Familial hypercholesterolemia identification through innovative use of existing laboratory data

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Primary objective: in this study, we want to investigate the performance of FH identification with a new approach using existing clinical laboratory data in which patients with severe hypercholesterolemia (LDL-C > 99.5th percentile for age and...

Ethical review	Approved WMO
Status	Pending
Health condition type	Arteriosclerosis, stenosis, vascular insufficiency and necrosis
Study type	Observational invasive

Summary

ID

NL-OMON51701

Source
ToetsingOnline

Brief title
PELICAN

Condition

- Arteriosclerosis, stenosis, vascular insufficiency and necrosis

Synonym
Cardiovascular disease

Research involving
Human

Sponsors and support

Primary sponsor: Medisch Diagnostische Centra Atalmedial
Source(s) of monetary or material Support: Medisch Diagnostische Centra Atalmedial

Intervention

Keyword: Clinical criteria, Familial Hypercholesterolemia, Laboratory data, Screening methods

Outcome measures

Primary outcome

The main outcome in this study will be prevalence of genetically confirmed FH

in a preselected population of individuals with severe hypercholesterolemia

(diagnostic yield). DNA genotyping in this study will be performed using a

customized Illumina GSA v3 array.

Secondary outcome

The secundary study parameter will be the diagnostic yield of existing clinical

criteria for genetically confirmed FH.

Study description

Background summary

Familial hypercholesterolemia (FH) is an autosomal dominant disorder characterized by lifelong elevated low-density lipoprotein cholesterol (LDL-C) resulting in an up to 25.8-fold increased risk for premature cardiovascular disease (CVD) in heterozygous FH (HeFH) patients. Based on an estimated 1:250 prevalence, the number of HeFH patients in the Netherlands is around 70.000. Unfortunately, over 50% of these patients remain unidentified. Novel screening methods and optimization of the use of clinical scoring systems are needed to identify these remaining undiagnosed FH patients.

Study objective

Primary objective: in this study, we want to investigate the performance of FH identification with a new approach using existing clinical laboratory data in which patients with severe hypercholesterolemia (LDL-C > 99.5th percentile for age and sex) are selected.

Our secondary outcome is the comparison of the following FH clinical scoring

systems in their ability to predict the presence of an FH causing mutation: Dutch Lipid Clinic Network (DLCN) criteria, Familial Hypercholesterolemia Case Ascertainment (FAMCAT) Tool, Make Early Diagnosis to Prevent Early Deaths (MEDPED) criteria, criteria of the Simon Broome registry (SB), and the more recently developed and validated criteria by Besseling et al.

Study design

This is a single-center, cross-sectional observational study.

Study burden and risks

The results of this study will contribute to optimization of the identification of FH patients, which has been a major issue with this disorder. This study will teach us whether central laboratories, like Atalmedial, can indeed play a role in identifying new FH patients, and subsequent cascade testing of first degree relatives. Early detection of FH patients will result in early benefit from lipid lowering therapies, greatly reducing CVD risk. In addition, the identification of these FH patients will enable cascade screening of their first degree family members, who in turn can be treated (at a young age) as well. The expected risk for participants in this study is low, as the only intervention will be a 10 ml venous blood withdrawal.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)

Inclusion criteria

Patients with severe hypercholesterolemia (LDL-cholesterol level above 99,5th percentile for age and sex).

Exclusion criteria

Three failed blood withdrawal attempts.

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):	01-07-2022
Enrollment:	600
Type:	Anticipated

Ethics review

Approved WMO Date:	31-10-2022
Application type:	First submission
Review commission:	METC Amsterdam UMC
Approved WMO Date:	24-11-2022
Application type:	Amendment
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	NL81583.018.22