# Coagulation in Lamin A-C mutations

Published: 16-09-2009
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To establish whether carriers of LMNA mutations have a hypercoagulable state compared to non-affected family members.

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|---|---|
| **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 |