A GLOBAL PROSPECTIVE OBSERVATIONAL REGISTRY OF PATIENTS WITH POMPE DISEASE

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Study Planned

Administrative details

EU PAS number

EUPAS107299

Study ID

107300

DARWIN EU® study

No

Study countries

Argentina

Australia

Austria

Belgium

Bosnia and Herzegovina
Brazil
Canada
Chile
Czechia
Denmark
Estonia
France
Germany
Greece
Hungary
Israel
Italy
Japan
Korea, Republic of
Netherlands
New Zealand
Poland
Portugal
Serbia
Slovakia
Slovenia
Spain
Sweden
Taiwan
Thailand
United Kingdom
United States

Study description

The goal of this registry is to assess clinical outcomes in patients with Pompe disease, including patients with late-onset Pompe disease (LOPD) or infantile-onset Pompe disease (IOPD), regardless of current or previous therapy.

Study status

Planned

Research institutions and networks

Institutions

Amicus Therapeutics

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Institution

Contact details

Study institution contact

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Study contact

jgiuliano@amicusrx.com

Primary lead investigator

Roberts Mark

Primary lead investigator

Study timelines

Date when funding contract was signed
Planned: 01/10/2023

Study start date
Planned: 01/10/2023

Date of final study report
Planned: 30/12/2033

Sources of funding

- Pharmaceutical company and other private sector

More details on funding

Amicus Therapeutics Inc

Regulatory

Was the study required by a regulatory body?

Yes

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

EU RMP category 3 (required)

Other study registration identification numbers and links

Methodological aspects

Study type

Study type list

Study topic:

Disease /health condition

Study type:

Non-interventional study

Scope of the study:

Disease epidemiology

Effectiveness study (incl. comparative)

Safety study (incl. comparative)

Main study objective:

- To evaluate the long-term safety of Pompe disease treatments through collection of data that describe the frequency of AEs/SAEs occurring in Pompe disease patients - To evaluate the long-term real-world effectiveness - To evaluate the long-term real-world impact of Pompe disease treatments on QOL and PROs - To describe the natural history of untreated Pompe disease

Study Design

Non-interventional study design

Cohort

Study drug and medical condition

Medicinal product name

POMBILITI

OPFOLDA

Population studied

Age groups

- 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)
- Adults (18 to < 46 years)
- Adults (46 to < 65 years)
- Adults (65 to < 75 years)
- Adults (75 to < 85 years)
- Adults (85 years and over)

Estimated number of subjects

500

Study design details

Data analysis plan

No formal hypotheses will be tested in this registry. Demographics and medical history (including but not limited to disease stage, time since diagnosis, prior and concomitant medication use, prior treatment with ERTs, and co-morbidities) will be summarized using descriptive statistics, with number and percent for categorical variables, and n, mean, SD, SE of the mean, median, minimum, and maximum for continuous variables. Quantitative analyses may include incidence rates of events, reasons for dropout and discontinuation, time-to-event profiles, and descriptions of clinical outcomes such as impact on physical function, HRQOL, and the occurrence of AEs, including but not limited to IARs, hypersensitivity reactions (including anaphylaxis), immune complex related reactions, and medication errors in the home infusion setting. Demographics and medical history will also be summarized using descriptive statistics on different subgroups.

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)

Disease registry

Electronic healthcare records (EHR)

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