TREAT-NMD
Core Datasets Implementation Workshops Report

19 and 20 May 2021
Virtual

Contents

1. DISCLOSURE AND CONFLICT OF INTEREST

These workshops were part of the TREAT-NMD Core Dataset Implementation Projects supported by Biogen (SMA Dataset Project) and Sarepta (DMD and LGMD Dataset Project). This report provides an overview of the discussions and recommendations made during the workshops; it does not necessarily represent the full perspectives of any individual attendee, Pharmaceutical Company or TREAT-NMD.

This report has been prepared by Janet Wilkins (DMD Dataset Project Coordinator), Joanna Das (SMA Dataset Project Coordinator) John McKenna (DMD Dataset Project Manager) and Sonia Segovia-Simon (LGMD Dataset Project Manager) with review and input from TGDOC chairs.

2. EXECUTIVE SUMMARY

TREAT-NMD is an international neuromuscular network, which coordinates a global network of neuromuscular registries. Registries covering one or more of the following diseases SMA, DMD and LGMD agreed to collect the common core dataset relevant for each disease.

Three virtual workshops were held across the 19th and 20th of May 2021, to provide an overview of the TREAT-NMD Core Datasets to registry curators participating across the projects. This workshop report provides a summary of the key discussion points, questions raised and any agreed next steps.

3. ABOUT THE WORKSHOP

3a. Projects Context

The DMD Core Dataset was established in 2007, expanded in 2013 and further expanded in 2019 (version 1). The rapid advancement of technologies in gene therapies and upcoming new treatments requires the collection of data which is comprehensible, suitable for clinical trial recruitment and post marketing surveillance. In 2020 funding was received for the piloting and implementation of the new expanded dataset. The dataset pilot was successful and following minor amendments and alignment with the SMA and LGMD dataset, the TREAT-NMD DMD Core Dataset v1.2 dataset is now approved and ready for registries use.

The SMA Core Dataset supports standardised data collection across all SMA registries in the TREAT-NMD network and this model has been in place for 10 years. The original dataset was minimal, designed to identify patients eligible for recruitment into clinical trials. However, with disease modifying therapies (DMT's) now available in many countries, TREAT-NMD expanded the SMA Core Dataset with a view to providing real-world data from patient registries to support post marketing surveillance. The TREAT-NMD SMA Core Dataset was established in 2009 (version 0), expanded in September 2018 (version 1) and revised in October 2020 (version 2). It supports data collection to inform on the natural history of SMA and provides data to support post-marketing surveillance (safety and effectiveness) of new treatments.

The LGMD Core Dataset has been developed to be fit for purpose and feasible for all LGMD registries to collect worldwide. The objectives of the LGMD Core Dataset are to clinically characterise the disease, support the longitudinal data collection for natural history studies and clinical trial recruitment. Version 0.1 of the LGMD Core Dataset was published in May 2021 and is pending a pilot test to assess the feasibility of data collection. Version 1.0 will be published by the end of October and available for all TGDOC registries.

3b. Workshop Aims and Objectives

The target audience for this workshop were registry curators participating in the Core Datasets Implementation projects. The workshop focused on providing training and guidance on the core datasets to support registry curators understanding and implementation. The dataset specification tool looks radically different to previous versions; however, the most significant changes lie in the presentation and description of the data items. The content (that is, the data to be collected for SMA and DMD) has not been drastically altered. In the case of LGMD there was no previous version of the dataset, so the current version follows the new structure.

Workshop Objectives:
- Provide a general overview of the core datasets structure and general principles.
- Explain the components of the dataset such as data item, value ID, longitudinal item, episode, reference period etc.
- Present examples of implementation of complex data items per disease (LGMD, DMD and SMA)

3c. Workshop agenda and structure

The workshop aimed to provide accurate and clear information and to ensure all participants had opportunities to:
- Learn from experience gained by registries who have implemented the SMA Core Dataset.
- Voice their opinion and ask questions.
- Encourage Q&A to aid the discussions.
- Share their experiences.
- Participate in disease dedicated workshop sessions.
- Workshop participants list (Appendix 1)

Workshop Agenda

5 min	**Welcome**	Michela Guglieri (TGDOC incoming Chair)
5 min	**Introduction**	Sonia Segovia (LGMD Project Manager) John Mckenna (DMD Project Manager)
30 min	**Dataset Specification Structure**	Marcel Heidemann (IT/ Data Expert)
5 min	**Dataset Appendices**	Joanna Das (SMA Project Coordinator)
10 min	**SMA Dataset Testimonials**	Robin Forbes (Australian NMD Registry) Lindsay Murphy (Global FKRP Registry) Vedrana Milic (Serbian DMD/SMA Patient Registry)
45min	**Working Examples**	Marcel Heidemann (IT/ Data expert)
20 min	**Comfort Break**	
20 min	**Disease Specific LGMD Session**	Marcel Heidemann (IT/ Data expert) Sonia Segovia (LGMD Project Manager)
20 min	**Disease Specific DMD Session**	Marcel Heidemann (IT/ Data expert) John Mckenna (DMD Project Manager)
20 min	**Disease Specific SMA Session**	Marcel Heidemann (IT/ Data expert) Joanna Das (SMA Project coordinator)
5 min	**Close**	

