Protocolised follow-up of Pompe patients receiving enzyme replacement therapy on a compassionate use basis

Submission date	Recruitment status	Prospectively registered
23/02/2007	Recruiting	Protocol
Registration date	Overall study status	Statistical analysis plan
23/02/2007	Ongoing	Results
Last Edited	Condition category	Individual participant data
01/07/2016	Nutritional, Metabolic, Endocrine	Record updated in last year

Plain English summary of protocol

Not provided at time of registration

Contact information

Type(s)

Scientific

Contact name

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Contact details

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Additional identifiers

Protocol serial number N/A

Study information

Scientific Title

Protocolised follow-up of Pompe patients receiving enzyme replacement therapy on a compassionate use basis

Study objectives

Enzyme therapy with recombinant human alpha glucosidase results in:

- 1. Prolonged survival
- 2. Improvement or stabilisation of cardiac hypertrophy and function
- 3. Improvement or stabilisation of pulmonary function
- 4. Improvement or stabilisation of muscle function and strength

Ethics approval required

Old ethics approval format

Ethics approval(s)

Ethics approval received from the local medical ethics committee

Study design

Protocolised follow up of parallel group trial

Primary study design

Observational

Study type(s)

Treatment

Health condition(s) or problem(s) studied

Pompe Disease

Interventions

Enzyme replacement therapy

Intervention Type

Drug

Phase

Not Specified

Drug/device/biological/vaccine name(s)

Recombinant human alpha glucosidase

Primary outcome(s)

- 1. Infantile: Survival
- 2. Late-onset: Improvement and/or stabilisation of muscle function

Key secondary outcome(s))

- 1. Infantile:
- a. improvement of cardiac hypertrophy and function
- b. achievement of motor milestones
- 2. Late-onset:
- a. improvement and/or stabilisation of pulmonary function
- b. improvement of quality of life

Completion date

01/01/2050

Eligibility

Key inclusion criteria

- 1. Confirmed diagnosis of Pompe Disease
- 2. Infantile-onset:
- 2.1. Age less than one year
- 2.2. Delayed motor milestones, and/or
- 2.3. Hypertrophic cardiomyopathy
- 3. Late-onset:
- 3.1. 24 hour/day artificial ventilation
- 3.2. Wheelchair bound
- 3.3. Previously enrolled in AGLU 1202 study

Participant type(s)

Patient

Healthy volunteers allowed

No

Age group

Not Specified

Sex

Not Specified

Key exclusion criteria

- 1. Infantile-onset:
- 1.1. congenital abnormalities
- 1.2. allergy to food and/or proteins
- 1.3. ventilator dependency
- 2. Late-onset:
- 2.1. developmental delays not explained by Pompe's Disease
- 2.2. allergies
- 2.3. severe co-morbidity

Date of first enrolment

01/01/1999

Date of final enrolment

01/01/2050

Locations

Countries of recruitment

Netherlands

Study participating centre Erasmus Medical Centre Rotterdam Netherlands 3000 CB

Sponsor information

Organisation

Erasmus Medical Centre (Netherlands)

ROR

https://ror.org/018906e22

Funder(s)

Funder type

Industry

Funder Name

Genzyme Corporation (Netherlands)

Results and Publications

Individual participant data (IPD) sharing plan

IPD sharing plan summary

Not provided at time of registration

Study outputs

Output type Details Date created Date added Peer reviewed? Patient-facing?

Participant information sheet Participant information sheet 11/11/2025 No Yes