EURO-NMD Registry

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

24 Jan 2024

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

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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

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EURO-NMD Registry: Download filled forms as PDF

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EURO-NMD Registry: Data exports

My Reports & Exports

		Report name	View/Export Options			Management Options		
A		**All data** (all records and fields)	🔍 View Report	📥 Export Data	📊 Stats & Charts			
B		**Selected instruments and/or events** (all records)	➤ Make custom selections					
1		date of birth, sex at birth, diagnosis orphacode	🔍 View Report	📥 Export Data	📊 Stats & Charts	✏ Edit	📋 Copy	✕ Delete
2		Group: Neuropathies	🔍 View Report	📥 Export Data	📊 Stats & Charts	✏ Edit	📋 Copy	✕ Delete
3		Group: Myopathies	🔍 View Report	📥 Export Data	📊 Stats & Charts	✏ Edit	📋 Copy	✕ Delete
4		Group: Mitochondrial Diseases	🔍 View Report	📥 Export Data	📊 Stats & Charts	✏ Edit	📋 Copy	✕ Delete
5		Group: Neuromuscular Junction Disorders	🔍 View Report	📥 Export Data	📊 Stats & Charts	✏ Edit	📋 Copy	✕ Delete
6		Group: Motorneuron Diseases	🔍 View Report	📥 Export Data	📊 Stats & Charts	✏ Edit	📋 Copy	✕ Delete
7		Group: Undiagnosed	🔍 View Report	📥 Export Data	📊 Stats & Charts	✏ Edit	📋 Copy	✕ Delete

EURO-NMD Registry: Data exports

date of birth, sex at birth, diagnosis orphacode

Search

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
308-28	Enrolment and first visit (Arm 4: Neuromuscular Junction Disorders)	1981-01-31	Male (male)	Myasthenia gravis (ORPHA:589)
308-34	Enrolment and first visit (Arm 4: Neuromuscular Junction Disorders)	2003-05-20	Female (female)	Myasthenia gravis (ORPHA:589)
308-98	Enrolment and first visit (Arm 4: Neuromuscular Junction Disorders)	1980-01-01	Male (male)	Myasthenia gravis (ORPHA:589)
13	Enrolment and first visit (Arm 4: Neuromuscular Junction Disorders)	2020-03-03	Female (female)	Myasthenia gravis (ORPHA:589)
308-5	Enrolment and first visit (Arm 4: Neuromuscular Junction Disorders)	1983-02-02	Male (male)	Myasthenia gravis (ORPHA:589)
308-9	Enrolment and first visit (Arm 4: Neuromuscular Junction Disorders)	1961-03-15	Female (female)	Lambert-Eaton myasthenic syndrome (ORPHA:43393)
308-10	Enrolment and first visit (Arm 4: Neuromuscular Junction Disorders)	2008-04-07	Female (female)	Congenital myasthenic syndrome (ORPHA:590)
308-19	Enrolment and first visit (Arm 4: Neuromuscular Junction Disorders)	2005-03-01	Male (male)	Congenital myasthenic syndrome (ORPHA:590)
320	Enrolment and first visit (Arm 4: Neuromuscular Junction Disorders)		Male (male)	Congenital myasthenic syndrome (ORPHA:590)

EURO-NMD Registry: Data exports

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]

	Name	Size	Time Uploaded	Comments	Share	Delete	doc_id / folder_id
☐	📄 308_testcenter1_20230816_15.56.pdf	715 KB	16.08.2023 15:59	Uploaded by jaegerd.	⬆	✕	20766
☐	📄 308_testcenter1_20230817_16.23.pdf	724.9 KB	17.08.2023 16:27	Uploaded by jaegerd.	⬆	✕	20776
☐	📄 308_testcenter1_20230817_17.06.pdf	735.7 KB	17.08.2023 17:10	Uploaded by jaegerd.	⬆	✕	20779
☐	📄 308_testcenter1_20230818_10.42.pdf	747.3 KB	18.08.2023 10:55	Uploaded by jaegerd.	⬆	✕	20785
☐	📄 308_testcenter1_20230818_11.23.pdf	753 KB	18.08.2023 11:39	Uploaded by jaegerd.	⬆	✕	20788
☐	📄 308_testcenter1_20230821_14.45.pdf	753.5 KB	21.08.2023 14:50	Uploaded by jaegerd.	⬆	✕	20803
☐	📄 308_testcenter1_20230822_13.54.pdf	753.3 KB	22.08.2023 13:58	Uploaded by jaegerd.	⬆	✕	20825
☐	📄 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

