

# EURO-NMD Registry

*ERN Plenary Session Feb 2024  
Medical Center – University of Freiburg (UKLFR)*

*24 Jan 2024*



EURO-NMD  
Registry Hub  
European Reference Network  
for neuromuscular diseases



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# EURO-NMD Registry - Overview

- Registry for all neuromuscular conditions
- Longitudinal data collection, annual follow-ups
- Data collection covers:
  - Mandatory set of common data elements for rare disease registration defined by the European Commission [1]
  - Common data elements for neuromuscular conditions
  - Disease-group specific data elements

[1] [https://eu-rd-platform.jrc.ec.europa.eu/sites/default/files/CDS/EU\\_RD\\_Platform\\_CDS\\_Final.pdf](https://eu-rd-platform.jrc.ec.europa.eu/sites/default/files/CDS/EU_RD_Platform_CDS_Final.pdf)

# EURO-NMD Registry - Overview

- Data subsets for disease groups:
  - Neuropathies
  - Myopathies
  - Mitochondrial diseases
  - Neuromuscular junction disorders
  - Motor neuron diseases
  - Undiagnosed cases
- Manual data entry and/or CSV-based bulk data import
- Implemented in REDCap, hosted by Medical Center – University of Freiburg

# What's in for data providers of the EURO-NMD Registry?

- contribute to research on how care for patients with rare neuromuscular conditions can be improved, become a better expert centre
- be listed as the author for publications about the registry
- compensation for data provision
- use your centre's data in the registry for your needs
  - download forms with data as PDF
  - create and configure custom data exports
  - data downloads in many formats available (CSV, SPSS, SAS, R, Stata,...)
- statistical reports provided by the registry team

# EURO-NMD Registry: Download filled forms as PDF

Actions: [Modify instrument](#) [Download PDF of instrument\(s\)](#) [Video: Basic data entry](#)

**Baseline**

Editing existing Patient ID 6.

Event: **Enrolment and first visit**

**Patient ID**

disease\_group

- This data entry form (blank)
- This data entry form with saved data
- This data entry form with saved data (via browser's *Save as PDF*)
- This data entry form with saved data (compact)
- All data entry forms (blank)
- All data entry forms with saved data
- All data entry forms with saved data (compact)

EURO-NMD Registry DEMO  
Patient ID 14 (Enrolment and first visit)  
Page 1

## Baseline

disease_group	<input checked="" type="checkbox"/> Neuropathies
<b>BASELINE</b>	
Sex at birth	<input checked="" type="checkbox"/> Male (Patient's sex at birth (Element 2.2. in the Set of common data elements for Rare Diseases Registration (link)))
Date of birth yyyy-mm-dd	2022-12-06 (Patient's date of birth (Element 2.1. in the Set of common data elements for Rare Diseases Registration (link)))
Date of baseline yyyy-mm-dd	2024-01-31
Age at baseline (years)	1 (Age of patient (years))
Age at baseline (months)	13 (Age of patient (months))
<b>DISEASE HISTORY</b>	
Date of first contact with specialised centre known	<input checked="" type="checkbox"/> Yes (Element 4.1. in the Set of common data elements for Rare Diseases Registration (link))

