Phenotyping and Identification of Biological Markers in STXBP1 Encephalopathy (FIBMEX)

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Study Planned

Administrative details

EU PAS number

EUPAS107644

Study ID

107645

DARWIN EU® study

No

Study countries

☐ Spain

Study description

This is a prospective observational study to evaluate the phenotype of 10 patients under 10 years of age with developmental epileptic encephalopathy due to STXBP1 gene mutation. The study will consist of a clinical and neurodevelopmental evaluation, magnetic resonance imaging, prolonged electroencephalogram, cardiological study, and cerebrospinal fluid biomarker analysis. A 3-year follow-up of these patients will be performed. The aim of the study is, knowing the baseline phenotype, to analyze the response to commonly used drugs and to anticipate the response to different drugs available on the market in this group of patients based on clinical and biomarker evaluation (EEG, MRI and study of specific proteins and neurotransmitters in plasma, urine and CSF).

Study status

Planned

Contact details

Study institution contact

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Study contact

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Primary lead investigator

Antonio Gil-Nagel

Primary lead investigator

Study timelines

Date when funding contract was signed
Planned: 14/11/2023

Study start date
Planned: 01/01/2024

Date of final study report
Planned: 31/12/2027

Sources of funding

- Other

More details on funding

Fundacion Iniciativa para las Neurociencias

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

Main study objective:

- To determine the expression pattern of biomarkers in cerebrospinal fluid in STXBP1. - Characterize electroencephalographic patterns and determine the presence of new markers in EEG signal. - In-depth characterization of MR imaging in patients with STXBP1 mutation

Study Design

Non-interventional study design

Cohort

Population studied

Age groups

- Infants and toddlers (28 days – 23 months)
- Children (2 to < 12 years)

Estimated number of subjects

10

Study design details

Data analysis plan

The study group will be compared with a cohort of patients with similar characteristics to those of the study. That is, in the case of CSF with healthy

subjects of the same age and in the case of MRI, EEG with patients affected by other EEDs and with healthy controls.

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

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