4. WORKSHOP DISCUSSIONS (Video Available)

4a. Welcome

Michela Guglieri, Anna Ambrosini and Craig Campbel welcomed everyone to the workshops and thanked everyone for attending. They highlighted that a lot of work had been done in the last 12 months by the dataset project teams and Marcel Heidemann on preparing and harmonising the web-based dataset specification.

4b. Introduction

Over the last 12 months TREAT-NMD had worked tirelessly on improving the quality of data provided by the existing core datasets (SMA and DMD) and establishing a new core dataset for LGMD. A lot of work has been invested in creating datasets specifications which will adhere to the FAIR (Findable, Accessible, Interoperable and Reusable) principles and harmonisation with other initiatives where possible.

Examples of issues identified by the SMA dataset project:
- Dataset presented as a data collection form (items appeared as questions, focusing only on what to collect, not on how/when to collect it)
- Mixed data types
- Too many free text fields, useful for registries but not TREAT-NMD
- Numbered items which were not unique or stable
- Included items that were not in remit of a core dataset

Key improvements made to the DMD and SMA Core Datasets and implemented into the LGMD Core Dataset:
- A clearly defined and structured technical dataset specification document which is machine readable
- Clear guidance on how to collect required data supported by example questions where relevant
- A dataset based on the FAIR data principles where possible
- Everything contained within one source file from which the specification website as well as PDF and Excel documents are generated
- Stable and unique identifiers for each data item
- Clearly defined data types with minimal free text fields

All of this will support more standardised data collection by the registries, and more efficient central aggregation and analysis of data by TREAT-NMD.

4c. Datasets Structure: general principles

Marcel Heidemann provided a comprehensive demonstration and explanation of the following topics:
- An explanation of naming conventions, data groups and items
- Use of unique identifiers in
- Data descriptions
- Data types
- Differences between longitudinal and datestamped items
- Explanation of 'reference periods' and 'episodes'
- Interactive example questions and example data representation

A full overview of is available via the video below:

4d. Testimonial

ANMDR SMA v2 Dataset implementation– Robin Forbes

The registry uses the REDCap platform and collects data from Australian patients. The implementation of the SMA Core Dataset v2 dataset took about six weeks. The most noticeable change the registry has experienced is the amount of time it takes add new patient on which currently takes at least an hour. As soon as a patient contacts the registry, registry staff will record their consent and fill out the questions that the patient is able to answer. They will then contact the patient's clinician for any missing clinical data. Paediatric patients are updated every six months and adult patients annually. A paediatric patient update takes approximately 45 minutes and an adult patient, not on treatment, takes around 30 minutes. Patients are suffering from registry fatigue, but the availability of new treatments and clinical trials present an opportunity. The registry is doing a lot of educational activities with different groups to explain the importance of data sharing. Using version 2 of the dataset has helped the registry to promote its activities and promote its collection of real-world activity and real-world data that can be used to respond to government and pharma queries regarding the effectiveness of treatments. What also helps the registry's credibility is being part of TREAT-NMD global dataset that is promoted across the world.

UK SMA Patient Registry – Lindsey Murphy

The registry was established in 2008 and collects data from patients in the UK and Ireland. The registry is a patient reported registry without clinicians' input. Any verification of genetic diagnosis is done by the registry curator. The UK registry is using the Munich platform which was created by Marcel Heidemann and used by the German registry. The implementation was therefore shared between the German and the UK registries. The Ethics approval was relatively pain free as the registry was already collecting the TREAT-NMD core dataset and the expansion of the dataset did not raise concerns. As for the dataset implementation, strong and sustained effort was and still is required to encourage participants engagement and re-engagement with the registry. The registry had been active for over 10 years and due to the severity of the disease some patients were unable to engage, and the activity of the registry was not as would have been desired. Contact with participants is via email and activities to raise the profile of the registry through patient organisation and clinics. As for the challenges with the collection of the expanded dataset, there is more data to manipulate, takes longer to download and is pushing the limits of Excel. There are challenges marrying the old data with the new. Patient engagement is a challenge and funding is a universal challenge. The pilot bursary was invaluable for the funding of the IT work and the establishment of the dataset.

