

# Metabolic Profiling of Neuromuscular Diseases (MetabNMD) – subproject SMA

**First published:** 30/10/2019

**Last updated:** 04/01/2022

Study

Planned

## Administrative details

### EU PAS number

EUPAS32033

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### Study ID

44920

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### DARWIN EU® study

No

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### Study countries

 Germany

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### Study description

This multi-center, prospective, controlled, non-randomized, non-interventional, open, unblinded study is aimed at identifying non-invasive diagnostic and prognostic biomarker profiles for neuromuscular diseases, providing a novel

method for screening, predicting disease severity and dynamic monitoring under pharmacotherapy. This will ultimately allow pre-symptomatic initiation of therapies, inform about the accurate time point of treatment initiation and provide tools for individual dosing adjustments.

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### **Study status**

Planned

## Research institutions and networks

### Institutions

[Heidelberg University Hospital](#)

**First published:** 01/02/2024

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**Institution**

[Hamburg Germany, Essen Germany, Gießen  
Germany, München Germany](#)

## Contact details

### **Study institution contact**

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## Study contact

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### Primary lead investigator

Andreas Ziegler

## Primary lead investigator

## Study timelines

### Date when funding contract was signed

Planned: 30/11/2019

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### Study start date

Planned: 01/05/2020

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### Data analysis start date

Planned: 30/11/2020

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### Date of final study report

Planned: 31/05/2022

## Sources of funding

- Pharmaceutical company and other private sector
- Other

## More details on funding

Biogen GmbH, own resources

## Regulatory

**Was the study required by a regulatory body?**

No

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

Not applicable

## Methodological aspects

### Study type

#### Study type list

**Study type:**

Non-interventional study

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**Scope of the study:**

Other

**If 'other', further details on the scope of the study**

Biomarker discovery

**Main study objective:**

To identify specific disease-defining metabolic profiles for SMA in urine, blood and CSF before and under therapy with Nusinersen (MetabNMD)

## Study Design

## **Non-interventional study design**

Other

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## **Non-interventional study design, other**

Intensive monitoring schemes

# Study drug and medical condition

## **Medical condition to be studied**

Spinal muscular atrophy

# Population studied

## **Age groups**

- Term newborn infants (0 - 27 days)
  - Infants and toddlers (28 days - 23 months)
  - Children (2 to < 12 years)
  - Adolescents (12 to < 18 years)
  - Adults (18 to < 46 years)
  - Adults (46 to < 65 years)
  - Adults (65 to < 75 years)
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## **Estimated number of subjects**

300

# Study design details

## **Outcomes**

1) to establish a prediction model of disease severity for SMA based on metabolic profiles providing “metabotype-phenotype” correlations and to inform about timepoints of therapy initiation  
2) to establish a biochemical tool for therapeutic monitoring of SMA under antisense-oligonucleotide therapy, gene therapy and further in the future approved therapies

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## **Data analysis plan**

Variables: diagnosis, age, gender, weight, motoric function measures (HFSME, RULM, CHOP-INTENT, 6-MWT). Descriptive statistics: variables will be summarized according to their measurement scale. For continuous variables, mean, median, min, max, quantiles and sd will be computed, for discrete variables frequency counts and percent will be provided. Analysis of basic NMR profiles with TopSpin software (provided by Bruker BioSpin GmbH) according to the publication of Dumas, M.E. and Davidovic, L. Parametric tests and models (e.g. t-Test, ANOVA and regression models) will be used to analyze continuous outcome variables with respect to various predictor variables. When parametric models are inappropriate (e.g. due to large skewness of outcome variables), a non-parametric pendant will be chosen, e.g. Mann-Whitney Test or aligned rank transform (ART) ANOVA. Count data will be analyzed with log-linear models. Multivariate statistical methods: Principal Component Analysis

## **Data management**

## **ENCePP Seal**

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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)

Other

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### Data sources (types), other

Prospective patient-based data collection

## Use of a Common Data Model (CDM)

### CDM mapping

No

## Data quality specifications

### Check conformance

Unknown

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### Check completeness

Unknown

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### Check stability

Unknown

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### Check logical consistency

Unknown

## Data characterisation

### **Data characterisation conducted**

No