamp within this reference period is provided.

Related items in previous version: 9.10

Non-spinal fracture

For each non-spinal fracture, the location and date date *must* be collected in an instance of this record. If provided, the datestamp of this record *must* be the date of the diagnosis of the fracture.

Non-spinal fracture date

Date of the diagnosis of the fracture.

Item type: date

Related items in previous version: 9.11

Non-spinal fracture location

Item type: single selection

Related items in previous version: 9.13

Value ID	Description
Upper arm	Upper arm
Lower arm	Lower arm
Upper leg	Upper leg
Lower leg	Lower leg
Hand	Hand

Value ID	Description
Finger	Finger(s)
Pelvis	Pelvis
Ankle	Ankle
Foot	Foot
Toe	Toe(s)
Skull	Skull
Jaw	Jaw
Rib	Rib(s)
Scapula	Scapula
Clavicle	Clavicle

Non-spinal fracture side

Specifies on which side of the body the fracture is located in case `Non-spinal fracture location` specifies a bone or body part that comes in pairs. For all other values of `Non-spinal fracture location` (`Pelvis`, `Skull` and `Jaw`), this value *must* be unspecified.

Item type: single selection

Related items in previous version: 9.12

Value ID	Description
Left	Left side
Right	Right side

Motor function

Motor ability

longitudinal

- **Sit without support** (WHO): Sits up straight with the head erect for at least 10 seconds. Does not use arms or hands to balance body or support position.
- **Transfer**: Can transfer from bed to a chair and vice versa without help from another person. Use of an assistive device such as a transfer board is permitted.
- **Stand without assistance** (WHO): Stands in upright position on both feet (not on the toes) with the back straight. The legs support 100
- **Get up from chair**: Can get up from a chair without help from another person. Use of armrests for support is permitted.
- **Rise from floor**: Can rise from the floor without help from another person and without contact to any object such as furniture. The starting position (e.g. sitting or lying) is irrelevant.
- **Walk without assistance** (WHO): Takes at least 5 steps independently in upright position with the back straight. One leg moves forward while the other supports most of the body weight. There is no contact with a person or object.

- **Climb stairs**: Climbs at least 4 stairs independently. Contact with a railing is permitted but there is no additional help from a person or other object.
- **Run**: Can move at a speed faster than a walk, never having both feet on the ground at the same time.
- **Useful function of hands** (RULM/PUL): Can use hands to hold pencil or pick up a token or drive a powered chair, use phone key pad. Corresponds to a score of 1 of the RULM/PUL entry item and to a grade of 5 on the Brooke scale.
- **Reach overhead in a sitting position** (RULM/PUL): Can raise both arms simultaneously above head whilst in a sitting position. Corresponds to a score of 5 or 6 on the RULM/PUL entry item and to a grade of 1 or 2 on the Brooke scale.
- **Raise hands to mouth in a sitting position** (RULM/PUL): Can raise one or two hands to mouth whilst in a sitting position. Corresponds to a score of 2 on the RULM entry item and to a grade of 4 on the Brooke scale.

Abilities marked with WHO are taken from the WHO Motor Milestones[15]; those marked with RULM/PUL are contained in the entry items of the Revised Upper Limb Module for SMA[16] and the Performance of the Upper Limb Module for DMD 2.0[17] as well as, with different grades, in the Brooke scale.

Motor ability

Item type: single selection

Value ID	Description
Sit without support	Sit without support
Transfer	Transfer from bed to chair
Stand without assistance	Stand without assistance
Get up from chair	Get up from chair
Rise from floor	Rise from floor without assistance
Walk without assistance	Walk without assistance
Climb stairs	Climb stairs
Run	Run
Useful function of hands	Useful function of hands
Reach overhead in a sitting position	Reach overhead in a sitting position
Raise hands to mouth in a sitting position	Raise hands to mouth in a sitting position

Motor ability status

Specifies whether the individual currently has the ability specified in `Motor ability`.

Item type: yes/no

Related items in previous version: 10.01, 10.05, 10.08

[15] <https://www.who.int/childgrowth/mgrs/en/fnb_motor_37_45.pdf?ua=1>
[16] <http://columbiasma.org/docs/cme-2010/RULM-Generic-Manual-16-Dec-2014.pdf>
[17] <https://www.musculardystrophyuk.org/wp-content/uploads/2017/06/PUL2.0-Worksheet-V1.0-1st-May-2016.pdf>

Motor measure

CR longitudinal

The collection of the following motor measures is **mandatory for clinician-reported registries**:

- 10MWT (10-Metre Walk Test)
- Brooke Scale of Upper Extremity Function
- RFF (Time to rise from the floor)

The collection of all other motor measures is non-mandatory.

Motor measure

CR

Item type: single selection

Value ID	Description
10MWT	10MWT (10-Metre Walk Test), provide time in seconds
100MTT	100MTT (100-Meter Timed Test), provide time in seconds
4SC	4SC (4-stair climb), provide time in seconds
Brooke	Brooke Scale of Upper Extremity Function, provide grade between 1 and 6
EK2	EK2 (Egen Klassification 2), provide score between 0 and 51
MFM-20	MFM-20 total score (Motor Function Measurement), provide total score as percentage between 0 and 100
MFM-20 D1	MFM-20 D1 (Motor Function Measurement), provide domain score as percentage between 0 and 100
MFM-20 D2	MFM-20 D2 (Motor Function Measurement), provide domain score as percentage between 0 and 100
MFM-20 D3	MFM-20 D3 (Motor Function Measurement), provide domain score as percentage between 0 and 100
MFM-32	MFM-32 total score (Motor Function Measurement), provide total score as percentage between 0 and 100
MFM-32 D1	MFM-32 D1 (Motor Function Measurement), provide domain score as percentage between 0 and 100
MFM-32 D2	MFM-32 D2 (Motor Function Measurement), provide domain score as percentage between 0 and 100
MFM-32 D3	MFM-32 D3 (Motor Function Measurement), provide domain score as percentage between 0 and 100
NSAA	NSAA (North Star Ambulatory Assessment), provide score between 0 and 34
PUL2	PUL2 (Performance of Upper Limb module for DMD 2.0), provide score between 0 and 42
RFF	RFF (Time to rise from the floor), provide time in seconds

Motor measure score

CR

The `Motor measure` value description specifies the unit in which the outcome *must* be provided.