Serbian DMD/SMA Patient Registry - Vedrana Milic

The registry is a clinician reported registry which uses Excel as its registry platform. They have added the SMA and DMD new items to their registry. The registry is interested in involving Serbian Patients Organisation in their work. Although the registry is a clinical registry that has access to the clinical data there are still gaps between the medical records and the datasets. Medical staff are struggling with keying in data at the same time as they are attending to the patient and their family. Data items that present a challenge are for example, hospitalisations, other medications, start and stop dates and other similar items. They are fully aware of the importance of these data item and the implications of collecting those items. Vedrana would like to use a different platform for the registry. The advantages of implementing the datasets are that by using the additional data items they are educating the next generation of medics on the importance of the data items and their collection. They are implementing functional tests but unable to use all recommended items.

Feedback

Following the workshop sessions an online was sent to all attendees. The following results were noted:

- 65.4% of attendees found the workshop very useful and liked the practical examples included in the workshop.
- 93.3.% strongly agreed/agreed that they have a better understanding of how to use the dataset specification tool.
- The majority of respondents stated that they would wish to see further support delivered via video tutorials (50%) or through one-to-one calls with the relevant dataset team (54.2%).

Next steps

- Produce video tutorials and post the in the dataset specification site for registries to view.
- Regular drop-in style session to be arranged via zoom for registries and project managers to attend to discuss any current topics.

Conclusions

- The workshops were well attended and well received with good engagement from curators.
- The format and design of the workshop successfully explained the most important aspects that help in the interpretation of the dataset. The examples of sections were very well received and appreciated as very illustrative.
- Presenting the datasets as a technical specification requires additional educational activity by TREAT-NMD. It is a new concept for most registries and is a change of approach that will require a structured training programme to address the more complex issues.
- Based on the feedback obtained, the development of training material is necessary to ensure the correct understanding of the dataset. In this sense we should offer workshops in the future, based on feedback received through the specification tool on topics of interest to registries.
- Most TGDOC registries are involved in more than one dataset implementation project. Unifying the presentation of the three datasets in one workshop has allowed us to address common concepts and avoid duplication of information and consequent registries burnout.
- More disease specific sessions looking at individual data items relevant to that disease would be beneficial to the registries.

APPENDIX 1: Workshops Organisers and Participants

<u>Chairs / Organisers:</u>

1. Anna Ambrosini, TGDOC Current Chair
2. Craig Campbell, TGDOC Outgoing Chair
3. Michela Guglieri, TGDOC Incoming Chair
4. Joanna Das, Project Coordinator
5. Marcel Heidemann
6. John McKenna, Project Manager
7. Sonia Segovia Simon, Project Manager
8. Lynsey Surtees Admin and Project Support Assistant
9. Janet Wilkins, Project Coordinator

<u>In attendance:</u>

1	Jessica Furlan	LGMD1F-D2
2	Esther Schrama	LUMC, Leiden, The Netherlands
3	Robin Forbes	AUSTRALIAN NEUROMUSCULAR DISEASE REGISTRY
4	Kim An	Australian Neuromuscular Disease Registry
5	Simone Thiele	TREAT-NMD Germany
6	Erin O'Ferrall	Canadian Neuromuscular Disease Registry
7	Marisol Montolio	REgistro de Pacientes Duchenne y Becker de EspaÃ±a
8	Ilaria Zito	REgistro Pazienti DMD/BMD Italia
9	Sureshkumar Sankaran	India
10	Claudia Sanchez	Registro unico de pacientes AME Colombia
11	Harumasa Nakamura	Remudy, Japan
12	Vitaliy Matyushenko	CSMA
13	Vesna Brankovic	SMA Serbia
14	LiÌvie MensovaÌ	ÄŒeskÃ¡ republika
15	Anna Cho	Korean DMD Registry
16	Ayesha Muhamed	MDA India
17	Viswanathan Venkataraman	Muscular Dystrophy Association India
18	Victoria Hodgkinson	Canadian Neuromusuclar Disease Registry
19	Noshin Koenig	Canadian Neuromuscular Disease Registry
20	Jessica Martin	Registro de afectados
21	Anna O'Malley	Australian Neuromuscular Disease Registry (ANMDR)
22	Francesc Pla Junca	Spanish registry of neuromuscular diseases
23	Mark Gowland	GRP TREAT-NMD
24	Andrew Corbett	Biogen
25	Cesar Canas	
26	Emma Faid	TGDOC TREAT-NMD
27	Lucy Imber	The Global Registry for COL6- Related Dystrophies and The International GNE Myopathy Registry
28	Lindsay Murphy	Global FKRP Registry
29	Ayse Karaduman	LHUKAM , Ankara Turkey
30	Marjan Cosyns	BNMDR
31	Alexandra Prufer de Queiroz Campos Araujo	Federal University of Rio de Janeiro, Brazil
32	Ana Paula Horokowski Kovacs	Federal University of Rio de Janeiro, Brazil
33	Teik-Beng Khoo	Malaysian SMA National Registry
34	Poorani Anandakeishnan	SMA Registry Malaysia
35	Marlene Jagut	BNMDR
36	IÌ‡pek GuÌˆrbuÌˆz	KUKAS