# EURO-NMD Registry: Data exports

My Reports & Exports			
	Report name	View/Export Options	Management Options
A	<b>All data</b> (all records and fields)	<a href="#">View Report</a> <a href="#">Export Data</a> <a href="#">Stats &amp; Charts</a>	
B	<b>Selected instruments and/or events</b> (all records)	<a href="#">Make custom selections</a>	
1	date of birth, sex at birth, diagnosis orphacode	<a href="#">View Report</a> <a href="#">Export Data</a> <a href="#">Stats &amp; Charts</a>	<a href="#">Edit</a> <a href="#">Copy</a> <a href="#">Delete</a>
2	Group: Neuropathies	<a href="#">View Report</a> <a href="#">Export Data</a> <a href="#">Stats &amp; Charts</a>	<a href="#">Edit</a> <a href="#">Copy</a> <a href="#">Delete</a>
3	Group: Myopathies	<a href="#">View Report</a> <a href="#">Export Data</a> <a href="#">Stats &amp; Charts</a>	<a href="#">Edit</a> <a href="#">Copy</a> <a href="#">Delete</a>
4	Group: Mitochondrial Diseases	<a href="#">View Report</a> <a href="#">Export Data</a> <a href="#">Stats &amp; Charts</a>	<a href="#">Edit</a> <a href="#">Copy</a> <a href="#">Delete</a>
5	Group: Neuromuscular Junction Disorders	<a href="#">View Report</a> <a href="#">Export Data</a> <a href="#">Stats &amp; Charts</a>	<a href="#">Edit</a> <a href="#">Copy</a> <a href="#">Delete</a>
6	Group: Motorneuron Diseases	<a href="#">View Report</a> <a href="#">Export Data</a> <a href="#">Stats &amp; Charts</a>	<a href="#">Edit</a> <a href="#">Copy</a> <a href="#">Delete</a>
7	Group: Undiagnosed	<a href="#">View Report</a> <a href="#">Export Data</a> <a href="#">Stats &amp; Charts</a>	<a href="#">Edit</a> <a href="#">Copy</a> <a href="#">Delete</a>



# EURO-NMD Registry: Data exports

date of birth, sex at birth, diagnosis orphacode








Patient ID record_id	Event Name redcap_event_name	Date of birth yyyy-mm-dd date_of_birth	Sex at birth sex_at_birth	Diagnosis of the neuromuscular disease (ORPHAcode) diagnosis_rd_orpha
<a href="#">308-28</a>	Enrolment and first visit (Arm 4: Neuromuscular Junction Disorders)	1981-01-31	Male (male)	Myasthenia gravis (ORPHA:589)
<a href="#">308-34</a>	Enrolment and first visit (Arm 4: Neuromuscular Junction Disorders)	2003-05-20	Female (female)	Myasthenia gravis (ORPHA:589)
<a href="#">308-98</a>	Enrolment and first visit (Arm 4: Neuromuscular Junction Disorders)	1980-01-01	Male (male)	Myasthenia gravis (ORPHA:589)
<a href="#">13</a>	Enrolment and first visit (Arm 4: Neuromuscular Junction Disorders)	2020-03-03	Female (female)	Myasthenia gravis (ORPHA:589)
<a href="#">308-5</a>	Enrolment and first visit (Arm 4: Neuromuscular Junction Disorders)	1983-02-02	Male (male)	Myasthenia gravis (ORPHA:589)
<a href="#">308-9</a>	Enrolment and first visit (Arm 4: Neuromuscular Junction Disorders)	1961-03-15	Female (female)	Lambert-Eaton myasthenic syndrome (ORPHA:43393)
<a href="#">308-10</a>	Enrolment and first visit (Arm 4: Neuromuscular Junction Disorders)	2008-04-07	Female (female)	Congenital myasthenic syndrome (ORPHA:590)
<a href="#">308-19</a>	Enrolment and first visit (Arm 4: Neuromuscular Junction Disorders)	2005-03-01	Male (male)	Congenital myasthenic syndrome (ORPHA:590)
<a href="#">320</a>	Enrolment and first visit (Arm 4: Neuromuscular Junction Disorders)		Male (male)	Congenital myasthenic syndrome (ORPHA:590)

# EURO-NMD Registry: Data exports

## Exporting "date of birth, sex at birth, diagnosis orphacode"

Select your export settings, which includes the export format (Excel/CSV, SAS, SPSS, R, Stata) and if you wish to perform de-identification on the data set.

### Choose export format

-  CSV / Microsoft Excel (raw data)
-  CSV / Microsoft Excel (labels)
-  SPSS Statistical Software
-  SAS Statistical Software
-  R Statistical Software
-  Stata Statistical Software
-  CDISC ODM (XML)

### De-identification options (optional)

The options below allow you to limit the amount of sensitive information that you are exporting out of the project. Check all that apply.