Item type: decimal

Related items in previous version: 10.02, 10.04, 10.07, 10.10, 10.12, 10.14, 10.16, 10.18, 10.20

Wheelchair usage

Wheelchair usage

CR PR longitudinal

Specifies whether the individual is currently using or has previously used a manual or powered wheelchair or similar assisted mobility device due to their neuromuscular condition.

Registries *may* assume that wheelchair usage is a permanent condition and include only the options `Currently` and `Never`, with suitable user-friendly wording, in their data collection forms.

Item type: single selection

Related items in previous version: 11.01

Value ID	Description
Currently	This is currently the case
Previously	This has previously been the case, but is not currently
Never	This has never been the case, neither previously nor currently
Sometime	This has been the case at some time, but it is unknown whether it is currently the case
Not currently	This is currently not the case, but it is unknown whether it has previously been the case

Wheelchair usage episode

CR PR episode

Episode that describes when the individual used or has been using a manual or powered wheelchair or similar assisted mobility device due to their neuromuscular condition, with the frequency specified in `Wheelchair usage frequency`.

Wheelchair usage frequency

CR PR

Specifies the frequency of wheelchair usage.

Item type: single selection

Related items in previous version: 11.03

Value ID	Description
Part-time	Part-time (the individual is sometimes able to get around without a wheelchair or similar device)
Full-time	Full-time (the individual is unable to get around at all without a wheelchair or similar device)

Nutrition

A gastric tube (G-tube or gastrostomy) is a surgical opening into the stomach, in this case to insert a flexible feeding tube through the abdominal wall and into the stomach to allow direct delivery of adequate nutrition. A gastric tube is sometimes referred to as a PEG (percutaneuos endoscopic gastrostomy). A nasal feeding tube (also called nasogastric tube) is one that goes through the nose and down into the stomach.

Feeding tube usage

CR PR longitudinal

Specifies whether the individual is currently using or has ever used a feeding tube for feeding due to their neuromuscular condition. If the value is `Currently`, `Previously`, or `Sometime`, one or more instances of the record `Feeding tube usage episode` *should* be provided to specify details.

Item type: single selection

Related items in previous version: 12.01

Value ID	Description
Currently	This is currently the case
Previously	This has previously been the case, but is not currently
Never	This has never been the case, neither previously nor currently
Sometime	This has been the case at some time, but it is unknown whether it is currently the case
Not currently	This is currently not the case, but it is unknown whether it has previously been the case

Feeding tube usage episode

CR episode

Episode that describes current or previous usage of a feeding tube for feeding due to the individual's neuromuscular condition. The start, stop and ongoing dates of this record apply to the feeding tube usage of the type specified in `Feeding tube usage type`. If the individual switched from one type of usage to another, two record instances must be provided.

Related items in previous version: 12.03, 12.05

Feeding tube usage type

CR PR

Item type: single selection

Related items in previous version: 12.02, 12.04

Value ID	Description
Exclusive	The individual is or was exclusively fed by a tube
Supplementary	The individual is or was supplementarily fed by a tube, e.g. for fluids

Pulmonary function

Please also see the online examples.

Non-invasive ventilation usage

CR PR longitudinal

Specifies whether the individual is currently using or has ever used non-invasive ventilation due to their neuromuscular condition over a period of two weeks or more. If the value is `Currently`, `Previously`, or `Sometime`, one or more instances of the record `Non-invasive ventilation episode` should be provided to specify details.

Non-invasive ventilation (NIV) uses airway support which is administered through a nose or face mask.

Item type: single selection

Value ID	Description
Currently	This is currently the case
Previously	This has previously been the case, but is not currently
Never	This has never been the case, neither previously nor currently
Sometime	This has been the case at some time, but it is unknown whether it is currently the case
Not currently	This is currently not the case, but it is unknown whether it has previously been the case

Non-invasive ventilation episode

episode

Episode that describes current or previous usage of non-invasive ventilation due to the individual's neuromuscular condition. Only periods of two weeks or more are to be added.

Non-invasive ventilation duration

Item type: single selection

Value ID	Description
Full-time	Full-time (the individual uses ventilation of the respective type for 16 hours or more per 24 hours)
Part-time awake and sleeping	Part-time, awake and sleeping (the individual uses ventilation of the respective type while awake and while sleeping, or only while awake, with a total duration of less than 16 hours per 24 hours)
Part-time sleeping	Part-time, only while sleeping (the individual uses ventilation of the respective type only while sleeping)

Invasive ventilation usage

CR PR longitudinal

Specifies whether the individual is currently using or has ever used invasive ventilation due to their neuromuscular condition over a period of two weeks or more. If the value is `Currently`, `Previously`, or `Sometime`, one or more instances of the record `Invasive ventilation episode` should be provided to specify details.

