Long-term clinical follow-up study of patients and families with choroideremia for future clinical trials

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The purpose of this study is to collect and complement long-term clinical follow-up data from patients and families with choroideremia. We hope to increase our understanding of the clinical course of choroideremia.

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Eye disorders congenital
Study type	Observational non invasive

Summary

ID

NL-OMON38958

Source
ToetsingOnline

Brief title
Follow-up choroideremia

Condition

- Eye disorders congenital
- Congenital eye disorders (excl glaucoma)

Synonym
CHM, Choroideremia, tapetochoroideal dystrophy (TCD)

Research involving
Human

Sponsors and support

Primary sponsor: Academisch Medisch Centrum
Source(s) of monetary or material Support: UitZicht

Intervention

Keyword: Choroideremia, follow-up

Outcome measures

Primary outcome

To study the correlation between the genotype and the phenotype (clinical symptoms) of choroideremia patients. The clinical symptoms of each patient were investigated during regular examination at the department of ophthalmology of the Academic Medical Centre Amsterdam. The clinical course of the disorder will be investigated on the basis of visual acuity, fundoscopy, fundus photography, fundus autofluorescence, spectral-domain optical coherence tomography (SD-OCT) and visual field tests from the past and the present.

Secondary outcome

Secundairy study parameters are the course of the complaints and subjective limitations in the activities of daily living. These parameters will be tested by a questionnaire, which contains subjective nyctalopia, color vision disorders, photofobia and subjective limitations of the visual field.

Study description

Background summary

Tapetoretinal dystrophy (TRD) is a clinical and genetic heterogeneous group of hereditary retinal disorders. Choroideremia (tapetochoroideal dystrophy: TCD) belongs to this group and is caused by degeneration of the choriocapillaris, retinal pigment epithelium and photoreceptors within the eye. Due to collection of data and careful clinical characterization of choroideremia families from the *Delleman archive* of the Netherlands Institute for Neuroscience, in 1994 the CHM-gene was discovered. The CHM-gene encodes the Rab escort protein-1 (REP-1). To date, this is the only gene associated with choroideremia. Future

and current therapies, such as gene therapy, are designed to prevent or restrain damage to the photoreceptors and therefore should be administered at young age. At this time, gene therapy is the most promising treatment option for incurable hereditary retinal disorders, such as Retinitis Pigmentosa (RP) and Stargardt*s disease (STGD). With this study, we collect and complement long-term clinical follow-up data for choroideremia. Gene therapy is in little applied in Great-Britain. Therapeutic proof-of-principal for gene therapy in mice was delivered and in the autumn of 2011 toxicity determination of gene therapy in a small group of choroideremia patients.

Study objective

The purpose of this study is to collect and complement long-term clinical follow-up data from patients and families with choroideremia. We hope to increase our understanding of the clinical course of choroideremia.

Study design

Longitudinal observiational study.

Study burden and risks

Participants will be asked to undergo routine ophthalmological examination and no invasive interventions will take place. Therefore, the strain and risk are negligible. Participants will be asked to fill in a questionnaire and to visit the hospital once.

Contacts

Public
Academisch Medisch Centrum

Meibergdreef 9
Amsterdam 1105 AZ
NL
Scientific
Academisch Medisch Centrum

Meibergdreef 9
Amsterdam 1105 AZ
NL

Trial sites

Listed location countries

Netherlands

Eligibility criteria

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

Inclusion criteria

Patients who are diagnosed with choroideremia based on ophthalmologic examination by an ophthalmologist and (often) confirmed by DNA-analysis. Patients were included if they are male gender, aged *18 years of age and able to provide written informed consent.

Exclusion criteria

There are no exclusion criteria.

Study design

Design

Study type: Observational non invasive

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

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	17-07-2013

Enrollment:	50
Type:	Actual

Ethics review

Approved WMO Date:	28-05-2013
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	NL44347.018.13