Drusen in hereditary angioedema patients, aged 50 years and older.

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What is the contribution of the different variations in the C1-inhibitor gene to the disease AMD?

Ethical review	Approved WMO
Status	Recruiting
Health condition type	Chromosomal abnormalities, gene alterations and gene variants
Study type	Observational invasive

Summary

ID

NL-OMON32467

Source
ToetsingOnline

Brief title
Drusen in HAE patients.

Condition

- Chromosomal abnormalities, gene alterations and gene variants
- Retina, choroid and vitreous haemorrhages and vascular disorders
- Connective tissue disorders (excl congenital)

Synonym
Drusen, retinal waste depositions AND/OR age related macular degeneration

Research involving
Human

Sponsors and support

Primary sponsor: Universitair Medisch Centrum Sint Radboud
Source(s) of monetary or material Support: Ministerie van OC&W,NWO

Intervention

Keyword: age related macular degeneration, C1-inhibitor, drusen, hereditary angioedema

Outcome measures

Primary outcome

The prevalence of AMD and/or drusen in HAE patients.

Secondary outcome

What kind of variations in the C1-inhibitor gene cause AMD and/or drusen.

Study description

Background summary

C1-inhibitor plays a crucial part in suppressing the activity of the classical pathway of the complement cascade. Inhibition of C1 prevents activation of complement components 2 and 4 (C2 and C4) and so has several downstream effects on the complement cascade. Different single-nucleotide polymorphisms in the C1-inhibitor gene are associated with hereditary angioedema. Genotypic variation in the C1-inhibitor gene also showed significant genotypic association with age-related macular degeneration (AMD). A follow-up study showed the absence of any association with AMD in two case-control studies. To test this further, we will test HEA patients, aged 50 years and older for drusen, a precursor of the disease and/or AMD itself.

Study objective

What is the contribution of the different variations in the C1-inhibitor gene to the disease AMD?

Study design

This is a cohort study of 15 HEA patients. The duration of the whole study is approximately 1,5 - 2 hours for each subject.
Before visiting the clinic the subjects receive a questionnaire about lifelong smoking habits, dietary habits, medical history, use of medication and family history of AMD. During their visit to the clinic they first undergo Snellen visual acuity measurement, ophthalmic examination and pupils are dilated with 1.0% tropicamide and 2.5% phenylephrine.
20 Minutes after pupil dilatation we take a venous blood sample for genomic DNA

extraction, make digital nonstereoscopic 30° color fundus photographs (Topcon TRC 50IX digital fundus camera), a Spectralis Domain OCT and a Fluorescein angiograph (Heidelberg Engineering Spectralis HRA2+OCT).

Study burden and risks

Before visiting the clinic the subjects receive a questionnaire at home, what they will return on their visit to the clinic. During their visit of 1,5-2 hours they get an ophthalmic examination, venapunction, fundus photograph, SD-OCT and a fluorescein angiograph. There are no direct benefits or great risks for the subjects.

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

Patients with hereditairy angioedema, aged 50 years and older.

Exclusion criteria

retinal pathology, other than drusen formation and/or age related macular degeneration.

Study design

Design

Study type: Observational invasive

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

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	01-08-2010
Enrollment:	15
Type:	Actual

Ethics review

Approved WMO

Date:	05-03-2010
Application type:	First submission
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)

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	NL30542.091.09