Invasive ventilation (IV) is surgery that creates an opening in the windpipe which allows breathing through a tracheostomy tube rather than through the nose and mouth.

Item type: single selection

Related items in previous version: 13.01

Value ID	Description
Currently	This is currently the case
Previously	This has previously been the case, but is not currently
Never	This has never been the case, neither previously nor currently
Sometime	This has been the case at some time, but it is unknown whether it is currently the case
Not currently	This is currently not the case, but it is unknown whether it has previously been the case

Invasive ventilation episode

episode

Episode that describes current or previous usage of invasive ventilation due to the individual's neuromuscular condition. Only periods of two weeks or more are to be added.

Related items in previous version: 13.02, 13.03, 13.04

Invasive ventilation duration

Item type: single selection

Value ID	Description
Full-time	Full-time (the individual uses ventilation of the respective type for 16 hours or more per 24 hours)
Part-time awake and sleeping	Part-time, awake and sleeping (the individual uses ventilation of the respective type while awake and while sleeping, or only while awake, with a total duration of less than 16 hours per 24 hours)
Part-time sleeping	Part-time, only while sleeping (the individual uses ventilation of the respective type only while sleeping)

Airway clearance assistance

CR PR longitudinal

Specifies how often the individual currently uses assistance in airway clearance and/or secretion mobilisation, for example using suction, chest percussion or a cough assist device.

Item type: single selection

Related items in previous version: 13.05, 13.07

Value ID	Description
Daily	Daily (used once or more in each 24-hour period)
Weekly	Weekly (used less frequently than daily, but used once or more in each 7-day period)
Occasionally	Occasionally (used less frequently than weekly)
Never	Never

Airway clearance technique

PR longitudinal

Types of airway clearance the individual is currently using.

Item type: multiple selection

Consistency rules: If a value is specified, `Airway clearance assistance` *must not* have the value `Never` for the same datestamp.

Related items in previous version: 13.06

Value ID	Description
Suction	Suction
Bag valve mask	Bag valve mask (Ambu bag)
Intrapulmonary percussive ventilation	Intrapulmonary percussive ventilation (IPV)
Mechanical insufflation-exsufflation	Mechanical insufflation-exsufflation (CoughAssist)

Pulmonary function test result

CR

At baseline, the result of the most recent test *must* be collected; at updates, the result of every test since the previous update *must* be collected. For clinician-reported registries, the date of the test or date of the report as well as the values for FVC and PCF *must* be collected, whereas for patient-reported registries it is sufficient to collect the date of the test or report.

Pulmonary function test date

CR

Date of the pulmonary function test in which the results specified in this record instance were obtained.

Item type: date

Forced vital capacity volume

CR

Forced vital capacity as absolute volume in litres.

Forced vital capacity (FVC) is the total amount of air exhaled during the FEV (Forced expiratory volume) test.

Item type: decimal

Unit: litres

Related items in previous version: 13.11

Forced vital capacity percentage

CR

Forced vital capacity as percentage of predicted value.

Item type: decimal

Unit: percent

Related items in previous version: 13.12

Spirometry position

Item type: single selection

Value ID	Description
Supine	Supine (lying on the back)
Sitting	Sitting
Standing	Standing

Peak cough flow

CR

Peak cough flow (PCF) in litres per minute.

Item type: decimal

Unit: litres per minute

Related items in previous version: 13.14

Cardiac function

Cardiac imaging performed

CR PR datestamped

Specifies whether the individual has ever had cardiac imaging, for example cardiac MRI or echocardiography.

Item type: yes/no

Related items in previous version: 14.01

Cardiac imaging result

CR

At baseline and registry updates, the results of the most recent imaging *must* be collected in this record; further results *may* be collected in addition. Furthermore, the date of the lastest cardiac MRI *must* also be collected in an instance of this record, if not already recorded.

Related items in previous version: 14.02

Cardiac imaging date

CR

Date of the cardiac imaging.

Item type: date

Cardiac imaging type

CR

Item type: single selection

Value ID	Description
Echocardiography	Echocardiography
Cardiac MRI	Cardiac MRI

Left ventricular ejection fraction

CR

Left ventricular ejection fraction as percentage.

Item type: decimal

Unit: percent

Related items in previous version: 14.04

Fractional shortening

CR

Fractional shortening as percentage.

Item type: decimal

Unit: percent

Related items in previous version: 14.08

Therapies and medication

Please also see the online examples.

DMT received

CR PR datestamped

Specifies whether the individual has ever received a disease-modifying therapy for their neuromuscular condition.

For DMD, these currently include the therapies listed as possible values for the item `DMT` below. Registries *should* ensure that the corresponding question in their data collection forms applies to all DMTs, even when further therapies are approved in the future. In case the question references specific therapies (e.g. "Have you received any of the following therapies?"), this means that the list *should* always be kept up to date (and if required, an ethics approval should cover such updates). Alternatively, the question can be worded in a way that does not rely on an exhaustive list of therapies (e.g. "Have you ever received a disease-modifying therapy?" together with an explanation and a non-exhaustive list of currently approved therapies).

Item type: yes/no

Related items in previous version: 15.31

DMT episode

CR PR episode

Related items in previous version: 15.34, 15.37, 15.38

DMT

CR PR

Specifies the disease-modifying therapy the individual is receiving or has received at some point.

When a new therapy is approved, this list will be updated in the dataset. However, there may be a time lag between the approval and the dataset update. Therefore, registries *should* either update their data collection forms as soon as possible after a relevant new therapy is approved without waiting for the addition to the dataset, or add a free-text field that can be used to specify any new therapy. As soon as the new value is added to the dataset, any entries containing a new therapy *should* be validated and mapped accordingly.

