Retrospective and prospective observational study on the systemic outcomes of lysosomal storage diseases with skeletal involvement in their natural history or during standard of care treatment

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Study Planned

Administrative details

EU PAS number

EUPAS1000000746

Study ID

1000000746

DARWIN EU® study

No

Study countries

Italy

Netherlands

Spain

United Kingdom

Study description

This is a multicenter, observational study with both retrospective and prospective components, designed to collect and analyze clinical data from patients affected by LSDs with skeletal and/or neurological involvement. The aim of the study is to collect and analyze data from these patients focusing both on natural history and outcomes of standard of care treatments.
At least 100 participants from different international centers will be enrolled. No additional procedures, laboratory tests, or questionnaires will be performed for the study; only data already collected as part of standard clinical management will be recorded.
Data will be collected from up to 9 visits per patient (maximum 5 retrospective visits and 4 prospective visits).

Study status

Planned

Research institutions and networks

Institutions

IRCCS Ospedale San Raffaele

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Institution

Fondazione IRCCS San Gerardo dei Tintori (Monza, IT)

Azienda Ospedaliera Universitaria Federico II (Napoli, IT)

Royal Manchester Children's Hospital (Manchester, UK)

Wilhelmina Kinderziekenhuis (Utrecht, NLD)

Vall d'Hebron Barcelona Hospital Campus (Barcelona, ES)

Contact details

Study institution contact

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Study contact

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Primary lead investigator

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Primary lead investigator

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Study timelines

Date when funding contract was signed
Planned: 01/09/2025

Study start date
Planned: 01/01/2026

Date of final study report
Planned: 15/01/2033

Sources of funding

- Non-for-profit organisation (e.g. charity)

More details on funding

Fondazione Telethon ETS

Regulatory

Was the study required by a regulatory body?

No

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

Not applicable

Other study registration identification numbers and links

Methodological aspects

Study type

Study type list

Study topic:

Disease /health condition

Study topic, other:

lysosomal storage diseases

Study type:

Non-interventional study

Scope of the study:

Other

If ‘other’, further details on the scope of the study

Natural history and standard of care study

Study design:

This is a multicenter, observational study with both retrospective and prospective components, designed to collect and analyze clinical data from patients affected by LSDs with skeletal and/or neurological involvement.
At least 100 participants from different international centers will be enrolled.

Main study objective:

The primary objective is to assess metabolic, skeletal, and neurological outcomes in patients with LSDs over time, using laboratory, clinical, and radiological parameters.
The secondary objective is to evaluate the systemic outcomes of LSDs, such as pulmonary, ocular, auditory, and cardiological manifestations.

Study Design

Non-interventional study design
Cohort

Study drug and medical condition

Medical condition to be studied
Lysosomal storage disorder

Population studied

Short description of the study population
Adult and pediatric patients with confirmed diagnosis of LSDs presenting skeletal and/or neurological manifestations in accordance with the inclusion/exclusion criteria.
Inclusion criteria:
1. Diagnosis of one of the following LSDs with skeletal and/or neurological manifestations from 2005 onwards:
• Mucopolysaccharidosis type II (MPSII)
• Mucopolysaccharidosis type IVA (MPSIVA)

• GLB1-related disorders (including MPSIVB and GM1 Gangliosidosis)
• Mucopolysaccharidosis type VI (MPSVI)
• Alpha Mannosidosis (MANN)
• Other LSDs, if data are available at the participating center.
2. Genetically confirmed diagnosis supported by biochemical and/or clinical evidence
3. Provision of written informed consent for participation in the study and for the processing of personal data

Exclusion criteria:
1. Patients who, in the opinion of the investigator, do not have enough available data to meet the objectives of the study.

Age groups

- **In utero**
- **Paediatric Population (< 18 years)**
  - Neonate
    - Preterm newborn infants (0 – 27 days)
    - Term newborn infants (0 – 27 days)
  - Infants and toddlers (28 days – 23 months)
  - Children (2 to < 12 years)
  - Adolescents (12 to < 18 years)
- **Adult and elderly population (≥18 years)**
  - Adults (18 to < 65 years)
    - Adults (18 to < 46 years)
    - Adults (46 to < 65 years)
  - Elderly (≥ 65 years)
    - Adults (65 to < 75 years)
    - Adults (75 to < 85 years)

- Adults (85 years and over)

Special population of interest

Frail population

Special population of interest, other

Pediatric patients

Estimated number of subjects

100

Study design details

Comparators

Not applicable

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 source(s), other

Patients clinical charts

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