A Prospective Study in Subjects with Late-onset Pompe Disease who are Currently Being Treated with Enzyme Replacement Therapy

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The purpose of the study is to evaluate changes in key clinical outcome measures (eg, motor, respiratory, fatigue) in adult subjects with late-onset Pompe disease (LOPD) receiving standard-of-care enzyme replacement therapy (ERT). Additionally,...

Ethical review	Approved WMO
Status	Pending
Health condition type	Musculoskeletal and connective tissue disorders congenital
Study type	Observational invasive

Summary

ID

NL-OMON46633

Source
ToetsingOnline

Brief title
Amicus POM003 Study

Condition

- Musculoskeletal and connective tissue disorders congenital
- Musculoskeletal and connective tissue disorders congenital

Synonym
Glycogen storage disease type II, Pompe disease

Research involving
Human

Sponsors and support

Primary sponsor: Amicus Therapeutics, Inc.
Source(s) of monetary or material Support: Amicus Therapeutics;Inc.

Intervention

Keyword: Enzyme Replacement Therapy, Late-onset Pompe disease, Prospective study

Outcome measures

Primary outcome

The following parameters will be assessed throughout the study. Historical data

for these parameters will also be collected.

* 6MWT

* Pulmonary Function Tests (FVC, VC, maximum inspiratory pressure [MIP], and

sniff nasal inspiratory pressure [SNIP])

* Motor Function Tests (Timed Up and Go [TUG] test and Gait, Stairs, Gower, and

Chair maneuver [GSGC])

* Muscle Strength Testing (manual muscle strength testing and hand-held

dynamometer measurements)

* Patient-reported Outcomes (PRO), including Rasch-built Pompe-specific

activity; Patient-reported Outcomes Measurement Information System (PROMIS®)

for dyspnea, fatigue, physical functioning, and upper extremity; and EuroQol-5

Dimensions-5 Levels (EQ-5D-5L)

* Adverse events

* Prior and concomitant medications

* Medical history

* Electrocardiograms (ECG)

* Physical examinations

* Vital signs, weight, and height

* Clinical laboratory assessments

* Pharmacodynamic assessments (creatine kinase [CK] and hexose tetrasaccharide [Hex4])

* Infusion-associated reactions

* Anti-recombinant human acid alpha-glucosidase (rhGAA) antibodies (total and neutralizing). The type of assay used and the facility performing the assay will be captured for retrospective antibody results.

* Physician*s Global Impression of Change

* Subject Global Impression of Change

Secondary outcome

Not applicable

Study description

Background summary

Pompe disease, also known as acid maltase deficiency or glycogen storage disease type II, is an autosomal recessive genetic disorder caused by mutations in the GAA gene that encodes acid *-glucosidase (GAA), an enzyme that catalyzes the breakdown of lysosomal glycogen.

The objective of this study is to evaluate the baseline characteristics and degree of change over time in clinical outcome measures commonly used to evaluate patients with LOPD.

Study objective

The purpose of the study is to evaluate changes in key clinical outcome measures (eg, motor, respiratory, fatigue) in adult subjects with late-onset Pompe disease (LOPD) receiving standard-of-care enzyme replacement therapy

(ERT). Additionally, information gained may be used in the design and conduct of future studies in LOPD subjects.

Study design

This is a prospective, multi-center, study

Study burden and risks

There will be 9 visits.

The following study procedures will be performed:
Physical evaluation
ECG
Blood draws including genetic test (venipuncture)
Urine collections
Pulmonary function tests(Spirometer, respiratory pressure meter)
Motor function tests
Muscle strength tests
Questionnaires
Pregnancy test

Risks: When taking the bloodsamples redness, swelling and/or pin can occur at the site of injection. When removing the ECG some irritation of the skin can occur. The genetic test for Pompe disease can be uncomfortable for the subjects.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

1. Subject has a diagnosis of Pompe disease based on documented deficiency of acid *-glucosidase (GAA) activity and a documented GAA mutation (the gene that encodes GAA).
2. Male and female subjects between 18 years and 75 years, inclusive and * 50 kg.
3. Subject must provide signed informed consent prior to performing any study-related procedures.
4. Subject must be currently receiving standard-of-care ERT (alglucosidase alfa) at the recommended dose (approximately 20 mg/kg dose) every other week and for the past 2 years or more
5. Subject must be able to perform pulmonary testing and muscle function testing in a seated position.
6. Subjects must have an upright forced vital capacity (FVC) within 35 to 90% of predicted normal (NHANES III reference values), based on the higher of the screening or baseline value, if their 6MWD is >200 m. Subject must have an upright FVC within 40 to 90% of redicted normal (NHANES III reference values), based on the higher of the screening or baseline value, if their 6MWD is *200m. If FVC is between 80 and 90% of predicted normal, the subject may enter the study if the percent predicted FVC value drops by 10% predicted or more in supine position.
7. Subject is able to walk at least 100 m in the 6-minute walk test (6MWT) and the assessment is noted as valid.

Exclusion criteria

1. Subject has received any investigational therapy or pharmacological treatment for Pompe disease, other than alglucosidase alfa, within 30 days or 5 half-lives, whichever is shorter, prior to theBaseline Visit or is anticipated to do so during the course of the study.

2. Subject is on any of the following prohibited medications within 30 days or 5 half-lives, whichever is shorter, prior to
Baseline, or is anticipated to do so during the course of the study:
* miglitol (eg, Glyset)
* miglustat (eg, Zavesca)
* acarbose (eg, Precose, Glucobay)
* voglibose (eg, Volix, Vocarb, Volibo)
3. Subject requires use of invasive or non-invasive ventilatory support for
> 6 hours a day while awake.
4. Subject has a medical or any other extenuating condition or circumstance that may, in the opinion of the investigator, pose an undue safety risk to the subject or compromise his/her ability to comply with protocol requirements. This includes clinical depression (as diagnosed by a psychiatrist or other mental health professional) with uncontrolled or poorly controlled symptoms.
5. Subject is breastfeeding, or is pregnant or planning to become pregnant within the next 2 years
6. Other exclusion criteria according to the Lumizyme/Myozyme instructions for use.

Study design

Design

Study phase:	4
Study type:	Observational invasive
Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Treatment

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	03-09-2018
Enrollment:	3
Type:	Anticipated

Ethics review

Approved WMO

Date:	28-08-2018
Application type:	First submission
Review commission:	METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam)

Study registrations

Followed up by the following (possibly more current) registration

No registrations found.

Other (possibly less up-to-date) registrations in this register

No registrations found.

In other registers

Register	ID
ClinicalTrials.gov	NCT03347253
CCMO	NL65102.078.18

Study results