#### Known Identifiers:

- Remove All Identifier Fields (tagged in Data Dictionary)
- Hash the Record ID field (converts record name to an unrecognizable value)

#### Free-form text:

- Remove unvalidated Text fields (i.e. Text fields other than dates, numbers, etc.)
- Remove Notes/Essay box fields

#### Date and datetime fields:

- Remove all date and datetime fields
- OR —
- Shift all dates by value between 0 and 364 days (shifted amount determined by algorithm for each record)  
[What is date shifting?](#)
  - Also shift all survey completion timestamps by value between 0 and 364 days (shifted amount determined by algorithm for each record)

[Deselect all options](#)

### Advanced data formatting options

#### Export blank values for gray Form Status?

All Form Status fields with a gray status icon can be exported either as a blank value or as "0" (Incomplete). Hint: Blank values are recommended if the data will be imported back into REDCap, in which this preserves the gray status icons for all the imported records.

Export gray Form Status fields with value of "0"

#### Set CSV delimiter character

Set the delimiter used to separate values in the CSV data file (only valid for CSV Raw Data and CSV Labels export formats):

, (comma) - default

#### Force all numbers into a specified decimal format?

You may choose to force all data values containing a decimal to have a specified decimal character (comma or period/full stop). This will be applied to all calculations and number-validated text values in the export file.

Use fields' native decimal format (default)

**NOTE:** Your data formatting selections above will be remembered in the future and will be pre-selected upon your next export.



# Statistical reports provided by the registry team

- Registry team @UKLFR configured an automated pipeline for data export, generating report, uploading results to centre-specific folders in the File Repository of EURO-NMD Registry
- Option for combined analysis with site-level data from Care and Trial Site Registry (CTSR)
- Content: basic data quality checks, summary statistics and key performance indicators (e.g. mean time from onset to diagnosis)
- Report-content can be expanded/adjusted, Registry Steering Committee decides

# Statistical reports provided by the registry team: Examples

All Files / Reports / TestCenter1 [DAG-Restricted: TestCenter1]

<input type="checkbox"/>	Name	Size	Time Uploaded	Comments	Share	Delete	doc_id / folder_id
<input type="checkbox"/>	 308_testcenter1_20230816_15.56.pdf	715 KB	16.08.2023 15:59	Uploaded by jaegerd.			20766
<input type="checkbox"/>	 308_testcenter1_20230817_16.23.pdf	724.9 KB	17.08.2023 16:27	Uploaded by jaegerd.			20776
<input type="checkbox"/>	 308_testcenter1_20230817_17.06.pdf	735.7 KB	17.08.2023 17:10	Uploaded by jaegerd.			20779
<input type="checkbox"/>	 308_testcenter1_20230818_10.42.pdf	747.3 KB	18.08.2023 10:55	Uploaded by jaegerd.			20785
<input type="checkbox"/>	 308_testcenter1_20230818_11.23.pdf	753 KB	18.08.2023 11:39	Uploaded by jaegerd.			20788
<input type="checkbox"/>	 308_testcenter1_20230821_14.45.pdf	753.5 KB	21.08.2023 14:50	Uploaded by jaegerd.			20803
<input type="checkbox"/>	 308_testcenter1_20230822_13.54.pdf	753.3 KB	22.08.2023 13:58	Uploaded by jaegerd.			20825
<input type="checkbox"/>	 308_testcenter1_20230822_16.26.pdf	753.4 KB	22.08.2023 16:29	Uploaded by jaegerd.			20829