Item type: single selection

Related items in previous version: 15.32, 15.35

Value ID	Description
Ataluren	Ataluren (Translarna)
Casimersen	Casimersen (Amondys 45)
Eteplirsen	Eteplirsen (Exondys 51)
Golodirsen	Golodirsen (Vyondys 53)
Viltolarsen	Viltolarsen (Viltepso)

DMT status

CR PR

Specifies whether this therapy is ongoing or stopped.

Item type: single selection

Consistency rules: If a value for `Stop date` of this record is provided, this item *must* have the value `Stopped`.

Background: The disease-modifying therapies are collected in an episode record which already encodes whether the therapy is ongoing or has stopped by providing a value for `Ongoing date` or `Stop date`, respectively. However, in case the therapy is stopped, but the stop date is not known or has not been entered by the user and thus no value for `Stop date` can be provided, it is still important to encode this information. Therefore, unlike the other episode records in the dataset, the DMT record contains this item as a fallback.

Related items in previous version: 15.31

Value ID	Description
Ongoing	The individual has been receiving the therapy as of the date specified in 'Ongoing date' and has been continuously receiving it from the date specified in 'Start date' on (if specified)
Stopped	The individual has stopped continuously receiving the therapy since the date specified in 'Start date'

DMT stopping reason

CR PR

Reason the individual has stopped receiving the therapy specified in DMT (in the case of a continuous treatment), or has received a different disease-modifying therapy after receiving a single-administration therapy specified in DMT.

In addition to the values listed for this item, registries *may* add further options in their data collection forms if relevant locally, and/or provide a free text field. However, additional options not specified in this dataset *must not* be provided in a data submission for a TREAT-NMD enquiry. Future revisions of this dataset may include additional values, and these can be suggested (along with justification) by using the Report an issue with this item function.

Item type: single selection

Related items in previous version: 15.39

Value ID	Description
Funding	Insurance coverage/funding
Availability	Availability
Side effects from drug	Side effects from the drug
Insufficient benefit	Insufficient benefit
Insufficient initial improvement	Insufficient initial improvement
Loss of response	Loss of response
Elective choice	Elective choice

Corticosteroid usage

CR PR longitudinal

Specifies whether the individual is currently taking or has previously taken corticosteroids for their neuromuscular condition.

Item type: single selection

Related items in previous version: 15.01

Value ID	Description
Currently	This is currently the case
Previously	This has previously been the case, but is not currently
Never	This has never been the case, neither previously nor currently
Sometime	This has been the case at some time, but it is unknown whether it is currently the case
Not currently	This is currently not the case, but it is unknown whether it has previously been the case

Corticosteroid episode

CR PR episode

At baseline, details for the most recent or ongoing corticosteroid usage *must* be given in an instance of this record. At registry updates, a previously ongoing episode *should* be updated and any further episodes added as additional instances.

The stop date *should* be the date of the last administration during this episode.

Related items in previous version: 15.07, 15.13, 15.14

Corticosteroid drug

CR PR

Drug taken during this episode, excluding the tapering period. In case the drug is changed during one treatment, multiple episodes *must* be provided.

Item type: single selection

Related items in previous version: 15.02, 15.08

Value ID	Description
prednisone oral	prednisone (oral)
prednisolone oral	prednisolone (oral)
deflazacort oral	deflazacort (oral)

Corticosteroid administration days on

CR PR

Integer item specifying the number of days in a row the individual has been taking the corticosteroid. In combination with the subsequent item `Corticosteroid administration days off`, it specifies the administration schedule. The following table shows examples for various administration frequencies, where the column "Days on" contains the value of this item and "Days off" contains the value of the item `Corticosteroid administration days off`:

Administration frequency	Days on	Days off
Daily	1	0
Every other day	1	1
Weekend dosing	2	5
10 days on/10 days off	10	10
20 days on/10 days off	20	10
etc.		

For daily administration, value of `Corticosteroid administration days on` *must* be 1. Registries *should* offer a predefined list of options for the most common administration frequencies (for example those shown in the table above) which are automatically mapped to the according values of the items `Corticosteroid administration days on` and `Corticosteroid administration days off`, while displaying number input fields only to cover further cases.

Item type: integer

Related items in previous version: 15.04, 15.05, 15.10, 15.11

Corticosteroid administration days off

CR PR

Number of days in a row the individual has **not** been taking the corticosteroid. See the description of `Corticosteroid administration days on` for examples.

Item type: integer

Related items in previous version: 15.04, 15.05, 15.10, 15.11

Corticosteroid dosage

CR PR

Daily dosage of the corticosteroid, relative to the individual's body weight. If the corticosteroid was not administered daily, the dosage given on the administration days *must* be specified. For example, if the individual took 2 mg/kg on two days of the week and nothing on the other days, then 2 *must* be provided as value.

Item type: decimal

Unit: milligrams per kilogram (mg/kg) of body weight

Related items in previous version: 15.06, 15.12

Corticosteroid stopping reason

CR PR

Reason(s) for stopping this treatment.

Item type: multiple selection

Consistency rules: *Must not* be provided in case `Ongoing date` is specified for this episode.

Related items in previous version: 15.15

Value ID	Description
Side effects	Side effects
Elective choice	Elective choice
Insufficient benefit	Insufficient benefit

Cardiac treatment usage

CR PR longitudinal

Specifies whether the individual is currently receiving or has previously received cardiac treatment.

