The incidence of late-onset Pompe disease in subjects with obstructive sleep apnea

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The aim of this study is to estimate the prevalence of late-onset inherited Pompe disease in a large cohort of patients with recently diagnosed obstructive sleep apnea syndrome (OSAS) in two different centres across Europe.

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Upper respiratory tract disorders (excl infections)
Study type	Observational invasive

Summary

ID

NL-OMON36188

Source
ToetsingOnline

Brief title
Pios

Condition

- Upper respiratory tract disorders (excl infections)

Synonym
rare muscle disorder, sleep disorder

Research involving
Human

Sponsors and support

Primary sponsor: Leids Universitair Medisch Centrum
Source(s) of monetary or material Support: Firma Genzyme

Intervention

Keyword: Pompe disease, Sleep apnea

Outcome measures

Primary outcome

deficiency of the lysosomal enzyme acid -glucosidase

Secondary outcome

none

Study description

Background summary

Patients with obstructive sleep apnea/hypopnea (OSA) have, on average, a narrower and more collapsible upper airway relative to others. They may overcome the abnormal upper airway mechanics during wakefulness by increased activation of upper airway dilators but develop OSA in sleep because of a reduction in basal upper airway muscle activity and attenuation of the mechanisms involved in activating upper airway dilators in response to negative pressure. Patients with abnormal passive upper airway mechanics are more dependent on upper airway muscle activation to maintain upper airway patency and, hence, are more vulnerable to sleep-related changes in neuromuscular control of pharyngeal muscles. Patients who have respiratory insufficiency related to neuromuscular disease are at high risk for sleep apnea and nocturnal hypoventilation, and these conditions often occur before the development of daytime respiratory insufficiency. Early detection of respiratory insufficiency is critical in the management of patients who have late-onset Pompe disease. This is a slowly progressive muscle disorder based on acid maltase deficiency, is a rare genetic lysosomal storage disorder caused by an absence or deficiency of the lysosomal enzyme acid -glucosidase (GAA). Late-onset Pompe disease is characterized by substantial involvement of skeletal muscle, which leads to progressive muscle weakness and respiratory insufficiency. In late-onset Pompe disease, death is usually due to respiratory complications.

Study objective

The aim of this study is to estimate the prevalence of late-onset inherited Pompe disease in a large cohort of patients with recently diagnosed obstructive

sleep apnea syndrome (OSAS) in two different centres across Europe.

Study design

This is a cross-sectional, explorative study to measure the prevalence of late-onset Pompe disease in subjects with an established diagnosis of obstructive sleep apnea syndrome. In two different study centers in Europe (Leiden, Copenhagen) patients will be asked to donate a blood sample for the detection of Acid -glucosidase enzyme activity. Patient information sheets will be mailed to all patients known in an OSAS database of the clinic. The medical record of each patient will be checked to know if the in- and exclusion criteria of this study are met.

Study burden and risks

n.v.t.

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

a.age over 18 years
b.a diagnosis of OSAS no older than 5 years
c. Diagnosis of OSAS confirmed by polysomnography with a apneu-hypopneu index higher than 15

Exclusion criteria

a.severe left-sided heart failure
b.active psychiater-diagnosed mental disorder, thereby not able to provide informed consent
c.clinical suspicion on vascular cerebral accident located in the ventral medulla of the brain stem.

Study design

Design

Study type: Observational invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Diagnostic

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	09-02-2012
Enrollment:	500
Type:	Actual

Ethics review

Approved WMO Date:	19-09-2011
Application type:	First submission
Review commission:	METC Leiden-Den Haag-Delft (Leiden)
	metc-ldd@lumc.nl

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
CCMO	NL37026.058.11