37	Numan Bulut	KUKAS
38	Dominique Baumann	Swiss Registry on Neuromuscular Disorder
39	Nagia Fahmy	LGMD
40	Khian Aun Tan	Malaysia
41	Ieva Micule	NMS datu kolekcija
42	Helen Walker	TGDOC
43	Shirley Ackerman Laufer	Little Steps Registry
44	Adrienna Dyck	Canadian Neuromuscular Disease Registry (CNDR)
45	Ana Kosac	
46	Caroline Ogden	Treat NMD
47	Susan Cardiff	Treat NMD
48	Jennifer Levy	LGMD2A/R1 Global Patient Registry
49	Rasha El Sherif	Egyptian neuromuscular registry
50	Beatrix Paĺlmafy	Hungarian SMA/DMD registry
51	Ann Martin	PPMD Duchenne Registry
52	Niki Armstrong	The Duchenne Registry
53	Lenka Mokraĺ	ÄŒeskÃ¡ republika
54	Sahar Hassanein	Abassia
55	Jung Hwan Lee	Korean NMD registry
56	Sarah Emmons	Dysferlin Registry
57	Stefania Pedroni	LGMD italian registry
58	Natalija Angelkova	Macedonia
59	Elena Sukarova Angelovska	Macedonia
60	Ulla Werlauff	The National Rehabilitation Centre for Neuromuscular Diseases
61	Miriam Rodrigues	NZ NMD Registry
62	Veronika Karcagi	Hungarian DMD Registry
63	Noshin Koenig	Canadian Neuromuscular Disease Registry
64	Grace Perez	CNDR
65	Nino Nana Tatishvili	Georgian registry pediatric
66	Ana Bedoshvili	Georgian NMD registry
67	Richard Roxburgh	NZ Neuromuscular Disease Registry
68	Vedrana Milic	Serbian DMD/SMA Patient Registry

TREAT-NMD Core Datasets Appendix
28 June 2021

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Introduction

This appendix has been prepared by the TREAT-NMD Dataset projects on behalf of the TGDOC (TREAT-NMD Global Database Oversight Committee), as a guidance document to support TGDOC member registries with the collection of the TREAT-NMD Core Datasets.

Any comments or questions about the dataset, or anticipated training or support requirements should be reported to the dataset project teams. Joanna Das for SMA Core Dataset, John McKenna for DMD Core Dataset and Sonia Segovia Simon for LGMD Core Dataset. Please see below the contact details.

Advised additional data collection (not part of core dataset)

The following data will never be requested nor accepted by TREAT-NMD for central submission, and as such do not form part of the Core Datasets. However, it is recommended that registries consider collecting the following information (where relevant) for their participants, for internal use only, to support local registry operations and communication with registry participants.

Identifiable data
First name / given name.
Last name / surname / family name
Current address and zip/post code
Current active email address
Telephone number
Contact preferences

Local registry ID
As best practise, each participant should be assigned upon enrolment a local registry ID (identifier) by the registry (or the IT platform), to assist the registry with safe data management. This ID would be for local use only. To conform with GDPR guidance, this ID must not contain any potentially identifiable information such as initials, date of birth or medical record number. For more information about what constitutes personal or identifiable data, please refer to GDPR guidance (https://gdpr.eu/eu-gdpr-personal-data/) and/or the 18 HIPAA identifiers (https://www.hipaajournal.com/considered-phi-hipaa/).

Consent and ethics

It is the responsibility of each individual registry to ensure that valid, informed legal consent is in place before sharing any individual's data with TREAT-NMD. This should be checked and confirmed before each data submission, in case of consent withdrawal or a change in consent requirements (e.g. an individual reaching the legal age of consent in their country).

It is also the responsibility of each individual registry to ensure that they have obtained appropriate ethical approval from any relevant committees, before participating in the TGDOC Global Registry and/or sharing any data with TREAT-NMD.

Suggested wording.
The wording in this section is provided to help registries describe their participation in the TREAT-NMD Global (SMA/DMD/ LGMD) Registry, and the implications for data usage. It may be useful for ethical applications, patient information materials, consent forms, and other similar documents. Please note, registries should ensure that the contents are adapted to suit their registry set-up, local ethical/consent requirements, and their patient cohorts.