Showing 1 to 8 of 8 entries

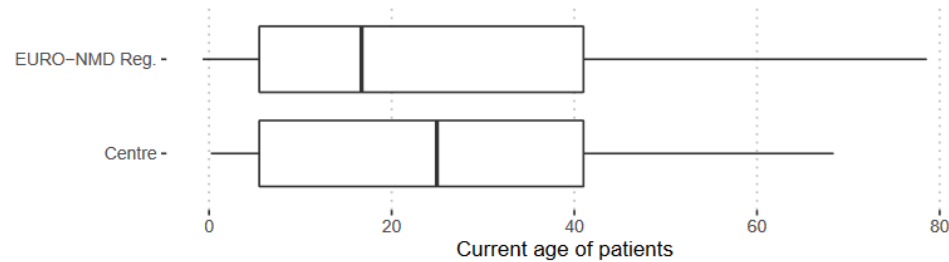
Previous 1 Next

# Statistical reports provided by the registry team: Examples

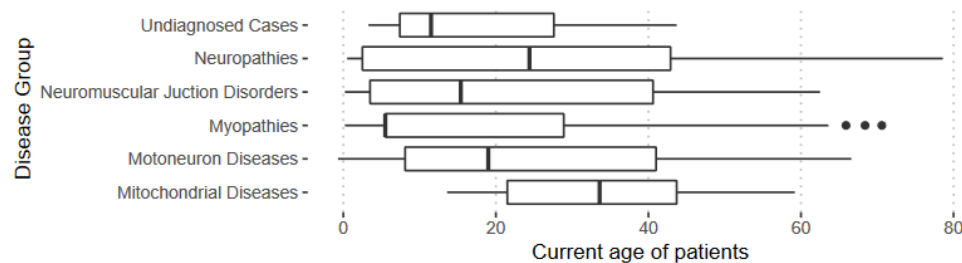
## Patients by age

currentAge calculated as  $\text{round}((\text{Sys.Date}() - \text{date\_of\_birth}) / 365, 1)$  with  $\text{Sys.Date}() = 16.08.2023$ .

Group	variable	n	min	median	mean	stdev	q25	q75	max
EURO-NMD Reg.	currentAge	290	-0.7	16.7	22.9	18.8	5.5	41	78.6
Centre	currentAge	42	0.2	25.0	25.2	20.9	5.5	41	68.4



## Patients by age and disease group (EURO-NMD Registry)

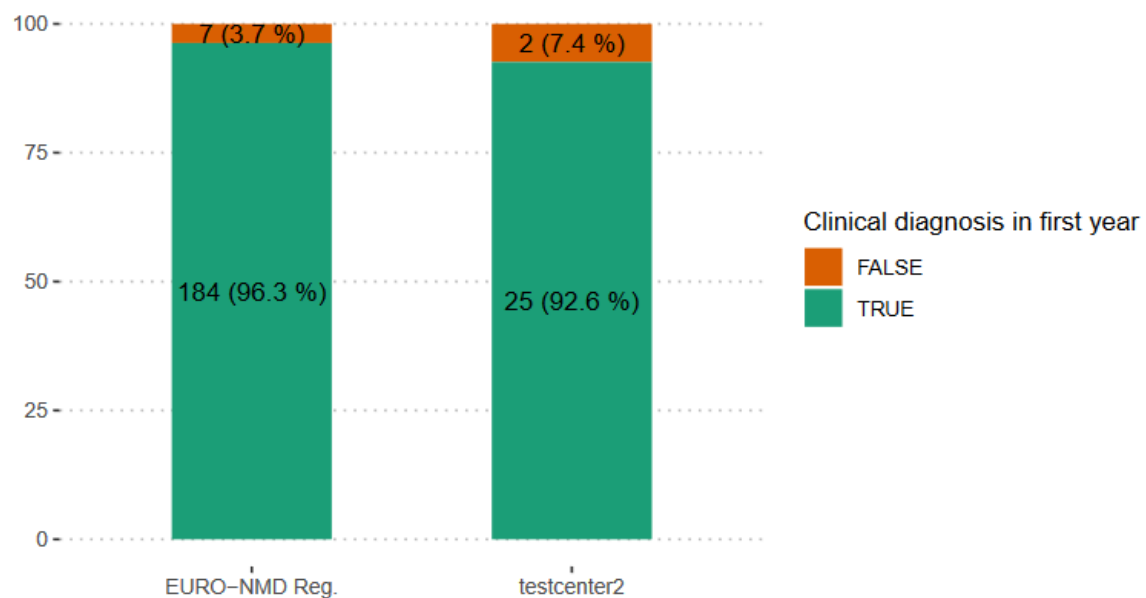


## Patients by age and disease group (TestCenter2)

# Statistical reports provided by the registry team: Examples

Patient records with clinical diagnosis in the the first year after first contact with speclised centre

Group	Clinical diagnosis within first year	N	n	%
EURO-NMD Reg.	FALSE	191	7	3.7
EURO-NMD Reg.	TRUE	191	184	96.3
testcenter2	FALSE	27	2	7.4
testcenter2	TRUE	27	25	92.6



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