Item type: single selection

Related items in previous version: 15.27

Value ID	Description
Currently	This is currently the case
Previously	This has previously been the case, but is not currently

Value ID	Description
Never	This has never been the case, neither previously nor currently
Sometime	This has been the case at some time, but it is unknown whether it is currently the case
Not currently	This is currently not the case, but it is unknown whether it has previously been the case

Cardiac treatment episode

CR episode

At baseline, details for all cardiac treatments in the last 12 months *should* be given in instances of this record. At registry updates, any previously ongoing episodes *should* be updated and any further episodes added as additional instances.

Related items in previous version: 15.28

Cardiac treatment

CR

Cardiac treatment this episode refers to.

Item type: single selection

Consistency rules: A value *must not* be specified for both this item and `Other cardiac drug` in the same episode.

Related items in previous version: 15.28

Value ID	Description
Angiotensin receptor blockers	Angiotensin receptor blockers
ACE inhibitors	ACE inhibitors
Beta blockers	Beta blockers
Eplerenone	Eplerenone
Spironolactone	Spironolactone
Other diuretics	Diuretics other than eplerenone and spironolactone

Other cardiac drug

CR

International Nonproprietary Name (INN)[18] of a cardiac drug that this episode refers to but which is not listed as a possible value of the item `Cardiac treatment`.

Item type: free text

[18] <https://www.who.int/medicines/services/inn/en/>

Related items in previous version: 15.29

Cardiac treatment stopping reason

CR

Reason(s) for stopping this treatment.

Item type: multiple selection

Consistency rules: *Must not* be provided in case `Ongoing date` is specified for this episode.

Related items in previous version: 15.46

Value ID	Description
Side effects	Side effects
Elective choice	Elective choice
Insufficient benefit	Insufficient benefit

Allopathic drugs

CR PR **reference period**

Allopathic drug usage

CR PR

Specifies whether the individual has taken any prescribed allopathic drugs (to manage symptoms - not disease-modifying) or supplements during this reference period. If the value is `Yes`, details for each drug *may* be specified in instances of the record `Allopathic drug episode`.

This item does **not** apply to the usage of corticosteroids as a treatment for the neuromuscular condition or to the usage of cardiac drugs. These usages are specified in the items `Corticosteroid usage` and `Cardiac treatment`, respectively.

Only the usage of drugs or supplements with a duration of 14 or more consecutive days *should* be considered (apart from immunizations).

Item type: yes/no

Related items in previous version: 15.17, 15.41

Allopathic drugs

CR PR

Prescribed allopathic drugs or supplements the individual has taken during some time in the period from `Begin date` to `End date`.

Please note that the inclusion of a drug or supplement in this list does not indicate TREAT-NMD endorsement.

The four options containing "not otherwise specified" in the description (`Bone health`, `Gastrointestinal`, `Respiratory` and `Supplement`) *should* only be used in case no more specific information is available. For

example, if a registry form contains the question "Have you taken any drugs for bone health?" and the reply is "Yes", but no further details are known, then the value would be `Bone health`. But if it is known that the individual has taken bisphosphonates, then only `Bisphosphonates` (and not `Bone health`) *should* be provided as value of this item.

Only the usage of drugs or supplements with a duration of 14 or more consecutive days *should* be considered (apart from immunizations).

Item type: multiple selection

Related items in previous version: 15.18, 15.19, 15.21, 15.22, 15.24

Value ID	Description
Vitamin D	Vitamin D
Calcium	Calcium
Bisphosphonates	Bisphosphonates
Bone health	Drugs for bone health, not otherwise specified
Gastroesophageal reflux	Drugs for gastroesophageal reflux
Constipation	Drugs for constipation
Gastrointestinal	Gastrointestinal drugs, not otherwise specified
Antibiotics	Antibiotics
Influenza immunizations	Annual influenza immunizations
Pneumococcal immunizations	Annual pneumococcal immunizations
Respiratory	Respiratory drugs, not otherwise specified
Testosterone	Testosterone
Supplement	Supplement, not otherwise specified

Other allopathic drugs

CR PR

Comma-separated list of the International Nonproprietary Names (INNs)[19] of other drugs.

Corticosteroids taken as a treatment for the neuromuscular condition as well as cardiac drugs *must not* be specified here. Instead, the items `Corticosteroid drug` and `Cardiac treatment` (or `Other cardiac drug`) *must* be used instead.

Item type: free text

Related items in previous version: 15.20, 15.22, 15.25, 15.30

Allopathic drug episode

CR PR episode

Details of the allopathic drugs or supplements the individual has taken.

[19] <https://www.who.int/medicines/services/inn/en/>

The usage of corticosteroids taken as a treatment for the neuromuscular condition as well as the usage of cardiac drugs *must not* be specified here. Instead, the episode records `Corticosteroid episode` and `Cardiac treatment` *must* be used instead.

See the item `Allopathic drugs` for information regarding the options containing "not otherwise specified". Registries *may* include a free-text field to collect the names of further supplements not listed here.

Only the usage of drugs or supplements with a duration of 14 or more consecutive days *should* be considered (apart from immunizations).

Related items in previous version: 15.19, 15.22, 15.25

Allopathic drug

CR PR

Prescribed allopathic drug or supplement the individual has taken.

Please note that the inclusion of a drug or supplement in this list does not indicate TREAT-NMD endorsement.