Patient Information sheets

[Registry name] is part of an international network called the TREAT-NMD Global (SMA/ DMD/ LGMD) registry, which collects information from independent (SMA, DMD, LGMD) registries across the world, including [registry name].

Being part of the Global Registries Network ensures that patients can be contacted by their local registry team if their profile fits a clinical trial's eligibility criteria. In addition, third parties such as researchers, pharmaceutical companies and regulators can request de-identified global data reports to answer important research questions about SMA/DMD/LGMD. The data provided in response to these research questions can help to monitor how well different treatments or interventions are working and support other activities to improve patients' quality of life, such as the assessment of standards of care in different countries.

We will not share your data with the TREAT-NMD Global (SMA/ DMD/ LGMD) Registry unless you give us explicit permission. Any data we do share will be de-identified and we will never transfer any of your personal identifiable details; if shared, your record will only be identifiable by an anonymous code. Researchers using the TREAT-NMD Global SMA/DMD/LGMD Registry therefore cannot identify you personally from the information they have.

TREAT-NMD will manage all data shared with the Global Registry in compliance with relevant data protection regulations, including GDPR. When a third party requests a data report from the TREAT-NMD Global Registry, an independent governing committee votes on whether it is an appropriate use of the data. If approved, the Curator of each registry is asked to securely send their de-identified data to the TREAT-NMD Global Registry, and it is then combined with data from other registries and formed into a report. Sometimes this request may be for individual (but always de-identified) patient-level data, and sometimes it will be for aggregated total patient numbers against certain criteria. Sometimes the request may come from academic or clinical researchers, and sometimes it may come from industry (pharmaceutical or biotechnology companies).

Patient consent forms

Please note this does not represent a complete consent form – these suggestions only cover consent to include an individual's data in a data submission for a TREAT-NMD Global SMA*/DMD/LGMD* Registry enquiry and should be added into your existing/main consent forms as appropriate. *[1]

Preferably the participant should initial each individual statement to show clear, explicit and informed consent. As a minimum, each statement should at least have a tick-box to indicate that it has been read and understood (rather than just a single signature at the end of the form)

☐ *The TREAT-NMD Global SMA/DMD/LGMD Registry has been explained to me and I have had the opportunity to ask questions and have them answered.*

I give permission for my data to be shared with the TREAT-NMD Global SMA/DMD/LGMD Registry, for the purposes described in [Patient Information Sheet name and version number], in the following way (choose only 1):
☐ ***All levels** (de-identified and included at individual level **and** included in aggregated numbers)*
☐ *Included in **aggregated numbers** only.*
☐ *No data to be shared with the TREAT-NMD Global SMA Registry*

  (If permission is given for any level of sharing:)
Once my data has been shared with the TREAT-NMD Global SMA/DMD/LGMD Registry, I give my permission for it to be used for (choose only 1):
☐ *All approved data enquiries*
☐ *Academic or clinical enquiries only (no industry enquiries)*

*[1] If your registry chooses to use the TREAT-NMD Global Registries Platform to collect and store data before submitting to TREAT-NMD, the consent forms provided in the system will cover this.

Protocols / ethical applications

[Registry name] is part of an international network called the TREAT-NMD Global SMA/DMD/LGMD Registry, which collects information from independent SMA/DMD/LGMD registries across the world, including [registry name].
The Global SMA/DMD/LGMD Registry can receive data enquiries from third parties (industry or academic). [Registry name] will only share de-identified data with relevant consent in place with the TREAT-NMD Global SMA/DMD/LGMD Registry.

Third parties wishing to enquire into the data in the TREAT-NMD Global Registry must first have the approval of both the TREAT-NMD Global Database Oversight Committee (TGDOC) and the [registry name] Principle Investigator/Steering Committee/ Manager [delete/amend as needed according to registry governance]. If approval is granted, TREAT-NMD requests the relevant data from the registries in the network and provides the third party with a report containing aggregated data.

Global Registries Platform Patient Information sheets
Clinician Reported Registry Consent Form

Dear Patient, Parent or Guardian,

To ensure that the care of people living with Duchene Muscular Dystrophy (DMD)/Spinal Muscular Atrophy (SMA)/Limb Girdle Muscular Dystrophy (LGMD) [delete as appropriate] continues to improve, it is essential that detailed information on the state of health and treatment of as many people with these conditions as possible is captured. This Registry works in collaboration with TREAT-NMD in the study of rare neuromuscular diseases with a hope to advance diagnosis, care and treatment for those living with neuromuscular diseases around the world.

