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

Study ID

44920

DARWIN EU® study

No

Study countries

☐ Germany

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.

Study status

Planned

Research institutions and networks

Institutions

Heidelberg University Hospital

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Institution

Hamburg Germany, Essen Germany, Gießen Germany, München Germany

Contact details

Study institution contact

Andreas Ziegler andreas.ziegler@med.uni-heidelberg.de

Study contact

andreas.ziegler@med.uni-heidelberg.de

Primary lead investigator

Andreas Ziegler

Primary lead investigator

Study timelines

Date when funding contract was signed
Planned: 30/11/2019

Study start date
Planned: 01/05/2020

Data analysis start date
Planned: 30/11/2020

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

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

Not applicable

Methodological aspects

Study type

Study type list

Study type:

Non-interventional study

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

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)

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 initiation2) 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 discreet 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

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

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

Check completeness

Unknown

Check stability

Unknown

Check logical consistency

Unknown

Data characterisation

Data characterisation conducted

No