Item type: single selection

Related items in previous version: 15.19, 15.21, 15.22, 15.24

Value ID	Description
Vitamin D	Vitamin D
Calcium	Calcium
Bisphosphonates	Bisphosphonates
Bone health	Drugs for bone health, not otherwise specified
Gastroesophageal reflux	Drugs for gastroesophageal reflux
Constipation	Drugs for constipation
Gastrointestinal	Gastrointestinal drugs, not otherwise specified
Antibiotics	Antibiotics
Influenza immunizations	Annual influenza immunizations
Pneumococcal immunizations	Annual pneumococcal immunizations
Respiratory	Respiratory drugs, not otherwise specified
Testosterone	Testosterone
Supplement	Supplement, not otherwise specified

Other allopathic drug

CR PR

International Nonproprietary Name (INN)[20] of the other drug taken.

Item type: free text

Related items in previous version: 15.20, 15.22, 15.25, 15.30

[20] <https://www.who.int/medicines/services/inn/en/>

Allopathic drug stopping reason

CR PR

Reason(s) for stopping this treatment.

Item type: multiple selection

Consistency rules: *Must not* be provided in case `Ongoing date` is specified for this episode.

Related items in previous version: 15.19, 15.22, 15.25

Value ID	Description
Side effects	Side effects
Elective choice	Elective choice
Insufficient benefit	Insufficient benefit

Rehabilitative interventions

reference period

Rehabilitative interventions usage

Specifies whether the individual has received any rehabilitative interventions in this period.

For the purposes of this dataset, rehabilitative interventions refers to the management of the individuals condition and quality of life through therapies from allied health professionals such as physiotherapists and speech therapists.

Item type: yes/no

Consistency rules: If `Rehabilitative interventions` has a value, this item *must* have the value `Yes`.

Related items in previous version: 15.44

Rehabilitative interventions

Rehabilitative interventions the individual has received at some time during the period from `Begin date` to `End date`.

In addition to the values listed for this item, registries *may* add further options in their data collection forms if relevant locally, and/or provide a free text field. However, additional options not specified in this dataset *must not* be provided in a data submission for a TREAT-NMD enquiry. Future revisions of this dataset may include additional values, and these can be suggested (along with justification) by using the Report an issue with this item function.

Item type: multiple selection

Related items in previous version: 15.44

Value ID	Description
Physiotherapy	Physiotherapy sessions (e.g. stretches)
Respiratory physiotherapy	Respiratory physiotherapy sessions
Massage	Massage

Value ID	Description
Home programme	Home programme (e.g. stretches/exercises)
Hydrotherapy	Hydrotherapy/water-based activity
Contracture management using orthotics	Management of contractures using orthotics (e.g. ankle foot orthoses)
Spinal brace	Spinal brace
Occupational therapy	Occupational therapy sessions, or input/equipment for home
Speech therapy	Speech and language therapy sessions

Hospitalisations and comorbidities

Please also see the online examples.

Hospitalisation period

reference period

Hospitalisation occurred

Specifies whether the individual has been admitted to hospital as an inpatient (that is, with an overnight stay) for any reason (irrespective of circumstances or connection to their SMA) and for any duration, sometime during the period from `Begin date` to `End date`. Note that these dates do **not** specify the begin and end of a hospitalisation; they only define the period to which this item applies. An inpatient stay in a rehabilitation hospital is to be included in this item and in the record `Hospitalisation`. If the value of this item is `Yes`, the details of all hospitalisations *must* be collected in instances of the record `Hospitalisation`.

Item type: yes/no

Related items in previous version: 16.01

Hospitalisation

Hospitalisation type

Type of hospitalisation.

The quoted definition of acute care is taken from Hirshon 2013[21].

Item type: single selection

Related items in previous version: 16.02

Value ID	Description
Planned	Planned (admission was scheduled in advance, e.g. planned surgery, scan, or administration of treatment)

[21] <https://doi.org/10.2471/blt.12.112664>

Value ID	Description
Acute	Acute (admission was in response to a sudden, often unexpected, urgent or emergent episode of injury and illness that can lead to death or disability without rapid intervention)

Hospitalisation admission date

Admission date of this hospitalisation.

Item type: date

Related items in previous version: 16.06, 16.12

Hospitalisation nights

Number of nights the individual spent in hospital during this hospitalisation.

Item type: integer

Consistency rules: Value must be positive

Related items in previous version: 16.07, 16.13

Hospitalisation acute reason code

Main reason for this hospitalisation as a code in the classification specified in `Hospitalisation acute reason classification`. This item is intended mainly for acute hospitalisations and *must* be collected if `Hospitalisation type` is `Acute`. However, it *may* also be provided if `Hospitalisation type` is `Planned`, if an emergency occurred during the planned hospitalisation which would otherwise have required an acute hospitalisation.

Registries, in particular patient-reported ones, *may* provide users with a list of predefined reasons which are mapped to their respective codes.

Item type: restricted text

Related items in previous version: 16.10, 16.11

Hospitalisation acute reason classification

Classification used for the value of `Hospitalisation acute reason code`. If a registry collects codes using a fixed classification, it is not required to collect the classification explicitly from the user; in this case, a fixed value for this item *may* be used instead.

Item type: single selection

Consistency rules: *Must* be provided in case `Hospitalisation acute reason code` is provided.

Related items in previous version: 16.10

Value ID	Description
ICD-10	ICD-10
ICD-11	ICD-11
MedDRA	MedDRA

Hospitalisation planned reason

Main reason for this hospitalisation. This item is only applicable to planned hospitalisations and *must* be collected only if `Hospitalisation type` is `Planned`.

In addition to the values listed for this item, registries *may* add further options in their data collection forms if relevant locally, and/or provide a free text field. However, additional options not specified in this dataset *must not* be provided in a data submission for a TREAT-NMD enquiry. Future revisions of this dataset may include additional values, and these can be suggested (along with justification) by using the Report an issue with this item function.