You are invited to participate with this Registry for (state what rare disease is being studied) where you will share your personal data with the Registry, who will be using the TREAT-NMD Global Registry Platform to store your data, as a central data repository but also to share your data. With your consent with the TREAT-NMD central data warehouse (CDW). As this is a global initiative, TREAT-NMD will invite the Registries to participate in sharing de-identified data to the CDW from many countries to provide more comprehensive information on how to improve the treatment for these rare diseases.

Regulations state that to include your data we require your written permission. We do hope you will agree to have your medical details being included by this Registry and shared with the CDW which will be managed by TREAT-NMD.

Your participation is entirely voluntary and any information that would allow you to be identified directly or indirectly will be removed. Also, you can withdraw your permission at any time. If you agree to participate please would you sign below as indicated.

Future Use of Private Information
Research using patient information is an important way to try to understand human disease. You are being given this information because the investigators want to save private information, by the Registry on the Global Registry Platform and to share your de-identified data with the TREAT-NMD CDW for other research campaigns globally.

The Registry will store your personal and medical patient data on the platform but it will be only the clinicians of your registry who will have access to this.

If you grant permission for your data to be used in wider research using the TREAT-NMD CDW, personal identifiers will be removed from identifiable private information and, after such removal, only then will the information be used for future research studies or distributed to another investigator for future research studies without additional informed consent from you.

<u>Withdrawal</u>
If you first agree to participate and then you change your mind, you are free to withdraw your consent and discontinue your participation at any time. Your decision will not affect your ability to receive medical care for your disease.

If you decide to withdraw your consent, please contact your registry curator immediately and this information can be updated on the platform.

Should you withdraw consent, please note that any data that has been shared with the CDW will not be able to be retrieved as it cannot be identified and therefore cannot be removed.

CONSENT FORM

After reading the information form, I hereby confirm that I consent to the following:

- ☐ *I accept that the Registry will hold my personal and medical data on the Universal Registry Platform, and hereby consent for the Registry to sore and use my data for any purpose related to my consultations*

I consent to the Registry providing my data to the TREAT-NMD Central Data Warehouse (singe selection):

- ☐ *All types of data – de-identified individual level data AND aggregate data*
- ☐ *Included in aggregated data only*
- ☐ *No data to be shared with the Central Data Warehouse*

I understand the types of research that my personal data can be used for and consent to (multiple selection):

- ☐ *Academic*
- ☐ *Industry*

- ☐ *I understand that I can withdraw consent at any time*

_____   _____
Signature of Adult Participant   *Date*

_____
Print Name of Adult Participant

_____   _____
Signature of Parent or Guardian   *Date*

_____
Print Name of Parent or Guardian

XXXXXXXXXXX Registry

Dear Patient, Parent or Guardian,

You are being invited to be part of a registry (research database). Before you agree to register in the xxxxxxxx Registry, it is important that you understand what is involved and what will be done with the information you provide.

The information below contains answers to some of the questions you might have, and underneath this information there are consent statements that you will need to complete, to confirm that you would like to participate.

If you have any questions after reading this information, please contact the Registry Curator.
Take time to decide whether you would like to take part or not.

You can also use the print button at the bottom of this page to download and print a copy of this information to keep.

1. What is a patient registry and why is one needed for [disease]?
Scientific advances over recent years have led to substantial changes in the treatment of many diseases. New therapeutic strategies are being developed and, for some of these treatments, plans for large studies involving patients from more than one country are already in place. When a clinical trial is being planned, it is very important that patients suitable for that trial can be found and contacted quickly. The best way of ensuring this can happen is to make sure that patients' details are all collected together in a single database or "registry" that contains all the information that researchers will need, including each patient's particular genetic defect and other key information about their disease.

This Registry works in collaboration with TREAT-NMD (www.treat-nmd.com) in the study of rare neuromuscular diseases with a hope to advance diagnosis, care and treatment for those living with neuromuscular diseases around the world. As part of this collaboration, individuals are approached to share data with the TREAT-NMD Global Registry also known as the central data warehouse (CDW). If you agree to share your data with the data warehouse, it will be de-identified and used to help researchers and support other activities to improve patient care, such as the assessment of standards of care. To ensure that the care of people with Duchene Muscular Dystrophy (DMD), Spinal Muscular Atrophy (SMA) and Limb Girdle Muscular Dystrophy (LGMD), continues to improve, it is essential that detailed information on the state of health and treatment of as many people with these conditions as possible is captured.

2. Whose data are you collecting in this registry?
This registry is for patients living with the disease Duchenne Muscular Dystrophy (DMD), Spinal Muscular Atrophy (SMA) or Limb Girdle Muscular Dystrophy (LGMD). Because one of the research aims is to understand the progress of the disease, the registry will be used to collect information on patients currently living with DMD/SMA/LGMD, and, as a record, on those who have died.
The registry is intended to collect data from patients who have been advised that they have DMD/SMA/LGMD, even if they do not yet have a confirmed genetic diagnosis. However, only when a patient has a confirmed genetic diagnosis will they be eligible for consideration for certain clinical trials and clinical studies.

