Coagulation in Lamin A-C mutations

Published: 16-09-2009
Last updated: 04-05-2024

To establish whether carriers of LMNA mutations have a hypercoagulable state compared to non-affected family members.

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

Summary

ID

NL-OMON33410

Source
ToetsingOnline

Brief title
Role of coagulation in patients with lamin A-C mutations

Condition

- Cardiac and vascular disorders congenital
- Embolism and thrombosis

Synonym
enhanced coagulation, hypercoagulation

Research involving
Human

Sponsors and support

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

Intervention

Keyword: coagulation, lamin A/C mutations

Outcome measures

Primary outcome

Determination of the coagulable state.

Secondary outcome

nvt

Study description

Background summary

A-type lamins (lamin A and lamin C) are nuclear envelope proteins which are encoded by the LMNA gene. Mutations in the LMNA gene are known to cause different conditions. The diversity of these phenotypes is striking with features such as premature ageing, axonal neuropathy, lipodystrophy and cardiac involvement. Recently an association between arterial thrombosis, independent of the presence of atrial fibrillation, and venous thrombosis and LMNA mutations was determined. The possible role of hypercoagulation however is not established yet.

Study objective

To establish whether carriers of LMNA mutations have a hypercoagulable state compared to non-affected family members.

Study design

Case-control

Study burden and risks

There is a direct benefit for the lamine A/C patients, since the assessment of a hypercoagulable state might prompt the lamine A/C working group to chance the guidelines concerning anticoagulant treatment. The non-carrier family members will benefit, since a general cardiovascular work-up will lead to a cardiovascular risk assessment and might lead to treatment if necessary, in case of high risk or subclinical atherosclerosis. There is no individual risk involved in this research project. The only inconvenience is the venapuncture.

Contacts

Public
Academisch Medisch Centrum

Meibergdreef 9
1105 AZ Amsterdam
Nederland
Scientific
Academisch Medisch Centrum

Meibergdreef 9
1105 AZ Amsterdam
Nederland

Trial sites

Listed location countries

Netherlands

Eligibility criteria

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

Inclusion criteria

Cases: Known LMNA mutation; adult men and women
Controls: non-carrier family members

Exclusion criteria

cases: LMNA mutations not known
controls: LMNA mutations not examinated

Study design

Design

Study type:	Observational invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)

Primary purpose: Basic science

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-06-2009
Enrollment:	90
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	NL28148.018.09