Description and assessment of fitness-for-purpose of real-world data (RWD) sources on Duchenne Muscular Dystrophy for regulatory decision-making

First published: 26/09/2025

Last updated: 13/04/2026

Study Ongoing

Administrative details

EU PAS number

EUPAS1000000748

Study ID

1000000748

DARWIN EU® study

No

Study countries

Australia

Austria

Belgium

- Brazil
- Bulgaria
- Canada
- China
- Croatia
- Czechia
- Denmark
- Finland
- France
- Germany
- Greece
- Hungary
- India
- Iran, Islamic Republic of
- Italy
- Japan
- Netherlands
- New Zealand
- Poland
- Portugal
- Russian Federation
- Saudi Arabia
- Serbia
- South Africa
- Spain
- Sweden
- Switzerland
- United Kingdom
- United States

Study description

Duchenne Muscular Dystrophy (DMD) is a severe and rare progressive inherited neuromuscular disorder for which there is currently no cure. Several new types of therapies have recently achieved conditional approval.

Therefore, there is a need to supplement the clinical trial data with long-term safety and effectiveness evidence to support regulatory decisions.

The aim of the current study is thus to map exiting data sources within DMD for future real-world evidence (RWE) studies.

This mapping will include the following key elements:

• Establishment of a list of core variables required for future RWE studies in DMD disease.

• A structured inventory of registries and other data sources relevant to DMD.

• Assessment of longitudinal completeness, core variable availability (e.g., genotype, functional
scores, medication use), and outcome definitions.

• Evaluation of completeness, consistency, and timeliness of data, as well as mechanisms for
ongoing data curation and quality assurance.

• A review of access procedures, legal and ethical considerations, and technical feasibility of
linkage with other data sources.

Finally, we will describe the overall findings and considerations in a manuscript for publication including recommendations on important operational, methodological and technical elements to consider when considering using real-world data (RWD) to address any research questions of interest.

Study status

Ongoing

Research institutions and networks

Institutions

Data Analytic Center (DAC), Danish Medicine Agency

Denmark

First published: 17/04/2023

Last updated: 17/04/2023

Institution | EU Institution/Body/Agency | ENCePP partner

The Danish Rehabilitation Centre for Neuromuscular Diseases (RCFM)

Networks

Translational Research in Europe - Assessment and Treatment of Neuromuscular Diseases (TREAT-NMD)

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Network

Contact details

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Study timelines

Date when funding contract was signed

Actual: 26/08/2025

Study start date

Planned: 17/08/2025

Actual: 26/08/2025

Data analysis start date

Actual: 26/08/2025

Date of interim report, if expected

Planned: 26/01/2026

Actual: 26/01/2026

Date of final study report

Planned: 26/08/2026

Sources of funding

- EMA

Study protocol

Feasibility Assessment Plan ROC39 Deliverable 3.pdf (752.67 KB)

Regulatory

Was the study required by a regulatory body?

Yes

Is the study required by a Risk Management Plan (RMP)?

Not applicable

Other study registration identification numbers and links

EMA/2020/46/TDA/L5.04 (ROC 39)

Methodological aspects

Study type

Study type list

Study topic:

Disease /health condition

Other

Study topic, other:

Real World Data feasibility study

Study type:

Not applicable

Scope of the study:

Feasibility analysis

Validation of study variables (exposure outcome covariate)

Data collection methods:

Secondary use of data

Study design:

na

Main study objective:

To identify a list of core variables to be used for future RWD studies in DMD patients with focus on the four regulatory research questions provided by EMA.

Study drug and medical condition

Medical condition to be studied

Duchenne muscular dystrophy

Population studied

Short description of the study population

Patients with Duchenne muscular dystrophy registered in a disease register (world-wide)

Estimated number of subjects

30.000

Study design details

Summary results

The final core variable list for the fit-for-purpose assessment includes variables that are both scientifically
and clinically relevant to collect in real-world data sources, particularly disease registries. A “question-led”
approach was used to identify variables of interest enabling selection of variables ensuring focus on the
regulatory research question of interest.

Documents

Abstract of study report

ROC39 Deliverable 2_version 1.1 (final)_clean.pdf (454.16 KB)

Data management

ENCePP Seal

The use of the ENCePP Seal has been discontinued since February 2025. The ENCePP Seal fields are retained in the display mode for transparency but are no longer maintained.

Data sources

Data sources (types)

Published literature

Use of a Common Data Model (CDM)

CDM mapping

No

Data quality specifications

Check conformance

Unknown

Check completeness

Unknown

Check stability

Unknown

Check logical consistency

Unknown

Data characterisation

Data characterisation conducted

No