3. Who will be holding the information I provide?
The registry is responsible for managing the access to the platform and your information. This Registry is using the TREAT-NMD Global Registry as its platform where your data will be stored and therefore it is TREAT-NMD's responsibility to keep your information secure. As your Registry is part of the TREAT-NMD Neuromuscular

*Network, your data can be shared with the TREAT-NMD central data warehouse but **only** if you provide your consent. If you do grant your permission, your data will be stored in the TREAT-NMD central data warehouse (CDW) and used in wider research, where your data will be de-identified, which means that all your personal identifiers will be removed. It is only after such removal, that the information will be used for future research studies or distributed to another investigator for future research studies without additional informed consent from you.*

If you have any questions or need further clarification on how your information is being used or would like to see a copy of the information which is held about you on the registry, please contact the Registry Curator.

4. How can I update my details if anything changes?
To make sure that the data in the registry is correct and up to date, you will receive an email reminder about once a year asking you to tell us about any changes in your medical condition. This can be done on-line by logging in with your username and password. We also ask you to inform your Registry about any major changes in your details that might occur in the period between updates, for example change of address or a change in the ability to walk.

5. How will I be identified in the registry?
Your personal details (name, address, email address, telephone number) have to be stored in the registry so that we can contact you if we need to inform you about possible clinical trials or anything else that might be relevant to your disease. This data will be stored in a secure manner and your records will be assigned a unique, anonymous code. It is this unique code that will be used to identify your record if data is being analysed. Only the staff in charge of the DMD/SMA/LGMD registry will be able to "de-code" the data to get access to personal details and that at no time will TREAT-NMD have access to your information unless it has been shared into the central data warehouse.

6. Will my data be kept confidential?
Yes. Your data is stored and managed by the registry curator XXXXXXX on the TREAT-NMD platform, and all data held is subject to protection under your Registry's data protection regulations whilst it is stored within the Registry. All information received from you will be treated confidentially and all of the information will be encrypted and stored on a secure server. If you give your permission to share your data with the TREAT-NMD central data warehouse and we publish any research or other documents based on data from the registry, this research will never identify you by name.

We may also share general (anonymous) statistical information and patient numbers from the registry with the TREAT-NMD community to add to their knowledge and improve information available to them as a precursor to planning clinical trials.

7. How will I benefit from registering?
This registry is intended as a public service for the benefit of patients living with xxxxx. You will not receive any payment or any other financial benefit as a result of submitting your data to the registry. The results of research facilitated by the registry may ultimately be patentable or may have commercial potential. However, you will not receive patent rights and will not receive financial benefits from future commercial development. Nevertheless, there may be other benefits to participating, including the following:
- *Your registry will inform you if (on the basis of the information you provide) you might be a suitable candidate for a certain clinical trial.*
- *The data collected from you might also provide benefits to other patients with xxxxxx, for example by revealing statistics on how many people have the same condition, or by providing general statistical information for researchers interested in the best standards of care and to help them make plans for clinical trials and other research initiatives.*
- *Your registry may publish some general statistical information from the registry, so you will be able to find out information about how SMA/DMD/LGMD affects other people.*

8. I want to be involved in a clinical trial. If I register, is this guaranteed?

No. Only patients with a confirmed genetic diagnosis will be eligible for certain clinical trials, should they become available in the future. However, even if your genetic diagnosis is not yet confirmed you can still participate in clinical studies.

Even with a confirmed genetic diagnosis there is no guarantee that registering your details will ensure you will be involved in a clinical trial. It is also important that you understand that even if the coordinators of a clinical trial believe that you might be eligible for that trial, based on the data about you stored in the registry, it is still possible that later on it will turn out that you do not meet the trial inclusion criteria after all.

9. I don't want to be involved in a clinical trial. Should I still register?

We hope you will be interested in registering even if you don't want to take part in a trial. The information you provide will still be useful to researchers who are trying to find out more about patients living with xxxxxxx, and we will still provide you with other information that might be relevant to your disease.

10. Do I have to participate in the registry and can I withdraw if I change my mind?

Your participation in this project is completely voluntary. The Regulation (EU) 2016/679 (General Data Protection Regulation) grants you the right to access to the data, and where appropriate, their rectification or erasure or the limitation of processing, or to object to the processing or the portability of the data in accordance with the provisions of the General Data Protection Regulation.

