Small fibre neuropathy in Fabry*s disease

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

Summary

ID

NL-OMON31065

Source
ToetsingOnline

Brief title
Small fibre neuropathy in Fabry*s disease

Condition

- Metabolic and nutritional disorders congenital
- Peripheral neuropathies

Synonym
painful neuropathy, small diameter nerve fibre neuropathy

Research involving
Human

Sponsors and support

Primary sponsor: Academisch Medisch Centrum
Source(s) of monetary or material Support: Ministerie van OC&W,Fonds lysosomale stapelingsziekten IAM12005

Intervention

Keyword: Fabry□s disease, Pathophysiology, Small fibre neuropathy

Outcome measures

Primary outcome

The QST protocol will be expressed as Z-score QST profiles. Skin biopsies will be expressed as intra-epidermal nerve fibre densities (IENFD). An IENFD of less than the 5th percentile of healthy humans is considered to be abnormal. Autonomic function will be expressed as changes in heart rate and blood pressure in response to standing up, forced breathing and the Valsalva*s manoeuvre. The results will be compared to well-established normative values per age-group.

Secondary outcome

None

Study description

Background summary

Small fibre neuropathy (SFN), an axonal sensory neuropathy affecting unmyelinated (C) and thinly myelinated (A*) fibres, is one of the key features of Fabry*s disease. Currently, the pathophysiology is poorly understood. By applying (pain) questionnaires, the QST protocol, skin biopsies and autonomic function tests to Fabry patients of different age groups, we want to get insight in the pathophysiology of small fibre neuropathy in male Fabry patients and female carriers.

Study objective

The objective is to get insight in the pathophysiology of small fibre neuropathy in Fabry patients.
Insight in the pathophysiology will be obtained by:
1. Describing results obtained by the QST protocol, skin biopsies and autonomic

function tests in male Fabry patients and female carriers;
2. Looking for associations between results obtained by the QST protocol, skin biopsies and autonomic function tests, stratified to different age-groups and specific measures of disease severity, i.e. pain severity, renal function, left ventricular mass and cerebral manifestations.

Study design

Cross sectional cohort study

Study burden and risks

Included patients will undergo questionnaires, a physical examination, quantitative sensory testing, a skin biopsy and autonomic function tests. The QST protocol and autonomic function tests take about 60 minutes each but are otherwise not harmful or distressing. Skin biopsies are temporarily painful but without risk.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adolescents (12-15 years)
Adolescents (16-17 years)
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

1. Age 12 years or older
2. A diagnosis of Fabry disease as proven by enzyme activity (males) or DNA mutation analysis (females)

Exclusion criteria

1. Pre-existent venous insufficiency, confirmed by echo Doppler or a history of ulcus cruris

Study design

Design

Study type: Observational invasive

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

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-03-2007
Enrollment:	60
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	NL16407.018.07