Assessment of cardio vascular risk by measurement of carotid intima-media thickness (cIMT) in adult type I Gaucher disease patients.

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To determine, by measuring cIMT's, whether the abnormal lipid profile of GD I carriers and patients is associated with atherosclerosis and an increased risk of CAD.

Ethical review	Approved WMO
Status	Pending
Health condition type	Metabolic and nutritional disorders congenital
Study type	Observational invasive

Summary

ID

NL-OMON29706

Source
ToetsingOnline

Brief title
Intima media thickness in Gaucher type I disease.

Condition

- Metabolic and nutritional disorders congenital

Synonym
Gaucher disease type I, glucocerebrosidase deficiency

Research involving
Human

Sponsors and support

Primary sponsor: Academisch Medisch Centrum
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: cardiovascular disease, cholesterol, Gaucher disease type I, intima media thickness

Outcome measures

Primary outcome

cIMT

Secondary outcome

total cholesterol, LDL, HDL.

Study description

Background summary

Gaucher disease type I (GD I) is the most common lysosomal storage disorder, caused by deficiency of the enzym glucocerebrosidase. Concentrations of total plasma cholesterol, LDL and HDL are reduced in a large proportion of patients with GD I. Also in GD carriers low HDL-c levels are found. In numerous epidemiologic studies it has been shown that low plasma HDL levels are associated with increased risk for coronary artery disease (CAD). A non-invasive validated biomarker for the status of atherosclerosis and present and future cardiovascular disease risk is the ultrasonographically measured carotid intima-media thickness (cIMT).
In order to study whether the abnormal lipid profile in GD I patients and carriers is associated with atherosclerosis and, potentially, an increased risk of cardiovascular disease, we will measure arterial cIMT in GD I patients, carriers and controls, who are matched for age, sex and smoking status.

Study objective

To determine, by measuring cIMT's, whether the abnormal lipid profile of GD I carriers and patients is associated with atherosclerosis and an increased risk of CAD.

Study design

A cross sectional, observational study will be performed in Gaucher disease type I patients, carriers and unaffected controls. Each participant will be subjected to a single IMT measurement. Blood samples will be obtained for lipid

profiling, DNA and biochemical analysis.

Study burden and risks

The burden for participants consists of a single cIMT, a venapuncture (max 39 mL) after an overnight fast, a questionaire and a physical examination.
The only risk involved is the risk associated with a venapuncture (hematoma, bleeding, inflammation).

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

-Patients, older than 18 years, with proven GD I, as evidenced by decreased plasma

glucocerebrosidase activity or genotyping.
-Carriers, older than 18 years, with proven heterozygosity for the GD I genotype.
-Controls, older than 18 years, with proven absence of the four most prevalent Gaucher-mutations, and, if related to a Gaucher patient, absence of the Gaucher mutations present in their family.
-Patients, carriers and controls have to provide written informed consent to participate in the study.

Exclusion criteria

none

Study design

Design

Study type:	Observational invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)
Control:	Active
Primary purpose:	Basic science

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-09-2006
Enrollment:	140
Type:	Anticipated

Ethics review

Approved WMO

Application type:	First submission
Review commission:	METC Amsterdam UMC

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	NL13350.018.06