Prospective observational study of natural history and standard of care in pediatric patients affected by lysosomal storage disorders and diagnosed through newborn screening

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Study Planned

Administrative details

EU PAS number

EUPAS1000001010

Study ID

1000001010

DARWIN EU® study

No

Study countries

Italy

Study description

This multicenter, observational study is designed to systematically characterize the natural history of lysosomal storage disorders (LSDs) from birth under standard of care (SoC). LSDs are rare inherited metabolic diseases caused by lysosomal enzyme deficiencies, leading to progressive accumulation of toxic substrates and multi-organ involvement, frequently affecting the central nervous system and skeletal system. Despite available treatments, including enzyme replacement therapy and hematopoietic stem cell transplantation, substantial unmet clinical needs remain, particularly for neurological and skeletal manifestations. Increasing evidence supports the benefit of early, pre-symptomatic intervention; however, diagnosis often occurs after irreversible disease progression. The implementation of newborn screening programs enables early identification of affected individuals, yet longitudinal real-world data on early disease onset, progression, and treatment response are limited. To address this gap, the study prospectively collects standardized clinical, biochemical, and functional data without altering routine clinical practice, thereby providing robust real-world evidence to improve disease understanding and support the development and evaluation of novel therapeutic strategies, including gene-based approaches.

Study status

Planned

Research institutions and networks

Institutions

IRCCS Ospedale San Raffaele

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Institution

Fondazione IRCCS San Gerardo dei Tintori-Monza

Contact details

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Primary lead investigator

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Study timelines

Date when funding contract was signed

Planned: 01/09/2026

Study start date

Planned: 01/09/2026

Date of final study report
Planned: 01/12/2032

Sources of funding

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

More details on funding

Fondazione Regionale per la Ricerca Biomedica

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

TIGET18-LSD-NBS

Methodological aspects

Study type

Study type list

Study topic:

Disease /health condition

Study topic, other:

Lysosomal storage disorder

Study type:

Non-interventional study

Scope of the study:

Disease epidemiology

Other

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

Natural history and standard of care study in patients identified after new born screening

Data collection methods:

Primary data collection

Study design:

This is a multicenter, observational study with prospective data collection, aimed at evaluating patients during their natural history and/or under standard of care, without modifying routine clinical practice.

Main study objective:

Characterize early clinical outcomes of LSD patients diagnosed through NBS, including disease evolution during natural history and under SoC, with a particular focus on skeletal, metabolic and neurological manifestations.

Study Design

Non-interventional study design

Case-only

Study drug and medical condition

Medical condition to be studied

Lysosomal storage disorder

Gaucher's disease

Mucopolysaccharidosis I

Mucopolysaccharidosis II

Mucopolysaccharidosis IV

Mucopolysaccharidosis VI

Alpha-mannosidosis

Population studied

Short description of the study population

Pediatric patients with a confirmed diagnosis of Lysosomal storage disorder (LSD) identified through newborn screening (NBS) and referred to participating clinical centers for follow-up under standard of care.

Special population of interest

Frail population

Special population of interest, other

Newborns

Estimated number of subjects

10

Study design details

Setting

All patients enrolled

Comparators

None

Outcomes

• Skeletal outcomes assessed through clinical examination, instrumental findings, and functional parameters

• Metabolic outcomes assessed through disease-specific biochemical markers.

• Neurological outcomes assessed through neurological and development assessments.

All outcomes will be assessed longitudinally both during the natural history phase and, when applicable, after initiation of standard of care treatment.

Data analysis plan

Descriptive analysis. Categorical and continuous variables will be summarized using standard descriptive statistics. Longitudinal data may be analyzed using either linear or nonlinear mixed effects models, depending on data distribution and feasibility.

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)

Hospital Information System

Data sources (types)

Clinical trial

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