Item type: single selection

Related items in previous version: 16.03

Value ID	Description
G-tube placement	Placement of g-tube
Sleep study	Sleep study
Scoliosis surgery	Scoliosis surgery
Hip surgery	Hip surgery
Contracture surgery	Surgical treatment of contractures
Disease-modifying therapy	Administration of disease-modifying therapy
Checkup	Routine checkup

Hospitalisation SAE

Specifies whether the reason for the hospitalisation specified in `Hospitalisation acute reason code` was classed as a serious adverse event (SAE) in relation to a disease-modifying therapy.

Registries *should* clearly inform the data provider that completing this data item does not replace the need to report SAEs immediately via their local reporting mechanisms.

SAE = Serious Adverse Event. Any untoward medical occurrence that at any dose:

- results in death,
- is life-threatening (NOTE: The term "life-threatening" in the definition of "serious" refers to an event in which the patient was at risk of death at the time of the event; it does not refer to an event which hypothetically might have caused death if it were more severe.
- requires inpatient hospitalisation or prolongation of existing hospitalisation,
- results in persistent or significant disability/incapacity, or
- is a congenital anomaly/birth defect.

(EMA ICH E2A[22])

Item type: yes/no

Related items in previous version: 16.14

Hospitalisation SAE DMT

Disease-modifying therapy to which this SAE was related.

[22] <https://www.ema.europa.eu/en/ich-e2a-clinical-safety-data-management-definitions-standards-expedited-reporting>

Item type: single selection

Related items in previous version: 16.15

Value ID	Description
Ataluren	Ataluren (Translarna)
Casimersen	Casimersen (Amondys 45)
Eteplirsen	Eteplirsen (Exondys 51)
Golodirsen	Golodirsen (Vyondys 53)
Viltolarsen	Viltolarsen (Viltepso)

Comorbidities period

CR PR reference period

Comorbidities diagnosed

CR PR

Specifies whether the individual has been diagnosed with other comorbidities during the period from `Begin date` to `End date`. This includes any comorbidities diagnosed earlier than `Begin date` which still held during this period. In this context comorbidities are defined as any additional medical conditions occurring alongside the neuromuscular condition.

Item type: yes/no

Related items in previous version: 16.19

Comorbidity

CR episode

Related items in previous version: 16.20

Comorbidity code

CR

Comorbidity as code of the classification specified in `Comorbidity classification`. Registries, in particular patient-reported registries, *may* provide common comorbidities as a list in their data collection forms or have users enter the diagnosis in a free-text field which is then coded by a curator.

It is *recommended* that registries use a dropdown menu or radio buttons containing at least the following diagnoses:

Diagnosis	ICD-10 code
Steroid-related comorbidities:	
Cataract	H26.3[23]
Obesity	E66.1[24]
Hypertension	I15.8[25]
Delayed puberty	E30.0[26]
Diabetes	E14[27]
DMD-related comorbidities:	
Autism	F84.0[28]
ADHD	F90.0[29]

If any of these options is selected, the value of this item *must* then be the respective ICD-10 code given above (and `Comorbidity classification` *must* have the value `ICD-10`). Additional or more specific pre-selected diagnoses *may* be provided. In order to collect other unlisted diagnoses, registries *should* allow entering an ICD-10 code (for clinician-reported registries) or offer a free-text field that is manually mapped to an ICD-10 code by a curator (for patient-reported registries).

Item type: restricted text

Related items in previous version: 16.20, 16.21

Comorbidity classification

CR

Classification used in the item `Comorbidity code`. The list of possible values may be amended in future versions of the dataset to enable the use of codes present only in local ICD-10 modifications.

Item type: single selection

Related items in previous version: 16.20, 16.21

Value ID	Description
ICD-10	ICD-10
ICD-11	ICD-11
MedDRA	MedDRA

Comorbidity SAE

Specifies whether this comorbidity was classed as a serious adverse event (SAE) in relation to a disease-modifying therapy.

Registries *should* clearly inform the data provider that completing this data item does not replace the need to report SAEs immediately via their local reporting mechanisms.

SAE = Serious Adverse Event. Any untoward medical occurrence that at any dose:

- results in death,
- is life-threatening (NOTE: The term "life-threatening" in the definition of "serious" refers to an event in which the patient was at risk of death at the time of the event; it does not refer to an event which

hypothetically might have caused death if it were more severe.
- requires inpatient hospitalisation or prolongation of existing hospitalisation,
- results in persistent or significant disability/incapacity, or
- is a congenital anomaly/birth defect.

(EMA ICH E2A[30])

Item type: yes/no

Related items in previous version: 16.22

Comorbidity SAE DMT

Disease-modifying therapy to which this SAE was related.

Item type: single selection

Related items in previous version: 16.23

Value ID	Description
Ataluren	Ataluren (Translarna)
Casimersen	Casimersen (Amondys 45)
Eteplirsen	Eteplirsen (Exondys 51)
Golodirsen	Golodirsen (Vyondys 53)
Viltolarsen	Viltolarsen (Viltepso)

Clinical research

Clinical trial participation

longitudinal

Specifies whether the individual is currently participating or has previously participated in an interventional clinical trial. In this context, participated means the individual has passed the screening period and has been either randomised (in randomised trial) or dosed (in non-randomised trial). If an individual fails the screening period, or is not randomised/dosed for some other reason (for example consent withdrawal, family relocation), this is not classed as participation.

