THE PREVALENCE OF FABRY DISEASE IN MALES WITH RENAL SYMPTOMS OF UNKOWN CAUSE, VISITING THE OUTPATIENT CLINIC OF A LARGE TEACHING HOSPITAL

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To determine the prevalence of α-Galactosidase A deficiency in male patients presenting with renal failure, proteinuria or micro-albuminuria of unknown cause, at the outpatient clinic of the department of internal medicine of a large teaching...

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

Summary

ID

NL-OMON29934

Source
ToetsingOnline

Brief title
Prevalence of renal disease in rmale patients with renal disease

Condition

- Metabolic and nutritional disorders congenital

Synonym
alpha-Galactosidase deficiency, storage diseases

Research involving
Human

Sponsors and support

Primary sponsor: Onze Lieve Vrouwe Gasthuis
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: Fabry, Prevalence, proteinuria, renal insufficiency

Outcome measures

Primary outcome

The prevalence of αGal A deficiency will be recorded.

Secondary outcome

none

Study description

Background summary

Fabry disease is an X-linked disorder caused by the deficiency of the lysosomal enzyme alpha Galactosidase A (*-Gal a), resulting in accumulation of specific glycosphingolipids in (vascular) endothelial cells. In childhood males suffer from severe pains in hands and feet (acroparesthesias), hypo- or anhydrosis and develop angiokeratoma at trunk and genitals. Later in life, they develop vascular complications due to the ongoing endothelial glycolipid accumulation, of which renal insufficiency often progressing to end stage renal failure requiring dialysis is a main feature 1. The clinical picture of the disorder is highly variable. In female carriers the clinical picture is even more heterogeneous, albeit with a more protracted course. It is possible that due to its variability in clinical expression, the above mentioned symptoms are not always recognised as Fabry disease, resulting in underdiagnosis and consequently underestimation of its prevalence. The prevalence of Fabry disease in large dialysis registry programs, was shown to be 0,019% and 0,017%, in Europe and the US, respectively 2 3. Reports on the prevalence of Fabry disease in such registries depend on a correct diagnosis. In contrast, Utsumi et al. diagnosed Fabry disease in 2 of 440 males (0,45%) with renal failure in Japan 4. In the Netherlands, screening of 508 male dialysis patients revealed only one Fabry disease patient, who was already known with Fabry disease 5.

To date, screening of patients with moderate renal function disorders,

micro-albuminuria or proteinuria has not been performed.

Study objective

To determine the prevalence of α-Galactosidase A deficiency in male patients presenting with renal failure, proteinuria or micro-albuminuria of unknown cause, at the outpatient clinic of the department of internal medicine of a large teaching hospital.

Study design

Observational, prospective.

Study burden and risks

none

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

• Male origine, AND
• A serum kreatinin > 100µmol/l, OR
• A calculated kreatinin-clearance <80 ml/min OR
• Proteinuria (>0,3 g/l), OR
• Microalbuminuria (>0,03 g/l).

Exclusion criteria

• Female .
• Known cause of renal disease, as documented by renal biopsy, laboratory evaluation or by clinical judgment by the physician.

Study design

Design

Study type: Observational non invasive

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

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-08-2006
Enrollment:	1000
Type:	Anticipated

Ethics review

Approved WMO	
Application type:	First submission
Review commission:	MEC-U: Medical Research Ethics Committees United (Nieuwegein)

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	NL13587.067.06