Identification of the molecular basis of premature atherosclerosis in high penetrance pedigrees

Published: 15-09-2009
Last updated: 04-05-2024

We aim to identify sequence variations within pedigrees and determine how these may result in CVD by studying the effect on cell phenotype.

Ethical review	Approved WMO
Status	Pending
Health condition type	Cardiac and vascular disorders congenital
Study type	Observational invasive

Summary

ID

NL-OMON33048

Source
ToetsingOnline

Brief title
PAS PEDIGREES

Condition

- Cardiac and vascular disorders congenital
- Arteriosclerosis, stenosis, vascular insufficiency and necrosis

Synonym
atherosclerosis, cardiovascular disease

Research involving
Human

Sponsors and support

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

Intervention

Keyword: Genetics, Pedigree studies, Premature Atherosclerosis

Outcome measures

Primary outcome

A) Identification of sequence variations resulting in CVD in pedigrees

B) Determining through which mechanism sequence variations result in disease.

Secondary outcome

none

Study description

Background summary

Myocardial infarction is a leading cause of mortality and morbidity worldwide. A number of well validated risk factors have been identified over the last decades for cardiovascular disease (CVD) such as smoking, hypertension, diabetes, obesity and dyslipidemias. In addition to these traditional factors, several studies have confirmed that a family history of CVD is an independent risk factor. However, the genetic basis for CVD is still not completely understood. Myocardial infarctions in younger individuals have been associated with substantially greater heritability. Thus, early-onset myocardial infarction is a promising phenotype for mapping of genetic risk factors for CVD4. At the Academic Medical Centre (AMC) in Amsterdam we have identified pedigrees with several cases of early-onset CVD. It is tempting to speculate that novel, high-penetrance mutation segregates in some of these pedigrees. We will investigate these pedigrees.

Study objective

We aim to identify sequence variations within pedigrees and determine how these may result in CVD by studying the effect on cell phenotype.

Study design

Observational Pedigree Analysis

Study burden and risks

The burden for participants is a venipuncture and a skin biopsy of 1mm. The risks are haematomas, a 1mm scar or bleeding. There is no direct benefit for the participants. However in general more insight will be created in the molecular basis of CVD.

Contacts

Public
Academisch Medisch Centrum

Meibergdreef 15
1105 AZ Amsterdam
NL
Scientific
Academisch Medisch Centrum

Meibergdreef 15
1105 AZ Amsterdam
NL

Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

Member of high penetrence family for cardiovascular disease

Exclusion criteria

None

Study design

Design

Study type: Observational invasive

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

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-06-2009
Enrollment:	1000
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	NL28156.018.09