Item type: single selection

Related items in previous version: 17.01

Value ID	Description
Currently	This is currently the case
Previously	This has previously been the case, but is not currently
Never	This has never been the case, neither previously nor currently
Sometime	This has been the case at some time, but it is unknown whether it is currently the case

[30] <https://www.ema.europa.eu/en/ich-e2a-clinical-safety-data-management-definitions-standards-expedited-reporting>

Value ID	Description
Not currently	This is currently not the case, but it is unknown whether it has previously been the case

Clinical trial

For each clinical trial the individual is currently participating or has previously participated in (as defined in the item `Clinical trial participation`), a record instance *must* be provided whenever the information is available.

If known, `Clinical trial NCT number` should be provided; in this case, `Clinical trial name` and `Clinical trial drug` *may* be left unspecified.

Clinical trial NCT number

NCT number (ClinicalTrials.gov identifier) of the clinical trial, including the prefix "NCT", for example "NCT00004451".

Item type: restricted text

Related items in previous version: 17.02

Clinical trial name

Full name of the clinical trial that the individual is currently participating or has previously participated in.

Item type: free text

Related items in previous version: 17.03

Clinical trial drug

Full name of the drug that the clinical trial named in `Clinical trial name` was evaluating.

Item type: free text

Related items in previous version: 17.03

Other registry participation

CR PR datestamped

Specifies whether the individual is currently part of another registry and/or natural history study, in addition to this registry.

A patient registry can be defined as an organized system that uses observational study methods to collect uniform data (clinical and other) to evaluate specified outcomes for a population defined by a particular disease, condition, or exposure, and that serves one or more predetermined scientific, clinical, or policy purposes. (Gliklich et al. 2014[31])

Natural history studies can be defined as epidemiological studies that focus on describing the frequency, features, and evolution of a disease by collecting real-world data from groups of patients suffering from this disease. (Bevan et al. 2019[32])

[31] <https://www.ncbi.nlm.nih.gov/books/NBK208616/>

[32] <https://www.evidera.com/natural-history-studies-in-rare-diseases-and-genetic-biomarkers/>

Item type: yes/no

Related items in previous version: 1.07, 17.04

Other registry

CR PR datestamped

Comma-separated list of the full name of each registry or natural history study in which the individual is currently participating.

For observational studies listed on ClinicalTrials.gov, the NCT number (ClinicalTrials.gov identifier) *may* be provided instead of the name.

Item type: free text

Related items in previous version: 1.08, 17.05, 17.06

Patient-reported outcome measures

Patient-reported outcome measure

longitudinal

Patient-reported outcome measure

Item type: single selection

Related items in previous version: 18.02, 18.06

Value ID	Description
DISABKIDS	DISABKIDS (Measurement of Quality of Life and Level of Distress)
INQoL	INQoL (Individualised Neuromuscular Quality of Life Questionnaires) 2.0, provide score between 0 and 274
KIDSCREEN-52	KIDSCREEN-52
PedsQL 4.0 Generic Core Scales	PedsQL 4.0 Generic Core Scales
PedsQL 3.0 Neurosmuscular Module	PedsQL 3.0 Neurosmuscular Module, provide score between 0 and 100
PedsQL Multidimensional Fatigue Scale	PedsQL Multidimensional Fatigue Scale, provide score between 0 and 72
PedsQL 3.0 Duchenne Muscular Dystrophy Module	PedsQL 3.0 Duchenne Muscular Dystrophy Module
PODCI	PODCI (Pediatric Outcomes Data Collection Instrument), provide score between 0 and 100

Patient-reported outcome measure score

The `Patient-reported outcome measure` value description specifies which type of score *must* be provided.

Item type: decimal

Related items in previous version: 18.04, 18.07

Biomarkers

DEXA performed

datestamped

Specifies whether the individual has ever had a DEXA (Dual Energy X-ray Absorptiometry) scan.

Bone densitometry, also called dual-energy x-ray absorptiometry, DEXA or DXA, uses a very small dose of ionizing radiation to produce pictures of the inside of the body (usually the lower (or lumbar) spine and hips) to measure bone loss. It is commonly used to diagnose osteoporosis, to assess an individual's risk for developing osteoporotic fractures. (RadiologyInfo.org[33])

Item type: yes/no

Related items in previous version: 19.01

Muscle imaging performed

datestamped

Specifies whether the individual has ever had any muscle imaging undertaken.

Commonly used skeletal muscle imaging techniques include radiography, ultrasound, computed tomography, and MRI. Newer techniques include T2 mapping, blood oxygenation level dependent imaging, diffusion tensor imaging, and magnetic resonance spectroscopy. (Kuo et al. 2007[34])

Item type: yes/no

Related items in previous version: 19.03

Muscle biopsy performed

datestamped

Specifies whether the individual has ever had a muscle biopsy.

Item type: yes/no

Related items in previous version: 19.05

Muscle biopsy

Muscle biopsy date

Date of the muscle biopsy.

Item type: date

Related items in previous version: 19.06

[33] <https://www.radiologyinfo.org/en/pdf/dexa.pdf>
[34] <https://doi.org/10.1097/bor.0b013e3282efdc66>

Muscle biopsy purpose

Purpose of this muscle biopsy.

Item type: single selection

Related items in previous version: 19.07

Value ID	Description
Diagnostic	Diagnostic
Clinical research	Clinical research

Muscle biopsy stored in biobank

Specifies whether the sample is stored in a biobank.

Item type: yes/no

Related items in previous version: 19.09

Muscle biopsy biobank

Name of the biobank in which the sample is stored.

Item type: free text

Consistency rules: If a value is provided, `Muscle biopsy stored in biobank` *must* have the value `Yes`.

Related items in previous version: 19.10