The Hamlet study. Fabry or not Fabry: Better diagnosis of Fabry disease.

No registrations found.

Ethical review	Not applicable
Status	Recruiting
Health condition type	-
Study type	Observational non invasive

Summary

ID

NL-OMON27748

Source
Nationaal Trial Register

Brief title
Hamlet study

Health condition

Fabry disease
Diagnosis
Algorithm

Sponsors and support

Primary sponsor: TIpharma consortium
Academia: Amc
pharma: Genzyme, a sanofy company
subsidizing party: Shire HGT
Source(s) of monetary or material Support: TIpharma consortium
Academia: Amc
pharma: Genzyme, a sanofy company
subsidizing party: Shire HGT

Intervention

Outcome measures

Primary outcome

Diagnostic criteria to determine if an individual has true Fabry disease or a non disease causing genetic variation.

These criteria will be incorporated in diagnostic algorithms per organ system (e.g. Heart, kidney).
These algorithms will serve to:

1. Improve early identification of true Fabry patients, who may benefit from treatment and counseling;

2. Avoid misdiagnosis and unjustified treatment in individuals who do not have Fabry disease;

3. Improve the understanding of the phenotypic variability of Fabry disease by exploring each organ system;

4. Improve the understanding of the value of biochemical and genetic characterization of Fabry patients.

Secondary outcome

N/A

Study description

Background summary

The aim of the current study is to improve the use of clinical, imaging and laboratory assessments for early identification of the truly affected Fabry patient. This patient might subsequently benefit from treatment and counseling, while an individual who does not have Fabry disease will not be subject to the burden of having a genetic disease and time consuming and very costly treatment.
The added value of assessing small fiber neuropathy for improved diagnosis is studied in a separate protocol that is part of the TI-pharma project T6-504, and is also registered in the 'Nederlands Trial Register'.

Study objective

Individuals with a single, non-specific symptom are often identified with a genetic variation of unknown significance in de AlfaGalactosidase A gene that is involved in Fabry disease. This

study aims to develop diagnostic algorithms to improve the identification of true Fabry patients.

Study design

3/2013: 2nd review of algorithms based upon new information. Report back to UMCs on study progress; Second newsletter.

9/2013: Meeting: Review outcomes, adjust algorithms, third newsletter.

12/2013: Final report/ publications in peer reviewed Medical journals, fourth newsletter.

Intervention

All organ systems will be explored using clinical and biochemical assessments that are part of the standard of care. There are no additional study interventions.

Contacts

Public
Dept of Internal Medicine, div of Endocrinology and Metabolism
F5-165
Academic Medical Center
Meibergdreef 9
Linda Tol, van der
Amsterdam 1105 AZ
The Netherlands
+31 (0)20 5666071
Scientific
Dept of Internal Medicine, div of Endocrinology and Metabolism
F5-165
Academic Medical Center
Meibergdreef 9
Linda Tol, van der
Amsterdam 1105 AZ
The Netherlands
+31 (0)20 5666071

Eligibility criteria

Inclusion criteria

Males:

Decrease in alpha-galactosidase A activity in leucocytes, plasma or fibroblasts according to local laboratory criteria AND presence of a mutation in the alpha-galactosidase A gene of uncertain clinical relevance.

Females:

Presence of a mutation in the alpha-galactosidase A gene of uncertain clinical relevance.

Exclusion criteria

Patient is unwilling to participate.

Study design

Design

Study type:	Observational non invasive
Intervention model:	Parallel
Allocation:	Non controlled trial
Masking:	Open (masking not used)
Control:	N/A , unknown

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	01-01-2012
Enrollment:	25
Type:	Anticipated

Ethics review

Not applicable
Application type: Not applicable

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
NTR-new	NL3670
NTR-old	NTR3840
Other	TI pharma : T6-504
ISRCTN	ISRCTN wordt niet meer aangevraagd.

Study results

Summary results
N/A