Should you wish to withdraw your data from the registry you will be free to do so without having to provide any explanation. Should you choose to withdraw your consent, please note that any data that has been shared with the central data warehouse will not be able to be retrieved as it cannot be identified and therefore cannot be removed.

If you wish to withdraw, you should contact the Registry Curator on xxxxxxxxxx.

11. How long will my information be kept on the registry?

There is no set time period for removing your data. Unless you ask us to remove your information from the registry, we shall keep it for as long as we consider necessary for the purposes described in this form. However you can contact the Registry Curator to remove your data at any time.

12. What if I have any concerns or further questions?

If you have any concerns, or other questions about this study or the way it has been carried out, you should contact the Registry Curator.

1. *I confirm that I have read and understand the above patient informed consent information (dated xxxxx 2021, version 1.1) for the XXXXXX Patient Registry. I have had the opportunity to consider the information and to ask questions.*
2. *I give consent for my data to be stored in the xxxxx Registry and for it to be transerred/extracted (in anonymous form only identifiable by a code) to the TREAT-NMD central data warehouse so it may be used for research and for the planning of clinical trials by medical companies.*
3. *I understand that allowing my data to be stored on the registry does not mean I will automatically be entered into future clinical trials.*
4. *I agree to be contacted periodically to provide an update about any changes in my medical condition, or about the management of my online account.*
5. *I agree to be contacted with information about relevant clinical trials, studies or surveys, to support research into my condition.*
6. *I give consent for the xxxxxx Registry team to obtain access to relevant parts of my medical records (if needed) to confirm my diagnosis, including genetic test results, muscle biopsy results, and related correspondence.*
7. *I understand that the results from future research may not have any direct implications for myself or my family.*
8. *I understand that my participation is voluntary and that I am free to withdraw my data from the registry at any time, without giving any reason, and without my medical care or legal rights being affected.*
9. *The nature of the registry has been fully explained to me. I have understood the patient information and I agree to participate in this registry.*

☐ *I have read and understood the information form.*

I hereby confirm that I consent to the following:

☐ *I give permission for the {local registry name} team to contact my doctor if needed, to request a copy of my genetic test results.*
☐ *I accept that the registry will hold my personal and medical data, and hereby consent for the registry to store and use my data for any purpose related to my consultations.*
☐ *I understand that allowing my data to be held with the registry does not mean I will automatically be entered into future clinical trials.*
☐ *I agree to be contacted periodically to provide an update about any changes in my medical condition, or about the management of my online account.*
☐ *I agree to be contacted with information about relevant clinical trials, studies or surveys, to support research into my condition.*
☐ *I understand that the results from future research may not have any direct implications for myself or my family.*

I consent to the Registry providing my data to the TREAT-NMD Central Data Warehouse (single selection):

☐ *All types of data – de-identified individual level data AND aggregate data*
☐ *Included in aggregated data only*
☐ *No data to be shared with the Central Data Warehouse*

I understand the types of research that my personal data can be used for and consent to (multiple selection):

☐ *Academic*
☐ *Industry*

☐ *I understand that I can withdraw consent at any time.*

_____   _____

Signature of Adult Participant   *Date*

_____

Print Name of Adult Participant

_____   _____

Signature of Parent or Guardian   *Date*

_____

Print Name of Parent or Guardian

_____

Print name of child (if signed by a Parent or Guardian)

Publications

TREAT-NMD and the TGDOC acknowledge that a great deal of hard work, resource and expertise goes into the collection of high quality patient data by its affiliated registries, and we are committed to ensuring that contributions towards the TREAT-NMD Global SMA/*DMD/LGMD* Registry are appropriately acknowledged wherever and whenever relevant. To this end, TGDOC have formed a Publications Committee who have a priority task to develop and have ratified a TREAT-NMD Global Registries Publications Policy.

If you would like to find out more or get involved in the Publications Committee, please contact the Committee Chair, Dr Rasha El Sherif (dr.rashaelsherif@gmail.com).

Stakeholder feedback and dataset revisions

A formal SMA/*DMD/LGMD* Dataset Revision Process has been developed to reflect TREAT-NMD's commitment to:
- ✓ ensure the core SMA/*DMD/LGMD* dataset remains appropriate, feasible and relevant.
- ✓ support collaboration and harmonisation with other data collection initiatives
- ✓ remain responsive to the needs of and consensus within the SMA/*DMD/LGMD* community
- ✓ acknowledge and manage the burden placed on registries and their participants with each change to the dataset.

Any formal dataset review will involve stakeholder consultation. In addition to the periodic formal reviews, feedback or discussion on the SMA/*DMD/LGMD* core dataset is welcome from stakeholders at any time.

More information on the datasets and how to provide feedback is available DMD, LGMD and SMA.

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