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Statistical reports provided by the registry team: Examples

Patients by age

currentAge calculated as `round((Sys.Date() - date_of_birth) / 365, 1)` with 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
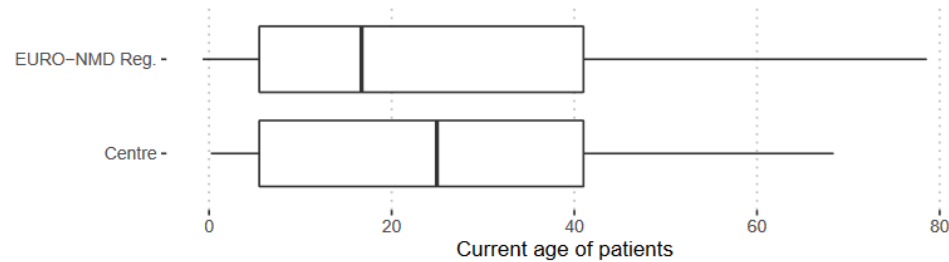
Patients by age and disease group (EURO-NMD Registry)
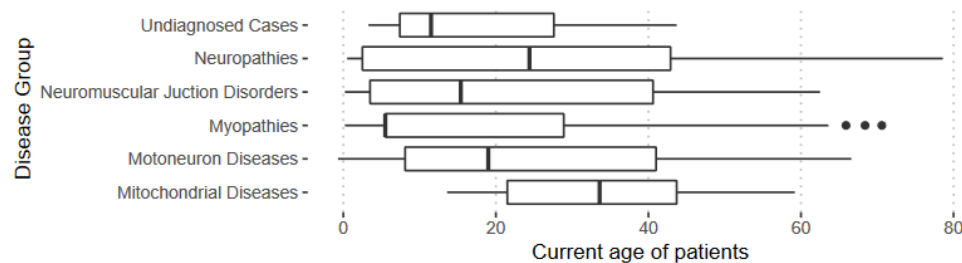
Patients by age and disease group (TestCenter2)

Statistical reports provided by the registry team: Examples

Patient records with clinical diagnosis in the the first year after first contact with specilised 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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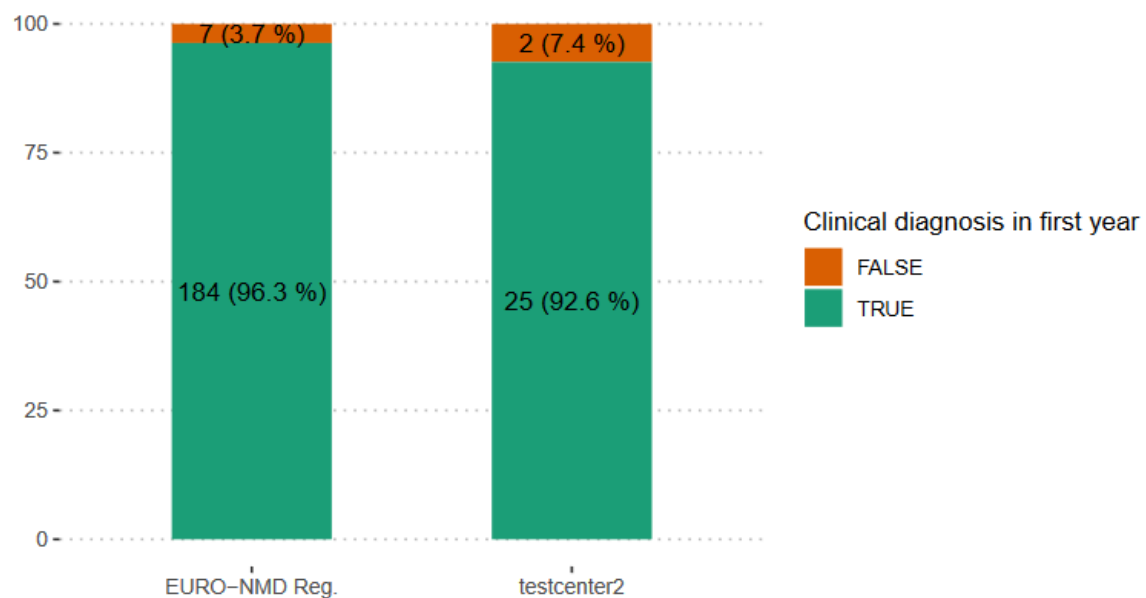
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