# Small fibre neuropathy in Fabry*s disease

Published: 27-02-2007
Last updated: 08-05-2024

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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

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|---|---|
| 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 |